CGT 250 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
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
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
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
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
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
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
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
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
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

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
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.6072delG, NM_018136.4:c.9677dupG, NM_018136.4:c.9685delA	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

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
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
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
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

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
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
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

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
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	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
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
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
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
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
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
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.Ã 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
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
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
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
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
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
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
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
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
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
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
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
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
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
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
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
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
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
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

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
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.505G>A, NM_000155.3:c.1006A>T, NM_000155.3:c.98ET>C	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
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	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

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
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
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
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
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
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
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
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
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
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
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
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
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

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
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
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
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
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
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
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
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
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
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
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
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
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

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
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
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
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
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

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
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.1117C>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.926G>A, NM_000277.1:c.509+1G>A	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
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
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
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
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
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

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
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
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
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
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
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
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

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
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
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

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

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
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
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
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

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
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
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

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
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
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
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
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
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
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

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
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
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

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
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
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
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

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
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
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
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
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
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