Group 1

Gene List

Gene	Condition	Type of Condition	More Information	Notes
45,X	Turner syndrome	Multisystem disorder (reproductive, endocrine, skeletal, cardiac, renal)	Turner syndrome: MedlinePlus Genetics	Added 6/24/2024
ABCC8	Hyperinsulinemic hypoglycemia, familial 1	Metabolic Disorder	Congenital hyperinsulinism: MedlinePlus Genetics	
ABCC8	Diabetes mellitus, transient and permanent neonatal	Neonatal Diabetes	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
ABCD1	Adrenoleukodystrophy	Metabolic Disorder	X-linked adrenoleukodystrophy: MedlinePlus Genetics	Males only
ACADM	Medium chain acyl-CoA dehydrogenase deficiency	Metabolic Disorder	Medium-chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics	
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	Metabolic Disorder	Very long-chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics	
ACAT1	Beta-ketothiolase deficiency	Metabolic Disorder	Beta-ketothiolase deficiency: MedlinePlus Genetics	
ADA	Severe Combined Immunodeficiency due to ADA Deficiency	Immunodeficiency Disorder	Adenosine deaminase deficiency: MedlinePlus Genetics	
ADAMTS13	Thrombotic thrombocytopenic purpura	Blood Disorder	Thrombotic thrombocytopenic purpura: MedlinePlus Genetics	Added 6/24/2024
ADGRV1	Usher syndrome, type 2C	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	Added 1/6/2025
AGXT	Primary hyperoxaluria type 1	Kidney Disorder	Primary hyperoxaluria: MedlinePlus Genetics	
AKR1D1	Congenital bile acid synthesis defect 2	Metabolic Disorder	Congenital bile acid synthesis defect type 2: MedlinePlus Genetics	
ALDH7A1	Pyridoxine-dependent epilepsy	Metabolic Disorder	Pyridoxine-dependent epilepsy: MedlinePlus Genetics	
ALDOB	Hereditary fructose intolerance	Metabolic Disorder	Hereditary fructose intolerance: MedlinePlus Genetics	
ALPL	Hypophosphatasia	Metabolic Disorder	Hypophosphatasia: MedlinePlus Genetics	
ARG1	Hyperargininemia	Metabolic Disorder	Arginase deficiency: MedlinePlus Genetics	

Continued

Gene	Condition	Type of Condition	More Information	Notes
ARSA	Metachromatic Leukodystrophy	Metabolic Disorder	Metachromatic leukodystrophy: MedlinePlus Genetics	Added 6/24/2024
ARSB	Mucopolysaccharidosis type 6	Metabolic Disorder	Mucopolysaccharidosis type VI: MedlinePlus Genetics	
ASL	Argininosuccinic aciduria	Metabolic Disorder	Argininosuccinic aciduria: MedlinePlus Genetics	
ASS1	Citrullinemia type I	Metabolic Disorder	Citrullinemia: MedlinePlus Genetics	
ATP6V0A4	Distal renal tubular acidosis type 3	Mutlisystem Disorder (kidney and hearing loss)	Renal Tubular Acidosis with Deafness: MedlinePlus Genetics	Added 6/24/2024
ATP6V1B1	Distal renal tubular acidosis type 2	Mutlisystem Disorder (kidney and hearing loss)	Renal Tubular Acidosis with Deafness: MedlinePlus Genetics	Added 6/24/2024
ATP7B	Wilson disease	Metabolic Disorder	Wilson disease: MedlinePlus Genetics	
BCKDHA	Maple syrup urine disease type 1A	Metabolic Disorder	Maple syrup urine disease: MedlinePlus Genetics	
BCKDHB	Maple syrup urine disease type 1B	Metabolic Disorder	Maple syrup urine disease: MedlinePlus Genetics	
BTD	Biotinidase deficiency	Metabolic Disorder	Biotinidase deficiency: MedlinePlus Genetics	
CASR	Neonatal severe primary hyperparathyroidism	Endocrine System Disorder	Familial isolated hyperparathyroidism: MedlinePlus Genetics	
CBLIF (GIF)	Intrinsic factor deficiency	Congenital Anemia	Congenital intrinsic factor deficiency - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
CBS	Classic homocystinuria	Metabolic Disorder	Homocystinuria: MedlinePlus Genetics	
CCDC103	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
CCDC39	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
CCDC40	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
CCNO	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
CDH23	Autosomal recessive nonsyndromic hearing loss 12	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
CDH23	Usher syndrome, type 1D/F	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	
CFAP300 (C11orf70)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025

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Gene	Condition	Type of Condition	More Information	Notes
CFTR	Cystic fibrosis	Respiratory System Disease	Cystic fibrosis: MedlinePlus Genetics	
CIB2	Autosomal recessive nonsyndromic hearing loss 48	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
CLDN14	Autosomal recessive nonsyndromic hearing loss 29	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
COL11A1	Stickler syndrome type 2	Multisystem disorder (hearing, vision, skeletal)	Stickler syndrome: MedlinePlus Genetics	
COL2A1	Stickler syndrome type 1	Multisystem disorder (hearing, vision, skeletal)	Stickler syndrome: MedlinePlus Genetics	
COL4A3	Alport syndrome, autosomal recessive	Multisystem disorder (hearing, vision, kidney)	Alport syndrome: MedlinePlus Genetics	
COL4A4	Alport syndrome, autosomal recessive	Multisystem disorder (hearing, vision, kidney)	Alport syndrome: MedlinePlus Genetics	
COL4A5	Alport Syndrome, X-linked	Multisystem disorder (hearing, vision, kidney)	Alport syndrome: MedlinePlus Genetics	Males and females
CPS1	Carbamoyl phosphate synthetase I deficiency disease	Metabolic Disorder	Carbamoyl phosphate synthetase I deficiency: MedlinePlus Genetics	
CPT1A	Carnitine palmitoyl transferase 1A deficiency	Metabolic Disorder	Carnitine palmitoyltransferase I deficiency: MedlinePlus Genetics	
CPT2	Carnitine palmitoyltransferase II deficiency	Metabolic Disorder	Carnitine palmitoyltransferase II deficiency: MedlinePlus Genetics	
CTNS	Cystinosis	Metabolic Disorder	Cystinosis: MedlinePlus Genetics	
CYBA	Chronic granulomatous disease	Immunodeficiency Disorder	Chronic granulomatous disease: MedlinePlus Genetics	
CYBB	Chronic granulomatous disease	Immunodeficiency Disorder	Chronic granulomatous disease: MedlinePlus Genetics	Males and females
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	Endocrine System Disorder	Congenital adrenal insuffiency with 46, XY sex reversal OR 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency - MedGen - NCBI (nih.gov)	Added 6/24/2024
CYP27A1	Cerebrotendinous xanthomatosis	Metabolic Disorder	Cerebrotendinous xanthomatosis: MedlinePlus Genetics	
CYP27B1	Vitamin D-dependent rickets, type I	Skeletal System Disorder	Vitamin D-dependent rickets: MedlinePlus Genetics	Added 6/24/2024; Reduced sensitivity

Continued

Gene	Condition	Type of Condition	More Information	Notes
DBT	Maple syrup urine disease type 2	Metabolic Disorder	Maple syrup urine disease: MedlinePlus Genetics	
DCLRE1C	Severe Combined Immunodeficiency due to DCLRE1C Deficiency	Immunodeficiency Disorder	Omenn syndrome: MedlinePlus Genetics	
DNAAF1	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAAF2	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAAF3	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAAF4 (DYX1C1)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAAF5	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAH11	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAH5	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAI1	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DNAI2	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
DUOX2	Thyroid dyshormonogenesis 6	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	Reduced sensitivity
DUOXA2	Thyroid dyshormonogenesis 5	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
ELN	Supravalvar aortic stenosis	Mulitsystem disorder (heart, skin)	Supravalvular aortic stenosis: MedlinePlus Genetics	
ESPN	Autosomal recessive nonsyndromic hearing loss 36	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	Reduced sensitivity
ETFA	Glutaric acidemia IIA	Metabolic Disorder	Glutaric acidemia type II: MedlinePlus Genetics	Added 6/24/2024
ETFB	Glutaric acidemia IIB	Metabolic Disorder	Glutaric acidemia type II: MedlinePlus Genetics	Added 6/24/2024
ETFDH	Glutaric acidemia IIC	Metabolic Disorder	Glutaric acidemia type II: MedlinePlus Genetics	Added 6/24/2024
F10	Factor X deficiency	Blood Disorder	Factor X deficiency: MedlinePlus Genetics	
F7	Factor VII deficiency	Blood Disorder	Factor VII deficiency: MedlinePlus Genetics	

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Gene	Condition	Type of Condition	More Information	Notes
F8	Hemophilia A	Blood Disorder	Hemophilia: MedlinePlus Genetics	<50% sensitivity as inversion cannot be called; males and females
F9	Hemophilia B	Blood Disorder	Hemophilia: MedlinePlus Genetics	Reduced sensitivity; males and females
FAH	Tyrosinemia type I	Metabolic Disorder	Tyrosinemia: MedlinePlus Genetics	
FBP1	Fructose-1, 6-bisphosphatase deficiency	Metabolic Disorder	https://rarediseases.org/gard-rare-disease/fructose-16-bisphosphatase-deficiency/	
FOXJ1	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
FZD4	Familial exudative vitreoretinopathy	Vision Disorder	Familial exudative vitreoretinopathy: MedlinePlus Genetics	Added 1/6/2025
G6PC1	Glycogen Storage Disease 1a	Metabolic Disorder	Glycogen storage disease type I: MedlinePlus Genetics	
G6PD	G6PD deficiency	Metabolic Disorder	Glucose-6-phosphate dehydrogenase deficiency: MedlinePlus Genetics	Males only unless females are homozygous
GAA	Glycogen storage disease II	Metabolic Disorder	Pompe disease: MedlinePlus Genetics	
GALK1	Galactokinase deficiency	Metabolic Disorder	Galactosemia: MedlinePlus Genetics	
GALNS	Mucopolysaccharidosis type 4A	Metabolic Disorder	Mucopolysaccharidosis type IV: MedlinePlus Genetics	
GALT	Galactosemia	Metabolic Disorder	Galactosemia: MedlinePlus Genetics	
GAMT	Guanidinoacetate methyltransferase deficiency	Metabolic Disorder	Guanidinoacetate methyltransferase deficiency: MedlinePlus Genetics	
GATM	AGAT deficiency	Metabolic Disorder	Arginine:glycine amidinotransferase deficiency	MedlinePlus Genetics	
GCDH	Glutaryl-CoA dehydrogenase deficiency	Metabolic Disorder	Glutaric acidemia type I: MedlinePlus Genetics	
GCH1	GTP cyclohydrolase I deficiency	Metabolic Disorder	Tetrahydrobiopterin deficiency: MedlinePlus Genetics	
GCK	Diabetes mellitus, permanent neonatal	Neonatal Diabetes	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
GIPC3	Autosomal recessive nonsyndromic hearing loss 15	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
GJB2	Autosomal recessive nonsyndromic hearing loss 1A	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	

Continued

Gene	Condition	Type of Condition	More Information	Notes
GLUD1	Hyperinsulinism hyperammonemia syndrome	Metabolic Disorder	Congenital hyperinsulinism: MedlinePlus Genetics	
GRHPR	Primary hyperoxaluria type 2	Kidney Disorder	Primary hyperoxaluria: MedlinePlus Genetics	
GRXCR1	Autosomal recessive nonsyndromic hearing loss 25	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
HADH	3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Metabolic Disorder	3-hydroxyacyl-CoA dehydrogenase deficiency: MedlinePlus Genetics	
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Metabolic Disorder	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: MedlinePlus Genetics	
HADHB	Mitochondrial trifunctional protein deficiency	Metabolic Disorder	Mitochondrial trifunctional protein deficiency: MedlinePlus Genetics	
HBB	Sickle cell disease	Blood Disorder	Sickle cell disease: MedlinePlus Genetics	
HBB	Beta thalassemia	Blood Disorder	Beta thalassemia: MedlinePlus Genetics	
HLCS	Holocarboxylase synthetase deficiency	Metabolic Disorder	Holocarboxylase synthetase deficiency: MedlinePlus Genetics	
HMGCL	3-hydroxy-3-methylglutaric aciduria	Metabolic Disorder	3-hydroxy-3-methylglutaryl-CoA lyase deficiency: MedlinePlus Genetics	
HOGA1	Primary hyperoxaluria type 3	Kidney Disorder	Primary hyperoxaluria: MedlinePlus Genetics	
HPS1	Hermansky-Pudlak syndrome 1	Multisystem disorder (vision, pigment, blood)	Hermansky-Pudlak syndrome: MedlinePlus Genetics	Cannot report common duplication
HPS4	Hermansky-Pudlak Syndrome 4	Multisystem disorder (vision, pigment, blood)	Hermansky-Pudlak syndrome: MedlinePlus Genetics	
HSD11B2	Apparent mineralocorticoid excess	Inherited Hypertension	Apparent mineralocorticoid excess - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	Endocrine System Disorder	3-beta-hydroxysteroid dehydrogenase deficiency: MedlinePlus Genetics	
HSD3B7	Congenital bile acid synthesis defect 1	Metabolic Disorder	Congenital bile acid synthesis defect type 1: MedlinePlus Genetics	
IDS	Mucopolysaccharidosis Type 2	Metabolic Disorder	Mucopolysaccharidosis type II: MedlinePlus Genetics	Reduced sensitivity; males only
IDUA	Mucopolysaccharidosis Type 1	Metabolic Disorder	Mucopolysaccharidosis type I: MedlinePlus Genetics	
IL2RG	Severe Combined Immunodeficiency, X-linked	Immunodeficiency Disorder	X-linked severe combined immunodeficiency: MedlinePlus Genetics	Males only

Continued

Gene	Condition	Type of Condition	More Information	Notes
IL7R	Severe Combined Immunodeficiency due to IL-7Ralpha Deficiency	Immunodeficiency Disorder	Omenn syndrome: MedlinePlus Genetics	
ILDR1	Autosomal recessive nonsyndromic hearing loss 42	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
INS	Diabetes mellitus, permanent neonatal	Neonatal Diabetes	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
IVD	Isovaleric acidemia	Metabolic Disorder	Isovaleric acidemia: MedlinePlus Genetics	
IYD	Thyroid dyshormonogenesis 4	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	Added 6/24/2024
JAK3	Severe Combined Immunodeficiency due to JAK3 Deficiency	Immunodeficiency Disorder	JAK3-deficient severe combined immunodeficiency: MedlinePlus Genetics	
KCNE1	Jervell and Lange-Nielsen syndrome 2	Multisystem disorder (hearing, heart)	Jervell and Lange-Nielsen syndrome: MedlinePlus Genetics	
KCNJ11	Hyperinsulinemic hypoglycemia, familial 2	Metabolic Disorder	Congenital hyperinsulinism: MedlinePlus Genetics	
KCNJ11	Diabetes mellitus, transient and permanent neonatal with neurologic features	Neonatal Diabetes	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
KCNQ1	Jervell and Lange-Nielsen syndrome 1	Multisystem disorder (hearing, heart)	Jervell and Lange-Nielsen syndrome: MedlinePlus Genetics	
KIF11	Familial exudative vitreoretinopathy	Vision Disorder	Familial exudative vitreoretinopathy: MedlinePlus Genetics	Added 1/6/2025
LHFPL5	Autosomal recessive nonsyndromic hearing loss 67	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
LIPA	Lysosomal acid lipase deficiency	Metabolic Disorder	Lysosomal acid lipase deficiency: MedlinePlus Genetics	
LMBRD1	Methylmalonic aciduria and homocystinuria type cblF	Metabolic Disorder	Methylmalonic acidemia with homocystinuria: MedlinePlus Genetics	
LOXHD1	Autosomal recessive nonsyndromic hearing loss 77	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
LRP5	Familial exudative vitreoretinopathy	Vision Disorder	Familial exudative vitreoretinopathy: MedlinePlus Genetics	Added 1/6/2025
LRRC6 (DNAAF11)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
LRTOMT	Autosomal recessive nonsyndromic hearing loss 63	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
MARVELD 2	Autosomal recessive nonsyndromic hearing loss 49	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	

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Gene	Condition	Type of Condition	More Information	Notes
MCCC1	3-Methylcrotonyl-CoA Carboxylase 1 deficiency	Metabolic Disorder	3-methylcrotonyl-CoA carboxylase deficiency: MedlinePlus Genetics	
MCCC2	3-Methylcrotonyl-CoA Carboxylase 2 deficiency	Metabolic Disorder	3-methylcrotonyl-CoA carboxylase deficiency: MedlinePlus Genetics	
MCIDAS	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
MITF	Waardenburg Syndrome type 2A	Multisystem disorder (hearing, pigment)	Waardenburg syndrome: MedlinePlus Genetics	Will not report genetic variant that increases risk for melanoma starting 6/24/2024
MMAA	Methylmalonic aciduria, cblA type	Metabolic Disorder	Methylmalonic acidemia: MedlinePlus Genetics	
MMAB	Methylmalonic aciduria, cblB type	Metabolic Disorder	Methylmalonic acidemia: MedlinePlus Genetics	
MMACHC	Methylmalonic aciduria and homocystinuria type cblC	Metabolic Disorder	Methylmalonic acidemia with homocystinuria: MedlinePlus Genetics	
MMADHC	Methylmalonic aciduria and homocystinuria type cblD	Metabolic Disorder	Methylmalonic acidemia with homocystinuria: MedlinePlus Genetics	
MMUT (MUT)	methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	Metabolic Disorder	Methylmalonic acidemia: MedlinePlus Genetics	
MPI	Congenital disorder of glycosylation type 1b	Metabolic Disorder	MPI-CDG - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
MSRB3	Autosomal recessive nonsyndromic hearing loss 74	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
MTHFR	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	Metabolic Disorder	Homocystinuria: MedlinePlus Genetics	
MTR	Methylcobalamin deficiency type cblG	Metabolic Disorder	Homocystinuria: MedlinePlus Genetics	
MTRR	Methylcobalamin deficiency type cblE	Metabolic Disorder	Homocystinuria: MedlinePlus Genetics	
MYO15A	Autosomal recessive nonsyndromic hearing loss 3	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
MYO7A	Autosomal recessive nonsyndromic hearing loss 2	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
MYO7A	Usher Syndrome type 1B	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	

Continued

Gene	Condition	Type of Condition	More Information	Notes
NAGS	N-acetylglutamate synthase deficiency	Metabolic Disorder	N-acetylglutamate synthase deficiency: MedlinePlus Genetics	
NDP	Familial exudative vitreoretinopathy	Vision Disorder	Familial exudative vitreoretinopathy: MedlinePlus Genetics	Added 1/6/2025
ODAD1 (CCDC114)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
ODAD2 (ARMC4)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025; Reduced sensitivity
ODAD3 (CCDC151)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
ODAD4 (TTC25)	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
OTC	Ornithine transcarbamylase deficiency	Metabolic Disorder	Ornithine transcarbamylase deficiency: MedlinePlus Genetics	Males and females
OTOF	Autosomal recessive nonsyndromic hearing loss 9	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
OTOG	Autosomal recessive B nonsyndromic hearing loss 18B	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
OTOGL	Autosomal recessive B nonsyndromic hearing loss 84B	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
PAH	Phenylketonuria	Metabolic Disorder	Phenylketonuria: MedlinePlus Genetics	
PAX3	Waardenburg Syndrome type 1	Multisystem disorder (hearing, pigment)	Waardenburg syndrome: MedlinePlus Genetics	
PAX8	Congenital hypothyroidism	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
PCCA	Propionic Acidemia	Metabolic Disorder	Propionic acidemia: MedlinePlus Genetics	
PCCB	Propionic Acidemia	Metabolic Disorder	Propionic acidemia: MedlinePlus Genetics	
PCDH15	Autosomal recessive nonsyndromic hearing loss 23	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
PCDH15	Usher Syndrome type 1F	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	
PDX1	Pancreatic agenesis 1	Endocrine System Disorder	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
PHEX	Hereditary hypophosphatemic rickets	Skeletal System Disorder	Hereditary hypophosphatemic rickets: MedlinePlus Genetics	Added 6/24/2024; Males and females
PHKA2	Glycogen Storage Disease, type IXa1/IXa2	Metabolic Disorder	Glycogen storage disease type IX: MedlinePlus Genetics	Males only

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Gene	Condition	Type of Condition	More Information	Notes
PHKB	Glycogen storage disease IXb	Metabolic Disorder	Glycogen storage disease type IX: MedlinePlus Genetics	
PHKG2	Glycogen storage disease IXc	Metabolic Disorder	Glycogen storage disease type IX: MedlinePlus Genetics	
PHOX2B	Central hypoventilation syndrome with or without Hirschsprung disease	Multisystem disorder (breathing, cardiac, digestion, vision)	Congenital central hypoventilation syndrome: MedlinePlus Genetics	Reduced sensitivity as only sequence variants can be detected, not polyalanine repeat expansions
PJVK (DFNB59)	Autosomal recessive nonsyndromic hearing loss 59	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
PLPBP	Epilepsy, early-onset, vitamin B6-dependent	Seizure Disorder	PLPBP Deficiency - GeneReviews® - NCBI Bookshelf (nih.gov)	Added 6/24/2024
PNPO	Pyridoxal phosphate-responsive seizures	Metabolic Disorder	Pyridoxal 5'-phosphate-dependent epilepsy: MedlinePlus Genetics	
POU1F1	Pituitary hormone deficiency, combined, 1	Neuroendocrine Disorder	Combined pituitary hormone deficiency: MedlinePlus Genetics	
PTPRQ	Autosomal recessive nonsyndromic hearing loss 84A	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	Reduced sensitivity
PTS	BH4-deficient hyperphenylalaninemia A	Metabolic Disorder	Tetrahydrobiopterin deficiency: MedlinePlus Genetics	
QDPR	Dihydropteridine reductase deficiency	Metabolic Disorder	Tetrahydrobiopterin deficiency: MedlinePlus Genetics	
RAG1	Severe Combined Immunodeficiency due to RAG1 Deficiency	Immunodeficiency Disorder	Omenn syndrome: MedlinePlus Genetics	
RAG2	Severe Combined Immunodeficiency due to RAG2 Deficiency	Immunodeficiency Disorder	Omenn syndrome: MedlinePlus Genetics	
RB1	Retinoblastoma	Inherited Cancer Presdisposition Syndrome	Retinoblastoma: MedlinePlus Genetics	
RDX	Autosomal recessive nonsyndromic hearing loss 24	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
RET	Multiple endocrine neoplasia 2B	Inherited Cancer Predisposition Syndrome	Multiple endocrine neoplasia: MedlinePlus Genetics	
RSPH1	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025

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Gene	Condition	Type of Condition	More Information	Notes
RSPH4A	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
RSPH9	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
SCNN1A	Pseudohypoaldosteronism type I	Metabolic Disorder	Pseudohypoaldosteronism type 1: MedlinePlus Genetics	
SCNN1B	Pseudohypoaldosteronism type I	Metabolic Disorder	Pseudohypoaldosteronism type 1: MedlinePlus Genetics	
SCNN1B	Liddle syndrome	Inherited Hypertension	Liddle syndrome: MedlinePlus Genetics	
SCNN1G	Liddle syndrome	Inherited Hypertension	Liddle syndrome: MedlinePlus Genetics	
SCNN1G	Pseudohypoaldosteronism type I	Metabolic Disorder	Pseudohypoaldosteronism type 1: MedlinePlus Genetics	
SLC19A3	Biotin-responsive basal ganglia disease	Metabolic Disorder	Biotin-thiamine-responsive basal ganglia disease: MedlinePlus Genetics	
SLC2A1	Dystonia 9, GLUT1 deficiency syndromes 1 and 2, autosomal dominant	Metabolic Disorder	GLUT1 deficiency syndrome: MedlinePlus Genetics	Added 6/24/2024
SLC22A5	Systemic primary carnitine deficiency disease	Metabolic Disorder	Primary carnitine deficiency: MedlinePlus Genetics	
SLC26A3	Congenital secretory chloride diarrhea 1	Gastrointestinal System Disorder	Congenital chloride diarrhea - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
SLC26A4	Autosomal recessive nonsyndromic hearing loss 4	Multisystem disorder (hearing, thyroid)	Pendred syndrome: MedlinePlus Genetics	
SLC37A4	Glycogen Storage Disease Ib/Ic	Metabolic Disorder	Glycogen storage disease type I: MedlinePlus Genetics	
SLC39A4	Acrodermatitis enteropathica	Metabolic Disorder	Acrodermatitis enteropathica - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
SLC5A5	Thyroid dyshormonogenesis 1	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
SLC52A2	Riboflavin transporter deficiency	Multisystem disorder (neurologic, hearing, vision, breathing)	Riboflavin transporter deficiency neuronopathy: MedlinePlus Genetics	Added 6/24/2024
SLC52A3	Riboflavin transporter deficiency	Multisystem disorder (neurologic, hearing, breathing)	Riboflavin transporter deficiency neuronopathy: MedlinePlus Genetics	Added 6/24/2024
SLC7A7	Lysinuric protein intolerance	Metabolic Disorder	Lysinuric protein intolerance: MedlinePlus Genetics	Added 6/24/2024

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Gene	Condition	Type of Condition	More Information	Notes
SMN1	Spinal muscular atrophy	Neuromuscular Disorder	Spinal muscular atrophy: MedlinePlus Genetics	Exon 7 deletions only
SOX10	Waardenburg syndrome type 2E	Multisystem disorder (hearing, pigment)	Waardenburg syndrome: MedlinePlus Genetics	
SPAG1	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025
SPR	Dopa responsive due to sepiapterin reductase deficiency	Metabolic Disorder	Dopa-responsive dystonia: MedlinePlus Genetics	
STAR	Congenital lipoid adrenal hyperplasia due to STAR deficiency	Endocrine System Disorder	Congenital lipoid adrenal hyperplasia due to STAR deficency - About the Disease - Genetic and Rare Diseases Information Center (nih.gov)	
TAFAZZIN (TAZ)	Barth Syndrome	Metabolic Disorder	Barth syndrome: MedlinePlus Genetics	Males only
TAT	Tyrosinemia, type II	Metabolic Disorder	Tyrosinemia: MedlinePlus Genetics	Added 6/24/2024
TBX19	Adrenocorticotropic hormone deficiency	Endocrine System Disorder	ACTH Deficiency - Symptoms, Causes, Treatment | NORD (rarediseases.org)	
TCN2	Transcobalamin II deficiency	Metabolic Disorder	Transcobalamin deficiency: MedlinePlus Genetics	
TECTA	Autosomal dominant nonsyndromic hearing loss 12	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TECTA	Autosomal recessive nonsyndromic hearing loss 21	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TG	Thyroid dyshormonogenesis 3	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
TH	TH-deficient dopa-responsive dystonia	Metabolic Disorder	Tyrosine hydroxylase deficiency: MedlinePlus Genetics	
TMC1	Autosomal recessive nonsyndromic hearing loss 7	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TMIE	Autosomal recessive nonsyndromic hearing loss 6	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TMPRSS3	Autosomal recessive nonsyndromic hearing loss 8	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TPO	Thyroid dyshormonogenesis 2A	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
TPRN	Autosomal recessive nonsyndromic hearing loss 79	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	
TRIOBP	Autosomal recessive nonsyndromic hearing loss 28	Hearing loss	Nonsyndromic hearing loss: MedlinePlus Genetics	1kb VNTR is excluded

Continued

Gene	Condition	Type of Condition	More Information	Notes
TSHB	Isolated thyroid-stimulating hormone deficiency	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
TSHR	Hypothyroidism due to TSH receptor mutations	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
TSPAN12	Familial exudative vitreoretinopathy	Vision Disorder	Familial exudative vitreoretinopathy: MedlinePlus Genetics	Added 1/6/2025
USH1C	Usher Syndrome type 1C	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	
USH1G	Usher Syndrome type 1G	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	
USH2A	Usher syndrome, type 2A	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	Added 1/6/2025
VWF	von Willibrand disease 3	Blood Disorder	Von Willebrand disease: MedlinePlus Genetics	Reduced sensitivity
WHRN	Usher syndrome, type 2D	Multisystem disorder (hearing, vision)	Usher syndrome: MedlinePlus Genetics	Added 1/62025
WT1	Wilms tumor	Inherited Cancer Predisposition Syndrome	Wilms Tumor: MedlinePlus	Added 1/6/2025
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	Immunodeficiency Disorder	ZAP70-related severe combined immunodeficiency: MedlinePlus Genetics	
ZMYND10	Primary ciliary dyskinesia	Respiratory System Disease	Primary ciliary dyskinesia: MedlinePlus Genetics	Added 1/6/2025