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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

Maternal inheritance of a mutation in the UBE3A gene causes Angelman syndrome*
If you need additional information, you can contact us at the following e-mail address: cgtspain@igenomix.com

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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

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