

CGT Bank v2.3

Gene	Disease
ABCD1	Adrenoleukodystrophy
AR	Androgen insensitivity syndrome, complete
ARSE	Chondrodysplasia punctata, brachytelephalangi
ATP7A	Menkes disease; Occipital horn syndrome
BTK	Agammaglobulinemia X-linked, type 1 Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CD40LG	
CFTR	Cystic fibrosis
CHM	Choroideremia
CYBB	Chronic granulomatous disease, X-linked
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
F8	Hemophilia A
F9	Hemophilia B
FMR1	Fragile X syndrome
G6PD	Hemolytic anemia, G6PD deficient (favism)
GJB2	Deafness, autosomal recessive, type 1A
GLA	Fabry disease
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)
HBA1	Thalassemia, alpha-
HBA2	Thalassemia, alpha-
HBB	HBB-related hemoglobinopathy
IDS	Mucopolysaccharidosis, type 2
IL2RG	Severe combined immunodeficiency, X-linked
L1CAM	L1 Syndrome
MECP2	Encephalopathy, neonatal severe
MTM1	Myotubular myopathy, X-linked
NR0B1	Adrenal hypoplasia, congenital
OCRL	Lowe Syndrome; Dent disease type 2
OTC	Ornithine transcarbamylase deficiency
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
SH2D1A	Lymphoproliferative syndrome, X-linked, type 1
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SMN1	Spinal muscular atrophy
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked