NURTUREGENOMICS

Childhood-Onset Conditions List

Screened by Nurture

Actionable, childhood-onset conditions and their associated genes.

17-alpha-hydroxylase/17,20-lyase deficiency	CYP17A1
3-Methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
3-Methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
3-methylglutaconic aciduria, type I	AUH
Abetalipoproteinemia	MTTP
Achalasia-addisonianism-alacrimia syndrome	AAAS
Acrodermatitis enteropathica	SLC39A4
Acyl-CoA dehydrogenase, medium chain, deficiency of	ACADM
Adenomatous polyposis coli	APC
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	CYP11B1
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	CYP21A2
Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2
Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	CYP11A1
Adrenocortical insufficiency	NR5A1
Adrenocorticotropic hormone deficiency	TBX19
Adrenoleukodystrophy	ABCD1
Afibrinogenemia, congenital	FGA
Afibrinogenemia, congenital	FGB
Afibrinogenemia, congenital	FGG
Agammaglobulinemia 3	CD79A
Agammaglobulinemia 4	BLNK
Agammaglobulinemia 6	CD79B
Agammaglobulinemia 7, autosomal recessive	PIK3R1
Agammaglobulinemia 8A, autosomal dominant	TCF3
Agammaglobulinemia 8B, autosomal recessive	TCF3
Agammaglobulinemia, X-linked 1	BTK
Aldosterone synthase deficiency	CYP11B2
Alpha-methylacetoacetic aciduria	ACAT1
Amelogenesis imperfecta, type IIA3	WDR72
Apparent mineralocorticoid excess	HSD11B2
Argininemia	ARG1
Argininosuccinic aciduria	ASL
Aromatic L-amino acid decarboxylase deficiency	DDC
Arterial calcification, generalized, of infancy, 1	ENPP1
Arterial calcification, generalized, of infancy, 2	ABCC6
Ataxia with isolated vitamin E deficiency	TTPA
Ataxia-pancytopenia syndrome	SAMD9L
B-cell expansion with NFKB and T-cell anergy	CARD11
Bare lymphocyte syndrome, type II, complementation group A	CIITA
Bare lymphocyte syndrome, type II, complementation group B	RFXANK
Bare lymphocyte syndrome, type II, complementation group C	RFX5
Bare lymphocyte syndrome, type II, complementation group D	RFXAP
Bare lymphocyte syndrome, type II, complementation group E	RFX5
Barth syndrome	TAFAZZIN
Bartter syndrome, type 1	SLC12A1

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Bartter syndrome, type 2	KCNJ1
Bartter syndrome, type 3	CLCNKB
Bartter syndrome, type 4a	BSND
Bartter syndrome, type 5, antenatal, transient	MAGED2
Basal cell nevus syndrome 1	PTCH1
Basal cell nevus syndrome 2	SUFU
Bile acid synthesis defect, congenital, 1	HSD3B7
Bile acid synthesis defect, congenital, 2	AKR1D1
Bile acid synthesis defect, congenital, 3	CYP7B1
Biotinidase deficiency	BTD
Bone marrow failure syndrome 3	DNAJC21
Bone marrow failure syndrome 4	MYSM1
Branched-chain keto acid dehydrogenase kinase deficiency	BCKDK
Brown-Vialetto-Van Laere syndrome 1	SLC52A3
Brown-Vialetto-Van Laere syndrome 2	SLC52A2
Carbamoylphosphate synthetase I deficiency	CPS1
Cardiac arrhythmia syndrome, with or without skeletal muscle weakness	TRDN
Carnitine deficiency, systemic primary	SLC22A5
Carnitine-acylcarnitine translocase deficiency	SLC25A20
Cerebral creatine deficiency syndrome 1	SLC6A8
Cerebral creatine deficiency syndrome 2	GAMT
Cerebral creatine deficiency syndrome 3	GATM
Cerebrotendinous xanthomatosis	CYP27A1
Ceroid lipofuscinosis, neuronal, 2	TPP1
Chediak-Higashi syndrome	LYST
Chronic granulomatous disease 1, autosomal recessive	NCF1
Chronic granulomatous disease 2, autosomal recessive	NCF2
Chronic granulomatous disease 3, autosomal recessive	NCF4
Chronic granulomatous disease 4, autosomal recessive	CYBA
Chronic granulomatous disease 5, autosomal recessive	CYBC1
Chronic granulomatous disease, X-linked	CYBB
Citrullinemia	ASS1
Citrullinemia, type II, neonatal-onset	SLC25A13
Coenzyme Q10 deficiency, primary, 1	COQ2
Coenzyme Q10 deficiency, primary, 2	PDSS1
Coenzyme Q10 deficiency, primary, 3	PDSS2
Coenzyme Q10 deficiency, primary, 4	COQ8A
Coenzyme Q10 deficiency, primary, 5	COQ9
Coenzyme Q10 deficiency, primary, 6	COQ6
Coenzyme Q10 deficiency, primary, 7	COQ4
Coenzyme Q10 deficiency, primary, 8	COQ7
Coenzyme Q10 deficiency, primary, 9	COQ5
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	MTHFD1
Congenital disorder of glycosylation, type Ib	MPI

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Congenital disorder of glycosylation, type IIc	SLC35C1
Congenital disorder of glycosylation, type IIk	TMEM165
Congenital disorder of glycosylation, type IIm	SLC35A2
Congenital disorder of glycosylation, type IIn	SLC39A8
Congenital disorder of glycosylation, type It	PGM1
CPT deficiency, hepatic, type IA	CPT1A
CPT II deficiency, infantile	CPT2
Cystic fibrosis	CFTR
Cystinosis, late-onset juvenile or adolescent nephropathic	CTNS
Cystinosis, nephropathic	CTNS
Developmental and epileptic encephalopathy 7	KCNQ2
Diabetes insipidus, nephrogenic, 1	AVPR2
Diabetes insipidus, nephrogenic, 2	AQP2
Diabetes mellitus, permanent neonatal 3, with or without neurologic features	ABCC8
Diabetes, permanent neonatal 2, with or without neurologic features	KCNJ11
Diamond Blackfan anemia 15 with mandibulofacial dysostosis	RPS28
Diamond-Blackfan anemia 1	RPS19
Diamond-Blackfan anemia 10	RPS26
Diamond-Blackfan anemia 11	RPL26
Diamond-Blackfan anemia 12	RPL15
Diamond-Blackfan anemia 13	RPS29
Diamond-Blackfan anemia 14 with mandibulofacial dysostosis	TSR2
Diamond-Blackfan anemia 16	RPL27
Diamond-Blackfan anemia 17	RPS27
Diamond-Blackfan anemia 18	RPL18
Diamond-Blackfan anemia 19	RPL35
Diamond-Blackfan anemia 20	RPS15A
Diamond-blackfan anemia 3	RPS24
Diamond-Blackfan anemia 4	RPS17
Diamond-Blackfan anemia 5	RPL35A
Diamond-Blackfan anemia 6	RPL5
Diamond-Blackfan anemia 7	RPL11
Diamond-Blackfan anemia 8	RPS7
Diamond-Blackfan anemia 9	RPS10
Diarrhea 1, secretory chloride, congenital	SLC26A3
Diarrhea 7, protein-losing enteropathy type	DGAT1
Diarrhea 8, secretory sodium, congenital	SLC9A3
Dihydrolipoamide dehydrogenase deficiency	DLD
Distal renal tubular acidosis 1	SLC4A1
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	ATP6V1B1
Distal renal tubular acidosis 3, with or without sensorineural hearing loss	ATP6V0A4
Distal renal tubular acidosis 4 with hemolytic anemia	SLC4A1
Duchenne muscular dystrophy	DMD
Dyskeratosis congenita, autosomal dominant 1	TERC
Dyskeratosis congenita, autosomal dominant 3	TINF2

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Dyskeratosis congenita, autosomal recessive 5	RTEL1
Dyskeratosis congenita, X-linked	DKC1
Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	SPR
Ehlers-Danlos syndrome, vascular type	COL3A1
Epilepsy, early-onset, vitamin B6-dependent	PLPBP
Fabry disease	GLA
Factor XIIIA deficiency	F13A1
Factor XIIIB deficiency	F13B
Familial cold autoinflammatory syndrome 4	NLRC4
Fanconi anemia, complementation group A	FANCA
Fanconi anemia, complementation group B	FANCB
Fanconi anemia, complementation group C	FANCC
Fanconi anemia, complementation group D2	FANCD2
Fanconi anemia, complementation group E	FANCE
Fanconi anemia, complementation group F	FANCF
Fanconi anemia, complementation group G	FANCG
Fanconi anemia, complementation group I	FANCI
Fanconi anemia, complementation group J	BRIP1
Fanconi anemia, complementation group L	FANCL
Fanconi anemia, complementation group N	PALB2
Fanconi anemia, complementation group O	RAD51C
Fanconi anemia, complementation group P	SLX4
Fanconi anemia, complementation group Q	ERCC4
Fanconi anemia, complementation group T	UBE2T
Fanconi anemia, complementation group V	MAD2L2
Fanconi anemia, complementation group W	RFWD3
Folate malabsorption, hereditary	SLC46A1
Fructose intolerance, hereditary	ALDOB
Fructose-1,6-bisphosphatase deficiency	FBP1
Galactokinase deficiency with cataracts	GALK1
Galactose epimerase deficiency	GALE
Galactosemia	GALT
Galactosemia IV	GALM
Gastrointestinal defects and immunodeficiency syndrome	TTC7A
GATA1 associated X-Linked Cytopenia	GATA1
Gaucher disease, type I	GBA
Gitelman syndrome	SLC12A3
Glucocorticoid deficiency 2	MRAP
Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	NNT
Glucocorticoid deficiency, due to ACTH unresponsiveness	MC2R
Glucose/galactose malabsorption	SLC5A1
GLUT1 deficiency syndrome 1, infantile onset, severe	SLC2A1
Glutaric acidemia IIC	ETFDH
Glutaricaciduria, type I	GCDH
Glycogen storage disease Ia	G6PC

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Glycogen storage disease Ib	SLC37A4
Glycogen storage disease II	GAA
Glycogen storage disease III	AGL
Glycogen storage disease IXc	PHKG2
Glycogen storage disease VI	PYGL
Glycogen storage disease, type IXa	PHKA2
Griscelli syndrome, type 2	RAB27A
Growth hormone deficiency, isolated, type IA	GH1
Growth hormone deficiency, isolated, type IB	GH1
Growth hormone deficiency, isolated, type II	GH1
Growth hormone deficiency, isolated, type IV	GHRHR
Growth hormone insensitivity with immune dysregulation 1, autosomal recessive	STAT5B
Growth retardation with deafness and mental retardation due to IGF1 deficiency	IGF1
Gyrate atrophy of choroid and retina with or without ornithinemia	OAT
Hemolytic anemia, G6PD deficient (favism)	G6PD
Hemophagocytic lymphohistiocytosis, familial, 2	PRF1
Hemophagocytic lymphohistiocytosis, familial, 3	UNC13D
Hemophagocytic lymphohistiocytosis, familial, 4	STX11
Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	STXBP2
Hemophilia B	F9
Hemosiderosis, systemic, due to aceruloplasminemia	CP
Hepatic venoocclusive disease with immunodeficiency	SP110
Hermansky-Pudlak syndrome 2	AP3B1
HMG-CoA lyase deficiency	HMGCL
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria due to MTHFR deficiency	MTHFR
Homocystinuria, B6-responsive and nonresponsive types	CBS
Homocystinuria-megaloblastic anemia, cbl E type	MTRR
Homocystinuria-megaloblastic anemia, cblG complementation type	MTR
Homocystinuria/Methylmalonic acidemia	MMADHC
Hutchinson-Gilford progeria	LMNA
Hyper-IgE recurrent infection syndrome, autosomal recessive	DOCK8
Hyperaldosteronism, familial, type III	KCNJ5
Hyperammonemia due to carbonic anhydrase VA deficiency	CA5A
Hypercholesterolemia, familial, 1	LDLR
Hypercholesterolemia, familial, 2	APOB
Hypercholesterolemia, familial, 3	PCSK9
Hypercholesterolemia, familial, 4	LDLRAP1
Hyperinsulinemic hypoglycemia, familial, 1	ABCC8
Hyperinsulinemic hypoglycemia, familial, 2	KCNJ11
Hyperinsulinemic hypoglycemia, familial, 3	GCK
Hyperinsulinemic hypoglycemia, familial, 7	SLC16A1
Hyperinsulinism-hyperammonemia syndrome	GLUD1
Hypermanganesemia with dystonia 1	SLC30A10
Hypermanganesemia with dystonia 2	SLC39A14

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Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15
Hyperoxaluria, primary, type II	GRHPR
Hyperoxaluria, primary, type III	HOGA1
Hyperphenylalaninemia, BH4-deficient, A	PTS
Hyperphenylalaninemia, BH4-deficient, C	QDPR
Hypobetalipoproteinemia	APOB
Hypomagnesemia 1, intestinal	TRPM6
Hypophosphatasia	ALPL
Hypophosphatemic rickets with hypercalciuria	SLC34A3
Hypophosphatemic rickets, X-linked dominant	PHEX
Immunodeficiency 11B with atopic dermatitis	CARD11
Immunodeficiency 15B	IKBKB
Immunodeficiency 23	PGM3
Immunodeficiency 26, with or without neurologic abnormalities	PRKDC
Immunodeficiency 40	DOCK2
Immunodeficiency 41 with lymphoproliferation and autoimmunity	IL2RA
Immunodeficiency 48	ZAP70
Immunodeficiency 67	IRAK4
Immunodeficiency 68	MYD88
Immunodeficiency 8	CORO1A
Immunodeficiency due to defect in MAPBP-interacting protein	LAMTOR2
Immunodeficiency due to purine nucleoside phosphorylase deficiency	PNP
Immunodeficiency with hyper IgM, type 5	UNG
Immunodeficiency with hyper-IgM, type 2	AICDA
Immunodeficiency with hyper-IgM, type 3	CD40
Immunodeficiency, common variable, 10	NFKB2
Immunodeficiency, common variable, 11	IL21
Immunodeficiency, common variable, 13	IKZF1
Immunodeficiency, common variable, 2	TNFRSF13B
Immunodeficiency, common variable, 8, with autoimmunity	LRBA
Immunodeficiency, X-linked, with hyper-IgM	CD40LG
Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	MAGT1
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3
Isobutyryl-CoA dehydrogenase deficiency	ACAD8
Isovaleric acidemia	IVD
Krabbe disease	GALC
Lacticacidemia due to PDX1 deficiency	PDHX
Laron dwarfism	GHR
LCHAD deficiency	HADHA
Leber congenital amaurosis 2	RPE65
Leukocyte adhesion deficiency	ITGB2
Leukocyte adhesion deficiency, type III	FERMT3
Li-Fraumeni syndrome	TP53
LIG4 syndrome	LIG4
Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	FLAD1

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Lipodystrophy, congenital generalized, type 1	AGPAT2
Lipodystrophy, congenital generalized, type 2	BSCL2
Lipoid adrenal hyperplasia	STAR
Lipoprotein lipase deficiency	LPL
Liver failure, transient infantile	TRMU
Loeys-Dietz syndrome 1	TGFBR1
Loeys-Dietz syndrome 2	TGFBR2
Loeys-Dietz syndrome 3	SMAD3
Lymphoproliferative syndrome 1	ITK
Lymphoproliferative syndrome 2	CD27
Lymphoproliferative syndrome 3	CD70
Lymphoproliferative syndrome, X-linked, 1	SH2D1A
Lymphoproliferative syndrome, X-linked, 2	XIAP
Lysinuric protein intolerance	SLC7A7
Malonyl-CoA decarboxylase deficiency	MLYCD
Mannosidosis, alpha-, types I and II	MAN2B1
Maple syrup urine disease, type Ia	BCKDHA
Maple syrup urine disease, type Ib	BCKDHB
Medullary thyroid carcinoma	RET
Megaloblastic anemia due to dihydrofolate reductase deficiency	DHFR
Megaloblastic anemia, folate-responsive	SLC19A1
Menkes disease	ATP7A
Metachromatic leukodystrophy	ARSA
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC
Methylmalonic aciduria and homocystinuria, cblC type, digenic	PRDX1
Methylmalonic aciduria and homocystinuria, cblF type	LMBRD1
Methylmalonic aciduria and homocystinuria, cblJ type	ABCD4
Methylmalonic aciduria, mut(0) type	MMUT
Methylmalonic aciduria, vitamin B12-responsive, cblA type	MMAA
Methylmalonic aciduria, vitamin B12-responsive, cblB type	MMAB
Methylmalonyl-CoA epimerase deficiency	MCEE
MIRAGE syndrome	SAMD9
Mitochondrial complex I deficiency, nuclear type 20	ACAD9
Mitochondrial DNA depletion syndrome 2 (myopathic type)	TK2
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1
Mitochondrial trifunctional protein deficiency 2	HADHB
MODY, type I	HNF4A
MODY, type III	HNF1A
Molybdenum cofactor deficiency A	MOCS1
Mucopolysaccharidosis Ih	IDUA
Mucopolysaccharidosis Ih/s	IDUA
Mucopolysaccharidosis II	IDS
Mucopolysaccharidosis Is	IDUA
Mucopolysaccharidosis IVA	GALNS
Mucopolysaccharidosis type IIIB (Sanfilippo B)	NAGLU

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Mucopolysaccharidosis type IIIC (Sanfilippo C)	HGSNAT
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	ARSB
Mucopolysaccharidosis VII	GUSB
Multiple endocrine neoplasia IIA	RET
Multiple endocrine neoplasia IIB	RET
Myasthenia, congenital, 12, with tubular aggregates	GFPT1
Myasthenic syndrome, congenital, 10	DOK7
Myasthenic syndrome, congenital, 13, with tubular aggregates	DPAGT1
Myasthenic syndrome, congenital, 14, with tubular aggregates	ALG2
Myasthenic syndrome, congenital, 15, without tubular aggregates	ALG14
Myasthenic syndrome, congenital, 1B, fast-channel	CHRNA1
Myasthenic syndrome, congenital, 6, presynaptic	CHAT
N-acetylglutamate synthase deficiency	NAGS
Nephrotic syndrome, type 14	SGPL1
Nephrotic syndrome, type 9	COQ8B
Neurodegeneration due to cerebral folate transport deficiency	FOLR1
NEUROG3 associated neonatal diabetes mellitus	NEUROG3
Neutropenia, severe congenital 1, autosomal dominant	ELANE
Neutropenia, severe congenital 2, autosomal dominant	GFI1
Neutropenia, severe congenital 3, autosomal recessive	HAX1
Neutropenia, severe congenital 4, autosomal recessive	G6PC3
Neutropenia, severe congenital, 5, autosomal recessive	VPS45
Neutropenia, severe congenital, 6, autosomal recessive	JAGN1
Neutropenia, severe congenital, 7, autosomal recessive	CSF3R
Neutropenia, severe congenital, 8, autosomal dominant	SRP54
Niemann-Pick disease, type A	SMPD1
Niemann-Pick disease, type B	SMPD1
Niemann-Pick disease, type C1	NPC1
Niemann-pick disease, type C2	NPC2
Nijmegen breakage syndrome	NBN
Obesity, adrenal insufficiency, and red hair due to POMC deficiency	POMC
Obesity, morbid, due to leptin deficiency	LEP
Omenn syndrome	DCLRE1C
Ornithine transcarbamylase deficiency	OTC
Orotic aciduria	UMPS
Osteopetrosis, autosomal recessive 1	TCIRG1
Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	CA2
Osteopetrosis, autosomal recessive 7	TNFRSF11A
Osteopetrosis, autosomal recessive 8	SNX10
Pancreatic agenesis 2	PTF1A
Pancreatic agenesis and congenital heart defects	GATA6
Panhypopituitarism, X-linked	SOX3
Periodic fever, immunodeficiency, and thrombocytopenia syndrome	WDR1
Phenylketonuria	PAH
Phosphorylase kinase deficiency of liver and muscle, autosomal recessive	PHKB

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Pituitary hormone deficiency, combined or isolated, 1	POU1F1
Pituitary hormone deficiency, combined or isolated, 7	RNPC3
Pituitary hormone deficiency, combined, 2	PROP1
Pituitary hormone deficiency, combined, 3	LHX3
Pituitary hormone deficiency, combined, 4	LHX4
Pituitary hormone deficiency, combined, 5	HESX1
Pleuropulmonary blastoma	DICER1
Polycystic kidney disease 1	PKD1
Polycystic kidney disease 4, with or without hepatic disease	PKHD1
Polycystic kidney disease with hyperinsulinemic hypoglycemia	PMM2
Porphyria, acute intermittent	HMBS
Primary aldosteronism, seizures, and neurologic abnormalities	CACNA1D
Propionicacidemia	PCCA
Propionicacidemia	PCCB
Protoporphyria, erythropoietic, 1	FECH
Protoporphyria, erythropoietic, X-linked	ALAS2
Pyridoxamine 5'-phosphate oxidase deficiency	PNPO
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1
Pyruvate dehydrogenase E1-beta deficiency	PDHB
Pyruvate dehydrogenase E2 deficiency	DLAT
Pyruvate dehydrogenase phosphatase deficiency	PDP1
Pyruvate kinase deficiency	PKLR
Radioulnar synostosis with amegakaryocytic thrombocytopenia 1	HOXA11
Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	MECOM
Renal cysts and diabetes syndrome	HNF1B
Renal tubular acidosis, proximal, with ocular abnormalities	SLC4A4
Reticular dysgenesis	AK2
Retinitis pigmentosa 20	RPE65
Retinoblastoma	RB1
Rett syndrome	MECP2
Rickets due to defect in vitamin D 25-hydroxylation deficiency	CYP2R1
Rickets, vitamin D-resistant, type IIA	VDR
Segawa syndrome, recessive	TH
Severe combined immunodeficiency due to ADA deficiency	ADA
Severe combined immunodeficiency, Athabascan type	DCLRE1C
Severe combined immunodeficiency, B cell-negative	RAG2
Shwachman-Diamond syndrome 1	SBDS
Shwachman-Diamond syndrome 2	EFL1
Sickle cell anemia	HBB
Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	TRNT1
Smith-Lemli-Opitz syndrome	DHCR7
Sodium-dependent multivitamin transporter deficiency	SLC5A6
Spinal muscular atrophy-1	SMN1
Succinic semialdehyde dehydrogenase deficiency	ALDH5A1

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Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1
Surfactant metabolism dysfunction, pulmonary, 2	SFTPC
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1
T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	STK4
Thalassemia, beta	HBB
Thalassemia-beta, dominant inclusion-body	HBB
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	SLC19A3
Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	SLC25A19
Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	TPK1
Thiamine-responsive megaloblastic anemia syndrome	SLC19A2
Thrombocytopenia, congenital amegakaryocytic	MPL
Thrombotic thrombocytopenic purpura, hereditary	ADAMTS13
Transcobalamin II deficiency	TCN2
Tuberous sclerosis-1	TSC1
Tuberous sclerosis-2	TSC2
Tyrosinemia, type I	FAH
Tyrosinemia, type II	TAT
Tyrosinemia, type III	HPD
Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome	ADA2
Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome	RYR2
Ventricular tachycardia, catecholaminergic polymorphic, 1	RYR2
Ventricular tachycardia, catecholaminergic polymorphic, 2	CASQ2
Vitamin D-dependent rickets, type I	CYP27B1
Vitamin K-dependent clotting factors, combined deficiency of, 1	GGCX
Vitamin K-dependent clotting factors, combined deficiency of, 2	VKORC1
VLCAD deficiency	ACADVL
Wilms tumor, type 1	WT1
Wilson disease	ATP7B
Wiskott-Aldrich syndrome	WAS
Wiskott-Aldrich syndrome 2	WIPF1
Wolfram syndrome 1	WFS1
Wolman disease	LIPA