Set 1

Gene List

Gene	Condition	Type of Condition	More Information	Notes
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	
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	
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	
ATP7B	Wilson disease	Metabolic Disorder	Wilson disease: MedlinePlus Genetics	
BCKDHA	Maple syrup urine disease type 1A	Metabolic Disorder	Maple syrup urine disease: MedlinePlus Genetics	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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	
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	Will report digenic inheritance with PCDH15
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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
CYP27A1	Cerebrotendinous xanthomatosis	Metabolic Disorder	Cerebrotendinous xanthomatosis: MedlinePlus Genetics	
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	
DUOX2	Thyroid dyshormonogenesis 6	Congenital Hypothyroidism	Congenital hypothyroidism: MedlinePlus Genetics	
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	
F10	Factor X deficiency	Blood Disorder	Factor X deficiency: MedlinePlus Genetics	
F7	Factor VII deficiency	Blood Disorder	Factor VII deficiency: MedlinePlus Genetics	
F8	Hemophilia A	Blood Disorder	Hemophilia: MedlinePlus Genetics	<50% sensitivity due to no inversion or CNV calling; males and females
F9	Hemophilia B	Blood Disorder	Hemophilia: MedlinePlus Genetics	<50% sensitivity due to no inversion or CNV calling; 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/	
G6PC1	Glycogen Storage Disease 1a	Metabolic Disorder	Glycogen storage disease type I: MedlinePlus Genetics	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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	
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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	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
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	
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
KCNQ1	Jervell and Lange-Nielsen syndrome 1	Multisystem disorder (hearing, heart)	Jervell and Lange-Nielsen syndrome: MedlinePlus Genetics	
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	
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	
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	
MITF	Waardenburg Syndrome type 2A	Multisystem disorder (hearing, pigment)	Waardenburg syndrome: MedlinePlus Genetics	May also detect genetic variant that increases risk for melanoma
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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	
NAGS	N-acetylglutamate synthase deficiency	Metabolic Disorder	N-acetylglutamate synthase deficiency: MedlinePlus Genetics	
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	Will report digenic inheritance with CDH23
PDX1	Pancreatic agenesis 1	Endocrine System Disorder	Permanent neonatal diabetes mellitus: MedlinePlus Genetics	
PHKA2	Glycogen Storage Disease, type IXa1/IXa2	Metabolic Disorder	Glycogen storage disease type IX: MedlinePlus Genetics	Males only
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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	
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	
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
SLC19A3	Biotin-responsive basal ganglia disease	Metabolic Disorder	Biotin-thiamine-responsive basal ganglia disease: MedlinePlus Genetics	
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	
SMN1	Spinal muscular atrophy	Neuromuscular Disorder	Spinal muscular atrophy: MedlinePlus Genetics	Copy number 0 only reported
SOX10	Waardenburg syndrome type 2E	Multisystem disorder (hearing, pigment)	Waardenburg syndrome: MedlinePlus Genetics	
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
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
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
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	
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	
VWF	von Willibrand disease 3	Blood Disorder	Von Willebrand disease: MedlinePlus Genetics	
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	Immunodeficiency Disorder	ZAP70-related severe combined immunodeficiency: MedlinePlus Genetics	