CGT Essential v1.1

chrom	OMIM (gene)	Gene	*Previous symbol*	OMIM (phen)	DISEASE	MOI
1	607008	ACADM		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
2	604285	AGXT		259900	Hyperoxaluria, primary, type 1	Autosomal recessive
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
3	609019	BTD		253260	Biotinidase deficiency	Autosomal recessive
21	613381	CBS		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
7	602421	CFTR		219700	Cystic fibrosis	Autosomal recessive
11	602858	DHCR7		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
X	300384	EMD		310300	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
X	309550	FMR1		300624	Fragile X syndrome	X-linked
X	305900	G6PD		300908	Hemolytic anemia, G6PD deficient (favism)	X-linked
17	606800	GAA		232300	Glycogen storage disease, type 2	Autosomal recessive
9	606999	GALT		230400	Galactosemia	Autosomal recessive
13	121011	GJB2		220290	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
X	300644	GLA		301500	Fabry disease	X-linked
2	600890	HADHA		609016; 609015	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
16	141800	HBA1		604131	Thalassemia, alpha-	Autosomal recessive
16	141850	HBA2		604131	Thalassemia, alpha-	Autosomal recessive
11	141900	HBB		603903	HBB-related hemoglobinopathy	Autosomal recessive
1	609831	MMACHC		277400	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
12	612349	PAH		261600	Phenylketonuria	Autosomal recessive
16	601785	PMM2		212065	Congenital disorder of glycosylation, type 1A	Autosomal recessive
5	606718	SLC26A2		600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
5	600354	SMN1		253300	Spinal muscular atrophy	Autosomal recessive