

chrom	OMIM (gene)	Gene	OMIM (phen)	DISEASE	MOI
X	300371	ABCD1	300100	Adrenoleukodystrophy	X-linked
X	313700	AR	300068	Androgen insensitivity syndrome, complete	X-linked
X	300180	ARSE	302950	Chondrodysplasia punctata, brachytelephalangi	X-linked
X	300011	ATP7A	309400; 304150	Menkes disease; Occipital horn syndrome	X-linked
X	300300	BTK	300755	Agammaglobulinemia X-linked, type 1	X-linked
X	300386	CD40LG	308230	Hyper-IgM syndrome, type 1 (immunodeficiency, with hyper-IgM, type 1)	X-linked
7	602421	CFTR	219700	Cystic fibrosis	Autosomal recessive
X	300390	CHM	303100	Choroideremia	X-linked
X	300481	CYBB	306400	Chronic granulomatous disease, X-linked	X-linked
X	300451	EDA	305100	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
X	300384	EMD	310300	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
X	300841	F8	306700	Hemophilia A	X-linked
X	300746	F9	306900	Hemophilia B	X-linked
X	309550	FMR1	300624	Fragile X syndrome	X-linked
X	305900	G6PD	300908	Hemolytic anemia, G6PD deficient (favism)	X-linked
13	121011	GJB2	220290	Deafness, autosomal recessive, type 1A	Autosomal recessive
X	300644	GLA	301500	Fabry disease	X-linked
X	300808	GPR143	300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
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
X	300823	IDS	309900	Mucopolysaccharidosis, type 2	X-linked
X	308380	IL2RG	300400	Severe combined immunodeficiency, X-linked	X-linked
X	308840	L1CAM	307000; 303350	L1 Syndrome	X-linked
X	300005	MECP2	300673	Encephalopathy, neonatal severe	X-linked
X	300415	MTM1	310400	Myotubular myopathy, X-linked	X-linked
X	300473	NR0B1	300200	Adrenal hypoplasia, congenital	X-linked
X	300535	OCRL	309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked

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X	300461	OTC	311250	Ornithine transcarbamylase deficiency	X-linked
X	300502	PDHA1	312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
X	300490	SH2D1A	308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
X	300036	SLC6A8	300352	Cerebral creatine deficiency syndrome, type 1	X-linked
5	600354	SMN1	253300	Spinal muscular atrophy	Autosomal recessive
X	300392	WAS	301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked