CGT 600 v1.1

Gene	Disease	Transcript	Mutations	Disease.description	products
ABCA4	Cone-rod dystrophy type 3	NM_000350.2	NM_000350.2:c.3540_3555delGTCTAAGGGTTTCTCC, NM_000350.2:c.2616_2617delCT, NM_000350.2:c.4793C>A, NM_000350.2:c.6179T>G, NM_000350.2:c.1222C>T, NM_000350.2:c.763C>T	Cone rod dystrophy type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ABCA4 gene located on chromosomal region 1p22. The age of onset is infantile. This disease is characterized by decreased visual acuity, color vision defects, photoaversion and decreased sensitivity in the central visual field, later followed by progressive loss in peripheral vision and night blindness. The prevalence is 1:100,000-9100,000.	250,6
ABCA4	Retinitis pigmentosa type 19	NM_000350.2	NM_000350.2:c.1848delA	Retinitis pigmentosa type 19 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ABCA4 gene located on chromosomal region 1p22. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000-5:10,000.	250,6
ABCA4	Stargardt disease type 1	NM_000350.2	NM_000350.2:c.1018T>G, NM_000350.2:c.4457C>T, NM_000350.2:c.1225delA, NM_000350.2:c.1622T>C, NM_000350.2:c.1715G>A, NM_000350.2:c.1755delA, NM_000350.2:c.1771delT, NM_000350.2:c.1804C>T, NM_000350.2:c.6449G>A, NM_000350.2:c.1938-1G>A, NM_000350.2:c.1964T>G, NM_000350.2:c.2160+1G>T, NM_000350.2:c.2588G>C, NM_000350.2:c.4469G>A, NM_000350.2:c.2690C>T, NM_000350.2:c.2791G>A, NM_000350.2:c.286A>G, NM_000350.2:c.2971G>C, NM_000350.2:c.3083C>T, NM_000350.2:c.3106G>A, NM_000350.2:c.3210_3211dupGT, NM_000350.2:c.3364G>A, NM_000350.2:c.6320G>A, NM_000350.2:c.3970delG, NM_000350.2:c.4139C>T, NM_000350.2:c.4429C>T, NM_000350.2:c.2300T>A, NM_000350.2:c.3322C>T, NM_000350.2:c.52C>T, NM_000350.2:c.5512delC, NM_000350.2:c.5819T>C, NM_000350.2:c.5881G>A, NM_000350.2:c.5882G>A, NM_000350.2:c.5912T>G, NM_000350.2:c.634C>T, NM_000350.2:c.5714+5G>A, NM_000350.2:c.6394G>T, NM_000350.2:c.67-2A>G, NM_000350.2:c.5461-10T>C, NM_000350.2:c.6089G>A, NM_000350.2:c.6118C>T, NM_000350.2:c.6148G>C, NM_000350.2:c.661G>A, NM_000350.2:c.5338C>G	Stargardt disease type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ABCA4 gene located on chromosomal region 1p22. The age of onset is infantile. This disease is characterized by progressive central vision loss, mild loss of color vision, delayed dark adaptation and macular atrophy with or without paramacular flecks and degeneration of the underlying retinal pigment ephitelium. The prevalence is 1:10,000- 5:10,000.	250,6
ABCB7	Sideroblastic anemia and ataxia, X-linked	NM_004299.4	NM_004299.4:c.1203T>G, NM_004299.4:c.1234G>C, NM_004299.4:c.1300G>A	X-linked sideroblastic anemia with ataxia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ABCB7 gene located on chromosomal region Xq13.3. The age of onset is neonatal/infantile. This disease is characterized by core neurological features including motor delay, ataxia evident from early childhood, and dysarthria and patients usually have a mild asymptomatic anaemia or a borderline decreased mean corpuscular volume. The prevalence is <1:1,000,000.	600
ACAD9	Acyl-CoA dehydrogenase type 9 deficiency	NM_014049.4	NM_014049.4:c.1240C>T, NM_014049.4:c.1249C>T, NM_014049.4:c.130T>A, NM_014049.4:c.1594C>T, NM_014049.4:c.23delT, NM_014049.4:c.358delT, NM_014049.4:c.797G>A, NM_014049.4:c.976G>C, NM_014049.4:c.453+1G>A	Acyl-CoA dehydrogenase type 9 deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACAD9 gene located on chromosomal region 3q21.3. The age of onset is neonatal/infantile. This disease is characterized by failure to thrive, hypertrophic cardiomyopathy, exercise intolerance and mild to severe neurological dysfunction.	250,6
ACADM	Acyl-CoA dehydrogenase deficiency, medium-chain	NM_000016.5	NM_000016.5:c.1102_1105delTTAG, NM_000016.5:c.1232_1233delAA, NM_000016.5:c.287-2A>G, NM_000016.5:c.362C>T, NM_000016.5:c.447G>A, NM_000016.5:c.447G>T, NM_000016.5:c.449_452delCTGA, NM_000016.5:c.616C>T, NM_000016.5:c.617G>A, NM_000016.5:c.683C>A, NM_000016.5:c.797A>G, NM_000016.5:c.799G>A, NM_000016.5:c.815_827delTTGCAATGGGAGC, NM_000016.5:c.890A>G, NM_000016.5:c.984delG, NM_000016.5:c.985A>G, NM_000016.5:c.127G>A, NM_000016.5:c.734C>T, NM_000016.5:c.250C>T	Medium chain acyl-CoA dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACADM gene located on chromosomal region 1p31. The age of onset is neonatal/infantile. This disease is characterized by a rapidly progressive metabolic crisis, often presenting as hypoketotic hypoglycemia, lethargy, vomiting, seizures and coma. The prevalence is 1:4,900-1:27,000 in Caucasian populations and 1:14,600 in worldwide populations.	250,6
ACADS	Acyl-CoA dehydrogenase deficiency, short-chain	NM_000017.2	NM_000017.2:c.1095G>T, NM_000017.2:c.1108A>G, NM_000017.2:c.1147C>T, NM_000017.2:c.136C>T, NM_000017.2:c.319C>T, NM_000017.2:c.417G>C, NM_000017.2:c.529T>C, NM_000017.2:c.561_568delCAATGCCT, NM_000017.2:c.826G>A, NM_000017.2:c.314T>A	Short chain acyl-CoA dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACADS gene located on chromosomal region 12q24.31. The age of onset is infantile. This disease is characterized by seizures, developmental delay, failure to grow with poor feeding, and usually muscle weakness and hypotonia. The prevalence is <1:50,000.	250,6

ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	NM_001609.3	NM_001609.3:c.1159G>A, NM_001609.3:c.443C>T, NM_001609.3:c.763C>T, NM_001609.3:c.621G>A, NM_001609.3:c.303+1G>A	2-Methylbutyryl-CoA dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACADSB gene located on chromosomal region 10q26.3. The age of onset is neonatal/infantile. This disease is characterized by muscle hypotonia, cerebral palsy, developmental delay, lethargy, hypoglycemia, and metabolic acidosis. The prevalence is <1:1,000,000.	250,6
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	NM_000018.3	NM_000018.3:c.1096C>T, NM_000018.3:c.1097G>A, NM_000018.3:c.1106T>C, NM_000018.3:c.1141_1143delGAG, NM_000018.3:c.1182+1G>A, NM_000018.3:c.1357C>T, NM_000018.3:c.1360G>A, NM_000018.3:c.1375dupC, NM_000018.3:c.1389dupG, NM_000018.3:c.1406G>A, NM_000018.3:c.1468G>C, NM_000018.3:c.1532+1G>A, NM_000018.3:c.1837C>T, NM_000018.3:c.1843C>T, NM_000018.3:c.1882delC, NM_000018.3:c.278-1G>A, NM_000018.3:c.298_299delCA, NM_000018.3:c.343delG, NM_000018.3:c.400C>T, NM_000018.3:c.477+1G>C, NM_000018.3:c.520G>A, NM_000018.3:c.685C>T, NM_000018.3:c.739A>C, NM_000018.3:c.753-2A>C, NM_000018.3:c.896_898delAGA, NM_000018.3:c.917T>C, NM_000018.3:c.1844G>A, NM_000018.3:c.848T>C	Very long chain acyl-CoA dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACADVL gene located on chromosomal region 17p13.1. The age of onset is neonatal/infantile. This disease is characterized by cardiomyopathy, hypoketotic hypoglycemia, liver disease, exercise intolerance and rhabdomyolysis. The prevalence is 1:100,000-9:100,000.	250,6
ACAT1	Beta-ketothiolase deficiency	NM_000019.3	NM_000019.3:c.1035_1037delAGA, NM_000019.3:c.1083dupA, NM_000019.3:c.1136G>T, NM_000019.3:c.1138G>A, NM_000019.3:c.2T>A, NM_000019.3:c.410_417delCTCAAAGT, NM_000019.3:c.547G>A, NM_000019.3:c.622C>T, NM_000019.3:c.905delA	Beta-ketothiolase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACAT1 gene located on chromosomal region 11q22.3. The age of onset is neonatal/infantile. This disease is characterized by normal early development followed by a progressive loss of mental and motor skills. The prevalence is < 1:1,000,000.	600
ACE	Renal tubular dysgenesis	NM_000789.3	NM_000789.3:c.1319_1322delTGGA, NM_000789.3:c.1510delC, NM_000789.3:c.3381-4C>T, NM_000789.3:c.798C>G, NM_000789.3:c.1486C>T, NM_000789.3:c.2371C>T, NM_000789.3:c.1587-2A>G	Renal tubular dysgenesis deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACE (chromosomal region 17q23.3), AGT (1q42.2) AGTR1 (3q24) and REN (1q32.1) genes. The age of onset is fetal. This disease is characterized by absent or poorly developed proximal tubules of the kidneys, persistent oligohydramnios, leading to Potter sequence, and skull ossification defects.	250,6
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	NM_004035.6	NM_004035.6:c.832A>G, NM_004035.6:c.532G>T, NM_004035.6:c.591delG	Peroxisomal acyl-CoA oxidase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACOX1 gene located on chromosomal region 17q25.1. The age of onset is neonatal/infantile. This disease is characterized by hypotonia and seizures in the neonatal period and neurological regression in early infancy. The prevalence is < 1:1,000,000.	600
ACTN4	Glomerulosclerosis, focal segmental, type 1	NM_004924.4	NM_004924.4:c.763A>G, NM_004924.4:c.2619_2620insC, NM_004924.4:c.776C>T, NM_004924.4:c.784T>C	Focal segmental glomerulosclerosis type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACTN4 gene located on chromosomal region 19q13.2. The age of onset is variable. This disease is characterized by severe proteinuria with low serum albumin and possible edemas.	600
ADA	Adenosine deaminase deficiency	NM_000022.2	NM_000022.2:c.226C>T, NM_000022.2:c.632G>A, NM_000022.2:c.890C>A, NM_000022.2:c.247G>A, NM_000022.2:c.320T>C, NM_000022.2:c.872C>T, NM_000022.2:c.956_960delAAGAG, NM_000022.2:c.986C>T	Adenosine deaminase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ADA gene located on chromosomal region 20q13.12. The age of onset is neonatal/infantile. This disease is characterized by profound lymphopenia and very low immunoglobulin levels of all isotypes resulting in severe and recurrent opportunistic infections. The annual incidence is 1:200,000-1:1,000,000. The prevalence is 1:100,000-9:100,000.	250,6
ADAMTS2	Ehlers-Danlos syndrome type 7C	NM_014244.4	NM_014244.4:c.2384G>A	Ehlers-Danlos syndrome type 7C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ADAMTS2 gene located on chromosomal region 5q35.3. The age of onset is neonatal/infantile. This disease is characterized by extremely fragile tissues, hyperextensible skin and easy bruising. The prevalence is <1:1,000,000.	600
ADAMTSL2	Geleophysic dysplasia type 1	NM_014694.3	NM_014694.3:c.338G>A, NM_014694.3:c.440C>T, NM_014694.3:c.661C>T, NM_014694.3:c.340G>A	Geleophysic dysplasia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ADAMTSL2 gene located on chromosomal region 9q34.2. The age of onset is infantile. This disease is characterized by extremely by short stature, prominent abnormalities in hands and feet, and a characteristic facial appearance. The prevalence is <1:1,000,000.	600
ADCK3	Primary coenzyme Q10 deficiency type 4	NM_020247.4	NM_020247.4:c.911C>T, NM_020247.4:c.815G>T, NM_020247.4:c.993C>T, NM_020247.4:c.1541A>G, NM_020247.4:c.1645G>A, NM_020247.4:c.1651G>A, NM_020247.4:c.1750_1752delACC, NM_020247.4:c.1813_1814insG, NM_020247.4:c.589-3C>G, NM_020247.4:c.637C>T, NM_020247.4:c.815G>A	Primary coenzyme Q10 deficiency type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ADCK3 gene located on chromosomal region 1q42.13. The age of onset is infantile. This disease is characterized by progressive ataxia, cerebellar atrophy, and often exercise intolerance with elevated lactate levels and mild intellectual deficit.	250,6

AGA	Aspartylglucosaminuria	NM_000027.3	NM_000027.3:c.488G>C, NM_000027.3:c.755G>A, NM_000027.3:c.214T>C, NM_000027.3:c.302C>T, NM_000027.3:c.800dupT, NM_000027.3:c.904G>A	Aspartylglucosaminuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AGA gene located on chromosomal region 4q34.3. The age of onset is infantile. This disease is characterized by slowly developping mental retardation, beginning with clumsiness, late speech, and hyperkinesia, mild facial dysmorphism, and slight kyphoscoliosis.	600
AGL	Glycogen storage disease type 3	NM_000642.2	NM_000642.2:c.1783C>T, NM_000642.2:c.18_19delGA, NM_000642.2:c.112A>G, NM_000642.2:c.1222C>T, NM_000642.2:c.1481G>A, NM_000642.2:c.1485delT, NM_000642.2:c.16C>T, NM_000642.2:c.4260-1G>T, NM_000642.2:c.3214_3215delGA, NM_000642.2:c.1999delC, NM_000642.2:c.2039G>A, NM_000642.2:c.2590C>T, NM_000642.2:c.4456delT, NM_000642.2:c.3216_3217delGA, NM_000642.2:c.3980G>A, NM_000642.2:c.4342G>C, NM_000642.2:c.4529dupA, NM_000642.2:c.294-2A>T, NM_000642.2:c.4260-12A>G	Glycogen storage disease type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AGL gene located on chromosomal region 1p21.2. The age of onset is infantile. This disease is characterized by hepatomegaly, growth retardation and occasional seizures related to hypoglycemia and frequently muscular hypotonia and hypertrophic cardiomyopathy.	250,6
AGPS	Rhizomelic chondrodysplasia punctata type 3	NM_003659.3	NM_003659.3:c.1256G>A, NM_003659.3:c.926C>T, NM_003659.3:c.1406T>C, NM_003659.3:c.1703C>T	Rhizomelic chondrodysplasia punctata type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AGPS gene located on chromosomal region 2q31.2. The age of onset is neonatal/infantile. This disease is characterized by shortness of the femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe intellectual deficit. The prevalence is 1:100,000-9:100,000.	600
AGT	Renal tubular dysgenesis	NM_000029.3	NM_000029.3:c.1124G>A, NM_000029.3:c.604C>T, NM_000029.3:c.1290_1291insT, NM_000029.3:c.1290delT	Renal tubular dysgenesis deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACE (chromosomal region 17q23.3), AGT (1q42.2) AGTR1 (3q24) and REN (1q32.1) genes. The age of onset is fetal. This disease is characterized by absent or poorly developed proximal tubules of the kidneys, persistent oligohydramnios, leading to Potter sequence, and skull ossification defects.	600
AGTR1	Renal tubular dysgenesis	NM_031850.3	NM_031850.3:c.481delC, NM_031850.3:c.259dupG, NM_031850.3:c.215dupT, NM_031850.3:c.481C>T	Renal tubular dysgenesis deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACE (chromosomal region 17q23.3), AGT (1q42.2) AGTR1 (3q24) and REN (1q32.1) genes. The age of onset is fetal. This disease is characterized by absent or poorly developed proximal tubules of the kidneys, persistent oligohydramnios, leading to Potter sequence, and skull ossification defects.	600
AGXT	Primary hyperoxaluria type 1	NM_000030.2	NM_000030.2:c.166-2A>G, NM_000030.2:c.121G>A, NM_000030.2:c.32C>A, NM_000030.2:c.245G>A, NM_000030.2:c.25_26insC, NM_000030.2:c.322T>C, NM_000030.2:c.508G>A, NM_000030.2:c.560C>T, NM_000030.2:c.590G>A, NM_000030.2:c.613T>C, NM_000030.2:c.697C>T, NM_000030.2:c.698G>A, NM_000030.2:c.731T>C, NM_000030.2:c.738G>A, NM_000030.2:c.836T>C, NM_000030.2:c.860G>A, NM_000030.2:c.33_34insC, NM_000030.2:c.454T>A, NM_000030.2:c.466G>A, NM_000030.2:c.248A>G	Primary hyperoxaluria type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AGXT gene located on chromosomal region 2q37.3. The age of onset is variable. This disease is characterized by variable clinical presentation, ranging from occasional symptomatic nephrolithiasis to nephrocalcinosis and end-stage renal disease with systemic involvement. The prevalence is 1:1,000,000-9:1,000,000.	250,6
AHI1	Joubert syndrome type 3	NM_017651.4	NM_017651.4:c.1303C>T, NM_017651.4:c.1484G>A, NM_017651.4:c.2295_2296insA, NM_017651.4:c.2295dupA, NM_017651.4:c.3257A>G, NM_017651.4:c.2168G>A, NM_017651.4:c.985C>T, NM_017651.4:c.989A>G, NM_017651.4:c.3263_3264delGG, NM_017651.4:c.1051C>T, NM_017651.4:c.1052G>T	El sÃ-ndrome de Joubert tipo 3 sigue un patrÃ³n de herencia autosÃ³mico recesivo y estÃ¡ causado por variantes patogÃ©nicas en el gen AHI1 localizado en la regiÃ³n cromosÃ³mica 6q23.3. La edad de aparicIÃ³n es neonatal/infantil con sÃ-ntomas como los rasgos neurolÃ³gicos del sÃ-ndrome de Joubert (hipotonÃ-a neonatal, retraso del desarrollo, discapacidad intelectural de leve a grave, ataxia, movimiento ocular anormal incluyendo apraxia oculomotora y nistagmo en posiciÃ³n primaria) asociados a una distrofia retiniana.	250,6
AIPL1	Cone-rod dystrophy	NM_014336.4	NM_014336.4:c.1053_1064delTGCAGAGCCACC	La distrofia de conos y bastones causada por variantes patogÃ©nicas en el gen AIPL1 localizado en la regiÃ³n cromosÃ³mica 17p13.2 sigue un patrÃ³n de herencia autosÃ³mico recesivo. La edad de aparicIÃ³n es temprana. Se caracteriza por una agudeza visual disminuida, defectos en la visiÃ³n de los colores, fotoaversiÃ³n y disminuciÃ³n de la sensibilidad en el centro del campo visual, seguido por una pÃ©rdida de la visiÃ³n perifÃ©rica y ceguera nocturna.	250,6
AIPL1	Leber congenital amaurosis type 4	NM_014336.4	NM_014336.4:c.905G>T, NM_014336.4:c.834G>A, NM_014336.4:c.589G>C, NM_014336.4:c.715T>C	Leber congenital amaurosis type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AIPL1gene located on chromosomal region 17p13.1. The age of onset is neonatal/infantile. This disease is characterized by blindness, nystagmus, roving eye movement and lack of detectable signals on an electroretinogram, leading to severe visual impairment within the first year of life. The prevalence is 1:100,000-9:100,000.	250,6

ALAS2	Erythropoietic protoporphyria	NM_000032.4	NM_000032.4:c.1699_1700delAT, NM_000032.4:c.1706_1709delAGTG	Erythropoietic protoporphyria caused by pathogenic variants in the ALAS2 gene located on chromosomal region Xp11.21 follows a dominant X-linked pattern of inheritance. The age of onset is neonatal/infancy. This disease is characterized by cutaneous manifestations of acute painful photosensitivity with erythema and edema, sometimes with petechiae, together with stinging and burning sensations without blistering, upon exposure to sunlight or artificial light (400-700 nm). These episodes have a variable severity depending on the exposure duration and may result in chronic permanent lesions on exposed skin. There is a risk of .cholelithiasis with obstructive episodes, and chronic liver disease that might evolve to acute liver failure. The global prevalence is ranging between 1/75,000 and 1/200,000.	600
ALAS2	Sideroblastic anemia, X-linked	NM_000032.4	NM_000032.4:c.1354C>T	X-linked sideroblastic anemia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ALAS2 gene located on chromosomal region Xp11.21. The age of onset is variable. This disease is characterized by clinical features of anemia and/or iron overload such as pallor, fatigue, weakness, and more rarely breathlessness, mild splenomegaly, cardiac problems, abnormal liver function, hyperglycemia, glucose intolerance and skin hyperpigmentation.	600
ALDH4A1	Hyperprolinemia type 2	NM_003748.3	NM_003748.3:c.1055C>T	Hyperprolinemia type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALDH4A1 gene located on chromosomal region 1p36. The age of onset is variable. This disease is characterized by seizures, intellectual deficit and mild developmental delay.	600
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	NM_001080.3	NM_001080.3:c.1234C>T, NM_001080.3:c.1226G>A, NM_001080.3:c.901A>G, NM_001080.3:c.1540C>T, NM_001080.3:c.1579C>T, NM_001080.3:c.612G>A, NM_001080.3:c.803G>A, NM_001080.3:c.862A>G	Succinic semialdehyde dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALDH451 gene located on chromosomal region 6p22. The age of onset is infantile. This disease is characterized by psychomotor retardation, delayed speech development, hypotonia and ataxia. It is a rare disease with around 350 cases reported.	600
ALDOA	Glycogen storage disease type 12	NM_000034.3	NM_000034.3:c.619G>A, NM_000034.3:c.386A>G	Glycogen storage disease type 12 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALDOA gene located on chromosomal region 16p11.2. The age of onset is neonatal/infantile. This disease is characterized by myopathy with exercise intolerance and rhabdomyolysis associated with hemolytic anaemia.Â	600
ALDOB	Hereditary fructose intolerance	NM_000035.3	NM_000035.3:c.1005C>G, NM_000035.3:c.178C>T, NM_000035.3:c.1027T>C, NM_000035.3:c.10C>T, NM_000035.3:c.136A>T, NM_000035.3:c.448G>C, NM_000035.3:c.2T>C, NM_000035.3:c.360_363delCAAA, NM_000035.3:c.442T>C, NM_000035.3:c.1013C>T, NM_000035.3:c.113-1_115delGGTA, NM_000035.3:c.1067C>A, NM_000035.3:c.612T>A, NM_000035.3:c.720C>A, NM_000035.3:c.524C>A	Hereditary fructose intolerance follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALDOB gene located on chromosomal region 9q21.3-q22.2. The age of onset is neonatal/infantile. This disease is characterized by severe abdominal pain, vomiting, and hypoglycemia following ingestion of fructose or other sugars metabolised through fructose-1-phosphate. The prevalence is 1:100,000-9:100,000.	250,6
ALG1	Congenital disorders of glycosylation type 1k	NM_019109.4	NM_019109.4:c.1187+1G>A, NM_019109.4:c.1079C>T, NM_019109.4:c.1129A>G, NM_019109.4:c.901+1G>A, NM_019109.4:c.434G>A, NM_019109.4:c.450C>G, NM_019109.4:c.773C>T	Congenital disorder of glycosylation type 1k follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALG1 gene located on chromosomal region 16p13.3. The age of onset is neonatal/infantile. This disease is characterized by psychomotor delay, seizures, microcephaly and coagulation anomalies. The prevalence is <1:1,000,000.	600
ALG6	Congenital disorders of glycosylation type Ic	NM_013339.3	NM_013339.3:c.897_899delAAT, NM_013339.3:c.998C>T, NM_013339.3:c.495-3C>G, NM_013339.3:c.53G>A, NM_013339.3:c.316C>T, NM_013339.3:c.482A>G, NM_013339.3:c.1432T>C	Congenital disorder of glycosylation type 1c follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALG6 gene located on chromosomal region 1p31.3. The age of onset is neonatal/infantile. This disease is characterized by psychomotor delay and muscular hypotonia, and possible coagulation anomalies, hormonal abnormalities and seizures. The prevalence is <1:1,000,000.	250,6
ALMS1	AlstrÃ¶m syndrome	NM_015120.4	NM_015120.4:c.11443C>T, NM_015120.4:c.10775delC, NM_015120.4:c.11316_11319delAGAG, NM_015120.4:c.2323C>T, NM_015120.4:c.11449C>T, NM_015120.4:c.11452_11453insA, NM_015120.4:c.1574_1576delCTCinsT, NM_015120.4:c.8383C>T, NM_015120.4:c.9612_9616delAACAG, NM_015120.4:c.10579_10580delAT, NM_015120.4:c.11610_11611delCT, NM_015120.4:c.12439C>T, NM_015120.4:c.12445C>T, NM_015120.4:c.891_907delTCAGCACCCGCTTATAG, NM_015120.4:c.9911-1G>A, NM_015120.4:c.11618_11619delCT, NM_015120.4:c.4245delC, NM_015120.4:c.5584C>T, NM_015120.4:c.8164C>T	AlstrÃ¶m syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALMS1 gene located on chromosomal region 2p13.1. The age of onset is neonatal/infantile. This disease is characterized by cone-rod dystrophy, hearing loss, obesity, insulin resistance and hyperinsulinemia, type 2 diabetes mellitus, dilated cardiomyopathy and progressive hepatic and renal dysfunction. The prevalence is 1:10,000-1:1,000,000.	250,6

ALPL	Hypophosphatasia	NM_000478.4	NM_000478.4:c.1001G>A, NM_000478.4:c.1366G>A, NM_000478.4:c.211C>T, NM_000478.4:c.212G>C, NM_000478.4:c.323C>T, NM_000478.4:c.346G>A, NM_000478.4:c.407G>A, NM_000478.4:c.526G>A, NM_000478.4:c.535G>A, NM_000478.4:c.571G>A, NM_000478.4:c.620A>C, NM_000478.4:c.1133A>T, NM_000478.4:c.1250A>G, NM_000478.4:c.1306T>C, NM_000478.4:c.98C>T, NM_000478.4:c.1574delG, NM_000478.4:c.892G>A, NM_000478.4:c.814C>T, NM_000478.4:c.881A>C	Childhood-onset hypophosphatasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALPL gene located on chromosomal region 1p36.12. The age of onset is infantile. This disease is characterized by clinical features ranging from stillbirth without mineralized bone to pathologic fractures of the lower extremities in later adulthood.	600
AMACR	Alpha-methylacyl-CoA racemase deficiency	NM_014324.5	NM_014324.5:c.857delT, NM_014324.5:c.320T>C, NM_014324.5:c.43delG, NM_014324.5:c.154T>C	Alpha-methylacyl-Coa racemase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AMACR gene located on chromosomal region 5p13. The age of onset is variable. This disease is characterized by mild cholestatic liver disease, fat malabsorption and/or neurological disease. The prevalence is <1:1,000,000.	600
AMT	Glycine encephalopathy	NM_000481.3	NM_000481.3:c.139G>A, NM_000481.3:c.125A>G, NM_000481.3:c.959G>A, NM_000481.3:c.574C>T, NM_000481.3:c.806G>A, NM_000481.3:c.826G>C, NM_000481.3:c.259-1G>C	Glycine encephalopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AMT and GLDC genes located on chromosomal regions 3p21.31 and 9p24.1 respectively. The age of onset is neonatal/infantile. This disease is characterized by lethargy or even coma, hypotonia, hiccups, myoclonic jerks, and breathing/swallowing disorders, with subsequent intellectual deficit, spasticity and intractable seizures. The prevalence is 1:1,000,000-9:1,000,000.	600
ANO5	Limb-girdle muscular dystrophy type 2L, autosomal recessive	NM_213599.2	NM_213599.2:c.155A>G, NM_213599.2:c.1622_1623insA, NM_213599.2:c.1407+5G>A, NM_213599.2:c.1887delA, NM_213599.2:c.1733T>C, NM_213599.2:c.692G>T, NM_213599.2:c.1627_1628insA, NM_213599.2:c.172C>T, NM_213599.2:c.206_207delAT, NM_213599.2:c.1210C>T, NM_213599.2:c.1295C>G, NM_213599.2:c.1914G>A, NM_213599.2:c.184_185insA, NM_213599.2:c.1898+1G>A, NM_213599.2:c.191_192insA	Autosomal recessive limb-girdle muscular dystrophy type 2L follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ANO5 gene located on chromosomal region 11p14.3. The age of onset is adult. This disease is characterized by weakness and wasting restricted to the limb musculature, proximal greater than distal, and muscle degeneration/regeneration on muscle biopsy. The prevalence is <1:1,000,000.	250,6
APTX	Ataxia with oculomotor apraxia type 1	NM_175073.2	NM_175073.2:c.167delT, NM_175073.2:c.788T>G, NM_175073.2:c.320delC, NM_175073.2:c.617C>T, NM_175073.2:c.659C>T, NM_175073.2:c.134-2A>G, NM_175073.2:c.166C>T, NM_175073.2:c.124C>T, NM_175073.2:c.875-1G>A, NM_175073.2:c.837G>A, NM_175073.2:c.596G>A	Ataxia with oculomotor apraxia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the APTX gene located on chromosomal region 9p13.1. The age of onset is infantile. This disease is characterized by a progressive cerebellar ataxia associated with oculomotor apraxia, choeroathetosis and severe peripheral neuropathy. The prevalence is 0,4:100.000 in Portugal.	250,6
AR	Androgen insensitivity syndrome	NM_000044.3	NM_000044.3:c.2650A>T, NM_000044.3:c.340C>T, NM_000044.3:c.1937C>A, NM_000044.3:c.2323C>T, NM_000044.3:c.2391G>A, NM_000044.3:c.2567G>A, NM_000044.3:c.1769-11T>A, NM_000044.3:c.1771A>T, NM_000044.3:c.2395C>G	Androgen insensitivity syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the AR gene located on chromosomal region Xq12. The age of onset is variable. This disease is characterized by the presence of female external genitalia in a 46,XY individual with normal testis development but undescended testes and unresponsiveness to age-appropriate levels of androgens. The prevalence is 2:100,000-5:100,000.	250,6
ARG1	Argininemia	NM_000045.3	NM_000045.3:c.61C>T, NM_000045.3:c.365G>A, NM_000045.3:c.413G>T, NM_000045.3:c.871C>T, NM_000045.3:c.32T>C, NM_000045.3:c.703G>C, NM_000045.3:c.869C>G	Argininemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARG1gene located on chromosomal region 6q23. The age of onset is neonatal/infantile. This disease is characterized by variable degrees of hyperammonemia, developing from about 3 years of age, and leading to progressive loss of developmental milestones and spasticity in the absence of treatment. The prevalence is 1:350,000-1:1,000,000.	600
ARL13B	Joubert syndrome type 8	NM_182896.2	NM_182896.2:c.1186C>G, NM_182896.2:c.246G>A, NM_182896.2:c.1252C>T, NM_182896.2:c.598C>T	Joubert syndrome type 8 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARL13B gene located on chromosomal region 3q11.1. The age of onset is neonatal/infantile. This disease is characterized by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis leading to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia and delay in achieving motor milestones. The prevalence is 1/80,000 to 1/100,000.	600
ARL6	Bardet-Biedl syndrome type 3	NM_177976.2	NM_177976.2:c.4G>T, NM_177976.2:c.92C>G, NM_177976.2:c.281T>C, NM_177976.2:c.92C>T, NM_177976.2:c.431C>T, NM_177976.2:c.364C>T	Bardet-Biedl syndrome type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARL6 gene located on chromosomal region 3q11.2. The age of onset is early. A syndrome characterized by usually severe pigmentary retinopathy, early-onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease.	600
ARL6	Retinitis pigmentosa type 55	NM_177976.2	NM_177976.2:c.266C>T	Retinitis pigmentosa type 55 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARL6 gene located on chromosomal region 3q11.2. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000-5:10,000.	600

ARSA	Metachromatic leukodystrophy	NM_000487.5	NM_000487.5:c.1241delC, NM_000487.5:c.1283C>T, NM_000487.5:c.346C>T, NM_000487.5:c.34delG, NM_000487.5:c.1210+1G>A, NM_000487.5:c.1232C>T, NM_000487.5:c.582delC, NM_000487.5:c.583delT, NM_000487.5:c.542dupT, NM_000487.5:c.542T>G, NM_000487.5:c.1408_1418delGCAGCTGTGAC, NM_000487.5:c.195delC, NM_000487.5:c.641C>T, NM_000487.5:c.1401_1411delGTTAGACGCAG, NM_000487.5:c.869G>A, NM_000487.5:c.869G>T, NM_000487.5:c.883G>A, NM_000487.5:c.899T>C, NM_000487.5:c.931G>A, NM_000487.5:c.937C>T, NM_000487.5:c.938G>A, NM_000487.5:c.979G>A, NM_000487.5:c.737G>A, NM_000487.5:c.739G>A, NM_000487.5:c.763G>A, NM_000487.5:c.827C>T, NM_000487.5:c.854+1G>A, NM_000487.5:c.1108-2A>G, NM_000487.5:c.1125_1126delCT, NM_000487.5:c.1150G>A, NM_000487.5:c.1174C>T, NM_000487.5:c.1175G>A, NM_000487.5:c.986C>T, NM_000487.5:c.991G>T, NM_000487.5:c.465+1G>A, NM_000487.5:c.257G>A, NM_000487.5:c.293C>T, NM_000487.5:c.302G>A	Metachromatic leukodystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARSA gene located on chromosomal region 22q13.33. The age of onset is variable. This disease is characterized by hypotonia, walking difficulties, optic atrophy and motor regression preceding mental impairment in the late infantile form, arrested intellectual development, followed by motor regression, epileptic seizures and ataxia in the juvenile form, and motor or psychiatric disorders, but with slow progression in the adult form. The incidence is 0.5:5,000-1:50,000Â and the prevalence is 1:10,000 -5/10,000.	250,6
ARSB	Mucopolysaccharidosis type 6	NM_000046.3	NM_000046.3:c.410G>T, NM_000046.3:c.427delG, NM_000046.3:c.349T>C, NM_000046.3:c.389C>T, NM_000046.3:c.937C>G, NM_000046.3:c.944G>A, NM_000046.3:c.971G>T, NM_000046.3:c.979C>T, NM_000046.3:c.1562G>A, NM_000046.3:c.629A>G, NM_000046.3:c.1143-1G>C, NM_000046.3:c.571C>T, NM_000046.3:c.589C>T, NM_000046.3:c.1178A>C, NM_000046.3:c.1214G>A, NM_000046.3:c.1143-8T>G, NM_000046.3:c.1161dupC, NM_000046.3:c.707T>C, NM_000046.3:c.753C>G, NM_000046.3:c.1366C>T, NM_000046.3:c.1438_1439insG, NM_000046.3:c.921delA	Mucopolysaccharidosis type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ARSB gene located on chromosomal region 5q14.1. The age of onset is infantile. This disease is characterized by educed pulmonary function, hepatosplenomegaly, hearing loss, sleep apnea, corneal clouding, carpal tunnel disease and occasionally central nervous system findings may include cervical cord compression caused by cervical spinal instability, meningeal thickening and/or bony stenosis, communicating hydrocephalus, optic nerve atrophy and blindness. The prevalence is 1:250,000-1:600,000 newborns.	250,6
ARSE	Chondrodysplasia punctata type 1, X-linked	NM_000047.2	NM_000047.2:c.119T>G, NM_000047.2:c.1429delG, NM_000047.2:c.1442C>T, NM_000047.2:c.1732C>T, NM_000047.2:c.1743G>A, NM_000047.2:c.24-1G>A, NM_000047.2:c.410G>C, NM_000047.2:c.410G>T	X-linked chondrodysplasia punctata type 1 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ARSE gene located on chromosomal region Xp22.33. The age of onset is neonatal. This disease is characterized by chondrodysplasia punctata (stippled epiphyses), brachytelephalangy (shortening of the distal phalanges), and nasomaxillary hypoplasia. The prevalence is 1:500,000.	250,6
ARX	Epileptic encephalopathy, early infantile, type 1	NM_139058.2	NM_139058.2:c.1058C>T	Early infantile epileptic encephalopathy type 1 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ARX gene located on chromosomal region Xp21.3. The age of onset is neonatal/infantile. This disease is characterized by poor suckling reflexes, hypotonia and generalized and symmetrical tonic spasms. The prevalence is 1:500,000.	600
ARX	Lissencephaly with abnormal genitalia, X-linked	NM_139058.2	NM_139058.2:c.980_983delAACA	X-linked lissencephaly with abnormal genitalia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ARX gene located on chromosomal region Xp21.3. The age of onset is neonatal/infantile. It is a severe neurological disorder that only manifests in genotypic males and includes lissencephaly with posterior-to-anterior gradient and only moderate increase in thickness of the cortex, absent corpus callosum, neonatal-onset severe epilepsy, hypothalamic dysfunction including defective temperature regulation, and ambiguous genitalia with micropenis and cryptorchidism.	600
ASL	Argininosuccinic aciduria	NM_000048.3	NM_000048.3:c.1135C>T, NM_000048.3:c.1060C>T, NM_000048.3:c.1255_1256delCT, NM_000048.3:c.1366C>T, NM_000048.3:c.1045_1057delGTCATCTCTACGC, NM_000048.3:c.578G>A, NM_000048.3:c.539T>G, NM_000048.3:c.544C>T, NM_000048.3:c.557G>A, NM_000048.3:c.1144-2A>G, NM_000048.3:c.602+1G>A, NM_000048.3:c.857A>G, NM_000048.3:c.925G>A, NM_000048.3:c.446+1G>A, NM_000048.3:c.505T>C, NM_000048.3:c.525-2A>T, NM_000048.3:c.532G>A, NM_000048.3:c.337C>T, NM_000048.3:c.346C>T, NM_000048.3:c.35G>A, NM_000048.3:c.1369dupG, NM_000048.3:c.437G>A, NM_000048.3:c.392C>T, NM_000048.3:c.1153C>T	Argininosuccinic aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ASL gene located on chromosomal region 7q11.21. The age of onset is infantile. This disease is characterized by severe hyperammonemic coma, hypotonia, growth failure, anorexia and chronic vomiting or behavioral disorders during childhood, and hyperammonemic coma or behavioral disorders that simulate psychiatric disorders later in life. The prevalence is 1:70,000 newborns.	250,6
ASPA	Canavan disease	NM_000049.2	NM_000049.2:c.838C>T, NM_000049.2:c.693C>A, NM_000049.2:c.654C>A, NM_000049.2:c.433-2A>G, NM_000049.2:c.854A>C, NM_000049.2:c.914C>A, NM_000049.2:c.212G>A, NM_000049.2:c.863A>G	Canavan disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ASPA gene located on chromosomal region 17p13.3. The age of onset is neonatal/infantile. This disease is characterized by a variable spectrum between severe forms with leukodystrophy, macrocephaly and severe developmental delay, and a very rare mild/juvenile form characterized by mild developmental delay. The prevalence is 1:6,400- 1:13,500 in Askenazis Jewis.	250,6

ASPM	Microcephaly primary, type 5, autosomal recessive	NM_018136.4	NM_018136.4:c.1002delA, NM_018136.4:c.3055C>T, NM_018136.4:c.2389C>T, NM_018136.4:c.2967G>A, NM_018136.4:c.1260_1266delTCAAGTC, NM_018136.4:c.10059C>A, NM_018136.4:c.1154_1155delAG, NM_018136.4:c.1179delT, NM_018136.4:c.1729_1730delAG, NM_018136.4:c.1959_1962delCAAA, NM_018136.4:c.1990C>T, NM_018136.4:c.3979C>T, NM_018136.4:c.4195dupA, NM_018136.4:c.4583delA, NM_018136.4:c.4795C>T, NM_018136.4:c.4858_4859delAT, NM_018136.4:c.5136C>A, NM_018136.4:c.5149delA, NM_018136.4:c.1366G>T, NM_018136.4:c.1406_1413delATCCTAAA, NM_018136.4:c.1590delA, NM_018136.4:c.6189T>G, NM_018136.4:c.6232C>T, NM_018136.4:c.6337_6338delAT, NM_018136.4:c.6732delA, NM_018136.4:c.719_720delCT, NM_018136.4:c.7491_7495delTATTA, NM_018136.4:c.7565T>G, NM_018136.4:c.7761T>G, NM_018136.4:c.7782_7783delGA, NM_018136.4:c.7860_7861delGA, NM_018136.4:c.7894C>T, NM_018136.4:c.8131_8132delAA, NM_018136.4:c.8230_8231insA, NM_018136.4:c.8378delT, NM_018136.4:c.8508_8509delGA, NM_018136.4:c.8668C>T, NM_018136.4:c.8844delC, NM_018136.4:c.9115_9118dupCATT, NM_018136.4:c.9159delA, NM_018136.4:c.9178C>T, NM_018136.4:c.3082G>A, NM_018136.4:c.3188T>G, NM_018136.4:c.3477_3481delCGCTA, NM_018136.4:c.349C>T, NM_018136.4:c.3527C>G, NM_018136.4:c.3663delG, NM_018136.4:c.3710C>G, NM_018136.4:c.3796G>T, NM_018136.4:c.3811C>T, NM_018136.4:c.3978G>A, NM_018136.4:c.9747_9748delCT, NM_018136.4:c.9754delA, NM_018136.4:c.9789T>A, NM_018136.4:c.8711_8712delAA, NM_018136.4:c.9190C>T, NM_018136.4:c.9238A>T, NM_018136.4:c.9319C>T, NM_018136.4:c.5439_5440delAG, NM_018136.4:c.577C>T, NM_018136.4:c.6073delG, NM_018136.4:c.9677dupG, NM_018136.4:c.9685delA, NM_018136.4:c.9697C>T, NM_018136.4:c.9730C>T, NM_018136.4:c.9557C>G, NM_018136.4:c.9492T>G, NM_018136.4:c.9539A>C	Primary autosomal recessive microcephaly type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ASPM gene located on chromosomal region 1q31. The age of onset is neonatal/infantile. This disease is characterized by a reduction in head circumference at birth, mild to moderate non-progressive intellectual impairment and delay in early motor milestones, speech delay and hyperactive behavior are common.Â The annual incidence is 1:1,000,000.	250,6
ASS1	Citrullinemia type 1	NM_000050.4	NM_000050.4:c.421-2A>G, NM_000050.4:c.40G>A, NM_000050.4:c.1088G>A, NM_000050.4:c.470G>A, NM_000050.4:c.1085G>T, NM_000050.4:c.1087C>T, NM_000050.4:c.257G>A, NM_000050.4:c.323G>T, NM_000050.4:c.349G>A, NM_000050.4:c.380G>A, NM_000050.4:c.836G>A, NM_000050.4:c.910C>T, NM_000050.4:c.928A>C, NM_000050.4:c.496-2A>G, NM_000050.4:c.535T>C, NM_000050.4:c.539G>A, NM_000050.4:c.53C>T, NM_000050.4:c.571G>A, NM_000050.4:c.787G>A, NM_000050.4:c.793C>T, NM_000050.4:c.794G>A, NM_000050.4:c.805G>A, NM_000050.4:c.835C>T, NM_000050.4:c.919C>T, NM_000050.4:c.970G>A, NM_000050.4:c.814C>T, NM_000050.4:c.970+5G>A, NM_000050.4:c.1168G>A, NM_000050.4:c.1194-1G>C, NM_000050.4:c.256C>T	Citrullinemia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ASS1 gene located on chromosomal region 9q34.1. The age of onset is variable. This disease is characterized by hyperammonemia, progressive lethargy, poor feeding and vomiting in the neonatal form and by variable hyperammonemia in the later-onset form. The prevalence is 1:100,000-9:100,000.	250,6
ATIC	AICA-ribosiduria	NM_004044.6	NM_004044.6:c.223+1G>A, NM_004044.6:c.1277A>G, NM_004044.6:c.625delG	AICA-ribosiduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ATIC gene located on chromosomal region 2q35. The age of onset is neonatal/infantile. This disease is characterized by profound intellectual deficit, epilepsy, dysmorphic features of the knees, elbows, and shoulders and congenital blindness. The prevalence is <1:1,000,000.	250,6
ATP7A	Menkes disease	NM_000052.6	NM_000052.6:c.2938C>T, NM_000052.6:c.2531G>A, NM_000052.6:c.1639C>T, NM_000052.6:c.1974_1977dupGTTT, NM_000052.6:c.3257_3258delAC, NM_000052.6:c.3294+2T>G, NM_000052.6:c.3915_3921delCTCCCCA, NM_000052.6:c.3931A>G	Menkes disease follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ATP7A gene located on chromosomal region Xq21.1. The age of onset is neonatal/infantile. This disease is characterized by progressive neurodegeneration and marked connective tissue anomalies as well as typical sparse abnormal steely hair. The birth incidence is 1:300,000 in Europe, 1:360,000 in Japan and 1:50,000-1:100,000 in Australia, and The prevalence is 1:100,000 newborns.	600
ATP7A	Occipital horn syndrome	NM_000052.6	NM_000052.6:c.3911A>G	Occipital horn syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ATP7A gene located on chromosomal region Xq21.1. The age of onset is variable. This disease is characterized by progressive neurodegeneration and connective tissue disorders due to a copper transport defect. The prevalence is 1:100,000 newborns.	600
ATP7A	Spinal muscular atrophy, distal, X-linked	NM_000052.6	NM_000052.6:c.2981C>T	Spinal muscular atrophy, distal follows an X-linked pattern of inheritance and is caused by pathogenic variants in the ATP7A gene located on chromosomal region Xq21.1. The age of onset is infantile. This disease is characterized by distal weakness, atrophy of the muscles of the lower limbs, particularly in the tibioperoneal compartment and pes cavus.	600

ATP7B	Wilson disease	NM_000053.3	NM_000053.3:c.2532delA, NM_000053.3:c.2356-2A>G, NM_000053.3:c.1285+5G>T, NM_000053.3:c.2305A>G, NM_000053.3:c.1145_1151delCCCAACT, NM_000053.3:c.1934T>G, NM_000053.3:c.2071G>A, NM_000053.3:c.2297C>G, NM_000053.3:c.2972C>T, NM_000053.3:c.2975C>T, NM_000053.3:c.3083delA, NM_000053.3:c.2605G>A, NM_000053.3:c.2621C>T, NM_000053.3:c.2755C>G, NM_000053.3:c.2755C>T, NM_000053.3:c.2762G>A, NM_000053.3:c.2795C>A, NM_000053.3:c.2804C>T, NM_000053.3:c.2807T>A, NM_000053.3:c.2906G>A, NM_000053.3:c.2930C>T, NM_000053.3:c.4301C>T, NM_000053.3:c.915T>A, NM_000053.3:c.98T>C, NM_000053.3:c.1745_1746delTA, NM_000053.3:c.2123T>C, NM_000053.3:c.2267C>T, NM_000053.3:c.4088C>T, NM_000053.3:c.4135C>T, NM_000053.3:c.1512_1513insT, NM_000053.3:c.19_20delCA, NM_000053.3:c.1922T>C, NM_000053.3:c.3955C>T, NM_000053.3:c.3990_3993delTTAT, NM_000053.3:c.4058G>A, NM_000053.3:c.3207C>A, NM_000053.3:c.3359T>A, NM_000053.3:c.3688A>G, NM_000053.3:c.3101A>G, NM_000053.3:c.3796G>A, NM_000053.3:c.3809A>G, NM_000053.3:c.562C>T, NM_000053.3:c.3694A>C, NM_000053.3:c.1846C>T	Wilson disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ATP7B gene located on chromosomal region 13q14.3. The age of onset is infantile. This disease is characterized by the toxic accumulation of copper, mainly in the liver and central nervous system, and symptomatic patients may present with hepatic, neurologic or psychiatric forms. The birth incidence is 1:30,000-1:100,000 in France and The prevalence is 1:10,000-1:30,000.	250,6
ATR	Seckel syndrome type 1	NM_001184.3	NM_001184.3:c.2341+1G>A, NM_001184.3:c.5645delA, NM_001184.3:c.6037_6038insA, NM_001184.3:c.6488delT, NM_001184.3:c.975_976delCT, NM_001184.3:c.5635G>T	Seckel syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ATR gene located on chromosomal region 3q23. The age of onset is neonatal/infantile. This disease is characterized by a proportionate dwarfism of prenatal onset, a severe microcephaly with a bird-headed like appearance and mental retardation.Â The prevalence is <1:1,000,000.	250,6
AUH	3-Methylglutaconic aciduria type 1	NM_001698.2	NM_001698.2:c.471delT, NM_001698.2:c.559G>A, NM_001698.2:c.589C>T, NM_001698.2:c.650G>A, NM_001698.2:c.895-1G>A, NM_001698.2:c.991A>T, NM_001698.2:c.656-2A>G, NM_001698.2:c.943-2A>G	3-Methylglutaconic aciduria type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AUH gene located on chromosomal region 9q22.31. The age of onset is neonatal/infantile. This disease is characterized by a variable clinical phenotype ranging from mildly delayed speech to psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia.Â The prevalence is <1:1,000,000.	600
B4GALT1	Congenital disorders of glycosylation type 2d	NM_001497.3	NM_001497.3:c.1031dupC	Congenital disorder of glycosylation type 2d follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the B4GALT1 gene located on chromosomal region 9p13. The age of onset is neonatal/infantile. This disease is characterized by macrocephaly, hydrocephaly, hypotonia, myopathy and coagulation anomalies. The prevalence is <1:1,000,000.	600
B9D2	Meckel syndrome type 10	NM_030578.3	NM_030578.3:c.301A>C	Meckel syndrome type 10 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the B9D2 gene located on chromosomal region 19q13.2. The age of onset is neonatal/infantile. This disease is characterized by a combination of renal cysts developmental anomalies of the central nervous system (usually occipital encephalocele), hepatic ductal dysplasia and polydactyly.Â The prevalence is <1:1,000,000.	600
BCKDHA	Maple syrup urine disease type 1A	NM_000709.3	NM_000709.3:c.1037G>A, NM_000709.3:c.1036C>T, NM_000709.3:c.1234G>A, NM_000709.3:c.14delT, NM_000709.3:c.761C>A, NM_000709.3:c.929C>G, NM_000709.3:c.964C>T, NM_000709.3:c.979G>A, NM_000709.3:c.905A>C, NM_000709.3:c.632C>T, NM_000709.3:c.659C>T, NM_000709.3:c.740_741insT, NM_000709.3:c.868G>A, NM_000709.3:c.909_910delGT, NM_000709.3:c.917delT, NM_000709.3:c.853G>C, NM_000709.3:c.796delA	Maple syrup urine disease type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BCKDHA gene located on chromosomal region 19q13.1-13.2. The age of onset is neonatal/infantile. This disease is characterized by poor feeding, lethargy, vomiting, a maple syrup odor in the cerumen and urine, encephalopathy and central respiratory failure if untreated. The prevalence is 1:1,000,000-9:1,000,000.	250,6
BCKDHB	Maple syrup urine disease type 1B	NM_183050.2	NM_183050.2:c.1046G>A, NM_183050.2:c.547C>T, NM_183050.2:c.509G>A, NM_183050.2:c.526A>T, NM_183050.2:c.344-1G>A, NM_183050.2:c.1114G>T, NM_183050.2:c.302G>A, NM_183050.2:c.342T>G, NM_183050.2:c.508C>A, NM_183050.2:c.508C>G, NM_183050.2:c.508C>T, NM_183050.2:c.748G>T, NM_183050.2:c.752T>C, NM_183050.2:c.799C>T, NM_183050.2:c.548G>C, NM_183050.2:c.884delT, NM_183050.2:c.902T>G, NM_183050.2:c.952-1G>A, NM_183050.2:c.853C>T, NM_183050.2:c.832G>A, NM_183050.2:c.356T>G, NM_183050.2:c.970C>T, NM_183050.2:c.488A>T, NM_183050.2:c.479T>G	Maple syrup urine disease type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BCKDHB gene located on chromosomal region 6q14.1. The age of onset is neonatal/infantile. This disease is characterized by poor feeding, lethargy, vomiting, a maple syrup odor in the cerumen and urine, encephalopathy and central respiratory failure if untreated. The prevalence is 1:10,000-5:10,000.	600
BCS1L	BjÃ¶rnstad syndrome	NM_004328.4	NM_004328.4:c.548G>A	BjÃ¶rnstadt syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BCS1L gene located on chromosomal region 2q33. The age of onset is neonatal/infantile. This disease is characterized by congenital sensorineural hearing loss and pili torti. The prevalence is <1:1,000,000.	250,6

BCS1L	GRACILE syndrome	NM_004328.4	NM_004328.4:c.232A>G	GRACILE syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BCS1L gene located on chromosomal region 2q33. The age of onset is neonatal/infantile. This disease is characterized by fetal growth restriction (GR), aminoaciduria (A), cholestasis (C), iron overload (I), lactacidosis (L) and early death (E). The birth incidence is 1:50,000 in Finland and the prevalence is <1:1,000,000.	250,6
BCS1L	Mitochondrial comlpex III deficiency, nuclear type 1	NM_004328.4	NM_004328.4:c.1057G>A, NM_004328.4:c.830G>A, NM_004328.4:c.133C>T, NM_004328.4:c.103G>C, NM_004328.4:c.696delT, NM_004328.4:c.148A>G, NM_004328.4:c.166C>T, NM_004328.4:c.550C>T, NM_004328.4:c.547C>T	Mitochondrial comlpex III deficiency, nuclear type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BCS1L gene located on chromosomal region 2q33. The age of onset is neonatal and it is characterized by lactic acidosis, hypotonia, hypoglycemia, failure to thrive, encephalopathy, and delayed psychomotor development.	250,6
BEST1	Bestrophinopathy	NM_004183.3	NM_004183.3:c.934G>A, NM_004183.3:c.598C>T, NM_004183.3:c.752G>A, NM_004183.3:c.949G>A, NM_004183.3:c.521_522delTG	Bestrophinopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BEST1 gene located on chromosomal region 11q13. The age of onset is variable. This disease is characterized by central visual loss in the first 2 decades of life associated with an absent electrooculogram light rise, and a reduced electroretinogram.	250,6
BEST1	Retinitis pigmentosa type 50	NM_004183.3	NM_004183.3:c.1383_1384insGCCTTGATGGA, NM_004183.3:c.1444delG, NM_004183.3:c.1491_1497dupCAAAGAC, NM_004183.3:c.1566_1576dupCTTGATGGAGC, NM_004183.3:c.341_342delTG, NM_004183.3:c.1308_1309insACCAAAG, NM_004183.3:c.1264delG, NM_004183.3:c.418C>G, NM_004183.3:c.614T>C, NM_004183.3:c.682G>A, NM_004183.3:c.344delG, NM_004183.3:c.524delG	Retinitis pigmentosa refers to a heterogeneous group of inherited ocular diseases that result in a progressive retinal degeneration. Type 50 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BEST1 gene located on chromosomal region 11q12.3. The age of onset is adult. This disease is characterized by night blindness, the development of tunnel vision, and slowly progressive decreased central vision. The global prevalence of all types of retinitis pigmentosa is 1/3,000 to 1/5,000.	250,6
BEST1	Vitelliform macular dystrophy type 2	NM_004183.3	NM_004183.3:c.122T>C, NM_004183.3:c.422G>A	Vitelliform macular dystrophy type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BEST1 gene located on chromosomal region 11q12.3. The age of onset is infancy or adolescence. This disease is characterized by normal vision at birth, then progresses through distinct stages that include an asymptomatic previtelliform phase (stage 1) followed by the formation of a yellow, egg yolk-like (vitelliform) lesion in the macula (stage 2). The contents become less homogenous and develop a "scrambled-egg" appearance (stage 2a). The lesion eventually develops a fluid, yellow-colored vitelline substance (pseudohypopyon or stage 3) and finally breaks down, leaving a scar that causes central visual acuity deterioration (20/200). This may be complicated by a subfoveal choroidal neovascular (CNV) membrane (rare in children). Anomalous color discrimination (mainly the protan axis) and metamorphopsia may be observed but patients retain normal peripheral vision and dark adaptation. Some affected individuals remain asymptomatic. The prevalence is 1/5,000 to 1/67,000.	250,6
BRCA2	Fanconi anemia, complementation group D1	NM_000059.3	NM_000059.3:c.1514T>C, NM_000059.3:c.4648G>T, NM_000059.3:c.8415A>T, NM_000059.3:c.7544C>T, NM_000059.3:c.7994A>G, NM_000059.3:c.5574_5577delAATT, NM_000059.3:c.4889C>G, NM_000059.3:c.4936_4939delGAAA, NM_000059.3:c.5066_5067insA, NM_000059.3:c.6024dupG, NM_000059.3:c.6860delG, NM_000059.3:c.7235C>A, NM_000059.3:c.9382C>T, NM_000059.3:c.9900dupA, NM_000059.3:c.3847_3848delGT, NM_000059.3:c.5718_5719delCT, NM_000059.3:c.5837_5838delCAinsAG, NM_000059.3:c.6023_6024insG, NM_000059.3:c.8503T>C, NM_000059.3:c.6486_6489delACAA, NM_000059.3:c.657_658delTG, NM_000059.3:c.6997_6998insT	Fanconi anemia, complementation group D1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BRCA2 gene located on chromosomal region 13q12.3. The age of onset is infantile. This disease is characterized by physical abnormalities, bone marrow failure, and increased risk of malignancy. The prevalence is 1:1,000,000-9:1,000,000.	250,6
BRIP1	Fanconi anemia, complementation group J	NM_032043.2	NM_032043.2:c.2990_2993delCAAA, NM_032043.2:c.1045G>C, NM_032043.2:c.2237_2240delTCAA, NM_032043.2:c.3209C>A, NM_032043.2:c.502C>T, NM_032043.2:c.139C>G, NM_032043.2:c.1702_1703delAA, NM_032043.2:c.2392C>T	Fanconi anemia, complementation group J follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BRIP1 gene located on chromosomal region 17q22.2. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1:1,000,000- 9:1,000,000.	250,6
BSCL2	Berardinelli-Seip congenital lipodystrophy	NM_032667.6	NM_032667.6:c.634G>C, NM_032667.6:c.412C>T, NM_032667.6:c.782_783insG, NM_032667.6:c.823C>T, NM_032667.6:c.985C>T, NM_032667.6:c.672-3C>G, NM_032667.6:c.974_975insG, NM_032667.6:c.671+5G>A	Berardinelli-Seip congenital lipodystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BSCL2 gene located on chromosomal region 11q13. The age of onset is neonatal/infantile. This disease is characterized the association of lipoatrophy, hypertriglyceridemia, hepatomegaly and acromegaloid features. The prevalence is 1.27:100,000.	600

BSCL2	Severe neurodegenerative syndrome with lipodystrophy	NM_032667.6	NM_032667.6:c.793C>T	Severe neurodegenerative syndrome with lipodystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BSCL2 gene located on chromosomal region 11q13. The age of onset is neonatal/infantile. This disease is characterized by developmental regression of motor and cognitive skills in the first years of life, often leading to death in the first decade, hyperactive behavior, seizures, tremor and ataxic gait. Patients may show a mild or typical lipodystrophic appearance. The prevalence is 1.27:100,000.	600
BSND	Bartter syndrome type 4A	NM_057176.2	NM_057176.2:c.1A>T, NM_057176.2:c.22C>T, NM_057176.2:c.3G>A, NM_057176.2:c.10G>T, NM_057176.2:c.23G>T, NM_057176.2:c.35T>C, NM_057176.2:c.23G>A, NM_057176.2:c.139G>A	Bartter syndrome type 4A with deafness follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BSND gene located on chromosomal region 1p32.3. The age of onset is neonatal/infantile. This disease is characterized by maternal polyhydramnios, premature delivery, polyuria, sensorineural deafness and is associated with hypokalemic alkalosis, increased levels of plasma renin and aldosterone, low blood pressure, and vascular resistance to angiotensin II.	250,6
BTD	Biotinidase deficiency	NM_000060.3	NM_000060.3:c.1531C>G, NM_000060.3:c.1508_1512delGGATG, NM_000060.3:c.1339C>T, NM_000060.3:c.1352G>A, NM_000060.3:c.1489C>T, NM_000060.3:c.643C>T, NM_000060.3:c.664G>A, NM_000060.3:c.755A>G, NM_000060.3:c.1368A>C, NM_000060.3:c.933delT, NM_000060.3:c.1595C>T, NM_000060.3:c.1612C>T, NM_000060.3:c.757C>T, NM_000060.3:c.1106C>T, NM_000060.3:c.1321delG, NM_000060.3:c.794A>T, NM_000060.3:c.595G>A, NM_000060.3:c.629A>G, NM_000060.3:c.631C>T, NM_000060.3:c.235C>T, NM_000060.3:c.334G>C, NM_000060.3:c.511G>A, NM_000060.3:c.184G>A, NM_000060.3:c.557G>A, NM_000060.3:c.583A>G, NM_000060.3:c.968A>G, NM_000060.3:c.528G>T, NM_000060.3:c.443G>A	Biotinidase deficiency an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the BTD gene located on chromosomal region 3p25. The age of onset is neonatal/infantile. This disease is characterized by seizures, breathing difficulties, hypotonia, skin rash, alopecia, hearing loss and delayed development.	250,6
BTK	Agammaglobulinemia, X-linked	NM_000061.2	NM_000061.2:c.1275C>A, NM_000061.2:c.1506C>A, NM_000061.2:c.1125T>G, NM_000061.2:c.1223T>C, NM_000061.2:c.1288A>G, NM_000061.2:c.1082A>G, NM_000061.2:c.763C>T, NM_000061.2:c.1516T>C, NM_000061.2:c.718G>T, NM_000061.2:c.755G>A, NM_000061.2:c.1766A>G, NM_000061.2:c.1773C>A, NM_000061.2:c.1558C>T, NM_000061.2:c.1559G>A, NM_000061.2:c.1889T>A, NM_000061.2:c.1906G>T, NM_000061.2:c.338T>A, NM_000061.2:c.1820C>A, NM_000061.2:c.862C>T, NM_000061.2:c.919A>G, NM_000061.2:c.1838G>A, NM_000061.2:c.1001A>C	X-linked agammaglobulinemia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the BTK gene located on chromosomal region Xq21.33-q22. The age of onset is infantile. This disease is characterized by recurrent bacterial infections during infancy. The prevalence is 3:1,000,000-6:1,000,000 men.	600
C10orf2	Infantile onset spinocerebellar ataxia	NM_021830.4	NM_021830.4:c.1523A>G	La ataxia espinocerebelosa infantil sigue un patrÃ³n de herencia autosÃ³mico recesivo y estÃ¡ causada por variantes patogÃ©nicas en el gen C10orf2 localizado en la regiÃ³n cromosÃ³mica 10q24. La edad de apariciÃ³n es neonatal/infantil con sÃ-ntomas como inicio temprano ataxia, atetosis y reducciÃ³n de los reflejos tendinosos.	600
C10orf2	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form	NM_021830.4	NM_021830.4:c.1287C>T, NM_021830.4:c.952G>A, NM_021830.4:c.1370C>T, NM_021830.4:c.524_525insG	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the C10orf2 gene located on chromosomal region 10q24. The age of onset is neonatal/infantile. It is a severe disease associated with mitochondrial dysfunction. Some patients are affected by progressive atrophy of the cerebellum, brain stem, the spinal cord, and sensory axonal neuropathy. Clinical features include hypotonia, athetosis, ataxia, ophthalmoplegia, sensorineural hearing deficit, sensory axonal neuropathy, epileptic encephalopathy and female hypogonadism. In some individuals liver dysfunction and multi-organ failure is present.	600
C10orf2	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis	NM_021830.4	NM_021830.4:c.955A>G	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the C10orf2 gene located on chromosomal region 10q24. This syndrome is characterised by adult-onset severe sensory ataxic neuropathy, dysarthria and chronic progressive external ophthalmoplegia. The prevalence is unknown. Other common features include progressive gait unsteadiness, absent deep tendon reflexes, the presence of Romberg's sign, a decreased sense of vibration and proprioception and detection of red ragged fibres on muscle biopsy.	600

C3	Atypical hemolytic-uremic syndrome with C3 anomaly	NM_000064.2	NM_000064.2:c.2562C>G	Atypical hemolytic-uremic syndrome with C3 anomaly follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the C3 gene located on chromosomal region 19p13.3-p13.2. An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. The prevalence is <1:1.000.000.	600
C3	C3 deficiency	NM_000064.2	NM_000064.2:c.2354+1G>A, NM_000064.2:c.4851-1G>A, NM_000064.2:c.1119+1G>T, NM_000064.2:c.3116dupT, NM_000064.2:c.3627_3628insGGGGCCC, NM_000064.2:c.1004-2A>T	C3 deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the C3 gene located on chromosomal region 19p13.3-p13.2. The age of onset is infantile. It is a rare defect of the complement classical pathway. Patients develop recurrent, severe, pyogenic infections because of ineffective opsonization of pathogens. Some patients may also develop autoimmune disorders, such as arthralgia and vasculitic rashes, lupus-like syndrome and membranoproliferative glomerulonephritis. The prevalence is <1:1.000.000.	600
CA2	Osteopetrosis, autosomal recessive, type 3	NM_000067.2	NM_000067.2:c.663+2T>C, NM_000067.2:c.319C>T, NM_000067.2:c.120T>G	Osteopetrosis, autosomal recessive, type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CA2 gene located on chromosomal region 8q22. The age of onset is neonatal/infantile. This disease is characterized by osteopetrosis, renal tubular acidosis, and neurological disorders related to cerebral calcifications. The prevalence is <1:1.000.000.	600
CAPN3	Limb-girdle muscular dystrophy type 2A	NM_000070.2	NM_000070.2:c.1838delA, NM_000070.2:c.2120A>G, NM_000070.2:c.1795_1796insA, NM_000070.2:c.1469G>A, NM_000070.2:c.1599_1602delGAGC, NM_000070.2:c.1715G>A, NM_000070.2:c.1743_1745+1delTGAG, NM_000070.2:c.257C>T, NM_000070.2:c.328C>T, NM_000070.2:c.549delA, NM_000070.2:c.2212C>T, NM_000070.2:c.223dupT, NM_000070.2:c.2243G>A, NM_000070.2:c.2251_2254dupGTCA, NM_000070.2:c.2257G>A, NM_000070.2:c.2306G>A, NM_000070.2:c.2361_2363delAGinsTCATCT, NM_000070.2:c.2361_2364delAGinsTCATCT, NM_000070.2:c.2362_2363delAGinsTCATCT, NM_000070.2:c.246G>A, NM_000070.2:c.676G>A, NM_000070.2:c.551C>T, NM_000070.2:c.580delT, NM_000070.2:c.133G>A, NM_000070.2:c.550delA, NM_000070.2:c.1468C>T, NM_000070.2:c.956C>T, NM_000070.2:c.1322delG, NM_000070.2:c.1466G>A, NM_000070.2:c.662G>T, NM_000070.2:c.855_864dupGTTGATTGCA, NM_000070.2:c.1610A>G, NM_000070.2:c.598_612delTTCTGGAGTGCTCTG	Limb-girdle muscular dystrophy type 2A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CAPN3 gene located on chromosomal region 15q15.1. The age of onset is variable. This disease is characterized by symmetrical and selective atrophy and weakness of proximal limb and girdle muscles. The prevalence is 1:100,000- 9:100,000.	250,6
CBS	Homocystinuria	NM_000071.2	NM_000071.2:c.1150A>G, NM_000071.2:c.1058C>T, NM_000071.2:c.1136G>A, NM_000071.2:c.341C>T, NM_000071.2:c.1006C>T, NM_000071.2:c.325T>C, NM_000071.2:c.1316G>A, NM_000071.2:c.374G>A, NM_000071.2:c.1265C>T, NM_000071.2:c.1280C>T, NM_000071.2:c.146C>T, NM_000071.2:c.1471C>T, NM_000071.2:c.1616T>C, NM_000071.2:c.162G>A, NM_000071.2:c.833T>C, NM_000071.2:c.904G>A, NM_000071.2:c.919G>A, NM_000071.2:c.393G>C, NM_000071.2:c.415G>A, NM_000071.2:c.430G>A, NM_000071.2:c.434C>T, NM_000071.2:c.502G>A, NM_000071.2:c.526G>T, NM_000071.2:c.572C>T, NM_000071.2:c.676G>A, NM_000071.2:c.689delT, NM_000071.2:c.797G>A, NM_000071.2:c.959T>C, NM_000071.2:c.969G>A, NM_000071.2:c.992C>A, NM_000071.2:c.1330G>A, NM_000071.2:c.1379C>T, NM_000071.2:c.1397C>T, NM_000071.2:c.304A>C	Homocystinuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CBS gene located on chromosomal region 21q22.3. The age of onset is infantile. This disease is characterized by the multiple involvement of the eye, skeleton, central nervous system and vascular system. The prevalence is 1:200,000-1:335,000.	250,6
CC2D2A	Joubert syndrome type 9	NM_001080522.2	NM_001080522.2:c.4179delG, NM_001080522.2:c.3594+1G>A, NM_001080522.2:c.3289delG, NM_001080522.2:c.4582C>T, NM_001080522.2:c.4667A>T, NM_001080522.2:c.2848C>T, NM_001080522.2:c.3364C>T, NM_001080522.2:c.4333C>T, NM_001080522.2:c.4181delG	Joubert syndrome type 9 defect follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CC2D2A gene located on chromosomal region 4p15.32. The age of onset is neonatal/infantile. This disease is characterized neonatal hypotonia, developmental delay, intellectrual disability, ataxia, and abnormal eye movements including oculomotor apraxia, primary position nystagmus and congenital hepatic fibrosis.	250,6
CC2D2A	Meckel syndrome type 6	NM_001080522.2	NM_001080522.2:c.3145C>T, NM_001080522.2:c.2486+1G>C	Meckel syndrome type 6 with hepatic defect follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CC2D2A gene located on chromosomal region 4p15.32. The age of onset is neonatal/infantile. This disease is characterized by a combination of renal cysts, developmental anomalies of the central nervous system (usually occipital encephalocele), hepatic ductal dysplasia and polydactyly.	250,6

CD2AP	Focal segmental glomerulosclerosis type 3	NM_012120.2	NM_012120.2:c.730-1delGinsCT, NM_012120.2:c.1575_1577delAGA, NM_012120.2:c.1488G>A	Focal segmental glomerulosclerosis type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CD2AP gene located on chromosomal region 6p12. The age of onset is variable. A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation.	600
CD40LG	Hyper IgM syndrome, X-linked	NM_000074.2	NM_000074.2:c.386A>G, NM_000074.2:c.368C>A, NM_000074.2:c.384T>A, NM_000074.2:c.632C>A, NM_000074.2:c.107T>G	X-linked hyper-IgM syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the CD40LG gene located on chromosomal region Xq26. The age of onset is neonatal/infantile. This disease is characterized by lower-respiratory tract bacterial infections, opportunistic infections, recurrent or protracted diarrhea associated with failure to thrive, neutropenia, thrombocytopenia and anemia. The prevalence is 2:1,000,000 male newborns.	600
CDH23	Deafness type 12, autosomal recessive	NM_022124.5	NM_022124.5:c.6442G>A, NM_022124.5:c.5663T>C, NM_022124.5:c.9565C>T, NM_022124.5:c.7823G>A, NM_022124.5:c.902G>A	Non-syndromic autosomal recessive deafness type 12 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CDH23 gene located on chromosomal region 10p22.1. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.	250,6
CDH23	Usher syndrome type 1D	NM_022124.5	NM_022124.5:c.288+1G>A, NM_022124.5:c.193delC, NM_022124.5:c.6050-9G>A, NM_022124.5:c.3141C>A, NM_022124.5:c.146-2A>G, NM_022124.5:c.4504C>T, NM_022124.5:c.3516_3519delATCC, NM_022124.5:c.3579+2T>C, NM_022124.5:c.3293A>G, NM_022124.5:c.9319+1_9319+4delGTAA, NM_022124.5:c.5237G>A, NM_022124.5:c.1858+2T>G, NM_022124.5:c.6392delC, NM_022124.5:c.7660G>A	Usher syndrome type 1D follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CDH23 gene located on chromosomal region 10p22.1. The age of onset is neonatal/infantile. This disease is characterized by sensorineural deafness, retinitis pigmentosa and progressive vision loss.	250,6
CDH3	Ectodermal dysplasia - ectrodactyly - macular dystrophy	NM_001793.4	NM_001793.4:c.455_456insC, NM_001793.4:c.981delG, NM_001793.4:c.1508G>A, NM_001793.4:c.965A>T, NM_001793.4:c.830delG, NM_001793.4:c.965A>G	Ectodermal dysplasia - ectrodactyly - macular dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CDH3 gene located on chromosomal region 16q22.1. The age of onset is neonatal/infantile. This disease is characterized by the association of ectodermal dysplasia, ectrodactyly, and macular dystrophy. The prevalence is <1:1,000,000.	600
CDHR1	Retinitis pigmentosa type 65	NM_033100.3	NM_033100.3:c.1485+2T>C, NM_033100.3:c.1463delG, NM_033100.3:c.1110delC, NM_033100.3:c.338delG, NM_033100.3:c.524dupA, NM_033100.3:c.1485+2T>G, NM_033100.3:c.1112delC, NM_033100.3:c.640delG	Retinitis pigmentosa refers to a heterogeneous group of inherited ocular diseases that result in a progressive retinal degeneration. Type 65 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CDHR1 gene located on chromosomal region 10q23.1. The age of onset is adult. This disease is characterized by night blindness, the development of tunnel vision, and slowly progressive decreased central vision. The global prevalence of all types of retinitis pigmentosa is 1/3,000 to 1/5,000.	250,6
CDK5RAP2	Microcephaly, primary, type 3, autosomal recessive	NM_018249.5	NM_018249.5:c.4661_4662insTATT, NM_018249.5:c.246T>A, NM_018249.5:c.4546G>T, NM_018249.5:c.127+1G>C, NM_018249.5:c.4672C>T, NM_018249.5:c.524_528delAGGCA, NM_018249.5:c.700G>T	Primary autosomal recessive microcephaly type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CDK5RAP2 gene located on chromosomal region 9q33.2. The age of onset is neonatal/infantile. This disease is characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment.	600
CENPJ	Microcephaly primary, type 6, autosomal recessive	NM_018451.4	NM_018451.4:c.3243_3246delTCAG, NM_018451.4:c.2614delT, NM_018451.4:c.3415G>T, NM_018451.4:c.3653C>T, NM_018451.4:c.2462C>T, NM_018451.4:c.3699_3702dupAATA, NM_018451.4:c.3568_3571dupGTCA, NM_018451.4:c.3843_3844insTA, NM_018451.4:c.757_760delGTCT, NM_018451.4:c.1952_1953insAGTG, NM_018451.4:c.3704A>T, NM_018451.4:c.232_236delCAGAA, NM_018451.4:c.2460_2463delGACG, NM_018451.4:c.2968_2972delAAAAA, NM_018451.4:c.40C>T, NM_018451.4:c.289dupA	Primary autosomal recessive microcephaly type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CENPJ gene located on chromosomal region 13q12.12.Â The age of onset is neonatal/infantile. This disease is characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment.	250,6
CEP152	Microcephaly, primary, type 9, autosomal recessive	NM_014985.3	NM_014985.3:c.794A>C, NM_014985.3:c.749_750delGA	Primary autosomal recessive microcephaly type 9 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP152 gene located on chromosomal region 15q21.1. The age of onset is neonatal/infantile. This disease is characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment.	600
CEP152	Seckel syndrome type 5	NM_014985.3	NM_014985.3:c.2034T>G, NM_014985.3:c.1578-1G>A	Seckel syndrome type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP152 gene located on chromosomal region 15q21.1. The age of onset is neonatal/infantile. This disease is characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation.	600

CEP290	Joubert syndrome, Senior-Loken type	NM_025114.3	NM_025114.3:c.5611_5614delCAAA, NM_025114.3:c.164_167delCTCA	Joubert syndrome, Senior-Loken type syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP290 gene located on chromosomal region 12q21.32. The age of onset is infantile. This disease is characterized by symptoms of nephronophthisis such as polyuria, polydipsia, secondary eneuresis and anemia. The progression of the disease can lead to acute or chronic renal insufficiency and finally to end-stage kidney disease. Ocular features include congenital or early-onset severe visual loss, due to retinal dystrophy. In rare occasions, other additional clinical signs may be observed like liver fibrosis, obesity and neurologic disorders.. The prevalence is <1:1,000,000.	250,6
CEP290	Joubert syndrome type 5	NM_025114.3	NM_025114.3:c.4656delA, NM_025114.3:c.21G>T, NM_025114.3:c.5668G>T	Joubert syndrome with oculorenal defect 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP290 gene located on chromosomal region 12q21.32. The age of onset is infantile. This disease is characterized by neonatal hypotonia, developmental delay, intellectrual disability, ataxia, abnormal eye movements including oculomotor apraxia, primary position nystagmus and renal and ocular disease.	250,6
CEP290	Leber congenital amaurosis type 10	NM_025114.3	NM_025114.3:c.7341_7342insA, NM_025114.3:c.4705-1G>T, NM_025114.3:c.4723A>T, NM_025114.3:c.4962_4963delAA, NM_025114.3:c.4916C>A, NM_025114.3:c.6624delG, NM_025114.3:c.6645+1G>A, NM_025114.3:c.7324G>T, NM_025114.3:c.6798G>A, NM_025114.3:c.7394_7395delAG, NM_025114.3:c.1681C>T, NM_025114.3:c.7341delA, NM_025114.3:c.6448_6455delCAGTTGAA, NM_025114.3:c.1665_1666delAA, NM_025114.3:c.384_387delTAGA, NM_025114.3:c.2249T>G, NM_025114.3:c.3185delT, NM_025114.3:c.4393C>T, NM_025114.3:c.1501G>T	Leber congenital amaurosis type 10 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP290 gene located on chromosomal region 12q21.32. The age of onset is infancy/neonatal. This disease is characterized by retinal dystrophy defined by blindness, nystagmus, roving eye movement and lack of detectable signals on an electroretinogram, leading to severe visual impairment within the first year of life.	250,6
CEP290	Meckel syndrome type 4	NM_025114.3	NM_025114.3:c.613C>T	Meckel syndrome type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CEP290 gene located on chromosomal region 12q21.32. The age of onset is neonatal. This disease is characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically occipital encephalocele), hepatic ductal dysplasia and cysts, and postaxial polydactyly. The prevalence is <1 / 1,000,000.	250,6
CERKL	Retinitis pigmentosa tipo 26	NM_201548.4	NM_201548.4:c.1012C>T, NM_201548.4:c.1090C>T, NM_201548.4:c.312delA, NM_201548.4:c.715C>T, NM_201548.4:c.769C>T, NM_201548.4:c.780delT, NM_201548.4:c.847C>T, NM_201548.4:c.1553_1569dupTTATCAGTCTTTATGGA	Retinitis pigmentosa 26 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CERKL gene located on chromosomal region 2q31.3. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000-5:10,000.	250,6
CFH	Complement factor H deficiency	NM_000186.3	NM_000186.3:c.3628C>T, NM_000186.3:c.2876G>A, NM_000186.3:c.380G>T, NM_000186.3:c.481G>T, NM_000186.3:c.1606T>C	Immunodeficiency with factor H anomaly follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CFH gene located on chromosomal region 1q32. This disease is characterized by recurrent bacterial infections and renal failure.	250,6

CFTR	Cystic fibrosis	NM_000492.3	NM_000492.3:c.1327_1330dupGATA, NM_000492.3:c.1210-7_1210-6delTT, NM_000492.3:c.125C>T, NM_000492.3:c.1301_1307delCACTTCT, NM_000492.3:c.1397C>A, NM_000492.3:c.1340delA, NM_000492.3:c.1364C>A, NM_000492.3:c.1393-1G>A, NM_000492.3:c.1438G>T, NM_000492.3:c.1466C>A, NM_000492.3:c.1475C>T, NM_000492.3:c.1477C>T, NM_000492.3:c.1516A>G, NM_000492.3:c.1519_1521delATC, NM_000492.3:c.1521_1523delCTT, NM_000492.3:c.1545_1546delTA, NM_000492.3:c.1624G>T, NM_000492.3:c.1692delA, NM_000492.3:c.1706A>G, NM_000492.3:c.1721C>A, NM_000492.3:c.178G>T, NM_000492.3:c.1970delG, NM_000492.3:c.200C>T, NM_000492.3:c.2012delT, NM_000492.3:c.2051_2052delAAinsG, NM_000492.3:c.2052_2053insA, NM_000492.3:c.2052delA, NM_000492.3:c.1000C>T, NM_000492.3:c.1007T>A, NM_000492.3:c.1013C>T, NM_000492.3:c.1021T>C, NM_000492.3:c.1022_1023insTC, NM_000492.3:c.1040G>A, NM_000492.3:c.1040G>C, NM_000492.3:c.1055G>A, NM_000492.3:c.1081delT, NM_000492.3:c.115C>T, NM_000492.3:c.2538G>A, NM_000492.3:c.254G>A, NM_000492.3:c.2551C>T, NM_000492.3:c.2583delT, NM_000492.3:c.262_263delTT, NM_000492.3:c.2657+5G>A, NM_000492.3:c.2668C>T, NM_000492.3:c.273+1G>A, NM_000492.3:c.2737_2738insG, NM_000492.3:c.2739T>A, NM_000492.3:c.274-1G>A, NM_000492.3:c.274G>A, NM_000492.3:c.274G>T, NM_000492.3:c.2780T>C, NM_000492.3:c.2834C>T, NM_000492.3:c.2855T>C, NM_000492.3:c.2869_2870insG, NM_000492.3:c.2875delG, NM_000492.3:c.2908G>C, NM_000492.3:c.292C>T, NM_000492.3:c.2939T>A, NM_000492.3:c.2989-1G>A, NM_000492.3:c.3067_3072delATAGTG, NM_000492.3:c.3140-26A>G, NM_000492.3:c.3194T>C, NM_000492.3:c.3196C>T, NM_000492.3:c.3197G>A, NM_000492.3:c.3230T>C, NM_000492.3:c.325_327delTATinsG, NM_000492.3:c.3266G>A, NM_000492.3:c.3276C>A, NM_000492.3:c.3276C>G, NM_000492.3:c.328G>C, NM_000492.3:c.328G>T, NM_000492.3:c.3302T>A, NM_000492.3:c.3310G>T, NM_000492.3:c.349C>T, NM_000492.3:c.350G>T, NM_000492.3:c.3528delC, NM_000492.3:c.3533_3536delCAAC, NM_000492.3:c.3587C>G, NM_000492.3:c.358G>A, NM_000492.3:c.3611G>A, NM_000492.3:c.3612G>A, NM_000492.3:c.3659delC, NM_000492.3:c.366T>A, NM_000492.3:c.3731G>A, NM_000492.3:c.3744delA, NM_000492.3:c.3752G>A, NM_000492.3:c.3761T>G, NM_000492.3:c.3764C>A	Cystic fibrosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CFTR gene located on chromosomal region 7q31.2. The age of onset of severe form is neonatal or infantile but there are also variants associated with moderate clinical or late onset. This disease is characterized by the production of sweat with a high salt content, mucus secretions with an abnormal viscosity, chronic bronchitis, pancreatic insufficiency, adolescent diabetes and, more rarely, stercoral obstruction and cirrhosis. Male sterility is a constant feature. Late-onset forms, which are usually only mild or monosymptomatic. The prevalence is 1:10,000-9:10,000.	250,6
CHST6	Macular corneal dystrophy	NM_021615.4	NM_021615.4:c.820G>T, NM_021615.4:c.853delC, NM_021615.4:c.993G>T, NM_021615.4:c.327_328delCT, NM_021615.4:c.392C>A	Macular corneal dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CHST6 gene located on chromosomal region 16q22. The age of onset is variable. This disease is characterized by bilateral ill-defined cloudy regions within a hazy stroma, and eventually severe visual impairment. The prevalence is 1:100,000-9:100,000.	250,6
CLCN1	Myotonia congenita, autosomal recessive	NM_000083.2	NM_000083.2:c.1453A>G, NM_000083.2:c.409T>G, NM_000083.2:c.568G>A, NM_000083.2:c.899G>A, NM_000083.2:c.1169G>A, NM_000083.2:c.1238T>G, NM_000083.2:c.871G>A, NM_000083.2:c.180+3A>T, NM_000083.2:c.225dupC, NM_000083.2:c.501C>G, NM_000083.2:c.2680C>T	Myotonia congenita (Becker disease) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLCN1 gene located on chromosomal region 7q35. The age of onset is neonatal/infantile. This disease is characterized by slow muscle relaxation, that it is relieved with exercise, associated with hyperexcitation of the muscle fibres. The prevalence is 1:100,000.	250,6
CLCN7	Osteopetrosis type 4, autosomal recessive	NM_001287.5	NM_001287.5:c.622C>T, NM_001287.5:c.781A>T	Osteopetrosis type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLCN7 gene located on chromosomal region 16p13. The age of onset is neonatal/infantile. This disease is characterized bone marrow failure, fractures and visual impairment. The incidence is 1:200,000 live births and the prevalence is 1:100,000.	600
CLDN14	Deafness type 29, autosomal recessive	NM_144492.2	NM_144492.2:c.254T>A, NM_144492.2:c.301G>A, NM_144492.2:c.398delT	Deafness type 29, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLDN14 gene located on chromosomal region 21q22.3. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.	600
CLDN19	Hypomagnesemia type 5, renal failure with severe ocular abnormalities	NM_148960.2	NM_148960.2:c.269T>C, NM_148960.2:c.425_437delCCCTGGTGACCCA, NM_148960.2:c.59G>A, NM_148960.2:c.169C>G, NM_148960.2:c.599G>A	Hypomagnesemia type 5, renal failure with severe ocular abnormalities follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLDN19 gene located on chromosomal region 1p34.2. The age of onset is infantile. This disease is characterized by excessive magnesium and calcium renal wasting, bilateral nephrocalcinosis, progressive renal failure and severe ocular abnormalities. The prevalence is <1:1,000,000.	250,6

CLN3	Ceroid-lipofuscinoses neuronal type 3	NM_001042432.1	NM_001042432.1:c.883G>A, NM_001042432.1:c.597C>A, NM_001042432.1:c.622_623insT, NM_001042432.1:c.1272delG, NM_001042432.1:c.1210C>A	Juvenile neuronal ceroid lipofuscinosis 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLN3 gene located on chromosomal region 16p12.1. The age of onset is infantile. This disease is characterized by onset at early school age with vision loss due to retinopathy, seizures and the decline of mental and motor capacities. The annual birth incidence is 1:217,000-1:450,000 in Sweden and 1:143,000 in Germany, and the prevalence is 1.5:1,000,000-9:1,000,000.	600
CLN5	Neuronal ceroid lipofuscinosis type 5	NM_006493.2	NM_006493.2:c.619T>C, NM_006493.2:c.335G>A, NM_006493.2:c.377G>A, NM_006493.2:c.620G>C, NM_006493.2:c.669dupC, NM_006493.2:c.335G>C, NM_006493.2:c.565C>T, NM_006493.2:c.575A>G, NM_006493.2:c.593T>C, NM_006493.2:c.595C>T, NM_006493.2:c.613C>T, NM_006493.2:c.919delA, NM_006493.2:c.924_925delAT, NM_006493.2:c.955_970delGGAAATGAAACATCTG, NM_006493.2:c.835G>A, NM_006493.2:c.526dupA, NM_006493.2:c.1026C>A, NM_006493.2:c.524T>G, NM_006493.2:c.433C>T	Neuronal ceroid lipofuscinosis type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLN5 gene located on chromosomal region 3q21.1-q32. The age of onset is infantile. This disease is characterized by onset during infancy or early childhood with decline of mental and motor capacities, epilepsy and vision loss through retinal degeneration. The prevalence is <1:1,000,000.	600
CLN6	Ceroid lipofuscinosis, neuronal, type 6	NM_017882.2	NM_017882.2:c.200T>C, NM_017882.2:c.214G>C, NM_017882.2:c.139C>T, NM_017882.2:c.307C>T, NM_017882.2:c.214G>T, NM_017882.2:c.663C>G	Late infantile neuronal ceroid lipofuscinosis 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLN6 gene located on chromosomal region 15q23. The age of onset is infantile. This disease is characterized by onset during infancy or early childhood with decline of mental and motor capacities, epilepsy and vision loss through retinal degeneration. The prevalence is <1:1,000,000.	600
CLN8	Ceroid lipofuscinosis, neuronal, type 8	NM_018941.3	NM_018941.3:c.88delG, NM_018941.3:c.789G>C, NM_018941.3:c.610C>T, NM_018941.3:c.88G>C	Late infantile neuronal ceroid lipofuscinosis 8 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLN8 gene located on chromosomal region 8p23.3. The age of onset is infantile. This disease is characterized by onset during infancy or early childhood with decline of mental and motor capacities, epilepsy, and vision loss through retinal degeneration. The prevalence is <1:1,000,000.	600
CLRN1	Retinitis pigmentosa type 61	NM_174878.2	NM_174878.2:c.92C>T	Retinitis pigmentosa refers to a heterogeneous group of inherited ocular diseases that result in a progressive retinal degeneration. Type 61 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLRN1gene located on chromosomal region 3q25.1. The age of onset is adult. This disease is characterized by night blindness, the development of tunnel vision, and slowly progressive decreased central vision. The global prevalence of all types of retinitis pigmentosa is 1/3,000 to 1/5,000.	250,6
CLRN1	Usher syndrome type 3A	NM_174878.2	NM_174878.2:c.591_592insT, NM_174878.2:c.630_631insT, NM_174878.2:c.118T>G, NM_174878.2:c.433+1061A>T, NM_174878.2:c.189C>A, NM_174878.2:c.144T>G	Usher syndrome type 3A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CLRN1 gene located on chromosomal region 3q25.1. The age of onset is neonatal/infantile. This disease is characterized by the association of sensorineural deafness with retinitis pigmentosa and progressive vision loss. The prevalence is 1:1.000.000- 9/1.000.000.	250,6
CNGA1	Retinitis pigmentosa type 49	NM_000087.3	NM_000087.3:c.1747C>T, NM_000087.3:c.1540C>T, NM_000087.3:c.2071T>C, NM_000087.3:c.1927C>T, NM_000087.3:c.1271G>A, NM_000087.3:c.1001G>A, NM_000087.3:c.959C>T, NM_000087.3:c.97_98insA, NM_000087.3:c.449+2T>C, NM_000087.3:c.1972delA, NM_000087.3:c.238G>T, NM_000087.3:c.794G>A, NM_000087.3:c.238G>A	Retinitis pigmentosa 49 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CNGA1 gene located on chromosomal region 4p12. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000-5:10,000.	250,6
CNGB1	Retinitis pigmentosa tipo 45	NM_001297.4	NM_001297.4:c.3150delG, NM_001297.4:c.2762_2765delACGA, NM_001297.4:c.2957A>T, NM_001297.4:c.413-1G>A, NM_001297.4:c.218-2A>G, NM_001297.4:c.2492+2T>G, NM_001297.4:c.3462+1G>A, NM_001297.4:c.2653delG, NM_001297.4:c.3425delT, NM_001297.4:c.1122-2A>T, NM_001297.4:c.1958-1G>A, NM_001297.4:c.952C>T	Retinitis pigmentosa 45 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CNGB1 gene located on chromosomal region 16q13. The age of onset is variable. This disease is characterized by night blindness, peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000 to 5:10,000.	250,6
CNGB3	Achromatopsia type 3	NM_019098.4	NM_019098.4:c.2011G>T, NM_019098.4:c.1063C>T, NM_019098.4:c.1208G>A, NM_019098.4:c.1672G>T, NM_019098.4:c.819_826delCAGACTCC, NM_019098.4:c.1148delC, NM_019098.4:c.886_890delACTTC, NM_019098.4:c.2048_2049delCA, NM_019098.4:c.446_447insT, NM_019098.4:c.893_897delCAAAA, NM_019098.4:c.887_896delCTTCTACAAA	Achromatopsia type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CNGB3 gene located on chromosomal region 8q21.3. The age of onset is neonatal/Infantile. This disease is characterized by reduced visual acuity, pendular nystagmus, increased sensitivity to light (photophobia), a small central scotoma, and reduced or complete loss of color discrimination. Most individuals have complete form, with total lack of function in all three types of cones. Rarely, individuals have incomplete form, with similar, but generally less severe symptoms. The prevalence is 1/30,000-1/50,000.	250,6
CNGB3	Macular degeneration, juvenile	NM_019098.4	NM_019098.4:c.1405T>G	Juvenile macular degeneration follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CNGB3 gene located on chromosomal region 8q21.3. The age of onset is infancy or adolescence. This disease is characterized by blurred or distorted central vision with dark areas. Normally, side vision is not affected, but the perception of color can vary during the later stages of the disease.	250,6

COL11A1	Stickler syndrome type 2	NM_001854.3	NM_001854.3:c.1750dupG, NM_001854.3:c.2350G>C, NM_001854.3:c.4606C>G, NM_001854.3:c.4642C>G, NM_001854.3:c.3709-1G>A	Stickler syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL11A1 gene located on chromosomal region 1p21. The age of onset is neonatal/infantile. Autosomal recessive Stickler syndrome is a rare type of Stickler syndrome manifesting with opthalmological (myopia, retinal detachment and cataracts), orofacial (micrognathia, midface hypoplasia and cleft palate) auditory (sensorineural hearing loss) and articular (epiphyseal dysplasia) symptoms.	600
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	NM_000494.3	NM_000494.3:c.1898G>A, NM_000494.3:c.3827_3828insC, NM_000494.3:c.2228-3_2235delCAGGTCCTGCTinsTTG, NM_000494.3:c.1706delC, NM_000494.3:c.2336-2A>G, NM_000494.3:c.3897_3900delATCT, NM_000494.3:c.3908G>A, NM_000494.3:c.2336-1G>T, NM_000494.3:c.2965delA, NM_000494.3:c.3043C>T, NM_000494.3:c.3067C>T, NM_000494.3:c.3277+1G>A, NM_000494.3:c.3676C>T, NM_000494.3:c.4319_4320insC, NM_000494.3:c.433C>T, NM_000494.3:c.520_521delAG, NM_000494.3:c.4003_4004delGG, NM_000494.3:c.2551+1G>T, NM_000494.3:c.3800delC, NM_000494.3:c.2564T>G, NM_000494.3:c.2430_2431insCCGA, NM_000494.3:c.2383C>T, NM_000494.3:c.2944_2947+1delGAAGG	Epidermolysis bullosa, junctional, non-Herlitz type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL17A1 gene located on chromosomal region 10q24.3. The age of onset is neonatal/infantile. This disease is characterized by a generalized skin blistering, atrophic scarring, nail dystrophy or nail absence, and enamel hypoplasia, with extracutaneous involvement.	250,6
COL18A1	Knobloch syndrome type 1	NM_030582.3	NM_030582.3:c.3367_3379delCCCCCAGGCCCAC, NM_030582.3:c.3493_3501delGGCCCCCCA, NM_030582.3:c.2797C>T, NM_030582.3:c.995_996insGACGTGAAAGAGGGG, NM_030582.3:c.3502_3511delGGCCCCCCAG, NM_030582.3:c.3618_3618+1delGG, NM_030582.3:c.994_995insGGACGTGAAAGAGGG, NM_030582.3:c.3517_3518delCC, NM_030582.3:c.1535_1536insGACGTGAAAGAGGGG, NM_030582.3:c.2589_2590delAG, NM_030582.3:c.4054_4055delCT, NM_030582.3:c.4463_4464insG	Knobloch syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL18A1 gene located on chromosomal region 21q22.3. The age of onset is neonatal/infantile. This disease is characterized by vitreoretinal and macular degeneration, and occipital encephalocele.Â The prevalence is <1:1,000,000.	250,6
COL1A2	Ehlers-Danlos syndrome, cardiac valvular type	NM_000089.3	NM_000089.3:c.3601G>T, NM_000089.3:c.1404+1G>A, NM_000089.3:c.559G>C, NM_000089.3:c.133-1G>A, NM_000089.3:c.1404+1G>C, NM_000089.3:c.240_247delGTATGATG, NM_000089.3:c.293_294insC	Ehlers-Danlos syndrome, cardiac valvular type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL1A2 gene located on chromosomal region 7q22.1. The age of onset is neonatal/infantile. This disease is characterized by joint hypermobility, skin hyperextensibility and cardiac valvular defects.Â The prevalence is 6/100,000 to 7/100,000.	600
COL2A1	Otospondylomegaepiphyseal dysplasia	NM_001844.4	NM_001844.4:c.1052delG, NM_001844.4:c.3106C>T	Otospondylomegaepiphyseal dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL2A1 gene located on chromosomal region 12q13.11. The age of onset is neonatal/infantile. This disease is characterized by sensorineural hearing loss, enlarged epiphyses, skeletal dysplasia with disproportionately short limbs, vertebral body anomalies and a characteristic facies. The prevalence is 1:7,500-1:9,000.	600
COL4A3	Alport syndrome, autosomal recessive	NM_000091.4	NM_000091.4:c.345delG, NM_000091.4:c.346C>A, NM_000091.4:c.898G>A, NM_000091.4:c.4421T>C, NM_000091.4:c.2110delC, NM_000091.4:c.343delG, NM_000091.4:c.4420_4424delCTTTT, NM_000091.4:c.5002_*6delAAAAGACACTGAAGCTAA, NM_000091.4:c.2083G>A, NM_000091.4:c.2954G>T, NM_000091.4:c.4484A>G, NM_000091.4:c.4571C>G, NM_000091.4:c.4441C>T	Alport syndrome, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL4A3 and COL4A4 genes located on chromosomal region 2q36.3. The age of onset is infantile. This disease is characterized by renal, cochlear, and ocular involvement. Renal disease progresses from microscopic hematuria to proteinuria, progressive renal insufficiency, and end-stage renal disease. Progressive sensorineural hearing loss is usually present by late childhood or early adolescence. Ocular findings include anterior lenticonus, maculopathy, corneal endothelial vesicles, and recurrent corneal erosion. The prevalence is 1:50,000 newborn.	250,6
COL4A4	Alport syndrome, autosomal recessive	NM_000092.4	NM_000092.4:c.3713C>A, NM_000092.4:c.4129C>T, NM_000092.4:c.4923C>A, NM_000092.4:c.3601G>A, NM_000092.4:c.2312delG, NM_000092.4:c.71+1G>A	Alport syndrome, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL4A3 and COL4A4 genes located on chromosomal region 2q36.3. The age of onset is infantile. This disease is characterized by renal, cochlear, and ocular involvement. Renal disease progresses from microscopic hematuria to proteinuria, progressive renal insufficiency, and end-stage renal disease. Progressive sensorineural hearing loss is usually present by late childhood or early adolescence. Ocular findings include anterior lenticonus, maculopathy, corneal endothelial vesicles, and recurrent corneal erosion. The prevalence is 1:50,000 newborn.	250,6

COL7A1	Epidermolysis bullosa dystrophica, Hallopeau-Siemens type	NM_000094.3	NM_000094.3:c.4039G>C, NM_000094.3:c.425A>G, NM_000094.3:c.336C>G, NM_000094.3:c.3809C>T, NM_000094.3:c.4119+1G>T, NM_000094.3:c.6205C>T, NM_000094.3:c.6527_6528insC, NM_000094.3:c.6573+1G>T, NM_000094.3:c.6187C>T, NM_000094.3:c.6752G>A, NM_000094.3:c.6859G>A, NM_000094.3:c.6946G>A, NM_000094.3:c.6670G>T, NM_000094.3:c.1907G>T, NM_000094.3:c.2471_2472insG, NM_000094.3:c.7440+4delC, NM_000094.3:c.7912G>T, NM_000094.3:c.7930-1G>C, NM_000094.3:c.7957G>A, NM_000094.3:c.8245G>A, NM_000094.3:c.8371C>T, NM_000094.3:c.8393T>A, NM_000094.3:c.8440C>T, NM_000094.3:c.8479C>T, NM_000094.3:c.8524_8527+10delGAAGGTGAGGACAG, NM_000094.3:c.887delG, NM_000094.3:c.933C>A, NM_000094.3:c.238G>T, NM_000094.3:c.3831+1G>T, NM_000094.3:c.4373C>T, NM_000094.3:c.6091G>A, NM_000094.3:c.4888C>T, NM_000094.3:c.5052+1G>A, NM_000094.3:c.5096C>T, NM_000094.3:c.4783G>C, NM_000094.3:c.5443G>C, NM_000094.3:c.5532+1G>A, NM_000094.3:c.5821-1G>A, NM_000094.3:c.5287C>T, NM_000094.3:c.706C>T, NM_000094.3:c.7345-1G>A, NM_000094.3:c.592G>A, NM_000094.3:c.7411C>T	Epidermolysis bullosa dystrophica, Hallopeau-Siemens type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL7A1 gene located on chromosomal region 3p21.1. The age of onset is neonatal/infantile. This disease is characterized by generalized cutaneous and mucosal blistering and scarring associated with severe deformities and major extracutaneous involvement. The prevalence is <1:1,000,000.	250,6
COL9A1	Stickler syndrome type 4	NM_001851.4	NM_001851.4:c.883C>T, NM_001851.4:c.706C>T	Stickler syndrome type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL9A1 gene located on chromosomal region 6q13. The age of onset is infantile. This disease is characterized by opthalmological (myopia, retinal detachment and cataracts), orofacial (micrognathia, midface hypoplasia and cleft palate) auditory (sensorineural hearing loss) and articular (epiphyseal dysplasia) symptoms. The prevalence is <1:1,000,000.	600
COL9A2	Stickler syndrome type 5	NM_001852.3	NM_001852.3:c.1918C>T, NM_001852.3:c.1097_1098insC, NM_001852.3:c.793-1G>C	Stickler syndrome type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL9A2 gene located on chromosomal region 1p33-p32. The age of onset is infantile. This disease is characterized by opthalmological (myopia, retinal detachment and cataracts), orofacial (micrognathia, midface hypoplasia and cleft palate) auditory (sensorineural hearing loss) and articular (epiphyseal dysplasia) symptoms. The prevalence is <1:1,000,000.	600
COQ2	Primary coenzyme Q10 deficiency type 1	NM_015697.7	NM_015697.7:c.683A>G, NM_015697.7:c.1197delT, NM_015697.7:c.590G>A, NM_015697.7:c.723delT, NM_015697.7:c.890A>G	Coenzyme Q10 deficiency, primary follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COQ2 gene located on chromosomal region 4q21.23. The age of onset is neonatal/infantile. The phenotypes include an encephalomyopathic form with seizures and ataxia; a multisystem infantile form with encephalopathy, cardiomyopathy and renal failure; a predominantly cerebellar form with ataxia and cerebellar atrophy; Leigh syndrome with growth retardation; and an isolated myopathic form.	250,6
CPS1	Carbamoylphosphate synthetase type 1 deficiency	NM_001875.4	NM_001875.4:c.1912C>T, NM_001875.4:c.697C>T, NM_001875.4:c.1631C>T, NM_001875.4:c.3556delA	Carbamoylphosphate synthetase deficiency type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CPS1 gene located on chromosomal region 2q35. The age of onset is infantile. This disease is characterized by congenital hyperammonemia and defective citrulline synthesis. The prevalence is 1:800,000 newborn in Japan.	600
CPT1A	Carnitine palmitoyltransferase type 1A deficiency	NM_001876.3	NM_001876.3:c.1216C>T, NM_001876.3:c.1241C>T, NM_001876.3:c.1361A>G, NM_001876.3:c.222C>A, NM_001876.3:c.1079A>G, NM_001876.3:c.1436C>T, NM_001876.3:c.1493A>G, NM_001876.3:c.335_336delCC, NM_001876.3:c.1393G>T, NM_001876.3:c.281+1G>A, NM_001876.3:c.1538C>T, NM_001876.3:c.298C>T	Carnitine palmitoyl transferase 1A deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CPT1A gene located on chromosomal region 11q13.2. The age of onset is neonatal/infantile. This disease is characterized by recurrent attacks of fasting-induced hypoketotic hypoglycemia and risk of liver failure. The prevalence is 1.3:1,000 newborn.	600
CPT2	Carnitine palmitoyltransferase deficiency, type 2	NM_000098.2	NM_000098.2:c.1239_1240delGA, NM_000098.2:c.1369A>T, NM_000098.2:c.1237C>T, NM_000098.2:c.680C>T, NM_000098.2:c.1437C>G, NM_000098.2:c.149C>A, NM_000098.2:c.1784delC, NM_000098.2:c.886C>T, NM_000098.2:c.1763C>G, NM_000098.2:c.359A>G, NM_000098.2:c.370C>T, NM_000098.2:c.1883A>C, NM_000098.2:c.1891C>T, NM_000098.2:c.1148T>A, NM_000098.2:c.638A>G, NM_000098.2:c.725_726delAC, NM_000098.2:c.452G>A, NM_000098.2:c.338C>T, NM_000098.2:c.481C>T, NM_000098.2:c.464dupT, NM_000098.2:c.520G>A	Carnitine palmitoyl transferase type 2 deficiency, infantile form follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CPT2 gene located on chromosomal region 1p32. The age of onset is neonatal/infantile. This disease is characterized by a severe fasting intolerance leading to metabolic derangements of hypoketotic hypoglycemia, resulting in coma and seizures, and hepatic encephalopathy leading to liver failure.Â The prevalence is <1:1,000,000.	250,6

CRB1	Leber congenital amaurosis type 8	NM_201253.2	NM_201253.2:c.3299T>G, NM_201253.2:c.3383delT, NM_201253.2:c.3419T>A, NM_201253.2:c.3094G>A, NM_201253.2:c.936T>G, NM_201253.2:c.493_501delGATGGAATT, NM_201253.2:c.3997G>T, NM_201253.2:c.498_506delAATTGATGG, NM_201253.2:c.2688T>A, NM_201253.2:c.613_619delATAGGAA, NM_201253.2:c.2401A>T, NM_201253.2:c.610_616delGAAATAG	Leber congenital amaurosis follows an autosomal recessive pattern of inheritance. Type 8 is caused by pathogenic variants in the CRB1 gene located on chromosomal region 1q31-q32.1. The age of onset is neonatal/infancy. This disease comprises a group of early-onset childhood retinal dystrophies characterized by vision loss, nystagmus, and severe retinal dysfunction. Patients usually present at birth with profound vision loss and pendular nystagmus. Electroretinogram responses are usually nonrecordable. Other clinical findings may include high hypermetropia, photodysphoria, oculodigital sign, keratoconus, cataracts, and a variable appearance to the fundus.	250,6
CRB1	Pigmented paravenous chorioretinal atrophy	NM_201253.2	NM_201253.2:c.484G>A	Pigmented paravenous chorioretinal atrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CRB1 gene located on chromosomal region 19p12. The age of onset is variable. This disease is characterized by an unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins.	250,6
CRB1	Retinitis pigmentosa type 12	NM_201253.2	NM_201253.2:c.3053_3054insTTATA, NM_201253.2:c.3122T>C, NM_201253.2:c.2416G>T, NM_201253.2:c.2843G>A, NM_201253.2:c.3299T>C, NM_201253.2:c.2983G>T, NM_201253.2:c.2290C>T	Retinitis pigmentosa 12 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CRB1 gene located on chromosomal region 1q31-q32.1. The age of onset is variable. This disease is characterized by night blindness, peripheral visual field impairment and over time loss of central vision. The prevalence is 1:10,000-5:10,000.	250,6
CRLF1	Cold-induced sweating syndrome	NM_004750.4	NM_004750.4:c.538C>T, NM_004750.4:c.303delC, NM_004750.4:c.413C>T, NM_004750.4:c.527+5G>A, NM_004750.4:c.226T>G, NM_004750.4:c.829C>T, NM_004750.4:c.397+1G>A, NM_004750.4:c.708_709delCCinsT, NM_004750.4:c.713_714insC, NM_004750.4:c.1125delG, NM_004750.4:c.676dupA, NM_004750.4:c.856-1G>A, NM_004750.4:c.852G>T, NM_004750.4:c.935G>A, NM_004750.4:c.1137C>G, NM_004750.4:c.845_846delTG	Cold-induced sweating syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CRLF1 gene located on chromosomal region 19p12. The age of onset is infantile. This disease is characterized by profuse sweating (involving the chest, face, arms and trunk) induced by cold ambient temperature kyphoscoliosis, a high-arched palate, depressed nasal bridge and impaired peripheral sensitivity to pain and temperature. The prevalence is <1:1,000,000.	600
CRTAP	Osteogenesis imperfecta type 7	NM_006371.4	NM_006371.4:c.826C>T, NM_006371.4:c.180G>A, NM_006371.4:c.561T>G, NM_006371.4:c.634C>T	Osteogenesis imperfecta type VII follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CRTAP gene located on chromosomal region 3p22.3. The age of onset is variable. This disease is characterized by increased bone fragility, low bone mass, and susceptibility to bone fractures with variable severity. The prevalence is 6:100,000-7:100,000.	600
CRX	Leber congenital amaurosis type 7	NM_000554.4	NM_000554.4:c.425A>G, NM_000554.4:c.196G>A, NM_000554.4:c.898T>C	Leber congenital amaurosis type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CRX gene located on chromosomal region 19q13.3. The age of onset is neonatal/infantile. This disease is characterized by blindness, nystagmus, roving eye movement and lack of detectable signals on an electroretinogram, leading to severe visual impairment within the first year of life. The prevalence is 2:100,000-3:100,000 newborn.	250,6
CSTB	Progressive myoclonic epilepsy type 1A	NM_000100.3	NM_000100.3:c.212A>C, NM_000100.3:c.202C>T	Progressive myoclonic epilepsy type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CSTB gene located on chromosomal region 21q22.3. The age of onset is infantile. This disease is characterized by severe, stimulus-sensitive myoclonus and tonic-clonic seizures. The onset, occurring between 6 and 13 years of age, is characterized by convulsions. Myoclonus begins 1 to 5 years later. The twitchings occur predominantly in the proximal muscles of the extremities and are bilaterally symmetrical, although asynchronous. At first small, they become late in the clinical course so violent that the victim is thrown to the floor. Mental deterioration and eventually dementia develop. The prevalence is 1:20,000 newborn.	600
CTNS	Cystinosis, ocular nonnephropathic	NM_004937.2	NM_004937.2:c.589G>A, NM_004937.2:c.853-3C>G	Ocular non nephropathic cystinosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CTNS gene located on chromosomal region 17p13. The age of onset is neonatal/infantile. It presents typical ocular findings of nephropathic cystinosis. However, systemic manifestations are absent and kidney disease does not occur. The prevalence is 1:100,000-1:200,000.	250,6
CTNS	Nephropathic cystinosis	NM_004937.2	NM_004937.2:c.416C>T, NM_004937.2:c.414G>A, NM_004937.2:c.124G>A, NM_004937.2:c.357_360delCAGC, NM_004937.2:c.397_398delAT, NM_004937.2:c.1015G>A, NM_004937.2:c.646dupA, NM_004937.2:c.283G>T, NM_004937.2:c.329G>T, NM_004937.2:c.506G>A	Nephropathic cystinosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CTNS gene located on chromosomal region 17p13. The age of onset is neonatal/infantile. This disease is characterized by hypothyroidism, insulin-dependent diabetes, hepatosplenomegaly with portal hypertension, and muscle, cerebral and ocular involvement, caused by cystine deposits in various organs. The prevalence is 1:100,000-1:200,000.	250,6

CTSD	Ceroid lipofuscinosis, neuronal, type 10	NM_001909.4	NM_001909.4:c.685T>A, NM_001909.4:c.1149G>C	Neuronal ceroid lipofuscinosis type 10 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CTSD gene located on chromosomal region 11p15.5. The age of onset is adult. This disease is characterized by dementia, seizures and loss of motor capacities, and sometimes associated with visual loss caused by retinal degeneration. The prevalence is 2:100,000-4:100,000 newborn.	600
CTSK	Pycnodysostosis	NM_000396.3	NM_000396.3:c.236G>A, NM_000396.3:c.154A>T, NM_000396.3:c.436G>C, NM_000396.3:c.926T>C, NM_000396.3:c.721C>T	Pycnodysostosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CTSK gene located on chromosomal region 1q21. The age of onset is variable. This disease is characterized by osteosclerosis, short stature or dwarfism, acroosteolysis of the distal phalanges, fragile bones associated with spontaneous fractures and dysplasia of the clavicles. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	-	hybrid 5'CYP21A1P/3'CYP21A2, hybrid 5'CYP21A2/3'CYP21A1P (Detection by MLPA)	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP21A2 gene located on chromosomal region 6p21.3. The age of onset is neonatal/infantile. This disease is characterized by simple virilizing or salt wasting forms that can manifest with genital ambiguity in females, and in both sexes with adrenal insufficiency with dehydration during the neonatal period, life threatening hypoglycemia and hyperandrogenia. The prevalence is 1/100,000 to 9/100,000.	600
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	NM_000500.7	NM_000500.7:c.518T>A, NM_000500.7:c.955C>T, NM_000500.7:c.1069C>T, NM_000500.7:c.719T>A, NM_000500.7:c.[713T>A;719T>A], NM_000500.7:c.293-13A/C>G, NM_000500.7:c.332_339del, NM_000500.7:c.[710T>A;719T>A], NM_000500.7:c.923_924insT, NM_000500.7:c.[710T>A;713T>A], NM_000500.7:c.713T>A, NM_000500.7:c.[710T>A;713T>A;719T>A], NM_000500.7:c.710T>A (Detection by minisequencing)	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP21A2 gene located on chromosomal region 6p21.3. The age of onset is neonatal/infantile. This disease is characterized by simple virilizing or salt wasting forms that can manifest with genital ambiguity in females, and in both sexes with adrenal insufficiency with dehydration during the neonatal period, life threatening hypoglycemia and hyperandrogenia. The prevalence is 1/100,000 to 9/100,000.	600
CYP21A2	Non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	NM_000500.7	NM_000500.7:c.92C>T, NM_000500.7:c.844G>T, NM_000500.7:c.1360C>T (Detection by minisequencing)	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP21A2 gene located on chromosomal region 6p21.3. This is a common milder form of congenital adrenal hyperplasia characterized by a later onset of androgen excess symptoms seen in females and precocious pseudopuberty in both sexes. Cortisol and aldosterone levels are normal but there is an increased amount of androgens. Disease onset occurs in adolescence with variable degrees of postnatal androgen excess (precocious pubarche, hirsutism, acne, alopecia, anovulation and menstrual irregularies and in the post-pubertal period it can mimic polycystic ovary syndrome. It is also sometimes asymptomatic. The prevalence ranges from 1/1,000-1/500 in the general Caucasian population, but up to 1-2% among inbred populations, such as Eastern European (Ashkenazi) Jews.	600
CYP4V2	Bietti crystalline corneoretinal dystrophy	NM_207352.3	NM_207352.3:c.1523G>A, NM_207352.3:c.130T>A, NM_207352.3:c.327+1G>A, NM_207352.3:c.332T>C	Bietti crystalline corneoretinal dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP4V2 gene located on chromosomal region 4q35.2. The age of onset is adult. This disease is characterized by nightblindness, decreased vision, paracentral scotoma, and, in the end stages of the disease, legal blindness.	250,6
CYP7B1	Congenital bile acid synthesis defect type 3	NM_004820.3	NM_004820.3:c.1162C>T	Congenital bile acid synthesis defect type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP7B1 gene located on chromosomal region 8q21.3. The age of onset is neonatal/infantile. This disease is characterized by severe neonatal cholestatic liver disease. The prevalence is below 1,000,000.	250,6
CYP7B1	Spastic paraplegia type 5A, autosomal recessive	NM_004820.3	NM_004820.3:c.1460_1461insT, NM_004820.3:c.321_324delACAA, NM_004820.3:c.825T>A, NM_004820.3:c.889A>G, NM_004820.3:c.1456C>T, NM_004820.3:c.187C>T	Autosomal recessive spastic paraplegia type 5A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP7B1 gene located on chromosomal region 8q21.3. The age of onset is neonatal/infantile. This disease is characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. The prevalence is below 1,000,000.	250,6

D2HGDH	D-2-Hydroxyglutaric aciduria	NM_152783.4	NM_152783.4:c.1315A>G, NM_152783.4:c.1276G>A, NM_152783.4:c.440T>G, NM_152783.4:c.1333_1334delAC, NM_152783.4:c.1123G>T, NM_152783.4:c.1331T>C	D-2-Hydroxyglutaric aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the D2HGDH gene located on chromosomal region 2q37.3. The age of onset is variable. This disease is characterized by extremely variable clinical manifestations, with severe cases characterized by neonatal or early infantile-onset epileptic encephalopathy, and marked hypotonia, and cerebral visual failure, developmental delay, seizures, involuntary movements, and cardiomyopathy are also common in these cases. The prevalence is below 1,000,000.	250,6
DBT	Maple syrup urine disease type 2	NM_001918.3	NM_001918.3:c.670G>T, NM_001918.3:c.827T>G, NM_001918.3:c.294C>G, NM_001918.3:c.581C>G, NM_001918.3:c.772+1G>A, NM_001918.3:c.272_275delCAGT, NM_001918.3:c.1281+1G>A, NM_001918.3:c.871C>T, NM_001918.3:c.901C>T, NM_001918.3:c.939G>C, NM_001918.3:c.126T>G	Maple syrup urine disease type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DBT gene located on chromosomal region 1p31. The age of onset is neonatal/infantile. This disease is characterized by a maple syrup odor in the cerumen at birth, poor feeding, lethargy and focal dystonia, followed by progressive encephalopathy and central respiratory failure if untreated. The prevalence is 1/10,000 to 5/10,000.	250,6
DCLRE1C	Omenn syndrome	NM_001033855.2	NM_001033855.2:c.2T>C	Omenn syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAG1 and RAG2 genes located on chromosomal region 11p12. The age of onset is early. This disease is characterized by erythroderma, desquamation, alopecia, chronic diarrhea, failure to thrive, lymphadenopathy, and hepatosplenomegaly, associated with severe combined immunodeficiency.	250,6
DCLRE1C	Severe combined immunodeficiency due to DCLRE1C deficiency	NM_001033855.2	NM_001033855.2:c.1558_1559insA, NM_001033855.2:c.597C>A, NM_001033855.2:c.780+1delG, NM_001033855.2:c.1639G>T, NM_001033855.2:c.1903_1904insA, NM_001033855.2:c.457G>A, NM_001033855.2:c.1559_1560insA	Severe combined immunodeficiency due to DCLRE1C deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DCLRE1C gene located on chromosomal region 10p13. The age of onset is neonatal/infantile. This disease is characterized by severe and recurrent infections, diarrhea, failure to thrive, and cell sensitivity to ionizing radiation. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
DDB2	Xeroderma pigmentosum complementation group E	NM_000107.2	NM_000107.2:c.730A>G, NM_000107.2:c.937C>T, NM_000107.2:c.818G>A, NM_000107.2:c.919G>T	Xeroderma pigmentosum complementation group E follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DDB2 gene located on chromosomal region 11p12-p11. The age of onset is variable. This disease is characterized by mild xeroderma pigmentosum symptoms and no neurological abnormalities. The prevalence is 1/1,000,000.	600
DDC	Aromatic L-amino acid decarboxylase deficiency	NM_000790.3	NM_000790.3:c.100delG, NM_000790.3:c.1040G>A, NM_000790.3:c.823G>A, NM_000790.3:c.304G>A, NM_000790.3:c.272C>T, NM_000790.3:c.749C>T	Aromatic L-amino acid decarboxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DDC gene located on chromosomal region 7p12.2. The age of onset is neonatal/infantile. This disease is characterized by severe developmental delay, weak muscle tone (hypotonia), muscle stiffness, difficulty moving, and involuntary writhing movements of the limbs (athetosis). The prevalence is below 1,000,000.	600
DFNB31	Deafness type 31, autosomal recessive	NM_015404.3	NM_015404.3:c.1135C>T, NM_015404.3:c.817C>T	Deafness, autosomal recessive type 31 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DFNB31 gene located on chromosomal region 9q32. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.Â	250,6
DFNB59	Deafness type 59, autosomal recessive	NM_001042702.3	NM_001042702.3:c.122delA, NM_001042702.3:c.420delT, NM_001042702.3:c.113dupT, NM_001042702.3:c.988delG, NM_001042702.3:c.726delT, NM_001042702.3:c.161C>T, NM_001042702.3:c.817_818insT	Autosomal recessive nonsyndromic sensorineural deafness type DFNB59 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DFNB59 gene located on chromosomal region 2q31.2. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.	600
DGUOK	Mitochondrial DNA depletion syndrome type 3	NM_080916.2	NM_080916.2:c.137A>G, NM_080916.2:c.707+2T>G, NM_080916.2:c.763G>T, NM_080916.2:c.425G>A, NM_080916.2:c.313C>T, NM_080916.2:c.494A>T	Mitochondrial DNA depletion syndrome type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DGUOK gene located on chromosomal region 2p13. The age of onset is neonatal/infantile. This disease is characterized by progressive liver failure, hypoglycemia and neurologic abnormalities including hypotonia, encephalopathy and peripheral neuropathy	250,6

DHCR7	Smith-Lemli-Opitz syndrome	NM_001360.2	NM_001360.2:c.1055G>A, NM_001360.2:c.1210C>T, NM_001360.2:c.1054C>T, NM_001360.2:c.461C>G, NM_001360.2:c.151C>T, NM_001360.2:c.1031G>A, NM_001360.2:c.453G>A, NM_001360.2:c.506C>T, NM_001360.2:c.356A>T, NM_001360.2:c.1228G>A, NM_001360.2:c.1A>G, NM_001360.2:c.976G>T, NM_001360.2:c.964-1G>C, NM_001360.2:c.682C>T, NM_001360.2:c.452G>A, NM_001360.2:c.1337G>A, NM_001360.2:c.1342G>A, NM_001360.2:c.730G>A, NM_001360.2:c.292C>T, NM_001360.2:c.904T>C, NM_001360.2:c.907G>A, NM_001360.2:c.841G>A, NM_001360.2:c.744G>T, NM_001360.2:c.724C>T, NM_001360.2:c.725G>A, NM_001360.2:c.866C>T, NM_001360.2:c.278C>T, NM_001360.2:c.839A>G, NM_001360.2:c.832-1G>C	Smith-Lemli-Opitz syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DHCR7 gene located on chromosomal region 11q13.4. The age of onset is neonatal/infantile. This disease is characterized by multiple congenital anomalies, intellectual deficit, and behavioral problems.Â The prevalence is 1/20,000 to 1/40,000 newborn.	250,6
DHDDS	Retinitis pigmentosa type 59	NM_024887.3	NM_024887.3:c.328delA, NM_024887.3:c.998C>G, NM_024887.3:c.124A>G	Retinitis pigmentosa type 59 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DHDDS gene located on chromosomal region 1p36.11. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1/10,000 to 5/10,000.	600
DKC1	Dyskeratosis congenita, X-linked	NM_001363.4	NM_001363.4:c.91C>A, NM_001363.4:c.214_215delCTinsTA, NM_001363.4:c.194G>C, NM_001363.4:c.838A>C, NM_001363.4:c.91C>G, NM_001363.4:c.196A>G	Dyskeratosis congenital, X-linked follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DKC1 gene located on chromosomal region Xq28. The age of onset is infantile. This disease is characterized by the mucocutaneous triad of abnormal skin pigmentation, nail dystrophy and mucosal leucoplakia. The prevalence is 1-9/1,000,000.	600
DKC1	Hoyeraal-Hreidarsson syndrome	NM_001363.4	NM_001363.4:c.200C>T, NM_001363.4:c.204C>A	Hoyeraal-Hreidarsson syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DKC1 gene located on chromosomal region Xq28. The age of onset is neonatal/infantile. This disease is characterized by intrauterine growth retardation, microcephaly, cerebellar hypoplasia, progressive combined immune deficiency and aplastic anemia. The prevalence is below 1/1,000,000.	600
DLD	Dihydrolipoamide dehydrogenase deficiency E3	NM_000108.4	NM_000108.4:c.916_926delTGTGATGTACT, NM_000108.4:c.105_106insA, NM_000108.4:c.1483A>G	Dihydrolipoamide dehydrogenase deficiency E3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DLD gene located on chromosomal region 7q31-q32. The age of onset is neonatal/infantile. This disease is characterized by poor feeding, lethargy, vomiting and a maple syrup odor in the cerumen (and later in urine) noted soon after birth, followed by progressive encephalopathy and central respiratory failure if untreated. The prevalence is 1/1,000,000 to 9/1,000,000.	600
DLL3	Spondylocostal dysostosis type 1	NM_016941.3	NM_016941.3:c.231C>A, NM_016941.3:c.712C>T	Spondylocostal dysostosis type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DLL3 gene located on chromosomal region 19q13. The age of onset is neonatal/infantile. This disease is associated with vertebral and rib segmentation defects and characterised by a short neck with limited mobility, winged scapulae, a short trunk, and short stature with multiple vertebral anomalies at all levels of the spine. The prevalence is below 1/1,000,000.	600
DMD	Becker muscular dystrophy	NM_004006.2	NM_004006.2:c.3432+3A>G, NM_004006.2:c.3432+1G>A, NM_004006.2:c.137A>T	Becker muscular dystrophy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DMD gene located on chromosomal region Xp21.2. The age of onset is infantile. This disease is characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle. The incidence is 1/18,000 to 1/31,000 male newborns and the prevalence is 1/10,000 to 5/10,000.	600
DMD	Becker muscular dystrophy	-	insBecker, delBecker (Detection by MLPA)	Becker muscular dystrophy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DMD gene located on chromosomal region Xp21.2. The age of onset is infantile. This disease is characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle. The incidence is 1/18,000 to 1/31,000 male newborns and the prevalence is 1/10,000 to 5/10,000.	600
DMD	Dilated cardiomyopathy type 3B	NM_004006.2	NM_004006.2:c.5922+3G>C	Dilated cardiomyopathy type 3B follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DMD gene located on chromosomal region Xp21.2. The age of onset is variable. This disease is characterized by ventricular dilatation and impaired systolic function. Patients with DCM suffer from heart failure, arrhythmia, and are at risk of premature death. The incidence is 1/3,300 male newborns and the prevalence is 1/16,000 to 1/125,000.	600

DMD	Duchenne muscular dystrophy	NM_004006.2	NM_004006.2:c.1261C>T, NM_004006.2:c.1286C>A, NM_004006.2:c.1070delC, NM_004006.2:c.10086+1G>A, NM_004006.2:c.1886C>A, NM_004006.2:c.1900A>T, NM_004006.2:c.10774delA, NM_004006.2:c.204dupC, NM_004006.2:c.1900_1903dupAAGT, NM_004006.2:c.10141C>T, NM_004006.2:c.10033C>T, NM_004006.2:c.10453_10454delCT, NM_004006.2:c.1012G>T, NM_004006.2:c.1048G>T, NM_004006.2:c.2302C>T, NM_004006.2:c.1734delA, NM_004006.2:c.2380+1G>C, NM_004006.2:c.2380+2T>C, NM_004006.2:c.2479delG, NM_004006.2:c.2482T>G, NM_004006.2:c.2484T>G, NM_004006.2:c.251delT, NM_004006.2:c.2523delA, NM_004006.2:c.10446_10447delCT, NM_004006.2:c.2650C>T, NM_004006.2:c.10454delT, NM_004006.2:c.2294_2297delCCAT, NM_004006.2:c.2803+1G>A, NM_004006.2:c.2803+1G>T, NM_004006.2:c.2804-1G>A, NM_004006.2:c.2804-2A>T, NM_004006.2:c.2815_2816delTT, NM_004006.2:c.1306dupG, NM_004006.2:c.1332-9A>G, NM_004006.2:c.133C>T, NM_004006.2:c.1341_1342dupAG, NM_004006.2:c.2547delT, NM_004006.2:c.1371delG, NM_004006.2:c.2755A>T, NM_004006.2:c.2758C>T, NM_004006.2:c.1529_1530delTC, NM_004006.2:c.160_162delCTC, NM_004006.2:c.3295C>T, NM_004006.2:c.6182delC, NM_004006.2:c.6226G>T, NM_004006.2:c.3639dupA, NM_004006.2:c.199G>T, NM_004006.2:c.3747delG, NM_004006.2:c.2125delC, NM_004006.2:c.137_138dupAT, NM_004006.2:c.4117C>T, NM_004006.2:c.412_413delAA, NM_004006.2:c.2281_2285delGAAAA, NM_004006.2:c.4375C>T, NM_004006.2:c.4405C>T, NM_004006.2:c.2332C>T, NM_004006.2:c.4471_4472delAA, NM_004006.2:c.4486delG, NM_004006.2:c.4500delA, NM_004006.2:c.4518+5G>A, NM_004006.2:c.4735G>T, NM_004006.2:c.4806A>T, NM_004006.2:c.4843A>T, NM_004006.2:c.489G>A, NM_004006.2:c.5287C>T, NM_004006.2:c.530+1delG, NM_004006.2:c.5313dupT, NM_004006.2:c.5353C>T, NM_004006.2:c.5363C>G, NM_004006.2:c.5530C>T, NM_004006.2:c.5554C>T, NM_004006.2:c.5570_5571dupAA, NM_004006.2:c.5640T>A, NM_004006.2:c.5671A>T, NM_004006.2:c.5697delA, NM_004006.2:c.5773G>T, NM_004006.2:c.5807T>A, NM_004006.2:c.583C>T, NM_004006.2:c.8944C>T, NM_004006.2:c.1489C>T, NM_004006.2:c.6000T>A, NM_004006.2:c.6014_6017delCTCA, NM_004006.2:c.615T>A, NM_004006.2:c.9346C>T, NM_004006.2:c.9361+1G>A	Duchenne muscular dystrophy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DMD gene located on chromosomal region Xp21.2. The age of onset is infantile. This disease is characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle. The incidence is 1/3,300 male newborns and the prevalence is 1/16,000 to 1/125,000.	600
DMD	Duchenne muscular dystrophy	-	insDuchenne, delDuchenne (Detection by MLPA)	Duchenne muscular dystrophy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the DMD gene located on chromosomal region Xp21.2. The age of onset is infantile. This disease is characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle. The incidence is 1/3,300 male newborns and the prevalence is 1/16,000 to 1/125,000.	600
DMP1	Hypophosphatemic rickets type 1, autosomal recessive	NM_004407.3	NM_004407.3:c.1A>G, NM_004407.3:c.55-1G>C, NM_004407.3:c.31delT, NM_004407.3:c.362delC	Hypophosphatemic rickets type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DMP1 gene located on chromosomal region 4q21. The age of onset is variable. This disease is associated with vertebral and rib segmentation defects and characterised by hypophosphatemia, rickets and/or osteomalacia and slow growth. The prevalence is below 1/20,000 newborns.	600
DNAJC19	Dilated cardiomyopathy with ataxia	NM_145261.3	NM_145261.3:c.300delA	Dilated cardiomyopathy with ataxia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DNAJC19 gene located on chromosomal region 3q26.33. The age of onset is infantile. This disease is characterised by severe early onset (before the age of three years) dilated cardiomyopathy with conduction defects (long QT syndrome), non-progressive cerebellar ataxia, testicular dysgenesis, and 3-methylglutaconic aciduria.	600
DPAGT1	Congenital disorder of glycosylation type 1j	NM_001382.3	NM_001382.3:c.791T>G, NM_001382.3:c.358C>A, NM_001382.3:c.643+1G>A, NM_001382.3:c.902G>A, NM_001382.3:c.349G>A, NM_001382.3:c.980_981delCT	Congenital disorder of glycosylation type 1j follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DPAGT1 gene located on chromosomal region 11q23.3. The age of onset is neonatal/infantile. This disease is characterised by severe psychomotor delay, seizures, hypotonia and dysmorphism (microcephaly, ocular exotropia, micrognathia and clinodactyly). The prevalence is below 1,000,000.	600
DPM1	Congenital disorders of glycosylation type 1e	NM_003859.1	NM_003859.1:c.564-1G>A, NM_003859.1:c.628delC, NM_003859.1:c.274C>G, NM_003859.1:c.679-1G>T, NM_003859.1:c.742T>C	Congenital disorder of glycosylation type 1e follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DPM1 gene located on chromosomal region 20q13.13. The age of onset is neonatal/infantile. This disease is characterised by psychomotor delay, seizures, hypotonia, facial dysmorphism and microcephaly. The prevalence is below 1,000,000.	600

DPYD	Dihydropyrimidine dehydrogenase deficiency	NM_000110.3	NM_000110.3:c.775A>G, NM_000110.3:c.1679T>G, NM_000110.3:c.299_302delTCAT, NM_000110.3:c.703C>T, NM_000110.3:c.1109_1110delTA, NM_000110.3:c.1905+1G>A, NM_000110.3:c.257C>T	Dihydropyrimidine dehydrogenase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DPYD gene located on chromosomal region 1p22. The age of onset is infantile. This disease is characterised by mental and motor retardation and convulsions.	250,6
DSP	Cardiomyopathy, arrhythmogenic	NM_004415.2	NM_004415.2:c.7000C>T, NM_004415.2:c.88G>A, NM_004415.2:c.6370_6371delCT, NM_004415.2:c.7180_7181delAG, NM_004415.2:c.643G>A, NM_004415.2:c.3098delA, NM_004415.2:c.8188C>T	Cardiomyopathy, arrhythmogenic follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DSP gene located on chromosomal region 6p24. The age of onset is neonatal/infantile. This disease is a heart condition in which the heart muscle fibers are gradually replaced by fibrous or fibro-fatty tissue, causing abnormal heart electrical rhythms and heart failure. Consequently pumping blood to the body is weakened and sometimes leads to sudden cardiac death. The prevalence is below 1,000,000.	250,6
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma	NM_004415.2	NM_004415.2:c.5513G>A	Cardiomyopathy, dilated, with woolly hair and keratoderma follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DSP gene located on chromosomal region 6p24. The age of onset is neonatal/infantile. This disease is characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. The prevalence is below 1,000,000.	250,6
DSP	Lethal acantholytic epidermolysis bullosa	NM_004415.2	NM_004415.2:c.5800C>T	Lethal acantholytic epidermolysis bullosa follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DSP gene located on chromosomal region 6p24. The age of onset is neonatal/infantile. This disease is characterised by generalized oozing erosions, usually in the absence of blisters. The prevalence is below 1,000,000.	250,6
DYSF	Dysferlinopathy	NM_003494.3	NM_003494.3:c.1398-2A>G, NM_003494.3:c.1392dupA, NM_003494.3:c.1398-1G>A, NM_003494.3:c.5266C>T, NM_003494.3:c.1620delA, NM_003494.3:c.1481-1G>A, NM_003494.3:c.3041A>G, NM_003494.3:c.3985C>G, NM_003494.3:c.4090C>T, NM_003494.3:c.5713C>T, NM_003494.3:c.1053+1G>A, NM_003494.3:c.200_201delTGinsAT, NM_003494.3:c.2869C>T, NM_003494.3:c.2870_2874delAGACC, NM_003494.3:c.458-390C>T, NM_003494.3:c.757C>T, NM_003494.3:c.3065G>A, NM_003494.3:c.393_394delCC, NM_003494.3:c.3859A>T, NM_003494.3:c.5429G>A, NM_003494.3:c.3130C>T, NM_003494.3:c.3444_3445delTGinsAA, NM_003494.3:c.1638+2T>A, NM_003494.3:c.4108_4109delGT, NM_003494.3:c.3641delC, NM_003494.3:c.1368C>A, NM_003494.3:c.4872_4876delGCCCGinsCCCC, NM_003494.3:c.5341-2A>C, NM_003494.3:c.509C>A, NM_003494.3:c.5836_5839delCAGC, NM_003494.3:c.5644C>T, NM_003494.3:c.1861G>C, NM_003494.3:c.5429+1G>T, NM_003494.3:c.3957delC, NM_003494.3:c.5998C>T, NM_003494.3:c.3724C>T, NM_003494.3:c.5525+1G>A, NM_003494.3:c.3477C>A, NM_003494.3:c.3708delA, NM_003494.3:c.5992G>T, NM_003494.3:c.3113G>C, NM_003494.3:c.1216T>C, NM_003494.3:c.3903delG	Dysferlinopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DYSF gene located on chromosomal region 2p13.3. The age of onset is adult. Dysferlinopathy includes a spectrum of muscle disease characterized by two main phenotypes: Miyoshi myopathy with primarily distal weakness and limb-girdle muscular dystrophy type 2B (LGMD2B) with primarily proximal weakness. Miyoshi myopathy (median age of onset 19 years) is characterized by muscle weakness and atrophy, most marked in the distal parts of the legs, especially the gastrocnemius and soleus muscles. Over a period of years, the weakness and atrophy spread to the thighs and gluteal muscles. The forearms may become mildly atrophic with decrease in grip strength; the small muscles of the hands are spared. LGMD2B is characterized by early weakness and atrophy of the pelvic and shoulder girdle muscles in adolescence or young adulthood, with slow progression. Other phenotypes are scapuloperoneal syndrome, distal myopathy with anterior tibial onset, elevated serum CK concentration only, and congenital muscular dystrophy. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
DYSF	Miyoshi myopathy	NM_003494.3	NM_003494.3:c.1555G>A, NM_003494.3:c.5509G>A, NM_003494.3:c.5077C>T, NM_003494.3:c.5698_5699delAG, NM_003494.3:c.3892A>G, NM_003494.3:c.286A>C, NM_003494.3:c.1120G>C, NM_003494.3:c.1284+2T>C, NM_003494.3:c.5497G>T, NM_003494.3:c.3478C>T, NM_003494.3:c.2997G>T, NM_003494.3:c.3121C>T, NM_003494.3:c.1813C>T, NM_003494.3:c.3181_3182insAGGCGG, NM_003494.3:c.937+1G>A, NM_003494.3:c.3158T>G, NM_003494.3:c.1276G>A, NM_003494.3:c.701G>A, NM_003494.3:c.610C>T, NM_003494.3:c.5594delG, NM_003494.3:c.3112C>T, NM_003494.3:c.4199C>A, NM_003494.3:c.5999G>A, NM_003494.3:c.4756C>T, NM_003494.3:c.6124C>T, NM_003494.3:c.2966C>T, NM_003494.3:c.663+1G>C, NM_003494.3:c.3175-2A>T, NM_003494.3:c.895G>T, NM_003494.3:c.4985C>T, NM_003494.3:c.6203C>T	Miyoshi myopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DYSF gene located on chromosomal region 2p13.3. The age of onset is adult. This disease is characterised by weakness in the distal lower extremity posterior compartment (gastrocnemius and soleus muscles) and is associated with difficulties in standing on tip toes. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
DYSF	Muscular dystrophy, limb girdle type 2B	NM_003494.3	NM_003494.3:c.5979dupA, NM_003494.3:c.565C>G, NM_003494.3:c.1663C>T, NM_003494.3:c.1873G>T, NM_003494.3:c.1834C>T, NM_003494.3:c.5201A>G, NM_003494.3:c.895G>A, NM_003494.3:c.3805G>T, NM_003494.3:c.4003G>A, NM_003494.3:c.4253G>A	Muscular dystrophy, limb girdle type 2B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the DYSF gene located on chromosomal region 2p13.3. The age of onset is adult. This disease is characterised by limb-girdle weakness and atrophy mostly in the shoulder pelvic girdle. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6

EDA	Hypohidrotic ectodermal dysplasia, X-linked	NM_001399.4	NM_001399.4:c.206G>T, NM_001399.4:c.463C>T, NM_001399.4:c.187G>A, NM_001399.4:c.573_574insT, NM_001399.4:c.466C>T, NM_001399.4:c.826C>T, NM_001399.4:c.183C>G, NM_001399.4:c.181T>C, NM_001399.4:c.467G>A, NM_001399.4:c.671G>C, NM_001399.4:c.1045G>A	Hypohidrotic ectodermal dysplasia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the EDA gene located on chromosomal region Xq12-q13.1. The age of onset is neonatal/infantile. This disease is characterized by malformation of ectodermal structures such as skin, hair, teeth and sweat glands. The prevalence is 1/5,000 to 1/10,000 newborns.	250,6
EDN3	Shah-Waardenburg syndrome type 4B	NM_207034.1	NM_207034.1:c.277C>G, NM_207034.1:c.568_569delGA, NM_207034.1:c.262_263delGCinsT, NM_207034.1:c.559_560insA, NM_207034.1:c.565_566insA, NM_207034.1:c.476G>T	Waardenburg-Shah syndrome type 4B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the EDN3 gene located on chromosomal region 20q13.2-q13.3. The age of onset is neonatal/infantile. This disease is characterised by the association of Waardenburg syndrome (sensorineural hearing loss and pigmentary abnormalities) and Hirschsprung disease (signs of intestinal obstruction). The prevalence is below 1/40,000.	600
EDNRB	Shah-Waardenburg syndrome type 4A	NM_000115.3	NM_000115.3:c.914C>A, NM_000115.3:c.548C>G, NM_000115.3:c.828G>T, NM_000115.3:c.-51-946delC	Waardenburg-Shah syndrome type 4A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the EDNRB gene located on chromosomal region 13q22. The age of onset is neonatal/infantile. This disease is characterised by the association of Waardenburg syndrome (sensorineural hearing loss and pigmentary abnormalities) and Hirschsprung disease (signs of intestinal obstruction). The prevalence is below 1/1,000,000.	600
EGR2	Charcot-Marie-Tooth disease type 4E	NM_000399.3	NM_000399.3:c.803T>A	Charcot-Marie-Tooth disease type 4E follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the EGR2 gene located on chromosomal region 10q21.1. The age of onset is neonatal/infantile. This disease is characterised by distal muscle weakness and atrophy associated with sensory loss and, frequently, pes cavus foot deformity. The prevalence is 15/100,000 to 20/100,000.	600
EIF2AK3	Wolcott-Rallison syndrome	NM_004836.5	NM_004836.5:c.994G>T, NM_004836.5:c.1763G>A	Wolcott-Rallison syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the EIF2AK3 gene located on chromosomal region 2p12. The age of onset is neonatal/infantile. This disease is characterised by permanent neonatal diabetes mellitus with multiple epiphyseal dysplasia and other clinical manifestations, including recurrent episodes of acute liver failure. The prevalence is above 1/10,000 newborns.	600
EMD	Emery-Dreifuss muscular dystrophy type 1, X-linked	NM_000117.2	NM_000117.2:c.547C>A, NM_000117.2:c.631_635delCGTGC	Emery-Dreifuss muscular dystrophy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the EMD gene located on chromosomal region Xq28. The age of onset is infantile. This disease is characterized by muscular weakness and atrophy, with early joint contractures and cardiomyopathy. The prevalence is 1/100,000.	600
ENO3	Glycogen storage disease type 13	NM_053013.3	NM_053013.3:c.667+1G>T, NM_053013.3:c.1121G>A, NM_053013.3:c.953delA, NM_053013.3:c.692_707dupTCCAGGCGGCTGGTTA, NM_053013.3:c.467G>A, NM_053013.3:c.1303T>C	Glycogen storage disease type 13 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ENO3 gene located on chromosomal region 17p13.2. The age of onset is adult. This disease is characterised by exercise intolerance and myalgia due to severe enolase deficiency in muscle. The prevalence is below 1/1,000,000.	250,6
ENPP1	Generalized arterial calcification of infancy and pseudoxanthoma elasticum	NM_006208.2	NM_006208.2:c.1612G>C	Idiopathic infantile arterial calcification follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ENPP1 gene located on chromosomal region 6q22-q23. The age of onset is neonatal/infancy. A severe autosomal recessive disorder characterized by calcification of the internal elastic lamina of muscular arteries and stenosis due to myointimal proliferation. The disorder is often fatal within the first 6 months of life because of myocardial ischemia resulting in refractory heart failure.	600
ENPP1	Hypophosphatemic rickets type 2, Autosomal recessive	NM_006208.2	NM_006208.2:c.797G>T, NM_006208.2:c.2702A>C	Hypophosphatemic rickets type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ENPP1 gene located on chromosomal region 6q22-q23. The age of onset is variable. This disease is characterised by hypophosphatemia, rickets and/or osteomalacia and slow growth.	600
ENPP1	Idiopathic infantile arterial calcification	NM_006208.2	NM_006208.2:c.1112A>T, NM_006208.2:c.1025G>T, NM_006208.2:c.783C>G, NM_006208.2:c.2677G>T, NM_006208.2:c.913C>A, NM_006208.2:c.2230C>T, NM_006208.2:c.900G>A	Idiopathic infantile arterial calcification follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ENPP1 gene located on chromosomal region 6q22-q23. The age of onset is neonatal/infancy. A severe autosomal recessive disorder characterized by calcification of the internal elastic lamina of muscular arteries and stenosis due to myointimal proliferation. The disorder is often fatal within the first 6 months of life because of myocardial ischemia resulting in refractory heart failure.	600

ERCC2	Trichothiodystrophy	NM_000400.3	NM_000400.3:c.1972C>T	Trichothiodystrophy is a heterogeneous group of disorders that follows an autosomal recessive pattern of inheritance. It is caused by pathogenic variants in the ERCC2 gene located on chromosomal region 19q13.32. The age of onset is neonatal or infantile. This disease is characterized by brittle and fragile hair, often combined with growth retardation and intellectual deficit, congenital ichthyosis and nail abnormalities, among other symptoms. The abnormalities are usually obvious at birth, with variable clinical expression.	250,6
ERCC2	Xeroderma pigmentosum complementation group D	NM_000400.3	NM_000400.3:c.1308-1G>A, NM_000400.3:c.1454T>C, NM_000400.3:c.1621A>C, NM_000400.3:c.1703_1704delTT, NM_000400.3:c.1381C>G, NM_000400.3:c.719-1G>A, NM_000400.3:c.2230_2233dupCTAG, NM_000400.3:c.183+2T>A, NM_000400.3:c.567G>A, NM_000400.3:c.1354C>T, NM_000400.3:c.2047C>T, NM_000400.3:c.1304T>G, NM_000400.3:c.2176C>T, NM_000400.3:c.950-2A>G, NM_000400.3:c.949+1G>A	Xeroderma pigmentosum complementation group D follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC2 gene located on chromosomal region 19q13.3. The age of onset is variable. This disease is characterised by typical xeroderma pigmentosum manifestations (photosensitivity of skin with burning, freckling, and dryness of skin, skin cancers) associated with a spectrum of neurological anomalies (from no abnormality to severe neurological disease).	250,6
ERCC3	Xeroderma pigmentosum complementation group B	NM_000122.1	NM_000122.1:c.1633C>T, NM_000122.1:c.1757_1758delAG, NM_000122.1:c.296T>C, NM_000122.1:c.1273C>T, NM_000122.1:c.1757delA, NM_000122.1:c.1858delG	Xeroderma pigmentosum complementation group B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC3 gene located on chromosomal region 2q21. The age of onset is variable. This disease is characterised by classic xeroderma pigmentosum features of varying severity (photosensitivity of skin with burning and freckling, skin and eye tumors) and mild neurological abnormalities, or in other cases classical xeroderma pigmentosum features with systemic and neurological manifestations of Cockayne syndrome such as short stature, bilateral sensorineural hearing loss and hyperreflexia. The prevalence is 1/1,000,000.	600
ERCC4	Xeroderma pigmentosum complementation group F	NM_005236.2	NM_005236.2:c.49G>T, NM_005236.2:c.1467_1468insA, NM_005236.2:c.2281_2284delTTTG, NM_005236.2:c.2T>C, NM_005236.2:c.538_539delAG, NM_005236.2:c.706T>C, NM_005236.2:c.2395C>T	Xeroderma pigmentosum complementation group F follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC4 gene located on chromosomal region 16p13.12. The age of onset is variable. This disease is characterised very mild skin symptoms and no ocular or neurological disease. The prevalence is 1/1,000,000.	250,6
ERCC5	Xeroderma pigmentosum complementation group G	NM_000123.3	NM_000123.3:c.2620G>A, NM_000123.3:c.463_464insA, NM_000123.3:c.526C>T, NM_000123.3:c.88+2T>C, NM_000123.3:c.2144dupA, NM_000123.3:c.2375C>T, NM_000123.3:c.381-2A>G, NM_000123.3:c.2573T>C, NM_000123.3:c.406C>T, NM_000123.3:c.215C>A, NM_000123.3:c.787C>T, NM_000123.3:c.2751delA	Xeroderma pigmentosum complementation group G follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC5 gene located on chromosomal region 13q33. The age of onset is variable. This disease is characterised by variable clinical manifestations, as some patients present with a mild xeroderma pigmentosum phenotype (UV sensitivity, hyper- or hypo-pigmented skin lesions and increased incidence of skin cancer) and others combine symptoms of xeroderma pigmentosum with systemic and neurological manifestations of Cockayne syndrome. The prevalence is 1/1,000,000.	250,6
ERCC6	Cerebrooculofacioskeletal syndrome tipo 1	NM_000124.3	NM_000124.3:c.2047C>T	Cerebrooculofacioskeletal syndrome tipo 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC6 gene located on chromosomal region 10q11.23. The age of onset is variable. This disease is characterised by congenital microcephaly, congenital cataract and/or microphthalmia, arthrogryposis, severe psychomotor developmental delay, height-weight growth delay (principally postnatal) and facial dysmorphism (prominent metopic suture, micrognathism). The prevalence is below 1,000,000.	250,6
ERCC6	Cockayne syndrome type B	NM_000124.3	NM_000124.3:c.207_208insG, NM_000124.3:c.2203C>T, NM_000124.3:c.1357C>T, NM_000124.3:c.48_49delCT, NM_000124.3:c.3592_3593insGA, NM_000124.3:c.422+1G>A, NM_000124.3:c.1550G>A, NM_000124.3:c.3284C>G, NM_000124.3:c.2587C>T, NM_000124.3:c.3862C>T	Cockayne syndrome type B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC6 gene located on chromosomal region 10q11.23. The age of onset is variable. This disease is characterised by growth failure at birth, with little or no postnatal neurologic development.	250,6
ERCC8	Cockayne syndrome type A	NM_000082.3	NM_000082.3:c.1103_1108delAGTTinsTTATATGAACCTTATATGAA, NM_000082.3:c.618-1G>A, NM_000082.3:c.593_594dupAT, NM_000082.3:c.613G>C, NM_000082.3:c.966C>A, NM_000082.3:c.37G>T	Cockayne syndrome type A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ERCC8 gene located on chromosomal region 5q12.1. The age of onset is variable. This disease is characterised by normal prenatal growth with the onset of growth and developmental abnormalities in the first two years. The prevalence is 2.7/1,000,000 newborns in Western Europe.	600
ESCO2	Roberts syndrome	NM_001017420.2	NM_001017420.2:c.1615T>G, NM_001017420.2:c.879_880delAG, NM_001017420.2:c.1597dupT, NM_001017420.2:c.505C>T, NM_001017420.2:c.291_292insGA, NM_001017420.2:c.308_309delAA, NM_001017420.2:c.876_879delCAGA, NM_001017420.2:c.874_877delGACA	Roberts syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ESCO2 gene located on chromosomal region 8p21.1. The age of onset is neonatal/infantile. This disease is characterised by pre- and postnatal growth retardation, severe symmetric limb reduction defects, craniofacial anomalies and severe intellectual deficit.	600

ESCO2	SC Phocomelia syndrome	NM_001017420.2	NM_001017420.2:c.1269G>A, NM_001017420.2:c.604C>T	SC phocomelia syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ESCO2 gene located on chromosomal region 8p21.1. The age of onset is neonatal/infantile. This disease has a milder phenotype than Roberts syndrome, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common.	600
ESPN	Deafness type 36, autosomal recessive	NM_031475.2	NM_031475.2:c.1988_1991delAGAG, NM_031475.2:c.2230G>A, NM_031475.2:c.2470_2473delTCAG	Autosomal recessive nonsyndromic sensorineural deafness type DFNB36 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ESPN gene located on chromosomal region 1p36.31. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.	600
ESRRB	Deafness type 35, autosomal recessive	NM_004452.3	NM_004452.3:c.329C>T	Autosomal recessive nonsyndromic sensorineural deafness type DFNB35 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ESRRB gene located on chromosomal region 14q24.3. The age of onset is neonatal/infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment.	600
ETFA	Glutaric acidemia type 2A	NM_000126.3	NM_000126.3:c.470T>G, NM_000126.3:c.797C>T	Glutaric acidemia type 2A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ETFA gene located on chromosomal region 15q23-q25. The age of onset is variable. This disease is characterized by clinically heterogeneous symptoms ranging from a severe neonatal presentation with metabolic acidosis, cardiomyopathy and liver disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure.	600
ETFB	Glutaric acidemia type 2B	NM_001985.2	NM_001985.2:c.278_279insG, NM_001985.2:c.490C>T, NM_001985.2:c.491G>A, NM_001985.2:c.382G>A, NM_001985.2:c.58-53_58-52insG, NM_001985.2:c.61C>T, NM_001985.2:c.614_616delAGA	Glutaric acidemia type 2B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ETFB gene located on chromosomal region 19q13.3. The age of onset is variable. This disease is characterized by clinically heterogeneous symptoms ranging from a severe neonatal presentation with metabolic acidosis, cardiomyopathy and liver disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure.	600
ETFDH	Glutaric acidemia type 2C	NM_004453.3	NM_004453.3:c.1823delG, NM_004453.3:c.1570_1571delCT, NM_004453.3:c.2T>C, NM_004453.3:c.1234G>T, NM_004453.3:c.250G>A, NM_004453.3:c.1351G>C, NM_004453.3:c.1367C>T, NM_004453.3:c.524G>T, NM_004453.3:c.1001T>C, NM_004453.3:c.1773_1774delAT, NM_004453.3:c.1832G>A, NM_004453.3:c.508G>T, NM_004453.3:c.413T>G, NM_004453.3:c.643G>A	Glutaric acidemia type 2C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ETFDH gene located on chromosomal region 4q32-q35. The age of onset is variable. This disease is characterized by clinically heterogeneous symptoms disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure.	600
ETHE1	Ethylmalonic encephalopathy	NM_014297.3	NM_014297.3:c.487C>T, NM_014297.3:c.554T>G, NM_014297.3:c.440_450delACAGCATGGCC, NM_014297.3:c.604dupG, NM_014297.3:c.221dupA, NM_014297.3:c.488G>A	Ethylmalonic encephalopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ETHE1 gene located on chromosomal region 19q13.31. The age of onset is neonatal/infantile. This disease is characterised by elevated excretion of ethylmalonic acid with recurrent petechiae, orthostatic acrocyanosis and chronic diarrhoea associated with neurodevelopmental delay, psychomotor regression and hypotonia with brain magnetic resonance imaging abnormalities. The prevalence is below 1/1,000,000, with total of 30 cases of patients reported worldwide, mainly for Mediterranean and Arab populations.	600
EYS	Retinitis pigmentosa type 25	NM_001142800.1	NM_001142800.1:c.5044G>T, NM_001142800.1:c.9036delT, NM_001142800.1:c.490C>T, NM_001142800.1:c.5928-2A>G, NM_001142800.1:c.571dupA, NM_001142800.1:c.4597_4613delTCAAGCAACCAGAGACT, NM_001142800.1:c.7822C>T, NM_001142800.1:c.5857G>T, NM_001142800.1:c.6170delA, NM_001142800.1:c.8569G>T, NM_001142800.1:c.232delT, NM_001142800.1:c.6102_6103insT, NM_001142800.1:c.8834G>A, NM_001142800.1:c.1211_1212insA, NM_001142800.1:c.4350_4356delTATAGCT, NM_001142800.1:c.4469_4470insAGCCCCTC, NM_001142800.1:c.8648_8655delCATGCAGA, NM_001142800.1:c.4120C>T, NM_001142800.1:c.863-4_863-3insT, NM_001142800.1:c.8629_8632dupACAG, NM_001142800.1:c.9299_9302delCTCA, NM_001142800.1:c.103C>T, NM_001142800.1:c.2826_2827delAT, NM_001142800.1:c.4045C>T, NM_001142800.1:c.5757_5758insT, NM_001142800.1:c.8408dupA, NM_001142800.1:c.7095T>G, NM_001142800.1:c.3329C>G, NM_001142800.1:c.9405T>A	Retinitis pigmentosa 25 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the EYS gene located on chromosomal region 6q12. The age of onset is variable. This disease is characterized by night blindness (nyctalopia), peripheral visual field impairment and over time loss of central vision. The prevalence is 1/10,000 to 5/10,000.	250,6

F11	Factor 11 deficiency	NM_000128.3	NM_000128.3:c.1613C>T, NM_000128.3:c.166T>C, NM_000128.3:c.403G>T, NM_000128.3:c.731A>G, NM_000128.3:c.809A>T, NM_000128.3:c.1693G>A, NM_000128.3:c.1211C>A, NM_000128.3:c.901T>C, NM_000128.3:c.595+3A>G, NM_000128.3:c.438C>A	Factor 11 deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the F11 gene located on chromosomal region 4q35. The age of onset is variable. This disease is characterized by reduced levels and activity of factor XI resulting in moderate bleeding symptoms, usually occurring after trauma or surgery. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
F5	Factor 5 deficiency	NM_000130.4	NM_000130.4:c.4876delA, NM_000130.4:c.439G>T, NM_000130.4:c.6419G>A, NM_000130.4:c.2401C>T, NM_000130.4:c.5521G>A, NM_000130.4:c.1083G>A, NM_000130.4:c.5189A>G, NM_000130.4:c.3799delC, NM_000130.4:c.6304C>T	Factor 5 deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the F5 gene located on chromosomal region 1q23. The age of onset is variable. This disease is characterized by mild to severe bleeding symptoms usually occurring after trauma or surgery. In severe forms of the disease, there can be a risk of intracranial, pulmonary or gastrointestinal bleedings. The severity of the bleeding manifestations correlates with the FV levels. The prevalence is 1/5,000.	250,6
F5	Thrombosis	NM_000130.4	NM_000130.4:c.1000A>G	Deep venous thrombosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the F5 gene located on chromosomal region 1q23. The age of onset is variable. This disease is characterized by a poor anticoagulant response to activated Protein C and an increased risk for venous thromboembolism. Deep venous thrombosis and pulmonary embolism are the most common manifestations, but thrombosis in unusual locations also occurs. The prevalence is 1/5,000.	250,6
F8	Hemophilia A	NM_000132.3	NM_000132.3:c.1075_1078delAATG, NM_000132.3:c.1042T>C, NM_000132.3:c.1078_1079delGA, NM_000132.3:c.120delC, NM_000132.3:c.1214T>G, NM_000132.3:c.1090G>A, NM_000132.3:c.1207C>G, NM_000132.3:c.1331_1332delAAinsT, NM_000132.3:c.1175C>A, NM_000132.3:c.1335dupC, NM_000132.3:c.1203G>A, NM_000132.3:c.128dupT, NM_000132.3:c.1331A>C, NM_000132.3:c.1301G>A, NM_000132.3:c.1234T>C, NM_000132.3:c.1316G>A, NM_000132.3:c.1293delG, NM_000132.3:c.1200_1201delTT, NM_000132.3:c.1310delG, NM_000132.3:c.1331_1332delAA, NM_000132.3:c.1410_1413delTTTA, NM_000132.3:c.1420G>T, NM_000132.3:c.143+1G>A, NM_000132.3:c.1432G>A, NM_000132.3:c.1438_1439delCT, NM_000132.3:c.1440_1441insA, NM_000132.3:c.144-11T>G, NM_000132.3:c.1442_1443dupTG, NM_000132.3:c.1175C>G, NM_000132.3:c.1324T>A, NM_000132.3:c.1324T>C, NM_000132.3:c.1325A>G, NM_000132.3:c.144-5C>G, NM_000132.3:c.1463C>G, NM_000132.3:c.1463C>T, NM_000132.3:c.1467_1472dupCAGACC, NM_000132.3:c.1477A>G, NM_000132.3:c.1538-1G>T, NM_000132.3:c.1538-2A>T, NM_000132.3:c.1560delT, NM_000132.3:c.1564_1565delATinsTA, NM_000132.3:c.1585A>G, NM_000132.3:c.1594T>G, NM_000132.3:c.1189_1190insC, NM_000132.3:c.1443+3A>C, NM_000132.3:c.1596G>A, NM_000132.3:c.1618C>A, NM_000132.3:c.1619C>G, NM_000132.3:c.1630G>A, NM_000132.3:c.1639T>C, NM_000132.3:c.1337G>A, NM_000132.3:c.1337G>C, NM_000132.3:c.1338delA, NM_000132.3:c.1348T>G, NM_000132.3:c.1357G>T, NM_000132.3:c.1390G>T, NM_000132.3:c.1595G>A, NM_000132.3:c.1596dupG, NM_000132.3:c.1400T>G, NM_000132.3:c.1406G>C, NM_000132.3:c.1736A>T, NM_000132.3:c.173delC, NM_000132.3:c.1752+5G>C, NM_000132.3:c.185C>G, NM_000132.3:c.1904-1G>A, NM_000132.3:c.1904-37G>A, NM_000132.3:c.1912G>A, NM_000132.3:c.1913G>A, NM_000132.3:c.1924_1927delGATA, NM_000132.3:c.1934A>C, NM_000132.3:c.1941_1944delAGTT, NM_000132.3:c.1943_1946delTTTG, NM_000132.3:c.1952A>C, NM_000132.3:c.195C>A, NM_000132.3:c.1985G>C, NM_000132.3:c.1988C>T, NM_000132.3:c.1990_1991delCA, NM_000132.3:c.1991A>C, NM_000132.3:c.199_200delAA, NM_000132.3:c.1992_1995dupGACT, NM_000132.3:c.1996_1999delGACT, NM_000132.3:c.1999delT, NM_000132.3:c.199A>G, NM_000132.3:c.1A>G, NM_000132.3:c.2009_2011delTCT	Hemophilia A follows an X-linked pattern of inheritance and is caused by pathogenic variants in the F8 gene located on chromosomal region Xq28. The age of onset is neonatal/infantile. This disease is characterized by spontaneous or prolonged hemorrhages due to factor VIII deficiency. The prevalence is 1/4,000 to 1/ 5,000 male newborns.	600
F8	Hemophilia A	-	Inv22 (Detection by PCR)	Hemophilia A follows an X-linked pattern of inheritance and is caused by pathogenic variants in the F8 gene located on chromosomal region Xq28. The age of onset is neonatal/infantile. This disease is characterized by spontaneous or prolonged hemorrhages due to factor VIII deficiency. The prevalence is 1/4,000 to 1/ 5,000 male newborns.	600
F9	Hemophilia B	NM_000133.3	NM_000133.3:c.1150C>T, NM_000133.3:c.52T>C, NM_000133.3:c.1031T>C, NM_000133.3:c.82T>C, NM_000133.3:c.1136G>A, NM_000133.3:c.79G>A, NM_000133.3:c.19A>T, NM_000133.3:c.80A>T	Hemophilia B follows an X-linked pattern of inheritance and is caused by pathogenic variants in the F9 gene located on chromosomal region Xq27.1-q27.2. The age of onset is neonatal/infantile. This disease is characterized by spontaneous or prolonged hemorrhages due to factor IX deficiency. The prevalence is 1/100,000 to 9/100,000.	250,6

FAH	Tyrosinemia type 1	NM_000137.2	NM_000137.2:c.1141A>G, NM_000137.2:c.1069G>T, NM_000137.2:c.1090G>T, NM_000137.2:c.401C>A, NM_000137.2:c.456G>A, NM_000137.2:c.192G>T, NM_000137.2:c.607-6T>G, NM_000137.2:c.707-1G>A, NM_000137.2:c.939delC, NM_000137.2:c.103G>A, NM_000137.2:c.982C>T, NM_000137.2:c.837+1G>A, NM_000137.2:c.1009G>A, NM_000137.2:c.47A>T, NM_000137.2:c.554-1G>T, NM_000137.2:c.1027G>T, NM_000137.2:c.1062+5G>A, NM_000137.2:c.786G>A, NM_000137.2:c.1021C>T, NM_000137.2:c.782C>T	Tyrosinemia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FAH gene located on chromosomal region 15q25.1. The age of onset is variable. This disease is characterized by progressive liver disease, renal tubular dysfunction, porphyria-like crises and a dramatic improvement in prognosis following treatment with nitisinone. The birth incidence is 1/100,000, notably in QuÃ©bec, Canada, and the prevalence is 1/100,000 to 1/120,000 newborns.	250,6
FAM126A	Hypomyelination and congenital cataract	NM_032581.3	NM_032581.3:c.191A>G, NM_032581.3:c.158T>C	Hypomyelination - congenital cataract follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FAM126A gene located on chromosomal region 7p15.3. The age of onset is neonatal/infantile. This disease is characterized by the onset of cataract either at birth or in the first two months of life, delayed psychomotor development by the end of the first year of life and moderate intellectual deficit. The prevalence is below 1/1,000,000.	600
FAM20C	Osteosclerotic bone dysplasia	NM_020223.3	NM_020223.3:c.1093G>C, NM_020223.3:c.773T>A, NM_020223.3:c.1364-5C>T, NM_020223.3:c.1163T>G, NM_020223.3:c.838G>A, NM_020223.3:c.1351G>A	Osteosclerotic bone dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FAM20C gene located on chromosomal region 7p22.3. The age of onset is neonatal/infantile. This disease is characterized by generalized osteosclerosis with periosteal bone formation, characteristic facial dysmorphism, brain abnormalities including intracerebral calcifications, and neonatal lethal course. The prevalence is below 1/1,000,000.	600
FANCA	Fanconi anemia, complementation group A	NM_000135.2	NM_000135.2:c.3788_3790delTCT, NM_000135.2:c.2303T>C, NM_000135.2:c.3558_3559insG, NM_000135.2:c.4130C>G, NM_000135.2:c.233_236delTTGA, NM_000135.2:c.3763G>T, NM_000135.2:c.1115_1118delTTGG, NM_000135.2:c.131_132insA	Fanconi anemia complementation group A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCA gene located on chromosomal region 16q24.3. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FANCC	Fanconi anemia, complementation group C	NM_000136.2	NM_000136.2:c.1642C>T, NM_000136.2:c.37C>T, NM_000136.2:c.996+1G>T, NM_000136.2:c.67delG, NM_000136.2:c.416G>A, NM_000136.2:c.1015delA, NM_000136.2:c.1487T>G, NM_000136.2:c.1103_1104delTG	Fanconi anemia complementation group C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCC gene located on chromosomal region 9q22.3. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FANCD2	Fanconi anemia, complementation group D2	NM_033084.3	NM_033084.3:c.1278+1delG, NM_033084.3:c.2152C>T, NM_033084.3:c.2494+2T>C, NM_033084.3:c.958C>T, NM_033084.3:c.2444G>A, NM_033084.3:c.782A>T, NM_033084.3:c.904C>T	Fanconi anemia complementation group D2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCD2 gene located on chromosomal region 3p26. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FANCE	Fanconi anemia, complementation group E	NM_021922.2	NM_021922.2:c.1501C>T, NM_021922.2:c.929_930insC, NM_021922.2:c.421C>T, NM_021922.2:c.1114-8G>A, NM_021922.2:c.922_923insC, NM_021922.2:c.355C>T	Fanconi anemia complementation group E follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCE gene located on chromosomal region 6p22-p21. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	600
FANCG	Fanconi anemia, complementation group G	NM_004629.1	NM_004629.1:c.1795_1804delTGGATCCGTC, NM_004629.1:c.313G>T, NM_004629.1:c.637_643delTACCGCC, NM_004629.1:c.1480+1G>C, NM_004629.1:c.1852_1853delAA, NM_004629.1:c.510+1G>A, NM_004629.1:c.1077-2A>G, NM_004629.1:c.908_909insCT	Fanconi anemia complementation group G follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCG gene located on chromosomal region 9p13. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FANCI	Fanconi anemia, complementation group I	NM_001113378.1	NM_001113378.1:c.3816+1G>A, NM_001113378.1:c.52C>T, NM_001113378.1:c.989_991delTAA, NM_001113378.1:c.2097C>G, NM_001113378.1:c.3466G>C, NM_001113378.1:c.2292-1G>A, NM_001113378.1:c.3492delG, NM_001113378.1:c.3853C>T, NM_001113378.1:c.3626_3627delGT, NM_001113378.1:c.3854G>A	Fanconi anemia complementation group I follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCI gene located on chromosomal region 15q26.1. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FANCL	Fanconi anemia, complementation group L	NM_018062.3	NM_018062.3:c.1051_1052delAG, NM_018062.3:c.1066_1067delAG, NM_018062.3:c.1096_1099dupATTA, NM_018062.3:c.1099_1100insATTA	Fanconi anemia complementation group L follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCL gene located on chromosomal region 2p16.1. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6

FANCM	Fanconi anemia, complementation group M	NM_020937.2	NM_020937.2:c.2171C>A, NM_020937.2:c.5766_5769delGACT, NM_020937.2:c.5101C>T, NM_020937.2:c.1072G>T, NM_020937.2:c.2996_2997insC, NM_020937.2:c.2586_2589delAAAA, NM_020937.2:c.5791C>T, NM_020937.2:c.624_625delAA, NM_020937.2:c.5569G>A, NM_020937.2:c.5764_5767delCTGA	Fanconi anemia complementation group M follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FANCM gene located on chromosomal region 14q21.2. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FGA	Congenital fibrinogen deficiency (gene FGA)	NM_021871.2	NM_021871.2:c.1039C>T, NM_021871.2:c.1441delG, NM_021871.2:c.*675_*676insC, NM_021871.2:c.1359dupC, NM_021871.2:c.*1086delG, NM_021871.2:c.1906_1907insC	Congenital fibrinogen deficiency (gene FGA) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FGA gene located on chromosomal region 4q28. The age of onset is variable. This disease is characterized by bleeding symptoms ranging from mild to severe resulting from reduced quantity and/or quality of circulating fibrinogen. The prevalence is 1/1,000,000 to 9/1,000,000.	600
FGB	Congenital afibrinogenemia	NM_005141.4	NM_005141.4:c.1289G>A, NM_005141.4:c.1148T>G, NM_005141.4:c.794C>T	Congenital afibrinogenemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FGB gene located on chromosomal region 4q28. The age of onset is variable. This disease is characterized by bleeding symptoms ranging from mild to severe resulting from reduced quantity and/or quality of circulating fibrinogen. The prevalence is 1/1,000,000 to 9/1,000,000.	250,6
FGD4	Charcot-Marie-Tooth disease type 4H	NM_139241.2	NM_139241.2:c.1325G>A, NM_139241.2:c.893T>G, NM_139241.2:c.893T>C, NM_139241.2:c.670C>T	Charcot-Marie-Tooth disease type 4H follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FGD4 gene located on chromosomal region 12p11.21. The age of onset is neonatal/infantile. This disease is characterized by slowly progressive muscle weakness in the distal extremities, and other common features include delayed walking, an abnormal gait, scoliosis and pes equines with toe retraction. The prevalence is 1/3,300.	600
FH	Fumaric aciduria	NM_000143.3	NM_000143.3:c.1067T>A, NM_000143.3:c.697C>T, NM_000143.3:c.698G>A, NM_000143.3:c.1236+1G>C, NM_000143.3:c.901dupA, NM_000143.3:c.320A>C, NM_000143.3:c.760C>T, NM_000143.3:c.1431_1433dupAAA, NM_000143.3:c.521C>G, NM_000143.3:c.1093A>G, NM_000143.3:c.1189G>A, NM_000143.3:c.1200delT, NM_000143.3:c.1394A>G, NM_000143.3:c.1255T>C, NM_000143.3:c.793G>A, NM_000143.3:c.40_41insC, NM_000143.3:c.1446_1449delAAAG, NM_000143.3:c.1293delA	Fumaric aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FH gene located on chromosomal region 1q42.1. The age of onset is neonatal/infantile. This disease is characterized byÂ hypotonia, severe psychomotor impairment, convulsions, respiratory distress, feeding difficulties and frequent cerebral malformations, along with a distinctive facies, although some patients present with only moderate intellectual impairment. The prevalence is below 1,000,000.	600
FHL1	Emery-Dreifuss muscular dystrophy type 6	NM_001449.4	NM_001449.4:c.625T>C	Emery-Dreifuss muscular dystrophy tipe 6 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the FHL1 gene located on chromosomal region Xq26. The age of onset is infantile. This disease is by muscular weakness and atrophy, with early joint contractures and cardiomyopathy. The prevalence is 1/1,000,000 to 9/1,000,000.	600
FHL1	Myopathy, reducing body	NM_001449.4	NM_001449.4:c.689-479G>A, NM_001449.4:c.310T>C	Reducing body myopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FH gene located on chromosomal region 1q42.1. The age of onset is neonatal/infantile. This disease is characterized by progressive muscle weakness and the presence of characteristic inclusion bodies in affected muscle fibres. The prevalence is below 1,000,000.	600
FIG4	Charcot-Marie-Tooth disease type 4J	NM_014845.5	NM_014845.5:c.592C>T, NM_014845.5:c.831_838delTAAATTTG, NM_014845.5:c.547C>T, NM_014845.5:c.501C>G, NM_014845.5:c.737G>A, NM_014845.5:c.122T>C, NM_014845.5:c.2296_2297insG	Charcot-Marie-Tooth disease type 4J follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FIG4 gene located on chromosomal region 6q.21. The age of onset is neonatal/infantile. This disease is characterized by rapidly progressive, asymmetric motor neuron degeneration with slow nerve conduction velocities, weakness and paralysis, without sensory loss. The prevalence is 4/100,000 to 8/100,000.	250,6
FIG4	Yunis-Varon syndrome	NM_014845.5	NM_014845.5:c.311G>A	Yunis-Varon syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FIG4 gene located on chromosomal region 6q.21. The age of onset is neonatal/infantile. This disease is characterized by skeletal defects, including cleidocranial dysplasia and digital anomalies, and severe neurologic involvement with neuronal loss. Enlarged cytoplasmic vacuoles are found in neurons, muscle, and cartilage. The disorder is usually lethal in infancy. The prevalence is 4/100,000 to 8/100,000.	250,6
FKRP	Congenital muscular dystrophy type 5B	NM_024301.4	NM_024301.4:c.235G>A, NM_024301.4:c.1343C>T, NM_024301.4:c.1387A>G, NM_024301.4:c.1154C>A	Congenital muscular dystrophy type 5B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FKRP gene located on chromosomal region 19q13.32. The age of onset is neonatal/infantile. This disease is characterized by hypotonia, muscle wasting, weakness or delayed motor milestones. The prevalence is 1/14,500 to 1/123,000.	250,6

FKRP	Limb-girdle muscular dystrophy type 2I, autosomal recessive	NM_024301.4	NM_024301.4:c.160C>T	Autosomal recessive limb-girdle muscular dystrophy type 2I follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FKRP gene located on chromosomal region 19q13.32. The age of onset is infantile. This disease is characterized by proximal limb girdle weakness predominant in the legs, together with bilateral moderate scapulae winging, abdominal muscle weakness, waddling gait, calf hypertrophy, cardiomyopathy and respiratory insufficiency. The prevalence is 1/14,500 to 1/123,000.	250,6
FKTN	Fukuyama congenital muscular dystrophy	NM_001079802.1	NM_001079802.1:c.1112A>G, NM_001079802.1:c.509C>A, NM_001079802.1:c.411C>A, NM_001079802.1:c.1167dupA	Fukuyama congenital muscular dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FKTN gene located on chromosomal region 9q31-q33. The age of onset is infantile. This disease is characterized by brain malformation (cobblestone lissencephaly), dystrophic changes in skeletal muscle, severe intellectual deficit, epilepsy and motor impairment. The annual incidence is 1/50,000 to 2:50,000 live births in Japans and the prevalence is 1:1,000,000- 9:1,000,000.	600
FKTN	Muscular dystrophy, limb girdle, type 2M	NM_001079802.1	NM_001079802.1:c.1380dupA, NM_001079802.1:c.766C>T, NM_001079802.1:c.527T>C, NM_001079802.1:c.340G>A	Limb-girdle muscular dystrophy type 2M follows autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FKTN gene located on chromosomal region 9q31-q33. The age of onset is from birth to early infancy. This disease is characterized by muscle weakness, joint contractures and spinal deformities. The prevalence is 1.92:100,000-3.68:100,000.	600
FLNA	Frontometaphyseal dysplasia	NM_001456.3	NM_001456.3:c.4447_4448insAT, NM_001456.3:c.760G>A, NM_001456.3:c.3476A>C, NM_001456.3:c.3557C>T	Fronto-metaphyseal dysplasia (FMD) follows an X-linked pattern of inheritance and is caused by pathogenic variants in the FLNA gene located on chromosomal region Xq28. The age of onset is neonatal/infancy This disease is characterized by a characteristic face (prominent supraorbital ridges, hypertelorism, downslanted palpebral fissures, broad nasal bridge, and micrognathia with anomalies of teeth) and skeletal anomalies (fusion of carpal bones, increased density of long bone diaphyses, flared metaphyses and scoliosis). The prevalence is <1 / 1,000,000.	600
FLNA	Periventricular heterotopia	NM_001456.3	NM_001456.3:c.4543C>T, NM_001456.3:c.7129C>T, NM_001456.3:c.7733-1G>C, NM_001456.3:c.7527_7528+6delAGGTGAGC, NM_001456.3:c.5108_5109delTCinsAA, NM_001456.3:c.2761C>T, NM_001456.3:c.4777_4778dupAA	Periventricular heterotopia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the FLNA gene located on chromosomal region Xq28. The age of onset is neonatal/infancy This disease is a brain malformation, due to abnormal neuronal migration, in which a subset of neurons fails to migrate into the developing cerebral cortex and remains as nodules that line the ventricular surface. Classical form is a rare X-linked dominant disorder far more frequent in females who present normal intelligence to borderline intellectual deficit, epilepsy of variable severity and extra-central nervous system signs, especially cardiovascular defects or coagulopathy. The disorder is generally associated with prenatal lethality in males. The prevalence is <1 / 1,000,000.	600
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	NM_014053.3	NM_014053.3:c.361A>G, NM_014053.3:c.574T>C, NM_014053.3:c.739-2delA	Posterior column ataxia - Retinitis pigmentosa follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FLVCR1 gene located on chromosomal region 1q32.3. The age of onset is childhood. This disease is characterized by sensory ataxia, proprioceptive loss and blindness. The prevalence is <1 / 1.000.000.	600
FMR1	Fragile X syndrome	-	(CGG)n pre-mutated allele (Detection by PCR and TP-PCR)	Fragile X syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the FMR1 gene located on chromosomal region Xq27.3. The symptoms are variable depending on the range of CGG triplet expansion. In complete mutation the onset is infantile in men and is characterized by intellectual disability, characteristic appearance (large head, long face, prominent forehead and chin, protruding ears) joint laxity and large testes after puberty. In carrier female, the symptoms are milder and include primary ovarian insufficiency. The prevalence is 1/2,500 (full mutation allele) to 1/4,000 (prevalence of symptomatic cases) for both genders.	250,6
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	NM_003593.2	NM_003593.2:c.763C>T	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FOXN1 gene located on chromosomal region 17q11.2. The age of onset is infantile. This disease is characterized by T-cell immunodeficiency, the skin disorder congenital alopecia, and nail dystrophy. The prevalence is <1:1,000,000.	600

FRAS1	Fraser syndrome	NM_025074.6	NM_025074.6:c.7813C>T, NM_025074.6:c.832_835delTGTG, NM_025074.6:c.11159_11166delAGCTGGAG, NM_025074.6:c.776T>G, NM_025074.6:c.6991_6992insGG, NM_025074.6:c.6433C>T, NM_025074.6:c.3799C>T, NM_025074.6:c.1071+1_1071+4delGTGA, NM_025074.6:c.4969+1_4969+2insTAGC, NM_025074.6:c.5605_5606insT	Fraser syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the genes FRAS1 (located on chromosomal region 4q21.21) and FREM2 (located on chromosomal region 13q13.3). The age of onset is early infancy. Twenty-five per cent of affected infants are stillborn, while 20 % die before the age of 1 year. This disease is characterized mainly by cryptophthalmos and syndactyly, besides urinary and genital anormalities. The prevalence is <1:1,000,000.	250,6
FREM2	Fraser syndrome	NM_207361.5	NM_207361.5:c.2361_2362insC, NM_207361.5:c.8409+1G>A, NM_207361.5:c.5914G>A, NM_207361.5:c.5920G>A, NM_207361.5:c.3792_3795delTTAT	Fraser syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the genes FRAS1 (located on chromosomal region 4q21.21) and FREM2 (located on chromosomal region 13q13.3). The age of onset is early infancy. Twenty-five per cent of affected infants are stillborn, while 20 % die before the age of 1 year. This disease is characterized mainly by cryptophthalmos and syndactyly, besides urinary and genital anormalities. The prevalence is <1:1,000,000.	600
FUCA1	Fucosidosis	NM_000147.4	NM_000147.4:c.244C>T, NM_000147.4:c.1279C>T, NM_000147.4:c.856C>T, NM_000147.4:c.648C>A, NM_000147.4:c.1229T>G, NM_000147.4:c.433T>C	Fucosidosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FUCA1 gene located on chromosomal region 1p36.11. The age of onset is infantile. This disease is characterized by facial dysmorphism, dysostosis multiplex, moderate hepatomegaly, severe intellectual deficit, deafness, and according to age, angiokeratomas. The prevalence is <1:1,000,000.	600
FXN	Friedreich ataxia	NM_000144.4	NM_000144.4:c.389G>T, NM_000144.4:c.460A>T, NM_000144.4:c.385-2A>G, NM_000144.4:c.317T>G	Friedreich ataxia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the FXN gene located on chromosomal region 9q21.11. The age of onset is between age 10 and 15 years and usually before age 25 years. This disease is characterized by slowly progressive ataxia, dysarthria, muscle weakness, spasticity in the lower limbs, scoliosis, bladder dysfunction, absent lower limb reflexes, and loss of position and vibration sense. The prevalence is 2:100,000-4:100,000.	600
G6PC	Glycogen storage disease type 1a	NM_000151.3	NM_000151.3:c.508C>T, NM_000151.3:c.551G>A, NM_000151.3:c.447-1G>A, NM_000151.3:c.1039C>T, NM_000151.3:c.562G>C, NM_000151.3:c.380_381insTA, NM_000151.3:c.497T>G, NM_000151.3:c.247C>T, NM_000151.3:c.113A>T, NM_000151.3:c.229T>C, NM_000151.3:c.230+1G>C, NM_000151.3:c.47C>G, NM_000151.3:c.883C>T, NM_000151.3:c.370G>A, NM_000151.3:c.626A>G, NM_000151.3:c.248G>A	Glycogen storage disease type 1a follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the G6PC gene located on chromosomal region 17q21.31. The age of onset is infantile. This disease is characterized by poor tolerance to fasting, significant hepatomegaly and growth retardation. The incidence is 1/100,000.	250,6
G6PC3	Severe congenital neutropenia type 4	NM_138387.3	NM_138387.3:c.346A>G, NM_138387.3:c.141C>G, NM_138387.3:c.778G>C, NM_138387.3:c.758G>A, NM_138387.3:c.935_936insT, NM_138387.3:c.784G>C	Severe congenital neutropenia type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the G6PC3 gene located on chromosomal region 17q21.31. This disease is characterized by familial pulmonary arterial hypertension, cardiac abnormalities including atrial septal defect, leukopenia including intermittent neutropenia, lymphopenia, monocytosis, and anemia. The prevalence is 1:100,000.	600
GAA	Glycogen storage disease type 2	NM_000152.3	NM_000152.3:c.118C>T, NM_000152.3:c.1316T>A, NM_000152.3:c.1799G>A, NM_000152.3:c.1827_1828insA, NM_000152.3:c.1846_1847insA, NM_000152.3:c.1115A>T, NM_000152.3:c.1552-3C>G, NM_000152.3:c.1445C>T, NM_000152.3:c.2238G>C, NM_000152.3:c.1327-2A>G, NM_000152.3:c.1650dupG, NM_000152.3:c.2238G>A, NM_000152.3:c.307T>G, NM_000152.3:c.230_240delCAGTGCCCACA, NM_000152.3:c.2512C>T, NM_000152.3:c.1431delT, NM_000152.3:c.1561G>A, NM_000152.3:c.1465G>A, NM_000152.3:c.1548G>A, NM_000152.3:c.546G>A, NM_000152.3:c.1064T>C, NM_000152.3:c.877G>A, NM_000152.3:c.925G>A, NM_000152.3:c.768_769insT, NM_000152.3:c.2560C>T, NM_000152.3:c.655G>A, NM_000152.3:c.1408_1410delAAC, NM_000152.3:c.953T>C, NM_000152.3:c.1933G>T, NM_000152.3:c.1935C>A, NM_000152.3:c.1585_1586delTCinsGT, NM_000152.3:c.1927G>A, NM_000152.3:c.2041-1G>A, NM_000152.3:c.2066_2070dupAGCCG, NM_000152.3:c.2105G>T, NM_000152.3:c.2237G>A, NM_000152.3:c.525delT, NM_000152.3:c.546+1_546+4delGTGG, NM_000152.3:c.2544delC, NM_000152.3:c.1912G>T, NM_000152.3:c.1634C>T, NM_000152.3:c.710C>T, NM_000152.3:c.2015G>A, NM_000152.3:c.546G>C, NM_000152.3:c.2012T>G, NM_000152.3:c.853C>T, NM_000152.3:c.697delA	Glycogen storage disease type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GAA gene located on chromosomal region 17q25.3. There are two forms: adult and infantile.The age of onset in this last form is before the age of three months. This disease is characterized by severe hypotonia, hypertrophic cardiomyopathy and progressive hepatomegaly. The incidence is 1/57,000 for the adult form and 1/138,000 for infantile form.	250,6

GALC	Krabbe disease	NM_000153.3	NM_000153.3:c.1591C>T, NM_000153.3:c.1161+2T>G, NM_000153.3:c.1586C>T, NM_000153.3:c.1592G>A, NM_000153.3:c.1489+1_1489+2delGT, NM_000153.3:c.582+1G>A, NM_000153.3:c.388G>A, NM_000153.3:c.430delA, NM_000153.3:c.1695delT, NM_000153.3:c.1472delA, NM_000153.3:c.1004A>G, NM_000153.3:c.1153G>T, NM_000153.3:c.658C>T, NM_000153.3:c.1543G>A, NM_000153.3:c.332G>A, NM_000153.3:c.334A>G, NM_000153.3:c.205C>T, NM_000153.3:c.1796T>G, NM_000153.3:c.1814dupA, NM_000153.3:c.1700A>C, NM_000153.3:c.1723_1724insT, NM_000153.3:c.1964delC, NM_000153.3:c.236G>A, NM_000153.3:c.1488_1489+2delTGGT, NM_000153.3:c.453G>A, NM_000153.3:c.1488_1489delTG, NM_000153.3:c.628A>T, NM_000153.3:c.655C>T, NM_000153.3:c.953C>G, NM_000153.3:c.2056T>C	Krabbe disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GALC gene located on chromosomal region 14q31.3. There are two forms of the disease: infantile form (2-6 months onset) more severe and adult form less severe. It is a degenerative disorder that affects the nervous system characterized by a muscle stiffness, blindness, deafness, and eventually death. The incidence is 1/100,000-1/250,000 and the prevalence is 1/100,000.	250,6
GALT	Galactosemia	NM_000155.3	NM_000155.3:c.130G>A, NM_000155.3:c.132delG, NM_000155.3:c.118G>T, NM_000155.3:c.265T>G, NM_000155.3:c.289_291delAAC, NM_000155.3:c.1138T>C, NM_000155.3:c.113A>C, NM_000155.3:c.152G>A, NM_000155.3:c.1048delA, NM_000155.3:c.290A>G, NM_000155.3:c.221T>C, NM_000155.3:c.253-2A>G, NM_000155.3:c.425T>A, NM_000155.3:c.428C>T, NM_000155.3:c.442C>T, NM_000155.3:c.143G>C, NM_000155.3:c.443G>A, NM_000155.3:c.158G>A, NM_000155.3:c.18delC, NM_000155.3:c.199C>T, NM_000155.3:c.200G>A, NM_000155.3:c.203A>C, NM_000155.3:c.218_219delCT, NM_000155.3:c.512T>C, NM_000155.3:c.547C>A, NM_000155.3:c.552C>A, NM_000155.3:c.563A>G, NM_000155.3:c.565_578delGTATGGGCCAGCAG, NM_000155.3:c.568T>C, NM_000155.3:c.580T>C, NM_000155.3:c.584T>C, NM_000155.3:c.598delC, NM_000155.3:c.601C>T, NM_000155.3:c.602G>A, NM_000155.3:c.1030C>A, NM_000155.3:c.510C>A, NM_000155.3:c.617A>G, NM_000155.3:c.619C>T, NM_000155.3:c.626A>G, NM_000155.3:c.634C>T, NM_000155.3:c.688-2A>C, NM_000155.3:c.692G>A, NM_000155.3:c.292G>A, NM_000155.3:c.329-2A>C, NM_000155.3:c.367C>T, NM_000155.3:c.377+7A>C, NM_000155.3:c.386T>C, NM_000155.3:c.607G>A, NM_000155.3:c.610C>T, NM_000155.3:c.413C>T, NM_000155.3:c.416T>G, NM_000155.3:c.41delinsTT, NM_000155.3:c.904+1G>T, NM_000155.3:c.905-2A>G, NM_000155.3:c.907G>A, NM_000155.3:c.442G>A, NM_000155.3:c.947G>A, NM_000155.3:c.443G>C, NM_000155.3:c.445dupG, NM_000155.3:c.997C>G, NM_000155.3:c.997C>T, NM_000155.3:c.998G>A, NM_000155.3:c.793C>G, NM_000155.3:c.820+13A>G, NM_000155.3:c.1052delC, NM_000155.3:c.844C>G, NM_000155.3:c.855G>T, NM_000155.3:c.719_728delTAGTACTGGT, NM_000155.3:c.772C>T, NM_000155.3:c.939G>A, NM_000155.3:c.71_72insA, NM_000155.3:c.404C>T, NM_000155.3:c.508-1G>C, NM_000155.3:c.775C>T, NM_000155.3:c.400delT, NM_000155.3:c.502_504delGTG, NM_000155.3:c.957C>A, NM_000155.3:c.823C>G, NM_000155.3:c.505C>A, NM_000155.3:c.1006A>T, NM_000155.3:c.985T>C, NM_000155.3:c.790delC, NM_000155.3:c.790_792delinsTAG	Galactosemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GALT gene located on chromosomal region 9p13.3. The age of onset is neonatal. This disease is characterized by feeding difficulties, lethargy, and severe liver disease. Long-term complications appear including cognitive impairments, motor deficits, and ovarian dysfunction with reduced fertility in women and diminished bone density. The prevalence is 1/40,000-1/60,000.	250,6
GAMT	Guanidinoacetate methyltransferase deficiency	NM_000156.5	NM_000156.5:c.506G>A, NM_000156.5:c.590T>C	Guanidinoacetate methyltransferase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GAMT gene located on chromosomal region 19p13.3. The age of onset is infantile. This disease is characterized by intellectual disability, seizures and behavioral problems, often in conjunction with pyramidal and/or extrapyramidal manifestations with muscular hypotony. Biochemical symptoms are also included with high urinary excretion of guanidinoacetate, low urinary excretion of creatinine and creatine depletion in brain and muscles.	600
GAN	Giant axonal neuropathy	NM_022041.3	NM_022041.3:c.1447C>T, NM_022041.3:c.1456G>A, NM_022041.3:c.1684C>G, NM_022041.3:c.1429C>T, NM_022041.3:c.601C>T, NM_022041.3:c.413G>A, NM_022041.3:c.505G>A, NM_022041.3:c.1268T>C	Giant axonal neuropathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GAN gene located on chromosomal region 16q23.2. The age of onset is infantile. This disease is characterized by a progressive motor and sensitive peripheral and central nervous system neuropathy. Twenty families have been reported with this disease but the frequency is likely to be under-estimated.	250,6

GBA	Gaucher disease	NM_001005741.2	NM_001005741.2:c.1093G>A, NM_001005741.2:c.1090G>A, NM_001005741.2:c.1043C>T, NM_001005741.2:c.1274dupA, NM_001005741.2:c.1098dupA, NM_001005741.2:c.1085C>T, NM_001005741.2:c.1102C>T, NM_001005741.2:c.1049A>G, NM_001005741.2:c.1240G>T, NM_001005741.2:c.1246G>A, NM_001005741.2:c.1301G>C, NM_001005741.2:c.1088T>C, NM_001005741.2:c.1348T>A, NM_001005741.2:c.1361C>G, NM_001005741.2:c.1342G>C, NM_001005741.2:c.1448T>C, NM_001005741.2:c.1448T>G, NM_001005741.2:c.1504C>T, NM_001005741.2:c.1447_1466delCTGGACGCAGTGGCACTGATinsTG, NM_001005741.2:c.254G>A, NM_001005741.2:c.259C>T, NM_001005741.2:c.1053G>T, NM_001005741.2:c.160G>T, NM_001005741.2:c.431T>G, NM_001005741.2:c.475C>T, NM_001005741.2:c.476G>A, NM_001005741.2:c.481C>T, NM_001005741.2:c.487delG, NM_001005741.2:c.497A>T, NM_001005741.2:c.508C>T, NM_001005741.2:c.1141T>G, NM_001005741.2:c.115+1G>A, NM_001005741.2:c.1171G>C, NM_001005741.2:c.1174C>G, NM_001005741.2:c.354G>C, NM_001005741.2:c.1060G>C, NM_001005741.2:c.1208G>C, NM_001005741.2:c.1228C>G, NM_001005741.2:c.123A>G, NM_001005741.2:c.1240G>C, NM_001005741.2:c.914delC, NM_001005741.2:c.517A>C, NM_001005741.2:c.1295G>T, NM_001005741.2:c.1307T>C, NM_001005741.2:c.1265_1319del, NM_001005741.2:c.1319C>T, NM_001005741.2:c.1309G>T, NM_001005741.2:c.1226A>G, NM_001005741.2:c.407C>A, NM_001005741.2:c.1343A>T, NM_001005741.2:c.84_85insG, NM_001005741.2:c.518C>T, NM_001005741.2:c.1391A>C, NM_001005741.2:c.509G>T, NM_001005741.2:c.1604G>A, NM_001005741.2:c.84dupG, NM_001005741.2:c.535G>C, NM_001005741.2:c.586A>C, NM_001005741.2:c.1297G>T, NM_001005741.2:c.1184C>T, NM_001005741.2:c.1192C>T	Gaucher disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GBA gene located on chromosomal region 1q22. Gaucher disease encompasses a continuum of clinical findings from a perinatal lethal disorder to an asymptomatic type. There are three major clinical types (1, 2, and 3) and two other subtypes (perinatal-lethal and cardiovascular). Type 1 is characterized by the presence of clinical or radiographic evidence of bone disease, hepatosplenomegaly, anemia and thrombocytopenia, lung disease, and the absence of primary central nervous system disease. GD types 2 and 3 are characterized by the presence of primary neurologic disease. Type 2 has an onset before age two years, limited psychomotor development, and a rapidly progressive course with death by age two to four years. Type 3 may have onset before age two years, but often have a more slowly progressive course, with survival into the third or fourth decade. The perinatal-lethal form is associated with ichthyosiform or collodion skin abnormalities or with nonimmune hydrops fetalis. The cardiovascular form is characterized by calcification of the aortic and mitral valves, mild splenomegaly, corneal opacities, and supranuclear ophthalmoplegia. Cardiopulmonary complications have been described with all the clinical subtypes, although varying in frequency and severity. The incidence is 1/60,000 and the prevalence is approximately 1/100,000.	250,6
GBE1	Glycogen storage disease type 4	NM_000158.3	NM_000158.3:c.1571G>A, NM_000158.3:c.1570C>T, NM_000158.3:c.1774G>T, NM_000158.3:c.771T>A, NM_000158.3:c.1543C>T, NM_000158.3:c.1883A>G, NM_000158.3:c.2052+1G>A, NM_000158.3:c.986A>C, NM_000158.3:c.466_470delCGTAT, NM_000158.3:c.1604A>G	Glycogen storage disease type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GBE1 gene located on chromosomal region 3p12.2. The age of onset is infantile. This disease is characterized by failure to thrive; hepatomegaly, liver dysfunction, and progressive liver cirrhosis; hypotonia; cardiomyopathy and, finally, death.	250,6
GBE1	Polyglucosan body disease, adult	NM_000158.3	NM_000158.3:c.986A>G	Polyglucosan body disease, adult form follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GBE1 gene located on chromosomal region 3p12.2. The age of onset is late-onset, slowly progressive disorder affecting the central and peripheral nervous systems. This disease is characterized by a variable combination of cognitive impairment, pyramidal tetraparesis, peripheral neuropathy, and neurogenic bladder. Other manifestations include cerebellar dysfunction and extrapyramidal signs.	250,6
GCDH	Glutaric acidemia type 1	NM_000159.3	NM_000159.3:c.1093G>A, NM_000159.3:c.1060G>C, NM_000159.3:c.542A>G, NM_000159.3:c.442G>A, NM_000159.3:c.1199dupT, NM_000159.3:c.572T>C, NM_000159.3:c.1060G>A, NM_000159.3:c.1247C>T, NM_000159.3:c.74C>A, NM_000159.3:c.947C>A, NM_000159.3:c.1168G>C, NM_000159.3:c.416C>T, NM_000159.3:c.1198G>A, NM_000159.3:c.636-1G>A, NM_000159.3:c.1204C>T, NM_000159.3:c.1244-2A>C, NM_000159.3:c.751C>T, NM_000159.3:c.1262C>T, NM_000159.3:c.1148G>A, NM_000159.3:c.680G>C, NM_000159.3:c.883T>C, NM_000159.3:c.1015A>G, NM_000159.3:c.764C>T, NM_000159.3:c.271+1G>A, NM_000159.3:c.743C>T, NM_000159.3:c.877G>A, NM_000159.3:c.914C>T, NM_000159.3:c.1002_1003delGA, NM_000159.3:c.383G>A, NM_000159.3:c.769C>T	Glutaric acidemia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GCDH gene located on chromosomal region 19p13.2. The age of onset is infantile or neonatal. This disease is characterized by encephalopathic crises resulting in striatal injury and a severe dystonic dyskinetic movement disorder. The prevalence is 1 in 100,000 births.	250,6
GCSH	Glycine encephalopathy (GCSH)	NM_004483.4	NM_004483.4:c.337dupT	Glycine encephalopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GCSH gene located on chromosomal region 16q23.2. The age of onset is neonatal. This disease is characterized by lethargy or even coma, hypotonia, hiccups, myoclonic jerks, and breathing/swallowing disorders, with subsequent intellectual deficit, spasticity and intractable seizures.	600
GDAP1	Charcot-Marie-Tooth disease type 4A	NM_018972.2	NM_018972.2:c.358C>T, NM_018972.2:c.487C>T, NM_018972.2:c.311-1G>A, NM_018972.2:c.844C>T, NM_018972.2:c.715C>T, NM_018972.2:c.92G>A	Charcot-Marie-Tooth disease type 4A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GDAP1 gene located on chromosomal region 8q21.11. It is a severe, early-onset form of demyelinating CMT peripheral sensorimotor polyneuropathy characterized by severe motor retardation and progressive scoliosis. Vocal cord paresis may also occur.	600

GFM1	Combined oxidative phosphorylation deficiency type 1	NM_024996.5	NM_024996.5:c.1294_1297delACAG, NM_024996.5:c.748C>T, NM_024996.5:c.139C>T, NM_024996.5:c.1528_1529delAG, NM_024996.5:c.521A>G	Combined oxidative phosphorylation deficiency type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GFM1 gene located on chromosomal region 3q25.32. The age of onset is from early infancy until adult. This disease is characterized by ptosis, external ophthalmoplegia, proximal myopathy and exercise intolerance, cardiomyopathy, sensorineural deafness, optic atrophy, pigmentary retinopathy, and diabetes mellitus.	600
GJA1	Oculodentodigital dysplasia	NM_000165.4	NM_000165.4:c.227G>A, NM_000165.4:c.97C>T	Oculodentodigital dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GJA1 gene located on chromosomal region 6q22.31. The age of onset is infantile. This disease is characterized by craniofacial, neurologic, limb and ocular abnormalities.	600
GJB2	Deafness type 1A, autosomal recessive	NM_004004.5	NM_004004.5:c.176_191delGCTGCAAGAACGTGTG, NM_004004.5:c.169C>T, NM_004004.5:c.270dupA, NM_004004.5:c.239A>C, NM_004004.5:c.269T>C, NM_004004.5:c.427C>T, NM_004004.5:c.299_300delAT, NM_004004.5:c.250G>T, NM_004004.5:c.230G>A, NM_004004.5:c.516G>A, NM_004004.5:c.439G>A, NM_004004.5:c.465T>A, NM_004004.5:c.229T>C, NM_004004.5:c.241C>G, NM_004004.5:c.235delC, NM_004004.5:c.238C>T, NM_004004.5:c.557C>T, NM_004004.5:c.269_270insT, NM_004004.5:c.617A>G, NM_004004.5:c.231G>A, NM_004004.5:c.310_323delAGGAAGTTCATCAA, NM_004004.5:c.313_326delAAGTTCATCAAGGG, NM_004004.5:c.358_360delGAG, NM_004004.5:c.35delG, NM_004004.5:c.249C>G, NM_004004.5:c.334_335delAA, NM_004004.5:c.402delG, NM_004004.5:c.413G>A, NM_004004.5:c.416G>A, NM_004004.5:c.299A>T, NM_004004.5:c.250G>C, NM_004004.5:c.550C>T, NM_004004.5:c.551G>C, NM_004004.5:c.503A>G, NM_004004.5:c.227T>C, NM_004004.5:c.380G>A, NM_004004.5:c.132G>A, NM_004004.5:c.365A>T, NM_004004.5:c.139G>T	Deafness, autosomal recessive type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GJB2 and GJB3 genes located on chromosomal regions 13q12.11 and 1p34.3 respectively. The age of onset is infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment. No other associated medical findings are present.	250,6
GJB3	Deafness type 1A, autosomal recessive	NM_024009.2	NM_024009.2:c.529T>G, NM_024009.2:c.580G>A, NM_024009.2:c.94C>T	Deafness, autosomal recessive type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GJB2 and GJB3 genes located on chromosomal regions 13q12.11 and 1p34.3 respectively. The age of onset is infantile. This disease is characterized by congenital, non-progressive, mild-to-profound sensorineural hearing impairment. No other associated medical findings are present.	250,6
GJB6	Deafness type 1B, autosomal recessive	NM_006783.4	NM_006783.4:c.261dupA, NM_006783.4:c.169C>T, NM_006783.4:c.485dupA, NM_006783.4:c.689dupA, NM_006783.4:c.14C>T, NM_006783.4:c.443delC, NM_006783.4:c.383_384delTA, NM_006783.4:c.689_690insA	Nonsyndromic sensorineural deafness, autosomal recessive type DFNB1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GJB6 gene located on chromosomal region 13q12.11. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment. No other associated medical findings are present.	250,6
GJC2	Pelizaeus-Merzbacher-like disease type 1	NM_020435.3	NM_020435.3:c.857T>C, NM_020435.3:c.814T>G, NM_020435.3:c.613C>T, NM_020435.3:c.787G>A, NM_020435.3:c.718C>T, NM_020435.3:c.268C>T	Pelizaeus-Merzbacher-like disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GJC2 gene located on chromosomal region 1q42.13. It is an autosomal recessive leukodystrophy sharing identical clinical and radiological features as X-linked Pelizaeus-Merzbacher disease. The age of onset is early infantile. This disease is characterized by nystagmus, delayed motor milestones, ataxia, progressive spasticity, partial seizures, mild peripheral neuropathy, and diffuse leukodystrophy. The prevalence is <1.27:100,000.	600
GLB1	GM1 Gangliosidosis	NM_000404.2	NM_000404.2:c.1369C>T, NM_000404.2:c.1370G>A, NM_000404.2:c.1452C>G, NM_000404.2:c.176G>A, NM_000404.2:c.276G>A, NM_000404.2:c.1733A>G, NM_000404.2:c.1355dupA, NM_000404.2:c.442C>A, NM_000404.2:c.202C>T, NM_000404.2:c.591_592insT, NM_000404.2:c.622C>T, NM_000404.2:c.1549G>T, NM_000404.2:c.442C>T, NM_000404.2:c.457+2T>C, NM_000404.2:c.947A>G, NM_000404.2:c.438_440delTCT, NM_000404.2:c.601C>T, NM_000404.2:c.602G>A, NM_000404.2:c.1068+1G>T, NM_000404.2:c.1174_1175delCT, NM_000404.2:c.1004C>T, NM_000404.2:c.1051C>T, NM_000404.2:c.171C>G, NM_000404.2:c.1321G>A, NM_000404.2:c.1325G>A, NM_000404.2:c.818G>T, NM_000404.2:c.152T>C, NM_000404.2:c.1456_1466dupGGTGCATATAT, NM_000404.2:c.145C>T, NM_000404.2:c.175C>T, NM_000404.2:c.901G>A, NM_000404.2:c.1646C>T, NM_000404.2:c.1577dupG, NM_000404.2:c.1310A>T	Gangliosidosis GM1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GLB1 gene located on chromosomal region 3p22.3. Although the three types differ in severity, their features can overlap significantly. The age of onset in type 1 is infantile, in type 2 is late-infantile or juvenile and adult in type3. This disease is characterized by arrest/regression of neurological development, hypotonia, visceromegaly, macular cherry-red spots, dysostosis and coarse facial features. The prevalence is 1:100,000 a 200,000 newborn.	250,6

GLB1	Mucopolysaccharidosis type 4B	NM_000404.2	NM_000404.2:c.1444C>T, NM_000404.2:c.1313G>A, NM_000404.2:c.817T>C, NM_000404.2:c.1445G>A, NM_000404.2:c.1223A>C	Mucopolysaccharidosis type 4B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GLB1 gene located on chromosomal region 3p22.3. The age of onset is variable infantile/juvenile. In addition to skeletal involvement, significant morbidity can result from respiratory compromise, obstructive sleep apnea, valvular heart disease, hearing impairment, corneal clouding, and spinal cord compression. The prevalence is 1:200,000-1:300,000.	250,6
GLDC	Glycine encephalopathy	NM_000170.2	NM_000170.2:c.322G>T, NM_000170.2:c.1229G>A, NM_000170.2:c.1545G>C, NM_000170.2:c.1691G>T, NM_000170.2:c.1166C>T, NM_000170.2:c.2113G>A, NM_000170.2:c.2284G>A, NM_000170.2:c.1705G>A, NM_000170.2:c.2216G>A, NM_000170.2:c.2405C>T	Glycine encephalopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the AMT and GLDC genes located on chromosomal regions 3p21.31 and 9p24.1 respectively. The age of onset is neonatal/infantile. This disease is characterized by lethargy or even coma, hypotonia, hiccups, myoclonic jerks, and breathing/swallowing disorders, with subsequent intellectual deficit, spasticity and intractable seizures. The prevalence is 1:1,000,000-9:1,000,000.	250,6
GLE1	Lethal arthrogryposis with anterior horn cell disease	NM_001003722.1	NM_001003722.1:c.2051T>C, NM_001003722.1:c.1412_1413delAG, NM_001003722.1:c.898-2A>G, NM_001003722.1:c.2069_2072delTTCT, NM_001003722.1:c.1807C>T	Lethal arthrogryposis with anterior horn cell disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GLE1 gene located on chromosomal region 9q34.11. The age of onset is fetal. This disease is characterized by fetal akinesia, arthrogryposis and motor neuron loss. The fetus often survives delivery, but dies early as a result of respiratory failure. Neuropathological findings resemble those of lethal congenital contracture syndrome type 1, but are less severe.	250,6
GM2A	GM2 Gangliosidosis	NM_000405.4	NM_000405.4:c.285delC, NM_000405.4:c.160G>T, NM_000405.4:c.506G>C	GM2-gangliosidosis, AB variant follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GM2A gene located on chromosomal region 5q33.1. The age of onset is infantile. This disease is characterized by a group of neurodegenerative disorders: seizures, blindness, spasticity, eventual total incapacitation, and death. The prevalence is <1:100.000.	600
GNE	Distal myopathy Nonaka type	NM_005476.5	NM_005476.5:c.2116T>C, NM_005476.5:c.2135T>C, NM_005476.5:c.2086G>A, NM_005476.5:c.478C>T, NM_005476.5:c.1844C>G, NM_005476.5:c.737G>A, NM_005476.5:c.385C>T, NM_005476.5:c.1714G>T, NM_005476.5:c.1798G>A, NM_005476.5:c.2086G>T, NM_005476.5:c.787C>T, NM_005476.5:c.2023T>C, NM_005476.5:c.1993G>A, NM_005476.5:c.673G>A, NM_005476.5:c.909T>A, NM_005476.5:c.1727G>A	Distal myopathy, Nonaka type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GNE gene located on chromosomal region 9p13.3. The age of onset is adult. This disease is characterized by progressive muscle weakness and joint deformity. The prevalence is 1:500-1:1,000.	250,6
GNPTAB	Mucolipidosis type 2/type 3	NM_024312.4	NM_024312.4:c.1931C>T, NM_024312.4:c.1799delC, NM_024312.4:c.3503_3504delTC, NM_024312.4:c.3173C>G, NM_024312.4:c.25C>T, NM_024312.4:c.3663delG, NM_024312.4:c.1906dupA, NM_024312.4:c.2383delG, NM_024312.4:c.732_733delAA, NM_024312.4:c.749dupA, NM_024312.4:c.2896delA, NM_024312.4:c.648_651delAGAA, NM_024312.4:c.3326dupA, NM_024312.4:c.3410T>A, NM_024312.4:c.10A>C, NM_024312.4:c.1000C>T, NM_024312.4:c.1196C>T, NM_024312.4:c.1759C>T, NM_024312.4:c.3565C>T, NM_024312.4:c.616_619delACAG, NM_024312.4:c.99delC, NM_024312.4:c.3598G>A, NM_024312.4:c.3560_3561delAG	Mucolipidosis type 2/type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GNPTAB gene located on chromosomal region 12q23.2. The age of onset is infantile. This disease is characterized by growth retardation, skeletal abnormalities, facial dysmorphism, stiff skin, developmental delay and cardiomegaly and that is lethal in childhood. The prevalence is 1:123,500-1:625,500.	250,6
GNS	Mucopolysaccharidosis type 3D	NM_002076.3	NM_002076.3:c.1063C>T, NM_002076.3:c.1226dupG, NM_002076.3:c.1169delA, NM_002076.3:c.1168C>T, NM_002076.3:c.413C>G	Mucolipidosis type 3D (Sanfilippo disease) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GNS gene located on chromosomal region 12q14.3. The age of onset is infantile. This disease is characterized by joint stiffness and pain initially in the shoulders, hips, and fingers; and gradual mild coarsening of facial features, cardiorespiratory complications and mild cognitive impairment. The incidence is 1:70,000 newborn.	600
GPR143	Ocular albinism, X-linked	NM_000273.2	NM_000273.2:c.992_993insCG, NM_000273.2:c.695C>A	X-linked recessive ocular albinism follows an X-linked pattern of inheritance and is caused by pathogenic variants in the GPR143 gene located on chromosomal region Xp22.2. The age of onset is infantile. This disease is characterized by ocular hypopigmentation, foveal hypoplasia, nystagmus, photodysphoria, and reduced visual acuity. The prevalence is 1/60,000 to 1/150,000 live male births.	600
GPR179	Night blindness, congenital stationary type 1E	NM_001004334.3	NM_001004334.3:c.1784+1G>A, NM_001004334.3:c.1368delT, NM_001004334.3:c.3656_3657delCT, NM_001004334.3:c.6847_6848delCT, NM_001004334.3:c.984delC, NM_001004334.3:c.1807C>T, NM_001004334.3:c.278_279insC, NM_001004334.3:c.5693_5694insT, NM_001004334.3:c.278delC, NM_001004334.3:c.1236G>A, NM_001004334.3:c.376G>C, NM_001004334.3:c.3233_3234delCT, NM_001004334.3:c.5763_5764delGA, NM_001004334.3:c.839_842delATCA, NM_001004334.3:c.4699_4700delAG	Congenital stationary night blindness type 1E follow an autosomal recessive, dominant or X-linked pattern of inheritance and is caused by pathogenic variants in the GPR179 gene located on chromosomal region 17q12. The age of onset is infantile. This disease is characterized by hemeralopia with a moderate loss of visual acuity.	250,6

GPR98	Usher syndrome type 2C	NM_032119.3	NM_032119.3:c.11377G>T, NM_032119.3:c.8713_8716dupAACA, NM_032119.3:c.2864C>A, NM_032119.3:c.18131A>G, NM_032119.3:c.2258_2270delAAGTGCTGAAATC, NM_032119.3:c.6275-1G>A, NM_032119.3:c.2636C>T, NM_032119.3:c.14973-1G>C, NM_032119.3:c.17668_17669delAT, NM_032119.3:c.5357_5358delAA, NM_032119.3:c.5747C>T, NM_032119.3:c.15196_15199dupCAAA, NM_032119.3:c.3151G>T, NM_032119.3:c.6901C>T, NM_032119.3:c.8790delC, NM_032119.3:c.5830G>A, NM_032119.3:c.6311_6312insT	Usher syndrome type 2C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GPR98 and PDZD7 genes located on chromosomal regions 5q14.3 and 10q24.32 respectively. The age of onset is infantile. This disease is characterized by the association of sensorineural prelingual deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. The prevalence is 1/30,000.	250,6
GRHPR	Primary hyperoxaluria type 2	NM_012203.1	NM_012203.1:c.103delG, NM_012203.1:c.295C>T, NM_012203.1:c.755dupA, NM_012203.1:c.622C>T, NM_012203.1:c.435G>A	Primary hyperoxaluria type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GRHPR gene located on chromosomal region 9p13.2. The age of onset is infantile. This disease is characterized by recurrent nephrolithiasis, nephrocalcinosis and end-stage renal disease with subsequent systemic oxalosis.	600
GRM6	Night blindness, congenital stationary type 1B	NM_000843.3	NM_000843.3:c.2341G>A, NM_000843.3:c.727_728insG, NM_000843.3:c.2213_2219delCCAGAGG, NM_000843.3:c.1861C>T, NM_000843.3:c.2560C>T, NM_000843.3:c.712C>T, NM_000843.3:c.2122C>T, NM_000843.3:c.719_720insG, NM_000843.3:c.1214T>C, NM_000843.3:c.1336C>T, NM_000843.3:c.1258C>T, NM_000843.3:c.1565G>A	Congenital stationary night blindness type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GRM6 gene located on chromosomal region 5q35.3. The age of onset is early infancy. This disease is characterized by hemeralopia with a moderate loss of visual acuity.	250,6
GRXCR1	Deafness type 25, autosomal recessive	NM_001080476.2	NM_001080476.2:c.229C>T, NM_001080476.2:c.190G>A, NM_001080476.2:c.710_711delAT	Autosomal recessive nonsyndromic sensorineural deafness type DFNB25 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GRXCR1 gene located on chromosomal region 4p13. The age of onset is infantile. This disease is characterized by hearing loss which is not associated visible abnormalities of the external ear or any related medical problems.	600
GSS	Glutathione synthetase deficiency	NM_000178.2	NM_000178.2:c.656A>G, NM_000178.2:c.847C>T, NM_000178.2:c.754C>T, NM_000178.2:c.799C>T, NM_000178.2:c.4delG, NM_000178.2:c.656A>C, NM_000178.2:c.491G>A, NM_000178.2:c.832C>T	Glutathione synthetase deficiency with 5-oxoprolinuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GSS gene located on chromosomal region 20q11.22. The severity and age of onset are variable. This disease is characterized by affectation of the neutrophil respiratory burst and can increase host susceptibility to infections, is associated with hemolytic anemia and intellectual disability. The prevalence is <1:1,000,000.	600
GUCY2D	Leber congenital amaurosis type 1	NM_000180.3	NM_000180.3:c.1694T>C, NM_000180.3:c.2735_2736delTT, NM_000180.3:c.456C>A, NM_000180.3:c.622delC, NM_000180.3:c.2734_2735delTT, NM_000180.3:c.2945delG	Leber congenital amaurosis type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GUCY2D gene located on chromosomal region 17p13.1. The age of onset is infantile. This disease is characterized by blindness, nystagmus, roving eye movement and severe visual impairment.	600
GUSB	Mucopolysaccharidosis type 7	NM_000181.3	NM_000181.3:c.1065+1G>T, NM_000181.3:c.1084G>A, NM_000181.3:c.1144C>T, NM_000181.3:c.1337G>A, NM_000181.3:c.1222C>T, NM_000181.3:c.1730G>T, NM_000181.3:c.1831C>T, NM_000181.3:c.1856C>T, NM_000181.3:c.1881G>T, NM_000181.3:c.442C>T, NM_000181.3:c.499C>T, NM_000181.3:c.526C>T, NM_000181.3:c.646C>T, NM_000181.3:c.820_821delAC, NM_000181.3:c.1061C>T, NM_000181.3:c.1050G>C, NM_000181.3:c.1534G>A, NM_000181.3:c.1244C>T, NM_000181.3:c.1219_1220insC, NM_000181.3:c.866G>A, NM_000181.3:c.1244+1G>A, NM_000181.3:c.1521G>A, NM_000181.3:c.1429C>T, NM_000181.3:c.1618G>T, NM_000181.3:c.1338G>A	Mucopolysaccharidosis type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GUSB gene located on chromosomal region 7q11.21. The age of onset is variable. There are prenatal forms with non-immune hydrops fetalis, and severe neonatal forms with dysmorphism, hernias, hepatosplenomegaly, club feet, dysostosis, severe hypotonia and neurological disorders that ultimately lead to profound intellectual deficit and small stature in patients that survive. Finally, there are also very mild cases that are discovered during adolescence or adulthood following presentation with thoracic kyphosis. The prevalence is 1:250,000 in newborn.	250,6
HADHA	Trifunctional protein deficiency	NM_000182.4	NM_000182.4:c.1918C>T, NM_000182.4:c.274_278delTCATC, NM_000182.4:c.2131C>A, NM_000182.4:c.1793_1794delAT, NM_000182.4:c.1620+2_1620+6delTAAGG, NM_000182.4:c.2027G>A, NM_000182.4:c.1678C>T, NM_000182.4:c.2132_2133insC, NM_000182.4:c.2146+1G>A, NM_000182.4:c.919-2A>G, NM_000182.4:c.1644delC, NM_000182.4:c.1132C>T, NM_000182.4:c.1528G>C, NM_000182.4:c.499delA, NM_000182.4:c.845T>A, NM_000182.4:c.1422dupT	Mitochondrial trifunctional protein deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HADHA and HADHB genes located on chromosomal region 2p23.3. The age of onset is neonatal/infancy. It is characterized by a wide clinical spectrum ranging from severe neonatal manifestations including cardiomyopathy, hypoglycemia, metabolic acidosis, skeletal myopathy and neuropathy, liver disease and death to a mild phenotype with peripheral polyneuropathy, episodic rhabdomyolysis and pigmentary retinopathy. The prevalence is <1 / 1,000,000.	250,6
HADHB	Trifunctional protein deficiency	NM_000183.2	NM_000183.2:c.788A>G, NM_000183.2:c.1364T>G, NM_000183.2:c.1331G>A	Mitochondrial trifunctional protein deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HADHA and HADHB genes located on chromosomal region 2p23.3. The age of onset is neonatal/infancy. It is characterized by a wide clinical spectrum ranging from severe neonatal manifestations including cardiomyopathy, hypoglycemia, metabolic acidosis, skeletal myopathy and neuropathy, liver disease and death to a mild phenotype with peripheral polyneuropathy, episodic rhabdomyolysis and pigmentary retinopathy. The prevalence is <1 / 1,000,000.	600

HAL	Histidinemia	NM_002108.3	NM_002108.3:c.890_891insT, NM_002108.3:c.146_152delATGACGC, NM_002108.3:c.1287+2T>C, NM_002108.3:c.102_103insGC, NM_002108.3:c.903+1G>A	Histidinemia follows a pattern of inheritance and is caused by pathogenic variants in the HAL gene located on chromosomal region 12q23.1. The age of onset is infantile ans is characterized by high concentration of Histidine in blood and urine. This disease seems to be benign in most of cases but 2/3 of the patients could show mild development delay.	600
HBA	Alpha-thalassemia	-	--MED, --SEA, --THAI, -Î±4.2, -Î±3.7, --FIL, -(Î±)20.5 (Detection by PCR)	Alpha-thalassemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HBA gene located on chromosomal region 16p13.3. The age of onset is infantile. It is characterized by impaired synthesis of alpha-globin chains leading to a variable clinical picture depending on the number of affected alleles. The disease can be classified into clinical subtypes of increasing severity: silent alpha thalassemia, alpha thalassemia trait (or alpha thalassemia minor), hemoglobin H disease (HbH), and Hb Bart's hydrops fetalis (see these terms). A rare form called alpha-thalassemia-intellectual deficit syndrome has also been identified (see these terms). Alpha thalassemia trait causes microcytosis and hypochromia with absent or mild anemia (often detected on routine blood tests), generally with no other symptoms. HbH patients develop moderate hemolytic anemia with variable amounts of HbH along with occasionally severe splenomegaly, sometimes complicated by hypersplenism. Hb Bart's hydrops fetalis involves a severe deficiency in alpha-globin with serious developmental implications. Alpha-thalassemia-intellectual deficit syndrome is characterized by very mild to severe anemia associated with developmental abnormalities. The prevalence is 1:10,000-5:10,000.	600
HBB	Beta-thalassemia	NM_000518.4	NM_000518.4:c.135delC, NM_000518.4:c.118C>T, NM_000518.4:c.217dupA, NM_000518.4:c.92+5G>C, NM_000518.4:c.208G>A, NM_000518.4:c.85_86insC, NM_000518.4:c.92+5G>A, NM_000518.4:c.27dupG, NM_000518.4:c.126_129delCTTT, NM_000518.4:c.93-23T>C, NM_000518.4:c.92+1G>A, NM_000518.4:c.-50-u32C>T, NM_000518.4:c.82G>T, NM_000518.4:c.315+1G>A, NM_000518.4:c.52A>T, NM_000518.4:c.380T>A, NM_000518.4:c.93-21G>A, NM_000518.4:c.79G>A, NM_000518.4:c.112delT, NM_000518.4:c.92+6T>C, NM_000518.4:c.59A>G, NM_000518.4:c.364G>A	Beta-thalassemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HBB gene located on chromosomal region 11p15.4. The age of onset is infantile. Three main types of BT have been described, thalassemia minor is usually asymptomatic, thalassemia major is associated with splenomegaly and microcytic and hypochromic anemia and thalassemia intermedia, in which the anemia is less severe. The incidence is 1/100,000.	250,6
HBB	Sickle cell anaemia	NM_000518.4	NM_000518.4:c.19G>A, NM_000518.4:c.20A>T	Sickle cell anemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HBB gene located on chromosomal region 11p15.4. The age of onset is infantile. This disease is characterized by chronic severe anemia, bacterial infections, and ischemic vaso-occlusive accidents. This results in tissue ischemia leading to acute and chronic pain as well as organ damage that can affect any organ in the body, including the bones, lungs, liver, kidneys, brain, eyes, and joints. The highest frequency of sickle cell disease is found in tropical regions, particularly sub-Saharan Africa, India and the Middle-East.	250,6
HESX1	Combined pituitary hormone deficiencies, genetic forms	NM_003865.2	NM_003865.2:c.374A>G, NM_003865.2:c.77T>C, NM_003865.2:c.445G>A, NM_003865.2:c.450_451delCA, NM_003865.2:c.18G>C	Combined pituitary hormone deficiencies, genetic forms follow an autosomal recessive pattern of inheritance and are caused by pathogenic variants in the HESX1 gene located on chromosomal region 3p14.3. The age of onset is infantile. These diseases are characterized by short stature, cognitive alterations or delayed puberty. The incidence is 1:3,000 and 1:4,000 births.	250,6

HEXA	Tay-Sachs disease	NM_000520.4	NM_000520.4:c.1176G>A, NM_000520.4:c.1495C>T, NM_000520.4:c.1177C>T, NM_000520.4:c.116T>G, NM_000520.4:c.1510delC, NM_000520.4:c.1496G>A, NM_000520.4:c.1260G>C, NM_000520.4:c.1351C>G, NM_000520.4:c.1511G>A, NM_000520.4:c.1499delT, NM_000520.4:c.1510C>T, NM_000520.4:c.380T>G, NM_000520.4:c.459+5G>A, NM_000520.4:c.508C>T, NM_000520.4:c.509G>A, NM_000520.4:c.532C>T, NM_000520.4:c.533G>A, NM_000520.4:c.533G>T, NM_000520.4:c.1528C>T, NM_000520.4:c.173G>A, NM_000520.4:c.1A>G, NM_000520.4:c.1A>T, NM_000520.4:c.1444G>A, NM_000520.4:c.1453T>C, NM_000520.4:c.739C>T, NM_000520.4:c.745C>T, NM_000520.4:c.749G>A, NM_000520.4:c.759_774dupGCTTGCAGAGTTTGAC, NM_000520.4:c.772G>C, NM_000520.4:c.1214_1215delinsG, NM_000520.4:c.78G>A, NM_000520.4:c.538T>C, NM_000520.4:c.540C>G, NM_000520.4:c.805G>A, NM_000520.4:c.915_917delCTT, NM_000520.4:c.254-1G>C, NM_000520.4:c.2T>C, NM_000520.4:c.1537C>T, NM_000520.4:c.1490A>G, NM_000520.4:c.77G>A, NM_000520.4:c.1422G>C, NM_000520.4:c.805+1G>A, NM_000520.4:c.805+1G>C, NM_000520.4:c.672+1G>A, NM_000520.4:c.629C>T, NM_000520.4:c.987G>A, NM_000520.4:c.632T>C, NM_000520.4:c.1278_1279insTATC, NM_000520.4:c.1274_1277dupTATC, NM_000520.4:c.986+3A>G, NM_000520.4:c.611A>G, NM_000520.4:c.1277_1278insTAT	Tay-Sachs disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HEXA gene located on chromosomal region 15q23. The age of onset is infantile. There are three forms, type 1 (infantile), with a psychomotor retardation which is associated with hypotonia, amaurosis and megalencephaly. Type 2 is characterized by locomotor ataxia, behavioural disorders, and progressive loss of intellectual capacities. Type three (chronic form) shows spinocerebellar ataxia or spinal amyotrophy. The prevalence is 1 case per 320 000 live births.	250,6
HEXB	Sandhoff disease	NM_000521.3	NM_000521.3:c.1310_1311delCA, NM_000521.3:c.1380G>A, NM_000521.3:c.1367A>C, NM_000521.3:c.1238_1242delCAAAG, NM_000521.3:c.298delC, NM_000521.3:c.1345delT, NM_000521.3:c.797A>G, NM_000521.3:c.1539_1540delCT, NM_000521.3:c.1375G>T, NM_000521.3:c.508C>T, NM_000521.3:c.1517_1529dupCAAGTGCTGTTGG, NM_000521.3:c.841C>T, NM_000521.3:c.202_203insGG, NM_000521.3:c.1250C>T, NM_000521.3:c.1619_1620insTTCATGTTATCTACAGACGTG, NM_000521.3:c.1537_1538delCT, NM_000521.3:c.170delG, NM_000521.3:c.115delG, NM_000521.3:c.171delG, NM_000521.3:c.850C>T	Sandhoff disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HEXB gene located on chromosomal region 5q13.3. The age of onset is adult or infantile. This disease is characterized by central nervous system degeneration, with startle reactions, early blindness, progressive motor and mental deterioration, macrocephaly and cherry-red spots on the macula. The prevalence is 1/130.000.	250,6
HFE	Haemochromatosis	NM_000410.3	NM_000410.3:c.18G>C, NM_000410.3:c.252G>A, NM_000410.3:c.989G>T, NM_000410.3:c.314T>C, NM_000410.3:c.193A>T, NM_000410.3:c.829G>A, NM_000410.3:c.277G>C	Hemochromatosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HFE gene located on chromosomal region 6p22.2. The age of onset is adult. This disease is characterized by chronic fatigue, bronzed skin pigmentation and tissue damage in the liver, pancreas, joints, bone, endocrine glands, heart. The prevalence is 1/200 - 1/1.000.	250,6
HGD	Alkaptonuria	NM_000187.3	NM_000187.3:c.140C>T, NM_000187.3:c.16-1G>A, NM_000187.3:c.342+1G>A, NM_000187.3:c.1111_1112insC, NM_000187.3:c.899T>G, NM_000187.3:c.1189-2A>G, NM_000187.3:c.674G>A, NM_000187.3:c.175delA, NM_000187.3:c.283-5delT, NM_000187.3:c.172A>T, NM_000187.3:c.873C>A, NM_000187.3:c.283-4C>T, NM_000187.3:c.808G>A, NM_000187.3:c.1102A>G, NM_000187.3:c.469+2T>C, NM_000187.3:c.688C>T, NM_000187.3:c.481G>A	Alkaptonuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HGD gene located on chromosomal region 3q13.33. The age of onset is infantile. This disease is characterized by darkening of the urine when it is left exposed to air, grey-blue colouration of the eye sclerae and the ear helix (ochronosis), and a disabling joint disease involving both the axial and peripheral joints (ochronotic arthropathy). The prevalence is 1:250,000-1:1.000.000 newborn.	250,6
HGF	Deafness type 39, autosomal recessive	NM_000601.4	NM_000601.4:c.2028delA, NM_000601.4:c.1597C>T	Autosomal recessive nonsyndromic sensorineural deafness type DFNB239 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HGF gene located on chromosomal region 7q21.11. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	600
HGSNAT	Mucopolysaccharidosis type 3C	NM_152419.2	NM_152419.2:c.1378-1G>A, NM_152419.2:c.1843G>A, NM_152419.2:c.607C>T, NM_152419.2:c.1250+1G>A, NM_152419.2:c.848C>T, NM_152419.2:c.1464+1G>A, NM_152419.2:c.1501delA, NM_152419.2:c.1030C>T, NM_152419.2:c.1503delA, NM_152419.2:c.1553C>T, NM_152419.2:c.1622C>T, NM_152419.2:c.493+1G>A	Mucopolysaccharidosis type 3C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HGSNAT gene located on chromosomal region 8p11.21. The age of onset is infantile. This disease is characterized by defective or missing enzymes to break down mucopolysaccharides are missing or are defective. The prevalence is <1:70.000 newborn.	250,6
HIBCH	3-Hydroxyisobutryl-CoA hydrolase deficiency	NM_014362.3	NM_014362.3:c.1012A>T, NM_014362.3:c.79-3C>G, NM_014362.3:c.494_495delTT, NM_014362.3:c.365A>G, NM_014362.3:c.220-9T>G	3-Hydroxyisobutryl-CoA hydrolase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HIBCH gene located on chromosomal region 2q32.2. The age of onset is infantile. This disease is characterized by delayed motor development, hypotonia and progressive neurodegeneration. The prevalence is <1:1,000,000.	600

HMGCL	3-hydroxy-3-methylglutaric aciduria	NM_000191.2	NM_000191.2:c.835G>A, NM_000191.2:c.230delT, NM_000191.2:c.122G>A, NM_000191.2:c.698A>G, NM_000191.2:c.206_207delCT, NM_000191.2:c.505_506delTC	3-hydroxy-3-methylglutaric aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HMGCL gene located on chromosomal region 1p36.11. The age of onset is infantile. This disease is is an organic aciduria, due to deficiency of 3-hydroxy-3-methylglutaryl-CoA-lyase (a key enzyme in ketogenesis and leucine metabolism) usually presenting in infancy with episodes of metabolic decompensation triggered by periods of fasting or infections, which when left untreated are life-threatening and may lead to neurological sequelae.	600
HPD	Tyrosinemia type 3	NM_002150.2	NM_002150.2:c.600C>G, NM_002150.2:c.774T>G, NM_002150.2:c.1005C>G, NM_002150.2:c.987delA	Tyrosinemia type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HPD gene located on chromosomal region 12q24.31. The age of onset is infantile. This disease is characterized by intellectual deficit and ataxia. The prevalence is 1:100,000-1:120,000 newborn.	250,6
HPRT1	Lesch-Nyhan syndrome	NM_000194.2	NM_000194.2:c.486-1G>A, NM_000194.2:c.508C>T, NM_000194.2:c.610-2A>G, NM_000194.2:c.532+2T>G	Lesch-Nyhan syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the HPRT1 gene located on chromosomal region Xq26.2-q26.3. The age of onset is infantile. This disease is characterized by acid overproduction, neurological troubles, and behavioral problems. The prevalence is 1:380,000.	600
HPS1	Hermansky-Pudlak syndrome type 1	NM_000195.3	NM_000195.3:c.972_973insC, NM_000195.3:c.972delC, NM_000195.3:c.1996G>T, NM_000195.3:c.398+5G>A, NM_000195.3:c.1472_1487dupCCAGCAGGGGAGGCCC, NM_000195.3:c.397G>T	Hermansky-Pudlak syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HPS1 gene located on chromosomal region 10q24.2. The age of onset is early childhood. This disease is characterized by oculocutaneous albinism, bleeding diathesis and, in some cases, neutropenia, pulmonary fibrosis, or granulomatous colitis. The prevalence is 1/500,000 - 1/1,000,000.	600
HSD17B4	D-bifunctional protein deficiency	NM_000414.3	NM_000414.3:c.1369A>T, NM_000414.3:c.46G>A, NM_000414.3:c.972+1G>T, NM_000414.3:c.317G>C	Bifunctional enzyme deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HSD17B4 gene located on chromosomal region 5q23. The age of onset is juvenile. This disease is characterized by slowly progressive cerebellar atrophy and ataxia, intellectual decline, hearing loss, hypogonadism, hyperreflexia, a demyelinating sensorimotor neuropathy.	600
HSD17B4	Perrault syndrome	NM_000414.3	NM_000414.3:c.650A>G	Perrault syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HSD17B4 gene located on chromosomal region 5q23.1. The age of onset is adult. This disease is characterized by ovarian dysgenesis in females with sensorineural hearing impairment and other neurological alterations. The prevalence is <1:1,000,000.	600
HSPD1	Leukodystrophy hypomyelinating type 4	NM_002156.4	NM_002156.4:c.1381C>G, NM_002156.4:c.292G>A	Leukodystrophy hypomyelinating type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HSPD1 gene located on chromosomal region 2q33.1. The age of onset is infantile. A severe autosomal recessive hypomyelinating leukodystrophy. Clinically characterized by infantile-onset rotary nystagmus, progressive spastic paraplegia, neurologic regression, motor impairment, profound mental retardation. Death usually occurs within the first two decades of life.	600
HSPG2	Schwartz-Jampel syndrome type 1	NM_005529.6	NM_005529.6:c.13075delC, NM_005529.6:c.1653_1654insT, NM_005529.6:c.10355G>A, NM_005529.6:c.1125C>A, NM_005529.6:c.9326delA, NM_005529.6:c.8464+4A>G	Schwartz-Jampel syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HSPG2 gene located on chromosomal region 1p36.12. The age of onset is infantile. This disease is characterized by myotonia and osteoarticular abnormalities. The prevalence is <1:1,000,000.	600
HTRA1	CARASIL syndrome	NM_002775.4	NM_002775.4:c.1108C>T, NM_002775.4:c.883G>A, NM_002775.4:c.904C>T, NM_002775.4:c.754G>A, NM_002775.4:c.889G>A	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL syndrome) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HTRA1 gene located on chromosomal region 10q26.13. The age of onset is adult. This disease is characterized by early-onset gait disturbances, premature scalp alopecia, ischemic stroke, acute mid to lower back pain and progressive cognitive disturbances leading to severe dementia. About 50 people diagnosed, mainly in Japan and China.	600
HYLS1	Hydrolethalus syndrome type 1	NM_145014.2	NM_145014.2:c.632A>G, NM_145014.2:c.724C>T, NM_145014.2:c.669G>A	Hydrolethalus syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HYLS1 gene located on chromosomal region 11q24.2. The age of onset is fetal. This disease is characterized by craniofacial dysmorphic features, central nervous system, cardiac, respiratory tract and limb abnormalities. The incidence is 1/20,000 in Finland and the prevalence is <1:1,000,000.	600
IDH3B	Retinitis pigmentosa type 46	NM_006899.3	NM_006899.3:c.395T>C, NM_006899.3:c.490C>T, NM_006899.3:c.589delA	Retinitis pigmentosa 46 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IDH3B gene located on chromosomal region 20p13. The age of onset is variable. This disease is characterized by night blindness, followed by a progressive loss of peripheral vision in the daylight period and leading to blindness. The prevalence is 1/3,000 to 1/5,000.	600

IDS	Mucopolysaccharidosis type 2	NM_000202.6	NM_000202.6:c.1464G>T, NM_000202.6:c.1466G>C, NM_000202.6:c.1505G>C, NM_000202.6:c.283A>G, NM_000202.6:c.208dupC, NM_000202.6:c.596_599delAACA, NM_000202.6:c.597delA, NM_000202.6:c.683C>T, NM_000202.6:c.1148delC, NM_000202.6:c.880-8A>G, NM_000202.6:c.937C>T, NM_000202.6:c.998C>T, NM_000202.6:c.690_691insT, NM_000202.6:c.1122C>T, NM_000202.6:c.587T>C, NM_000202.6:c.314_317dupTCAA, NM_000202.6:c.278delC, NM_000202.6:c.514C>T, NM_000202.6:c.1508T>A, NM_000202.6:c.388_389insG, NM_000202.6:c.240+1G>A, NM_000202.6:c.404A>G	Mucopolysaccharidosis type 2 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the IDS gene located on chromosomal region Xq28. The age of onset is infantile. This disease is characterized by distinctive coarse facial features, short stature, cardio-respiratory involvement and skeletal abnormalities. The prevalence is 1:100,000-1:170,000 mannewborn.	600
IFT80	Short-rib thoracic dysplasia type 2 with or without polydactyly	NM_020800.2	NM_020800.2:c.701C>G, NM_020800.2:c.721G>C, NM_020800.2:c.315C>G	Short-rib thoracic dysplasia type 2 with or without polydactyly an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IFT80 gene located on chromosomal region 3q25.33. The age of onset is antenatal/neonatal. This is, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a trident appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. The incidence is 1-5/500,000.	600
IGF1	Growth delay due to insulin-like growth factor type 1 deficiency	NM_000618.3	NM_000618.3:c.274G>A	Growth delay due to insulin-like growth factor type 1 deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IGF1 gene located on chromosomal region 12q23.2. The age of onset is infantile. This disease is characterized by the association of intrauterine and postnatal growth retardation with sensorineural deafness and intellectual deficit, addition clinical features include microcephaly, adiposity, and insulin resistance. The prevalence is <1:1,000,000.	600
IGHMBP2	Spinal muscular atrophy, distal, type 1, autosomal recessive	NM_002180.2	NM_002180.2:c.1488C>A, NM_002180.2:c.2611+1G>T, NM_002180.2:c.1540G>A, NM_002180.2:c.1738G>A, NM_002180.2:c.661delA, NM_002180.2:c.121C>T, NM_002180.2:c.1101_1116delCTACTTCGACGTGGTG, NM_002180.2:c.2922T>G, NM_002180.2:c.1107C>G, NM_002180.2:c.2362C>T, NM_002180.2:c.638A>G	Autosomal recessive distal spinal muscular atrophy type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IGHMBP2 gene located on chromosomal region 11q13.3. The age of onset is infantile. This disease is characterized by neuromuscular disorder characterized by progressive weakness and atrophy of the diaphragm and skeletal muscles, leading to death in childhood. The prevalence is 4:100,000-10:100,000.	250,6
IKBKAP	Familial dysautonomia	NM_003640.3	NM_003640.3:c.3332delC, NM_003640.3:c.1460+2T>C, NM_003640.3:c.2204+6T>C, NM_003640.3:c.2328delT, NM_003640.3:c.2741C>T, NM_003640.3:c.2087G>C, NM_003640.3:c.2087G>A	Familial dysautonomia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IKBKAP gene located on chromosomal region 9q31.3. The age of onset is infantile. This disease is characterized by sensory dysfunction and severe impairment of the autonomic nervous system activity, resulting in multisystem dysfunction. The prevalence is <1:1,000,000	600
IL2RG	Severe combined immunodeficiency T-B+; X-linked	NM_000206.2	NM_000206.2:c.454+1G>A, NM_000206.2:c.452T>C, NM_000206.2:c.186T>A, NM_000206.2:c.664C>T, NM_000206.2:c.343T>C, NM_000206.2:c.854G>A, NM_000206.2:c.341G>A, NM_000206.2:c.355A>T	T-B+ severe combined immunodeficiency, X-linked follows an X-linked pattern of inheritance and is caused by pathogenic variants in the IL2RG gene located on chromosomal region Xq13.1. The age of onset is infantile. This disease is characterized by absent or markedly reduced numbers of T cells, leading to recurrent infections. The prevalence is 1:50,000-1:100,000.	600
IMPDH1	Retinitis pigmentosa type 10	NM_000883.3	NM_000883.3:c.1057G>A, NM_000883.3:c.1390delC, NM_000883.3:c.931G>A	Retinitis pigmentosa type 10 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IMPDH1 gene located on chromosomal region 7q32.1. The age of onset is infantile. This disease is characterized by progressive loss of the photoreceptors and retinal pigment epithelium and resulting in blindness usually after several decades. The prevalence is 1/4,000.	250,6
IMPG2	Retinitis pigmentosa type 56	NM_016247.3	NM_016247.3:c.635C>G, NM_016247.3:c.3262C>T, NM_016247.3:c.502-1G>C, NM_016247.3:c.2890C>T	Retinitis pigmentosa type 56 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IMPG2 gene located on chromosomal region 3q12.3. The age of onset is infantile. This disease is characterized by progressive loss of the photoreceptors and retinal pigment epithelium and resulting in blindness usually after several decades. The prevalence is 1/4.000.	600

INPP5E	Joubert syndrome type 1	NM_019892.4	NM_019892.4:c.1132C>T, NM_019892.4:c.855_856insCG, NM_019892.4:c.1688G>A, NM_019892.4:c.1543C>T, NM_019892.4:c.1304G>A	Joubert syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the INPP5E gene located on chromosomal region 9q34.3. The age of onset is early infantile. This disease is characterized by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis leading to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia, and delay in achieving motor milestones.. The prevalence is 1/100.000.	250,6
INPP5E	MORM syndrome	NM_019892.4	NM_019892.4:c.1879C>T	MORM syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the INPP5E gene located on chromosomal region 9q34.3. The age of onset is early infantile. This disease is characterized by the association of intellectual deficit, truncal obesity, retinal dystrophy and micropenis. The prevalence is 1/100.000.	250,6
INSR	Diabetes mellitus, insulin-resistant	NM_000208.2	NM_000208.2:c.3079C>T, NM_000208.2:c.3680G>C, NM_000208.2:c.3034G>A, NM_000208.2:c.1114C>T, NM_000208.2:c.1378A>G	Diabetes mellitus, insulin-resistant follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the INSR gene located on chromosomal region 19p13.2. The age of onset is infantile. This disease is characterized by the triad of hyperinsulinemia, acanthosis nigricans (skin lesions associated with insulin resistance), and signs of hyperandrogenism in females without lipodystrophy and who are not overweight. It is generally diagnosed in young women with marked signs of hyperandrogenism, but insulin resistance and acanthosis nigricans may be observed in men and in childhood. Acromegaloid facies or muscular cramps are sometimes associated. Hyperinsulinemia, a biological marker for insulin resistance, is often associated with glucose tolerance defects over the course of the disease, and diabetes progressively sets in. Hyperandrogenism (associated with polycystic ovarian syndrome (see this term) or ovarian hyperthecoses) leads to fertility problems. The prevalence is <1:1,000,000.	250,6
INSR	Leprechaunism	NM_000208.2	NM_000208.2:c.2668C>T, NM_000208.2:c.172G>A	Leprechaunism follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the INSR gene located on chromosomal region 19p13.2. The age of onset is infantile. This disease is characterized by intrauterine and mainly postnatal severe growth retardation, extreme insulin resistance. The prevalence is <1:1,000,000.	250,6
IQCB1	Senior-Loken syndrome type 5	NM_001023570.2	NM_001023570.2:c.1036G>T, NM_001023570.2:c.817G>T, NM_001023570.2:c.1381C>T, NM_001023570.2:c.1465C>T, NM_001023570.2:c.1090C>T, NM_001023570.2:c.1518_1519delCA, NM_001023570.2:c.1069C>T	Senior-Loken syndrome type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IQCB1 gene located on chromosomal region 3q13.33. The age of onset is infantile. This disease is characterized by the association of nephronophthisis (NPHP), a chronic kidney disease, with retinal dystrophy. The prevalence is 1/1.000.000.	600
ISCU	Myopathy with deficiency of ISCU	NM_213595.3	NM_213595.3:c.338_339+2delCGGT, NM_213595.3:c.149G>A	Hereditary myopathy with lactic acidosis due to ISCU deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ISCU gene located on chromosomal region 12q23.3. The age of onset is infantile. This disease is characterized by myopathy with severe exercise intolerance.	600
ITGA6	Epidermolysis bullosa, junctional with pyloric atresia	NM_000210.2	NM_000210.2:c.791delC, NM_000210.2:c.1255dupA	Junctional epidermolysis bullosa with pyloric atresia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ITGA6 and ITGB4 genes located on chromosomal regions 2q31.1 and 17q25.1 respectively. The age of onset is early infantile. This disease is characterized by generalized blistering at birth and congenital atresia of the pylorus and rarely of other portions of the gastrointestinal tract.	600
ITGB4	Epidermolysis bullosa, junctional with pyloric atresia	NM_001005731.1	NM_001005731.1:c.112T>C, NM_001005731.1:c.1684T>C, NM_001005731.1:c.1150delG, NM_001005731.1:c.1544G>A, NM_001005731.1:c.3977-19T>A, NM_001005731.1:c.4410delG, NM_001005731.1:c.4433G>A, NM_001005731.1:c.5119+2T>C, NM_001005731.1:c.3321_3331delACTGGACCGGA, NM_001005731.1:c.4618C>T, NM_001005731.1:c.182G>A, NM_001005731.1:c.2607delC, NM_001005731.1:c.3801_3802insT, NM_001005731.1:c.3841C>T, NM_001005731.1:c.2608delC, NM_001005731.1:c.3793+1G>A, NM_001005731.1:c.1660C>T, NM_001005731.1:c.3674G>A	Junctional epidermolysis bullosa with pyloric atresia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ITGA6 and ITGB4 genes located on chromosomal regions 2q31.1 and 17q25.1 respectively. The age of onset is early infantile. This disease is characterized by generalized blistering at birth and congenital atresia of the pylorus and rarely of other portions of the gastrointestinal tract.	250,6
ITGB4	Epidermolysis bullosa, without pyloric atresia	NM_001005731.1	NM_001005731.1:c.2792G>A	Junctional epidermolysis bullosa with piloric atresia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ITGB4 gene located on chromosomal region 17q25.1. The age of onset is neonatal. This disease is characterized by generalized blistering at birth and congenital atresia of the pylorus and rarely of other portions of the gastrointestinal tract. More than 100 cases have been reported around the world.	250,6

IVD	Isovaleric acidemia	NM_002225.3	NM_002225.3:c.158G>C, NM_002225.3:c.1208A>G, NM_002225.3:c.157C>T, NM_002225.3:c.1141T>C, NM_002225.3:c.243+1G>A, NM_002225.3:c.1147+1_1147+4delGTGA, NM_002225.3:c.367G>A, NM_002225.3:c.605G>T, NM_002225.3:c.1145_1147+4delTTGGTGA, NM_002225.3:c.559+1G>A, NM_002225.3:c.134T>C, NM_002225.3:c.941C>T, NM_002225.3:c.627delT, NM_002225.3:c.793+1G>A, NM_002225.3:c.2T>G, NM_002225.3:c.1183C>T, NM_002225.3:c.390delT, NM_002225.3:c.406_407delTG, NM_002225.3:c.158G>A, NM_002225.3:c.593G>A, NM_002225.3:c.507delG, NM_002225.3:c.1188delT, NM_002225.3:c.465+2T>C, NM_002225.3:c.434_437dupATGA, NM_002225.3:c.860G>A, NM_002225.3:c.994_995delAT, NM_002225.3:c.1192C>T, NM_002225.3:c.478_479insGT	Isovaleric academia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the IVD gene located on chromosomal region 15q15.1. The age of onset is neonatal. This disease is characterized by vomiting, dehydration, coma and abnormal movements. The prevalence is 1/100,000.	250,6
JAK3	Severe combined immunodeficiency T-B+NK-	NM_000215.3	NM_000215.3:c.452C>G, NM_000215.3:c.1765G>A, NM_000215.3:c.1333C>T, NM_000215.3:c.1172_1173insG, NM_000215.3:c.1837C>T, NM_000215.3:c.299A>G, NM_000215.3:c.1695C>A	Severe combined immunodeficiency T-B+NK- follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the JAK3 gene located on chromosomal region 19p13.11. The age of onset is infantile. This disease is characterized by chronic diarrhea, failure to thrive, recurrent respiratory infections and/or generalized infections due to opportunistic pathogens. The incidence is 1/100,000 and 1/1,000,000.	250,6
KCNJ1	Bartter syndrome type 2	NM_000220.4	NM_000220.4:c.1012C>T, NM_000220.4:c.1070T>C, NM_000220.4:c.592G>A, NM_000220.4:c.322G>C, NM_000220.4:c.372T>A, NM_000220.4:c.500G>A, NM_000220.4:c.237C>G, NM_000220.4:c.1014delA, NM_000220.4:c.641C>T, NM_000220.4:c.657C>G, NM_000220.4:c.996_999delAAAG, NM_000220.4:c.942T>G	Bartter syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the KCNJ1 gene located on chromosomal region 11q24.3. The age of onset is antenatal. This disease is characterized by severe polyhydramnios in mother leading to premature delivery, postnatally newborns suffer from recurrent episodes of severe dehydration and electrolyte imbalance which can lead to fatal outcome.	250,6
KCNJ13	Leber congenital amaurosis type 16	NM_002242.4	NM_002242.4:c.722T>C	Leber congenital amaurosis type 16 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the KCNJ13 gene located on chromosomal region 2q37.1. The age of onset is early infantile. This disease is characterized by retinal dystrophy defined by blindness, nystagmus, roving eye movement, leading to severe visual impairment within the first year of life.	600
KCNV2	Retinal cone dystrophy type 3B	NM_133497.3	NM_133497.3:c.1016_1024delACCTGGTGG, NM_133497.3:c.1376G>A, NM_133497.3:c.427G>T, NM_133497.3:c.226C>T, NM_133497.3:c.325C>T, NM_133497.3:c.357_358insC, NM_133497.3:c.1480A>C, NM_133497.3:c.1132_1133insT, NM_133497.3:c.854T>G, NM_133497.3:c.491T>C, NM_133497.3:c.767C>G, NM_133497.3:c.916G>T, NM_133497.3:c.778A>T, NM_133497.3:c.442G>T	Retinal cone dystrophy type 3B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the KCNV2 gene located on 9p24.2. The age of onset is in the first or second decade of life. This disease is characterized by is characterized by onset in the first or second decade of life of very marked photophobia, myopia, reduced color vision along the red-green axis with relatively preserved tritan discrimination, and central scotomata with peripheral widespread sensitivity loss predominating in the superior visual field. Nyctalopia is a later feature of the disorder. There is often retinal pigment epithelium disturbance at the macula with a normal retinal periphery.	250,6
KIF7	Acrocallosal syndrome	NM_198525.2	NM_198525.2:c.2473G>T, NM_198525.2:c.687delG, NM_198525.2:c.3001C>T, NM_198525.2:c.460C>T, NM_198525.2:c.61C>T, NM_198525.2:c.2896_2897delGC, NM_198525.2:c.3778_3779insC	Acrocallosal syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the KIF7 gene located on chromosomal region 15q26.1. The age of onset is infantile. This disease is characterized by y agenesis of corpus callosum (CC), distal anomalies of limbs, minor craniofacial anomalies and intellectual deficit. The prevalence is <1:1,000,000.	600
L1CAM	MASA sindrome/hydrocephalus	NM_000425.4	NM_000425.4:c.3489_3490delTG, NM_000425.4:c.3201T>G, NM_000425.4:c.719C>T, NM_000425.4:c.3581C>T, NM_000425.4:c.2879delA, NM_000425.4:c.791G>A, NM_000425.4:c.2092G>A, NM_000425.4:c.924C>T, NM_000425.4:c.536T>G, NM_000425.4:c.23delT, NM_000425.4:c.2254G>A, NM_000425.4:c.800dupA, NM_000425.4:c.772C>T, NM_000425.4:c.1354G>A, NM_000425.4:c.551G>A, NM_000425.4:c.1792G>A, NM_000425.4:c.1108G>A	MASA sindrome/hydrocephalus follows an X-linked pattern of inheritance and is caused by pathogenic variants in the L1CAM gene located on chromosomal region Xq28. The age of onset is infantile. This disease is characterized by adducted thumbs, hypotonia progressing to spasticity or spastic paraplegia, delayed development of speech, mild to moderate intellectual deficit, and mild to moderate distension of the cerebral ventricles. Hydrocephalus appears frequently. The prevalence is 1:25,000-1:60,000 male.	600

LAMA2	Congenital muscular dystrophy type 1A	NM_000426.3	NM_000426.3:c.184G>T, NM_000426.3:c.1612C>T, NM_000426.3:c.3718C>T, NM_000426.3:c.2750-1G>C, NM_000426.3:c.2049_2050delAG, NM_000426.3:c.5050G>T, NM_000426.3:c.1634T>A, NM_000426.3:c.2045_2046delAG, NM_000426.3:c.4645C>T, NM_000426.3:c.2962C>T, NM_000426.3:c.2098_2099delTT, NM_000426.3:c.4437-5T>A, NM_000426.3:c.2901C>A, NM_000426.3:c.112+1G>A, NM_000426.3:c.7732C>T, NM_000426.3:c.6038delT, NM_000426.3:c.7888C>T, NM_000426.3:c.825delC, NM_000426.3:c.8314delA, NM_000426.3:c.3976C>T, NM_000426.3:c.9101_9104dupAACA, NM_000426.3:c.9253C>T, NM_000426.3:c.2323-2A>T, NM_000426.3:c.8748delA, NM_000426.3:c.6334A>T, NM_000426.3:c.1050delT, NM_000426.3:c.7536delC, NM_000426.3:c.8705delT, NM_000426.3:c.9221delA, NM_000426.3:c.5227G>T, NM_000426.3:c.6429+1G>A, NM_000426.3:c.6617delT, NM_000426.3:c.2451-2A>G, NM_000426.3:c.6011delA, NM_000426.3:c.7810C>T, NM_000426.3:c.8684C>G, NM_000426.3:c.3630delT, NM_000426.3:c.3215delG, NM_000426.3:c.3623_3645delAGGGCATTGTTTTTCAACATCCA, NM_000426.3:c.6955C>T, NM_000426.3:c.7279_7280delCT, NM_000426.3:c.725G>A, NM_000426.3:c.7147C>T, NM_000426.3:c.3237C>A	Congenital muscular dystrophy type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LAMA2 gene located on chromosomal region 6q22.33. The age of onset is early infancy. This disease is characterized by hypotonia, muscle weakness and muscle wasting and motor development delayed. The prevalence is 1/30,000.	250,6
LAMA3	Epidermolysis bullosa, junctional	NM_000227.3	NM_000227.3:c.-122061G>T, NM_000227.3:c.335delG, NM_000227.3:c.4878dupT, NM_000227.3:c.2116A>T, NM_000227.3:c.4135C>T, NM_000227.3:c.751G>T, NM_000227.3:c.1981C>T, NM_000227.3:c.3350+2T>G, NM_000227.3:c.1182delG, NM_000227.3:c.4335_4336insA, NM_000227.3:c.2662C>T	Junctional epidermolysis bullosa follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LAMB3 and LAMC2 genes located on chromosomal regions 1q32.2 and 1q25.3 respectively. The age of onset is infantile. It is a lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo-epidermal basement. In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic.	600
LAMB3	Epidermolysis bullosa, junctional	NM_000228.2	NM_000228.2:c.1587_1588delAG, NM_000228.2:c.124C>T, NM_000228.2:c.1438_1442delCCGTG, NM_000228.2:c.1830G>A, NM_000228.2:c.565-2A>G, NM_000228.2:c.2806C>T, NM_000228.2:c.904delT, NM_000228.2:c.1357delT, NM_000228.2:c.3228+1G>T, NM_000228.2:c.628+1delG, NM_000228.2:c.496C>T, NM_000228.2:c.1903C>T, NM_000228.2:c.628G>A, NM_000228.2:c.3228+1G>A, NM_000228.2:c.727C>T	Junctional epidermolysis bullosa follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LAMB3 and LAMC2 genes located on chromosomal regions 1q32.2 and 1q25.3 respectively. The age of onset is infantile. It is a lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo-epidermal basement. In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic.	250,6
LAMC2	Epidermolysis bullosa, junctional	NM_005562.2	NM_005562.2:c.1659C>A, NM_005562.2:c.3069+1G>A, NM_005562.2:c.1782_1783delGC, NM_005562.2:c.2137_2143delCAGAACC, NM_005562.2:c.283C>T, NM_005562.2:c.343C>T, NM_005562.2:c.3512_3513insA, NM_005562.2:c.405-1G>A, NM_005562.2:c.3120_3121insA	Junctional epidermolysis bullosa follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LAMB3 and LAMC2 genes located on chromosomal regions 1q32.2 and 1q25.3 respectively. The age of onset is infantile. It is a lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo-epidermal basement. In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic.	600
LARGE	Muscular dystrophy-dystroglycanopathy type 6	NM_004737.4	NM_004737.4:c.1102C>T, NM_004737.4:c.992C>T, NM_004737.4:c.1525G>A, NM_004737.4:c.1483T>C	Muscular dystrophy-dystroglycanopathy type 6follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LARGE gene located on chromosomal region 22q12.3. The age of onset is infantile. There are two subtypes. Subtype 6A is associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. Subtype 6B is associated with profound mental retardation, white matter changes and structural brain abnormalities. Skeletal muscle biopsies show reduced immunolabeling of alpha-dystroglycan. The prevalence is 1:100,000-9:100,000.	600
LBR	Greenberg skeletal dysplasia	NM_002296.3	NM_002296.3:c.1748G>A, NM_002296.3:c.1402delT, NM_002296.3:c.1114C>T, NM_002296.3:c.32_35delTGGT	Greenberg dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LBR gene located on chromosomal region 1q42.12. The age of onset is fetal. This disease is characterized by fetal hydrops, short limbs and abnormal chondro-osseous calcification. The prevalence is <1:1,000,000.	600

LDHA	Glycogen storage disease type 11	NM_005566.3	NM_005566.3:c.126+1_126+4delGTAA, NM_005566.3:c.126+1G>A, NM_005566.3:c.310G>T, NM_005566.3:c.126+1_126+4del, NM_005566.3:c.397G>T, NM_005566.3:c.213+1_213+4delGTAA, NM_005566.3:c.640_641delCT	Glycogen storage disease type 11 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LDHA gene located on chromosomal region 11p15.1. The age of onset is infantile. This disease is characterized by hepatic glycogenosis and renal Fanconi syndrome.	600
LEPRE1	Osteogenesis imperfecta type 8	NM_022356.3	NM_022356.3:c.2055+13_2055+31del19, NM_022356.3:c.1656C>A, NM_022356.3:c.1365_1366delAGinsC, NM_022356.3:c.2068_2086delCGAGCGGGTGAGAGCAGCT, NM_022356.3:c.747delC, NM_022356.3:c.1102C>T, NM_022356.3:c.1473+1G>T	Osteogenesis imperfecta type 8 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LEPRE1 gene located on chromosomal region 1p34.2. The age of onset is infantile. This disease is characterized by bone fragility, low bone mass and susceptibility to bone fractures. The prevalence is 6:100,000-7:100,000.	600
LHFPL5	Deafness type 67, autosomal recessive	NM_182548.3	NM_182548.3:c.494C>T, NM_182548.3:c.476G>A, NM_182548.3:c.649delG, NM_182548.3:c.250delC, NM_182548.3:c.380A>G	Autosomal recessive nonsyndromic sensorineural deafness type DFNB67 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LHFPL5 gene located on chromosomal region 6p21.31. The age of onset is infantile, etc/. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	600
LHX3	Combined pituitary hormone deficiency type 3	NM_014564.3	NM_014564.3:c.687G>A, NM_014564.3:c.347A>G	Combined pituitary hormone deficiency type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LHX3 gene located on chromosomal region 9q34.3. The age of onset is infantile, etc/. This disease is characterized by somatolactotroph, thyrotroph and gonadotroph deficiencies, limited head and neck rotation associated with spinal abnormalities. The prevalence is <1 /1,000,000.	600
LIFR	Stuve-Wiedemann syndrome	NM_002310.5	NM_002310.5:c.2013_2014insT, NM_002310.5:c.653dupT, NM_002310.5:c.1018_1022delAATTG, NM_002310.5:c.2503G>T, NM_002310.5:c.171_174delTAAC, NM_002310.5:c.1789C>T	StÃ¼ve-Wiedemann syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LIFR gene located on chromosomal region 5p13.1. The age of onset is neonatal. This disease is characterized by small stature, congenital bowing of the long bones and campodactyly.	600
LIG4	LIG4 syndrome	NM_002312.3	NM_002312.3:c.1271_1275delAAAGA, NM_002312.3:c.833G>A, NM_002312.3:c.1738C>T, NM_002312.3:c.2440C>T, NM_002312.3:c.1406G>A, NM_002312.3:c.1455_1456delTG, NM_002312.3:c.1369_1372delGGAC, NM_002312.3:c.1512_1513delTC	LIG4 syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LIG4 gene located on chromosomal region 10p13. The age of onset is infantile. It is associated with impaired DNA double-strand break repair mechanisms and characterized by microcephaly, unusual facial features (''bird-like''), growth and developmental delay, skin anomalies including photosensitivity and psoriatic-like lesions, and pancytopenia. The disease is associated with immunodeficiency. Some patients have been reported as having telangiectasias, leukemia, lymphoma, bone marrow abnormalities, and type 2 diabetes. The prevalence 1-9/1.000.000.	600
LMNA	Cardiomyopathy, dilated type 1A	NM_170707.3	NM_170707.3:c.1366A>C, NM_170707.3:c.1930C>T, NM_170707.3:c.1567G>A, NM_170707.3:c.1786G>A	Dilated cardiomyopathy type 1A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LMNA gene located on chromosomal region 1q22. The age of onset is neonatal/fetal. This disease is characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.	250,6
LMNA	Hutchinson-Gilford progeria syndrome	NM_170707.3	NM_170707.3:c.1579C>T, NM_170707.3:c.1411C>T, NM_170707.3:c.1824C>T, NM_170707.3:c.1626G>C	Hutchinson-Gilford progeria syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LMNA gene located on chromosomal region 1q22. The age of onset is neonatal/infancy. It is characterized by growth reduction, failure to thrive, a typical facial appearance (prominent forehead, protuberant eyes, thin nose with a beaked tip, thin lips, micrognathia and protruding ears) and distinct dermatologic features (generalized alopecia, aged-looking skin, sclerotic and dimpled skin over the abdomen and extremities, prominent cutaneous vasculature, dyspigmentation, nail hypoplasia and loss of subcutaneous fat).	250,6
LMNA	Lipodystrophy, familial partial, type 2	NM_170707.3	NM_170707.3:c.1318G>A	Lipodystrophy, familial partial, type2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LMNA gene located on chromosomal region 1q22. The age of onset is neonatal/fetal. This disease is characterized by the loss of subcutaneous adipose tissue in the lower parts of the body (limbs, buttocks, trunk). It is accompanied by an accumulation of adipose tissue in the face and neck causing a double chin, fat neck, or cushingoid appearance. Adipose tissue may also accumulate in the axillae, back, labia majora, and intraabdominal region. Affected patients are insulin-resistant and may develop glucose intolerance and diabetes mellitus after age 20 years,hypertriglyceridemia, and low levels of high density lipoprotein cholesterol.	250,6

LMNA	Mandibuloacral dysplasia	NM_170707.3	NM_170707.3:c.1586C>T, NM_170707.3:c.1580G>A, NM_170707.3:c.1585G>A, NM_170707.3:c.1228C>T	Mandibuloacral dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LMNA gene located on chromosomal region 1q22. The age of onset is neonatal/. This disease is characterized by postnatal growth retardation, craniofacial anomalies, skeletal malformations, and mottled cutaneous pigmentation. The prevalence is 1:2,700-1:5,000.	250,6
LMNA	Muscular dystrophy, Emery-Dreifuss type 3	NM_170707.3	NM_170707.3:c.1072G>A, NM_170707.3:c.419T>C, NM_170707.3:c.1488+1G>A, NM_170707.3:c.1583C>A	Emery-Dreifuss muscular dystrophy type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LMNA gene located on chromosomal region 1q22. The age of onset is neonatal/fetal. This disease is characterized by weakness and atrophy of muscle without involvement of the nervous system, early contractures of the elbows, Achilles tendons and spine, and cardiomyopathy associated with cardiac conduction defects.	250,6
LOXHD1	Deafness type 77, autosomal recessive	NM_144612.6	NM_144612.6:c.2008C>T, NM_144612.6:c.2T>A, NM_144612.6:c.3874C>T, NM_144612.6:c.4526G>A, NM_144612.6:c.3924C>A, NM_144612.6:c.512-1G>A, NM_144612.6:c.4524_4525delAG, NM_144612.6:c.4714C>T, NM_144612.6:c.457_461dupCGCCA	Autosomal recessive nonsyndromic sensorineural deafness type DFNB77 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LOXHD1 gene located on chromosomal region 18q21.1. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	600
LRAT	Leber congenital amaurosis type 14	NM_004744.3	NM_004744.3:c.217_218delAT, NM_004744.3:c.588dupT	Leber congenital amaurosis 14 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRAT gene located on chromosomal region 4q32.1. The age of onset is infantile. This disease is characterized by blindness, nystagmus, roving eye movement. The prevalence is 2:100,000-3:100,000 newborn.	600
LRAT	Retinal dystrophy, early-onset severe	NM_004744.3	NM_004744.3:c.525T>A	Retinal dystrophy, early-onset severe follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRAT gene located on chromosomal region 4q32.1. The age of onset is infantile. It is a mild form of Leber congenital amaurosis that is characterized by a severe night blindness, nystagmus, and sluggish pupil responses. A relatively good central vision well into the second decade of life and blindness by the age of 30 years is generally observed. The prevalence is 2:100,000-3:100,000 newborn.	600
LRP2	Donnai-Barrow syndrome	NM_004525.2	NM_004525.2:c.11469_11472delTTTG, NM_004525.2:c.2640-1G>A, NM_004525.2:c.13139_13140insC, NM_004525.2:c.1093C>T, NM_004525.2:c.11663G>A, NM_004525.2:c.7564T>C, NM_004525.2:c.8519_8522delATTT, NM_004525.2:c.1341+2T>G, NM_004525.2:c.10769-2A>G, NM_004525.2:c.9484_9485delGT, NM_004525.2:c.13388+2T>C, NM_004525.2:c.11636-1G>T	Donnai-Barrow syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRP2 gene located on chromosomal region 2q31.1. The age of onset is infantile. This disease is characterized by diaphragmatic hernia, ocular findings, hipertelorism, agenesis of the corpus callosum, hearing loss and facial dimorphism. The prevalence is <1:1,000,000.	600
LRP5	Exudative vitreoretinopathy type 4	NM_002335.3	NM_002335.3:c.2254C>G, NM_002335.3:c.518C>T, NM_002335.3:c.1709G>A, NM_002335.3:c.804_813delGGGGAAGAGG, NM_002335.3:c.4099G>A	Exudative vitreoretinopathy type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRP5 gene located on chromosomal region 11q13.2. The age of onset is infantile or juvenile. This disease is characterized by abnormal or incomplete vascularization of the peripheral retina leading to variable clinical manifestations ranging from no effects to minor anomalies, or even retinal detachment with blindness.	250,6
LRP5	Isolated polycystic liver disease	NM_002335.3	NM_002335.3:c.4651G>A	Isolated polycystic liver disease due to LRP5 gene located on chromosomal region 11q13.2 follows an autosomal recessive pattern of inheritance. The age of onset is variable. This disease is characterized by the appearance of numerous cysts spread throughout the liver.	250,6
LRP5	Osteoporosis-pseudoglioma syndrome	NM_002335.3	NM_002335.3:c.1481G>A, NM_002335.3:c.1453G>T, NM_002335.3:c.1468delG, NM_002335.3:c.2305delG, NM_002335.3:c.2202G>A, NM_002335.3:c.1708C>T, NM_002335.3:c.3107G>A, NM_002335.3:c.2557C>T	Osteoporosis-pseudoglioma syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRP5 gene located on chromosomal region 11q13.2. The age of onset is infantile. This disease is characterized by congenital or infancy-onset blindness and severe juvenile-onset osteoporosis and spontaneous fractures. The prevalence is 1:2,000,000.	250,6
LRPPRC	Leigh syndrome, French-Canadian type	NM_133259.3	NM_133259.3:c.1061C>T, NM_133259.3:c.3830_3839delGTGGTGCAATinsAG	French-Canadian type Leigh syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRPPRC gene located on chromosomal region 2p21. The age of onset is infantile. This disease is characterized by chronic metabolic acidosis, hypotonia, facial dysmorphism and delayed development. The prevalence is 1:2,000 newborn.	600
LRTOMT	Deafness type 63, utosomal recessive	NM_001145308.4	NM_001145308.4:c.242G>A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB63 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the LRTOMT gene located on chromosomal region 11q13.4. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	600

MAK	Retinitis pigmentosa type 62	NM_001242957.1	NM_001242957.1:c.37G>A, NM_001242957.1:c.719_720dupAG, NM_001242957.1:c.1087_1088delAG, NM_001242957.1:c.718C>T, NM_001242957.1:c.388A>C	Retinitis pigmentosa type 62 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MAK gene located on chromosomal region 6p24.2. The age of onset is infantile. This disease is characterized by progressive loss of the photoreceptors and retinal pigment epithelium and resulting in blindness usually after several decades.	600
MAN2B1	Alpha-mannosidosis	NM_000528.3	NM_000528.3:c.215A>T, NM_000528.3:c.2401G>T, NM_000528.3:c.2278C>T, NM_000528.3:c.2368C>T, NM_000528.3:c.2119C>T, NM_000528.3:c.2013delT, NM_000528.3:c.1A>G, NM_000528.3:c.1067C>G, NM_000528.3:c.384G>A, NM_000528.3:c.2398G>A, NM_000528.3:c.1915C>T, NM_000528.3:c.2426T>C, NM_000528.3:c.2436+2T>C, NM_000528.3:c.1259G>T, NM_000528.3:c.1780C>T, NM_000528.3:c.1929G>A, NM_000528.3:c.2686_2687delCTinsG, NM_000528.3:c.1830+1G>C	Alpha-mannosidosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MAN2B1 gene located on chromosomal region 19p13.2. The age of onset is infantile. This disease is characterized by immunodeficiency, facial and skeletal abnormalities, hearing impairment and intellectual disability. The prevalence is 1:1,000,000-9:1,000,000.	250,6
MARVELD2	Deafness type 49, autosomal recessive	NM_001038603.2	NM_001038603.2:c.1363C>T, NM_001038603.2:c.1183-1G>A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB49 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MARVELD2 gene located on chromosomal region 5q13.2. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	600
MAT1A	Methionine adenosyltransferase deficiency	NM_000429.2	NM_000429.2:c.1006G>A, NM_000429.2:c.1070C>T, NM_000429.2:c.595C>T, NM_000429.2:c.1043_1044delTG, NM_000429.2:c.790C>T, NM_000429.2:c.827_828insG, NM_000429.2:c.538_539insTG, NM_000429.2:c.914T>C, NM_000429.2:c.791G>A, NM_000429.2:c.966T>G	Methionine adenosyltransferase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MAT1A gene located on chromosomal region 10q23.1. This disease is characterized by brain demyelination (rarely leading to neurological disorders) and isolated hepatic hypermethioninemia. The prevalence is <1:1,000,000.	600
MATN3	Multiple epiphyseal dysplasia type 5	NM_002381.4	NM_002381.4:c.1405+2T>C, NM_002381.4:c.910T>A, NM_002381.4:c.1303G>A, NM_002381.4:c.693G>C	Multiple epiphyseal dysplasia type 5 type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MATN3 gene located on chromosomal region 2p24.1. The age of onset is infantile. It is relatively mild and clinically variable. It is primarily characterized by delayed and irregular ossification of the epiphyses and early-onset osteoarthritis.The prevalence is <1:1,000,000.	600
MBTPS2	Ichthyosis follicularis-atrichia-photophobia	NM_015884.3	NM_015884.3:c.1286G>A, NM_015884.3:c.1424T>C, NM_015884.3:c.677G>T, NM_015884.3:c.261G>A	Ichthyosis follicularis - alopecia â€“ photophobia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the MBTPS2 gene located on chromosomal region Xp22.12-p22.11. The age of onset is infantile. This disease is characterized by the triad of ichthyosis follicularis, alopecia, and photophobia. The prevalence is 1:200,000.	600
MCCC1	3-Methylcrotonyl-CoA carboxylase type 1 deficiency	NM_020166.4	NM_020166.4:c.1155A>C, NM_020166.4:c.1930G>T, NM_020166.4:c.2079delA, NM_020166.4:c.388G>A, NM_020166.4:c.559T>C, NM_020166.4:c.343C>T, NM_020166.4:c.640-2A>G, NM_020166.4:c.1942G>A, NM_020166.4:c.1905delA, NM_020166.4:c.640-1G>A, NM_020166.4:c.1074delG, NM_020166.4:c.1114C>T, NM_020166.4:c.558delA, NM_020166.4:c.1277T>C, NM_020166.4:c.1526delG, NM_020166.4:c.1380T>G, NM_020166.4:c.310C>T, NM_020166.4:c.1310T>C	3-methylcrotonyl-CoA carboxylase deficiency type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MCCC1 gene located on chromosomal region 3q27.1. The age of onset is neonatal. This disease is characterized by a highly variable clinical picture ranging from neonatal onset with severe neurological involvement to asymptomatic adults. The prevalence is 1:75,000 newborn.	600
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, type 2	NM_022132.4	NM_022132.4:c.295G>C, NM_022132.4:c.380C>G, NM_022132.4:c.1309A>G, NM_022132.4:c.515_516insT, NM_022132.4:c.1015G>A, NM_022132.4:c.464G>A, NM_022132.4:c.641delG, NM_022132.4:c.1576_1577insT, NM_022132.4:c.735_736insC, NM_022132.4:c.517_518insT, NM_022132.4:c.838G>T, NM_022132.4:c.499T>C, NM_022132.4:c.1367C>T, NM_022132.4:c.929C>G, NM_022132.4:c.1065A>T, NM_022132.4:c.1580G>A, NM_022132.4:c.994C>T, NM_022132.4:c.1072+1G>A	3-methylcrotonyl-CoA carboxylase deficiency type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MCCC2 gene located on chromosomal region 5q13.2. The age of onset is neonatal. This disease is characterized by a highly variable clinical picture ranging from neonatal onset with severe neurological involvement to asymptomatic adults. The prevalence is 1:75,000 newborn.	250,6
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	NM_032601.3	NM_032601.3:c.178A>C, NM_032601.3:c.2T>C, NM_032601.3:c.139C>T	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MCEE gene located on chromosomal region 2p13.3. The age of onset is neonatal. This disease is characterized by lethargy, vomiting, hypotonia, hypothermia, respiratory distress, severe ketoacidosis, hyperammonemia, neutropenia, and thrombocytopenia. The prevalence is 1:50,000-1:80,000.	600

MCOLN1	Mucolipidosis type 4	NM_020533.2	NM_020533.2:c.1084G>T, NM_020533.2:c.304C>T, NM_020533.2:c.1207C>T, NM_020533.2:c.964C>T	Mucolipidosis type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MCOLN1 gene located on chromosomal region 19p13.2. The age of onset is infantile. This disease is characterized by psychomotor retardation and visual abnormalities including corneal clouding, retinal degeneration, or strabismus. The prevalence is 1:40,000.	600
MCPH1	Microcephaly, primary, type 1, autosomal recessive	NM_024596.3	NM_024596.3:c.1973+1G>A, NM_024596.3:c.2221C>T, NM_024596.3:c.427_428insA, NM_024596.3:c.1561G>T, NM_024596.3:c.1935+1G>T, NM_024596.3:c.215C>T, NM_024596.3:c.1249_1250insT	Autosomal recessive primary microcephaly type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MCPH1 gene located on chromosomal region 8p23.1. The age of onset is neonatal. This disease is characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment. The incidence is 1/1,000,000.	600
MECP2	Rett syndrome	NM_004992.3	NM_004992.3:c.1048_1050delAGC, NM_004992.3:c.215dupC, NM_004992.3:c.611C>G, NM_004992.3:c.1282G>A, NM_004992.3:c.916C>T, NM_004992.3:c.806delG, NM_004992.3:c.502C>T, NM_004992.3:c.880C>T, NM_004992.3:c.674C>T, NM_004992.3:c.964C>T, NM_004992.3:c.705G>A, NM_004992.3:c.808C>T, NM_004992.3:c.730C>T, NM_004992.3:c.683C>G, NM_004992.3:c.753delC, NM_004992.3:c.965C>T, NM_004992.3:c.763C>T	Rett syndrome an X -linked pattern of inheritance and is caused by pathogenic variants in the MECP2 gene located on chromosomal region Xq28. The age of onset is neonatal. An X-linked dominant neurodevelopmental disorder, and one of the most common causes of mental retardation in females. Patients appear to develop normally until 6 to 18 months of age, then gradually lose speech and purposeful hand movements, and develop microcephaly, seizures, autism, ataxia, mental retardation and stereotypic hand movements. After initial regression, the condition stabilizes and patients usually survive into adulthood. The prevalence is 1:8,500.	600
MED12	Ohdo syndrome	NM_005120.2	NM_005120.2:c.3493T>C, NM_005120.2:c.5185C>A, NM_005120.2:c.3443G>A	Ohdo syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the MED12 gene located on chromosomal region Xq13.1. The age of onset is infantile. It is characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia.	600
MED25	Charcot-Marie-Tooth disease type 2B2	NM_030973.3	NM_030973.3:c.316delG, NM_030973.3:c.1366C>T, NM_030973.3:c.1004C>T	Charcot-Marie-Tooth disease type 2B2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MED25 gene located on chromosomal region 19q13.33. The age of onset is adult. This disease is characterized by symmetric moderate to severe weakness of the distal muscles, predominantly affecting the lower extremities. Marked sensory deficits were also reported.	250,6
MEFV	Familial mediterranean fever	NM_000243.2	NM_000243.2:c.163_164insA, NM_000243.2:c.1437C>G, NM_000243.2:c.2282G>A, NM_000243.2:c.163dupA, NM_000243.2:c.2076_2078delAAT, NM_000243.2:c.1958G>A, NM_000243.2:c.443A>T, NM_000243.2:c.656_657insG, NM_000243.2:c.688G>A, NM_000243.2:c.800C>T, NM_000243.2:c.1223G>A, NM_000243.2:c.501G>C, NM_000243.2:c.2040G>A, NM_000243.2:c.2040G>C, NM_000243.2:c.2084A>G, NM_000243.2:c.1141C>T, NM_000243.2:c.1016C>T, NM_000243.2:c.2177T>C, NM_000243.2:c.1772T>C, NM_000243.2:c.2080A>G, NM_000243.2:c.2082G>A, NM_000243.2:c.2230G>T	Familial Mediterranean fever follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MEFV gene located on chromosomal region 16p13.3. The age of onset is infantile or adult (before the age of 30). This disease is characterized by recurrent short episodes of fever and serositis resulting in pain in the abdomen, chest, joints and muscles. The prevalence is 1:10,000-5:10,000.	250,6
MERTK	Retinitis pigmentosa type 38	NM_006343.2	NM_006343.2:c.2189+1G>T, NM_006343.2:c.1605-2A>G, NM_006343.2:c.2070_2074delAGGAC, NM_006343.2:c.2784_2785insTA, NM_006343.2:c.2785_2786dupTA, NM_006343.2:c.2323C>T, NM_006343.2:c.2207_2210delCTGT	Retinitis pigmentosa type 38 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MERTK gene located on chromosomal region 2q13. The age of onset is infantile. This disease is characterized by. This disease is characterized by night blindness, followed by a progressive loss of peripheral vision in the daylight period and leading to blindness.	250,6
MFRP	Microphthalmia - Retinitis pigmentosa - foveoschisis - optic disc drusen	NM_031433.3	NM_031433.3:c.498delC, NM_031433.3:c.523C>T, NM_031433.3:c.629G>T, NM_031433.3:c.1150_1151insC, NM_031433.3:c.545T>C, NM_031433.3:c.1124+1G>T	Microphthalmia - Retinitis pigmentosa - foveoschisis - optic disc drusen follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MFRP gene located on chromosomal region 11q23.3. The age of onset is infantile. This disease is characterized by posterior microphthalmos, retinitis pigmentosa, foveoschisis, and optic disk drusen.	250,6
MFSD8	Ceroid lipofuscinosis, neuronal, type 7	NM_152778.2	NM_152778.2:c.1286G>A, NM_152778.2:c.999-2A>G, NM_152778.2:c.1235C>T, NM_152778.2:c.1090delA, NM_152778.2:c.362A>G, NM_152778.2:c.881C>A, NM_152778.2:c.929G>A, NM_152778.2:c.1525_1526delCT, NM_152778.2:c.894T>G	Neuronal ceroid lipofuscinosis type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MFSD8 gene located on chromosomal region 4q28.2. The age of onset is late infantile. This disease is characterized by decline of mental and motor capacities, epilepsy, and vision loss through retinal degeneration. The prevalence is 0.56:100,000-3.9:100,000.	600

MGAT2	Congenital disorders of glycosylation 2a type	NM_002408.3	NM_002408.3:c.869C>T, NM_002408.3:c.785A>G, NM_002408.3:c.1017T>A	Congenital disorder of glycosylation type 2a follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MGAT2 gene located on chromosomal region 14q21.3. The age of onset is infantile. This disease is characterized by severe psychomotor delay, postnatal growth retardation, facial dysmorphology and bleeding tendency. It has been described in four children.	600
MKKS	Bardet-Biedl/McKusick-Kaufman syndrome	NM_018848.3	NM_018848.3:c.353delG	Bardet-Biedl syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKKS gene located on chromosomal region 20p12.2. The age of onset is antenatal or infacy. This disease is characterized by a combination of clinical signs: obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, and learning disabilities, many of which appear several years after disease onset. Clinical expression is variable but most patients manifest the majority of clinical signs during the disease course. McKusick-Kaufman syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKKS gene located on chromosomal region 20p12.2. The age of onset is fetal. This disease is characterized by hydrometrocolpos, post-axial polydactyly, and to a lesser extent cardiac defects.	250,6
MKKS	Bardet-Biedl syndrome type 6	NM_018848.3	NM_018848.3:c.830T>C, NM_018848.3:c.1436C>G	Bardet-Biedl syndrome type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKKS gene located on chromosomal region 20p12.2. The age of onset is antenatal or infacy. This disease is characterized by a combination of clinical signs: obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, and learning disabilities, many of which appear several years after disease onset. Clinical expression is variable but most patients manifest the majority of clinical signs during the disease course.	250,6
MKKS	McKusick-Kaufman syndrome	NM_018848.3	NM_018848.3:c.250C>T, NM_018848.3:c.1225_1226delGG, NM_018848.3:c.724G>T	McKusick-Kaufman syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKKS gene located on chromosomal region 20p12.2. The age of onset is fetal. This disease is characterized by hydrometrocolpos, post-axial polydactyly, and to a lesser extent cardiac defects.	250,6
MKS1	Bardet-Biedl syndrome type 13	NM_017777.3	NM_017777.3:c.1349T>C	Bardet-Biedl syndrome type 13 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKS1 gene located on chromosomal region 17q22. The age of onset is antenatal or infacy. This disease is characterized by a combination of clinical signs: obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, and learning disabilities, many of which appear several years after disease onset. Clinical expression is variable but most patients manifest the majority of clinical signs during the disease course.	250,6
MKS1	Meckel type 1/Bardet-Biedl syndrome	NM_017777.3	NM_017777.3:c.1024+1G>A, NM_017777.3:c.857A>G, NM_017777.3:c.1319T>C, NM_017777.3:c.814G>C, NM_017777.3:c.508C>T, NM_017777.3:c.1319G>C	Meckel syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKS1 gene located on chromosomal region 17q22. The age of onset is infantile, etc/. This disease is characterized by a combination of renal cysts and variably associated features, including developmental anomalies of the central nervous system (usually occipital encephalocele), hepatic ductal dysplasia and cysts, and polydactyly.. The prevalence is 1:1,000,000-9:1,000,000. Bardet-Biedl syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MKS1 gene located on chromosomal region 17q22. The age of onset is antenatal or infacy. This disease is characterized by a combination of clinical signs: obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, and learning disabilities, many of which appear several years after disease onset. Clinical expression is variable but most patients manifest the majority of clinical signs during the disease course.	250,6
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1	NM_015166.3	NM_015166.3:c.135_136insC, NM_015166.3:c.206C>T, NM_015166.3:c.278C>T, NM_015166.3:c.424-2A>C, NM_015166.3:c.422A>G, NM_015166.3:c.274C>T, NM_015166.3:c.839C>T, NM_015166.3:c.423C>A, NM_015166.3:c.33_34insC	Megalencephalic leukoencephalopathy with subcortical cysts follows an autosomal recessive patternof inheritance and is caused by pathogenic variants in the MLC1 gene located on chromosomal region 22q13.33. The age of onset is infantile. This disease is characterized by ataxia followed by progressive signs of pyramidal tract involvement and mental deterioration.	600

MLYCD	Malonyl-CoA decarboxylase deficiency	NM_012213.2	NM_012213.2:c.758delT, NM_012213.2:c.679delinsATGAAGC, NM_012213.2:c.560C>G	Malonyl-CoA decarboxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MLYCD gene located on chromosomal region 16q23.3. The age of onset is early infantile. This disease is characterized by developmentally delayed with other features that include hypotonia, seizures, hypoglycaemia, metabolic acidosis, cardiomyopathy and diarrhoea. The prevalence is <1:1,000,000.	600
MMAA	Vitamin B12-responsive methylmalonic acidemia type cblA	NM_172250.2	NM_172250.2:c.1034delT, NM_172250.2:c.283C>T, NM_172250.2:c.440G>A, NM_172250.2:c.451delC, NM_172250.2:c.592_595delCTGA, NM_172250.2:c.503delC, NM_172250.2:c.450_451insG, NM_172250.2:c.811G>T, NM_172250.2:c.586C>T, NM_172250.2:c.387C>A, NM_172250.2:c.620A>G	Vitamin B12-responsive methylmalonic acidemia type cblA follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MMAA gene located on chromosomal region 4q31.21. The age of onset is early infantile. This disease is characterized by developmentally delayed with other features that include hypotonia, seizures, hypoglycaemia, metabolic acidosis, cardiomyopathy and diarrhoea. The prevalence is <1:1,000,000.	600
MMAB	Vitamin B12-responsive methylmalonic acidemia type cblB	NM_052845.3	NM_052845.3:c.557G>A, NM_052845.3:c.556C>T, NM_052845.3:c.569G>A, NM_052845.3:c.568C>T, NM_052845.3:c.577G>A, NM_052845.3:c.197-1G>T, NM_052845.3:c.700C>T, NM_052845.3:c.548A>T, NM_052845.3:c.220G>T, NM_052845.3:c.197-1G>A	Vitamin B12-responsive methylmalonic acidemia type cbl B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MMAB gene located on chromosomal region 12q24.31. The age of onset is early infantile. This disease is characterized by developmentally delayed with other features that include hypotonia, seizures, hypoglycaemia, metabolic acidosis, cardiomyopathy and diarrhoea. The prevalence is <1:1,000,000.	600
MMACHC	Methylmalonic aciduria cblC type, with homocystinuria	NM_015506.2	NM_015506.2:c.389A>G, NM_015506.2:c.388T>C, NM_015506.2:c.482G>A, NM_015506.2:c.609G>A, NM_015506.2:c.688C>T, NM_015506.2:c.394C>T, NM_015506.2:c.440G>C, NM_015506.2:c.608G>A, NM_015506.2:c.481C>T, NM_015506.2:c.619_620insG, NM_015506.2:c.547_548delGT, NM_015506.2:c.347T>C, NM_015506.2:c.658_660delAAG, NM_015506.2:c.388_390delTAC, NM_015506.2:c.615C>A, NM_015506.2:c.331C>T, NM_015506.2:c.616C>T, NM_015506.2:c.270_271insA, NM_015506.2:c.271dupA, NM_015506.2:c.615C>G	Methylmalonic acidemia with homocystinuria, type cblC follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MMACHC gene located on chromosomal region 1p34.1. The age of onset is infantile. This disease is characterized by failure to thrive, acute neurological deterioration, intellectual deficit, lethargy, seizures, microcephaly, a salt-and-pepper retinopathy, and signs of megaloblastic anemia. The prevalence is <1:1,000,000.	250,6
MMADHC	methylmalonic aciduria cblD type, with homocystinuria	NM_015702.2	NM_015702.2:c.545C>A, NM_015702.2:c.746A>G, NM_015702.2:c.419dupA, NM_015702.2:c.748C>T, NM_015702.2:c.795_796insT, NM_015702.2:c.57_64delCTCTTTAG, NM_015702.2:c.737A>G, NM_015702.2:c.776T>C, NM_015702.2:c.478+1G>T	Methylmalonic acidemia with homocystinuria, type cblD follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MMADHC gene located on chromosomal region 2q23.2. The age of onset is variable (infantile to adult). This disease is characterized by developmental delay, severe learning difficulties, seizures, movement and gait abnormalities, behavioral problems and signs of megaloblastic anemia (pallor, fatigue, anorexia). The prevalence is 1:50,000-1:80,000.	600
MOCS1	Molybdenum cofactor deficiency type A	NM_005943.5	NM_005943.5:c.218G>A, NM_005943.5:c.397_406delCCGGACGTGG, NM_005943.5:c.217C>T, NM_005943.5:c.956G>A, NM_005943.5:c.1027C>T	Molybdenum cofactor deficiency type A (gene MOCS1) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MOCS1 gene located on chromosomal region 6p21.2. The age of onset is infantile. This disease is characterized by severe neurological abnormalities, dislocated ocular early death.	600
MOCS2	Molybdenum cofactor deficiency type B	NM_176806.3	NM_176806.3:c.106_107delAT, NM_176806.3:c.*297+1G>A, NM_176806.3:c.58delT, NM_176806.3:c.245delT, NM_176806.3:c.190G>A, NM_176806.3:c.16C>T, NM_176806.3:c.*487A>C, NM_176806.3:c.*422G>A, NM_176806.3:c.*26_*27delAT, NM_176806.3:c.539_540delAA, NM_176806.3:c.*459_*460delAA	Molybdenum cofactor deficiency type B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MOCS2 gene located on chromosomal region 5q11.2. This disease is characterized by severe neurological abnormalities, dislocated ocular early death.	250,6
MPI	Congenital disorders of glycosylation type 1b	NM_002435.2	NM_002435.2:c.305C>T, NM_002435.2:c.656G>A, NM_002435.2:c.982C>T, NM_002435.2:c.413T>C, NM_002435.2:c.884G>A, NM_002435.2:c.1016_1019delACCC	Congenital disorder of glycosylation type 1b follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MPI gene located on chromosomal region 15q24.1. The age of onset is infantile. This disease is characterized by hepatic-intestinal manifestations (diarrhoea, vomiting, and hepatomegaly associated with hepatic fibrosis).	600
MPV17	Mitochondrial DNA depletion syndrome type 6	NM_002437.4	NM_002437.4:c.263_265delAGA, NM_002437.4:c.148C>T, NM_002437.4:c.149G>A, NM_002437.4:c.263A>T, NM_002437.4:c.284_285insG, NM_002437.4:c.498C>A, NM_002437.4:c.462-2A>C, NM_002437.4:c.70G>T, NM_002437.4:c.359G>A	Mitochondrial DNA depletion syndrome type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MPV17 gene located on chromosomal region 2p23.3. The age of onset is infantile. It is a disease due to mitochondrial dysfunction. It is characterized by infantile onset of progressive liver failure, often leading to death in the first year of life, peripheral neuropathy, corneal scarring, acral ulceration and osteomyelitis leading to autoamputation, cerebral leukoencephalopathy, failure to thrive, and recurrent metabolic acidosis with intercurrent infections.	600

MPZ	Dejerine-Sottas syndrome (MPZ)	NM_000530.6	NM_000530.6:c.661G>A, NM_000530.6:c.380G>A, NM_000530.6:c.123_125delTGT, NM_000530.6:c.407T>A, NM_000530.6:c.560_563dupAGGC, NM_000530.6:c.355_356insTCTACT, NM_000530.6:c.661_662dupGC, NM_000530.6:c.411C>T, NM_000530.6:c.188C>G, NM_000530.6:c.506delT, NM_000530.6:c.190_192delTTC, NM_000530.6:c.496_499delCTCGinsTCC, NM_000530.6:c.372_377delGTTCAC, NM_000530.6:c.89T>C, NM_000530.6:c.499G>C, NM_000530.6:c.368G>A	Dejerine-Sottas syndrome follows autosomal recessive and dominant patterns of inheritance and is caused by pathogenic variants in the MPZ gene located on chromosomal region 1q23.3. The age of onset is infantile. A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. It is characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, and delayed age of walking as well as areflexia.	600
MPZ	Neuropathy, congenital hypomyelinating or amyelinating	NM_000530.6	NM_000530.6:c.588dupT, NM_000530.6:c.626_630delCGTCG, NM_000530.6:c.392A>G, NM_000530.6:c.578G>A, NM_000530.6:c.397C>T, NM_000530.6:c.164G>T, NM_000530.6:c.150C>G, NM_000530.6:c.142C>G, NM_000530.6:c.130_137delTCCCGGGT, NM_000530.6:c.128G>T, NM_000530.6:c.549dupG, NM_000530.6:c.106A>G, NM_000530.6:c.410G>C, NM_000530.6:c.332C>T, NM_000530.6:c.371C>A, NM_000530.6:c.368_382delGCACGTTCACTTGTG, NM_000530.6:c.382G>A, NM_000530.6:c.393C>A, NM_000530.6:c.103G>T, NM_000530.6:c.88A>T, NM_000530.6:c.106A>T, NM_000530.6:c.419C>G	Neuropathy, congenital hypomyelinating or amyelinating follows an autosomal recessive or dominant patterns of inheritance and is caused by pathogenic variants in the MPZ gene located on chromosomal region 1q23.3. The age of onset is infantile. A severe degenerating neuropathy that results from a congenital impairment in myelin formation. It is clinically characterized by early onset of hypotonia, areflexia, distal muscle weakness, and very slow nerve conduction velocities (as low as 3m/s). Some patients manifest nearly complete absence of spontaneous limb movements, respiratory distress at birth, and complete absence of myelin shown by electron microscopy of peripheral nerves.	600
MRPS16	Combined oxidative phosphorylation deficiency type 2	NM_016065.3	NM_016065.3:c.2T>C, NM_016065.3:c.331C>T	Combined oxidative phosphorylation defect type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MRPS16 gene located on chromosomal region 10q22.2. The age of onset is infantile. This disease is characterized by agenesis of corpus callosum, dismorphism and fatal lactic acidosis.	600
MRPS22	Combined oxidative phosphorylation deficiency type 5	NM_020191.2	NM_020191.2:c.509G>A, NM_020191.2:c.644T>C, NM_020191.2:c.40_41insA	Combined oxidative phosphorylation defect type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MRPS22 gene located on chromosomal region 3q23. The age of onset is infantile. This disease is characterized by evere hypotonia, lactic academia and congenital hyperammonaemia.	600
MTHFR	Homocystinuria due to MTHFR deficiency	NM_005957.4	NM_005957.4:c.1743G>A, NM_005957.4:c.3G>A, NM_005957.4:c.547C>T, NM_005957.4:c.1129C>T, NM_005957.4:c.1768delC, NM_005957.4:c.968T>C, NM_005957.4:c.971A>G, NM_005957.4:c.439C>T	Homocystinuria due to methylene tetrahydrofolate reductase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MTHFR gene located on chromosomal region 1p36.22. There are some forms with onset during childhood, adolescence, or adulthood beginning with mental regression, ataxia, and, most often, common psychiatric disorders of the schizophrenic type that may be linked to cerebrovascular accidents. Other symptoms are recurrent apnoea, microcephaly and convulsions.	600
MTM1	Myotubular myopathy, X-linked	NM_000252.2	NM_000252.2:c.1415_1416delGT, NM_000252.2:c.1357_1358delCC, NM_000252.2:c.461T>G, NM_000252.2:c.420C>G, NM_000252.2:c.595_599delCCTGC, NM_000252.2:c.1261-10A>G, NM_000252.2:c.780T>A, NM_000252.2:c.670C>T, NM_000252.2:c.1306_1310dupCCTAT, NM_000252.2:c.969delA, NM_000252.2:c.721C>T, NM_000252.2:c.70C>T, NM_000252.2:c.969dupA	X-linked centronuclear myopathy follows an X-linked pattern of inheritance and is caused by pathogenic variants in the MTM1 gene located on chromosomal region Xq28. The age of onset is infantile, etc/. This disease is characterized by severe phenotype in males presenting at birth with marked weakness, hypotonia and respiratory failure. The incidence is 1/50,000 newborn man.	600
MTMR2	Charcot-Marie-Tooth disease type 4B1	NM_016156.5	NM_016156.5:c.88C>T, NM_016156.5:c.304C>T, NM_016156.5:c.1276C>T, NM_016156.5:c.88A>T	Charcot-Marie-Tooth disease type 4B1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MTMR2 gene located on chromosomal region 11q21. The age of onset is early infantile. This disease is characterized by distal and proximal muscular weakness starting in the lower extremities, sensory loss and cranial nerve involvement, foot deformities and diaphragmatic and facial involvement.	600
MTTP	Abetalipoproteinemia	NM_000253.3	NM_000253.3:c.1769G>T, NM_000253.3:c.2030delC, NM_000253.3:c.1619G>A, NM_000253.3:c.2593G>T, NM_000253.3:c.708_709delCA, NM_000253.3:c.1867+1G>A, NM_000253.3:c.703_704delAC	Abetalipoproteinemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MTP gene located on chromosomal region 4q23. The age of onset is infantile. This disease is characterized by growth delay, malabsorption, hepatomegaly, and neurological and neuromuscular manifestations. The prevalence is <1:1,000,000.	250,6
MUT	Methylmalonic acidemia	NM_000255.3	NM_000255.3:c.1420C>T, NM_000255.3:c.1445-2A>G, NM_000255.3:c.2080C>T, NM_000255.3:c.1867G>A, NM_000255.3:c.607G>A, NM_000255.3:c.1658delT, NM_000255.3:c.1280G>A, NM_000255.3:c.1399C>T, NM_000255.3:c.914T>C, NM_000255.3:c.643G>A, NM_000255.3:c.655A>T, NM_000255.3:c.1741C>T, NM_000255.3:c.1106G>A, NM_000255.3:c.1871A>G, NM_000255.3:c.1924G>C, NM_000255.3:c.682C>T, NM_000255.3:c.572C>A, NM_000255.3:c.313T>C, NM_000255.3:c.1181T>A, NM_000255.3:c.278G>A, NM_000255.3:c.678_679insAATTTATG, NM_000255.3:c.794dupT, NM_000255.3:c.671_678dupAATTTATG, NM_000255.3:c.2150G>T, NM_000255.3:c.280G>A, NM_000255.3:c.91C>T, NM_000255.3:c.1207C>T	Methylmalonic acidemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MUT gene located on chromosomal region 6p12.3. The age of onset is very early infantile. This disease is characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12.	250,6

MVK	Hyper-IgD syndrome	NM_000431.3	NM_000431.3:c.829C>T, NM_000431.3:c.803T>C, NM_000431.3:c.185G>A, NM_000431.3:c.494C>T, NM_000431.3:c.59A>C, NM_000431.3:c.1129G>A	Hyper IgD syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MVK gene located on chromosomal region 12q24.11. The age of onset is infantile. This disease is characterized by periodic attacks of fever and a systemic inflammatory reaction (cervical lymphadenopathy, abdominal pain, vomiting, diarrhea, arthralgias and skin signs).	250,6
MVK	Mevalonic aciduria	NM_000431.3	NM_000431.3:c.1000G>A, NM_000431.3:c.902A>C, NM_000431.3:c.928G>A	Mevalonic aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MVK gene located on chromosomal region 12q24.11. The age of onset is infantile. This disease is characterized by psychomotor retardation, failure to thrive, progressive cerebellar ataxia, dysmorphic features, progressive visual impairment and recurrent febrile crises. The prevalence is <1:1,000,000.	250,6
MYO15A	Deafness type 3, autosomal recessive	NM_016239.3	NM_016239.3:c.3385C>T, NM_016239.3:c.6003delG, NM_016239.3:c.6004delG, NM_016239.3:c.10573delA, NM_016239.3:c.3313G>T, NM_016239.3:c.3336delG, NM_016239.3:c.755dupA, NM_016239.3:c.5492G>T, NM_016239.3:c.4351G>A, NM_016239.3:c.6864_6874delGGACCTGGAGC, NM_016239.3:c.4751_4752dupTC, NM_016239.3:c.625G>T, NM_016239.3:c.3693-2A>G, NM_016239.3:c.6614C>T, NM_016239.3:c.6743C>T, NM_016239.3:c.6046+2T>G, NM_016239.3:c.5326C>T, NM_016239.3:c.3756+1G>T, NM_016239.3:c.8410A>T, NM_016239.3:c.8429_8447delGCGGGCAGCTGCGGGTCCT, NM_016239.3:c.8148G>T, NM_016239.3:c.9958_9961delGACT, NM_016239.3:c.4750_4751insTC, NM_016239.3:c.8548C>T	Deafness autosomal recessive type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO15A gene located on chromosomal region 17p11.2. The age of onset is infantile, etc/. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	250,6
MYO3A	Deafness type 30, autosomal recessive	NM_017433.4	NM_017433.4:c.1086T>G, NM_017433.4:c.2793+2T>A, NM_017433.4:c.4586+2T>G, NM_017433.4:c.4730+1G>A, NM_017433.4:c.1A>G, NM_017433.4:c.2506-1G>A, NM_017433.4:c.1777-12G>A, NM_017433.4:c.1952delC, NM_017433.4:c.1193C>A, NM_017433.4:c.770C>G, NM_017433.4:c.3154C>T, NM_017433.4:c.585+5G>C, NM_017433.4:c.2243delA, NM_017433.4:c.3112-2A>G, NM_017433.4:c.732-2A>G	Deafness autosomal recessive type 30 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO3A gene located on chromosomal region 10p12.1. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	250,6
MYO5A	Griscelli syndrome type 1	NM_000259.3	NM_000259.3:c.1145delC, NM_000259.3:c.2332C>T	Griscelli disease type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO5A gene located on chromosomal region 15q21.2. The age of onset is infantile. This disease is characterized by is characterised by silvery gray sheen of the hair and hypopigmentation of the skin which can be associated to neurological impairment. The prevalence is <1:1,000,000.	600
MYO6	Deafness type 37, autosomal recessive	NM_004999.3	NM_004999.3:c.2897_2899delAAG, NM_004999.3:c.2840G>A, NM_004999.3:c.647A>T, NM_004999.3:c.3496C>T, NM_004999.3:c.3808C>T, NM_004999.3:c.1446_1447insT	Deafness autosomal recessive type 37 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO6 gene located on chromosomal region 6q14.1. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	250,6
MYO7A	Deafness type 2, autosomal recessive	NM_000260.3	NM_000260.3:c.1797G>A, NM_000260.3:c.2023C>T, NM_000260.3:c.731G>C, NM_000260.3:c.3596dupT, NM_000260.3:c.1184G>A, NM_000260.3:c.133-2A>G	Deafness autosomal recessive type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO7A gene located on chromosomal region 11q13.5. The age of onset is infantile. This disease is characterized by mild-to-profound sensorineural hearing impairment with no associated visible abnormalities of the external ear or any related medical problems.	250,6
MYO7A	Usher syndrome type 1B	NM_000260.3	NM_000260.3:c.1996C>T, NM_000260.3:c.1884C>A, NM_000260.3:c.448C>T, NM_000260.3:c.2476G>A, NM_000260.3:c.4024delT, NM_000260.3:c.2617C>T, NM_000260.3:c.5227C>T, NM_000260.3:c.1344-1G>A, NM_000260.3:c.5507T>G, NM_000260.3:c.5886_5889delCTTT, NM_000260.3:c.3504-1G>C, NM_000260.3:c.3508G>A, NM_000260.3:c.4018G>A, NM_000260.3:c.5392C>T, NM_000260.3:c.640G>A, NM_000260.3:c.3134T>C, NM_000260.3:c.5824G>T, NM_000260.3:c.3G>A, NM_000260.3:c.494C>T, NM_000260.3:c.5618G>A, NM_000260.3:c.5884_5887delTTCT, NM_000260.3:c.634C>T, NM_000260.3:c.3719G>A, NM_000260.3:c.5967C>G, NM_000260.3:c.3763delA, NM_000260.3:c.635G>A, NM_000260.3:c.6025delG	Usher syndrome type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO7A gene located on chromosomal region 11q13.5. The age of onset is infantile. This disease is characterized by congenital, bilateral, profound sensorineural hearing loss, vestibular areflexia, and adolescent-onset retinitis pigmentosa. The prevalence is 1:100,000-9:100,000.	250,6

NAGA	Schindler disease	NM_000262.2	NM_000262.2:c.973G>A, NM_000262.2:c.985C>T, NM_000262.2:c.986G>A, NM_000262.2:c.577G>T	Schindler disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NAGA gene located on chromosomal region 22q13.2. The age of onset is infantile. This disease is characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzazi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties.	250,6
NAGS	Hyperammonemia due to N-acetylglutamate synthetase deficiency	NM_153006.2	NM_153006.2:c.971G>A, NM_153006.2:c.1289T>C, NM_153006.2:c.1025delG, NM_153006.2:c.916-2A>T, NM_153006.2:c.1307dupT, NM_153006.2:c.1299G>C	Hyperammonemia due to N-acetylglutamate synthetase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NAGS gene located on chromosomal region 17q21.31. The age of onset is infantile, etc/. This disease is characterized by vomiting, hyperactivity or lethargy, diarrhoea, poor feeding, seizures, hypotonia, delayed psychomotor development and respiratory distress. The prevalence is <1:1,000,000.	600
NDRG1	Charcot-Marie-Tooth disease, type 4D	NM_006096.3	NM_006096.3:c.16C>T, NM_006096.3:c.-18-2_-18-1delAG, NM_006096.3:c.538-1G>A, NM_006096.3:c.-18-2_-18-1del1, NM_006096.3:c.928C>T, NM_006096.3:c.442C>T	Charcot-Marie-Tooth disease type 4D follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NDRG1 gene located on chromosomal region 8q24.22. The age of onset is infantile, etc/. This disease is characterized by demyelination and hearing loss.	600
NEB	Nemaline myopathy type 2	NM_004543.4	NM_004543.4:c.11474_11475delTG, NM_004543.4:c.19119_19120delGA, NM_004543.4:c.19306-1G>A, NM_004543.4:c.19606G>T, NM_004543.4:c.6105dupT, NM_004543.4:c.3191A>G, NM_004543.4:c.18318_18319delAG, NM_004543.4:c.11473_11474delAT, NM_004543.4:c.2173G>T, NM_004543.4:c.19097_19098delTT, NM_004543.4:c.19836+1_19836+2insATGGA, NM_004543.4:c.18825+1370C>T, NM_004543.4:c.5567G>A, NM_004543.4:c.6105_6106insT, NM_004543.4:c.16842+1G>A, NM_004543.4:c.843T>G, NM_004543.4:c.8031_8041delAAATAAACGAG, NM_004543.4:c.14182_14183delGCinsAA, NM_004543.4:c.15973C>T	Nemaline myopathy type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NEB gene located on chromosomal region 2q23.3. The age of onset is infantile or adult. This disease is characterized by hypotonia, weakness and depressed or absent deep tendon reflexes, with pathologic evidence of nemaline bodies (rods) on muscle biopsy. The prevalence is 1:100,000-9:100,000 and the incidence is 1/50.000 newborn.	250,6
NEFL	Charcot-Marie-Tooth disease type 1F	NM_006158.3	NM_006158.3:c.418G>T, NM_006158.3:c.628G>T, NM_006158.3:c.361G>T	Charcot-Marie-Tooth disease type 1F follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NEFL gene located on chromosomal region 8p21.2. The age of onset is infantile. This disease is characterized by a progressive peripheral motor and sensory neuropathy with variable clinical, distal weakness and wasting of the muscles of the lower limbs. The prevalence is 15:100,000-20:100,000.	600
NEUROG3	Diarrhea type 4, malabsorptive, congenital	NM_020999.3	NM_020999.3:c.319C>A, NM_020999.3:c.278G>T	Congenital malabsorptive diarrhea type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NEUROG3 gene located on chromosomal region 10q22.1. The age of onset is early infantile. This disease is characterized by generalized malabsorption, a paucity of enteroendocrine cells, those symptoms lead to vomiting, diarrhea, dehydration, and a severe hyperchloremic metabolic acidosis after the ingestion of standard cow's milk-based formula. The prevalence is <1:1,000,000.	600
NHP2	Dyskeratosis congenita type 2, autosomal recessive	NM_017838.3	NM_017838.3:c.415T>C, NM_017838.3:c.460T>A, NM_017838.3:c.289_290delAT	Dyskeratosis congenita 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NHP2 gene located on chromosomal region 5q35.3. The age of onset is variable from infancy to adult. It is a multisystem disorder caused by defective telomere maintenance. Clinical manifestations include mucocutaneous abnormalities, bone marrow failure, and an increased predisposition to cancer, among other variable features.	600
NMNAT1	Leber congenital amaurosis type 9	NM_022787.3	NM_022787.3:c.451G>T, NM_022787.3:c.25G>A, NM_022787.3:c.457C>G, NM_022787.3:c.507G>A, NM_022787.3:c.710G>T, NM_022787.3:c.619C>T, NM_022787.3:c.769G>A	Leber congenital amaurosis type 9 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NMNAT1 gene located on chromosomal region 1p36.22. The age of onset is early infantile. This disease is characterized by blindness, nystagmus, roving eye movement, leading to severe visual impairment.	250,6
NOP10	Dyskeratosis congenita type 1, autosomal recessive	NM_018648.3	NM_018648.3:c.34G>C, NM_018648.3:c.100C>T	Dyskeratosis congenita autosomal recessive type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NOP10 gene located on chromosomal region 15q14. The age of onset is infantile. This disease is characterized by the mucocutaneous triad of abnormal skin pigmentation, nail dystrophy and mucosal leucoplakia. The prevalence is 1:1,000,000.	600

NPC1	Niemann-Pick disease type C1	NM_000271.4	NM_000271.4:c.1042C>T, NM_000271.4:c.2842G>A, NM_000271.4:c.1628C>T, NM_000271.4:c.2974G>T, NM_000271.4:c.3019C>G, NM_000271.4:c.1211G>A, NM_000271.4:c.2072C>T, NM_000271.4:c.2324A>C, NM_000271.4:c.337T>C, NM_000271.4:c.3107C>T, NM_000271.4:c.530G>A, NM_000271.4:c.743G>T, NM_000271.4:c.3611_3614delTTAC, NM_000271.4:c.813_815delCAT, NM_000271.4:c.2932C>T, NM_000271.4:c.3425T>C, NM_000271.4:c.2761C>T, NM_000271.4:c.3104C>T, NM_000271.4:c.3662delT, NM_000271.4:c.2972_2973delAG, NM_000271.4:c.2974G>A, NM_000271.4:c.2873G>A, NM_000271.4:c.352_353delAG, NM_000271.4:c.3182T>C, NM_000271.4:c.3467A>G, NM_000271.4:c.3175C>T, NM_000271.4:c.2861C>T, NM_000271.4:c.2848G>A	Niemann-Pick disease type C1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPC1 gene located on chromosomal region 18q11.2. The age of onset varies between the perinatal period and the age of 50 years or more. This disease is characterized by hepatosplenomegaly and progressive neurological involvement. The prevalence is 1/130,000.	250,6
NPC2	Niemann-Pick disease type C2	NM_006432.3	NM_006432.3:c.115G>A, NM_006432.3:c.190+5G>A, NM_006432.3:c.27delG, NM_006432.3:c.352G>T, NM_006432.3:c.58G>T, NM_006432.3:c.358C>T, NM_006432.3:c.295T>C, NM_006432.3:c.441+1G>A, NM_006432.3:c.436C>T	Niemann-Pick disease type C2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPC2 gene located on chromosomal region 14q24.3. The age of onset varies between the perinatal period and the age of 50 years or more. This disease is characterized by hepatosplenomegaly and progressive neurological involvement. The prevalence is 1/130,000.	250,6
NPHP1	Nephronophthisis type 1	NM_000272.3	NM_000272.3:c.80T>A, NM_000272.3:c.1delA, NM_000272.3:c.555_556insA, NM_000272.3:c.829C>T, NM_000272.3:c.455C>G, NM_000272.3:c.1884+1G>T, NM_000272.3:c.1184dupC	Nephronophthisis type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPHP1 gene located on chromosomal region 2q13. The age of onset is juvenile. This disease is characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent.	600
NPHP3	Nephronophthisis type 3	NM_153240.4	NM_153240.4:c.1817G>A, NM_153240.4:c.434_437delAAAG, NM_153240.4:c.1119-2A>G, NM_153240.4:c.1729C>T, NM_153240.4:c.2694-2A>G, NM_153240.4:c.1985+5G>A, NM_153240.4:c.3406C>T, NM_153240.4:c.3373C>T, NM_153240.4:c.1381G>T, NM_153240.4:c.2694-2_2694-1delAG, NM_153240.4:c.1157A>G, NM_153240.4:c.2369T>C, NM_153240.4:c.3550G>A, NM_153240.4:c.2541delG, NM_153240.4:c.2570+1G>T, NM_153240.4:c.3156_3157insA, NM_153240.4:c.3662C>T	Nephronophthisis type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPHP3 gene located on chromosomal region 3q22.1. The age of onset is adult. This disease is characterized by polyuria, polydipsia, anemia. Onset of terminal renal failure occur significantly later (median age, 19 years) than in juvenile nephronophthisis. Renal pathology is characterized by alterations of tubular basement membranes, tubular atrophy and dilation, sclerosing tubulointerstitial nephropathy, and renal cyst development predominantly at the corticomedullary junction. The prevalence is <1:1,000,000.	250,6
NPHP4	Nephronophthisis type 4	NM_015102.4	NM_015102.4:c.4179T>A, NM_015102.4:c.3767_3768insAA, NM_015102.4:c.3674C>T, NM_015102.4:c.556_557insT, NM_015102.4:c.2940_2944dupGCTCC, NM_015102.4:c.3231+1G>C, NM_015102.4:c.517C>T, NM_015102.4:c.2335C>T, NM_015102.4:c.2219G>A, NM_015102.4:c.7G>T, NM_015102.4:c.1972C>T, NM_015102.4:c.1120-1G>C	Nephronophthisis type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPHP4 gene located on chromosomal region 1p36.31. The age of onset is infantile. This disease results in end-stage renal disease at age ranging between 6 and 35 years. It is a progressive tubulo-interstitial kidney disorder characterized by polydipsia, polyuria, anemia and growth retardation. The prevalence is 1:1,000,000-9:1,000,000.	250,6
NPHS1	Nephrotic syndrome type 1	NM_004646.3	NM_004646.3:c.59-5C>G, NM_004646.3:c.3109+1G>A, NM_004646.3:c.3478C>T, NM_004646.3:c.121_122delCT, NM_004646.3:c.1481delC, NM_004646.3:c.2456A>T, NM_004646.3:c.2491C>T, NM_004646.3:c.2464G>A, NM_004646.3:c.1307_1308dupAC, NM_004646.3:c.3250delG, NM_004646.3:c.3325C>T, NM_004646.3:c.2928G>T, NM_004646.3:c.3250_3251insG, NM_004646.3:c.2746G>T, NM_004646.3:c.1715G>A	Nephrotic syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NPHS1 gene located on chromosomal region 19q13.12. The age of onset is fetal- infantile. This disease is characterized by fetal proteinuria and nephritic infantile syndrome. The prevalence is 1 in 8 200 births.	250,6
NR0B1	Cytomegalic congenital adrenal hypoplasia	NM_000475.4	NM_000475.4:c.513G>A, NM_000475.4:c.591C>A, NM_000475.4:c.315G>C, NM_000475.4:c.873G>C, NM_000475.4:c.800G>C, NM_000475.4:c.788T>A, NM_000475.4:c.704G>A, NM_000475.4:c.1319A>T, NM_000475.4:c.388_389delTA, NM_000475.4:c.273C>A, NM_000475.4:c.813C>G, NM_000475.4:c.847C>T, NM_000475.4:c.1107G>A, NM_000475.4:c.890T>C, NM_000475.4:c.1316T>G	Cytomegalic congenital adrenal hypoplasia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the NR0B1 gene located on chromosomal region Xp21.2. The age of onset is infantile. This disease is characterized by adrenal insufficiency with vomiting, feeding difficulty, dehydration, and shock caused by a salt-wasting episode and hypoglycemia.	600
NR2E3	Enhaced S-Cone Syndrome	NM_014249.3	NM_014249.3:c.119-2A>C, NM_014249.3:c.297_298delGT, NM_014249.3:c.932G>A, NM_014249.3:c.226C>T, NM_014249.3:c.361G>A, NM_014249.3:c.227G>A, NM_014249.3:c.1034_1038delTGCAG	Enhaced S-Cone Syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NR2E3 gene located on chromosomal region 15q23. The age of onset is infantile. This disease is characterized by night blindness, reduced bilateral visual acuity, and typical fundus findings (progressive pigmentary degenerative changes, macular edema, retinoschisis).	250,6
NTRK1	Hereditary sensory and autonomic neuropathy type 4	NM_001012331.1	NM_001012331.1:c.1076A>G, NM_001012331.1:c.1711G>C, NM_001012331.1:c.1741A>G, NM_001012331.1:c.1908_1909insT, NM_001012331.1:c.1709delT, NM_001012331.1:c.1942C>T, NM_001012331.1:c.1852C>T, NM_001012331.1:c.1456G>A, NM_001012331.1:c.2321G>C, NM_001012331.1:c.2066C>T	Hereditary sensory and autonomic neuropathy type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NTRK1 gene located on chromosomal region 1q23.1. The age of onset is infantile. This disease is characterized by anhidrosis, insensitivity to pain, self-mutilating behavior and episodes of fever.	600

NUP62	Infantile striatal degeneration	NM_153719.3	NM_153719.3:c.1172A>C	Infantile striatal degeneration follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NUP62 gene located on chromosomal region 19q13.33. The age of onset is infantile. This disease is characterized by choreoathetosis, dystonia, rigidity, spasticity, dysphagia, optic atrophy, intellectual deficit, developmental regression of motor and verbal skills, failure to thrive, myoclonus, quadriparesis, cerebellar ataxia and nystagmus. The prevalence is <1:1,000,000.	600
NYX	Night blindness, congenital stationary , type 1A, X-linked	NM_022567.2	NM_022567.2:c.1049G>A	Congenital stationary night blindness follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the NYX gene located on chromosomal region Xp11.4. The age of onset is infantile. This disease is characterized by hemeralopia with a moderate loss of visual acuity.	600
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	NM_000274.3	NM_000274.3:c.1250C>T, NM_000274.3:c.159delC, NM_000274.3:c.1205T>C, NM_000274.3:c.901-2A>G, NM_000274.3:c.533G>A, NM_000274.3:c.1276C>T, NM_000274.3:c.824G>A, NM_000274.3:c.268C>G, NM_000274.3:c.952delG, NM_000274.3:c.994G>A, NM_000274.3:c.955C>T, NM_000274.3:c.677C>T, NM_000274.3:c.278G>T, NM_000274.3:c.627T>A, NM_000274.3:c.812G>A, NM_000274.3:c.952G>A, NM_000274.3:c.539G>C, NM_000274.3:c.596C>A	Gyrate atrophy of choroid and retina follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OAT gene located on chromosomal region 10q26.13. The age of onset is infantile. This disease is characterized by gyrate atrophy of the choroid and retina that begins during childhood with myopia and night blindness, followed by concentric shrinking of the visual field (tunnel vision) and a peculiar aspect of retinopathy on the funduscopy.	600
OCA2	Oculocutaneous albinism type 2	NM_000275.2	NM_000275.2:c.1610A>G, NM_000275.2:c.1960delG, NM_000275.2:c.2359G>A, NM_000275.2:c.819_822delCTGGinsGGTC, NM_000275.2:c.2228C>T, NM_000275.2:c.1025A>G, NM_000275.2:c.1842+1G>T, NM_000275.2:c.157delA, NM_000275.2:c.1182G>A, NM_000275.2:c.1182+2T>C, NM_000275.2:c.1441G>A, NM_000275.2:c.79G>A, NM_000275.2:c.1465A>G, NM_000275.2:c.1327G>A, NM_000275.2:c.1364+1G>T	Oculocutaneous albinism type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OCA2 gene located on chromosomal region 15q12-q13. The age of onset is infantile. This disease is characterized by variable hypopigmentation of the skin and hair, numerous characteristic ocular changes and misrouting of the optic nerves at the chiasm. The prevalence is 1/38,000-1/40,000	250,6
OCRL	Lowe syndrome	NM_000276.3	NM_000276.3:c.2299C>T, NM_000276.3:c.909_910delAG, NM_000276.3:c.2535delA, NM_000276.3:c.2403dupA, NM_000276.3:c.1499G>A, NM_000276.3:c.2530C>T	Lowe syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the OCRL gene located on chromosomal region Xq25-q26. The age of onset is infantile. This disease is characterized by proximal tubule dysfunction, hypercalcinuria, mild intellectual impairment, hypotonia and sub-clinical cataract. The prevalence is 1:100,000-10:100,000 newborn.	600
OFD1	Joubert syndrome type 10	NM_003611.2	NM_003611.2:c.2582dupT, NM_003611.2:c.2321_2322insT, NM_003611.2:c.277G>T	Joubert syndrome type 10 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the OFD1 gene located on chromosomal region Xp22.2. The age of onset is early. A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease.	600
OFD1	Orofaciodigital syndrome type 1	NM_003611.2	NM_003611.2:c.43_44delAG, NM_003611.2:c.52G>T, NM_003611.2:c.65dupA, NM_003611.2:c.221C>T, NM_003611.2:c.274T>C, NM_003611.2:c.616_617delGA, NM_003611.2:c.260A>G, NM_003611.2:c.13-10T>A, NM_003611.2:c.312+1delG, NM_003611.2:c.224A>C, NM_003611.2:c.294_312delTGGTTTGGCAAAAGAAAAG, NM_003611.2:c.62_63insT, NM_003611.2:c.275_276delCT, NM_003611.2:c.614_617delGAGA, NM_003611.2:c.225C>G, NM_003611.2:c.602delA, NM_003611.2:c.628C>T, NM_003611.2:c.653delA, NM_003611.2:c.1365_1368delACAA, NM_003611.2:c.619_624delATAGAA, NM_003611.2:c.1303A>C, NM_003611.2:c.1318delC, NM_003611.2:c.654+2_654+3delTA, NM_003611.2:c.1268_1272delAAAAC, NM_003611.2:c.1323_1326delAGAA, NM_003611.2:c.1360_1363delCTTA, NM_003611.2:c.1358T>A, NM_003611.2:c.1840delG, NM_003611.2:c.1612C>T, NM_003611.2:c.1757delG, NM_003611.2:c.1821delG, NM_003611.2:c.1319delT, NM_003611.2:c.1859_1860delCCinsG, NM_003611.2:c.2261-1G>T, NM_003611.2:c.235G>A, NM_003611.2:c.607_610delTATA, NM_003611.2:c.594_598delAAAGC, NM_003611.2:c.247C>T, NM_003611.2:c.2387+1G>C, NM_003611.2:c.290A>G, NM_003611.2:c.454C>T, NM_003611.2:c.312+2_312+7delTAAAGT, NM_003611.2:c.413-10T>G, NM_003611.2:c.2349delC, NM_003611.2:c.518-1G>A, NM_003611.2:c.1322_1326delAAGAA, NM_003611.2:c.541dupG, NM_003611.2:c.243C>G, NM_003611.2:c.241C>G	Orofaciodigital syndrome type 1 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the OFD1 gene located on chromosomal region Xp22.2. It is a group of heterogeneous disorders characterized by abnormalities in the oral cavity, face, and digits and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD1 is X-linked dominant syndrome, lethal in males. Craniofacial findings consist of facial asymmetry, hypertelorism, median cleft, or pseudocleft of the upper lip, hypoplasia of the alae nasi, oral clefts and abnormal frenulea, tongue anomalies (clefting, cysts, hamartoma), and anomalous dentition involving missing or extra teeth. Asymmetric brachydactyly and/or syndactyly of the fingers and toes occur frequently. Approximately 50% of OFD1 females have some degree of intellectual disability. Some patients have structural central nervous system anomalies such as agenesis of the corpus callosum, cerebellar agenesis, or a Dandy-Walker malformation. Patients with OFD1 can develop fibrocystic disease of the liver and pancreas, in addition to polycystic kidneys.	600

OPA3	3-methylglutaconic aciduria type 3	NM_025136.3	NM_025136.3:c.*24136delG, NM_025136.3:c.221delG	3-methylglutaconic aciduria type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OPA3 gene located on chromosomal region 19q13.32. The age of onset is infantile. This disease is characterized by the association of optic atrophy and choreoathetosis with 3-methylglutaconic aciduria. The prevalence is 1:10,000-5:10,000.	600
OSTM1	Osteopetrosis type 5, autosomal recessive	NM_014028.3	NM_014028.3:c.415_416delAG	Osteopetrosis, autosomal recessive type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OSTM1 gene located on chromosomal region 6p21. The age of onset is infantile. This disease is characterized by osteopetrosis, agenesis del cuerpo calloso, atrofia cerebral e hipocampo pequeÃ±o.	600
OTC	Ornithine transcarbamylase deficiency	NM_000531.5	NM_000531.5:c.119G>A, NM_000531.5:c.259G>A, NM_000531.5:c.118C>T, NM_000531.5:c.617T>G, NM_000531.5:c.646C>G, NM_000531.5:c.148G>T, NM_000531.5:c.589G>T, NM_000531.5:c.77G>A, NM_000531.5:c.829C>T, NM_000531.5:c.674C>T, NM_000531.5:c.717+2T>C, NM_000531.5:c.563G>T, NM_000531.5:c.275G>A, NM_000531.5:c.245T>G, NM_000531.5:c.460G>T, NM_000531.5:c.134T>C, NM_000531.5:c.332T>C, NM_000531.5:c.238A>G, NM_000531.5:c.421C>T	Ornithine transcarbamylase deficiency follows an X-linked pattern of inheritance and is caused by pathogenic variants in the OTC gene located on chromosomal region Xp11.4. The age of onset is infantile. This disease is characterized by severe neonatal hyperammonemic coma that generally proves to be fatal, in males. Females may be also affected by symptoms with various degrees of intensity, ranging from dislike for proteins to chronic vomiting, growth retardation, hypotonia, psychomotor retardation, hyperammonemic coma, or psychiatric disorders. The prevalence is 1:80,000.	600
OTOA	Deafness type 22, autosomal recessive	NM_144672.3	NM_144672.3:c.2301+1G>T, NM_144672.3:c.2359G>T, NM_144672.3:c.121-1G>A, NM_144672.3:c.827delT, NM_144672.3:c.1725_1726delCA	Deafness, autosomal recessive type 22 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OTOA gene located on chromosomal region 16p12.2. The age of onset is infantile. This disease is characterized by hearing loss with no associated visible abnormalities of the external ear or any related medical problems.	250,6
OTOF	Deaffness type 9, autosomal recessive	NM_194248.2	NM_194248.2:c.149G>A, NM_194248.2:c.1867G>A, NM_194248.2:c.1669G>A, NM_194248.2:c.2381G>A, NM_194248.2:c.1498C>T, NM_194248.2:c.1544T>C, NM_194248.2:c.5473C>G, NM_194248.2:c.1150G>A, NM_194248.2:c.1778delT, NM_194248.2:c.5103+2T>A, NM_194248.2:c.227+2T>C, NM_194248.2:c.5474_5475delCC, NM_194248.2:c.5332G>A, NM_194248.2:c.584-1G>C, NM_194248.2:c.98G>A, NM_194248.2:c.2348delG, NM_194248.2:c.3032T>C, NM_194248.2:c.4559G>A, NM_194248.2:c.4491T>A, NM_194248.2:c.5816G>A, NM_194248.2:c.766-2A>G, NM_194248.2:c.2485C>T, NM_194248.2:c.2401G>T	Deaffness, autosomal recessive type 9 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the OTOF gene located on chromosomal region 2p23.3. The age of onset is infantile. This disease is characterized by hearing loss with no associated visible abnormalities of the external ear or any related medical problems.	250,6
PAH	Phenylketonuria	NM_000277.1	NM_000277.1:c.1139C>T, NM_000277.1:c.1066-3C>T, NM_000277.1:c.117C>G, NM_000277.1:c.1166delC, NM_000277.1:c.1068C>A, NM_000277.1:c.1315+1G>A, NM_000277.1:c.1162G>A, NM_000277.1:c.143T>C, NM_000277.1:c.1243G>A, NM_000277.1:c.1169A>G, NM_000277.1:c.136G>A, NM_000277.1:c.1184C>A, NM_000277.1:c.194T>C, NM_000277.1:c.1199+17G>A, NM_000277.1:c.232G>A, NM_000277.1:c.1045T>C, NM_000277.1:c.1197A>T, NM_000277.1:c.441+1G>A, NM_000277.1:c.442-1G>A, NM_000277.1:c.442-5C>G, NM_000277.1:c.450_451insA, NM_000277.1:c.472C>T, NM_000277.1:c.204A>T, NM_000277.1:c.482T>C, NM_000277.1:c.250G>T, NM_000277.1:c.261C>A, NM_000277.1:c.1030G>A, NM_000277.1:c.1199+1G>A, NM_000277.1:c.1238G>C, NM_000277.1:c.1241A>G, NM_000277.1:c.673C>G, NM_000277.1:c.688G>A, NM_000277.1:c.721C>T, NM_000277.1:c.722delG, NM_000277.1:c.722G>A, NM_000277.1:c.727C>T, NM_000277.1:c.728G>A, NM_000277.1:c.733G>C, NM_000277.1:c.734T>C, NM_000277.1:c.737C>A, NM_000277.1:c.745C>T, NM_000277.1:c.1042C>G, NM_000277.1:c.638T>C, NM_000277.1:c.764T>C, NM_000277.1:c.782G>A, NM_000277.1:c.806delT, NM_000277.1:c.809G>A, NM_000277.1:c.814G>T, NM_000277.1:c.818C>T, NM_000277.1:c.823C>T, NM_000277.1:c.829T>G, NM_000277.1:c.898G>T, NM_000277.1:c.912+1G>A, NM_000277.1:c.284_286delTCA, NM_000277.1:c.754C>T, NM_000277.1:c.755G>A, NM_000277.1:c.357delC, NM_000277.1:c.1217T>C, NM_000277.1:c.1222C>T, NM_000277.1:c.157C>T, NM_000277.1:c.158G>A, NM_000277.1:c.165T>G, NM_000277.1:c.473G>A, NM_000277.1:c.490A>G, NM_000277.1:c.503delA, NM_000277.1:c.508C>G, NM_000277.1:c.533A>G, NM_000277.1:c.665A>G, NM_000277.1:c.838G>A, NM_000277.1:c.842+5G>A, NM_000277.1:c.896T>G, NM_000277.1:c.320A>G, NM_000277.1:c.441+5G>T, NM_000277.1:c.311C>A, NM_000277.1:c.527G>T, NM_000277.1:c.529G>A, NM_000277.1:c.47_48delCT, NM_000277.1:c.1208C>T, NM_000277.1:c.331C>T, NM_000277.1:c.926C>T, NM_000277.1:c.955G>T, NM_000277.1:c.926C>A, NM_000277.1:c.509+1G>A, NM_000277.1:c.1033G>T, NM_000277.1:c.611A>G, NM_000277.1:c.1066-11G>A, NM_000277.1:c.569T>C	Phenylketonuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PAH gene located on chromosomal region 12q23.2. The age of onset is neonatal. This disease is characterized by gradual developmental delay, stunted growth, microcephaly, seizures, tremors, eczema, vomiting, and musty odor. Untreated patients subsequently develop intellectual disability, behavioral disorders (hyperactivity) and motor disorders. The prevalence is 1:2,600-1:200,000.	250,6

PALB2	Fanconi anemia, complementation group N	NM_024675.3	NM_024675.3:c.1882_1890delAAGTCCTGC, NM_024675.3:c.2962C>T, NM_024675.3:c.50T>G, NM_024675.3:c.3116delA, NM_024675.3:c.3287A>G, NM_024675.3:c.3549C>G, NM_024675.3:c.3113G>A, NM_024675.3:c.2816T>G, NM_024675.3:c.1240C>T, NM_024675.3:c.557_558insA	Fanconi anemia, complementation group N follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PALB2 gene located on chromosomal region 16p12.2. The age of onset is infantile. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1:1,000,000-9:1,000,000.	250,6
PANK2	Pantothenate kinase-associated neurodegeneration	NM_153638.2	NM_153638.2:c.1561G>A, NM_153638.2:c.688G>A, NM_153638.2:c.790C>T, NM_153638.2:c.821_822delCT, NM_153638.2:c.1583C>T, NM_153638.2:c.1211A>T	Pantothenate kinase-associated neurodegeneration follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PANK2 gene located on chromosomal region 20p13. The age of onset is infantile. This disease is characterized by progressive extrapyramidal dysfunction (dystonia, rigidity, choreoathetosis), iron accumulation on the brain and axonal spheroids in the central nervous system. The prevalence is 1-2/1,000,000.	250,6
PAX3	Waardenburg syndrome type 3	NM_181457.3	NM_181457.3:c.268T>C, NM_181457.3:c.251C>T	Waardenburg syndrome type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PAX3 gene located on chromosomal region 2q36.1. The age of onset is infantile. This disease is characterized by limb anomalies in association with congenital hearing loss, minor defects in structures arising from neural crest resulting in pigmentation anomalies of eyes, hair, and skin.	600
PAX6	Aniridia	NM_000280.4	NM_000280.4:c.1032+3A>T, NM_000280.4:c.978_979delCA, NM_000280.4:c.949C>T, NM_000280.4:c.1124C>A, NM_000280.4:c.1032+3_1032+6delAAGT, NM_000280.4:c.917-2A>T, NM_000280.4:c.1000_1001insTGGCATATAACCT, NM_000280.4:c.891delA, NM_000280.4:c.889dupC, NM_000280.4:c.890_905delAACCAATTCCACAACC, NM_000280.4:c.921_924dupCTCC, NM_000280.4:c.888C>G, NM_000280.4:c.887dupA, NM_000280.4:c.889C>T, NM_000280.4:c.888C>A, NM_000280.4:c.868_871dupAGTT, NM_000280.4:c.847_854dupAGTCATAT, NM_000280.4:c.879dupC, NM_000280.4:c.875G>T, NM_000280.4:c.839delA, NM_000280.4:c.829C>T, NM_000280.4:c.818delA, NM_000280.4:c.844_845delCC, NM_000280.4:c.812_813delTG, NM_000280.4:c.808A>T, NM_000280.4:c.799A>T, NM_000280.4:c.818dupA, NM_000280.4:c.794G>A, NM_000280.4:c.781C>T, NM_000280.4:c.775dupT, NM_000280.4:c.795G>A, NM_000280.4:c.1031A>G, NM_000280.4:c.1016_1019delATAA, NM_000280.4:c.1017delT, NM_000280.4:c.766-1G>C, NM_000280.4:c.766-2delA, NM_000280.4:c.760_765+9delATACAGGTACCGAGA, NM_000280.4:c.765G>C, NM_000280.4:c.765G>T, NM_000280.4:c.763C>T, NM_000280.4:c.742_752delGATCTACCTGA, NM_000280.4:c.745delC, NM_000280.4:c.916+2T>G, NM_000280.4:c.847_848dupAG, NM_000280.4:c.916+1G>C, NM_000280.4:c.689delA, NM_000280.4:c.683-2A>C, NM_000280.4:c.683-5_683-4delTTinsAAC, NM_000280.4:c.683-6T>A, NM_000280.4:c.683-9C>G, NM_000280.4:c.682+2T>A, NM_000280.4:c.673delC, NM_000280.4:c.668delA, NM_000280.4:c.656_665delAAGAGCAAAT, NM_000280.4:c.661C>T, NM_000280.4:c.658G>T, NM_000280.4:c.655C>T, NM_000280.4:c.646T>C, NM_000280.4:c.642A>C, NM_000280.4:c.639_640delTA, NM_000280.4:c.640A>G, NM_000280.4:c.640A>T, NM_000280.4:c.631C>T, NM_000280.4:c.623G>A, NM_000280.4:c.622C>T, NM_000280.4:c.613C>T, NM_000280.4:c.607C>T, NM_000280.4:c.601C>T, NM_000280.4:c.595G>T, NM_000280.4:c.580G>T, NM_000280.4:c.773T>C, NM_000280.4:c.771G>A, NM_000280.4:c.770G>A, NM_000280.4:c.766-1G>A, NM_000280.4:c.535C>T, NM_000280.4:c.534G>T, NM_000280.4:c.532C>T, NM_000280.4:c.524-2A>G, NM_000280.4:c.520C>T, NM_000280.4:c.500_501delCGinsGA, NM_000280.4:c.490_500delCCGGGGACTTCinsTCGGTA, NM_000280.4:c.500C>A, NM_000280.4:c.495delG, NM_000280.4:c.491delC, NM_000280.4:c.489T>G, NM_000280.4:c.480delT, NM_000280.4:c.475delC, NM_000280.4:c.470delC, NM_000280.4:c.468C>A, NM_000280.4:c.467C>A	Aniridia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PAX6 gene located on chromosomal region 11p13. The age of onset is infantile. A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time.	600
PC	Pyruvate carboxylase deficiency	NM_000920.3	NM_000920.3:c.434T>C, NM_000920.3:c.1748G>T, NM_000920.3:c.496G>A	Pyruvate carboxylase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PC gene located on chromosomal region 11q13.2. The age of onset is infantile. This disease is characterized by metabolic acidosis, failure to thrive, developmental delay, and recurrent seizures. The prevalence is 1:250,000.	250,6
PCCA	Propionic acidemia type 1	NM_000282.3	NM_000282.3:c.1598_1601delTTGT, NM_000282.3:c.412G>A, NM_000282.3:c.1226_1227delTT, NM_000282.3:c.1891G>C, NM_000282.3:c.1899+1_1899+4delGTAA, NM_000282.3:c.1284+1G>A, NM_000282.3:c.229C>T, NM_000282.3:c.1023dupT, NM_000282.3:c.600+1G>A, NM_000282.3:c.261_262insT, NM_000282.3:c.1118T>A, NM_000282.3:c.862A>T	Propionic acidemia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PCCA gene located on chromosomal region 13q32.3. The age of onset is infantile. This disease is characterized by life threatening episodes of metabolic decompensation, neurological dysfunction and may be complicated by cardiomyopathy. The prevalence is 1:100,000.	250,6

PCCB	Propionic acidemia type 2	NM_000532.4	NM_000532.4:c.1279_1291delGTTCCCinsAA, NM_000532.4:c.1283C>T, NM_000532.4:c.337C>T, NM_000532.4:c.1538_1540dupCCC, NM_000532.4:c.990dupT, NM_000532.4:c.1304A>G, NM_000532.4:c.1228C>T, NM_000532.4:c.1229_1230insT, NM_000532.4:c.1606A>G, NM_000532.4:c.1223_1226delTCAT, NM_000532.4:c.1490C>T, NM_000532.4:c.1534C>T, NM_000532.4:c.1173_1174insT, NM_000532.4:c.1540_1541insCCC, NM_000532.4:c.331C>T, NM_000532.4:c.683C>T, NM_000532.4:c.797G>T, NM_000532.4:c.737G>T, NM_000532.4:c.1218_1231delinsTAGAGCACAGGA, NM_000532.4:c.502G>A, NM_000532.4:c.562G>A, NM_000532.4:c.1219_1224delGGCATCinsAA	Propionic acidemia type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PCCB gene located on chromosomal region 3q22.3. The age of onset is infantile. This disease is characterized by life threatening episodes of metabolic decompensation, neurological dysfunction and may be complicated by cardiomyopathy.. The prevalence is 1:100,000.	250,6
PCDH15	Usher syndrome type 1F	NM_033056.3	NM_033056.3:c.1583T>A, NM_033056.3:c.4885delA, NM_033056.3:c.4961_4962insTGAT, NM_033056.3:c.5659A>T, NM_033056.3:c.4937_4940dupTGAT, NM_033056.3:c.785G>A, NM_033056.3:c.5622_5624delAAC, NM_033056.3:c.400C>T, NM_033056.3:c.5724_5755delACGCACAAATGTTTCAGAACTTCAAACTATGT, NM_033056.3:c.4864delA, NM_033056.3:c.1737C>G, NM_033056.3:c.1021C>T, NM_033056.3:c.1088delT, NM_033056.3:c.1006C>T, NM_033056.3:c.1940C>G, NM_033056.3:c.400C>G, NM_033056.3:c.3718-2A>G, NM_033056.3:c.4548_4551dupATCT, NM_033056.3:c.7C>T, NM_033056.3:c.2645_2646delAT	Usher syndrome type 1F follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PCDH15 gene located on chromosomal region 10q21.1. The age of onset is early. This disease is characterized by congenital, bilateral, profound sensorineural hearing loss, vestibular areflexia, and adolescent onset retinitis pigmentosa. The prevalence is 4.4:100,000.	250,6
PDE6A	Retinitis pigmentosa type 43	NM_000440.2	NM_000440.2:c.1683G>A, NM_000440.2:c.1113+1G>T, NM_000440.2:c.718-4_718-3insT, NM_000440.2:c.1749C>G, NM_000440.2:c.2053G>A, NM_000440.2:c.1560_1561insA, NM_000440.2:c.304C>A, NM_000440.2:c.1040C>T, NM_000440.2:c.1113+1G>A	Retinitis pigmentosa 43 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDE6A gene located on chromosomal region 5q32. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	250,6
PDE6B	Retinitis pigmentosa type 43	NM_000283.3	NM_000283.3:c.1580T>C, NM_000283.3:c.655T>C, NM_000283.3:c.1540delC, NM_000283.3:c.1572delC, NM_000283.3:c.1920+2T>C, NM_000283.3:c.1669C>T, NM_000283.3:c.892C>T	Retinitis pigmentosa 43 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDE6A gene located on chromosomal region 5q32. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	250,6
PDE6C	Cone dystrophy type 4	NM_006204.3	NM_006204.3:c.1682dupA, NM_006204.3:c.1805A>T, NM_006204.3:c.2283+1G>C, NM_006204.3:c.256_257insAG, NM_006204.3:c.1066G>T, NM_006204.3:c.881G>A, NM_006204.3:c.481-12T>A, NM_006204.3:c.1363A>G, NM_006204.3:c.2457T>A, NM_006204.3:c.826C>T, NM_006204.3:c.633G>C, NM_006204.3:c.2036+1G>T, NM_006204.3:c.85C>T, NM_006204.3:c.180_186delCCTGTGC	Progressive cone dystrophy 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDE6C gene located on chromosomal region 10q23.33. The age of onset is infantile. This disease is characterized by reduced visual acuity, pendular nystagmus, increased sensitivity to light (photophobia), a small central scotoma, eccentric fixation, and reduced or complete loss of color discrimination.	600
PDE6G	Retinitis pigmentosa type 57	NM_002602.3	NM_002602.3:c.187+1G>T	Retinitis pigmentosa type 57 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDE6G gene located on chromosomal region 17q25.3. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife.	600
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	NM_000284.3	NM_000284.3:c.262C>T, NM_000284.3:c.787C>G, NM_000284.3:c.871G>A, NM_000284.3:c.773A>C	Pyruvate dehydrogenase E1-alpha deficiency follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PDHA1 gene located on chromosomal region Xp22.12. The age of onset is variable. This disease is characterized by primary lactic acidosis in children. It is associated with a broad clinical spectrum ranging from fatal lactic acidosis in the newborn to chronic neurologic dysfunction with structural abnormalities in the central nervous system without systemic acidosis.The prevalence is >1:40,000 newborn.	600
PDP1	Pyruvate dehydrogenase phosphatase deficiency	NM_018444.3	NM_018444.3:c.597_601delCTTTA, NM_018444.3:c.1606C>T, NM_018444.3:c.277G>T, NM_018444.3:c.803delC, NM_018444.3:c.669_673delTTACT, NM_018444.3:c.672_676delCTTTA, NM_018444.3:c.878delC, NM_018444.3:c.851_853delTTC	Pyruvate dehydrogenase phosphatase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDP1 gene located on chromosomal region 8q22.1. The age of onset is neonatal. This disease is characterized by lactic acidosis and hypotonia.	600
PDSS1	Coenzyme Q10 deficiency, primary, type 2	NM_014317.3	NM_014317.3:c.319dupT, NM_014317.3:c.924T>G	Coenzyme Q10 deficiency, primary, type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDSS1 gene located on chromosome 10p12.1. The age of onset is neonatal/infantile. This disease is characterized by multisystem disorder with early-onset deafness, optic atrophy, mild mental retardation, peripheral neuropathy, obesity and cardiac valvulopathy.	600

PDSS2	Coenzyme Q10 deficiency, primary, type 3	NM_020381.3	NM_020381.3:c.964C>T, NM_020381.3:c.129_130insC, NM_020381.3:c.1145C>T	Coenzyme Q10 deficiency, primary, type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDSS2 gene located on chromosomal region 6q21. The age of onset is infantile. This disease is characterized by onset of symptoms typically between age three and 12 months, often following a viral infection. Neurologic features include hypotonia, spasticity, movement disorders, cerebellar ataxia, and peripheral neuropathy.	600
PDX1	Pancreatic agenesis	NM_000209.3	NM_000209.3:c.532G>A, NM_000209.3:c.533A>G, NM_000209.3:c.492G>T	Pancreatic agenesis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PDX1 gene located on chromosomal region 13q12.1. The age of onset is infantile. This disease is characterized by the congenital absence of a critical mass of pancreatic tissue, pancreaticobiliary duct anomalies, leading to acute or chronic pancreatitis, hyperglycemia (50% of cases) and polysplenia.	600
PDZD7	Usher syndrome type 2C	NM_001195263.1	NM_001195263.1:c.1543C>T, NM_001195263.1:c.166_167insC, NM_001195263.1:c.2107delA, NM_001195263.1:c.144_145insA	Usher syndrome type 2C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the GPR98 and PDZD7 genes located on chromosomal regions 5q14.3 and 10q24.32 respectively. The age of onset is infantile. This disease is characterized by the association of sensorineural prelingual deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. The prevalence is 1/30,000.	600
PEX1	Peroxisome biogenesis disorder type 1A	NM_000466.2	NM_000466.2:c.2097dupT, NM_000466.2:c.2916delA, NM_000466.2:c.1842delA, NM_000466.2:c.1991T>C, NM_000466.2:c.1239+1G>T	Peroxisome biogenesis disorder type 1A (Zellweger) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX1 gene located on chromosomal region 7q21.2. The age of onset is early. This disease is characterized by neuronal migration defects in the brain, dysmorphic craniofacial features, profound hypotonia, neonatal seizures, and liver dysfunction. The prevalence is 1:1,000,000.	250,6
PEX1	Peroxisome biogenesis disorder type 1B	NM_000466.2	NM_000466.2:c.2097_2098insT, NM_000466.2:c.1952_1960dupCAGTGTGGA, NM_000466.2:c.877C>T, NM_000466.2:c.3505_3517delCAGTTGTTTTCAC, NM_000466.2:c.2528G>A	Peroxisome biogenesis disorder type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX1 gene located on chromosomal region 7q21.2. The age of onset is early. This disease includes neonatal adrenoleukodystrophy and infantile Refsum disease, two milder manifestations of the Zellweger disease spectrum. The clinical course of patients is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment.	250,6
PEX12	Peroxisome biogenesis disorder complementation group 6	NM_000286.2	NM_000286.2:c.959C>T, NM_000286.2:c.894delC, NM_000286.2:c.888_889delCT, NM_000286.2:c.538C>T, NM_000286.2:c.455_459dupGGAAA, NM_000286.2:c.771delC	Peroxisome biogenesis disorder complementation group 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX12 gene located on chromosomal region 17q12. The age of onset is early. This is a peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease, and classical rhizomelic chondrodysplasia punctata.	600
PEX2	Peroxisome biogenesis disorder complementation group 5	NM_000318.2	NM_000318.2:c.163G>A, NM_000318.2:c.789_790delCT	Peroxisome biogenesis disorder complementation group 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX2 gene located on chromosomal region 8q21.11. The age of onset is early. This is a peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease, and classical rhizomelic chondrodysplasia punctata.	600
PEX26	Peroxisome biogenesis disorder type 7	NM_017929.5	NM_017929.5:c.292C>T, NM_017929.5:c.254dupT, NM_017929.5:c.265G>A, NM_017929.5:c.353C>G	Peroxisome biogenesis disorder complementation group 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX26 gene located on chromosomal region 22q11.21. The age of onset is early. This is a peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease, and classical rhizomelic chondrodysplasia punctata. The prevalence is 1:1,000,000.	600

PEX5	Peroxisome biogenesis disorder type 2	NM_001131025.1	NM_001131025.1:c.1578T>G, NM_001131025.1:c.1279C>T, NM_001131025.1:c.*20G>C	Peroxisome biogenesis disorder complementation group 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX26 gene located on chromosomal region 22q11.21. The age of onset is early. This is a peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease, and classical rhizomelic chondrodysplasia punctata. The prevalence is 1:1,000,000.	600
PEX7	Rhizomelic chondrodysplasia punctata type 1	NM_000288.3	NM_000288.3:c.694C>T, NM_000288.3:c.649G>A, NM_000288.3:c.618G>A, NM_000288.3:c.722A>T, NM_000288.3:c.875T>A, NM_000288.3:c.653C>T, NM_000288.3:c.854A>G, NM_000288.3:c.532C>T, NM_000288.3:c.903+1G>C	Rhizomelic chondrodysplasia punctata type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PEX7 gene located on chromosomal region 6q23.3. The age of onset is early. This disease is characterized by proximal shortening of the humerus and to a lesser degree the femur (rhizomelia), punctate calcifications in cartilage with epiphyseal and metaphyseal abnormalities (chondrodysplasia punctata), coronal clefts of the vertebral bodies, cataracts, postnatal growth deficiency is profound, intellectual disability is severe, seizures. The prevalence is <1:100,000.	250,6
PGM1	Congenital disorder of glycosylation, type 1T	NM_002633.2	NM_002633.2:c.343A>G, NM_002633.2:c.361G>C, NM_002633.2:c.1507C>T, NM_002633.2:c.300+1G>A, NM_002633.2:c.787G>T	Congenital disorder of glycosylation, type 1T follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PGM1 gene located on chromosomal region 1p31.3. The age of onset is infantile. It is a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. The prevalence is <1:1,000,000.	600
PHKG2	Glycogen storage disease type 9C	NM_000294.2	NM_000294.2:c.958C>T, NM_000294.2:c.553C>T, NM_000294.2:c.130C>T, NM_000294.2:c.393-2A>G	Glycogen storage disease type 9C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PHKG2 gene located on chromosomal region 16p12.1-p11.2. The age of onset is infantile. This disease is characterized by hepatomegaly, growth retardation, and mild delay in motor development during childhood. The incidence is <1:100.000 births.	600
PHYH	Refsum disease	NM_006214.3	NM_006214.3:c.135-2A>G, NM_006214.3:c.497-2A>G, NM_006214.3:c.135-1G>C, NM_006214.3:c.805A>C, NM_006214.3:c.678+5G>T, NM_006214.3:c.823C>T, NM_006214.3:c.530A>G, NM_006214.3:c.164delT, NM_006214.3:c.678+2T>G, NM_006214.3:c.824G>A	Refsum disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PHYH gene located on chromosomal region 10p13. The age of onset is variable. This disease is characterized by hemeralopia (loss of vision in the dark), followed by episods of chronic distal motor polyneuropathy. Other associated signs include perceptive deafness, anosmia, cerebellous ataxia and sometimes, severe intellectual deficiency. Over the course of time cutaneous signs appear (ichtyosis), along with polyepiphyseal dysplasia, myocardiopathy, elevated protein in cerebrospinal fluid, and pigmentary retinitis that may result in blindness. The prevalence is 1:1.000,000-9:1.000,000.	250,6
PKHD1	Polycystic kidney disease, autosomal recessive	NM_138694.3	NM_138694.3:c.10515C>A, NM_138694.3:c.11363_11372delCTTCCCTGGA, NM_138694.3:c.10585G>C, NM_138694.3:c.107C>T, NM_138694.3:c.10452dupT, NM_138694.3:c.2452C>T, NM_138694.3:c.2747A>C, NM_138694.3:c.12027C>G, NM_138694.3:c.11284C>A, NM_138694.3:c.3367G>A, NM_138694.3:c.353delG, NM_138694.3:c.2827_2828delGA, NM_138694.3:c.2854G>A, NM_138694.3:c.1342G>C, NM_138694.3:c.1409G>A, NM_138694.3:c.11611T>C, NM_138694.3:c.3761_3762delCCinsG, NM_138694.3:c.2414C>T, NM_138694.3:c.5895_5896insA, NM_138694.3:c.5895dupA, NM_138694.3:c.6499C>T, NM_138694.3:c.664A>G, NM_138694.3:c.682A>G, NM_138694.3:c.6854G>A, NM_138694.3:c.370C>T, NM_138694.3:c.8407T>C, NM_138694.3:c.3766delC, NM_138694.3:c.3940delA, NM_138694.3:c.1486C>T, NM_138694.3:c.2341C>T, NM_138694.3:c.10219C>T, NM_138694.3:c.9107T>G, NM_138694.3:c.930delC, NM_138694.3:c.9370C>T, NM_138694.3:c.9530T>C, NM_138694.3:c.9689delA, NM_138694.3:c.982C>T, NM_138694.3:c.9866G>T, NM_138694.3:c.10036T>C, NM_138694.3:c.3229-2A>C, NM_138694.3:c.4870C>T, NM_138694.3:c.4165C>A, NM_138694.3:c.9719G>A, NM_138694.3:c.5325_5326delAG, NM_138694.3:c.5498C>T, NM_138694.3:c.8824C>T, NM_138694.3:c.85G>T, NM_138694.3:c.10412T>G, NM_138694.3:c.8518C>T, NM_138694.3:c.8408G>A, NM_138694.3:c.8317G>T, NM_138694.3:c.8870T>C	Autosomal recessive polycystic kidney disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PKHD1 gene located on chromosomal region 6p12.3-p12.2. The age of onset is early. This disease is characterized by the development of cysts affecting the collecting ducts. It is frequently associated with hepatic involvement. After birth, in addition to nephromegaly, arterial hypertension and urinary tract infections are common and often severe. The prevalence is 1:10,000-1:40,000.	250,6

PKLR	Hemolytic anemia due to red cell pyruvate kinase deficiency	NM_000298.5	NM_000298.5:c.1151C>T, NM_000298.5:c.1706G>A, NM_000298.5:c.1529G>A, NM_000298.5:c.1528C>T, NM_000298.5:c.1595G>A, NM_000298.5:c.721G>T, NM_000298.5:c.1076G>A, NM_000298.5:c.1675C>T, NM_000298.5:c.1261C>A, NM_000298.5:c.1436G>A, NM_000298.5:c.1456C>T	Hemolytic anemia due to red cell pyruvate kinase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PKLR gene located on chromosomal region 1q22. The age of onset is early. This disease is characterized by highly variable degree of chronic hemolysis, with severe neonatal jaundice and fatal anemia at birth, severe transfusion-dependent chronic hemolysis, and moderate hemolysis with exacerbation during infection. The prevalence is 1:20,000.	250,6
PLA2G6	Infantile neuroaxonal dystrophy	NM_003560.2	NM_003560.2:c.1634A>C, NM_003560.2:c.238G>A, NM_003560.2:c.1903C>T, NM_003560.2:c.109C>T, NM_003560.2:c.1612C>T, NM_003560.2:c.2370T>G, NM_003560.2:c.929T>A, NM_003560.2:c.1894C>T, NM_003560.2:c.2239C>T	Infantile neuroaxonal dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLA2G6 gene located on chromosomal region 22q13.1. The age of onset is infantile. This disease is a type of neurodegeneration with brain iron accumulation characterized by psychomotor delay and regression, increasing neurological involvement with symmetrical pyramidal tract signs and spastic tetraplegia. INAD may be classic or atypical and patients present with symptoms anywhere along a continuum between the two.	600
PLCE1	Nephrotic syndrome type 3	NM_016341.3	NM_016341.3:c.3346C>T, NM_016341.3:c.4808delA, NM_016341.3:c.3846delG, NM_016341.3:c.3736C>T, NM_016341.3:c.5560C>T, NM_016341.3:c.4451C>T, NM_016341.3:c.5669C>T, NM_016341.3:c.961C>T	Nephrotic syndrome type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLCE1 gene located on chromosomal region 10q23.33. The age of onset is variable. This disease is characterized by low blood protein levels, high cholesterol levels, high triglyceride levels, and presence of protein in the urine. The prevalence is 2:100,000-7:100,000 Children; 3:100,000 adults.	250,6
PLEC	Epidermolysis bullosa simplex with muscular dystrophy	NM_000445.4	NM_000445.4:c.6955C>T, NM_000445.4:c.9250_9251delCT, NM_000445.4:c.10971_10972delGA, NM_000445.4:c.2493+1G>C	Epidermolysis bullosa simplex with muscular dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLEC gene located on chromosomal region 8q24. The age of onset is early. This disease is characterized by generalized blistering associated with muscular dystrophy, dystrophic nails, and focal keratoderma of the palms and soles. The prevalence is 1:30,000-1:50,000.	600
PLEC	Epidermolysis bullosa simplex with pyloric atresia	NM_000445.4	NM_000445.4:c.9085C>T, NM_000445.4:c.913C>T, NM_000445.4:c.906+1G>A, NM_000445.4:c.12043_12044insG, NM_000445.4:c.11446G>T	Epidermolysis bullosa simplex with pyloric atresia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLEC gene located on chromosomal region 8q24. The age of onset is early. This disease is characterized by generalized severe blistering with widespread congenital absence of skin and pyloric atresia. The prevalence is <1:1,000,000.	600
PLEKHG5	Charcot-Marie-Tooth disease, intermediate type C	NM_020631.4	NM_020631.4:c.440-2A>G, NM_020631.4:c.3166C>T, NM_020631.4:c.1940T>C, NM_020631.4:c.2935C>T	Charcot-Marie-Tooth disease, intermediate type C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLEKHG5 gene located on chromosomal region 1p36.31. The age of onset is infantile. This disease is a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec.	600
PLG	Congenital plasminogen deficiency type 1	NM_000301.3	NM_000301.3:c.704G>A, NM_000301.3:c.1848G>A, NM_000301.3:c.1435G>T, NM_000301.3:c.693_695delGAA, NM_000301.3:c.1120G>T, NM_000301.3:c.112A>G	Plasminogen deficiency type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLG gene located on chromosomal region 6q26. The age of onset is infantile.This disease is characterized by markedly impaired extracellular fibrinolysis leading to the formation of ligneous (fibrin-rich) pseudomembranes on mucosae during wound healing. The prevalence is 1:1,000,000-9:1,000,000.	250,6
PLOD1	Ehlers-Danlos syndrome, type 6	NM_000302.3	NM_000302.3:c.955C>T, NM_000302.3:c.2032G>A, NM_000302.3:c.1533C>G, NM_000302.3:c.1836G>C, NM_000302.3:c.2008C>T, NM_000302.3:c.466+1G>A	Ehlers-Danlos syndrome type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PLOD1 gene located on chromosomal region 1p36. The age of onset is early. This disease is characterized by progressive scoliosis from birth, severe muscle hypotonia, hyperextensible joints and fragile eyeballs. The weakness can lead to ocular retinal hemorrhage, glaucoma, coloring of the sclera or even to rupture of the globe. The prevalence is <1:5,000.	600
PLP1	Pelizaeus-Merzbacher disease	NM_000533.3	NM_000533.3:c.128C>T, NM_000533.3:c.593delG, NM_000533.3:c.231_232insC, NM_000533.3:c.3G>A, NM_000533.3:c.487T>C, NM_000533.3:c.725C>T, NM_000533.3:c.737G>C, NM_000533.3:c.169G>T	Pelizaeus-Merzbacher disease follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PLP1 gene located on chromosomal region Xq22.2. The age of onset is infantile. It is a hypomyelinative leukodystrophy in which myelin is not formed properly in the central nervous system. It is characterized clinically by nystagmus, spastic quadriplegia, ataxia, and developmental delay.	600

PLP1	Spastic paraplegia type 2, X-linked	NM_000533.3	NM_000533.3:c.409C>T	Spastic paraplegia type 2, X-linked follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PLP1 gene located on chromosomal region Xq22.2. The age of onset is infantile. This disease is characterized by spastic gait and autonomic dysfunction.	600
PMM2	Congenital disorders of glycosylation type 1a	NM_000303.2	NM_000303.2:c.349G>C, NM_000303.2:c.357C>A, NM_000303.2:c.255+2T>C, NM_000303.2:c.127G>C, NM_000303.2:c.395T>C, NM_000303.2:c.415G>A, NM_000303.2:c.368G>A, NM_000303.2:c.385G>A, NM_000303.2:c.470T>C, NM_000303.2:c.484C>T, NM_000303.2:c.422G>A, NM_000303.2:c.442G>A, NM_000303.2:c.623G>C, NM_000303.2:c.647A>T, NM_000303.2:c.652C>G, NM_000303.2:c.323C>T, NM_000303.2:c.677C>G, NM_000303.2:c.691G>A, NM_000303.2:c.710C>G, NM_000303.2:c.669C>G, NM_000303.2:c.95_96delTAinsGC, NM_000303.2:c.95T>G, NM_000303.2:c.53C>G, NM_000303.2:c.710C>T, NM_000303.2:c.620T>C, NM_000303.2:c.97C>T, NM_000303.2:c.193G>T, NM_000303.2:c.338C>T, NM_000303.2:c.563A>G, NM_000303.2:c.131T>C, NM_000303.2:c.26G>A, NM_000303.2:c.109C>T, NM_000303.2:c.317A>T, NM_000303.2:c.190delT, NM_000303.2:c.256-1G>C	Congenital disorder of glycosylation type 1a follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PMM2 gene located on chromosomal region 16p13.2. The age of onset is infantile. This disease is characterized by highly variable clinical manifestations that may include feeding problems, vomiting, and diarrhea with failure to thrive in infants, and severe encephalopathy with axial hypotonia, abnormal eye movement, marked psychomotor retardation, peripheral neuropathy, cerebellar hypoplasia, stroke-like episodes, and retinitis pigmentosa in late infancy, childhood or adulthood.	250,6
PNPO	PNPO deficiency	NM_018129.3	NM_018129.3:c.685C>T, NM_018129.3:c.674G>A	PNPO deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PNPO gene located on chromosomal region 17q21.32. The age of onset is early. This disease is characterized by onset of severe seizures within hours of birth that are not responsive to anticonvulsants.	600
POLG	Mitochondrial DNA depletion syndrome, Alpers type	NM_002693.2	NM_002693.2:c.2617G>T, NM_002693.2:c.1120C>T, NM_002693.2:c.830A>T, NM_002693.2:c.3218C>T, NM_002693.2:c.3630dupC	Mitochondrial DNA depletion syndrome, Alpers type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POLG gene located on chromosomal region 15q26.1. The age of onset is early. This disease is characterized by the clinical triad of psychomotor regression, seizures, and liver disease. The prevalence is 1:1,600 newborn.	250,6
POLG	Progressive external ophthalmoplegia	NM_002693.2	NM_002693.2:c.1437C>G, NM_002693.2:c.2591A>G, NM_002693.2:c.1754G>A, NM_002693.2:c.1399G>A, NM_002693.2:c.1491G>C, NM_002693.2:c.3151G>C, NM_002693.2:c.803G>C, NM_002693.2:c.3286C>T, NM_002693.2:c.2794C>T, NM_002693.2:c.752C>T, NM_002693.2:c.3644-1G>A, NM_002693.2:c.1879C>T, NM_002693.2:c.2605C>T, NM_002693.2:c.911T>G, NM_002693.2:c.1760C>T, NM_002693.2:c.2542G>A, NM_002693.2:c.1550G>T, NM_002693.2:c.2557C>T, NM_002693.2:c.2207A>G, NM_002693.2:c.2243G>C, NM_002693.2:c.2209G>C	Progressive external ophthalmoplegia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POLG gene located on chromosomal region 15q26.1. The age of onset is early. This disease is characterized by ptosis, paralysis of the extraocular muscles, oropharyngeal weakness, and variably severe proximal limb weakness.	250,6
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A3	NM_017739.3	NM_017739.3:c.1425G>A, NM_017739.3:c.1545delC, NM_017739.3:c.1274G>C, NM_017739.3:c.1864delC, NM_017739.3:c.1411A>T, NM_017739.3:c.1469G>A, NM_017739.3:c.1539+1G>A, NM_017739.3:c.92dupA, NM_017739.3:c.1539+1G>T, NM_017739.3:c.932G>A, NM_017739.3:c.794G>A, NM_017739.3:c.880-1G>A, NM_017739.3:c.652+1G>A, NM_017739.3:c.931C>T, NM_017739.3:c.1666G>A, NM_017739.3:c.1814G>C, NM_017739.3:c.636C>T, NM_017739.3:c.187C>T	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A3 which includes both the more severe Walker-Warburg syndrome (WWS) and the slightly less severe muscle-eye-brain disease (MEB), follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POMGNT1 gene located on chromosomal region 1p34.1. The age of onset is infantile. This disease is characterized by generalized severe hypotonia, muscle weakness, absent psychomotor development, eye involvement and seizures. The prevalence is 1-9:100,000.	250,6
POMT1	Congenital muscular dystrophy with intellectual disability type B1	NM_007171.3	NM_007171.3:c.598G>C, NM_007171.3:c.193G>A, NM_007171.3:c.1770G>C, NM_007171.3:c.2005G>A, NM_007171.3:c.2163C>A, NM_007171.3:c.1746G>C, NM_007171.3:c.793C>T	Congenital muscular dystrophy with intellectual disability type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POMT1 gene located on chromosomal region 9q34.13. The age of onset is early. This disease is associated with mental retardation and mild structural brain abnormalities.	250,6
POMT1	Walker-Warburg syndrome	NM_007171.3	NM_007171.3:c.1540C>T, NM_007171.3:c.226G>A, NM_007171.3:c.1611C>G, NM_007171.3:c.1242-2A>G, NM_007171.3:c.907C>T, NM_007171.3:c.2163_2164insG, NM_007171.3:c.2167dupG, NM_007171.3:c.1153C>T, NM_007171.3:c.1261_1262insC, NM_007171.3:c.831C>G, NM_007171.3:c.1545C>G, NM_007171.3:c.1280_1281delAGinsTC	Walker-Warburg syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POMT1 and POMT2 genes located on chromosomal regions 9q34.13 and 14q24.3 respectively. The age of onset is infantile. This disease is characterized by generalized severe hypotonia, muscle weakness, absent or very poor psychomotor development, eye involvement and seizures. The prevalence is 1:100,000-9:100,000.	250,6

POMT2	Congenital muscular dystrophy with intellectual disability type A2	NM_013382.5	NM_013382.5:c.2243G>C, NM_013382.5:c.1997A>G, NM_013382.5:c.2242T>C, NM_013382.5:c.1445G>T, NM_013382.5:c.2177G>A, NM_013382.5:c.1238G>C, NM_013382.5:c.1941G>A, NM_013382.5:c.1057G>A, NM_013382.5:c.551C>T	Congenital muscular dystrophy with intellectual disability type A2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POMT2 gene located on chromosomal region 14q24.3. The age of onset is early. This disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease.	250,6
POMT2	Walker-Warburg syndrome	NM_013382.5	NM_013382.5:c.1726-2A>G, NM_013382.5:c.1417C>T, NM_013382.5:c.1912C>T, NM_013382.5:c.1608_1609delCA, NM_013382.5:c.1045_1052delinsG	Walker-Warburg syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POMT1 and POMT2 genes located on chromosomal regions 9q34.13 and 14q24.3 respectively. The age of onset is infantile. This disease is characterized by generalized severe hypotonia, muscle weakness, absent or very poor psychomotor development, eye involvement and seizures. The prevalence is 1:100,000-9:100,000.	250,6
POU1F1	Pituitary hormone deficiency, combined, type 1	NM_000306.3	NM_000306.3:c.472G>C, NM_000306.3:c.428G>A, NM_000306.3:c.577T>C, NM_000306.3:c.514C>T, NM_000306.3:c.515G>A, NM_000306.3:c.71C>T, NM_000306.3:c.433A>T, NM_000306.3:c.715C>T, NM_000306.3:c.515G>C, NM_000306.3:c.391G>T, NM_000306.3:c.688G>A, NM_000306.3:c.793C>T, NM_000306.3:c.748G>T, NM_000306.3:c.811C>T, NM_000306.3:c.404T>G	Pituitary hormone deficiency, combined, type 1. Genetic forms follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the POU1F1 gene located on chromosomal region 3p11.2. The age of onset is early. This disease is characterized by short stature, cognitive alterations or delayed puberty. The prevalence is 1:8,000.	600
POU3F4	Deafness type 2, X-linked	NM_000307.4	NM_000307.4:c.604A>T, NM_000307.4:c.499C>T	X-linked deafness type 2 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the POU3F4 gene located on chromosomal region Xq21.1. The age of onset is infantile. This disease is characterized by both conductive hearing loss resulting from stapes (perilymphatic gusher) fixation, and progressive sensorineural deafness.	600
PPT1	Neuronal ceroid-lipofuscinoses type 1	NM_000310.3	NM_000310.3:c.29T>A, NM_000310.3:c.223A>C, NM_000310.3:c.627+1G>T, NM_000310.3:c.169_170insA, NM_000310.3:c.451C>T, NM_000310.3:c.541G>T, NM_000310.3:c.840_841insA	Neuronal ceroid lipofuscinoses, type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PPT1 gene located on chromosomal region 1p32. The age of onset is adult. This disease is characterized by dementia, seizures and loss of motor capacities, and sometimes associated with visual loss caused by retinal degeneration. The prevalence is 1.5:1,000,000-9:1,000,000.	250,6
PRCD	Retinitis pigmentosa 36	NM_001077620.2	NM_001077620.2:c.64C>T, NM_001077620.2:c.52C>T	Retinitis pigmentosa 36 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PRCD gene located on chromosomal region 17q25.1. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife.	600
PRKRA	Dystonia tipo 16	NM_003690.4	NM_003690.4:c.665C>T	Dystonia 16 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PRKRA gene located on chromosomal region 2q31.2. The age of onset is early. This disease is characterized by progressive limb dystonia, laryngeal and oromandibular dystonia and parkinsonism.	600
PRODH	Hyperprolinemia type 1	NM_016335.4	NM_016335.4:c.865T>A, NM_016335.4:c.1331G>A	Hyperprolinemia type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PRODH gene located on chromosomal region 22q11.2. The age of onset is variable. This disease is characterized by benign symptoms, but associations with renal abnormalities, epileptic seizures, and other neurological manifestations, as well as certain forms of schizophrenia have been reported.	250,6
PROM1	Retinitis pigmentosa type 41	NM_006017.2	NM_006017.2:c.1841delG, NM_006017.2:c.1354_1355insT, NM_006017.2:c.1726C>T, NM_006017.2:c.199C>T, NM_006017.2:c.2490-2A>G, NM_006017.2:c.1177_1178delAT	Retinitis pigmentosa 41 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PROM1 gene located on chromosomal region 4p15.32. The age of onset is early. This disease is characterized by night blindness often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 19:100,000-27:100,000.	250,6
PROP1	Pituitary hormone deficiency, combined, type 2	NM_006261.4	NM_006261.4:c.2T>C, NM_006261.4:c.150delA, NM_006261.4:c.349T>A, NM_006261.4:c.112_124delTCGAGTGCTCCAC, NM_006261.4:c.310delC, NM_006261.4:c.343-11C>G, NM_006261.4:c.217C>T, NM_006261.4:c.218G>A, NM_006261.4:c.373C>T, NM_006261.4:c.157delA, NM_006261.4:c.295C>T, NM_006261.4:c.469_470insT, NM_006261.4:c.301_302delAG, NM_006261.4:c.358C>T, NM_006261.4:c.247C>T, NM_006261.4:c.263T>C, NM_006261.4:c.4delG	Pituitary hormone deficiency, combined, type 2, genetic forms follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PROP1 gene located on chromosomal region 5q35.3. The age of onset is early. This disease is characterized by short stature, cognitive alterations or delayed puberty.	600

PRPS1	Charcot-Marie-Tooth disease type 5, X-linked recessive	NM_002764.3	NM_002764.3:c.344T>C	X-linked Charcot-Marie-Tooth disease type 5 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PRPS1 gene located on chromosomal region Xq22.3. The age of onset is infantile. This disease is characterized by peripheral neuropathy, early-onset (prelingual) bilateral profound sensorineural hearing loss, and optic neuropathy. The prevalence is <1:1,000,000.	600
PRPS1	Deafness, X-linked	NM_002764.3	NM_002764.3:c.916G>A, NM_002764.3:c.869T>C	Sensorineural deafness, nonsyndromic, X-linked follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PRPS1 gene located on chromosomal region Xq22.1-q24. The age of onset is early. This is a form of deafness characterized by progressive, severe-to-profound sensorineural hearing loss in males. Females manifest mild to moderate hearing loss.	600
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	NM_002764.3	NM_002764.3:c.398A>C, NM_002764.3:c.455T>C	Phosphoribosylpyrophosphate synthetase superactivity follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PRPS1 gene located on chromosomal region Xq22.1-q24. This disease is characterized by excessive purine production, gout and uric acid urolithiasis.	600
PRPS1	Sensorineural deafness, nonsyndromic, X-linked	NM_002764.3	NM_002764.3:c.193G>A	Sensorineural deafness, nonsyndromic, X-linked follows an X-linked pattern of inheritance and is caused by pathogenic variants in the PRPS1 gene located on chromosomal region Xq22.1-q24. The age of onset is early. This is a form of deafness characterized by progressive, severe-to-profound sensorineural hearing loss in males. Females manifest mild to moderate hearing loss.	600
PRX	Charcot-Marie-Tooth disease type 4F	NM_181882.2	NM_181882.2:c.2553_2556delTCTC, NM_181882.2:c.1362delA, NM_181882.2:c.1951G>A, NM_181882.2:c.3208C>T, NM_181882.2:c.2145T>A, NM_181882.2:c.2098delG	Charcot-Marie-Tooth disease, type 4F follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PRX gene located on chromosomal region 19q13.2. The age of onset is infantile. This disease is characterized by delayed motor milestones, and proximal and distal muscle weakness. The prevalence is <1:1,000,000.	600
PRX	Dejerine-Sottas syndrome (PRX)	NM_181882.2	NM_181882.2:c.247delC, NM_181882.2:c.1102C>T, NM_181882.2:c.2857C>T	There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. Autosomal recessive form is caused by pathogenic variants in the PRX gene located on chromosomal region 19q13.2. The age of onset is infantile. This disease is characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, and delayed age of walking as well as areflexia. The prevalence is <1:1,000,000.	600
PSAP	Atypical Gaucher disease due to saposin C deficiency	NM_002778.2	NM_002778.2:c.643A>C, NM_002778.2:c.607C>T, NM_002778.2:c.1A>T, NM_002778.2:c.1046T>C, NM_002778.2:c.1288C>T	Gaucher disease, atypical, due to saposin C deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PSAP gene located on chromosomal region 10q21. The age of onset is early. This disease is characterized by marked glucosylceramide accumulation in the spleen without having a deficiency of glucosylceramide-beta glucosidase characteristic of classic Gaucher disease. Gaucher disease is a lysosomal storage disorder characterized by skeletal deterioration, hepatosplenomegaly, and organ dysfunction. There are several subtypes based on the presence and severity of neurological involvement.	600
PSAT1	Phosphoserine aminotransferase deficiency	NM_058179.3	NM_058179.3:c.1029_1030delCT, NM_058179.3:c.299A>C	Phosphoserine aminotransferase deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PSAT1 gene located on chromosomal region 9q21.2. The age of onset is early. This disease is characterized by acquired microcephaly, psychomotor retardation, intractable seizures and hypertonia. The prevalence is <1:1,000,000.	600
PYGM	McArdle disease	NM_005609.2	NM_005609.2:c.1628A>C, NM_005609.2:c.1466C>G, NM_005609.2:c.1094C>T, NM_005609.2:c.1827G>A, NM_005609.2:c.13_14delCT, NM_005609.2:c.1A>G, NM_005609.2:c.2009C>T, NM_005609.2:c.2128_2130delTTC, NM_005609.2:c.393delG, NM_005609.2:c.2392T>C, NM_005609.2:c.148C>T, NM_005609.2:c.1621G>T, NM_005609.2:c.613G>A, NM_005609.2:c.1963G>A, NM_005609.2:c.2262delA, NM_005609.2:c.1722T>G, NM_005609.2:c.255C>A, NM_005609.2:c.280C>T, NM_005609.2:c.1768+1G>A, NM_005609.2:c.501dupT, NM_005609.2:c.481C>T, NM_005609.2:c.1726C>T	McArdle disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PYGM gene located on chromosomal region 11q13.1. The age of onset is infantile. This disease is characterized by muscular exercise intolerance with myalgia, cramps, fatigue, and muscle weakness.	250,6

RAB23	Carpenter syndrome	NM_183227.2	NM_183227.2:c.407dupC, NM_183227.2:c.434T>A	Carpenter syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAB23 gene located on chromosomal region 6p11.2. The age of onset is early. This disease is characterized by acrocephaly, peculiar facies, brachydactyly and syndactyly in the hands, and preaxial polydactyly and syndactyly of the toes. The prevalence is <1:1,000,000.	600
RAB27A	Griscelli syndrome, type 2	NM_004580.4	NM_004580.4:c.259G>C, NM_004580.4:c.382dupA, NM_004580.4:c.217T>G, NM_004580.4:c.352C>T, NM_004580.4:c.389T>C, NM_004580.4:c.454G>C	Griscelli disease type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAB27A gene located on chromosome 15q15-q21.1. The age of onset is infantile. This disease is characterized by immune system abnormalities in addition to having hypopigmented skin and hair. Affected individuals are prone to recurrent infections. They also develop an immune condition called hemophagocytic lymphohistiocytosis, in which the immune system produces too many activated immune cells called T-lymphocytes and macrophages (histiocytes).	600
RAB3GAP1	Warburg micro syndrome type 1	NM_012233.2	NM_012233.2:c.1734G>A, NM_012233.2:c.496_497delTT, NM_012233.2:c.937dupA, NM_012233.2:c.1393_1396delTGTA, NM_012233.2:c.1410C>A, NM_012233.2:c.899+1G>A, NM_012233.2:c.2011C>T, NM_012233.2:c.748+1G>A	Warburg micro syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAB3GAP1 gene located on chromosomal region 2q21.3. The age of onset is infantile. This disease is characterized by microcephaly, microphthalmia, microcornia, congenital cataracts, optic atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, and hypogonadism. The prevalence is <1:1,000,000.	600
RAB3GAP2	Warburg micro syndrome type 2	NM_012414.3	NM_012414.3:c.1648C>T, NM_012414.3:c.1485C>A, NM_012414.3:c.325_328delAAAG, NM_012414.3:c.1276C>T	Warburg micro syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAB3GAP2 gene located on chromosomal region 1q41. The age of onset is early. This disease is characterized by microcephaly, microphthalmia, microcornia, congenital cataracts, optic atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, and hypogonadism. The prevalence is <1:1,000,000.	600
RAD51C	Fanconi anemia, complementation group O	NM_058216.2	NM_058216.2:c.706-2A>G, NM_058216.2:c.838-2A>T, NM_058216.2:c.133G>A, NM_058216.2:c.773G>A	Fanconi anemia complementation group O follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAD51C gene located on chromosomal region 17q22. The age of onset is early. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1:360,000 newborn.	600
RAG1	Immunodeficiency severe combined B cell-negative	NM_000448.2	NM_000448.2:c.2333G>A, NM_000448.2:c.2320G>T, NM_000448.2:c.2164G>A, NM_000448.2:c.940C>T, NM_000448.2:c.2814T>G, NM_000448.2:c.2923C>T, NM_000448.2:c.2326C>T	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell negative, NK cell positive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAG1 gene located on chromosomal region 11p13. The age of onset is early. This disease is characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development.	250,6
RAG1	Omenn syndrome	NM_000448.2	NM_000448.2:c.983G>A, NM_000448.2:c.3016A>G, NM_000448.2:c.256_257delAA, NM_000448.2:c.1682G>A, NM_000448.2:c.1681C>T	Omenn syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAG1 and RAG2 genes located on chromosomal region 11p12. The age of onset is early. This disease is characterized by erythroderma, desquamation, alopecia, chronic diarrhea, failure to thrive, lymphadenopathy, and hepatosplenomegaly, associated with severe combined immunodeficiency.	250,6
RAG2	Combined immunodeficiency with skin granulomas	NM_000536.3	NM_000536.3:c.115A>G, NM_000536.3:c.686G>A, NM_000536.3:c.283G>A, NM_000536.3:c.601C>T, NM_000536.3:c.230C>A, NM_000536.3:c.1352G>C	Combined immunodeficiency with skin granulomas follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAG2 gene located on chromosome 11p13. This disease is characterized by severe infections, hypogammaglobulinemia and defective T-cell function, multiple facial papulonodular.	600
RAG2	Omenn syndrome	NM_000536.3	NM_000536.3:c.685C>T, NM_000536.3:c.1504A>G	Omenn syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAG1 and RAG2 genes located on chromosomal region 11p12. The age of onset is early. This disease is characterized by erythroderma, desquamation, alopecia, chronic diarrhea, failure to thrive, lymphadenopathy, and hepatosplenomegaly, associated with severe combined immunodeficiency.	600

RAPSN	Congenital myasthenic syndrome	NM_005055.4	NM_005055.4:c.484G>A, NM_005055.4:c.264C>A, NM_005055.4:c.807C>A, NM_005055.4:c.848T>C, NM_005055.4:c.490C>T, NM_005055.4:c.603C>A	Congenital myasthenic syndromes follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAPSN gene located on chromosomal region 11p11.2. The age of onset is early. This disease is characterized by fatigable weakness of skeletal muscle (e.g., ocular, bulbar, limb muscles) with onset at or shortly after birth or in early childhood; rarely, symptoms may not manifest until later in childhood. Cardiac and smooth muscle are not involved. Severity and course of disease are highly variable, ranging from minor symptoms to progressive disabling weakness. The prevalence is 1:3,000.	250,6
RAPSN	Fetal akinesia deformation sequence	NM_005055.4	NM_005055.4:c.416T>C, NM_005055.4:c.566C>T	Fetal akinesia deformation sequence follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAPSN gene located on chromosomal region 11p11.2. The age of onset is early. This disease is characterized by multiple joint contractures, facial anomalies and pulmonary hypoplasia. The prevalence is 1:3,000.	250,6
RAX	Isolated microphthalmia type 3	NM_013435.2	NM_013435.2:c.909C>G, NM_013435.2:c.18C>A, NM_013435.2:c.197G>C, NM_013435.2:c.439C>T, NM_013435.2:c.383_384delAG	Isolated microphthalmia type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RAX gene located on chromosomal region 18q21.32. Microphthalmia designates a heterogeneous group of ocular malformations with a more or less evident reduction in the size of the eyeball. Additional features include high hypermetropia and a short axial length.	250,6
RDH12	Leber congenital amaurosis type 13	NM_152443.2	NM_152443.2:c.184C>T, NM_152443.2:c.146C>T, NM_152443.2:c.152T>A, NM_152443.2:c.451C>A, NM_152443.2:c.295C>A, NM_152443.2:c.377C>T, NM_152443.2:c.379G>T, NM_152443.2:c.565C>T, NM_152443.2:c.677A>G, NM_152443.2:c.805_809delGCCCT, NM_152443.2:c.164C>T, NM_152443.2:c.210dupC, NM_152443.2:c.448+1_448+4delGTAA, NM_152443.2:c.451C>G, NM_152443.2:c.464C>T, NM_152443.2:c.523T>C	Leber congenital amaurosis type 13 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RDH12 gene located on chromosomal region 14q24.1. The age of onset is early. This disease is characterized by blindness, nystagmus, roving eye movement and lack of detectable signals on an electroretinogram, leading to severe visual impairment within the first year of life.	250,6
RDX	Deafness type 24, autosomal recessive	NM_002906.3	NM_002906.3:c.1405dupG, NM_002906.3:c.342_346delGATAT	Autosomal recessive nonsyndromic sensorineural deafness type DFNB24 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RDX gene located on chromosomal region 11q22.3. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	600
RELN	Lissencephaly syndrome, Norman-Roberts type	NM_005045.3	NM_005045.3:c.6646C>T, NM_005045.3:c.5615-1G>A	Lissencephaly syndrome, Norman-Roberts type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RELN gene located on chromosomal region 7q22. The age of onset is early. This disease is characterized by craniofacial anomalies (severe microcephaly, a low sloping forehead, a broad and prominent nasal bridge and widely set eyes) and postnatal growth retardation, severe intellectual deficit, spasticity and epilepsy. The prevalence is 1:1,000,000-9:1,000,000.	600
REN	Renal tubular dysgenesis	NM_000537.3	NM_000537.3:c.404C>A, NM_000537.3:c.127C>T, NM_000537.3:c.145C>T	Renal tubular dysgenesis deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ACE (chromosomal region 17q23.3), AGT (1q42.2) AGTR1 (3q24) and REN (1q32.1) genes. The age of onset is fetal. This disease is characterized by absent or poorly developed proximal tubules of the kidneys, persistent oligohydramnios, leading to Potter sequence, and skull ossification defects.	600
RGR	Retinitis pigmentosa type 44	NM_001012720.1	NM_001012720.1:c.196A>C, NM_001012720.1:c.249_250insGGCTCGGA, NM_001012720.1:c.261_262insGGCTCGGA, NM_001012720.1:c.454C>A, NM_001012720.1:c.865C>T, NM_001012720.1:c.877C>T	Retinitis pigmentosa type 44 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RGR gene located on chromosomal region 10q23.1. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	250,6
RHO	Retinitis pigmentosa type 4	NM_000539.3	NM_000539.3:c.152G>C, NM_000539.3:c.173C>T, NM_000539.3:c.448G>A, NM_000539.3:c.620T>G, NM_000539.3:c.670G>A, NM_000539.3:c.745G>T, NM_000539.3:c.659T>G	Retinitis pigmentosa type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RHO gene located on chromosomal region 3q22.1. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	250,6
RLBP1	Retinitis punctata albescens	NM_000326.4	NM_000326.4:c.333T>G, NM_000326.4:c.452G>A, NM_000326.4:c.700C>T, NM_000326.4:c.875C>T	Retinitis punctata albescens follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RLBP1 gene located on chromosomal region 15q26.1. The age of onset is early. This disease is characterized by night blindness from early childhood, delay in the regeneration of cone and rod photopigments in young adults, followed by macular degeneration and a decrease in visual acuity that led to legal blindness in early adulthood.	250,6

RP2	Retinitis pigmentosa type 2	NM_006915.2	NM_006915.2:c.235delG, NM_006915.2:c.305_306insT, NM_006915.2:c.352delC, NM_006915.2:c.353G>A, NM_006915.2:c.353G>T, NM_006915.2:c.358C>T, NM_006915.2:c.453C>G, NM_006915.2:c.453delC, NM_006915.2:c.631delC	Retinitis pigmentosa type 2 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the RP2 gene located on chromosomal region Xp11.23. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	600
RPE65	Leber congenital amaurosis type 2	NM_000329.2	NM_000329.2:c.1067delA, NM_000329.2:c.1301C>T, NM_000329.2:c.1292A>G, NM_000329.2:c.272G>A, NM_000329.2:c.907A>T, NM_000329.2:c.514_515delGT	Leber congenital amaurosis 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RPE65 gene located on chromosomal region 1p31.3-p31.2. The age of onset is variable. This disease is characterized by a severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus.	250,6
RPE65	Retinitis pigmentosa type 20	NM_000329.2	NM_000329.2:c.1022T>C, NM_000329.2:c.1087C>A, NM_000329.2:c.1102T>C, NM_000329.2:c.271C>T, NM_000329.2:c.1355T>G, NM_000329.2:c.1543C>T, NM_000329.2:c.394G>A, NM_000329.2:c.881A>C	Retinitis pigmentosa type 20 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RPE65 gene located on chromosomal region 1p31.3-p31.2. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	250,6
RPGR	Retinitis pigmentosa type 3	NM_001034853.1	NM_001034853.1:c.155-2A>G, NM_001034853.1:c.173_174insA, NM_001034853.1:c.179G>T, NM_001034853.1:c.296C>A, NM_001034853.1:c.389T>G, NM_001034853.1:c.505G>T, NM_001034853.1:c.517G>C, NM_001034853.1:c.642_656delTGGAGAACCTGAGAAinsC, NM_001034853.1:c.654_655delGA, NM_001034853.1:c.674_675delCC, NM_001034853.1:c.703C>T, NM_001034853.1:c.806G>A, NM_001034853.1:c.823G>A, NM_001034853.1:c.846_847delAA	Retinitis pigmentosa type 3 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the RPGR gene located on chromosomal region Xp11.4. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1:10,000-5:10,000.	600
RPGRIP1L	Joubert syndrome type 7	NM_015272.2	NM_015272.2:c.1177G>A, NM_015272.2:c.1326_1329delAAAA, NM_015272.2:c.1329_1330insA, NM_015272.2:c.1843A>C, NM_015272.2:c.1975T>C, NM_015272.2:c.2030C>T, NM_015272.2:c.2050C>T, NM_015272.2:c.2413C>T, NM_015272.2:c.757C>T, NM_015272.2:c.3548C>G, NM_015272.2:c.697A>T, NM_015272.2:c.3634_3637delGAAA, NM_015272.2:c.776+1G>A, NM_015272.2:c.2794_2795delTT	Joubert syndrome type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RPGRIP1L gene located on chromosomal region 16q12.2. The age of onset is early. This disease is characterized by cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease.	250,6
RPGRIP1L	Meckel syndrome type 5	NM_015272.2	NM_015272.2:c.394A>T, NM_015272.2:c.3706C>T, NM_015272.2:c.2614C>T	Meckel syndrome, type 5 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RPGRIP1L gene located on chromosomal region 16q12.2. The age of onset is early. This disease is characterized by a combination of renal cysts and variably associated features, including developmental anomalies of the central nervous system (usually occipital encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. The prevalence is <1:1,000,000.	250,6
RYR1	Central core disease	NM_000540.2	NM_000540.2:c.1021G>A, NM_000540.2:c.10343C>T, NM_000540.2:c.10579C>T, NM_000540.2:c.10616G>A, NM_000540.2:c.11798A>G, NM_000540.2:c.1205T>C, NM_000540.2:c.13480G>T, NM_000540.2:c.13513G>C, NM_000540.2:c.14365-2A>T, NM_000540.2:c.14511+1_14511+2delGT, NM_000540.2:c.14545G>A, NM_000540.2:c.1739_1742dupATCA, NM_000540.2:c.1841G>T, NM_000540.2:c.325C>T, NM_000540.2:c.4076delG, NM_000540.2:c.4178A>G, NM_000540.2:c.4405C>T, NM_000540.2:c.487C>T, NM_000540.2:c.5036G>A, NM_000540.2:c.5333C>A, NM_000540.2:c.5726_5727delAG, NM_000540.2:c.6082C>T, NM_000540.2:c.6104A>T, NM_000540.2:c.631+2T>C, NM_000540.2:c.6961A>G, NM_000540.2:c.7025A>G, NM_000540.2:c.7268T>A, NM_000540.2:c.7300G>A, NM_000540.2:c.7360C>T, NM_000540.2:c.7373G>A, NM_000540.2:c.738T>G, NM_000540.2:c.7463_7475delCAAAGATGTCAGC, NM_000540.2:c.9000+1G>T, NM_000540.2:c.14126C>T, NM_000540.2:c.1655G>A, NM_000540.2:c.4729G>A, NM_000540.2:c.7781C>A, NM_000540.2:c.7836-1G>A, NM_000540.2:c.8360C>G, NM_000540.2:c.9868G>A, NM_000540.2:c.9905_9906insC, NM_000540.2:c.1186G>T, NM_000540.2:c.6721C>T	Central core disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the RYR1 gene located on chromosomal region 19q13.2. Typical central core disease is a relatively mild congenital myopathy, usually characterized by motor developmental delay and signs of mild proximal weakness most pronounced in the hip girdle musculature. Orthopedic complications, particularly congenital dislocation of the hips and scoliosis, are common, and patients are at risk of having malignant hyperthermia. Onset is usually in childhood, although adult onset has also been reported.	250,6

SACS	Spastic ataxia, Charlevoix-Saguenay type	NM_014363.5	NM_014363.5:c.10907G>A, NM_014363.5:c.10954C>A, NM_014363.5:c.11624G>A, NM_014363.5:c.12160C>T, NM_014363.5:c.517C>T, NM_014363.5:c.6355C>T, NM_014363.5:c.6781C>A, NM_014363.5:c.7504C>T, NM_014363.5:c.8107C>T, NM_014363.5:c.8844delT, NM_014363.5:c.994A>T, NM_014363.5:c.13237C>T, NM_014363.5:c.3198T>A, NM_014363.5:c.4933C>T, NM_014363.5:c.5618_5619delAT, NM_014363.5:c.6563T>A	Spastic ataxia, Charlevoix-Saguenay type follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SACS gene located on chromosomal region 13q11. The age of onset is early. This disease is characterized by early-onset cerebellar ataxia with spasticity, a pyramidal syndrome and peripheral neuropathy. The prevalence is 1:1,500-1:2,000.	250,6
SAG	Oguchi disease	NM_000541.4	NM_000541.4:c.293_294insG, NM_000541.4:c.523C>T, NM_000541.4:c.577C>T, NM_000541.4:c.874C>T, NM_000541.4:c.916G>T, NM_000541.4:c.926delA, NM_000541.4:c.993C>G	Oguchi disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SAG gene located on chromosomal region 2q37. The age of onset is infantile. This disease is characterized by congenital stationary night blindness and the Mizuo-Nakamura phenomenon which is a unique morphological and functional abnormality of the retina that presents with a typical golden-yellow or silver-gray discoloration of the fundus in the presence of light that disappears after dark-adaptation and appears again after the onset of light.	250,6
SBDS	Shwachman-Diamond syndrome	NM_016038.2	NM_016038.2:c.120delG, NM_016038.2:c.127G>T, NM_016038.2:c.183_184delTAinsCT, NM_016038.2:c.184A>T, NM_016038.2:c.377G>C, NM_016038.2:c.505C>T, NM_016038.2:c.652C>T, NM_016038.2:c.258+2T>C	Shwachman-Diamond syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SBDS gene located on chromosomal region 7q11.21. The age of onset is infantile. This disease is characterized by chronic and usually mild neutropenia, pancreatic exocrine insufficiency associated with steatorrhea and growth failure, skeletal dysplasia with short stature, and an increased risk of bone marrow aplasia or leukemic transformation, cutaneous (eczema or ichthyosis) and dental anomalies, and psychomotor retardation. The prevalence is 1:76,000 newborn.	250,6
SBF2	Charcot-Marie-Tooth disease type 4B2	NM_030962.3	NM_030962.3:c.1459C>T, NM_030962.3:c.2875C>T, NM_030962.3:c.3586C>T, NM_030962.3:c.3154A>T, NM_030962.3:c.5539_5540insATCT	Charcot-Marie-Tooth disease, type 4B2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SBF2 gene located on chromosomal region 11p15.4. The age of onset is infantile. This disease is characterized by muscle weakness, sensory loss, reduced nerve conduction velocities, characteristic myelin outfoldings and a severe disease course.	600
SC5D	Lathosterolosis	NM_006918.4	NM_006918.4:c.86G>A	Lathosterolosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SC5D gene located on chromosomal region 11q23.3. The age of onset is early. This disease is characterized by malformations, intellectual deficit and liver disease. The prevalence is <1:1,000,000.	600
SCNN1A	Pseudohypoaldosteronism, type 1	NM_001038.5	NM_001038.5:c.1305delC, NM_001038.5:c.1482delC, NM_001038.5:c.1522C>T, NM_001038.5:c.1765C>T, NM_001038.5:c.1834C>T, NM_001038.5:c.203_204delTC, NM_001038.5:c.340G>A	Pseudohypoaldosteronism type 1, follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SCNN1A (12p13), SCNN1B (16p12.2-p12.1) and SCNN1G (16p12) genes. The age of onset is early. This disease is characterized by severe dehydration, vomiting and failure to thrive occurring in the first weeks of life, the clinical picture may be complicated by cardiac dysrhythmias, collapse, shock or cardiac arrest.	600
SCNN1B	Pseudohypoaldosteronism, type 1	NM_000336.2	NM_000336.2:c.109G>A	Pseudohypoaldosteronism type 1, follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SCNN1A (12p13), SCNN1B (16p12.2-p12.1) and SCNN1G (16p12) genes. The age of onset is early. This disease is characterized by severe dehydration, vomiting and failure to thrive occurring in the first weeks of life, the clinical picture may be complicated by cardiac dysrhythmias, collapse, shock or cardiac arrest.	250,6
SCNN1G	Pseudohypoaldosteronism, type 1	NM_001039.3	NM_001039.3:c.1373+2T>C, NM_001039.3:c.1570-1G>A, NM_001039.3:c.1627delG, NM_001039.3:c.598_599insA	Pseudohypoaldosteronism type 1, follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SCNN1A (12p13), SCNN1B (16p12.2-p12.1) and SCNN1G (16p12) genes. The age of onset is early. This disease is characterized by severe dehydration, vomiting and failure to thrive occurring in the first weeks of life, the clinical picture may be complicated by cardiac dysrhythmias, collapse, shock or cardiac arrest.	250,6
SEMA4A	Retinitis pigmentosa type 35	NM_022367.3	NM_022367.3:c.1033G>C, NM_022367.3:c.1049T>G	Retinitis pigmentosa type 35 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SEMA4A gene located on chromosomal region 1q22. The age of onset is variable. This disease is characterized by night blindness (nyctalopia) due to loss of rod function, often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife.	600

SEPN1	Muscular dystrophy, rigid spine, type 1	NM_020451.2	NM_020451.2:c.1315C>T, NM_020451.2:c.818G>A, NM_020451.2:c.883G>A, NM_020451.2:c.943G>A, NM_020451.2:c.943G>C, NM_020451.2:c.1384T>G, NM_020451.2:c.713_714insA, NM_020451.2:c.871C>T	Rigid spine syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SEPN1 gene located on chromosomal region 1p36.11. The age of onset is infantile. This disease is characterized by contractures of the spinal extensor muscles associated with abnormal posture (limitation of neck and trunk flexure), progressive scoliosis of the spine, early marked cervico-axial muscle weakness with relatively preserved strength and function of the extremities and progressive respiratory insufficiency. The prevalence is 3.5:100,000â€“5:100,000.	600
SERPINA1	Alpha1-antitrypsin deficiency	NM_000295.4	NM_000295.4:c.1177C>T, NM_000295.4:c.187C>T, NM_000295.4:c.194T>C, NM_000295.4:c.230C>T, NM_000295.4:c.250G>A, NM_000295.4:c.272G>A, NM_000295.4:c.347T>A, NM_000295.4:c.415G>A, NM_000295.4:c.514G>A, NM_000295.4:c.514G>T, NM_000295.4:c.739C>T, NM_000295.4:c.839A>T, NM_000295.4:c.1093G>A, NM_000295.4:c.848A>T	Alpha-1-antitrypsin deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SERPINA1 gene located on chromosomal region 14q32.13. The age of onset is variable. This disease is characterized by emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. The prevalence is 1:1,500-1:3,500 in European.	250,6
SETX	Spinocerebellar ataxia with axonal neuropathy type 2	NM_015046.5	NM_015046.5:c.1027G>T, NM_015046.5:c.1166T>C, NM_015046.5:c.1807A>G, NM_015046.5:c.2602C>T, NM_015046.5:c.3880C>T, NM_015046.5:c.4087C>T, NM_015046.5:c.5630delG, NM_015046.5:c.5927T>G, NM_015046.5:c.6848_6851delCAGA, NM_015046.5:c.994C>T, NM_015046.5:c.5308_5311delGAGA, NM_015046.5:c.5549-1G>T, NM_015046.5:c.6834_6839delAACAAA	Spinocerebellar ataxia with axonal neuropathy type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SETX gene located on chromosomal region 9q34.13. The age of onset is infantile. This disease is characterized by progressive cerebellar ataxia, axonal sensorimotor neuropathy with oculomotor apraxia, fixation instability, extrapyramidal features and an elevated serum alpha-fetoprotein level. The prevalence is 4:100,000-8:100,000.	250,6
SGCA	Limb-girdle muscular dystrophy type 2D	NM_000023.2	NM_000023.2:c.101G>A, NM_000023.2:c.229C>T, NM_000023.2:c.371T>C, NM_000023.2:c.518T>C, NM_000023.2:c.574C>T, NM_000023.2:c.850C>T, NM_000023.2:c.662G>A, NM_000023.2:c.739G>A, NM_000023.2:c.904_905insCC	Autosomal recessive limb-girdle muscular dystrophy type 2D follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SGCA gene located on chromosomal region 4q12. The age of onset is variable. This disease is characterized by limb-girdle weakness and calf pseudohypertrophy. The prevalence is 1:1,000,000-9:1,000,000.	250,6
SGCB	Muscular dystrophy, limb-girdle, type 2E	NM_000232.4	NM_000232.4:c.272G>C, NM_000232.4:c.272G>T, NM_000232.4:c.299T>A, NM_000232.4:c.323T>G, NM_000232.4:c.341C>T, NM_000232.4:c.452C>G, NM_000232.4:c.552T>G	Autosomal recessive limb-girdle muscular dystrophy type 2E follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SGCB gene located on chromosomal region 4q12. The age of onset is variable. This disease is characterized by limb-girdle weakness, particularly of the pelvic girdle muscles.	600
SGCG	Limb-girdle muscular dystrophy type 2C	NM_000231.2	NM_000231.2:c.195+4_195+7delAGTA, NM_000231.2:c.505+1G>A, NM_000231.2:c.787G>A, NM_000231.2:c.848G>A, NM_000231.2:c.88delG, NM_000231.2:c.521delT	Autosomal recessive limb-girdle muscular dystrophy type 2C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SGCG gene located on chromosomal region 13q12.12. The age of onset is variable. This disease is characterized by limb-girdle weakness, calf hypertrophy, diaphragmatic weakness, and variable cardiac abnormalities.	250,6
SGSH	Mucopolysaccharidosis type 3A (Sanfilippo disease type A)	NM_000199.3	NM_000199.3:c.1167C>A, NM_000199.3:c.1298G>A, NM_000199.3:c.130G>A, NM_000199.3:c.1339G>A, NM_000199.3:c.1380delT, NM_000199.3:c.197C>G, NM_000199.3:c.220C>T, NM_000199.3:c.235A>C, NM_000199.3:c.320delT, NM_000199.3:c.337_345delinsGCACAGGTGAG, NM_000199.3:c.364G>A, NM_000199.3:c.383C>T, NM_000199.3:c.416C>T, NM_000199.3:c.449G>A, NM_000199.3:c.466A>T, NM_000199.3:c.617G>C, NM_000199.3:c.752G>C, NM_000199.3:c.757delG, NM_000199.3:c.877C>T, NM_000199.3:c.892T>C	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SGSH gene located on chromosomal region 17q25.3. The age of onset is infantile. This disease is characterized by behavioural disorders (hyperkinesia, aggressiveness) and intellectual deterioration, sleep disorders and very mild dysmorphism. The prevalence is >1:70,000 newborn.	250,6
SH2D1A	Lymphoproliferative syndrome type 1, X-linked	NM_002351.4	NM_002351.4:c.163C>T, NM_002351.4:c.164G>T, NM_002351.4:c.172C>T, NM_002351.4:c.203C>T, NM_002351.4:c.302C>T, NM_002351.4:c.3G>T, NM_002351.4:c.95G>C	X-linked lymphoproliferative disease follows an X-linked pattern of inheritance and is caused by pathogenic variants in the SH2D1A gene located on chromosomal region Xq25. The age of onset is infantile. This disease is characterized by an inadequate immune response to infection with the Epstein-Barr virus: fulminant infectious mononucleosis, macrophage-activation syndrome or hemophagocytic lymphohistiocytosis (HLH) (see these terms), and/or progressive hypogammaglobulinemia and/or lymphomas. The prevalence is 1:1, 000,000 men.	600
SH3TC2	Charcot-Marie-Tooth disease type 4C	NM_024577.3	NM_024577.3:c.1586G>A, NM_024577.3:c.1747_1748delAG, NM_024577.3:c.1969G>A, NM_024577.3:c.1972C>T, NM_024577.3:c.1982T>C, NM_024577.3:c.217_227delGCTGCTCGGAGinsCCAGTAA, NM_024577.3:c.2191delG, NM_024577.3:c.2491_2492delAG, NM_024577.3:c.2710C>T, NM_024577.3:c.2829T>G, NM_024577.3:c.2860C>T, NM_024577.3:c.28delG, NM_024577.3:c.2993_2994insC, NM_024577.3:c.3325C>T, NM_024577.3:c.3326G>C, NM_024577.3:c.3341delC, NM_024577.3:c.3601C>T, NM_024577.3:c.3686A>T, NM_024577.3:c.505T>C, NM_024577.3:c.52+1delG, NM_024577.3:c.530-2A>G, NM_024577.3:c.735G>A, NM_024577.3:c.920G>A, NM_024577.3:c.3676-1G>A, NM_024577.3:c.1724T>A, NM_024577.3:c.53-1G>C	Charcot-Marie-Tooth disease, type 4C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SH3TC2 gene located on chromosomal region 5q32. The age of onset is infantile. This disease is characterized by scoliosis or kyphoscoliosis, neuropathy, foot deformities, respiratory insufficiency, hypoacousis and deafness.	250,6

SIL1	Marinesco-SjÃ¶gren syndrome	NM_022464.4	NM_022464.4:c.1312C>T, NM_022464.4:c.274C>T, NM_022464.4:c.331C>T	Marinesco-SjÃ¶gren syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SIL1 gene located on chromosomal region 5q31.2. The age of onset is infantile. This disease is characterized by dysarthria, nystagmus, muscle weakness and hypotonia. The prevalence is <1:1,000,000.	600
SIX6	Anophthalmia or microphthalmia, isolated	NM_007374.2	NM_007374.2:c.493A>G, NM_007374.2:c.532_536delAACCG, NM_007374.2:c.635C>T, NM_007374.2:c.725G>T	Anophthalmia or microphthalmia, isolated follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SIX6 gene located on chromosomal region 14q23.1. The age of onset is infantile. A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scarring of the retina and choroid, and other abnormalities may also be present.	600
SLC12A1	Bartter syndrome type 1	NM_000338.2	NM_000338.2:c.1875G>A, NM_000338.2:c.1942G>A, NM_000338.2:c.2805_2806insA, NM_000338.2:c.347G>A, NM_000338.2:c.611T>C, NM_000338.2:c.628+2T>C, NM_000338.2:c.814G>T, NM_000338.2:c.223C>T, NM_000338.2:c.2952_2955delCAAA	Bartter syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC12A1 gene located on chromosomal region 15q15-21. The age of onset is infantile. This disease is characterized by polyhydramnios, premature delivery, polyuria, dehydration, hypercalciuria and nephrocalcinosis. The prevalence is 1:1,000,000.	250,6
SLC12A6	Agenesis of the corpus callosum with neuropathy	NM_133647.1	NM_133647.1:c.1584_1585delCTinsG, NM_133647.1:c.2023C>T, NM_133647.1:c.3031C>T, NM_133647.1:c.619C>T, NM_133647.1:c.316+1G>A, NM_133647.1:c.366T>G	Corpus callosum agenesis with neuronopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC12A6 gene located on chromosomal region 15q13-q14. The age of onset is early. This disease is characterized by a delay in developmental milestones, a severe sensory-motor polyneuropathy with areflexia, a variable degree of agenesis of the corpus callosum, amyotrophy, hypotonia, and cognitive impairment. The prevalence is 1:2,117.	600
SLC17A5	Sialic acid storage disease	NM_012434.4	NM_012434.4:c.115C>T, NM_012434.4:c.406A>G, NM_012434.4:c.43G>T, NM_012434.4:c.918T>G, NM_012434.4:c.1259+1G>A, NM_012434.4:c.500T>C	Sialic acid storage diseases, follow an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC17A5 gene located on chromosomal region 6q13. The age of onset is from infantile to adult forms. The main symptoms are hypotonia, cerebellar ataxia, and mental retardation; visceromegaly and coarse features are also present in the infantile cases.	250,6
SLC24A1	Night blindness, congenital stationary type 1D	NM_004727.2	NM_004727.2:c.1963C>T	Night blindness, congenital stationary type 1D follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC24A1 gene located on chromosomal region 15q22.31. The age of onset is early. This disease is characterized by hemeralopia with a moderate loss of visual acuity.	250,6
SLC25A13	Citrullinemia type 2	NM_014251.2	NM_014251.2:c.1078C>T, NM_014251.2:c.1177+1G>A, NM_014251.2:c.1311+1G>A, NM_014251.2:c.1592G>A, NM_014251.2:c.1799dupA, NM_014251.2:c.1801G>A, NM_014251.2:c.1801G>T, NM_014251.2:c.1813C>T, NM_014251.2:c.615+1G>C, NM_014251.2:c.615+5G>A, NM_014251.2:c.674C>A, NM_014251.2:c.674C>T, NM_014251.2:c.852_855delTATG, NM_014251.2:c.1231-1G>A, NM_014251.2:c.1411_1412delCT	Citrullinemia type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC25A13 gene located on chromosomal region 7q21.3. The age of onset is early. This disease is characterized by hyperammonemia and associated neuropsychiatric symptoms such as nocturnal delirium, confusion, restlessness, disorientation, drowsiness, memory loss, abnormal behavior (aggression, irritability, and hyperactivity), seizures, and coma. The prevalence is 1:17,000-1:230,000.	600
SLC25A15	Hyperornithinemia - hyperammonemia - homocitrullinuria syndrome	NM_014252.3	NM_014252.3:c.110T>G, NM_014252.3:c.212T>A, NM_014252.3:c.535C>T, NM_014252.3:c.538G>A, NM_014252.3:c.562_564delTTC, NM_014252.3:c.569G>A, NM_014252.3:c.658G>A, NM_014252.3:c.815C>T, NM_014252.3:c.824G>A, NM_014252.3:c.44C>T	Hyperornithinemia-hyperammonemia-homocitrullinuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC25A15 gene located on chromosomal region 13q14.11. The age of onset is early. This disease is characterized by coma due to hyperammonemia, convulsions, and hypotonia. The prevalence is 1:5,500.	600
SLC25A22	Epileptic encephalopathy, early infantile, type 3	NM_024698.5	NM_024698.5:c.617C>T, NM_024698.5:c.706G>T	Early infantile epileptic encephalopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC25A22 gene located on chromosomal region 11p15.5. The age of onset is early. This disease is characterized by the onset of tonic spasms within the first 3 months of life leading to psychomotor impairment and death. The prevalence is <1:1,000,000.	600
SLC26A2	Achondrogenesis type 1B	NM_000112.3	NM_000112.3:c.1020_1022delTGT, NM_000112.3:c.1273A>G, NM_000112.3:c.532C>T, NM_000112.3:c.2033G>T	Achondrogenesis type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A2 gene located on chromosomal region 5q32. The age of onset is early. This disease is characterized by severe micromelia with very short fingers and toes, a flat face, a short neck, thickened soft tissue around the neck, hypoplasia of the thorax, protuberant abdomen, a hydropic fetal appearance and distinctive histological features of the cartilage. The prevalence is 1:20,000.	250,6

SLC26A2	Atelosteogenesis type 2	NM_000112.3	NM_000112.3:c.1535C>A, NM_000112.3:c.835C>T	Atelosteogenesis type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A2 gene located on chromosomal region 5q32. The age of onset is early. This disease is characterized by limb shortening, normal sized skull with cleft palate, hitchhiker thumbs, distinctive facial dysmorphism and radiographic skeletal features. The prevalence is 1:20,000.	250,6
SLC26A2	Diastrophic dysplasia	NM_000112.3	NM_000112.3:c.1724delA, NM_000112.3:c.1878delG, NM_000112.3:c.1361A>C, NM_000112.3:c.767T>C, NM_000112.3:c.833delC, NM_000112.3:c.496G>A, NM_000112.3:c.1957T>A	Diastrophic dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A2 gene located on chromosomal region 5q32. The age of onset is early. This disease is characterized by short stature with short extremities (final adult height is 120cm), and joint malformations leading to multiple joint contractures (principally involving the shoulders, elbows, interphalangeal joints and hips). The prevalence is 1:20,000.	250,6
SLC26A4	Deafness type 4, autosomal recessive	NM_000441.1	NM_000441.1:c.1001G>T, NM_000441.1:c.1034T>A, NM_000441.1:c.2162C>T, NM_000441.1:c.1975G>C, NM_000441.1:c.1174A>T, NM_000441.1:c.2131G>A, NM_000441.1:c.1454C>T, NM_000441.1:c.1468A>C, NM_000441.1:c.2211G>C, NM_000441.1:c.269C>T, NM_000441.1:c.916dupG, NM_000441.1:c.281C>T, NM_000441.1:c.1634T>G, NM_000441.1:c.1707+5G>A, NM_000441.1:c.1489G>A, NM_000441.1:c.961A>T, NM_000441.1:c.2048T>C, NM_000441.1:c.898A>C, NM_000441.1:c.918+2T>C, NM_000441.1:c.1001+1G>T, NM_000441.1:c.970A>T, NM_000441.1:c.563T>C	Autosomal recessive nonsyndromic sensorineural deafness type DFNB4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A4 gene located on chromosomal region 7q22.3. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	250,6
SLC26A4	Pendred syndrome	NM_000441.1	NM_000441.1:c.1246A>C, NM_000441.1:c.1826T>G, NM_000441.1:c.1229C>T, NM_000441.1:c.1263+1G>A, NM_000441.1:c.1061T>C, NM_000441.1:c.1790T>C, NM_000441.1:c.2168A>G, NM_000441.1:c.1151A>G, NM_000441.1:c.1226G>A, NM_000441.1:c.1003T>C, NM_000441.1:c.919-2A>G, NM_000441.1:c.554G>C, NM_000441.1:c.626G>T, NM_000441.1:c.1334T>G, NM_000441.1:c.1198delT, NM_000441.1:c.412G>T, NM_000441.1:c.707T>C	Pendred syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A4 gene located on chromosomal region 7q22.3. The age of onset is early. The main presenting clinical sign is prelingual sensorineural deafness, although occasionally the hearing loss develops later in childhood. The degree of hearing loss is variable: it can be mild-to-moderate and progressive in some patients, and severe-to-profound in others. Fluctuations in hearing are also common and may be associated with or preceded by vertigo. The onset and presentation of euthyroid goiter (75%) is highly variable within and between families, with thyroid enlargement usually developing in late childhood or early adulthood. The thyromegaly reflects a defect in iodide transport from the thyrocyte to the colloid, although organification itself is not impaired. Hypothyroidism may develop if nutritional iodide intake is low.	250,6
SLC26A5	Deafness type 61, autosomal recessive	NM_198999.2	NM_198999.2:c.209G>A, NM_198999.2:c.390A>C, NM_198999.2:c.152+1G>A, NM_198999.2:c.1A>G	Autosomal recessive nonsyndromic sensorineural deafness type DFNB16 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC26A5 gene located on chromosomal region 7q22.1. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	600
SLC35A1	Congenital disorder of glycosylation type 2F	NM_006416.4	NM_006416.4:c.277_280delGTGCinsTG	Congenital disorder of glycosylation type 2F follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC35A1 gene located on chromosomal region 6q15. The age of onset is early. This disease is characterized by repeated hemorrhagic incidents, including severe pulmonary hemorrhage.	600
SLC35C1	Congenital disorder of glycosylation 2c	NM_018389.4	NM_018389.4:c.439C>T, NM_018389.4:c.91G>T, NM_018389.4:c.923C>G, NM_018389.4:c.290dupG, NM_018389.4:c.503_505delTCT	Congenital disorder of glycosylation type 2c follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC35C1 gene located on chromosomal region 11p11.2. The age of onset is infantile. This disease is characterized by recurrent bacterial infections, severe growth delay and severe intellectual deficit.	600
SLC35D1	Schneckenbecken dysplasia	NM_015139.2	NM_015139.2:c.319C>T, NM_015139.2:c.932G>A	Schneckenbecken dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC35D1 gene located on chromosomal region 1p31.3. The age of onset is fetal. This disease is characterized by nail-like configuration of the hypoplastic iliac bone, flattened hypoplastic vertebral bodies, short ribs, short and wide fibulae, short and broad long bones with a dumbbell-like appearance, and precocious ossification of the tarsus.	600

SLC37A4	Glycogen storage disease types 1b, 1c and 1d	NM_001164278.1	NM_001164278.1:c.1042_1043delCT, NM_001164278.1:c.1081G>T, NM_001164278.1:c.1082G>A, NM_001164278.1:c.1108_1109delCT, NM_001164278.1:c.1129G>T, NM_001164278.1:c.1190-2_1190-1delAG, NM_001164278.1:c.1309C>T, NM_001164278.1:c.287G>A, NM_001164278.1:c.352T>C, NM_001164278.1:c.593A>T, NM_001164278.1:c.706_708delGTG, NM_001164278.1:c.83G>A, NM_001164278.1:c.899G>A	Glycogen storage disease due to glucose-6-phosphatase deficiency types 1b, 1c and 1d follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC37A4 gene located on chromosomal region 11q23. The age of onset is early. This disease is characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia. Glycogen storage disease type 1B patients also present a tendency towards infections associated with neutropenia, relapsing aphthous gingivostomatitis, and inflammatory bowel disease. The incidence is 1:100,000.	250,6
SLC45A2	Albinism, oculocutaneous, type 4	NM_016180.3	NM_016180.3:c.1121delT, NM_016180.3:c.469G>A, NM_016180.3:c.986delC	Oculocutaneous albinism type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC45A2 gene located on chromosomal region 5p13.2. The age of onset is early. This disease is characterized by skin and hair hypopigmentation, numerous ocular changes and misrouting of the optic nerves at the chiasm. The prevalence is 1:100.000.	600
SLC4A11	Congenital hereditary endothelial dystrophy type 2	NM_032034.3	NM_032034.3:c.1038_1039insA, NM_032034.3:c.1391G>A, NM_032034.3:c.2318C>T, NM_032034.3:c.1466C>T, NM_032034.3:c.1813C>T, NM_032034.3:c.2264G>A, NM_032034.3:c.2605C>T, NM_032034.3:c.2399C>T, NM_032034.3:c.554_561delGCTTCGCC, NM_032034.3:c.2606G>A	Congenital hereditary endothelial dystrophy type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC4A11 gene located on chromosomal region 20p13. The age of onset is early. This disease is characterized by a diffuse ground-glass appearance of the corneas and marked corneal thickening from birth with nystagmus, and blurred vision.	250,6
SLC4A11	Corneal dystrophy and perceptive deafness	NM_032034.3	NM_032034.3:c.2528T>C, NM_032034.3:c.1463G>A, NM_032034.3:c.473_480delGCTTCGCC, NM_032034.3:c.2566A>G, NM_032034.3:c.637T>C, NM_032034.3:c.625C>T, NM_032034.3:c.2224G>A, NM_032034.3:c.2240_2240+1insTATGACAC	Corneal dystrophy - perceptive deafness follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLC4A11 gene located on chromosomal region 20p13. The age of onset is early. This disease is characterized by the association of congenital hereditary endothelial dystrophy with progressive, postlingual sensorineural hearing loss.	250,6
SLC6A8	Cerebral creatine deficiency syndrome type 1	NM_005629.3	NM_005629.3:c.1011C>G, NM_005629.3:c.1141G>C, NM_005629.3:c.1222_1224delTTC, NM_005629.3:c.1540C>T, NM_005629.3:c.321_323delCTT, NM_005629.3:c.395G>T	X-linked creatine transporter deficiency follows an X-linked pattern of inheritance and is caused by pathogenic variants in the SLC6A8 gene located on chromosomal region Xq28. The age of onset is infantile. This disease is characterized by intellectual deficit, seizures, severe speech delay and sometimes midface hypoplasia, microcephaly, long, thin face, and prominent chin in the cases of affected male patients reported to date. The prevalence is 11:1,000.	600
SLX4	Fanconi anemia, complementation group P	NM_032444.2	NM_032444.2:c.1093delC, NM_032444.2:c.286delA, NM_032444.2:c.4921_4922insG, NM_032444.2:c.5097_5098delTC, NM_032444.2:c.5408_5409insAC, NM_032444.2:c.4739+1G>T, NM_032444.2:c.2808_2809delAG	Fanconi anemia complementation group P follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SLX4 gene located on chromosomal region 16p13.3. The age of onset is early. This disease is characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors. The prevalence is 1:160,000.	250,6
SMN1	Spinal muscular atrophy	-	del ex7, del ex7-8, del ex8 (Detection by MLPA)	Spinal muscular atrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SMN1 gene located on chromosomal region 5q13.2. The age of onset is variable. This disease comprise a group of neuromuscular disorders characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7-skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of patients bear one SMN1 copy with an intragenic mutation. Type 1 is a severe form, with onset before 6 months of age. Patients never achieve the ability to sit. Type 2 has intermediate severity, with onset between 6 and 18 months. Patients do not reach the motor milestone of standing, and survive into adulthood. Type 3 onset is after 18 months. Patients develop ability to stand and walk and survive into adulthood. Type 4 onset is in adulthood, disease progression is slow, and patients can stand and walk. The incidence is 1:10,000 and the prevalence is 1:80,000.	250,6

SMPD1	Niemann-Pick disease	NM_000543.4	NM_000543.4:c.103_118delCTGGTGCTGGCGCTGG, NM_000543.4:c.103_119delCTGGTGCTGGCGCTGGC, NM_000543.4:c.103_107delCTGGT, NM_000543.4:c.103_113delCTGGTGCTGGCGinsCTGGTG, NM_000543.4:c.1092-1G>C, NM_000543.4:c.1117C>T, NM_000543.4:c.106delG, NM_000543.4:c.108_124delGCTGGCGCTGGCGCTGGC, NM_000543.4:c.1267C>T, NM_000543.4:c.1299T>G, NM_000543.4:c.1327C>T, NM_000543.4:c.1420_1421delCT, NM_000543.4:c.1426C>T, NM_000543.4:c.1624C>T, NM_000543.4:c.1630delA, NM_000543.4:c.1805G>A, NM_000543.4:c.354delC, NM_000543.4:c.475T>C, NM_000543.4:c.551C>T, NM_000543.4:c.557C>T, NM_000543.4:c.558_559insC, NM_000543.4:c.558_574delGCCCCCCAAACCCCCTA, NM_000543.4:c.564delC, NM_000543.4:c.573delT, NM_000543.4:c.689G>A, NM_000543.4:c.730G>A, NM_000543.4:c.739G>A, NM_000543.4:c.740delG, NM_000543.4:c.742G>A, NM_000543.4:c.757G>C, NM_000543.4:c.785_807delTGTTGAGTGGGCTGGGCCCAGCC, NM_000543.4:c.788T>A, NM_000543.4:c.842_849dupTCCCCGCA, NM_000543.4:c.911T>C, NM_000543.4:c.940G>A, NM_000543.4:c.96G>A, NM_000543.4:c.996delC, NM_000543.4:c.688C>T, NM_000543.4:c.995C>G, NM_000543.4:c.1829_1831delGCC, NM_000543.4:c.1264-1G>T, NM_000543.4:c.1152G>A	Niemann-Pick disease follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SMPD1 gene located on chromosomal region 11p15.4. The clinical phenotype ranges from a severe infantile form with neurologic degeneration resulting in death usually by 3 years of age (type A) to a later-onset nonneurologic form (type B) that is compatible with survival into adulthood.	250,6
SNAI2	Waardenburg syndrome type 2	NM_003068.4	NM_003068.4:c.357C>A	Waardenburg syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SNAI2 gene located on chromosome 8q11.21. The age of onset is early. This disease is characterized by pigmentary abnormalities of the hair, skin, and eyes, congenital sensorineural hearing loss and the lateral displacement of the inner canthus of each eye. The prevalence is 1:100,000-9:100,000.	600
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	NM_004782.3	NM_004782.3:c.487dupA	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SNAP29 gene located on chromosomal region 22q11.2. The age of onset is early. This disease is characterized by severe developmental abnormalities of the nervous system and aberrant differentiation of the epidermis. The prevalence is <1:1,000,000.	600
SPG11	Spastic paraplegia type 11	NM_025137.3	NM_025137.3:c.118C>T, NM_025137.3:c.529_533delATATT, NM_025137.3:c.5623C>T, NM_025137.3:c.1339_1342dupGGCT, NM_025137.3:c.342delT, NM_025137.3:c.7152-1G>C, NM_025137.3:c.733_734delAT, NM_025137.3:c.6805_6806delCT, NM_025137.3:c.1736-1G>C, NM_025137.3:c.6100C>T, NM_025137.3:c.6848_6849insTC	Spastic paraplegia type 11 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SPG11 gene located on chromosomal region 13q13.3. The age of onset is infantile. This disease is characterized by progressive spasticity and weakness of the lower limbs frequently associated with the following: mild intellectual disability with learning difficulties in childhood and/or progressive cognitive decline; peripheral neuropathy; pseudobulbar involvement; and increased reflexes in the upper limbs. The prevalence is 5:100.000.	250,6
SPG20	Spastic paraplegia type 20, autosomal recessive	NM_015087.4	NM_015087.4:c.1110delA, NM_015087.4:c.364_365delAT	Autosomal recessive spastic paraplegia type 20 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SPG20 gene located on chromosomal region 13q13.3. The age of onset is infantile. This disease is characterized by progressive spastic paraparesis, dysarthria, and pseudobulbar palsy; distal amyotrophy; motor and cognitive delays; short stature; and subtle skeletal abnormalities.	600
SPG7	Spastic paraplegia type 7	NM_003119.3	NM_003119.3:c.1457G>A, NM_003119.3:c.1529C>T, NM_003119.3:c.2075G>C, NM_003119.3:c.233T>A, NM_003119.3:c.1676delA, NM_003119.3:c.1749G>C, NM_003119.3:c.773_774delTG, NM_003119.3:c.1045G>A, NM_003119.3:c.1124delG, NM_003119.3:c.679C>T, NM_003119.3:c.758+2T>C, NM_003119.3:c.286+1G>T	Spastic paraplegia type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SPG7 gene located on chromosomal region 16q24.3. The age of onset is adult. This disease is characterized by insidiously progressive bilateral lower limb weakness and spasticity. The prevalence is 1:100,000-9:100,000.	250,6
STAR	Lipoid adrenal hyperplasia	NM_000349.2	NM_000349.2:c.545G>T, NM_000349.2:c.559G>A, NM_000349.2:c.545G>A, NM_000349.2:c.749G>A, NM_000349.2:c.772C>T, NM_000349.2:c.562C>T, NM_000349.2:c.577C>T	Congenital lipoid adrenal hyperplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the STAR gene located on chromosomal region 8p11.23. The age of onset is early. This disease is characterized by severe adrenal insufficiency and sex reversal in males. The prevalence is 1:300,000.	600

STIL	Microcephaly primary, type 7, autosomal recessive	NM_003035.2	NM_003035.2:c.3655delG, NM_003035.2:c.3715C>T, NM_003035.2:c.3843_3846delACAG, NM_003035.2:c.2392T>G, NM_003035.2:c.2826+1G>A	Autosomal recessive primary microcephaly type 7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the STIL gene located on chromosomal region 1p33. The age of onset is early. This disease is characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment. The incidence is 1:1,000,000.	600
STRA6	Syndromic microphthalmia type 9	NM_022369.3	NM_022369.3:c.1678G>C, NM_022369.3:c.1699C>T, NM_022369.3:c.147delC, NM_022369.3:c.1521-1G>A, NM_022369.3:c.1964G>A, NM_022369.3:c.277_278insCC, NM_022369.3:c.1931C>T, NM_022369.3:c.1963C>T, NM_022369.3:c.69G>A, NM_022369.3:c.878C>T, NM_022369.3:c.910_911delGGinsAA, NM_022369.3:c.52_53delTAinsC, NM_022369.3:c.527_528insG	Syndromic microphthalmia type 9 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the STRA6 gene located on chromosomal region 15q24.1. The age of onset is early. This disease is characterized by anophthalmia or severe microphthalmia, and pulmonary hypoplasia or aplasia. The prevalence is 1:10,000.	600
STRC	Deafness type 16, autosomal recessive	NM_153700.2	NM_153700.2:c.4561_4562insC, NM_153700.2:c.5188C>T, NM_153700.2:c.3556C>T, NM_153700.2:c.5168_5171delTTCT, NM_153700.2:c.5185C>T, NM_153700.2:c.4545+1G>C	Autosomal recessive nonsyndromic sensorineural deafness type DFNB16 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the STRC gene located on chromosomal region 15q15.3. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	250,6
SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria	NM_003849.3	NM_003849.3:c.152_153delAT, NM_003849.3:c.626C>A	Fatal infantile lactic acidosis with methylmalonic aciduria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SUCLG1 gene located on chromosomal region 2p11.2. The age of onset is early. This disease is characterized by polypnea, severe hypotonia, lethargy, and vomiting, after a silent period during which the children were considered as normal. Facial dysmorphism and cerebral malformations may be noted, as well as diverse organ involvement such as hypertrophic myocardiopathy, tubulopathy, or liver insufficiency.	600
SUOX	Sulfocysteinuria	NM_000456.2	NM_000456.2:c.37C>T, NM_000456.2:c.894_895delCT, NM_000456.2:c.650G>A, NM_000456.2:c.794C>A	Sulfocysteinuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SUOX gene located on chromosome 12q13.13. The age of onset is early. This disease is characterized by seizures, reduced muscle tone, psychomotor retardation, lens dislocation.	600
TAF1	Dystonia-Parkinsonism, X-linked	NM_004606.4	NM_004606.4:c.417_418insCATAATCTATGATAATGATAAT	X-linked dystonia-parkinsonism follows an X-linked pattern of inheritance and is caused by pathogenic variants in the TAF1 gene located on chromosomal region Xq13.1. The age of onset is adult. This disease is characterized by adult-onset Parkinsonism that is frequently accompanied by focal dystonia, which becomes generalized over time, and that has a highly variable clinical course. The prevalence is 1:300,000.	600
TAT	Tyrosinemia type 2	NM_000353.2	NM_000353.2:c.1249C>T, NM_000353.2:c.236-5A>G, NM_000353.2:c.668C>G, NM_000353.2:c.1297C>T, NM_000353.2:c.169C>T	Tyrosinemia type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TAT gene located on chromosomal region 16q22.1. The age of onset is early. This disease is characterized by hypertyrosinemia with oculocutaneous manifestations and, in some cases, intellectual deficit. The prevalence is 1:100,000-1:120,000 newborn.	600
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	NM_003193.4	NM_003193.4:c.1491_1492insGTAAA	Hypoparathyroidism - retardation - dysmorphism syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TBCE gene located on chromosomal region 1q42.3. The age of onset is early. This disease is characterized by congenital hypoparathyroidism, growth retardation, intellectual deficit, seizures, and dysmorphic features including microcephaly, facial, ocular and dental abnormalities, and short hands and feet.	600
TCAP	Cardiomyopathy, hypertrophic, type 25	NM_003673.3	NM_003673.3:c.260G>A, NM_003673.3:c.316C>T	Cardiomyopathy, hypertrophic, type 25 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TCAP gene located on chromosomal region 17q12. The age of onset is variable. This disease is characterized by dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death.	250,6
TCAP	Limb-girdle muscular dystrophy type 2G	NM_003673.3	NM_003673.3:c.157C>T	Autosomal recessive limb-girdle muscular dystrophy type 2G follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TCAP gene located on chromosomal region 17q12. The age of onset is variable. This disease is characterized by muscle weakness in the four limbs, mild scapular winging, severe atrophy of the quadriceps and anterior tibialis muscles, calf hypertrophy, and lack of respiratory and cardiac involvement.	250,6

TCIRG1	Osteopetrosis type 1, autosomal recessive	NM_006019.3	NM_006019.3:c.1331G>T, NM_006019.3:c.1674-1G>A, NM_006019.3:c.179A>G, NM_006019.3:c.2236+1G>A, NM_006019.3:c.2415-3C>G, NM_006019.3:c.112_113delAG, NM_006019.3:c.1213G>A	Autosomal recessive osteopetrosis type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TCIRG1 gene located on chromosomal region 11q13.2. The age of onset is early. This disease is characterized by bone marrow failure, fractures and visual impairment. The incidence is 1:200.000 live births and the prevalence is 1:250,000.	250,6
TECTA	Deafness type 21, autosomal recessive	NM_005422.2	NM_005422.2:c.2428C>T, NM_005422.2:c.2941+1G>A, NM_005422.2:c.651_652insC, NM_005422.2:c.4370_4371insTCAGTGCGACCCGC, NM_005422.2:c.4601G>A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB21 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TECTA gene located on chromosomal region 11q23.3. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	600
TERT	Dyskeratosis congenita, autosomal recessive	NM_198253.2	NM_198253.2:c.1234C>T, NM_198253.2:c.835G>A, NM_198253.2:c.2701C>T, NM_198253.2:c.2431C>T	Dyskeratosis congenita, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TERT gene located on chromosomal region 5p15.33. The age of onset is early. This disease has a wide phenotypic spectrum and age onset. It usually manifests during childhood with the triad of dysplastic nails, lacy reticular pigmentation and atrophy of the skin at the level of the neck and upper chest, and oral leukoplakia. Patients show an increased risk for progressive bone marrow failure and may develop myelodysplastic syndrome or acute myelogenous leukemia at any age (the risk increasing with age). There is also an increased risk for solid tumors, typically squamous cell carcinoma of head and neck (see this term) or anogenital cancer. Various additional clinical findings have been reported and may include: developmental delay, short stature, microcephaly, blepharitis, epiphora, periodontal disease, taurodontism, decreased teeth/root ratio, esophageal stenosis, liver disease, urethral stenosis, osteoporosis, avascular necrosis of femur and/or humerus, premature hair greying/alopecia, or abnormal eyelashes. Individuals with DC are at high risk of pulmonary fibrosis. The prevalence is 1:1.000.000.	250,6
TFR2	Hemochromatosis, type 3	NM_003227.3	NM_003227.3:c.1330G>A, NM_003227.3:c.1403G>A, NM_003227.3:c.1469T>G, NM_003227.3:c.1235_1237delACA, NM_003227.3:c.1861_1872delGCCGTGGCCCAG, NM_003227.3:c.2343G>A, NM_003227.3:c.313C>T, NM_003227.3:c.1665delC, NM_003227.3:c.750C>G, NM_003227.3:c.840C>G, NM_003227.3:c.949C>T, NM_003227.3:c.515T>A, NM_003227.3:c.1632_1633delGA, NM_003227.3:c.2014C>T, NM_003227.3:c.2374G>A, NM_003227.3:c.1473+1G>A, NM_003227.3:c.1186C>T	Hemochromatosis type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TFR2 gene located on chromosomal region 7q22.1. The age of onset is adult. This disease is characterized by excessive tissue iron deposition of genetic origin, liver disease, hypogonadism, arthritis, diabetes and skin pigmentation. The prevalence is <1:1,000,000.	250,6
TH	Segawa syndrome, autosomal recessive	NM_000360.3	NM_000360.3:c.1141C>A, NM_000360.3:c.614T>C, NM_000360.3:c.733A>C, NM_000360.3:c.1388C>T, NM_000360.3:c.605G>A, NM_000360.3:c.917G>A	Segawa syndrome, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TH gene located on chromosomal region 11p15.5. This disease is characterized by dystonia presenting in infancy or early childhood. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Some cases present with parkinsonian symptoms in infancy. Unlike all other forms of dystonia, it is an eminently treatable condition, due to a favorable response to L-DOPA. The prevalence is 1:1,000,000-9:1,000,000.	600
TIMM8A	Mohr-Tranebjaerg syndrome	NM_004085.3	NM_004085.3:c.198C>G, NM_004085.3:c.238C>T, NM_004085.3:c.112C>T	Mohr-Tranebjaerg syndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the TIMM8A gene located on chromosomal region Xq22. The age of onset is infantile. This disease is characterized by hearing loss, followed by adolescent onset progressive dystonia or ataxia, visual impairment from early adulthood onwards and dementia from the 4th decade onwards. The prevalence is <1:1,000,000.	600
TK2	Mitochondrial DNA depletion syndrome type 2	NM_004614.4	NM_004614.4:c.323C>T, NM_004614.4:c.361C>A, NM_004614.4:c.373C>T, NM_004614.4:c.500G>A, NM_004614.4:c.604_606delAAG, NM_004614.4:c.635T>A, NM_004614.4:c.623A>G, NM_004614.4:c.159C>G, NM_004614.4:c.268C>T	Mitochondrial DNA depletion syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TK2 gene located on chromosomal region 16q21. The age of onset is infantile. This disease is characterized by generalized hypotonia, proximal muscle weakness, loss of previously acquired motor skills, poor feeding, and respiratory difficulties leading to respiratory failure and death within a few years after diagnosis. The prevalence is 1.2:100,000.	250,6
TMC1	Deafness type 7, autosomal recessive	NM_138691.2	NM_138691.2:c.1763+3A>G, NM_138691.2:c.1842G>A, NM_138691.2:c.100C>T, NM_138691.2:c.1165C>T, NM_138691.2:c.425G>A, NM_138691.2:c.454-1G>C, NM_138691.2:c.1960A>G	Autosomal recessive nonsyndromic sensorineural deafness type DFNB7 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMC1 gene located on chromosomal region 9q21.13. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	600

TMEM216	Joubert syndrome type 2	NM_001173990.2	NM_001173990.2:c.218G>T, NM_001173990.2:c.218G>A, NM_001173990.2:c.79_82delAACG	Joubert syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMEM216 gene located on chromosomal region 11q12.2. The age of onset is early. This disease is characterized by the neurological features associated with both renal and ocular disease; retinal involvement (manifesting with either Leber congenital amaurosis or progressive retinal dystrophy) and nephronophthisis (usually juvenile). The prevalence is 1:80,000-1:100,000.	600
TMEM216	Meckel syndrome type 2	NM_001173990.2	NM_001173990.2:c.230G>C, NM_001173990.2:c.253C>T, NM_001173990.2:c.341T>G	Meckel syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMEM216 gene located on chromosomal region 11q12.2. The age of onset is antenatal. It is a disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. The prevalence is 1:80,000-1:100,000.	600
TMEM67	COACH syndrome	NM_153704.5	NM_153704.5:c.1769T>C, NM_153704.5:c.2498T>C	COACH syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMEM67 gene located on chromosomal region 8q22. The age of onset is variable. This disease is characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Other features, such as coloboma and renal cysts, may be variable.	250,6
TMEM67	Joubert syndrome type 6	NM_153704.5	NM_153704.5:c.130C>T, NM_153704.5:c.148_149insTAAT, NM_153704.5:c.1538A>G	Joubert syndrome type 6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMEM67 gene located on chromosomal region 8q22. The age of onset is infantile. This disease is characterized by an irregular breathing pattern (episodic tachypnea and/or apnea), and nystagmus. During infancy, hypotonia may appear. Cerebellar ataxia (staggering gait and imbalance) may develop later. Delayed acquisition of motor milestones is common. Cognitive abilities are variable, ranging from severe intellectual deficit to normal intelligence. Neuro-ophthalmologic examination may show oculomotor apraxia. In some cases, seizures occur. Careful examination of the face shows a characteristic appearance: large head, prominent forehead, high rounded eyebrows, epicanthal folds, ptosis (occasionally), an upturned nose with prominent nostrils, an open mouth (which tends to have an oval shape early on, a 'rhomboid' appearance later, and finally can appear triangular with downturned angles), tongue protrusion and rhythmic tongue motions, and occasionally low-set and tilted ears. Other features sometimes present in Joubert syndrome include retinal dystrophy, nephronophthisis, and polydactyly. The prevalence is 1:80,000-1:100,000.	250,6
TMEM67	Meckel syndrome type 3	NM_153704.5	NM_153704.5:c.1309C>G, NM_153704.5:c.755T>C, NM_153704.5:c.1046T>C, NM_153704.5:c.653G>C, NM_153704.5:c.406+1402_406+1403insTAAT, NM_153704.5:c.622A>T	Meckel syndrome type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMEM67 gene located on chromosomal region 8q22. The age of onset is variable. This disease is characterized multiple congenital anomaly disorder characterized by the triad of brain malformation mainly occipital encephalocele (see this term), large polycystic kidneys, and polydactyly as well as associated abnormalities that may include cleft lip/palate (see these terms), cardiac and genital anomalies, central nervous system (CNS) malformations, liver fibrosis, and bone dysplasia.	250,6
TMIE	Deafness type 6, autosomal recessive	NM_147196.2	NM_147196.2:c.241C>T, NM_147196.2:c.250C>T, NM_147196.2:c.170G>A, NM_147196.2:c.257G>A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB6 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMIE gene located on chromosomal region 21q22.3. The age of onset is early. This disease is characterized by hearing loss and deafness.	600
TMPRSS3	Deafness types 8/10, autosomal recessive	NM_024022.2	NM_024022.2:c.1211C>T, NM_024022.2:c.1276G>A, NM_024022.2:c.1159G>A, NM_024022.2:c.413C>A, NM_024022.2:c.446+1G>T, NM_024022.2:c.647G>T, NM_024022.2:c.753G>C, NM_024022.2:c.646C>T, NM_024022.2:c.208delC, NM_024022.2:c.242C>G	Autosomal recessive nonsyndromic sensorineural deafness type DFNB10 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TMPRSS3 gene located on chromosomal region 21q22.3. The age of onset is early. This disease is characterized by hearing loss and deafness.	250,6
TNNT1	Nemaline myopathy type 5	NM_003283.5	NM_003283.5:c.538G>T	Nemaline myopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TNNT1 gene located on chromosomal region 19q13.4. The age of onset is from birth to adulthood. This disease is characterized by hypotonia, weakness and depressed or absent deep tendon reflexes, with pathologic evidence of nemaline bodies (rods) on muscle biopsy. The prevalence is 1:50,000 newborn.	600

TPP1	Neuronal ceroid-lipofuscinoses type 2	NM_000391.3	NM_000391.3:c.1093T>C, NM_000391.3:c.616C>T, NM_000391.3:c.622C>T, NM_000391.3:c.1340G>A, NM_000391.3:c.141_144delGAGT, NM_000391.3:c.827A>T, NM_000391.3:c.509-1G>C, NM_000391.3:c.851G>T	Neuronal ceroid lipofuscinosis type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TPP1 gene located on chromosomal region 11p15.4. Age of onset is infantile. This disease is characterized by epilepsy, followed by regression of developmental milestones, myoclonic ataxia, and pyramidal signs. Visual impairment typically appears at age four to six years and rapidly progresses to light/dark awareness only. Life expectancy ranges from age six years to early teenage. The prevalence is 1.5:1,000,000-9:1,000,000.	250,6
TPRN	Deafness type 79, autosomal recessive	NM_001128228.2	NM_001128228.2:c.1427delC, NM_001128228.2:c.1239G>A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB79 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TPRN gene located on chromosomal region 9q34.3. The age of onset is early. This disease is characterized by hearing loss and deafness.	600
TREX1	Aicardi-Goutieres syndrome type 1	NM_033629.4	NM_033629.4:c.341G>A, NM_033629.4:c.144_145insC, NM_033629.4:c.490C>T, NM_033629.4:c.506G>A	Aicardi-GoutiÃ¨res syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TREX1 gene located on chromosomal region 3p21.31. The age of onset is early. This disease is characterized by subacute encephalopathy (feeding problems, irritability and psychomotor regression or delay) associated with epilepsy (53% of cases), chilblain skin lesions on the extremities (43% of cases) and episodes of aseptic febrile illness (40% of cases). The prevalence is <1:1,000,000.	600
TRIM32	Limb-girdle muscular dystrophy type 2H	NM_012210.3	NM_012210.3:c.1459G>A, NM_012210.3:c.1560delC	Autosomal recessive limb-girdle muscular dystrophy type 2H follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TRIM32 gene located on chromosomal region 9q31-q33. The age of onset is variable. This disease is characterized by proximal muscle weakness and facial muscle wasting.	600
TRIM37	Mulibrey nanism	NM_015294.3	NM_015294.3:c.1346_1347insA, NM_015294.3:c.1411C>T, NM_015294.3:c.1037_1040dupAGAT, NM_015294.3:c.2056C>T, NM_015294.3:c.2212delG, NM_015294.3:c.227T>C, NM_015294.3:c.326G>C, NM_015294.3:c.496_500delAGGAA, NM_015294.3:c.745C>T, NM_015294.3:c.965G>T, NM_015294.3:c.1478_1479delAG, NM_015294.3:c.1668-1G>C	Mulibrey nanism follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TRIM37 gene located on chromosomal region 17q22. The age of onset is prenatal. This disease is characterized by pre- and postnatal growth restriction, characteristic craniofacial features with scaphocephaly, triangular face, high and broad forehead, low nasal bridge and yellowish dots in retinal mid peripheral region. The prevalence is <1:1,000,000.	600
TRIOBP	Deafness type 28, autosomal recessive	NM_001039141.2	NM_001039141.2:c.2362C>T, NM_001039141.2:c.3194delT, NM_001039141.2:c.1039C>T, NM_001039141.2:c.1741C>T, NM_001039141.2:c.4577C>G, NM_001039141.2:c.2639_2640insTCAC, NM_001039141.2:c.5316G>A, NM_001039141.2:c.3202C>T, NM_001039141.2:c.4429_4430insG	Deafness autosomal recessive type 28 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TRIOBP gene located on chromosomal region 22q13.1. The age of onset is early. This disease is characterized by hearing loss and deafness, no associated visible abnormalities of the external ear or any related medical problems.	250,6
TSEN54	Pontocerebellar hypoplasia	NM_207346.2	NM_207346.2:c.670_671delAA, NM_207346.2:c.736C>T, NM_207346.2:c.1027C>T, NM_207346.2:c.1039A>T, NM_207346.2:c.887G>A, NM_207346.2:c.919G>T	Pontocerebellar hypoplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TSEN54 gene located on chromosomal region 17q25.1. Pontocerebellar hypoplasia (PCH) refers to a group of severe neurodegenerative disorders affecting growth and function of the brainstem and cerebellum, resulting in little or no development. Different types were classified based on the clinical picture and the spectrum of pathologic changes.Â	250,6
TSFM	Combined oxidative phosphorylation deficiency type 3	NM_001172696.1	NM_001172696.1:c.1_2delAT, NM_001172696.1:c.580delC, NM_001172696.1:c.919C>T, NM_001172696.1:c.21_22delGC	Combined oxidative phosphorylation deficiency type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TSFM gene located on chromosomal region 12q14.1. The age of onset is early. This disease is characterized by hypotonia, lactic acidosis, and hepatic insufficiency, with progressive encephalomyopathy or hypertrophic cardiomyopathy.	250,6
TSHB	Isolated thyroid-stimulating hormone deficiency	NM_000549.4	NM_000549.4:c.94G>T, NM_000549.4:c.205C>T, NM_000549.4:c.145G>A	Isolated thyroid-stimulating hormone deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TSHB gene located on chromosomal region 1p13. The age of onset is early. This disease is characterized by decreased activity and increased sleep, feeding difficulty and constipation, prolonged jaundice, myxedematous facies, large fontanels (especially posterior), macroglossia, a distended abdomen with umbilical hernia and hypotonia. Slow linear growth and developmental delay are usually apparent by 4-6 months of age.	600

TSHR	Hypothyroidism	NM_000369.2	NM_000369.2:c.100G>A, NM_000369.2:c.1170T>G, NM_000369.2:c.484C>G, NM_000369.2:c.500T>A, NM_000369.2:c.122G>C, NM_000369.2:c.326G>A, NM_000369.2:c.1741_1742insC, NM_000369.2:c.202C>T	Hypothyroidism due to TSH receptor mutations follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TSHR gene located on chromosomal region 14q31. The age of onset is early. This disease is characterized by decreased activity and increased sleep, feeding difficulty and constipation, prolonged jaundice, myxedematous facies, large fontanels (especially posterior), macroglossia, a distended abdomen with umbilical hernia, and hypotonia. Slow linear growth and developmental delay are usually apparent by 4-6 months of age. Without treatment CH results in severe intellectual deficit and short stature. The prevalence is 1:3,000-1:4,000 newborn.	250,6
TTN	Cardiomyopathy, dilated/Tibial muscular dystrophy	NM_133378.4	NM_133378.4:c.13149C>A, NM_133378.4:c.22246G>A, NM_133378.4:c.31780G>A, NM_133378.4:c.40211dupT, NM_133378.4:c.44668delG, NM_133378.4:c.52977dupT, NM_133378.4:c.61640C>G, NM_133378.4:c.84669_84675delTGAATTC, NM_133378.4:c.94567C>T, NM_133378.4:c.96388C>T, NM_133378.4:c.96388delC, NM_133378.4:c.98366_98367delAT, NM_133378.4:c.12064C>T, NM_133378.4:c.28739-1G>A, NM_133378.4:c.3165-1G>T, NM_133378.4:c.4724_4728delTGAAA, NM_133378.4:c.48944-1G>A, NM_133378.4:c.91114_91117delTCCA, NM_133378.4:c.100185delA, NM_133378.4:c.40549delA, NM_133378.4:c.24568_24571delAGCA	Cardiomyopathy, dilated/Tibial muscular dystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TTN gene located on chromosomal region 2q31.2. The age of onset is variable. This disease is characterized by slowly progressive weakness and atrophy of the anterior tibial muscles with decreased dorsiflexion. Sometimes it could be companied by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. The prevalence is >9:100,000.	250,6
TTPA	Ataxia with vitamin E deficiency	NM_000370.3	NM_000370.3:c.661C>T, NM_000370.3:c.744delA, NM_000370.3:c.575G>A	Ataxia with vitamin E deficiency follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TTPA gene located on chromosomal region 8q13. The age of onset is variable. This disease is characterized by progressive spino-cerebellar ataxia, loss of proprioception, areflexia, and is associated with a marked deficiency in vitamin E. The prevalence is 0.56:1,000,000-3.5:1,000,000.	250,6
TULP1	Leber congenital amaurosis type 15	NM_003322.4	NM_003322.4:c.1198C>T, NM_003322.4:c.1204G>T	Leber congenital amaurosis 15 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TULP1 gene located on chromosomal region 6p21.31. The age of onset is early. This disease is characterized by blindness, nystagmus, roving eye movement and lack of detectable signals on an electroretinogram, leading to severe visual impairment within the first year of life.	600
TULP1	Retinitis pigmentosa type 14	NM_003322.4	NM_003322.4:c.1259G>C, NM_003322.4:c.1318C>T, NM_003322.4:c.1471T>C, NM_003322.4:c.1511_1521delTGCAGTTCGGC, NM_003322.4:c.1376T>A, NM_003322.4:c.1444C>T	Retinitis pigmentosa type 14 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TULP1 gene located on chromosomal region 6p21.31. The age of onset is early. This disease is characterized by night blindness often in adolescence or earlier. They then develop peripheral visual field impairment, and overtime loss of central vision, usually at late stages, often around midlife. The prevalence is 1/4,000.	600
TYR	Oculocutaneous albinism type 1	NM_000372.4	NM_000372.4:c.1012_1013insC, NM_000372.4:c.1146C>A, NM_000372.4:c.1164delT, NM_000372.4:c.1177delG, NM_000372.4:c.1147G>A, NM_000372.4:c.115T>G, NM_000372.4:c.1255G>A, NM_000372.4:c.1265G>A, NM_000372.4:c.1209G>T, NM_000372.4:c.1217C>T, NM_000372.4:c.140G>A, NM_000372.4:c.1467dupT, NM_000372.4:c.1501dupC, NM_000372.4:c.164G>A, NM_000372.4:c.1A>G, NM_000372.4:c.230G>A, NM_000372.4:c.242C>T, NM_000372.4:c.265T>C, NM_000372.4:c.272G>A, NM_000372.4:c.286dupA, NM_000372.4:c.533G>A, NM_000372.4:c.1336G>A, NM_000372.4:c.1342G>A, NM_000372.4:c.646T>A, NM_000372.4:c.650G>A, NM_000372.4:c.823G>T, NM_000372.4:c.896G>A, NM_000372.4:c.1111A>G, NM_000372.4:c.1118C>A, NM_000372.4:c.325G>A, NM_000372.4:c.572delG, NM_000372.4:c.616G>A	Oculocutaneous albinism type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TYR gene located on chromosomal region 11q14.2. The age of onset is early. This disease is characterized by white hair and skin, blue, fully translucent irises, nystagmus and misrouting of the optic nerves.	250,6
TYRP1	Oculocutaneous albinism type 3	NM_000550.2	NM_000550.2:c.107delT, NM_000550.2:c.1103delA, NM_000550.2:c.1057_1060delAACA, NM_000550.2:c.1067G>A, NM_000550.2:c.1557T>G, NM_000550.2:c.176C>G, NM_000550.2:c.497C>G, NM_000550.2:c.1120C>T, NM_000550.2:c.1369_1370insCAGA	Type 3 oculocutaneous albinism follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the TYRP1 gene located on chromosomal region 9p23. The age of onset is early. This disease is characterized by rufous or brown albinism and occurring mainly in the African population. The prevalence is of 1/8,500 individuals in Africa.	250,6
UBA1	Spinal muscular atrophy type 2, X-linked	NM_003334.3	NM_003334.3:c.1731C>T	X-linked spinal muscular atrophy type 2 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the UBA1 gene located on chromosomal region Xp11.23. The age of onset is infantile. This disease is characterized by congenital hypotonia and areflexia and evidence of degeneration and loss of anterior horn cells (i.e., lower motor neurons) in the spinal cord and brain stem. Often congenital contractures and/or fractures are present. Life span is shortened because of progressive ventilatory insufficiency resulting from chest muscle involvement.	600

UBR1	Johanson-Blizzard syndrome	NM_174916.2	NM_174916.2:c.869C>G, NM_174916.2:c.4254G>A, NM_174916.2:c.1281+1G>T, NM_174916.2:c.1537C>T	Johanson-Blizzard syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UBR1 gene located on chromosomal region 15q15.2. The age of onset is early. This disease is characterized by congenital exocrine pancreatic insufficiency and aplasia/hypoplasia of alae nasi, together with a variety of other abnormalities including aplasia cutis, anorectal anomalies and failure to thrive. The prevalence is <1:1,000,000.	600
UGT1A1	Crigler-Najjar syndrome type 1	NM_000463.2	NM_000463.2:c.1021C>T, NM_000463.2:c.1070A>G	Crigler-Najjar syndrome type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UGT1A1 gene located on chromosomal region 2q37. The age of onset is early. This disease is characterized by severe neonatal unconjugated hyperbilirubinemia, persistent jaundice and bilirubin encephalopathy manifesting as hypotonia, deafness, oculomotor palsy and lethargy. Neurologic defects can occur, generally associated with intellectual and motor impairment.	250,6
UGT1A1	Crigler-Najjar syndrome type 2	NM_000463.2	NM_000463.2:c.1207C>T, NM_000463.2:c.674T>G, NM_000463.2:c.1130G>T, NM_000463.2:c.524T>A, NM_000463.2:c.44T>G	Crigler-Najjar syndrome type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UGT1A1 gene located on chromosomal region 2q37. The age of onset is early. This disease is characterized by unconjugated hyperbilirubinemia due to reduced and inducible activity of hepatic bilirubin glucuronosyltransferase with pigmented bile that contains bilirubin glucuronides, and generally do not present neurologic or intellectual impairment. Bilirubin encephalopathy may develop in later life when patients experience a superimposed infection or stress.	250,6
UGT1A1	Gilbert syndrome	NM_000463.2	NM_000463.2:c.1211T>C, NM_000463.2:c.1456T>G	Gilbert syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UGT1A1 gene located on chromosomal region 2q37. The age of onset is early. This disease is characterized by jaundice due to unconjugated hyperbilirubinemia, resulting a partial deficiency in hepatic bilirubin glucuronosyltransferase activity.	250,6
UQCRB	Mitochondrial complex III deficiency, nuclear type 3	NM_006294.4	NM_006294.4:c.306_309delAAAA	Mitochondrial complex III deficiency, nuclear type 3 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UQCRB gene located on chromosomal region 8q22.1. The age of onset is infantile. This disease is characterized by a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance.	600
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	NM_014402.4	NM_014402.4:c.134C>T	Mitochondrial complex III deficiency, nuclear type 4 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the UQCRQ gene located on chromosomal region 5q31.1. The age of onset is infantile. This disease is characterized by severe psychomotor retardation and extrapyramidal signs. Neurologic features included dystonia, athetoid movements, ataxia, mild axial hypotonia, increased tone, hyperreflexia, and inability to walk unsupported.	600
USH1C	Usher syndrome type 1C	NM_153676.3	NM_153676.3:c.216G>A, NM_153676.3:c.2362G>A, NM_153676.3:c.2622_2623delCA, NM_153676.3:c.2688_2695dupAATTCACC, NM_153676.3:c.238_239insC, NM_153676.3:c.238delC, NM_153676.3:c.2547-1G>T, NM_153676.3:c.2695_2696insAATTCACC, NM_153676.3:c.388G>A	Usher syndrome type 1C follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the USH1C gene located on chromosomal region 11p15.1. The age of onset is infantile. This disease is characterized by the association of sensorineural deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. The prevalence is 3:100,000-4:100,000.	250,6
USH1G	Usher syndrome type 1G	NM_173477.4	NM_173477.4:c.186_187delCA, NM_173477.4:c.394_395insG, NM_173477.4:c.832_851delTCGGACGAGGACAGCGTCTC, NM_173477.4:c.649C>T, NM_173477.4:c.805C>T	Usher syndrome type 1G follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the USH1G gene located on chromosomal region 17q25.1. The age of onset is infantile. This disease is characterized by the association of sensorineural deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. The prevalence is 4.4:100,000.	600
USH2A	Retinitis pigmentosa type 39	NM_206933.2	NM_206933.2:c.10073G>A, NM_206933.2:c.2296T>C, NM_206933.2:c.14519T>C, NM_206933.2:c.7364G>A, NM_206933.2:c.12574C>T, NM_206933.2:c.2276G>T	Retinitis pigmentosa refers to a heterogeneous group of inherited ocular diseases that result in a progressive retinal degeneration. Type 39 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the USH2A gene located on chromosomal region 1q41. The age of onset is adult. This disease is characterized by night blindness, the development of tunnel vision, and slowly progressive decreased central vision. The global prevalence of all types of retinitis pigmentosa is 1/3,000 to 1/5,000.	250,6

USH2A	Usher syndrome type 2A	NM_206933.2	NM_206933.2:c.10636G>A, NM_206933.2:c.10561T>C, NM_206933.2:c.15371delT, NM_206933.2:c.2167+5G>A, NM_206933.2:c.11864G>A, NM_206933.2:c.14803C>T, NM_206933.2:c.2898delG, NM_206933.2:c.3491_3492delCT, NM_206933.2:c.11549-5_11549-4insT, NM_206933.2:c.2299delG, NM_206933.2:c.5975A>G, NM_206933.2:c.6670G>T, NM_206933.2:c.6862G>T, NM_206933.2:c.5743_5744delAG, NM_206933.2:c.779T>G, NM_206933.2:c.820C>T, NM_206933.2:c.8981G>A, NM_206933.2:c.956G>A, NM_206933.2:c.9799T>C, NM_206933.2:c.15089C>A, NM_206933.2:c.2135delC, NM_206933.2:c.4338_4339delCT, NM_206933.2:c.5573-2A>G, NM_206933.2:c.920_923dupGCCA, NM_206933.2:c.13709delG, NM_206933.2:c.14926G>A, NM_206933.2:c.15520-1G>A, NM_206933.2:c.8431C>A, NM_206933.2:c.12234_12235delGA, NM_206933.2:c.14442C>A	Usher syndrome type 2A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the USH2A gene located on chromosomal region 1q41. The age of onset is infantile. This disease is characterized by the association of sensorineural deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. The prevalence is 3:100,000-4:100,000.	250,6
VDR	Rickets, vitamin D-resistant, type 2A	NM_001017535.1	NM_001017535.1:c.137G>A, NM_001017535.1:c.149G>A, NM_001017535.1:c.885C>A, NM_001017535.1:c.88C>T, NM_001017535.1:c.239G>A, NM_001017535.1:c.821G>T, NM_001017535.1:c.88C>G, NM_001017535.1:c.915C>G, NM_001017535.1:c.985G>A	Vitamin D-dependent rickets type 2A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the VDR gene located on chromosomal region 12q13.11. The age of onset is early. This disease is characterized by hypocalcemia, severe rickets and in many cases alopecia. The prevalence is 1:10,000-5:10,000.	600
VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 1	NM_003383.3	NM_003383.3:c.2339delT, NM_003383.3:c.2302_2303delGA, NM_003383.3:c.844C>T, NM_003383.3:c.769C>T	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the VLDLR gene located on chromosomal region 9p24.2. The age of onset is early. This disease is characterized by non-progressive congenital ataxia that is predominantly truncal and results in delayed ambulation, moderate-to-profound intellectual disability, dysarthria, strabismus, and seizures. The prevalence is 1:100,000-9:100,000.	600
VPS13A	Choreoacanthocytosis	NM_033305.2	NM_033305.2:c.622C>T, NM_033305.2:c.9109C>T, NM_033305.2:c.2898T>G, NM_033305.2:c.3091delG, NM_033305.2:c.9275+1G>T	Chorea-acanthocytosis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the VPS13A gene located on chromosomal region 9q21. The age of onset is adult. This disease is characterized by progressive neurological symptoms including movement disorders, psychiatric manifestations and cognitive disturbances.	600
VPS33B	Arthrogryposis-renal dysfunction-cholestasis type 1	NM_018668.4	NM_018668.4:c.1246C>T, NM_018668.4:c.1312C>T, NM_018668.4:c.1498G>A, NM_018668.4:c.440_449delCTCTTGATGT, NM_018668.4:c.1594C>T, NM_018668.4:c.1480-1G>T, NM_018668.4:c.603+2T>A	Arthrogryposis - renal dysfunction â€“ cholestasis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the VPS33B gene located on chromosomal region 15q26.1. The age of onset is early. This disease is characterized by neurogenic arthrogryposis multiplex congenita, renal tubular dysfunction and neonatal cholestasis. The prevalence is <1:1,000,000.	600
WAS	Neutropenia, severe congenital, X-linked	NM_000377.2	NM_000377.2:c.881T>C, NM_000377.2:c.809T>C, NM_000377.2:c.814T>C	X-linked severe congenital neutropenia follows an X-linked pattern of inheritance and is caused by pathogenic variants in the WAS gene located on chromosomal region Xp11.23. The age of onset is early. This disease is characterized by recurrent major bacterial infections, severe congenital neutropenia, and monocytopenia. The prevalence is <1:1,000,000.	600
WAS	Thrombocytopaenia type 1	NM_000377.2	NM_000377.2:c.167C>T, NM_000377.2:c.173C>G, NM_000377.2:c.1442T>A	Thrombocytopaenia type 1 follows an X-linked pattern of inheritance and is caused by pathogenic variants in the WAS gene located on chromosomal region Xp11.23. The age of onset is early. Thrombocytopenia is defined by a decrease in the number of platelets in circulating blood, resulting in the potential for increased bleeding and decreased ability for clotting. The prevalence is <1:1,000,000.	600
WAS	Wiskott-Aldrich syndrome	NM_000377.2	NM_000377.2:c.134C>T	Wiskott-Aldrich sÃ-ndrome follows an X-linked pattern of inheritance and is caused by pathogenic variants in the WAS gene located on chromosomal region Xp11.23. The age of onset is infantile. This disease is characterized by microthrombocytopenia, eczema, infections and an increased risk for autoimmune manifestations and malignancies. The incidence is less than 1 in 100,000 live births and the prevalence is 1:1,000,000-10:1,000,000 men.	600
WDR62	Microcephaly primary, type 2, autosomal recessive	NM_001083961.1	NM_001083961.1:c.1313G>A, NM_001083961.1:c.3514+1delG, NM_001083961.1:c.3574delA, NM_001083961.1:c.1408C>T, NM_001083961.1:c.193G>A, NM_001083961.1:c.702dupG, NM_001083961.1:c.671G>C, NM_001083961.1:c.557G>A	Autosomal recessive primary microcephalyc type 2 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WDR62 gene located on chromosomal region 19q13.12. The age of onset is early. This disease is characterized by reduced head circumference at birth without gross anomalies of brain architecture and variable degrees of intellectual impairment. The incidence is 1:1,000,000.	600

WFS1	Wolfram syndrome	NM_006005.3	NM_006005.3:c.1234_1237delGTCT, NM_006005.3:c.1511C>T, NM_006005.3:c.2168T>C, NM_006005.3:c.2171C>T, NM_006005.3:c.1944G>A, NM_006005.3:c.2084G>T, NM_006005.3:c.577A>C, NM_006005.3:c.676C>T, NM_006005.3:c.2327A>T, NM_006005.3:c.407_408insGGGCCGTCGCGAGGCT, NM_006005.3:c.2576G>A, NM_006005.3:c.2643_2644delCT, NM_006005.3:c.616C>T, NM_006005.3:c.1060_1062delTTC, NM_006005.3:c.400G>A, NM_006005.3:c.1943G>A, NM_006005.3:c.1230_1233delCTCT	Wolfram syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WFS1 gene located on chromosomal region 4p16.1. The age of onset is infantile. This disease is characterized by diabetes mellitus type I, diabetes insipidus, optical atrophy and neurological signs. The prevalence is 1:1,000,000-9:1,000,000.	250,6
WNT10A	Hypohidrotic ectodermal dysplasia, autosomal recessive	NM_025216.2	NM_025216.2:c.347T>C, NM_025216.2:c.383G>A, NM_025216.2:c.321C>A	Hypohidrotic ectodermal displasia, autosomal recessive follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WNT10A gene located on chromosomal region 2q35. The age of onset is infantile. This disease is characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. The prevalence is <1:1,000,000.	250,6
WNT10A	Odontoonychodermal dysplasia	NM_025216.2	NM_025216.2:c.697G>T	Odonto-onycho-dermal dysplasia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WNT10A gene located on chromosomal region 2q35. The age of onset is infantile. This disease is characterized by hyperkeratosis and hyperhidrosis of the palms and soles, atrophic malar patches, hypodontia, conical teeth, onychodysplasia, and dry and sparse hair. The prevalence is <1:1,000,000.	250,6
WNT7A	Fuhrmann syndrome	NM_004625.3	NM_004625.3:c.874C>T	Fuhrmann syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WNT7A gene located on chromosomal region 3p25.1. The age of onset is neonatal/infancy. This disease is characterized by bowing of the femora, aplasia or hypoplasia of the fibulae and poly-, oligo-, and syndactyly. Most of the patients also have a hypoplastic pelvis and hypoplasia of the fingers and fingernails. Some had congenital dislocation of the hip, absence or fusion of tarsal bones, absence of various metatarsals, and hypoplasia and aplasia of the toes. The prevalence is <1:1,000,000.	600
WNT7A	Ulna and fibula, absence of, with sever limb deficiency	NM_004625.3	NM_004625.3:c.325G>A	Aplasia/hypoplasia of limbs and pelvis follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the WNT7A gene located on chromosomal region 3p25.1. The age of onset is early. This disease is characterized by intercalary limb deficiencies (phocomelia sometimes combined with polydactyly, oligodactyly or ectrodactyly), absent or hypoplastic pelvic bones (including sacral agenesis or hypoplasia), skull defects (frequently a defect of the occipital bone with or without meningocele)	600
XPA	Xeroderma pigmentosum Group A	NM_000380.3	NM_000380.3:c.323G>T, NM_000380.3:c.619C>T, NM_000380.3:c.727C>T, NM_000380.3:c.731A>G, NM_000380.3:c.348T>A, NM_000380.3:c.501delG	Xeroderma pigmentosum complementation group A follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the gene XPA located on chromosomal region 9q22.33. The age of onset is variable. This disease is characterized by photosensitivity of skin with burning, freckling, and skin cancers. It is associated with a spectrum of mild to severe neurological anomalies (e.g. cognitive deterioration, dysarthria, balance disturbance, areflexia) and sometimes delay of growth and sexual development. The prevalence is 1:1,000,000.	600
ZFYVE26	Spastic paraplegia type 15, autosomal recessive	NM_015346.3	NM_015346.3:c.3206G>A, NM_015346.3:c.3642_3643insCCACACTTAG, NM_015346.3:c.1477C>T, NM_015346.3:c.2887G>C, NM_015346.3:c.5422C>T, NM_015346.3:c.5485-1G>A, NM_015346.3:c.4312C>T, NM_015346.3:c.4936C>T, NM_015346.3:c.3182delT, NM_015346.3:c.2114_2115insC	Spastic paraplegia type 15 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ZFYVE26 gene located on chromosomal region 14q24.1. The age of onset is infancy. This disease is characterized by progressive spasticity primarily affecting the lower limbs. It is a complex form of spastic paraplegia, associated with other neurologic dysfunction, including variable mental retardation, hearing and visual defects, and thin corpus callosum. The prevalence is <1 / 1,000,000.	250,6
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	NM_005857.4	NM_005857.4:c.121C>T, NM_005857.4:c.1263dupT, NM_005857.4:c.1018T>C, NM_005857.4:c.955-1G>A, NM_005857.4:c.1349G>A	Mandibuloacral dysplasia with type B lipodystrophy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ZMPSTE24 gene located on chromosomal region 1p34.2. The age of onset is early. This disease is characterized by postnatal growth retardation, craniofacial anomalies and skeletal malformations, such as mandibular and clavicular hypoplasia; mottled cutaneous pigmentation and generalized lipoatrophy.	600
ZMPSTE24	Restrictive dermopathy, lethal	NM_005857.4	NM_005857.4:c.1076_1077insT, NM_005857.4:c.54dupT, NM_005857.4:c.1085_1086insT	Lethal restrictive dermopathy follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ZMPSTE24 gene located on chromosomal region 1p34.2. The age of onset is early. This disease is characterized by the skin being drawn tightly over the face causing a narrow, pinched nose, small mouth, limited jaw mobility, and entropion. Ears are malformed, with the auricle attached to the skin of scalp. Most infants die after a few hours from pulmonary distress. The prevalence is <1:1,000,000.	600

ZNF469	Brittle cornea syndrome	NM_001127464.1	NM_001127464.1:c.6673delC, NM_001127464.1:c.11452_11453insC, NM_001127464.1:c.4174G>T	Brittle cornea syndrome follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ZNF469 gene located on chromosomal region 16q24.2. The age of onset is infantile. This disease is characterized by severe ocular manifestations due to extreme corneal thinning and fragility with rupture in the absence of significant trauma. BCS generally progresses to blindness. The prevalence is <1:1,000,000.	600