

chrom	OMIM (gene)	gene	previous	OMIM (phen)	disease name (phenotype)	inheritance
X	300371	ABCD1		300100	Adrenoleukodystrophy	X-linked
1	607008	ACADM		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
2	604285	AGXT		259900	Hyperoxaluria, primary, type 1	Autosomal recessive
X	300629	AP1S2		304340	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
X	313700	AR		300068	Androgen insensitivity syndrome, complete	X-linked
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
X	300180	ARSL	ARSE	302950	Chondrodysplasia punctata, brachytelephalanic	X-linked
X	300382	ARX		308350; 300215; 309510	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
X	300011	ATP7A		309400; 304150	Menkes disease; Occipital horn syndrome	X-linked
X	300504	ATRX		309580; 301040	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
X	300553	BRWD3		300659	Mental retardation, X-linked, type 93	X-linked
3	609019	BTD		253260	Biotinidase deficiency	Autosomal recessive
X	300300	BTK		300755	Agammaglobulinemia X-linked, type 1	X-linked
21	613381	CBS		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
X	300386	CD40LG		308230	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
7	602421	CFTR		219700	Cystic fibrosis	Autosomal recessive
X	300390	CHM		303100	Choroideremia	X-linked
X	303630	COL4A5		301050	Alport syndrome, X-linked	X-linked
X	300304	CUL4B		300354	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
X	300481	CYBB		306400	Chronic granulomatous disease, X-linked	X-linked
X	300121	DCX		300067	Lissencephaly, X-linked, type 1	X-linked
11	602858	DHCR7		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
X	300126	DKC1		305000	Dyskeratosis congenita, X-linked	X-linked
X	300189	DLG3		300850	Mental retardation, X-linked, type 90	X-linked
X	300377	DMD		310200; 300376	Duchenne/Becker muscular dystrophy	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	300546	FGD1		305400	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
X	309550	FMR1		300624	Fragile X syndrome	X-linked
X	300499	FTSJ1		309549	Mental retardation, X-linked 44	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
X	304040	GJB1		302800	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
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
X	300808	GPR143		300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
2	600890	HADHA		609016; 609015	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive

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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	300019	HCFC1		309541	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cbIX type)	X-linked
X	308000	HPRT1		300322	Lesch-Nyhan syndrome	X-linked
X	300256	HSD17B10		300438	HSD10 mitochondrial disease	X-linked
X	300823	IDS		309900	Mucopolysaccharidosis, type 2	X-linked
X	300206	IL1RAPL1		300143	Mental retardation, X-linked, type 21/34	X-linked
X	308380	IL2RG		300400	Severe combined immunodeficiency, X-linked	X-linked
X	314690	KDM5C		300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
X	308840	L1CAM		307000; 303350; 304100	L1 Syndrome	X-linked
X	300005	MECP2		300673; 312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
1	609831	MMACHC		277400	Methylmalonic aciduria and homocystinuria, cbIC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
X	300415	MTM1		310400	Myotubular myopathy, X-linked	X-linked
X	300658	NDP		310600	Norrie disease	X-linked
X	300473	NR0B1		300200	Adrenal hypoplasia, congenital	X-linked
X	300535	OCRL		309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked
X	300127	OPHN1		300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
X	300461	OTC		311250	Ornithine transcarbamylase deficiency	X-linked
12	612349	PAH		261600	Phenylketonuria	Autosomal recessive
X	300142	PAK3		300558	Mental retardation, X-linked, type 30	X-linked
X	300502	PDHA1		312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
X	311800	PGK1		300653	Phosphoglycerate kinase 1 deficiency	X-linked
X	300560	PHF8		300263	Mental retardation syndrome, X-linked, Siderius type	X-linked
X	300401	PLP1		312080	Pelizaeus-Merzbacher disease	X-linked
16	601785	PMM2		212065	Congenital disorder of glycosylation, type 1A	Autosomal recessive
X	300039	POU3F4		304400	Deafness, X-linked, type 2	X-linked
X	300463	PQBP1		309500	Renpenning syndrome	X-linked
X	311850	PRPS1		300661; 304500; 311070; 301835	PRPS1-related disorders	X-linked
X	300757	RP2		312600	Retinitis pigmentosa, type 2, X-linked	X-linked
X	312610	RPGR		300029; 304020	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
X	300839	RS1		312700	Retinoschisis	X-linked
X	300490	SH2D1A		308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
X	300095	SLC16A2		300523	Allan-Herndon-Dudley syndrome	X-linked
5	606718	SLC26A2		600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
X	300036	SLC6A8		300352	Cerebral creatine deficiency syndrome, type 1	X-linked
5	600354	SMN1		253300	Spinal muscular atrophy	Autosomal recessive
X	313440	SYN1		300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
X	300395	THOC2		300957	Mental retardation, X-linked 12	X-linked

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X	300298	UPF3B		300676	Mental retardation, X-linked, syndromic, type 14	X-linked
X	300392	WAS		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
X	300646	ZDHHC9		300799	Mental retardation, X-linked syndromic, Raymond type	X-linked
X	314990	ZNF711		300803	Mental retardation, X-linked, type 97	X-linked