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chrom	OMIM (gene)	Gene	Approved symbol	OMIM (phen)	DISEASE	MOI
X	300135	ABCB7		301310	X-linked sideroblastic anemia and ataxia (XLSA/A)	X-linked
1	607008	ACADM		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
X	301300	ALAS2		300751	X-linked sideroblastic anemia, type 1 (XLSA or SIDBA1)	X-linked
X	313700	AR		300068	Androgen insensitivity syndrome, complete	X-linked
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
X	300180	ARSE	ARSL	302950	Chondrodysplasia punctata, brachytelephalangic	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
13	606882	ATP7B		277900	Wilson disease	Autosomal recessive
X	300300	BTK		300755	Agammaglobulinemia X-linked, type 1	X-linked
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
11	602858	DHCR7		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
X	300126	DKC1		305000	Dyskeratosis congenita, X-linked	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

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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	300163	FHL1		300696	Emery-Dreifuss muscular dystrophy, type 6, X-linked	X-linked
X	300017	FLNA		305620	FLNA-related disorders	X-linked
X	309550	FMR1		300624	Fragile X syndrome	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
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	308000	HPRT1		300322	Lesch-Nyhan syndrome	X-linked
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; 304100	L1 Syndrome	X-linked
X	300294	MBTPS2		308205; 301014	IFAP/BRESHECK syndrome; Osteogenesis imperfecta, type 19	X-linked
X	300005	MECP2		300673; 312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
X	300188	MED12		309520	Lujan-Fryns syndrome	X-linked
X	300415	MTM1		310400	Myotubular myopathy, X-linked	X-linked

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X	300473	NR0B1		300200	Adrenal hypoplasia, congenital	X-linked
X	300278	NYX		310500	Night blindness, congenital stationary (complete), type 1A, X-linked	X-linked
X	300535	OCRL		309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked
X	300170	OFD1		311200; 300209; 300804	Orofaciodigital syndrome, type 1; Simpson-Golabi-Behmel syndrome, type 2; Joubert syndrome, type 10	X-linked
X	300461	OTC		311250	Ornithine transcarbamylase deficiency	X-linked
X	300502	PDHA1		312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
6	606702	PKHD1		263200	Polycystic kidney disease type 4	Autosomal recessive
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	311850	PRPS1		300661; 304500; 311070; 301835	PRPS1-related disoders	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	300490	SH2D1A		308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
X	300036	SLC6A8		300352	Cerebral creatine deficiency syndrome, type 1	X-linked

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5	600354	SMN1		253300	Spinal muscular atrophy	Autosomal recessive
X	300356	TIMM8A		304700	Mohr-Tranebjaerg syndrome	X-linked
X	300392	WAS		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked

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