GUARDIAN Gene List Version 1:

Active from September 6th, 2022 until February 29th, 2024

Group 1:

Disease association	Gene name
ABCD1-related adrenoleukodystrophy	ABCD1
Methylmalonic aciduria and homocystinuria, cblJ type	ABCD4
Isobutyryl-CoA dehydrogenase deficiency	ACAD8
ACADM-related medium-chain acyl-CoA dehydrogenase deficiency	ACADM
ACADS-related short-chain acyl-CoA dehydrogenase deficiency	ACADS
2-methylbutyrylglycinuria	ACADSB
ACADVL-related very long-chain acyl-CoA dehydrogenase deficiency	ACADVL
ACAT1-related alpha-methylacetoacetic aciduria	ACAT1
ACTG1-related nonsyndromic hearing loss	ACTG1
ADA-related adenosine deaminase deficiency	ADA
AGXT-related primary hyperoxaluria	AGXT
AHCY-related S-adenosylhomocysteine hydrolase deficiency	AHCY
AK2-related reticular dysgenesis	AK2
ALDH4A1-related hyperprolinemia	ALDH4A1
ALDH7A1-related pyridoxine-dependent epilepsy	ALDH7A1
ALDOB-related hereditary fructose intolerance	ALDOB
ALPL-related hypophosphatasia	ALPL
ALPL-related hypophosphatasia	ALPL
APOB-related hypercholesterolemia	APOB
APOB-related hypobetalipoproteinemia	APOB
APOB-related hypercholesterolemia	APOB
APOB-related hypobetalipoproteinemia	APOB
Adenine phosphoribosyltransferase deficiency	APRT
AQP2-related nephrogenic diabetes insipidus	AQP2
AQP2-related nephrogenic diabetes insipidus	AQP2
ARG1-related arginase deficiency	ARG1
ARSB-related mucopolysaccharidosis type 6	ARSB
ASL-related argininosuccinic aciduria	ASL
ASS1-related argininosuccinate synthetase deficiency	ASS1
ATP6V1B1-related renal tubular acidosis with hearing loss	ATP6V1B1
ATP7A-related Menkes spectrum disorder	ATP7A
ATP7A-related distal motor neuropathy	ATP7A
ATP7B-related Wilson disease	ATP7B
AUH-related 3-methylglutaconic aciduria	AUH

AVPR2-related nephrogenic diabetes insipidus	AVPR2
AVPR2-related nephrogenic syndrome of inappropriate antidiuresis	AVPR2
BCKDHA-related maple syrup urine disease	BCKDHA
BCKDHB-related maple syrup urine disease	BCKDHB
BTD-related biotinidase deficiency 2	BTD
Homocystinuria, B6-responsive and nonresponsive types	CBS
Immunodeficiency 19	CD3D
Immunodeficiency 18	CD3E
CFTR-related cystic fibrosis and congenital absence of the vas deferens	CFTR
CPS1-related carbamoylphosphate synthetase I deficiency	CPS1
CPT1A-related carnitine palmitoyltransferase 1A deficiency	CPT1A
CPT2-related carnitine palmitoyltransferase deficiency	CPT2
CTNS-related cystinosis	CTNS
DBT-related maple syrup urine disease	DBT
DCLRE1C-related immunodeficiency	DCLRE1C
DUOX2-related hypothyroidism	DUOX2
DUOXA2-related hypothyroidism	DUOXA2
ETFA-related multiple acyl-CoA dehydrogenase deficiency	ETFA
ETFB-related multiple acyl-CoA dehydrogenase deficiency	ETFB
ETFDH-related multiple acyl-CoA dehydrogenase deficiency	ETFDH
FAH-related tyrosinemia	FAH
FGFR3-related skeletal dysplasia	FGFR3
Bamforth-Lazarus syndrome	FOXE1
G6PC1-related glycogen storage disease	G6PC
G6PD-related glucose-6-phosphate dehydrogenase deficiency	G6PD
GAA-related glycogen storage disease	GAA
GALC-related Krabbe disease	GALC
Galactose epimerase deficiency	GALE
GALNS-related mucopolysaccharidosis type 4	GALNS
GALT-related galactosemia	GALT
GAMT-related guanidinoacetate methyltransferase (GAMT) deficiency	GAMT
GATM-related arginine:glycine amidinotransferase (AGAT) deficiency	GATM
GCDH-related glutaryl-CoA dehydrogenase deficiency	GCDH
GCH1-related dopa-responsive dystonia	GCH1
GJB2-related sensorineural hearing loss	GJB2
GLA-related Fabry disease	GLA
Diabetes mellitus, neonatal, with congenital hypothyroidism	GLIS3
Glycine N-methyltransferase deficiency	GNMT
GUSB-related mucopolysaccharidosis type 7	GUSB
HADH-related 3-hydroxyacyl-CoA dehydrogenase deficiency	HADH
HADHA-related trifunctional protein deficiency	HADHA

HADHA-related long chain hydroxyacyl-CoA dehydrogenase deficiency	HADHA
HADHB-related mitochondrial trifunctional protein deficiency	HADHB
HBB-related sickle cell disease	HBB
HCFC1-methylmalonic acidemia and homocysteinemia, cblX type	HCFC1
HIBCH-related 3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH
HLCS-related holocarboxylase synthetase deficiency	HLCS
HMGCL-related HMG-CoA lyase deficiency	HMGCL
HMGCS2-related HMG-CoA synthase-2 deficiency	HMGCS2
HPD-related tyrosinemia	HPD
HSD17B4-related D-bifunctional protein deficiency	HSD17B4
IDS-related mucopolysaccharidosis type 2	IDS
IDUA-related mucopolysaccharidosis type 1	IDUA
IL2RG-related immune dysregulation disorder	IL2RG
IL7R-related immunodeficiency	IL7R
IVD-related isovaleric acidemia	IVD
SCID, autosomal recessive, T-negative/B-positive type	JAK3
KCNH2-related long QT syndrome	KCNH2
KCNH2-related short QT syndrome	KCNH2
KCNQ1-related Jervell and Lange-Nielsen syndrome	KCNQ1
KCNQ1-related arrhythmia	KCNQ1
LDLR-related familial hypercholesterolemia	LDLR
LIAS-related pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS
LIG4 syndrome	LIG4
LIPA-related cholesteryl ester storage disease	LIPA
LMBRD1-related methylmalonic aciduria and homocystinuria, cblF type 2	LMBRD1
MAT1A-related methionine adenosyltransferase I/III deficiency	MAT1A
MCCC1-related 3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
MCCC2-related 3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
MCEE-related methylmalonyl-CoA epimerase deficiency	MCEE
MLYCD-related malonyl-CoA decarboxylase deficiency	MLYCD
MMAA-related methylmalonic aciduria	MMAA
MMAB-related methylmalonic aciduria, cblB type	MMAB
MMACHC-related methylmalonic aciduria and homocystinuria, cblC type	MMACHC
MMADHC-related homocystinuria, cblD type	MMADHC
MMUT-related methylmalonic aciduria	MMUT
MOCS1-related molybdenum cofactor deficiency	MOCS1
MTHFR-related MTHFR deficiency	MTHFR
MTR-related methylcobalamin deficiency, cblG type	MTR
MTRR-related methionine synthase reductase deficiency, cbl E type	MTRR

NAGS-related N-acetylglutamate synthase deficiency	NAGS
Severe combined immunodeficiency with microcephaly, growth retardation and sensitivity to ionizing radiation	NHEJ1
NKX2-1-related movement disorder	NKX2-1
NKX2-5-related heart defect	NKX2-5
NKX2-5-related congenital hypothyroidism	NKX2-5
OAT-related gyrate atrophy of the choroid and retina	OAT
OTC-related ornithine transcarbamylase deficiency	OTC
Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1
PAH-related phenylketonuria	PAH
PAX8-related hypothyroidism	PAX8
PC-related pyruvate carboxylase deficiency	PC
PCBD1-related BH4-deficient hyperphenylalaninemia	PCBD1
PCCA-related propionic acidemia	PCCA
PCCB-related propionic acidemia	PCCB
PCSK9-related familial hypercholesterolemia	PCSK9
PHEX-related hypophosphatemic rickets	PHEX
Immunodeficiency due to purine nucleoside phosphorylase deficiency	PNP
PNPO-related pyridoxamine 5'-phosphate oxidase deficiency	PNPO
Methylmalonic aciduria and homocystinuria, cblC type, epigenic	PRDX1
PRKDC-related immune dysregulation disorder	PRKDC
PTS-related BH4-deficient hyperphenylalaninemia	PTS
QDPR-related BH4-deficient hyperphenylalaninemia	QDPR
RAG1-related immune dysregulation	RAG1
RAG1-related immune dysregulation	RAG2
RPE65-related retinal dystrophy	RPE65
SLC22A5-related primary/systemic carnitine deficiency	SLC22A5
SLC25A13-related citrullinemia	SLC25A13
SLC25A15-related hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	SLC25A15
SLC25A20-related carnitine-acylcarnitine translocase deficiency	SLC25A20
SLC26A4-related hearing loss	SLC26A4
SLC2A1-related GLUT1 deficiency	SLC2A1
SLC37A4-related glycogen storage disease 1	SLC37A4
Acrodermatitis enteropathica zinc-deficiency type 1	SLC39A4
SLC52A2-related riboflavin transporter deficiency	SLC52A2
SLC52A3-related riboflavin transporter deficiency	SLC52A3
SLC5A5-related hypothyroidism	SLC5A5
SMN1-related spinal muscular atrophy	SMN1
SPR-related sepiapterin reductase deficiency	SPR
TAT-related tyrosinemia	TAT
Transcobalamin II deficiency	TCN2

TG-related hypothyroidism	TG
TPO-related hypothyroidism	TPO
TPP1-related ceroid lipofuscinosis	TPP1
TSHR-related congenital hypothyroidism	TSHR
WAS-related Wiskott-Aldrich syndrome	WAS
WAS-related neutropenia	WAS
Immunodeficiency 48	ZAP70

Group 2:

Disease association	**Gene name**
ACBD5-related retinal dystrophy and leukodystrophy spectrum disorder	ACBD5
ACOX1-related peroxisomal acyl-CoA oxidase deficiency	ACOX1
ACOX1-related progressive myeloneuropathy with sensorineural hearing loss (AD)	ACOX1
ACTB-Related Disorder	ACTB
ACTB-related Baraitser-Winter syndrome	ACTB
ACTG1-related Baraitser-Winter syndrome	ACTG1
ACTL6B-related developmental and epileptic encephalopathy	ACTL6B
ACTL6B-related neurodevelopmental disorder	ACTL6B
ADNP-related neurodevelopmental disorder with multiple anomalies	ADNP
ADSL-related adenylosuccinate lyase deficiency and epileptic encephalopathy	ADSL
AHDC1-related neurodevelopmental disorder	AHDC1
ALDH5A1-related succinic semialdehyde dehydrogenase deficiency	ALDH5A1
AMT-related glycine encephalopathy	AMT
ANKRD11-related KBG syndrome	ANKRD11
ARID1B-related neurodevelopmental disorder	ARID1B
ASH1L-related neurodevelopmental disorder with multiple anomalies	ASH1L
ASXL3-related neurodevelopmental disorder with multiple anomalies	ASXL3
AUTS2-related neurodevelopmental disorder	AUTS2
BCL11A-related neurodevelopmental disorder with persistence of fetal hemoglobin	BCL11A
CASK-related neurodevelopmental disorder with multiple anomalies 160	CASK
CDKL5-related developmental and epileptic encephalopathy	CDKL5
CHAMP1-related neurodevelopmental disorder	CHAMP1
CHD2-related developmental and epileptic encephalopathy	CHD2
CHD3-related neurodevelopmental disorder	CHD3
CHD7-related CHARGE spectrum disorder	CHD7
CHD8-related neurodevelopmental disorder	CHD8

CLPB-related caseinolytic peptidase B deficiency	CLPB
CNOT3-related neurodevelopmental disorder	CNOT3
CREBBP-related Rubinstein-Taybi syndrome	CREBBP
CREBBP-related neurodevelopmental disorder with multiple anomalies	CREBBP
CSDE1-related neurodevelopmental disorder with ocular anomalies	CSDE1
CSNK2A1-related neurodevelopmental disorder	CSNK2A1
CTBP1-related neurodevelopmental disorder	CTBP1
CTCF-related neurodevelopmental disorder with multiple anomalies	CTCF
DDX3X-related neurodevelopmental disorder	DDX3X
DEAF1-related neurodevelopmental and movement disorder	DEAF1
DEAF1-related neurodevelopmental disorder	DEAF1
DHCR7-related Smith-Lemli-Opitz syndrome	DHCR7
DHPS-related neurodevelopmental disorder	DHPS
DLD-related dihydrolipoamide dehydrogenase deficiency	DLD
3-methylglutaconic aciduria, type V	DNAJC19
DYRK1A-related neurodevelopmental disorder	DYRK1A
EBF3-related neurodevelopmental disorder with multiple anomalies	EBF3
EHMT1-related Kleefstra syndrome	EHMT1
ETHE1-related ethylmalonic encephalopathy	ETHE1
FOXG1-related Rett spectrum disorder	FOXG1
FOXP1-related neurodevelopmental disorder	FOXP1
FOXP3-related immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3
GLDC-related glycine encephalopathy	GLDC
GRIN2B-related neurodevelopmental disorder	GRIN2B
GRIN2B-related developmental and epileptic encephalopathy	GRIN2B
HECW2-related neurodevelopmental disorder	HECW2
HIVEP2-related neurodevelopmental disorder	HIVEP2
HNRNPH2-related neurodevelopmental disorder with multiple anomalies	HNRNPH2
HNRNPU-related developmental and epileptic encephalopathy	HNRNPU
HSD17B10-related 17-beta-hydroxysteroid dehydrogenase deficiency	HSD17B10
KAT6A-related neurodevelopmental disorder with multiple anomalies	KAT6A
KIF1A-related neurodegenerative spectrum disorder	KIF1A
KMT5B-related neurodevelopmental disorder	KMT5B
MBD5-related neurodevelopmental disorder	MBD5
MECP2-related Rett spectrum disorder	MECP2
MED13-related neurodevelopmental disorder with multiple anomalies	MED13
MED13L-related neurodevelopmental disorder with multiple anomalies	MED13L
NFU1-related multiple mitochondrial dysfunctions syndrome	NFU1
PACS1-related neurodevelopmental disorder with multiple anomalies	PACS1
PEX1-related Zellweger spectrum disorder	PEX1
PEX10-related Zellweger spectrum disorder	PEX10

PEX12-related Zellweger spectrum disorder	PEX12
PEX13-related Zellweger spectrum disorder	PEX13
PEX14-related Zellweger spectrum disorder	PEX14
PEX16-related Zellweger spectrum disorder	PEX16
PEX19-related Zellweger spectrum disorder	PEX19
PEX2-related Zellweger spectrum disorder	PEX2
PEX26-related Zellweger spectrum disorder	PEX26
PEX3-related Zellweger spectrum disorder	PEX3
PEX5-related Zellweger spectrum disorder	PEX5
PEX6-related Zellweger spectrum disorder	PEX6
PHIP-related neurodevelopmental disorder with multiple anomalies	PHIP
PPP2R5D-related neurodevelopmental disorder with multiple anomalies	PPP2R5D
Hyperprolinemia, type I	PRODH
SCN1A-related epilepsy	SCN1A
SCN1A-related familial hemiplegic migraine	SCN1A
SCN2A-related developmental and epileptic encephalopathy	SCN2A
SCN2A-related epilepsy	SCN2A
SCN8A-related neurodevelopmental and movement disorder	SCN8A
SCN8A-related epilepsy	SCN8A
SERAC1-related 3-methylglutaconic aciduria	SERAC1
SETBP1-related neurodevelopmental disorder	SETBP1
SETBP1-related Schinzel-Giedion syndrome	SETBP1
SLC6A1-related neurodevelopmental disorder	SLC6A1
Glycine encephalopathy with normal serum glycine	SLC6A9
SLC9A6-related Christianson spectrum disorder	SLC9A6
STXBP1-related neurodevelopmental disorder	STXBP1
SUCLA2-related mitochondrial DNA depletion syndrome	SUCLA2
SUCLG1-related mitochondrial DNA depletion syndrome	SUCLG1
SYNGAP1-related neurodevelopmental disorder	SYNGAP1
Barth syndrome	TAZ
TRIO-related neurodevelopmental disorder	TRIO
USP9X-related neurodevelopmental disorder with multiple anomalies	USP9X