TarCET®
IVD kits

MULTIPLE ASSAYS IN ONE SINGLE RUN

Disorders covered by TarCET IVD kits

MEDICOVER GENETICS

Carrier Core Kit

Disorder	Genes
Alpha Thalassemia	HBA1, HBA2
Beta Thalassemia	HBB
Bloom Syndrome	BLM
Canavan Disease	ASPA
Cystic Fibrosis	CFTR
Duchenne Muscular Dystrophy, X-linked	DMD
Familial Dysautonomia	ELP1
Fanconi Anemia, Type C	FANCC
Galactosemia	GALT
Gaucher Disease	GBA
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM
Mucolipidosis, Type IV	MCOLN1
Niemann-Pick Disease, Types A/B	SMPD1
Phenylalanine Hydroxylase Deficiency	PAH
Sickle-Cell Disease	HBB
Smith-Lemli-Opitz Syndrome	DHCR7
Tay-Sachs Disease	HEXA
Non-Syndromic Hearing Loss GJB2-Related and GJB6-Related	GJB2, GJB6

Carrier Comprehensive Kit

Disorder	Genes
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	MCCC1
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	MCCC2
3-Methylglutaconic Aciduria, Type 3 [Costeff Syndrome]	OPA3
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	PTS
Abetalipoproteinemia	MTTP
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2
Achromatopsia (CNGB3-related)	CNGB3
Acute Infantile Liver Failure (TRMU-related)	TRMU
Acyl-CoA Oxidase I Deficiency	ACOX1
Adrenoleukodystrophy, X-Linked	ABCD1
Aicardi-Goutières Syndrome	SAMHD1
Alpha Thalassemia	HBA1, HBA2
Alport Syndrome (COL4A3-related)	COL4A3
Alport Syndrome, X-Linked	COL4A5
Alstrom Syndrome	ALMS1
Andermann Syndrome	SLC12A6
Argininosuccinate Lyase Deficiency	ASL
Aromatase Deficiency	CYP19A1
Arthrogryposis, impaired intellectual development, and seizures	SLC35A3
Asparagine Synthetase Deficiency	ASNS
Aspartylglycosaminuria	AGA
Ataxia with Vitamin E Deficiency	TTPA
Ataxia-Telangiectasia	ATM
Autoimmune Polyglandular Syndrome, Type 1	AIRE
Autosomal Recessive Polycystic Kidney Disease	PKHD1
Autosomal Recessive Spastic Ataxia of Charlevoix- Saguenay	SACS
Bardet-Biedl Syndrome (BBS1-related)	BBS1
Bardet Biedl syndrome (BBS12-related)	BBS12
Bare Lymphocyte Syndrome (CIITA-related)	CIITA
Bartter Syndrome (BSND-related)	BSND
Batten Disease (CLN3-related)	CLN3
Beta Thalassemia	HBB
Biotinidase Deficiency	BTD
Bloom Syndrome	BLM
Canavan Disease	ASPA

Carnitine Palmitoyltransferase IA Deficiency	CPT1A
Carnitine Palmitoyltransferase II Deficiency	CPT2
Carpenter Syndrome	RAB23
Cartilage-Hair Hypoplasia	RMRP
Cerebrotendinous Xanthomatosis	CYP27A1
Choreacanthocytosis	VPS13A
Choroideremia, X-Linked	CHM
Chronic Granulomatous Disease, X-Linked	CYBB
Citrullinemia	SLC25A13
Citrullinemia, Type 1	ASS1
Combined Malonic and Methylmalonic Aciduria	ACSF3
Combined Oxidative Phosphorylation Deficiency 1	GFM1
Combined Oxidative Phosphorylation Deficiency 3	TSFM
Combined Pituitary Hormone Deficiency 2	PROP1
Congenital Disorder of Glycosylation, Type 1A (PMM2- related)	PMM2
Congenital Disorder of Glycosylation, Type 1B	MPI
Congenital Disorder of Glycosylation Type 1C	ALG6
Congenital Finnish Nephrosis	NPHS1
Congenital Insensitivity to Pain with Anhidrosis	NTRK1
Congenital Myasthenic Syndrome (CHRNE-related)	CHRNE
Congenital Myasthenic Syndrome (RAPSN-related)	RAPSN
Congenital Neutropenia (HAX1-related)	HAX1
Congenital Neutropenia (VPS45-related)	VPS45
Corneal Dystrophy and Perceptive Deafness	SLC4A11
Corticosterone Methyloxidase Deficiency	CYP11B2
CRB1-related Retinal Dystrophies	CRB1
Creatine Transporter Defect [Cerebral Creatine Deficiency Syndrome 1] X-Linked	SLC6A8
Crigler Najjar syndrome, Type I/II	UGT1A1
Cystic Fibrosis	CFTR
Cystinosis	CTNS
D-Bifunctional Protein Deficiency	HSD17B4
Deafness, Autosomal Recessive 77	LOXHD1
Duchenne Muscular Dystrophy, X-linked	DMD
Dystrophic Epidermolysis Bullosa (COL7A1-related)	COL7A1
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	EMD
Enhanced S-Cone Syndrome	NR2E3

Ethylmalonic Encephalopathy	ETHE1
Fabry Disease, X-Linked	GLA
Factor IX Deficiency, X-Linked	F9
Factor V Leiden Thrombophilia	F5
Factor XI Deficiency	F11
Familial Dysautonomia	ELP1
Familial Hypercholesterolemia (LDLR-related)	LDLR
Familial Mediterranean Fever	MEFV
Nephrogenic Diabetes Insipidus (AQP2- related)	AQP2
Fanconi Anemia, Type G	FANCG
Fanconi Anemia, Type C	FANCC
Galactokinase Deficiency [Galactosemia, Type II]	GALK1
Galactosemia	GALT
Gaucher Disease	GBA
Glutaric Acidemia, Type 1	GCDH
Glutaric Acidemia, Type 2A	ETFA
Glycine Encephalopathy (GLDC-related)	GLDC
Glycine Encephalopathy (AMT-related)	AMT
Glycogen Storage Disease, Type 1A	G6PC
Glycogen Storage Disease, Type 1	SLC37A4
Glycogen Storage Disease, Type 2 [Pompe Disease]	GAA
Glycogen Storage Disease, Type 3	AGL
Glycogen Storage Disease, Type 4	GBE1
Glycogen Storage Disease, Type 5 [McArdle Disease]	PYGM
Glycogen Storage Disease, Type 7	PFKM
GRACILE Syndrome	BCS1L
Hemochromatosis, Type 2A	HJV
Hemochromatosis, Type 3 (TFR2-related)	TFR2
Hereditary Fructose Intolerance	ALDOB
Hermansky-Pudlak Syndrome (HPS1-related)	HPS1
Hermansky-Pudlak Syndrome (HPS3-related)	HPS3
Holocarboxylase Synthetase Deficiency	HLCS
Homocystinuria (CBS-related)	CBS
Homocystinuria, Type cblE	MTRR
Hydrolethalus Syndrome	HYLS1
Hypohidrotic Ectodermal Dysplasia, X-Linked	EDA
Hypophosphatasia (ALPL-related)	ALPL
Inclusion Body Myopathy Type 2	GNE

Isovaleric Acidemia	IVD
Joubert Syndrome, Type 2	TMEM216
Meckel syndrome 2	TMEM216
Junctional Epidermolysis Bullosa, Herlitz type	LAMC2
Juvenile Retinoschisis, X-Linked	RS1
Krabbe Disease	GALC
Lamellar Ichthyosis, Type 1	TGM1
Leber Congenital Amaurosis (LCA5-related)	LCA5
Leber Congenital Amaurosis, Type CEP290	CEP290
Leigh Syndrome, French-Canadian Type	LRPPRC
Leukoencephalopathy with Vanishing White Matter	EIF2B5
Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	LHCGR
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB
Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]	DLD
Lipoid Adrenal Hyperplasia	STAR
Lipoprotein Lipase Deficiency	LPL
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA
Lysinuric Protein Intolerance	SLC7A7
Maple Syrup Urine Disease, Type 1B	BCKDHB
Meckel-Gruber Syndrome, Type 1	MKS1
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1
Metachromatic Leukodystrophy (ARSA-related)	ARSA
Metachromatic Leukodystrophy (PSAP-related)	PSAP
Methylmalonic Aciduria (MMAA-related)	MMAA
Methylmalonic Aciduria (MMAB-related)	MMAB
Methylmalonic Acidemia and Homocystinuria, Type cblC	MMACHC
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC
Methylmalonic Aciduria, Type mut(0)	MMUT
Microphthalmia/Anophthalmia (VSX2-related)	VSX2
Mitochondrial Complex 1 Deficiency (ACAD9-related)	ACAD9
Mitochondrial Complex 1 Deficiency (NDUFAF5- related)	NDUFAF5
Mitochondrial Complex 1 Deficiency (NDUFS6-related)	NDUFS6

Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	PUS1
Mucolipidosis II/III	GNPTAB
Mucolipidosis III Gamma	GNPTG
Mucolipidosis, Type IV	MCOLN1
Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	IDS
Mucopolysaccharidosis, Type IIIB [Sanfilippo B]	NAGLU
Mucopolysaccharidosis, Type IIIC [Sanfilippo C]	HGSNAT
Mucopolysaccharidosis IIID [Sanfilippo D]	GNS
Mucopolysaccharidosis, Type IX	HYAL1
Multiple Sulfatase Deficiency	SUMF1
Myoneurogastrointestinal Encephalopathy (MNGIE)	TYMP
Myotubular Myopathy, X-Linked	MTM1
N-acetylglutamate Synthase Deficiency	NAGS
Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]	MPV17
Neurological Ceroid Lipofuscinosis, TPP1-related	TPP1
Neuronal Ceroid Lipofuscinosis (MFSD8-related)	MFSD8
Neuronal Ceroid Lipofuscinosis (CLN5-related)	CLN5
Neuronal Ceroid Lipofuscinosis (CLN6-related)	CLN6
Neuronal Ceroid Lipofuscinosis (CLN8-related)	CLN8
Neuronal Ceroid Lipofuscinosis (PPT1-related)	PPT1
Niemann-Pick Disease, Types A/B	SMPD1
Niemann-Pick Disease, Type C1/D	NPC1
Niemann-Pick Disease, Type C2	NPC2
Nijmegen Breakage Syndrome	NBN
Non-Syndromic Hearing Loss (GJB2-related, GJB6-related)	GJB2, GJB6
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz- Passarge Syndrome	WNT10A
Omenn Syndrome, RAG2-related	RAG2
Ornithine Aminotransferase Deficiency	OAT
Ornithine Transcarbamylase Deficiency	OTC
Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]	SLC25A15
Pendred Syndrome	SLC26A4
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	PEX1
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	PEX2
Phenylalanine Hydroxylase Deficiency	PAH
Pituitary Hormone Deficiency, Combined 3	LHX3

Pontocerebellar Hypoplasia, RARS2-related	RARS2
Pontocerebellar Hypoplasia, Type 1A	VRK1
Pontocerebellar Hypoplasia, Type 2D	SEPSECS
Pontocerebellar Hypoplasia, Type 2E	VPS53
Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5
Primary Ciliary Dyskinesia, DNAI1-related	DNAI1
Primary Ciliary Dyskinesia, DNAI2-related	DNAI2
Primary Hyperoxaluria, Type 1	AGXT
Primary Hyperoxaluria, Type 2	GRHPR
Primary Hyperoxaluria, Type 3	HOGA1
Pycnodysostosis	CTSK
Pyruvate Dehydrogenase Deficiency (PDHB-related)	PDHB
Pyruvate Dehydrogenase Deficiency, X-Linked	PDHA1
Renal Tubular Acidosis and Deafness (ATP6V1B11- related)	ATP6V1B1
Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]	RLBP1
Retinitis Pigmentosa 59 (DHDDS-related)	DHDDS
Retinitis Pigmentosa 25 (EYS-related)	EYS
Retinitis Pigmentosa 26	CERKL
Retinitis Pigmentosa 28	FAM161A
Retinitis Pigmentosa, X-linked	RPGR
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS
Roberts Syndrome	ESCO2
Salla Disease	SLC17A5
Sandhoff Disease	HEXB
Schimke Immunoosseous Dysplasia	SMARCAL1
Segawa Syndrome, (TH-related)	TH
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C
Severe Combined Immunodeficiency, X-Linked	IL2RG
Sickle-Cell Disease	HBB
Sjögren-Larsson Syndrome	ALDH3A2
Smith-Lemli-Opitz Syndrome	DHCR7
Steroid-Resistant Nephrotic Syndrome	NPHS2
Stuve-Wiedemann Syndrome	LIFR
Tay-Sachs Disease	HEXA
Tyrosinemia, Type 1	FAH
Usher Syndrome, Type 1C	USH1C
Usher Syndrome, Type 1F	PCDH15

Usher Syndrome, Type 2A	USH2A
Usher Syndrome, Type 3	CLRN1
Wilson Disease	ATP7B
Wolman Disease	LIPA
Zellweger Spectrum Disorders, (PEX6-related)	PEX6
Zellweger Spectrum Disorders (PEX10-related)	PEX10

Hereditary Cancer Kit

Disorder	Genes
Ataxia-telangiectasia syndrome	ATM
BAP1 mutation associated disease	BAP1
Constitutional mismatch repair syndrome	EPCAM, MSH2, MSH6, MLH1, PMS2
DICER 1 syndrome	DICER1
Familial Adenomatous Polyposis / Attenuated Familial Adenomatous Polyposis	APC
Fanconi anemia syndrome	FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, SLX4, ERCC4, BRCA1, BRCA2, BRIP1, PALB2, RAD51C
Hereditary breast & ovarian cancer syndrome	BRCA1, BRCA2
Hereditary diffuse gastric syndrome	CDH1
Hereditary mixed polyposis syndrome	GREM1
Hereditary melanoma-pancreatic cancer syndrome	CDKN2A, CDK4
Hereditary Paraganglioma – Pheochromocytoma syndrome	SDHD, SDHAF2, SDHC, SDHB
Juvenile polyposis syndrome	SMAD4, BMPR1A
Li-Fraumeni syndrome	TP53
Li-Fraumeni syndrome 2	CHEK2
Lynch syndrome	EPCAM, MLH1, MSH2, MSH6, PMS2
Multiple endocrine neoplasia type 1	MEN1
Multiple endocrine neoplasia type 2	RET
MUTYH-associated polyposis syndrome	MUTYH
Peutz-Jeghers syndrome	STK11
Polymerase proofreading associated syndrome	POLD1, POLE
PTEN hamartoma syndrome	PTEN
Retinoblastoma	RB1
Von-Hippel Lindau syndrome	VHL
Xeroderma Pigmentosum syndrome	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC

Cancers associated with this panel

Breast, Ovarian, Endometrial or associated cancers, Colorectal, Blood, Gastric (stomach), Prostate, Pancreatic, Renal (kidney), Skin, Paraganglioma or Pheochromocytoma, Familial melanoma skin, Parathyroid, Medullary thyroid

Infertility Kit

Female Infertility

Disorder	Gene
Primary Ovarian Insufficiency	BMP15
	CYP19A1
	FOXL2
	GALT
	GNAS
	GNRHR
	KISS1R
	NOBOX
	STAG3
	ZP1
	EIF2B2
	EIF2B3
	FIGLA
	POF1B
	POLG
	PSMC3IP
	WT1
	FSHB
	NR5A1
	LHB
	LHCGR
	FSHR
	CYP17A1
	GDF9
	AIRE

Polycystic ovary syndrome	LHCGR
	FSHR
	CYP17A1
	GDF9
	GNRH1
	INS
	INSR
	IRS1
	IRS2
	KISS1

Polycystic ovary syndrome	TACR3
	CAPN10
	CYP11A1
	DENND1A
	THADA

Hypogonadotropic Hypogonadism	GNRHR
	KISS1R
	FSHB
	LHB
	GNRH1
	KISS1
	TACR3
	ANOS1
	CHD7
	DUSP6
	FEZF1
	FGF17
	FGF8
	FGFR1
	FLRT3
	HESX1
	HS6ST1
	IL17RD
	NSMF
	PROK2
	PROKR2
	SEMA3A
	SPRY4
	TAC3
	WDR11

Ovarian Hyperstimulation syndrome	FSHR

Male Infertility

Spermatogenic failure (azoospermia/oligospermia etc)	ANOS1 (KAL1)
	AR

Spermatogenic failure (azoospermia/oligospermia etc)	AURKC
	CATSPER1
	CFTR
	CHD7
	DAZL
	DDX25
	FGF8
	FSHB
	FSHR
	NR5A1
	PRM1
	USP26
	USP9Y

Disorders of sexual development	AR
	LHCGR
	NR5A1
	SRY
	SRD5A1

Congenital bilateral absence of the vas deferens (CBAVD)	CFTR

Hypogonadotropic hypogonadism (HH)	ANOS1 (KAL1)
	CHD7
	DUSP6
	FEZF1
	FGF17
	FGF8
	FLRT3
	FSHB
	GNRH1
	GNRHR
	HESX1
	HS6ST1
	IL17RD
	KISS1
	KISS1R

Hypogonadotropic hypogonadism (HH)	LHB
	NR5A1
	NSMF
	PROK2
	PROKR2
	SEMA3A
	SPRY4
	TAC3
	TACR3
	WDR11
	FGFR1
	LHCGR

Neonatal Kit

Disorder	Genes
ENDOCRINE	
Congenital Adrenal Hyperplasia	CYP11B1, CYP17A1, HSD3B2, POR, STAR
Congenital Hypothyroidism	PAX8, THRA, SLC5A5, TG, TPO, TSHB, TSHR
Pendred Syndrome	SLC26A4
HEAMOGLOBIN	
Beta-Thalassemia	HBB
S, Beta-Thalassemia (Sickle Cell Beta-Thalassemia)	HBB
S,C Disease (Sickle Cell Disease)	HBB
S,S Disease (Sickle Cell Disease, Sickle Cell Anemia)	HBB
HEARING LOSS	
NON-SYNDROMIC HEARING LOSS	CDH23, MYO15A, OTOF, TMIE, TMPRSS3, TPRN, TRIOBP, GJB2, GJB6, TECTA
SYNDROMIC HEARING LOSS	
Jervell and Lange-Nielsen Syndrome	KCNE1, KCNQ1
Pendred Syndrome	SLC26A4
Shah-Waardenburg Syndrome	SOX10
Usher Syndrome Type 1C	USH1C
Usher Syndrome 1G	USH1G
Usher Syndrome Type 2A	USH2A
Usher Syndrome IID	WHRN
Waardenburg Syndrome	PAX3
METABOLIC	
2,4 Dienoyl-CoA Reductase Deficiency	NADK2
2-Methyl-3-Hydroxybutyric Aciduria	HSD17B10
2-Methylbutyrylglycinuria	ACADSB
3-Methylcrotonyl-CoA Carboxylase Deficiency	MCCC1, MCCC2
3-Methylglutaconic Aciduria Type I	AUH
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH
Abetalipoproteinemia	MTTP

Argininemia	ARG1
Argininosuccinic Aciduria	ASL
Biopterin Defect In Cofactor Biosynthesis	GCH1
Biotinidase Deficiency	BTD
Carnitine Acylcarnitine Translocase Deficiency	SLC25A20
Carnitine Palmitoyltransferase Type I Deficiency	CPT1A
Carnitine Palmitoyltransferase Type II Deficiency	CPT2
Carnitine Uptake Defect/Carnitine Transport Defect	SLC22A5
Cerebral Creatine Deficiency Syndrome	GAMT, GATM
Cerebrotendinous Xanthomatosis	CYP27A1
Citrullinemia, Type I	ASS1
Citrullinemia, Type II	SLC25A13
Classic Galactosemia	GALT
Combined Pituitary Hormone Deficiency	LHX3, PROP1
Congenital Disorder of Glycosylation 1b	MPI
Corticosterone Methyloxidase Deficiency	CYP11B2
Crigler-Najjar Syndrome	UGT1A1
Cystinosis	CTNS
Fabry Disease	GLA
Galactoepimerase Deficiency	GALE
Galactokinase Deficiency	GALK1
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD
Glutaric Acidemia Type I	GCDH
Glutaric Acidemia Type II	ETFA, ETFB, ETFDH
Glycogen Storage Disease Ia	G6PC
Glycogen Storage Disease IIIa	AGL
Glycogen Storage Disease Type 0	GYS2
Glycogen Storage Disease Type Ib	SLC37A4
Glycogen Storage Disease Type II (Pompe)	GAA
Glycogen Storage Disease VI	PYGL
Hereditary Fructose Intolerance	ALDOB
HMG-CoA Lyase Deficiency	HMGCL
Holocarboxylase Synthase Deficiency	HLCS
Homocystinuria	CBS
Homocystinuria (Cobalamin Disorders)	MMADHC, MTR, MTRR
Hypercholesterolemia	LDLR
Hypermethioninemia	AHCY, GNMT, MAT1A
Hypophosphatasia	ALPL

Isobutyrylglycinuria	ACAD8
Isovaleric Acidemia	IVD
Krabbe Disease	GALC
Lipoprotein Lipase Deficiency (LPL)	LPL
Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD Deficiency)	HADHA
Lysinuric Protein Intolerance	SLC7A7
Lysosomal Acid Lipase Deficiency	LIPA
Malonyl-CoA decaMalonyl-CoA decarboxylase deficiency	MLYCD
Maple Syrup Urine Disease	BCKDHA, BCKDHB, DBT
Maple Syrup Urine Disease Type III	DLD
3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADH
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	ACADM
Metachromatic Leukodystrophy	ARSA
Methylmalonic Acidemia (Cobalamin Disorders)	MMAA, MMAB, MMADHC
Methylmalonic Acidemia with Homocystinuria	ABCD4, LMBRD1, MMACHC, MMADHC, HCFC1
Methylmalonic Acidemia, Type mut (0)	MMUT
Methylmalonyl-CoA Epimerase Deficiency	MCEE
Mucopolysaccharidosis Type 1	IDUA
Mucopolysaccharidosis Type II (Hunter Syndrome)	IDS
N-Acetylglutamate Synthase Deficiency	NAGS
Nephrogenic Diabetes Insipidus Type II	AQP2
Niemann-Pick Disease Type A/B	SMPD1
Niemann-Pick Disease Type C1/D	NPC1
Ornithine Transcarbamylase Deficiency	OTC
Ornithine Translocase Deficiency; Triple H Syndrome	SLC25A15
Phenylalanine Hydroxylase Deficiency (Phenylketonuria)	PAH
Primary Hyperoxaluria Type I	AGXT
Primary Hyperoxaluria Type II	GRHPR
Primary Hyperoxaluria Type III	HOGA1
Propionic Acidemia	PCCA, PCCB
Short-Chain Acyl-CoA Dehydrogenase Deficiency	ACADS
Alpha-methylacetoacetic aciduria	ACAT1
Transient Infantile Liver Failure	TRMU
Trifunctional Protein Deficiency	HADHA, HADHB
Segawa syndrome	TH

Tyrosinemia, Type I	FAH
Tyrosinemia, Type II	TAT
Tyrosinemia, Type III	HPD
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	ACADVL
Wilson Disease	ATP7B
X-Linked Adrenoleukodystrophy	ABCD1

OTHER – Genetic, Immunodeficiency, Pulmonary, Musculoskeletal

Cystic Fibrosis	CFTR
Severe Combined Immunodeficiencies	ADA, IL7R, JAK3, IL2RG
Immunodeficiency 14A/B	PIK3CD

Cardiomyopathy Kit

Disorder	Gene
Arrhytmogenic Cardiopathy (ACM)/ Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	CTNNA3
	DES
	DSC2
	DSG2
	DSP
	FLNC
	JUP
	LMNA
	PKP2
	PLN
	RYR2
	TGFB3
	TMEM43
	TTN
Cardiomyopathy dilated	ABCC9
	ACTC1
	ACTN2
	ANKRD1
	BAG3
	CASQ2
	CRYAB
	CSRP3
	DES
	DMD
	DSC2
	DSG2
	DSP
	DTNA
	EMD
	FKTN
	FLNC
	GATAD1
	ILK
	JPH2
	JUP
	LAMA4
	LAMP2

Cardiomyopathy dilated	LDB3
	LMNA
	MYBPC3
	MYH6
	MYH7
	MYL2
	MYL3
	MYPN
	NEBL
	NEXN
	PDLIM3
	PKP2
	PLN
	PRDM16
	RAF1
	RBM20
	RYR2
	SCN5A
	SGCD
	TAFAZZIN
	TCAP
	TMEM43
	TNNC1
	TNNI3
	TNNT2
	TPM1
	TTN
	TTR
	VCL
Cardiomyopathy hypertrophic (HCM)	AARS2
	ABCC9
	ACAD9
	ACADVL
	ACTA1
	ACTC1
	AGK
	ALMS1
	ALPK3

Cardiomyopathy with onset in neonatal period infancy or childhood	BAG3
	COA5
	COA6
	COX15
	CPT2
	CRYAB
	DNAJC19
	DOLK
	DSP
	ELAC2
	FLNC
	GAA
	GBE1
	GLA
	GTPBP3
	HADHA
	HADHB
	HRAS
	KARS1
	LAMP2
	MRPL3
	MRPL44
	MTO1
	MYBPC3
	MYH7
	MYPN
	NDUFB11
	NDUFV2
	PPA2
	PRKAG2
	RAF1
	SCO2
	SLC22A5
	SLC25A20
	SLC25A3
	SLC25A4
	TAFAZZIN
	TK2

Cardiomyopathy with onset in neonatal period infancy or childhood	TMEM70
	TNNI3
	TNNT2
	TPM1
	TSFM
Left Ventricular Non compaction Cardiomyopathy (NCCM/LVNC)	ACTC1
	DTNA
	HCN4
	LDB3
	LMNA
	MIB1
	MYBPC3
	MYH7
	PKP2
	PRDM16
	TAFAZZIN
	TNNT2
	TPM1
	TTN
Cardiomyopathy restrictive	ACTC1
	BAG3
	CRYAB
	DES
	FLNC
	MYBPC3
	MYH7
	MYL2
	MYL3
	MYPN
	TNNI3
	TNNT2
	TPM1
	TTR
Cardiomyopathy hypertrophic (HCM)	ACTC1
	ACTN2
	AKAP9
	ANKRD1
	CAV3

Cardiomyopathy hypertrophic (HCM)	CRYAB
	CSRP3
	DES
	FHL1
	FLNC
	GAA
	GLA
	JPH2
	LAMP2
	LDB3
	MYBPC3
	MYH6
	MYH7
	MYL2
	MYL3
	MYLK2
	MYOZ2
	MYPN
	NEXN
	PLN
	PRKAG2
	TCAP
	TNNC1
	TNNI3
	TNNT2
	TPM1
	TTN
	TTR
	VCL

Arrhythmia Kit

Disorder	Gene
Brugada-Syndrome (BrS)	ABCC9
	CACNA1C
	CACNA2D1
	CACNB2
	GPD1L
	HCN4
	KCNAB2
	KCND3
	KCNE3
	KCNH2
	KCNJ8
	RYR2
	SCN10A
	SCN1B
	SCN2B
	SCN3B
	SCN5A
	TRPM4
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	ANK2
	CALM1
	CALM2
	CALM3
	CASQ2
	GNAI2
	HCN4
	KCNJ2
	KCNQ1
	LMNA
	RYR2
	SCN5A
	TECRL
	TRDN
Long QT Syndrome (LQTS)	AKAP9
	ANK2
	CACNA1C

Long QT Syndrome (LQTS)	CALM1
	CALM2
	CALM3
	CAV3
	KCNE1
	KCNE2
	KCNH2
	KCNJ2
	KCNJ5
	KCNQ1
	RYR2
	SCN4B
	SCN5A
	SNTA1
	TECRL
	TRDN
Atrial Fibrillation and Short QT Syndrome	ABCC9
	CACNA1C
	CACNA2D1
	CACNB2
	GJA5
	HCN4
	KCNA5
	KCNE1
	KCNE2
	KCNH2
	KCNJ2
	KCNQ1
	LMNA
	MYL4
	NPPA
	SCN1B
	SCN2B
	SCN3B
	SCN4B
	SCN5A

Atrial Fibrillation and Short QT Syndrome	TBX5
	TNNI3

Aortopathy Kit

Disorder	Gene
Aortopathy *(Includes disorders such as Marfan syndrome, Ehlers-Danlos Syndrome, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysms and Dissections, and Bicuspid Aortic Valve Disease)*	ABCC6
	ACTA2
	ACVR1
	ADAMTS2
	ALDH18A1
	ATP6V0A2
	ATP6V1A
	ATP6V1E1
	B3GALT6
	B4GALT7
	BGN
	C1R
	C1S
	CBS
	CHST14
	COL1A1
	COL1A2
	COL3A1
	COL4A1
	COL5A1
	COL5A2
	DSE
	EFEMP2
	ELN
	FBLN5
	FBN1
	FBN2
	FKBP14
	FOXE3
	GORAB
	LOX
	LTBP4
	MFAP5
	MYH11
	MYLK
	PLOD1
	PRDM5

Aortopathy *(Includes disorders such as Marfan syndrome, Ehlers-Danlos Syndrome, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysms and Dissections, and Bicuspid Aortic Valve Disease)*	PYCR1
	SKI
	SLC2A10
	SLC39A13
	SMAD3
	SMAD4
	TGFB2
	TGFB3
	TGFBR1
	TGFBR2
	ZNF469

Congenital Heart Defects Kit

Disorder	Gene
Isolated and syndromal congenital heart defects *(Includes disorders such as Heterotaxy, Allagille Syndrome, Atrioventricular Septal defect, Atrial Septal defect, Ventricular Septal defect, Aortic Stenosis, and Tetralogy of Fallot)*	ACTA2
	ACTC1
	ACVR2B
	AFF4
	BMPR2
	CAD
	CFAP53
	CDK13
	CELSR1
	CELSR2
	CELSR3
	CHD4
	CHD7
	CITED2
	CREBBP
	CRELD1
	DNAH11
	DNAH5
	DNAH6
	DNAI1
	DTNA
	EHMT1
	ELN
	EVC
	EVC2
	FBN1
	FLNA
	FOXC1
	FOXH1
	GANAB
	GATA4
	GATA5
	GATA6
	GDF1
	GPC3
	HAND1
	HAND2

Isolated and syndromal congenital heart defects *(Includes disorders such as Heterotaxy, Allagille Syndrome, Atrioventricular Septal defect, Atrial Septal defect, Ventricular Septal defect, Aortic Stenosis, and Tetralogy of Fallot)*	HRAS
	JAG1
	KDM5B
	KMT2D
	MED13L
	MMP21
	MYH11
	MYH6
	MYH7
	NIPBL
	NKX2-5
	NKX2-6
	NME7
	NODAL
	NOTCH1
	NOTCH2
	NR2F2
	PITX2
	PKD1L1
	PLD1
	POGZ
	PRDM6
	PRKD1
	RABGAP1L
	RBFOX2
	RBM10
	SALL4
	SEMA3D
	SEMA3E
	SMAD6
	TAB2
	TBX1
	TBX20
	TBX5
	TFAP2B
	TGFBR1
	TGFBR2
	TLL1

Isolated and syndromal congenital heart defects *(Includes disorders such as Heterotaxy, Allagille Syndrome, Atrioventricular Septal defect, Atrial Septal defect, Ventricular Septal defect, Aortic Stenosis, and Tetralogy of Fallot)*	TMEM260
	TPM1
	ZEB2
	ZFPM2
	ZIC3

FH, PH and RASopathies Kit

Disorders	Genes
RASopathies *(Includes disorders such as Cardio-Facio-Cutaneous Syndrome, Costello Syndrome, Legius Syndrome, Neurofibromatosis Type 1, Noonan Syndrome, Noonan with multiple lentigines, and Noonan-syndrome like disorder with loose anagen hair)*	AKT3
	BRAF
	CBL
	CCND2
	EPHB4
	HRAS
	KRAS
	LZTR1
	MAP2K1
	MAP2K2
	MRAS
	NF1
	NF2
	NRAS
	PIK3CA
	PIK3R2
	PPP1CB
	PTPN11
	RAF1
	RASA1
	RASA2
	RIT1
	RRAS
	SASH1
	SHOC2
	SMARCB1
	SOS1
	SOS2
	SPRED1
	STAMBP
Familial Hypercholesterolemia (FH)	ABCA1
	ABCG5
	ABCG8
	APOA5
	APOB
	APOE
	LDLR

Familial Hypercholesterolemia (FH)	LDLRAP1
	LIPA
	LPL
	PCSK9
Pulmonary Hypertension (PH)	ACVRL1
	BMPR1B
	BMPR2
	CAV1
	EIF2AK4
	ENG
	KCNA5
	KCNK3
	SMAD4
	SMAD9
	TBX4

Metabolic Kit

Disorder	Genes
The list of disorders mentioned above is not exhaustive	
Hyperinsulinemic hypoglycemia, familiar,1	ABCC8
Adrenoleukodystrophy	ABCD1
Methylmalonic acidemia and homocystinuria, cblJ Type	ABCD4
Isobutyryl-CoA dehydrogenase deficiency (Other names: IBD Deficiency, Deficiency of Acyl-CoA Dehydrogenase Family Member 8 (ACAD8 Deficiency)	ACAD8
Mitochondrial complex I deficiency, nuclear type 20 (Other names: Mitochondrial Complex Deficiency due to ACAD9 Defciency, Akyl-CoA Dehydrogenase 9 Deficiency, ACAD9 Deficiency)	ACAD9
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (Other names: ACADM Deficiency,MCAD Deficiency, MCADH Deficiency, Carnitine Deficiency Secondary to Medium-chain Acyl-CoA)	ACADM
Short chain acyl-CoA dehydrogenase (SCHAD) deficiency (Other names: ACADS Deficiency, Lipid-Storage Myopathy Secondary to short-chain Acyl-CoA, Dehydrogenase Deficiency, SCADH Deficiency, SCAD Deficiency)	ACADS
Short/Branched Chain Acyl-CoA Dehydrogenase Deficiency-SBCADD (Other names: 2-Methylbutyryl-CoA Dehydrogenase Deficiency, 2-methulbutyrylglycinuria)	ACADSB
Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency (Other names: VLCAD Deficiency)	ACADVL
Peroxisomal acyl-CoA oxidase deficiency (Other names: Straight-Chain Acyl-CoA Oxidase Deficiency, Pseudoneonatal Adrenoleukodystrophy)	ACOX1
Combined malonic and methylmalonic acidemia	ACSF3
Aspartylglucosaminuria	AGA
Glycogen storage disease III (a and b) (Other names: Forbes Disease, Cori disease, Limit Dextrinosis, Amylo-1,6-Glucosidase Deficiency, AGL Deficiency, Glycogen Debrancher Deficiency, GDE Deficiency)	AGL
Rhizomelic chondrodysplasia punctata, type 3 (Other names: Alkyldihydroxyacetonephosphate Synthase Deficiency, Alkylglycerone-Phosphate Synthase Deficiency, AGPS Deficiency)	AGPS
Methylmalonate semialdehyde dehydrogenase deficiency (Other names: MMSDH Deficiency)	ALDH6A1
Glycogen storage disease XII (Other names: GSD XII, Aldolase A Deficiency, ALDOA Deficiency, Aldolase Deficiency-Red Cell, Red Cell Aldolase Deficiency)	ALDOA
Fructose intolerance, hereditary (Other names: Fructosemia, Fructose-1 Phosphate Aldolase Deficiency, Fructose-1,6 Bisphosphate Aldolase B Deficiency, Aldolase B Deficiency, ALDOB Deficiency)	ALDOB
Congenital disorder of glycosylation, type Ik	ALG1
Congenital disorder of glycosylation, type Ip	ALG11
Congenital disorder of glycosylation, type Ig	ALG12

Congenital disorder of glycosylation, type Is (Other names: Developmental and epileptic encephalopathy 36, Epileptic Encephalopathy, Early Infantile, 36)	ALG13
Congenital disorder of glycosylation, type Ii	ALG2
Congenital disorder of glycosylation, type Id (Other names: CDG Id-CDGId, Carbohydrate-Deficient Glycoprotein Syndrome, Type IV, Formerly, CDGS4 Formerly)	ALG3
Congenital disorder of glycosylation, type Ic (Other names: CDG-Ic, Carbohydrate-Deficient Glycoprotein Syndrome Type I with deficient glycosylation of dolichol-linked oligosaccharide, Formerly Carbohydrate-Deficient Glycoprotein Syndrome, Type V, CDGS5 Formerly)	ALG6
Congenital disorder of glycosylation, type Ih-CDG1H	ALG8
Congenital disorder of glycosylation, type I1-CDGI1	ALG9
Gillessen-Kaesbach-Nishimura syndrome (Other names: Polycystic Kidney Disease, Autosomal Recessive with Microbrachycephaly, Hypertelorism and Brachymelia)	
Alpha-methylacyl-CoA racemase deficiency (AMACR Deficiency)	AMACR
Congenital Bile acid synthesis defect 4 (Other names: Cholestasis Intrahepatic with defective conversion of trihydroxycoprostanic acid to cholic acid, Trihydroxycoprostanic Acid in Bile)	
Glycine encephalopathy (Other names: Hyperglycinemia nonketotic-NKH)	AMT
Argininemia (Other names: Arginase Deficiency, Hyperargininemia, ARG1 Defciency)	ARG1
Metachromatic leukodystrophy-MLD (Metachromatic Leukoencephalopathy, Cerebral Sclerosis Diffuse Metachromatic Form, Sulfatide Lipidosis, Arylsulfatase A Deficiency, ARSA Deficiency, Cerebroside Sulfatase Deficiency)	ARSA
Mucopolysaccharidosis type VI (Maroteaux-Lamy) (Other names : MPS VI, Maroteaux-Lamy Syndrome, Arylsulfatase B deficiency, ARSB Deficiency, N-Acetylgalactosamine-4-Sulfatase Deficiency)	ARSB
Argininosuccinic aciduria (Other names: Argininosuccinase Deficiency, Argininosuccinate Lyase Deficiency, ASL Deficiency, Argininosuccinic Acid Lyase Deficiency)	ASL
Canavan disease (Other names: Canavan-Van Bogaert-Bertrand Disease, Spongy Degeneration of Central Nervous System, Aspartoacylase Deficiency, ASPA Deficiency, ASP Deficiency, Aminoacylase 2 Deficiency, ACY2 Deficiency)	ASPA
Citrullinemia (Other names: Citrullinemia Type I-CTLN1, Citrullinuria, Argininosuccinate Synthetase Deficiency, ASS Deficiency)	ASS1
3-methylglutaconic aciduria, type I-MGCA1 (Other names: MGA Type I-MGA1, 3-Methylglutaconyl-CoA Hydratase Deficiency, 3-MG-CoA Hydratase Deficiency)	AUH
Congenital disorder of glycosylation, type IId	B4GALT1

Maple syrup urine disease, type Ia (Other names: Branched-Chain Ketoaciduria, Branched-Chain Alpha-Keto Acid Dehydrogenase Deficiency, BCDK Deficiency, Keto Acid Decarboxylase Deficiency)	BCKDHA
Maple syrup urine disease, type Ib (Other names: Branched-Chain Ketoaciduria, Branched-Chain Alpha-Keto Acid Dehydrogonase Deficiency, BCKD Deficiency, Keto Acid Decarboxylase Deficiency)	BCKDHB
Biotinidase deficiency (Other names: BTD Deficiency, Multiple Carboxylase Deficiency Late-Onset, Multiple Carboxylase Deficiency Juvenile-Onset)	BTD
Developmental and Epileptic encephalopathy 50 (Other names: Epileptic Encephalopathy early infantile 50-EIEE50, Congenital Disorder of Glyxosylation type Iz formerly-CDG1Z)	CAD
Congenital disorder of glycosylation, type IIo	CCDC115
Methylmalonic aciduria, transient, due to transcobalamin receptor defect (Other names: Methylmalonic Acidemia, TCblR Type)	CD320
Neuronal ceroid lipofuscinosis 3 (Other names: Neuronal Ceroid Lipofuscinosis Juvenile, Batten Disease, Vogt-Spielmeyer Disease, Spielmeyer-Sjogren Disease)	CLN3
Neuronal ceroid lipofuscinosis 5 (Other names: Neuronal ceroid lipofuscinosis 5, Variable age at onset)	CLN5
Neuronal ceroid lipofuscinosis 6A (Other names: Neuronal Ceroid Lipofuscinosis, late infantile variant- vLINCL)	CLN6
Neuronal ceroid lipofuscinosis 6B	
Neuronal ceroid lipofuscinosis 8	CLN8
Neuronal ceroid lipofuscinosis 8, Northern epilepsy variant (Other names: Epilepsy Progressive with Mental Retardation -EPMR)	
3-methylglutaconic aciduria type VIIA (Other names: 3-Methylglutaconinc Aciduria with Neurologic Involvement and Neutropenia)	CLPB
3-methylglutaconic aciduria type VIIB (Other names: MGCA7, 3-Methylglutaconic Aciduria with Cataracts,Neurologic Involvement and Neutropenia-MEGCANN)	
Severe congenital Neutropenia 9	
Congenital disorder of glycosylation, type IIg	COG1
Congenital disorder of glycosylation, type IIq-CDG IIq	COG2
Congenital disorder of glycosylation, type Iij- cdg IIj	COG4
Saul-Wilson syndrome (Other names: Microcephalic Osteodysplastic Dysplasia)	
Congenital disorder of glycosylation, type IIi- CDG IIi	COG5
Congenital disorder of glycosylation, type IIl- CDG Iil	COG6
Shaheen syndrome	
Congenital disorder of glycosylation, type IIe	COG7
Congenital disorder of glycosylation, type IIh-CDG IIh	COG8
Carbamoylphosphate synthetase I deficiency (Other names: CPS I Deficiency)	CPS1

Carnitine palmitoyl transferase type I A deficiency (Other names: CPT I Deficiency, CPT Deficiency, Hepatic, Type I)	CPT1A
Carnitine palmitoyltransferase II deficiency, infantile (Other names: Carnitine Palmitoultransferase II Deficiency with Hypoketotic Hypoglycemia, Carnitine Palmitoultransferase II Deficiency Hepatocardiomuscular, CPT II Deficiency Hepatic, CPT2 Deficiency Infantile)	CPT2
Carnitine palmitoyltransferase II deficiency, lethal neonatal (Carnitine Palmitoultransferase II Deficiency Neonatal, Carnitine Palmitoultransferase II Deficiency Antenatal, CPT II Deficiency Lethal Neonatal, CPT2 Deficiency Lethal Neonatal)	
Carnitine palmitoyltransferase II deficiency, myopathic, stress-induced (Other names: Carnitine Palmitoultransferase II Deficiency Myopathic, Carnitine Palmitoultransferase II Deficiency Adult-Onset, CPT II Deficiency Myopathic, CPT2 Deficiency Late-Onset)	
Cystinosis, atypical nephropathic or nephropathic (Other names: Defect of Lysosomal Cystine Transport Protein, Defect Of Cystinosin)	CTNS
Cystinosis, late-onset juvenile or adolescent nephropathic (Other names: Cystinosis Intermediate)	
Cystinosis, adult nonnephropathic (Other names: Cystinosis Ocular Nonnephropathic, Cystinosis Benign Nonnephropathic)	
Galactosialidosis (Other names: Goldberg Syndrome, Neuraminidase Deficiency with Beta-Galactosidase Deficiency, Neuraminidase/Beta-Galactosidase Expression-NGBE, Lysosomal Protective Protein Deficiency, Cathepsin A Deficiency, Protective Protein/Cathepsin A Deficiency, PPCA Deficiency)	CTSA
Pycnodysostosis (Other names: Pyknodysostosis, PYCD)	CTSK
Maple syrup urine disease, type II (Other names: Branched-Chain Ketoaciduria, Branched-Chain Alpha-Keto Acid Dehydrogenase Deficiency, BCKD Deficiency, Keto Acid Decarboxylase Deficiency)	DBT
Congenital disorder of glycosylation, type Ir- CDG Ir	DDOST
Smith-Lemli-Opitz syndrome (Other names: SLO Syndrome, RSH Syndrome, Rytledge Lethal Multiple Congenital Anomaly Syndrome, Polydactyly Sex Reversal Renal Hypoplasia and unilobar Lung Lethal Acrodysgenital Syndrome)	DHCR7
Congenital disorder of glycosylation type 1bb - Retinitis pigmentosa 59	DHDDS
Developmental delay and seizures with or without movement abnormalities	
Maple syrup urine disease, type III (Other names: Dihydrolipoamide dehydrogenase deficiency, DLD deficiency)	DLD
Hyperphenylalaninemia, mild, non-BH4-deficient	DNAJC12
3-methylglutaconic aciduria, type V-MGCA5 (Other names: Dilated cardiomyopathy with ataxia (DCMA))	DNAJC19
Congenital disorder of glycosylation, type Im (Other names: Dolichol Kinase Deficiency, DK1 deficiency)	DOLK
Congenital disorder of glycosylation, type Ij-CDG Ij	DPAGT1

Congenital Myasthenic syndrome, 13, with tubular aggregates (Other names: Congenital Myasthenic Syndrome with tubular aggregates 2-CMSTA2)	DPAGT1
Congenital disorder of glycosylation, type Ie- CDG Ie	DPM1
Congenital disorder of glycosylation, type Iu- CDG Iu	DPM2
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B,15 (Other names: Muscular Dystrophy Congenital DPM3-Related)	DPM3
Muscular dystroph-dystroglycanopathy (limb-girdle), type C, 15-MDDGC15 (Other names: Muscular Dystrophy-Dystroglycanopathy, Limb-Girdle DPM3-Related Congenital Disorder of Glycosylation- Type Io -CDG1O, CDG Io, CDG1)	
Glycogen storage disease XIII (13) (Other names: GSD XIII, Enolase 3 Deficiency, Enolase Beta Deficiency)	ENO3
Multiple Acyl-CoA dehydrogenase deficiency, MADD (Other names: Glutaric acidemia type IIA, Glutaric Aciduria IIA, GA2A, Ethylmalonic-Adipicaciduria-EMA)	ETFA
Multiple Acyl-CoA dehydrogenase deficiency, MADD (Other names: Glutaric acidemia type IIB, Glutaric Aciduria IIB, GA2B, Ethylmalonic-Adipicaciduria-EMA)	ETFB
Multiple Acyl-CoA dehydrogenase deficiency, MADD (Other names: Glutaric acidemia type IIC, Glutaric Aciduria IIC, GA2C, Ethylmalonic-Adipicaciduria-EMA)	ETFDH
Fructose-1,6-bisphosphatase deficiency	FBP1
Fucosidosis (Other names: Alpha-L-Fucosidase Deficiency)	FUCA1
Congenital disorder of glycosylation with defective fucosylation 1-CDGF	FUT8
Glycogen storage disease Ia (GSD Ia, Glycogen Storage Disease I-GSD1, Von Gierke Disease, Hepatorenal Form of Glycogen Storage Disease, Glucose-6-Phosphatase Deficiency, Hepatorenal Glycogenosis)	G6PC
Glycogen storage disease II (Other names: GSD II, Acid Alpha-Glucosidase Deficiency, GAA Deficiency, Pompe Disease, Glycogenosis, Generalized Cardiac Form, Cardiomegalia Glycogenica Diffusa, Acid Maltase Deficiency-AMD, Alpha-1,4-Glucosidase Deficiency)	GAA
Krabbe disease (Other names: Globoid Cell Leukodystrophy-GLD, Globoid Cell Leukoencephalopathy, Galactosylceramide Beta-Galactosidase Deficiency, Galactocerebrosidase Deficiency, Galc Deficiency)	GALC
Mucopolysaccharidosis type IVA (Other names: Morquio Syndrome A, MPS IVA, Morquio A Disease, Galactosamine-6-Sulfatase Deficiency, GALNS Deficiency)	GALNS
Cerebral creatine deficiency syndrome 2-CCDS2 (Other names: Guanidinoacetate Methyltransferase Deficiency, GAMT Deficiency, Creatine Deficiency Syndrome Due to GAMT Deficiency)	GAMT

Cerebral creatine deficiency syndrome 3-CCDS3 (Other names: Arginine:Glycine Amidinotransferase Deficiency, AGAT Deficiency, GATM Deficiency, Creatine Deficiency Syndrome due to AGAT Deficiency)	GATM
Gaucher Disease, Perinatal lethal (Other names: Gaucher Disease, Collodion Type)	GBA
Gaucher disease,Type I (Other names: GD I, Gaucher Disease, Noncerebral Juvenile, Glucocerebrosidase Deficiency, Acid Beta-Glucosidase Deficiency, GBA Deficiency)	
Gaucher disease,Type II (Other names: Gaucher Disease, Accute Neuronopathic Type)	
Gaucher disease,Type III (Other names: Gaucher Disease, Subaccute Neuronopathic Type, Gaucher disease, Chronic neuropathic type, Gaucher Disease, juvenile and adult cerebral)	
Gaucher disease,Type IIIC	
Glycogen storage disease IV (Other names: GSD IV, Glycogen Branching Enzyme Deficiency, GBE1 Deficiency, Andersen Disease, Brancher Deficiency, Glycogenosis IV, Amylopectinosis, Cirrhosis, Familial with Deposition of Abnormal Glycogen)	GBE1
Polyglucosan body disease, adult form (Other names: Polyglucosan Body Neuropathy adult form-APBN)	
Glutaricaciduria, type I (Other names: Glutaric Acidemia I-GA1, Glutaric Aciduria I, GA I, Glutaryl-CoA Dehydrogenase Deficiency)	GCDH
Hyperphenylalaninemia, BH4-deficient, B-HPABH4B (Other names: Hyperphenylalaninemia Tetrahydrobiopterin-Deficient Due to GTP, Cyclohydrolase I Deficiency), GTP Cyclohydrolase I Deficiency)	GCH1
Dystonia, DOPA-responsive, with or without Hyperphenylalaninemia (Other names: Dystonia Dopa-Responsive-DRD, Dystonia 5-DYT5, Dystonia Progressive with Diurnal Variation, Dystonia-Parkinsonism with Diurnal Fluctuation, Segawa Syndrome Autosomal Dominant, Dystonia Dopa-Responsive Autosomal Dominant, Dopa-Responsive Dystonia Autosomal Dominant)	
Hyperinsulinemic hypoglycemia, familial, 3	GCK
Glycine encephalopathy-GCE (Other names: Hyperglycinemia Nonketotic-NKH)	GCSH
Fabry disease (Other names: Angiokeratoma Corporis Diffusum, Anderson-Fabry Disease, Hereditary Dystopic Lipidosis, Alpha-Galactosidase A Deficiency, GLA Deficiency, Ceramide Trihexosidase Deficiency)	GLA
GM1-gangliosidosis, type I (Other names: Gangliosidosis Generalized GMA1 Type 1, Ganliosidosis Generalized GM1 Infantile Form, Gangliosidosis Generalized GM1 Type 1, Beta-Galactosidase-1 Deficiency, GLB1 Deficiency)	GLB1
GM1-gangliosidosis, type II (Other names: Gangliosidosis Generalized GM1 Juvenile Type, Gangliosidosis Generalized GM1 Type II, Gangliosidosis Generalized GM1 Type 2)	

GM1-gangliosidosis, type III (Other names: Gangliosidosis Generalized GM1 Adult Type, Gangliosidosis Generalized GM1 Chronic Type, Ganlgiosidosis Generalized GM1Type III, Gangliosidosis Generalized GM1, Type 3)	GLB1
Mucopolysaccharidosis type IVB (Morquio) (Other names: Morquio Syndrome B, MPS IVB)	
Glycine encephalopathy (Other names: Hyperglycinemia Nonketotic-NKH)	GLDC
Hyperinsulinism-hyperammonemia syndrome (Other names: Hyperinsulinemic Hypoglycemia Familial 6-HHF6)	GLUD1
GM2-gangliosidosis, AB variant (Other names: Hexosaminidase Activator Deficiency, GM2 Activator Deficiency, AB Variant GM2-Gangliosidosis, Tay-Sachs Disease AB Variant)	GM2A
Alacrima, achalasia, and mental retardation syndrome	GMPPA
Nonaka myopathy (other names: Nonanak Distal Myopathy, Myopathy Distal with or without Rimmed Vacuoles, Inclusion Body Hereditary Myopathy Autosomal Recessive-HIBM, Inclusion Body Myopathy Quadriceps-Sparing-QSM, GNE Myopathy, Inclusion Body Myopathy 2 Autosomal Recessive Formerly IBM2)	GNE
Sialuria (Other names: Sialuria French Type)	
Mucolipidosis type II alpha/beta (Other names: I-Cell Disease-ICD)	GNPTAB
Mucolipidosis type III alpha/beta (Other names: Pseudo-Hurler Polydystrophy)	
Mucolipidosis III gamma	GNPTG
Mucopolysaccharidosis type IIID (Other names: N-Acetylglucosamine-6-Sulfatase Deficiency, Sanfilippo syndrome D)	GNS
Mucopolysaccharidosis VII (mps vii, SLY Syndrome, Beta-Glucuronidase Deficiency, GUSB Deficiency)	GUSB
Glycogen storage disease XV (Other names: GSD XV, Glycogenin Deficiency, GYG1 Deficiency)	GYG1
Polyglucosan body myopathy 2	
Glycogen storage disease 0, muscle (Other names: GSD 0b, Muscle Glycogen Storage Disease 0, Muscle Glycogen Synthase Deficiency)	GYS1
Glycogen storage disease 0, liver (Other names: GSD 0a, Hypoglycemia with Deficiency of Glycogen Synthetase in the liver, Liver Glycogen Synthase Deficiency, Liver Glycogen Storage Disease 0)	GYS2
Hyperinsulinemic hypoglycemia, familial, 4	HADH
3-hydroxyacyl-CoA dehydrogenase deficiency (Other names: HADH Deficiency, SCHAD Deficiency Formerly)	
long chain 3-hydroxyacyl-CoA dehydrogenase (Other names: LCHAD Deficiency)	HADHA
Mitochondrial trifunctional protein deficiency-MTPD (Other names: Trifunctional Protein Deficiency)	
Mitochondrial trifunctional protein (TFP) deficiency-MTPD (Other names: Trifunctional Protein Deficiency)	HADHB

Methylmalonic acidemia and homocysteinemia, cblX type (Other names: Intellectual Developmental Disorder, X-Linked 3. Mental Retardation X-Linked 3)	HCFC1
Tay-Sachs disease (Other names: GM2-Gangliosidosis Type I, B Variant GM2-Gangliosidosis, Hexosaminidase A Deficiency, Hexa Deficiency)	HEXA
Sandhoff disease (Other names: GM2-Gangliosidosis Type II, Hexosaminidases A and B Deficiency)	HEXB
Mucopolysaccharidosis type IIIC (Sanfilippo C) (Other names: MPS IIIC, Sanfilippo Syndrome C, AcetYL-CoA:Alpha-Glucosaminide N-Acetyltransferase Deficiency)	HGSNAT
Retinitis pigmentosa 73	
HMG-CoA Lyase deficiency (Other names: HMGCL Deficiency, HL Deficiency, Hydroxymethylglutaric Aciduria)	HMGCL
3-hydroxy-3-methylglutaryl-CoA synthase 2 (HMGCS2D) deficiency (Other names: Mitochondrial HMG-CoA Synthase Deficiency, HMGCS2 Deficiency)	HMGCS2
Tyrosinemia, type III (Other names: 4-Hydroxyphenylpyruvic Acid Oxidase Deficiency, 4-Hydroxyphenylpyruvate Dioxygenase Deficiency)	HPD
Hawkinsinuria	
Costello syndrome (Other names: Faciocutaneoskeletal Syndrome, FCS Syndrome, Congenital myopathy with excess of muscle spindles)	HRAS
2-methyl-3-hydroxybutyric aciduria (HSD17B10 Deficiency, 17-Beta-Hydroxysteroid Dehydrogenase X Deficiency, 3-Hydroxyacyl-CoA Dehydrogenase II Deficiency, 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency, MHBD Deficiency, Mental Retardation X-Linked Syndromic 10-MRXS10, Choreoathetosis with Mental Retardation And Abnormal Behavior-CAMR, Mental Retardation with Choreoathetosis and Abnormal Behavior)	HSD17B10
D-bifunctional protein deficiency (Other names: 17-Beta-Hydroxysteroid Dehydrogenase IV Deficiency, DBP Deficiency, Peroxisomal Bifunctional Enzyme Deficiency, PBFE Deficiency)	HSD17B4
Mucopolysaccharidosis type IX (Other names: MPS IX, Hyaluronidase Deficiency)	HYAL1
Mucopolysaccharidosis type II (Other names: MPS II, Hunter Syndrome, Iduronate 2-Sulfatase Deficiency, IDS Deficiency, Sulfoiduronate Sulfatase Deficiency, SIDS Deficiency)	IDS
Mucopolysaccharidosis type Ih (Other names: Hurler Syndrome)	IDUA
Mucopolysaccharidosis type Ih/s (Other names: Hurler-Scheie Syndrome)	
Mucopolysaccharidosis type Is (Other names: Scheie Syndrome, Mucopolysaccharidosis Type V Formerly, MPS V Formerly)	
Hyperinsulinemic hypoglycemia, familial, 5	INSR

Hyperinsulinemic hypoglycemia, familial, 2-HHF2 (Other names: Persistent Hyperinsulinemic Hypoglycemia of Infancy-PHHI, Hyperinsulinemic Hypoglycemia Persistent, Hyperinsulinemic Hypoglycemia Due to Focal Adenomatous, Hyperplasia, Hyperinsulinism Neonatal, Hyperinsulinism Congenital, Hyperinsulinism Familial, Nesidioblastosis)	KCNJ11
Danon disease (Other names: Formerly glycogen storage disease IIb)	LAMP2
Glycogen storage disease XI (Other names: Lactate dehydrogenase A deficiency, LDH SUBUNIT M)	LDHA
Hyperglycinemia, lactic acidosis, and seizures-HGCLAS (Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency-PDHLD)	LIAS
Lysosomal Acid Lipase Deficiency (Other names: Wolman disease, Cholesteryl Ester Storage Disease-CESD, Lipa Deficiency, LAL Deficiency, Cholesterol Ester Hydrolase Deficiency)	LIPA
Methylmalonic Acidemia and Homocystinuria, cblF Type. (Other names: Methylmalonic Aciduria Due to Vitamin B12-Release Defect. Vitamin B12 Lysosomal Release Defect. Cobalamin Defect in Lysosomal Release of Vitamin B12 Storage Disease. Cobalamin F Disease)	LMBRD1
Rafiq syndrome	MAN1B1
Alpha-Mannosidosis (Other names: Mannosidosis Alpha B Lysosomal-MANSA, Lysosomal Alpha-D-Mannosidase Deficiency, Alpha-Mannosidase B Deficiency)	MAN2B1
Beta-Mannosidosis (Other names: Mannosidase beta A Lysosomal-MANBA, MANB1, Mannanase,Mannase)	MANBA
Methylmalonyl-CoA epimerase deficiency (Other names: Methylmalonul-CoA Racemase Deficiency, Methylmalonic Aciduria III Formerly)	MCEE
Mucolipidosis type IV (Other names: ML IV, Sialolipidosis)	MCOLN1
Neuronal ceroid lipofuscinosis 7	MFSD8
Macular dystrophy with central cone involvement	
Congenital disorder of glycosylation, type IIa (Other names: CDG IIa, Alkuraya Syndrome, Mental Retardation Growth Retardation Prominent Columella and Open Mouth, Carbohydrate-Deficient Glycoprotein Syndrome Type II Formerly, CDGS2 Formerly)	MGAT2
Malonyl-CoA decarboxylase deficiency	MLYCD
Methylmalonic Acidemia, cblA Type (Other names: Methylmalonic Aciduria Vitamin B12-Responsive, due to defect in Synthesis of Adenosylcobalamin cblA Type)	MMAA
Methylmalonic Acidemia, cblB Type (Other names: Methylmalonic Aciduria Vitamin B12-Responsive, due to defect in Synthesis of Adenosylcobalamin cblB Type)	MMAB

Methylmalonic Acidemia and Homocystinuria, cblC Type (Other names: Methylmalonic aciduria and homocystinuria, vitamin B12-Responsive. Vitamin B12 Metabolic Defect with Combined Deficiency of Methylmalonyl-CoA Mutase and Homocysteine:Methyltetrahydrofolate Methyltransferase)	MMACHC
Methylmalonic acidemia and homocystinuria, cblD type (Other names: Methylmalonic aciduria and homocystinuria cblD Type. Methylmalonic Aciduria, cblH Type Formerly. Methylmalonic Acidemia, cblH Type Formerly)	MMADHC
Congenital disorder of glycosylation, type IIb (CDG IIb, Glucosidase I Deficiency)	MOGS
Congenital disorder of glycosylation, type If (CDG If)	MPDU1
Congenital disorder of glycosylation, type Ib (Other names: CDG Ib, CDGIb, CDG Gastrointestinal Type, Mannosephosphate Isomerase Deficiency, MPI Deficiency, Protein-Losing Enteropathy-Hepatic Fibrosis Syndrome, Saguenay-Lac Saint-Jean Syndrome, SLSJ Syndrome)	MPI
Homocystinuria-Megaloblastic Anemia, cblG Type. (Other names: Methylcobalamin Deficiency cblG Type. Methionine Synthase Deficiency)	MTR
Homocystinuria-Megaloblastic Anemia, cblE Type. (Other names: Vitamin B12-Responsive Homocystinuria cblE Type. Methylcobalamin Deficiency cblE Type)	MTRR
Methylmalonic acidemia due to Methylmalonyl-CoA Mutase Deficiency (Other names: Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase Deficiency, MMA due to MCM Deficiency, Methylmalonic Aciduria mut Type)	MMUT
Schindler disease (Other names: Alpha-N-acetylgalactosaminidase (alpha-NAGA) deficiency)	NAGA
Mucopolysaccharidosis IIIB (Sanfilippo B) (Other names: Mucopolysaccharidosis Type IIIB-MPS3B, MPS IIIB, Sanfilippo Syndrome B, n-Acetyl-Alpha-D-Glucosaminidase Deficiency, NAGLU Deficiency)	NAGLU
N-acetylglutamate synthase (NAGS) deficiency (Other names: Hyperammonemia Due to N-Acetylglutamate Synthetase Deficiency, N-Acetylglutamate Synthetase Deficiency, NAGS Deficiency)	NAGS
Sialidosis type I and II (Other names: Sialidosis Type I, Mucolipidosis I, ML I, Lipomucopolysaccharidosis, Sialidase Deficiency, Glycoprotein Neuraminidase Deficiency, NEUG Deficiency, Neuraminidase 1 Deficiency, NEU Deficiency, NEU1 Deficiency)	NEU1
Congenital disorder of deglycosylation 1-CDDG1 (Other names: Congenital Disorder of Deglycosylation-CDDG, Congenital Disorder of Glycosylation Type Iv Formerly-CDG1V Formerly)	NGLY1

Niemann-Pick disease, type C1/type D (Other names: Niemann-Pick Disease Type C-NPC, Niemman-Pick Disease with Cholesterol Esterification Block, Niemann-Pick Disease Subacute Juvenile Form, Niemann-Pick Disease Chronic Neuronopathic Form, Niemann-Pick Disease Without Sphingomyelinase Deficiency, Neurovisceral Storage Disease with Vertical Supranuclear Ophthalmoplegia)	NPC1
Niemann-pick disease, type C2	NPC2
Congenital disorder of glycosylation, type 1aa	NUS1
Intellectual Developmental Disorder Autosomal Dominant 55 with Seizures-MRD55 (Other names: Mental retardation, autosomal dominant 55, with seizures)	
3-methylglutaconic aciduria, type III-MGCA3 (Other names: MGA Type III-MGA3, Optic Atrophy Plus Syndrome, Optic Atrophy Infantile with Chorea and Spastic Paraplegia, Iraqi-Jewish Optic Atrophy Plus, Costeff Syndrome, Optic Atrophy 3 Autosomal Recessive)	OPA3
Optic atrophy 3 with cataract (Other names: Optic Atrophy 3 Autosomal Dominant-OPA3, Optic Atrophy and Cataract Autosomal Dominant)	
Ornithine transcarbamylase (OTC) deficiency (Other names: Hypermammonemia due to Ornithine Transcarbamylase Deficiency, Ornithine Carbamoyltransferase Deficiency, OTC Deficiency)	OTC
Phenylalanine Hydroxylase Deficiency (Other names: Phenylketonuria, PAH Deficiency, Oligophrenia Phenylpyruvica, Folling Disease, Hyperphenylalaninemia non-PKU mild)	PAH
Hyperphenylalaninemia, BH4-deficient, D-HPABH4D (Other names: Hyperphenylalaninemia, Tetrahydrobiopterin-Deficient due to Pterin-4Alpha-Carbinolamine Dehydratase Deficiency, Hyperphenylalaninemia with Primapterinuria, CADH Deficiency, PCBD Deficiency)	PCBD1
Peroxisome biogenesis disorder 1A (Zellweger) (Other names: ZS-ZWS, Cerebrohepatorenal Syndrome-CHR)	PEX1
Peroxisome biogenesis disorder 1B (NALD/IRD) (Other names: Peroxisome Biogenesis Disorder (Neonatal Adrenoleukodystrophy/Infantile Refsum Disease), Peroxisome Biogenesis Disorder (NALD/IRD), Adrenoleukodystrophy Autosomal Neonatal, Refsum Disease Infantile, Infantile Phytanic Acid Storage Disease)	
Heimler syndrome 1 (Other names: Hearing loss sensorineural with enamel hypoplasia and nail defects peroxisome biogenesis disorder 1C-PBD1C)	
Peroxisome biogenesis disorder 6A (Zellweger)	PEX10
Peroxisome biogenesis disorder 6B	
Peroxisome biogenesis disorder 14B	PEX11B
Peroxisome biogenesis disorder 3A (Zellweger)	PEX12
Peroxisome biogenesis disorder 3B	

Peroxisome biogenesis disorder 11A (Zellweger)	PEX13
Peroxisome biogenesis disorder 11B	PEX13
Peroxisome biogenesis disorder 13A (Zellweger)	PEX14
Peroxisome biogenesis disorder 8A (Zellweger)	PEX16
Peroxisome biogenesis disorder 8B	
Peroxisome biogenesis disorder 12A (Zellweger)	PEX19
Peroxisome biogenesis disorder 5A (Zellweger)	PEX2
Peroxisome biogenesis disorder 5B	
Peroxisome biogenesis disorder 7A (Zellweger)	PEX26
Peroxisome biogenesis disorder 7B	
Peroxisome biogenesis disorder 10A (Zellweger)	PEX3
Perixosome biogenesis disorder 10B	
Peroxisome biogenesis disorder 2A (Zellweger)	PEX5
Peroxisome biogenesis disorder 2B	
Rhizomelic chondrodysplasia punctata, type 5	
Heimler syndrome 2 (Other names: Peroxisome Biogenesis Disorder 4C-PBD4C)	PEX6
Peroxisome biogenesis disorder 4A (Zellweger)	
Peroxisome biogenesis disorder 4B	
Peroxisome biogenesis disorder 9B (Other names: Refsum Disease Adult 2, Peroxisome Biogenesis Disorder Pex7-Related Atypical)	PEX7
Rhizomelic chondrodysplasia punctata, type 1 (Other names: Peroxisome Biogenesis Disorder 9-pbd9, Chondrodysplasia Puncata Rhizomelic Form-CDPR, Chondrodystrophia Calcificans Punctata)	
Glycogen storage disease VII (Other names: GSD VII, Muscle Phosphofructokinase Deficiency, PFKM Deficiency, Tarui Disease)	PFKM
Glycogen storage disease X (Other names: GSD X, Phosphoglycerate Mutase Muscle Deficiency of Myopathy due to Phosphoglycerate Mutase Deficiency, PGAMM Deficiency)	PGAM2
Congenital disorder of glycosylation, type It-CDG1T (Other names: CDG It, Phosphoglucomutase 1 Deficiency, PGM1 Deficiency, Glycogen Storage Disease XIV-GSD13, GSD XIV)	PGM1
Glycogen storage disease IXd (Other names: GSD IXd, Muscle Phosphorylase Kinase Deficiency, Muscle Glycogenosis X-Linked)	PHKA1
Glycogen storage disease IXa (Other names: Liver Glycogenosis X-Linked Type I-XLG1, Glycogen Storage Disease VIII Formerly, GSD VIII Formerly-GSD8)	PHKA2
Glycogen storage disease IXb (Other names: GSD IXb, Glycogenosis of Liver and Muscle Autosomal Recessive, Phosphorylase Kinase Deficiency of Liver and Muscle Autosomal Recessive)	PHKB
Glycogen storage disease IXc-GSD IXc	PHKG2

Refsum disease (Other names: Refsum Disease Adult 1, Phytanic Acid Oxidase Deficiency, Heredopathia Atactica Polyneuritiformis, Hereditary Motor and Sensory Neuropathy IV-HMSN4, HMSN IV)	PHYH
Congenital disorder of glycosylation, type Ia-CDG Ia (Other names: Jaeken Syndrome, Phosphomannomutase 2 Deficiency, Carbohydrate-Deficient Glycoprotein Syndrome Type Ia Formerly)	PMM2
Maple syrup urine disease, mild variant	PPM1K
Neuronal ceroid lipofuscinosis 1 (Other names: Ceroid Lipofuscinosis Neuronal 1 Variable Age at onset)	PPT1
Glycogen storage disease of heart, lethal congenital (Other names: [hosphorylase Kinase Deficiency of Heart, Glycogen Storage Disease of Heart)	PRKAG2
Combined SAP deficiency (Other names: Prosaposin Deficiency, Combined Saposin Deficiency)	PSAP
Gaucher disease, atypical (Other names: Saposin C Deficiency)	
Krabbe disease, atypical (Other names: Saposin A Deficiency)	
Metachrpomatic leukodystrophy due to SAP-B deficiency (Other names: Metachromatic Leukodystrophy due to Cerebroside Sulfatase Activator Deficiency, Saposin B deficiency)	
Hyperphenylalaninemia, BH4-deficient, A (Other names: Hyperphenylalaninemia Tetrahydrobiopterin-Deficient due to PTS Deficiency, 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency-PTSD, PTS Deficiency)	PTS
Glycogen storage disease VI	PYGL
McArdle disease (Other names: Myophosphorylase deficiency)	PYGM
Hyperphenylalaninemia, BH4-deficient, C (Other names: Hyperphenylalaninemia Tetrahydrobiopterin-Deficienct due to DHPR Deficiency, Dihydropteridine Reductase Deficiency, DHPR Deficiency, Quinoid Dihydropteridine Reductase Deficiency, QDPR Deficiency)	QDPR
Congenital disorder of glycosylation, type In	RFT1
Leukoencephalopathy with dystonia and motor neuropathy (Other names: Sterol Carrier Protein 2 Deficiency)	SCP2
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome-MEGDEL (Other names: 3-Methylglutaconic Aciduria Type VI-MGCA6)	SERAC1
Mucopolysaccharidosis type IIIA (Sanfilippo A) (Other names: MPS IIIA, Sanfilippo Syndrome A, Heparan Sulfate Sulfatase Deficiency, Sulfamidase Deficiency)	SGSH
Hyperinsulinemic hypoglycemia, familial, 7-HHF7 (Other names: Hyperinsulinemic Hypoglycemia Exercise-Induced)	SLC16A1
Sialic acid storage disorder Infantile (Other names: Sialuria Infantile Form, N-Acetylneuraminic Acid Storage Disease, NANA Storage Disease-NSD)	SLC17A5
Salla disease (Other names: Sialuria Finnish Type)	

Systemic primary carnitine deficiency (Other names: Systemic Carnitine Deficiency-SCD, Systemic Carnitine Deficiency due to Defect in Renal Reabsorption of Carnitine, Carnitine Deficiency Primary, Deficiency of Carnitine Transporter Plasma-membrane, Carnitine Uptake Defect-CUD)	SLC22A5
Citrullinemia, adult-onset type II (Other names: Citrin Deficiency)	SLC25A13
Citrullinemia type II neonatal-onset (Other names: Citrullinemia Type II Neonatal-onset With or Without Failure to Thrive and Dyslipidemia, Cholestasis Neonatal Intrahepatic caused by Citrin Deficiency-NICCD)	
Hyperammonemia-hyperornithinemia-homocitrullinuria (HHH) syndrome (Other nameS: HHH Syndrome, Ornithine Translocase Deficiency)	SLC25A15
Carnitine-acylcarnitine translocase (CACT) deficiency (Other names: CACT Deficiency)	SLC25A20
Fanconi-Bickel syndrome-FBS (Other names: Hepatorenal Glycogenosis with Renal Falconi Syndrome, Hepatic Glycogenosis with Fanconi Nephropathy, Hepatic Glycogenosis with Amino Aciduria and Glucosuria, FANCONI Syndrome with Intestinal Malabsorption and Galactose Intorelance, Pseudo-phlorizin Diabetes, Glycogenosis Fanconi Type, Glycogen Storage Disease XI)	SLC2A2
Congenital disorder of glycosylation, type IIf- CDG IIf	SLC35A1
Congenital disorder of glycosylation, type IIm (Other names: CDG IIm, Developmental and Epileptic Encephalopathy 22-DEE22, Epileptic Encephalopathy Early Infantile 22-EIEE22)	SLC35A2
Congenital disorder of glycosylation, type IIc-CDG IIc (Other names: Leukocyte Adhesion Deficiency Type II-LAD2, Rambam-Hasharon Syndrome-RHS)	SLC35C1
Congenital disorder of glycosylation type IIw	SLC37A4
Glycogen storage disease Ib - GSD Ib (Other names: Glucose-6-Phosphate Transport Defect)	
Glycogen storage disease Ic - GSD Ic	
Congenital disorder of glycosylation, type IIn- CDG IIn	SLC39A8
Cerebral creatine deficiency syndrome 1-CCDS1 (Other names: Creatine Deficiency Syndrome X-Linked, Creatine Transporter Defect, Mental Retardation X-Linked with seizures, short stature and midface hypoplasia, Mental Retardation X-Linked with Creatine Transport Deficiency)	SLC6A8
Glycine encephalopathy with normal serum glycine	SLC6A9
Niemann Pick type A (Other names: Sphingomyelin Lipidosis, Shingomyelinase Deficiency, Acid Sphingomyelinase Deficiency Neurovisceral Type, ASMD Neurovisceral Type)	SMPD1
Niemann Pick type B (Acid Sphingomyealinase Deficiency Visceral Type, ASMD Visceral Type)	
Congenital disorder of glycosylation, type Iq (Other names: Coloboma Ocular with Ichthyosis brain malformations and Endocrine Abnormalities, CDG Iq)	SRD5A3

Kahrizi syndrome (Other names: Mental Retardation Cataract,Coloboma and Kyphosis Autosomal Recessive)	SRD5A3
Congenital disorder of glycosylation, type Iy- CDG IY	SSR4
Congenital disorder of glycosylation, type Iw	STT3A
Congenital disorder of glycosylation, type Iw	
Congenital disorder of glycosylation, type Ix-CDG Ix	STT3B
Mitochondrial DNA depletion syncdrome 5 (Other names: Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with or without Methylmalonic Aciduria Autosomal Recessive SUCLA2-Related)	SUCLA2
Mitochondrial DNA depletion syndrome 9 (Other names: Lactic Acidosis Fatal Infantile Formerly)	SUCLG1
Multiple sulfatase deficiency (Other names: Mucosulfatidosis, Sulfatidosis Juvenile Austin Type)	SUMF1
Barth Syndrome (Other names: Cardioskeletal Myopathy with Neutropenia and Abnormal Mitochondria, 3-Methylgutaconic Aciduria Type II-MGCA2, MGA Type II)	TAFAZZIN
3-methylglutaconic aciduria, type IX (MGCA9)	TIMM50
Congenital disorder of glycosylation, type IIk-CDK IIk	TMEM165
Congenital disorder of glycosylation, type IIp- CDK IIp	TMEM199
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 (MC5DN2) (Other names: TMEM70 Type Encephalocardiomyopathy Mitochrondrial Neonatal due to ATP Synthase Deficiency)	TMEM70
Neuronal ceroid lipofuscinosis 2 (CLN2) (Other names: Ceroid Lipofuscinosis Neuronal 2 Variable Age at Onset, Jansky-Bielschowsky Disease)	TPP1
Spinocerebellar ataxia, autosomal recessive 7	
Intellectual developmental disorder, autosomal recessive 7-MRT7 (Other names: Intellectual Developmental Disorder 22-MRT22, Mental Retardation Autosomal Recessive 7-MRT7, Mental Retardation Autosomal Recessive 22)	TUSC3
Mucopolysaccharidosis-plus syndrome	VPS33A

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