devACT® Panel – Gene List by Gene

Gene	Disease Association	Category
AASS	Alpha-aminoadipic acid semialdehyde dehydrogenase deficiency	Neurotransmitter disorders
ABAT	GABA-transaminase deficiency	Neurotransmitter disorders
ABCD1	Adrenoleukodystrophy	Peroxisomal disorder
ABCD3	Zellweger syndrome 2	Peroxisomal disorder
ABCD4	Methylmalonic acidemia and homocystinuria type CblJ	Organic acidemia
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	Organic acidemia
ACAD9	Mitochondrial complex I deficiency	Mitochondria-related disorder
ACADM	Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	Fatty Acid oxidation disorders
ACADS	Short chain acyl-CoA dehydrogenase (SCAD) deficiency	Fatty Acid oxidation disorders
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)	Fatty Acid oxidation disorders
ACAT1	Acetoacetyl-CoA-thiolase deficiency (ketothiolase deficiency)	Organic acidemia
ACOX1	Peroxisomal acyl-CoA oxidase deficiency (pseudoneonatal adrenoleukodystrophy)	Peroxisomal disorder
ADCK3	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
ADSL	Adenylosuccinate lyase (ADSL) deficiency, adenylosuccinase deficiency, autistic spectrum disorder, succinylpurinemic autism	Purine/pyrimidine metabolism
AGA	Lysosomal storage disease aspartylglycosaminuria	Lysosomal storage disorder
ALDH3A2	Sjogren-Larsson syndrome	Lipid metabolism disorder
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria)	Neurotransmitter disorders
ALDH7A1	Pyridoxine dependent epilepsy	Cofactor disorder
ALG1	Congenital disorder of glycosylation 1k	Congenital disorder of glycosylation
ALG11	Congenital disorder of glycosylation 1p	Congenital disorder of glycosylation
ALG12	Congenital disorder of glycosylation 1g	Congenital disorder of glycosylation
ALG13	Congenital disorder of glycosylation 1s	Congenital disorder of glycosylation
ALG2	Congenital disorder of glycosylation 1i	Congenital disorder of glycosylation
ALG3	Congenital disorder of glycosylation 1d	Congenital disorder of glycosylation
ALG6	Congenital disorder of glycosylation 1c	Congenital disorder of glycosylation
ALG8	Congenital disorder of glycosylation 1h	Congenital disorder of glycosylation
ALG9	Congenital disorder of glycosylation 1L	Congenital disorder of glycosylation
AMN	Megaloblastic anemia due to inborn errors of metabolism	Cofactor disorder
AMT	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
APTX	Ataxia-Oculomotor Apraxia	Coenzyme Q10 deficiency
ARG1	Arginase deficiency	Urea cycle disorders
ARSA	Metachromatic leukodystrophy	Lysosomal storage disorder
ARSB	Mucopolysaccharidosis type 6 (MPS 6)	Lysosomal storage disorder
ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Lysosomal storage disorder
ASL	Spinal muscular atrophy with progressive myoclonic epilepsy	Urea cycle disorders
ASPA	Canavan disease	Leukodystrophy
ASS1	Citrullinemia - Several forms: acute neonatal, milder later onset, pregnancy/postpartum in women, asymptomatic	Urea cycle disorders
ATP13A2	Neuronal ceroid lipofuscinosis 12 (CLN 12) - Juvenile NCL	Neuronal ceroid lipofuscinosis
ATP7A	Menkes disease - classic and mild forms	Copper metabolism disorder
ATP7B	Wilson disease	Copper metabolism disorder
AUH	3-methylglutaconic aciduria type I (MCGA1) disorder	Organic acidemia
B4GALT1	Congenital disorder of glycosylation IId	Congenital disorder of glycosylation
BCKDHA	Maple syrup urine disease type 1a	Organic acidemia
BCKDHB	Maple syrup urine disease type 1b	Organic acidemia

Gene	Disease Association	Category
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency (2-methylbutaryl-CoA dehydrogenase deficiency, SBCADD)	Organic acidemia
BTD	Biotinidase deficiency	Cofactor disorder
CACNA1H	Epilepsy, childhood absence, 6; epilepsy, idopathic generalized, 6	Brain malformation and seizure disorders
CBS	Homocystinuria, B6-responsive and nonresponsive types; thrombosis, hyperhomocysteinemia	Cofactor disorder
CHAT	Myasthenic syndrome, congenital, associated with episodic apnea	Neurotransmitter disorders
CISD2	Wolfram syndrome 2	Mitochondria-related disorder
CLN3	Neuronal ceroid lipofuscinosis 3 (CLN 3) - Classic juvenile NCL ("Spielmeyer-Sjögren"), adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
CLN5	Neuronal ceroid lipofuscinosis 5 (CLN 5) - Late infantile NCL ("Finnish variant"), Juvenile NCL, Adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
CLN6	Neuronal ceroid lipofuscinosis 6 (CLN 6) - Late infantile NCL ("Lake-Cavanagh or Indian variant"), adult NCL (Kufs disease type A)	Neuronal ceroid lipofuscinosis
CLN8	Neuronal ceroid lipofuscinosis 8 (CLN 8) - Late infantile NCL, Northern epilepsy (progressive epilepsy with mental retardation or EPMR)	Neuronal ceroid lipofuscinosis
COG1	Congenital disorder of glycosylation IIg	Congenital disorder of glycosylation
COG4	Congenital disorder of glycosylation IIj	Congenital disorder of glycosylation
COG5	Congenital disorders of glycosylation IIi	Congenital disorder of glycosylation
COG6	Congenital disorder of glycosylation IIL	Congenital disorder of glycosylation
COG7	Congenital disorder of glycosylation IIe	Congenital disorder of glycosylation
COG8	Congenital disorder of glycosylation IIh	Congenital disorder of glycosylation
COQ2	Coenzyme Q10 deficiency; COQ2 nephropathy	Coenzyme Q10 deficiency
COQ9	Autosomal-recessive neonatal-onset primary coenzyme Q10 deficiency.	Mitochondria-related disorder
CP	Deficiency of ferroxidase (aceruloplasminemia)	Copper metabolism disorder
CPS1	Carbamoylphosphate synthetase 1 deficiency, hyperammonemia	Urea cycle disorders
CPT1A	Carnitine palmitoyltransferase 1 deficiency - Hepatic encephalopathy form, adult myopathy form, or acute fatty liver of pregnancy form	Fatty Acid oxidation disorders
CPT2	Carnitine palmitoyltransferase 2 deficiency - Lethal neonatal form, severe infantile hepatocardiomuscular form, or myopathic form	Fatty Acid oxidation disorders
CTNS	Cystinosis	Lysosomal storage disorder
CTSA	Galactosialidosis	Lysosomal storage disorder
CTSD	Neuronal ceroid lipofuscinosis 10 (CLN 10) - Congenital NCL, Late-infantile NCL, or Teenage-/adult-onset NCL	Neuronal ceroid lipofuscinosis
CTSF	Neuronal ceroid lipofuscinosis 13 (CLN 13) - Adult NCL (Kufs disease type B)	Neuronal ceroid lipofuscinosis
CUBN	Megaloblastic anemia due to inborn errors of metabolism	Congenital disorder of glycosylation
CYP27A1	Cerebrotendinous xanthomatosis	Brain malformation and seizure disorders
DBH	Dopamine beta-hydroxylase deficiency	Neurotransmitter disorders
DBT	Maple syrup urine disease type 2	Organic acidemia
DDC	aromatic l-amino acid decarboxylase deficiency	Neurotransmitter disorders
DDOST	Congenital Disorders of Glycosylation-1r	Congenital disorder of glycosylation
DHCR7	Smith-Lemli-Opitz syndrome	Lipid metabolism disorder
DHDDS	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency	Folate metabolism
DLAT	Pyruvate dehydrogenase E2 deficiency	Mitochondria-related disorder
DLD	Dihydrolipoamide dehydrogenase deficiency	Mitochondria-related disorder
DNAJC5	Neuronal ceroid lipofuscinosis 4 (CLN 4) - Adult NCL (Parry disease)	Neuronal ceroid lipofuscinosis
DOLK	Congenital disorder of glycosylation, Cardiomyopathy, dilated	Congenital disorder of glycosylation
DPAGT1	Congenital disorder of glycosylation 1j	Congenital disorder of glycosylation
DPM1	Congenital disorder of glycosylation 1e	Congenital disorder of glycosylation
DPM2	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
DPM3	Alpha-dystroglycanopathy	Congenital disorder of glycosylation
DPYD	Dihydropyrimidine dehyrogenase deficiency	Purine/pyrimidine metabolism
EIF2B1	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B2	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B3	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter

Gene	Disease Association	Category
EIF2B4	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B5	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
ETFA	Glutaric Acidemia IIA (Multiple Acyl-CoA Dehydrogenase Deficiency)	Fatty Acid oxidation disorders
ETFB	Glutaric acidemia IIB	Fatty Acid oxidation disorders
ETFDH	Glutaric acidemia IIC	Fatty Acid oxidation disorders
ETHE1	Encephalopathy, ethylmalonic	Organic acidemia
FKRP	Muscular dystrophy-dystroglycanopathy (MDDG): a severe congenital form with brain and eye anomalies (type A5; MDDGA5), formerly designated Walker-Warburg syndrome (WWS) or muscle-eye-brain disease (MEB); a less severe congenital form with or without mental retardation (type B5; MDDGB5); and a milder limb-girdle form (type C5; MDDGC5), previously designated LGMD2I.	Congenital disorder of glycosylation
FKTN	Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X).	Congenital disorder of glycosylation
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	Folate metabolism
FOLR2	Potential disorder folate metabolism	Folate metabolism
FPGS	Potential disorder of folate metabolism	Folate metabolism
FTCD	Glutamate formiminotransferase deficiency	Folate metabolism
FUCA1	Fucosidosis	Lysosomal storage disorder
GAA	Pompe disease	Lysosomal storage disorder
GALC	Krabbe disease	Lysosomal storage disorder
GALNS	Mucopolysaccharidosis type IV-A (MPS 4A); Morquio syndrome	Lysosomal storage disorder
GAMT	cerebral creatine deficiency syndrome 2	Creatine metabolism
GATM	cerebral creatine deficiency syndrome 3	Creatine metabolism
GCDH	Glutaric acidemia I	Organic acidemia
GCH1	GTP cyclohydrolase deficiency (Hyperphenylalaninemia, BH4-deficient, B)	Neurotransmitter disorders
GCSH	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
GFAP	Alexander disease	Leukodystrophy
GIF	Intrinsic factor deficiency	Cofactor disorder
GK	Glycerol kinase deficiency	Lipid metabolism disorder
GLA	Fabry disease	Lysosomal storage disorder
GLB1	GM1 gangliosidosis	Lysosomal storage disorder
GLDC	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
GLUD1	Hyperinsulinism-hyperammonemia syndrome	Fatty Acid oxidation disorders
GM2A	GM2 gangliosidoses type AB	Lysosomal storage disorder
GMPPA	Congenital disorder of glycosylation	Congenital disorder of glycosylation
GNE	Sialuria; autosomal recessive inclusion body myopathy; Nonaka myopathy	Lysosomal storage disorder
GNPAT	Rhizomelic chondrodysplasia punctata type II - a peroxisomal disorder	Mitochondria-related disorder
GNPTAB	Mucolipidoses II (I-cell disease) & III (pseudo-Hurler polydystrophy)	Lysosomal storage disorder
GNPTG	Mucolipidosis III gamma	Lysosomal storage disorder
GNS	Mucopolysaccharidosis type IIID, Sanfilippo syndrome D	Lysosomal storage disorder
GRN	Neuronal ceroid lipofuscinosis 11 (CLN 11) - Adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
GUSB	Mucopolysaccharidosis VII	Lysosomal storage disorder
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency; Hypoglycemia, hyperinsulinemic	Fatty Acid oxidation disorders
HADHA	Long-chain-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); trifunctional protein deficiency	Fatty Acid oxidation disorders
HADHB	Long-chain-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); trifunctional protein deficiency	Fatty Acid oxidation disorders
HEXA	Tay-Sachs disease (GM2 gangliosidosis B)	Lysosomal storage disorder
HEXB	Sandhoff disease (GM2 gangliosidosis type O)	Lysosomal storage disorder
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)	Lysosomal storage disorder
HLCS	Holocarboxylase synthetase deficiency	Cofactor disorder
HMGCL	HMG-CoA lyase deficiency	Fatty Acid oxidation disorders
HMGCS2	HMG-CoA synthase-2 deficiency	Fatty Acid oxidation disorders
HPRT1	HPRT-related gout; Lesch-Nyhan syndrome	Purine/pyrimidine metabolism

Gene	Disease Association	Category
HSD17B10	Hydroxysteroid (17-beta) dehydrogenase 10; 3-hydroxy-2-methylbutyryl-CoA dehydrogenase deficiency	Amino acid disorder
HYAL1	Mucopolysaccharidosis IX	Lysosomal storage disorder
IDS	Mucopolysaccharidosis II, Hunter disease	Lysosomal storage disorder
IDUA	Hurler-Scheie syndromes	Lysosomal storage disorder
ISPD	Walker-Warburg syndrome	Congenital disorder of glycosylation
IVD	Isovaleric acidemia	Organic acidemia
KCTD7	Neuronal ceroid lipofuscinosis 14 (CLN 14) - Late infantile NCL; progressive myoclonic epilepsy 3	Neuronal ceroid lipofuscinosis
L2HGDH	L-2-hydroxyglutaric aciduria	Organic acidemia
LARGE	Congenital muscular dystrophy type 1D (MDC1D)	Congenital disorder of glycosylation
LARS2	Perrault syndrome	Mitochondria-related disorder
LIPA	Cholesteryl ester storage disease; Wolman disease	Endoplasmic reticulum
LMBRD1	Methylmalonic aciduria and homocystinuria, cobalamin F type	Organic acidemia
MAGT1	X-linked immunodeficiency with Mg(2+)defect, EBV infection and neoplasia	Transport disorders
MAN1B1	Non-syndromic Autosomal Recessive Intellectual Disability 15 (N-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
MAN2B1	α-Mannosidosis	Lysosomal storage disorder
MANBA	β-Mannosidosis	Lysosomal storage disorder
MCCC1	Elevated 3-hydroxyisovalerylcarnitine / 3-methylcrotonylglycine levels	Organic acidemia
MCCC2	3-methylcrotonyl-CoA carboxylase deficiency	Organic acidemia
MCEE	Methylmalonic coA epimerase deficiency	Organic acidemia
MFSD8	Neuronal ceroid lipofuscinosis 7 (CLN7) - Late infantile NCL ("Turkish variant")	Neuronal ceroid lipofuscinosis
MGAT2	Congenital disorder of glycosylation 2a	Congenital disorder of glycosylation
MLYCD	Malonyl-CoA decarboxylase deficiency	Organic acidemia
MMAA	Methylmalonic acidaemia	Organic acidemia
MMAB	Methylmalonic aciduria	Organic acidemia
MMACHC	Methylmalonic aciduria and homocystinuria, cobalamin C type	Organic acidemia
MMADHC	Cobalamin disorders	Organic acidemia
MOCS1	Molybdenum cofactor deficiency A	Organic acidemia
MOCS2	Combined molybdenum cofactor deficiency/MOCS2 (type B deficiency).	Cofactor disorder
MOGS	Congenital disorder of glycosylation	Congenital disorder of glycosylation
MPDU1	Congenital disorder of glycosylation 1f	Congenital disorder of glycosylation
MPI	Congenital disorder of glycosylation 1b	Congenital disorder of glycosylation
MTHFR	Homocystinuria due to MTHFR deficiency; neural tube defects, susceptibility to; schizophrenia, susceptibility to; thromboembolism, susceptibility to; vascular disease, susceptibility to	Folate metabolism
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type; neural tube defects, folate-sensitive, susceptibility to	Folate metabolism
MTRR	Homocystinuria-megaloblastic anemia, cbl E type; neural tube defect, folate sensitive, susceptibility to	Folate metabolism
MUT	methylmalonic acidemia	Organic acidemia
NAGA	Schindler disease; Kanzaki disease	Lysosomal storage disorder
NAGLU	mucopolysaccharidosis type IIIB (Sanfilippo B)	Lysosomal storage disorder
NAGS	NAGS deficiency	Urea cycle disorders
NDUFA1	Mitochondrial complex 1 deficiency	Mitochondria-related disorder
NEU1	Sialidosis	Lysosomal storage disorder
NPC1	Niemann-Pick type C disease	Lysosomal storage disorder
NPC2	Niemann-pick disease, type C2	Lysosomal storage disorder
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency	Purine/pyrimidine metabolism
OTC	Ornithine transcarbamylase deficiency	Urea cycle disorders
OXCT1	Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency	Organic acidemia
PAH	Phenylketonuria (PKU), hyperphenylalaninemia	Amino acid disorder
PANK2	Pantothenate kinase-associated neurodegeneration/HARP syndrome	Neurodegeneration
PCBD1	Tetrahydrobiopterin (BH4)-deficient hyperphenylalaninemia (HPA)	Amino acid disorder
PCCA	Propionic acidemia	Organic acidemia
PCCB	Propionic acidemia	Organic acidemia
PDHA1	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDHB	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder

Gene	Disease Association	Category
PDHX	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDP1	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDSS1	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
PDSS2	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
PEX1	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX10	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX12	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX13	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX14	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX16	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX19	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX2	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX26	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX3	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX5	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX6	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX7	peroxisome biogenesis disorder 9B; rhizomelic chondrodysplasia punctata, type 1	Peroxisomal disorder
PGK1	Phosphoglycerate kinase deficiency	Glycolysis disorder
PGM1	Congenital disorder of glycosylation, type It	Congenital disorder of glycosylation
PHGDH	Phosphoglycerate dehydrogenase deficiency	Amino acid disorder
PHYH	Refsum disease (hereditary motor sensory neuropathy type IV, heredopathia atactica polyneuritiformis)	Peroxisomal disorder
PMM2	Phosphomannomutase 2 deficiency (congenital defect of glycosylation type 1a)	Congenital disorder of glycosylation
PNPO	Pyridoxamine phosphate oxidase deficiency	Cofactor disorder
POLG	Progressive external ophthalmoplegia with mitochondrial DNA deletions 1 (PEOA1); Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis (SANDO); alpers-huttenlocher syndrome (AHS); and mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE)	Mitochondria-related disorder
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3; Muscular dystrophy-dystroglycanopathy (congenital with intellectual disability), type B, 3; Muscular dystrophy-dystroglycanopathy (limbe-girdle), type C, 3 (O-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1; Muscular dystrophy-dystroglycanopathy (congenital with intellectual disability), type B, 1; Muscular dystrophy-dystroglycanopathy (limbe-girdle), type C, 1 (O-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2; congenital muscular dystrophy-dystroglycanopathy with intellectual disability; limb-girdle muscular dystrophy-dystroglycanopathy	Congenital disorder of glycosylation
PPT1	Neuronal ceroid lipofuscinosis 1 (CLN 1) - Infantile classic NCL ("Haltia-Santavuori" type), late infantile NCL, juvenile NCL, adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	Purine/pyrimidine metabolism
PSAP	Krabbe disease; Gaucher disease	Lysosomal storage disorder
PSAT1	Phosphoserine aminotransferase deficiency	Amino acid disorder
PSPH	Deficiency of phosphoserine phosphatase	Amino acid disorder
PTS	6-pyruvoyltetrahydropterin synthase deficiency	Pterin disorder
QDPR	Quinoid dihydropteridine reductase deficiency	Pterin disorder
RFT1	N-linked glycosylation disorder	Congenital disorder of glycosylation
SCN1A	Dravet syndrome; epilepsy, generalized, with febrile seizures plus, type 2; febrile seizures, familial, 3A; migraine, familial hemiplegic, 3	Brain malformation and seizure disorders
SGSH	Mucopolysaccharidosis type IIIA (sanfilippo A)	Lysosomal storage disorder
SLC16A2	Allan-Herndon-Dudley syndrome	Transport disorders
SLC17A5	Sialic Acid Storage Disease; Salla Disease	Lysosomal storage disorder
SLC19A1	Cerebral folate transport deficiency	Transport disorders
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	Cofactor disorder

Gene	Disease Association	Category
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2); can appear as Leigh disease or Wernicke encephalopathy	Transport disorders
SLC22A5	Carnitine deficiency, systemic primary	Fatty Acid oxidation disorders
SLC25A12	Global cerebral hypomyelination	Transport disorders
SLC25A13	Citrullinemia, type II, neonatal-onset; Citrullinemia, adult-onset type II	Transport disorders
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome.	Amino acid disorder
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Transport disorders
SLC25A32	Mitochondrial inner membrane folate transporter deficiency	Transport disorders
SLC2A1	Glucose transporter deficiency syndrome 1 (GLUT1); GLUT1 deficiency syndrome 2; dystonia 9; susceptibility to idiopathic generalized epilepsy 12	Transport disorders
SLC35A1	Congenital disorder of glycosylation, type 2f	Congenital disorder of glycosylation
SLC35A2	Congenital disorder of glycosylation, type 2m	Congenital disorder of glycosylation
SLC35C1	Congenital disorder of glycosylation 2c	Congenital disorder of glycosylation
SLC46A1	Hereditary folate malabsorption	Transport disorders
SLC6A3	Hereditary dopamine transporter deficiency syndrome	Neurotransmitter disorders
SLC6A4	Serotonin transporter deficiency	Transport disorders
SLC6A8	Cerebral creatine deficiency syndrome 1	Transport disorders
SMPD1	Niemann-Pick Types A & B	Lysosomal storage disorder
SPR	Sepiapterin reductase deficiency	Pterin disorder
SRD5A3	Congenital Disordes of Glycosylation-1q	Congenital disorder of glycosylation
SSR4	Congenital disorder of glycosylation	Congenital disorder of glycosylation
ST3GAL3	Early infantile epileptic encephalopathy 15; Autosomal recessive intellectual disability 12	Congenital disorder of glycosylation
STT3A	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
STT3B	Congenital disorders of glycosylation	Congenital disorder of glycosylation
SUMF1	Multiple sulfatase deficiency	Lysosomal storage disorder
SUOX	Sulfite oxidase deficiency	Amino acid disorder
TAT	Tyrosinemia (type II, Richner-Hanhart syndrome), a disorder accompanied by major skin and corneal lesions, with possible mental retardation, X-linked	Amino acid disorder
TH	Tyrosine hydroxylase deficiency	Neurotransmitter disorders
TIMM8A	Mohr-Tranebjaerg syndrome, Jensen syndrome	Mitochondria-related disorder
TMEM165	Congenital Disorders of Glycosylation-IIk	Congenital disorder of glycosylation
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency	Carnitine synthesis disorder
TPH1	Tryptophan hydroxylase 1 deficiency	Neurotransmitter disorders
TPH2	Tryptophan hydroxylase 2 deficiency	Neurotransmitter disorders
TPP1	Neuronal ceroid lipofuscinosis 2 (CLN 2) - Late infantile classic NCL ("Janský-Bielschowsky" type), juvenile NCL, possibly later-onset type NCL	Neuronal ceroid lipofuscinosis
TRAP1	***TRAP1 related disorder (T1ReD)	Mitochondria-related disorder
TUBA1A	Lissencephaly 3	Brain malformation and seizure disorders
TUSC3	Congenital disorder of glycosylation type 1t	Congenital disorder of glycosylation

devACT® Panel – Gene List by Category

Gene	Disease Association	Category
AMT	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
GCSH	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
GLDC	Glycine encephalopathy (non-ketotic hyperglycinemia)	Amino acid disorder
HSD17B10	Hydroxysteroid (17-beta) dehydrogenase 10; 3-hydroxy-2-methylbutyryl-CoA dehydrogenase deficiency	Amino acid disorder
PAH	Phenylketonuria (PKU), hyperphenylalaninemia	Amino acid disorder
PCBD1	Tetrahydrobiopterin (BH4)-deficient hyperphenylalaninemia (HPA)	Amino acid disorder
PHGDH	Phosphoglycerate dehydrogenase deficiency	Amino acid disorder
PSAT1	Phosphoserine aminotransferase deficiency	Amino acid disorder
PSPH	Deficiency of phosphoserine phosphatase	Amino acid disorder
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome.	Amino acid disorder
SUOX	Sulfite oxidase deficiency	Amino acid disorder
TAT	Tyrosinemia (type II, Richner-Hanhart syndrome), a disorder accompanied by major skin and corneal lesions, with possible mental retardation, X-linked	Amino acid disorder
CACNA1H	Epilepsy, childhood absence, 6; epilepsy, idopathic generalized, 6	Brain malformation and seizure disorders
CYP27A1	Cerebrotendinous xanthomatosis	Brain malformation and seizure disorders
SCN1A	Dravet syndrome; epilepsy, generalized, with febrile seizures plus, type 2; febrile seizures, familial, 3A; migraine, familial hemiplegic, 3	Brain malformation and seizure disorders
TUBA1A	Lissencephaly 3	Brain malformation and seizure disorders
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency	Carnitine synthesis disorder
ADCK3	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
APTX	Ataxia-Oculomotor Apraxia	Coenzyme Q10 deficiency
COQ2	Coenzyme Q10 deficiency; COQ2 nephropathy	Coenzyme Q10 deficiency
PDSS1	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
PDSS2	Coenzyme Q10 deficiency	Coenzyme Q10 deficiency
ALDH7A1	Pyridoxine dependent epilepsy	Cofactor disorder
AMN	Megaloblastic anemia due to inborn errors of metabolism	Cofactor disorder
BTD	Biotinidase deficiency	Cofactor disorder
CBS	Homocystinuria, B6-responsive and nonresponsive types; thrombosis, hyperhomocysteinemia	Cofactor disorder
GIF	Intrinsic factor deficiency	Cofactor disorder
HLCS	Holocarboxylase synthetase deficiency	Cofactor disorder
MOCS2	Combined molybdenum cofactor deficiency/MOCS2 (type B deficiency).	Cofactor disorder
PNPO	Pyridoxamine phosphate oxidase deficiency	Cofactor disorder
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	Cofactor disorder
ALG1	Congenital disorder of glycosylation 1k	Congenital disorder of glycosylation
ALG11	Congenital disorder of glycosylation 1p	Congenital disorder of glycosylation
ALG12	Congenital disorder of glycosylation 1g	Congenital disorder of glycosylation
ALG13	Congenital disorder of glycosylation 1s	Congenital disorder of glycosylation
ALG2	Congenital disorder of glycosylation 1i	Congenital disorder of glycosylation
ALG3	Congenital disorder of glycosylation 1d	Congenital disorder of glycosylation
ALG6	Congenital disorder of glycosylation 1c	Congenital disorder of glycosylation
ALG8	Congenital disorder of glycosylation 1h	Congenital disorder of glycosylation
ALG9	Congenital disorder of glycosylation 1L	Congenital disorder of glycosylation
B4GALT1	Congenital disorder of glycosylation IId	Congenital disorder of glycosylation
COG1	Congenital disorder of glycosylation IIg	Congenital disorder of glycosylation
COG4	Congenital disorder of glycosylation IIj	Congenital disorder of glycosylation
COG5	Congenital disorders of glycosylation IIi	Congenital disorder of glycosylation
COG6	Congenital disorder of glycosylation IIL	Congenital disorder of glycosylation
COG7	Congenital disorder of glycosylation IIe	Congenital disorder of glycosylation
COG8	Congenital disorder of glycosylation IIh	Congenital disorder of glycosylation
CUBN	Megaloblastic anemia due to inborn errors of metabolism	Congenital disorder of glycosylation
DDOST	Congenital Disorders of Glycosylation-1r	Congenital disorder of glycosylation
DHDDS	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
DOLK	Congenital disorder of glycosylation, Cardiomyopathy, dilated	Congenital disorder of glycosylation
DPAGT1	Congenital disorder of glycosylation 1j	Congenital disorder of glycosylation

Gene	Disease Association	Category
DPM1	Congenital disorder of glycosylation 1e	Congenital disorder of glycosylation
DPM2	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
DPM3	Alpha-dystroglycanopathy	Congenital disorder of glycosylation
FKRP	Muscular dystrophy-dystroglycanopathy (MDDG): a severe congenital form with brain and eye anomalies (type A5; MDDGA5), formerly designated Walker-Warburg syndrome (WWS) or muscle-eye-brain disease (MEB); a less severe congenital form with or without mental retardation (type B5; MDDGB5); and a milder limb-girdle form (type C5; MDDGC5), previously designated LGMD2I.	Congenital disorder of glycosylation
FKTN	Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X).	Congenital disorder of glycosylation
GMPPA	Congenital disorder of glycosylation	Congenital disorder of glycosylation
ISPD	Walker-Warburg syndrome	Congenital disorder of glycosylation
LARGE	Congenital muscular dystrophy type 1D (MDC1D)	Congenital disorder of glycosylation
MAN1B1	Non-syndromic Autosomal Recessive Intellectual Disability 15 (N-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
MGAT2	Congenital disorder of glycosylation 2a	Congenital disorder of glycosylation
MOGS	Congenital disorder of glycosylation	Congenital disorder of glycosylation
MPDU1	Congenital disorder of glycosylation 1f	Congenital disorder of glycosylation
MPI	Congenital disorder of glycosylation 1b	Congenital disorder of glycosylation
PGM1	Congenital disorder of glycosylation, type It	Congenital disorder of glycosylation
PMM2	Phosphomannomutase 2 deficiency (congenital defect of glycosylation type 1a)	Congenital disorder of glycosylation
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3; Muscular dystrophy-dystroglycanopathy (congenital with intellectual disability), type B, 3; Muscular dystrophy-dystroglycanopathy (limbe-girdle), type C, 3 (O-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1; Muscular dystrophy-dystroglycanopathy (congenital with intellectual disability), type B, 1; Muscular dystrophy-dystroglycanopathy (limbe-girdle), type C, 1 (O-linked congenital disorder of glycosylation)	Congenital disorder of glycosylation
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2; congenital muscular dystrophy-dystroglycanopathy with intellectual disability; limb-girdle muscular dystrophy-dystroglycanopathy	Congenital disorder of glycosylation
RFT1	N-linked glycosylation disorder	Congenital disorder of glycosylation
SLC35A1	Congenital disorder of glycosylation, type 2f	Congenital disorder of glycosylation
SLC35A2	Congenital disorder of glycosylation, type 2m	Congenital disorder of glycosylation
SLC35C1	Congenital disorder of glycosylation 2c	Congenital disorder of glycosylation
SRD5A3	Congenital Disordes of Glycosylation-1q	Congenital disorder of glycosylation
SSR4	Congenital disorder of glycosylation	Congenital disorder of glycosylation
ST3GAL3	Early infantile epileptic encephalopathy 15; Autosomal recessive intellectual disability 12	Congenital disorder of glycosylation
STT3A	Congenital Disorders of Glycosylation	Congenital disorder of glycosylation
STT3B	Congenital disorders of glycosylation	Congenital disorder of glycosylation
TMEM165	Congenital Disorders of Glycosylation-IIk	Congenital disorder of glycosylation
TUSC3	Congenital disorder of glycosylation type 1t	Congenital disorder of glycosylation
ATP7A	Menkes disease - classic and mild forms	Copper metabolism disorder
ATP7B	Wilson disease	Copper metabolism disorder
CP	Deficiency of ferroxidase (aceruloplasminemia)	Copper metabolism disorder
GAMT	cerebral creatine deficiency syndrome 2	Creatine metabolism
GATM	cerebral creatine deficiency syndrome 3	Creatine metabolism
LIPA	Cholesteryl ester storage disease; Wolman disease	Endoplasmic reticulum
ACADM	Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	Fatty Acid oxidation disorders
ACADS	Short chain acyl-CoA dehydrogenase (SCAD) deficiency	Fatty Acid oxidation disorders
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)	Fatty Acid oxidation disorders
CPT1A	Carnitine palmitoyltransferase 1 deficiency - Hepatic encephalopathy form, adult myopathy form, or acute fatty liver of pregnancy form	Fatty Acid oxidation disorders

Gene	Disease Association	Category
CPT2	Carnitine palmitoyltransferase 2 deficiency - Lethal neonatal form, severe infantile hepatocardiomuscular form, or myopathic form	Fatty Acid oxidation disorders
ETFA	Glutaric Acidemia IIA (Multiple Acyl-CoA Dehydrogenase Deficiency)	Fatty Acid oxidation disorders
ETFB	Glutaric acidemia IIB	Fatty Acid oxidation disorders
ETFDH	Glutaric acidemia IIC	Fatty Acid oxidation disorders
GLUD1	Hyperinsulinism-hyperammonemia syndrome	Fatty Acid oxidation disorders
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency; Hypoglycemia, hyperinsulinemic	Fatty Acid oxidation disorders
HADHA	Long-chain-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); trifunctional protein deficiency	Fatty Acid oxidation disorders
HADHB	Long-chain-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); trifunctional protein deficiency	Fatty Acid oxidation disorders
HMGCL	HMG-CoA lyase deficiency	Fatty Acid oxidation disorders
HMGCS2	HMG-CoA synthase-2 deficiency	Fatty Acid oxidation disorders
SLC22A5	Carnitine deficiency, systemic primary	Fatty Acid oxidation disorders
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency	Folate metabolism
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	Folate metabolism
FOLR2	Potential disorder folate metabolism	Folate metabolism
FPGS	Potential disorder of folate metabolism	Folate metabolism
FTCD	Glutamate formiminotransferase deficiency	Folate metabolism
MTHFR	Homocystinuria due to MTHFR deficiency; neural tube defects, susceptibility to; schizophrenia, susceptibility to; thromboembolism, susceptibility to; vascular disease, susceptibility to	Folate metabolism
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type; neural tube defects, folate-sensitive, susceptibility to	Folate metabolism
MTRR	Homocystinuria-megaloblastic anemia, cbl E type; neural tube defect, folate sensitive, susceptibility to	Folate metabolism
PGK1	Phosphoglycerate kinase deficiency	Glycolysis disorder
ASPA	Canavan disease	Leukodystrophy
GFAP	Alexander disease	Leukodystrophy
EIF2B1	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B2	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B3	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B4	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
EIF2B5	Leukodystrophy vanishing white matter	Leukodystrophy with vanishing white matter
ALDH3A2	Sjogren-Larsson syndrome	Lipid metabolism disorder
DHCR7	Smith-Lemli-Opitz syndrome	Lipid metabolism disorder
GK	Glycerol kinase deficiency	Lipid metabolism disorder
AGA	Lysosomal storage disease aspartylglycosaminuria	Lysosomal storage disorder
ARSA	Metachromatic leukodystrophy	Lysosomal storage disorder
ARSB	Mucopolysaccharidosis type 6 (MPS 6)	Lysosomal storage disorder
ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Lysosomal storage disorder
CTNS	Cystinosis	Lysosomal storage disorder
CTSA	Galactosialidosis	Lysosomal storage disorder
FUCA1	Fucosidosis	Lysosomal storage disorder
GAA	Pompe disease	Lysosomal storage disorder
GALC	Krabbe disease	Lysosomal storage disorder
GALNS	Mucopolysaccharidosis type IV-A (MPS 4A); Morquio syndrome	Lysosomal storage disorder
GLA	Fabry disease	Lysosomal storage disorder
GLB1	GM1 gangliosidosis	Lysosomal storage disorder
GM2A	GM2 gangliosidoses type AB	Lysosomal storage disorder
GNE	Sialuria; autosomal recessive inclusion body myopathy; Nonaka myopathy	Lysosomal storage disorder
GNPTAB	Mucolipidoses II (I-cell disease) & III (pseudo-Hurler polydystrophy)	Lysosomal storage disorder

Gene	Disease Association	Category
GNPTG	Mucolipidosis III gamma	Lysosomal storage disorder
GNS	Mucopolysaccharidosis type IIID, Sanfilippo syndrome D	Lysosomal storage disorder
GUSB	Mucopolysaccharidosis VII	Lysosomal storage disorder
HEXA	Tay-Sachs disease (GM2 gangliosidosis B)	Lysosomal storage disorder
HEXB	Sandhoff disease (GM2 gangliosidosis type O)	Lysosomal storage disorder
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)	Lysosomal storage disorder
HYAL1	Mucopolysaccharidosis IX	Lysosomal storage disorder
IDS	Mucopolysaccharidosis II, Hunter disease	Lysosomal storage disorder
IDUA	Hurler-Scheie syndromes	Lysosomal storage disorder
MAN2B1	α-Mannosidosis	Lysosomal storage disorder
MANBA	β-Mannosidosis	Lysosomal storage disorder
NAGA	Schindler disease; Kanzaki disease	Lysosomal storage disorder
NAGLU	mucopolysaccharidosis type IIIB (Sanfilippo B)	Lysosomal storage disorder
NEU1	Sialidosis	Lysosomal storage disorder
NPC1	Niemann-Pick type C disease	Lysosomal storage disorder
NPC2	Niemann-pick disease, type C2	Lysosomal storage disorder
PSAP	Krabbe disease; Gaucher disease	Lysosomal storage disorder
SGSH	Mucopolysaccharidosis type IIIA (sanfilippo A)	Lysosomal storage disorder
SLC17A5	Sialic Acid Storage Disease; Salla Disease	Lysosomal storage disorder
SMPD1	Niemann-Pick Types A & B	Lysosomal storage disorder
SUMF1	Multiple sulfatase deficiency	Lysosomal storage disorder
ACAD9	Mitochondrial complex I deficiency	Mitochondria-related disorder
CISD2	Wolfram syndrome 2	Mitochondria-related disorder
COQ9	Autosomal-recessive neonatal-onset primary coenzyme Q10 deficiency.	Mitochondria-related disorder
DLAT	Pyruvate dehydrogenase E2 deficiency	Mitochondria-related disorder
DLD	Dihydrolipoamide dehydrogenase deficiency	Mitochondria-related disorder
GNPAT	Rhizomelic chondrodysplasia punctata type II - a peroxisomal disorder	Mitochondria-related disorder
LARS2	Perrault syndrome	Mitochondria-related disorder
NDUFA1	Mitochondrial complex 1 deficiency	Mitochondria-related disorder
PDHA1	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDHB	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDHX	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
PDP1	Pyruvate dehydrogenase complex deficiency	Mitochondria-related disorder
POLG	Progressive external ophthalmoplegia with mitochondrial DNA deletions 1 (PEOA1); Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis (SANDO); alpers-huttenlocher syndrome (AHS); and mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE)	Mitochondria-related disorder
TIMM8A	Mohr-Tranebjaerg syndrome, Jensen syndrome	Mitochondria-related disorder
TRAP1	***TRAP1 related disorder (T1ReD)	Mitochondria-related disorder
PANK2	Pantothenate kinase-associated neurodegeneration/HARP syndrome	Neurodegeneration
ATP13A2	Neuronal ceroid lipofuscinosis 12 (CLN 12) - Juvenile NCL	Neuronal ceroid lipofuscinosis
CLN3	Neuronal ceroid lipofuscinosis 3 (CLN 3) - Classic juvenile NCL ("Spielmeyer-Sjögren"), adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
CLN5	Neuronal ceroid lipofuscinosis 5 (CLN 5) - Late infantile NCL ("Finnish variant"), Juvenile NCL, Adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
CLN6	Neuronal ceroid lipofuscinosis 6 (CLN 6) - Late infantile NCL ("Lake-Cavanagh or Indian variant"), adult NCL (Kufs disease type A)	Neuronal ceroid lipofuscinosis
CLN8	Neuronal ceroid lipofuscinosis 8 (CLN 8) - Late infantile NCL, Northern epilepsy (progressive epilepsy with mental retardation or EPMR)	Neuronal ceroid lipofuscinosis
CTSD	Neuronal ceroid lipofuscinosis 10 (CLN 10) - Congenital NCL, Late-infantile NCL, or Teenage-/adult-onset NCL	Neuronal ceroid lipofuscinosis
CTSF	Neuronal ceroid lipofuscinosis 13 (CLN 13) - Adult NCL (Kufs disease type B)	Neuronal ceroid lipofuscinosis
DNAJC5	Neuronal ceroid lipofuscinosis 4 (CLN 4) - Adult NCL (Parry disease)	Neuronal ceroid lipofuscinosis
GRN	Neuronal ceroid lipofuscinosis 11 (CLN 11) - Adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis
KCTD7	Neuronal ceroid lipofuscinosis 14 (CLN 14) - Late infantile NCL; progressive myoclonic epilepsy 3	Neuronal ceroid lipofuscinosis
MFSD8	Neuronal ceroid lipofuscinosis 7 (CLN7) - Late infantile NCL ("Turkish variant")	Neuronal ceroid lipofuscinosis
PPT1	Neuronal ceroid lipofuscinosis 1 (CLN 1) - Infantile classic NCL ("Haltia-Santavuori" type), late infantile NCL, juvenile NCL, adult NCL (Kufs disease)	Neuronal ceroid lipofuscinosis

Gene	Disease Association	Category
TPP1	Neuronal ceroid lipofuscinosis 2 (CLN 2) - Late infantile classic NCL ("Janský-Bielschowsky" type), juvenile NCL, possibly later-onset type NCL	Neuronal ceroid lipofuscinosis
AASS	Alpha-aminoadipic acid semialdehyde dehydrogenase deficiency	Neurotransmitter disorders
ABAT	GABA-transaminase deficiency	Neurotransmitter disorders
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria)	Neurotransmitter disorders
CHAT	Myasthenic syndrome, congenital, associated with episodic apnea	Neurotransmitter disorders
DBH	Dopamine beta-hydroxylase deficiency	Neurotransmitter disorders
DDC	aromatic l-amino acid decarboxylase deficiency	Neurotransmitter disorders
GCH1	GTP cyclohydrolase deficiency (Hyperphenylalaninemia, BH4-deficient, B)	Neurotransmitter disorders
SLC6A3	Hereditary dopamine transporter deficiency syndrome	Neurotransmitter disorders
TH	Tyrosine hydroxylase deficiency	Neurotransmitter disorders
TPH1	Tryptophan hydroxylase 1 deficiency	Neurotransmitter disorders
TPH2	Tryptophan hydroxylase 2 deficiency	Neurotransmitter disorders
ABCD4	Methylmalonic acidemia and homocystinuria type CblJ	Organic acidemia
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	Organic acidemia
ACAT1	Acetoacetyl-CoA-thiolase deficiency (ketothiolase deficiency)	Organic acidemia
AUH	3-methylglutaconic aciduria type I (MCGA1) disorder	Organic acidemia
BCKDHA	Maple syrup urine disease type 1a	Organic acidemia
BCKDHB	Maple syrup urine disease type 1b	Organic acidemia
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency (2-methylbutaryl-CoA dehydrogenase deficiency, SBCADD)	Organic acidemia
DBT	Maple syrup urine disease type 2	Organic acidemia
ETHE1	Encephalopathy, ethylmalonic	Organic acidemia
GCDH	Glutaric acidemia I	Organic acidemia
IVD	Isovaleric acidemia	Organic acidemia
L2HGDH	L-2-hydroxyglutaric aciduria	Organic acidemia
LMBRD1	Methylmalonic aciduria and homocystinuria, cobalamin F type	Organic acidemia
MCCC1	Elevated 3-hydroxyisovalerylcarnitine / 3-methylcrotonylglycine levels	Organic acidemia
MCCC2	3-methylcrotonyl-CoA carboxylase deficiency	Organic acidemia
MCEE	Methylmalonic coA epimerase deficiency	Organic acidemia
MLYCD	Malonyl-CoA decarboxylase deficiency	Organic acidemia
MMAA	Methylmalonic acidaemia	Organic acidemia
MMAB	Methylmalonic aciduria	Organic acidemia
MMACHC	Methylmalonic aciduria and homocystinuria, cobalamin C type	Organic acidemia
MMADHC	Cobalamin disorders	Organic acidemia
MOCS1	Molybdenum cofactor deficiency A	Organic acidemia
MUT	methylmalonic acidemia	Organic acidemia
OXCT1	Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency	Organic acidemia
PCCA	Propionic acidemia	Organic acidemia
PCCB	Propionic acidemia	Organic acidemia
ABCD1	Adrenoleukodystrophy	Peroxisomal disorder
ABCD3	Zellweger syndrome 2	Peroxisomal disorder
ACOX1	Peroxisomal acyl-CoA oxidase deficiency (pseudoneonatal adrenoleukodystrophy)	Peroxisomal disorder
PEX1	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX10	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX12	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX13	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX14	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX16	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX19	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX2	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX26	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX3	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX5	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX6	Disorders of peroxisomal biogenesis (Zellweger syndrome spectrum)	Peroxisomal disorder
PEX7	peroxisome biogenesis disorder 9B; rhizomelic chondrodysplasia punctata, type 1	Peroxisomal disorder

Gene	Disease Association	Category
PHYH	Refsum disease (hereditary motor sensory neuropathy type IV, heredopathia atactica polyneuritiformis)	Peroxisomal disorder
PTS	6-pyruvoyltetrahydropterin synthase deficiency	Pterin disorder
QDPR	Quinoid dihydropteridine reductase deficiency	Pterin disorder
SPR	Sepiapterin reductase deficiency	Pterin disorder
ADSL	Adenylosuccinate lyase (ADSL) deficiency, adenylosuccinase deficiency, autistic spectrum disorder, succinylpurinemic autism	Purine/pyrimidine metabolism
DPYD	Dihydropyrimidine dehyrogenase deficiency	Purine/pyrimidine metabolism
HPRT1	HPRT-related gout; Lesch-Nyhan syndrome	Purine/pyrimidine metabolism
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency	Purine/pyrimidine metabolism
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	Purine/pyrimidine metabolism
MAGT1	X-linked immunodeficiency with Mg(2+)defect, EBV infection and neoplasia	Transport disorders
SLC16A2	Allan-Herndon-Dudley syndrome	Transport disorders
SLC19A1	Cerebral folate transport deficiency	Transport disorders
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2); can appear as Leigh disease or Wernicke encephalopathy	Transport disorders
SLC25A12	Global cerebral hypomyelination	Transport disorders
SLC25A13	Citrullinemia, type II, neonatal-onset; Citrullinemia, adult-onset type II	Transport disorders
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Transport disorders
SLC25A32	Mitochondrial inner membrane folate transporter deficiency	Transport disorders
SLC2A1	Glucose transporter deficiency syndrome 1 (GLUT1); GLUT1 deficiency syndrome 2; dystonia 9; susceptibility to idiopathic generalized epilepsy 12	Transport disorders
SLC46A1	Hereditary folate malabsorption	Transport disorders
SLC6A4	Serotonin transporter deficiency	Transport disorders
SLC6A8	Cerebral creatine deficiency syndrome 1	Transport disorders
ARG1	Arginase deficiency	Urea cycle disorders
ASL	Spinal muscular atrophy with progressive myoclonic epilepsy	Urea cycle disorders
ASS1	Citrullinemia - Several forms: acute neonatal, milder later onset, pregnancy/postpartum in women, asymptomatic	Urea cycle disorders
CPS1	Carbamoylphosphate synthetase 1 deficiency, hyperammonemia	Urea cycle disorders
NAGS	NAGS deficiency	Urea cycle disorders
OTC	Ornithine transcarbamylase deficiency	Urea cycle disorders