

gene	OMIM (gene)	Disease name (phenotype)	Inheritance
ABCD1	300371	Adrenoleukodystrophy	X-linked
AFF2	300806	Mental retardation, X-linked, FRAXE type	X-linked
AP1S2	300629	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
AR	313700	Androgen insensitivity syndrome, complete	X-linked
ARSL	300180	Chondrodysplasia punctata, brachytelephalangi	X-linked
ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
ATP7A	300011	Menkes disease; Occipital horn syndrome	X-linked
ATRX	300504	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
BRWD3	300553	Mental retardation, X-linked, type 93	X-linked
BTK	300300	Agammaglobulinemia X-linked, type 1	X-linked
CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
CFTR	602421	Cystic fibrosis	Autosomal recessive
CHM	300390	Choroideremia	X-linked
COL4A5	303630	Alport syndrome, X-linked	X-linked
CUL4B	300304	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
CYBB	300481	Chronic granulomatous disease, X-linked	X-linked
DCX	300121	Lissencephaly, X-linked, type 1	X-linked
DKC1	300126	Dyskeratosis congenita, X-linked	X-linked
DLG3	300189	Mental retardation, X-linked, type 90	X-linked
DMD	300377	Duchenne/Becker muscular dystrophy	X-linked
EDA	300451	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
EMD	300384	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
F8	300841	Hemophilia A	X-linked
F9	300746	Hemophilia B	X-linked
FGD1	300546	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
FMR1	309550	Fragile X syndrome	X-linked
FTSJ1	300499	Mental retardation, X-linked 44	X-linked
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)	X-linked
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
GJB2	121011	Deafness, autosomal recessive, type 1A	Autosomal recessive
GLA	300644	Fabry disease	X-linked
GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
HBA1	141800	Thalassemia, alpha-	Autosomal recessive
HBA2	141850	Thalassemia, alpha-	Autosomal recessive
HBB	141900	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies	Autosomal recessive
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocystinemia, cbIX type )	X-linked
HPRT1	308000	Lesch-Nyhan syndrome	X-linked
HSD17B10	300256	HSD10 mitochondrial disease	X-linked
IDS	300823	Mucopolysaccharidosis, type 2	X-linked
IL1RAPL1	300206	Mental retardation, X-linked, type 21/34	X-linked
IL2RG	308380	Severe combined immunodeficiency, X-linked	X-linked
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
L1CAM	308840	L1 Syndrome	X-linked
MECP2	300005	Encephalopathy, neonatal severe; Rett syndrome	X-linked
MID1	300552	Opitz GBBB syndrome, type 1	X-linked
MTM1	300415	Myotubular myopathy, X-linked	X-linked
NDP	300658	Norrie disease	X-linked

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NR0B1	300473	Adrenal hypoplasia, congenital	X-linked
OCRL	300535	Lowe Syndrome; Dent disease type 2	X-linked
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
OTC	300461	Ornithine transcarbamylase deficiency	X-linked
PAK3	300142	Mental retardation, X-linked, type 30	X-linked
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
PGK1	311800	Phosphoglycerate kinase 1 deficiency	X-linked
PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	X-linked
PLP1	300401	Pelizaeus-Merzbacher disease	X-linked
POU3F4	300039	Deafness, X-linked, type 2	X-linked
PQBP1	300463	Renpenning syndrome	X-linked
PRPS1	311850	PRPS1-related disorders	X-linked
RP2	300757	Retinitis pigmentosa, type 2, X-linked	X-linked
RPGR	312610	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
RS1	300839	Retinoschisis	X-linked
SH2D1A	300490	Lymphoproliferative syndrome, X-linked, type 1	X-linked
SLC16A2	300095	Allan-Herndon-Dudley syndrome	X-linked
SLC6A8	300036	Cerebral creatine deficiency syndrome, type 1	X-linked
SMN1	600354	Spinal muscular atrophy	Autosomal recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
THOC2	300395	Mental retardation, X-linked 12	X-linked
UPF3B	300298	Mental retardation, X-linked, syndromic, type 14	X-linked
WAS	300392	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
ZDHC9	300646	Mental retardation, X-linked syndromic, Raymond type	X-linked
ZNF711	314990	Mental retardation, X-linked, type 97	X-linked