Genes classified under different Inborn Errors of Metabolism (IEM) classes and Subclasses under different IEM terms [Source : The Monarch Initative Database]

Genes have variants present in ClinIndb
Genes have variants absent in ClinIndb

Gene	IEM class	Disease
AASS	AA_metabolism	hyperlysinemia (disease) / saccharopinuria
ABAT	AA_metabolism	GABA aminotransferase deficiency
ABCC6	AA_metabolism	arterial calcification, generalized, of infancy, 2
ABCC8	Carbohydrate Metabolism	hyperinsulinemic hypoglycemia, familial, 1
ABCD3	AA_metabolism	congenital bile acid synthesis defect 5
ABCD4	AA_metabolism	methylmalonic acidemia with homocystinuria, type cblJ
ACAD8	AA_metabolism	isobutyryl-CoA dehydrogenase deficiency
ACAD9	AA_Energy_metabolism	acyl-CoA dehydrogenase 9 deficiency
ACADL	AA_Energy_metabolism	long chain acyl-CoA dehydrogenase deficiency
ACADM	AA_Energy_metabolism	medium chain acyl-CoA dehydrogenase deficiency
ACADS	AA_Energy_metabolism	short chain acyl-CoA dehydrogenase deficiency
ACADSB	AA_metabolism	2-methylbutyryl-CoA dehydrogenase deficiency
ACADVL	Energy_metabolism	very long chain acyl-CoA dehydrogenase deficiency
ACAN	Carbohydrate Metabolism	spondyloepiphyseal dysplasia, Kimberley type
ACAT1	AA_Energy_metabolism	beta-ketothiolase deficiency
ACO2	AA_metabolism	infantile cerebellar-retinal degeneration
ACOX2	AA_metabolism	congenital bile acid synthesis defect 6
ACSF3	AA_metabolism	combined malonic and methylmalonic acidemia
ACY1	AA_metabolism	neurological conditions associated with aminoacylase 1 deficiency
ADK	AA_metabolism	hypermethioninemia due to adenosine kinase deficiency
ADSL	AA_metabolism	adenylosuccinate lyase deficiency
AGA	Carbohydrate Metabolism	aspartylglucosaminuria
AGL	Carbohydrate Metabolism	glycogen storage disease III
AGXT	Carbohydrate Metabolism	primary hyperoxaluria type 1
AHCY	AA_metabolism	hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase

AKR1D1	AA_metabolism	congenital bile acid synthesis defect 2
ALDH18A1	AA_metabolism	ALDH18A1-related de Barsy syndrome
ALDH4A1	AA_metabolism	hyperprolinemia type 2
ALDH5A1	AA_metabolism	succinic semialdehyde dehydrogenase deficiency
ALDH6A1	AA_metabolism	developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency
ALDOA	AA_Carbohydrate_Metabolism	glycogen storage disease due to aldolase A deficiency
ALDOB	Carbohydrate Metabolism	hereditary fructose intolerance
ALG1	Carbohydrate Metabolism	ALG1-CDG
ALG11	Carbohydrate Metabolism	ALG11-CDG
ALG12	Carbohydrate Metabolism	ALG12-CDG
ALG13	Carbohydrate Metabolism	ALG13-CDG
ALG14	Carbohydrate Metabolism	congenital myasthenic syndromes with glycosylation defect
ALG2	Carbohydrate Metabolism	ALG2-CDG
ALG3	Carbohydrate Metabolism	ALG3-CDG
ALG6	Carbohydrate Metabolism	congenital disorder of glycosylation type 1C
ALG8	Carbohydrate Metabolism	ALG8-CDG
ALG9	Carbohydrate Metabolism	ALG9-CDG
AMACR	AA_metabolism	congenital bile acid synthesis defect 4
AMT	AA_metabolism	glycine encephalopathy
ANO5	Carbohydrate Metabolism	autosomal recessive limb-girdle muscular dystrophy type 2L
AP3B1	AA_metabolism	Hermansky-Pudlak syndrome 2
AP3D1	AA_metabolism	Hermansky-Pudlak syndrome 10; HPS10
APRT	AA_metabolism	adenine phosphoribosyltransferase deficiency
ARG1	AA_metabolism	hyperargininemia
ARSB	Carbohydrate Metabolism	mucopolysaccharidosis type 6
ASL	AA_metabolism	argininosuccinic aciduria
ASNS	AA_metabolism	congenital microcephaly - severe encephalopathy - progressive cerebral atrophy syndrome
ASPA	AA_metabolism	Canavan disease
ASS1	AA_metabolism	citrullinemia type I
ATP6V0A2	AA_Carbohydrate_Metabolism	autosomal recessive cutis laxa type 2, classic type,ALG9-CDG
ATP6V1A	AA_Carbohydrate_Metabolism	autosomal recessive cutis laxa type 2d
ATP6V1E1	AA_Carbohydrate_Metabolism	autosomal recessive cutis laxa type 2c
AUH	AA_metabolism	3-methylglutaconic aciduria type 1
B3GALNT2	Carbohydrate Metabolism	muscle-eye-brain disease

B3GALT6	Carbohydrate Metabolism	spondyloepimetaphyseal dysplasia with joint laxity
B3GAT3	Carbohydrate Metabolism	Larsen-like syndrome, B3GAT3 type
B3GLCT	Carbohydrate Metabolism	Peters plus syndrome
B4GALT1	Carbohydrate Metabolism	B4GALT1-CDG
B4GALT7	Carbohydrate Metabolism	Ehlers-Danlos syndrome progeroid type
B4GAT1	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13
BAAT	AA_metabolism	familial hypercholanemia
BCKDHA	AA_metabolism	maple syrup urine disease
BCKDHB	AA_metabolism	maple syrup urine disease
BCKDK	AA_metabolism	branched-chain keto acid dehydrogenase kinase deficiency
BLOC1S3	AA_metabolism	Hermansky-Pudlak syndrome 8
BLOC1S6	AA_metabolism	Hermansky-Pudlak syndrome 9
BOLA3	AA_metabolism	multiple mitochondrial dysfunctions syndrome 2
BPGM	AA_Carbohydrate_Metabolism	hemolytic anemia due to diphosphoglycerate mutase deficiency
BTD	AA_Carbohydrate_Metabolism	biotinidase deficiency
CA5A	AA_Carbohydrate_Metabolism	hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency
CAD	Carbohydrate Metabolism	CAD-CDG
CBS	AA_metabolism	classic homocystinuria
CCDC115	Carbohydrate Metabolism	CCDC115-CDG
CD320	AA_metabolism	methylmalonic acidemia due to transcobalamin receptor defect
CHST14	Carbohydrate Metabolism	Ehlers-Danlos syndrome, musculocontractural type
CHST3	Carbohydrate Metabolism	spondyloepiphyseal dysplasia with congenital joint dislocations
CHSY1	Carbohydrate Metabolism	temtamy preaxial brachydactyly syndrome
CLDN19	AA_metabolism	renal hypomagnesemia 5 with ocular involvement
CLPB	AA_metabolism	3-methylglutaconic aciduria with cataracts, neurologic involvement and neutropenia
COG1	Carbohydrate Metabolism	COG1-CDG
COG2	Carbohydrate Metabolism	COG2-CDG
COG4	Carbohydrate Metabolism	COG4-CDG
COG5	Carbohydrate Metabolism	COG5-CDG
COG6	Carbohydrate Metabolism	COG6-CGD
COG7	Carbohydrate Metabolism	COG7-CDG
COG8	Carbohydrate Metabolism	COG8-CDG
COL2A1	Carbohydrate Metabolism	spondyloepiphyseal dysplasia congenita

COL4A1	Carbohydrate Metabolism	Walker-Warburg syndrome
CPS1	AA_metabolism	carbamoyl phosphate synthetase I deficiency disease
CPT1A	AA_Energy_metabolism	carnitine palmitoyl transferase 1A deficiency
CPT2	Energy_metabolism	carnitine palmitoyl transferase II deficiency, severe infantile form
CRPPA	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 7
CTH	AA_metabolism	cystathioninuria (disease)
CTSA	Carbohydrate Metabolism	galactosialidosis
CYP27A1	AA_metabolism	cerebrotendinous xanthomatosis
CYP7A1	AA_metabolism	hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency
CYP7B1	AA_metabolism	congenital bile acid synthesis defect 3
D2HGDH	AA_metabolism	D-2-hydroxyglutaric aciduria
DAG1	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A9
DBT	AA_metabolism	maple syrup urine disease
DCXR	Carbohydrate Metabolism	pentosuria
DDOST	Carbohydrate Metabolism	DDOST-CDG
DHFR	AA_metabolism	constitutional megaloblastic anemia with severe neurologic disease
DHTKD1	AA_metabolism	2-aminoadipic 2-oxoadipic aciduria
DLAT	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase E2 deficiency
DLD	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase E3 deficiency
DLL3	Carbohydrate Metabolism	autosomal recessive spondylocostal dysostosis
DMGDH	AA_metabolism	dimethylglycine dehydrogenase deficiency
DNAJC12	AA_metabolism	hyperphenylalaninemia, mild, non-bh4-deficient
DNAJC19	AA_metabolism	3-methylglutaconic aciduria type 5
DOLK	Carbohydrate Metabolism	DK1-CDG
DPAGT1	Carbohydrate Metabolism	DPAGT1-CDG
DPM1	Carbohydrate Metabolism	congenital disorder of glycosylation type 1E
DPM2	Carbohydrate Metabolism	congenital muscular dystrophy with intellectual disability and severe epilepsy
DPM3	Carbohydrate Metabolism	DPM3-CDG
DSE	Carbohydrate Metabolism	Ehlers-Danlos syndrome, musculocontractural type
DTNBP1	AA_metabolism	Hermansky-Pudlak syndrome 7
DUOX2	Thyroid metabolism	familial thyroid dyshormonogenesis

DUOXA2	Thyroid metabolism	familial thyroid dyshormonogenesis
ENO3	Carbohydrate Metabolism	glycogen storage disease due to muscle beta-enolase deficiency
EPHX1	AA_metabolism	familial hypercholanemia
ETFA	AA_Energy_metabolism	multiple acyl-CoA dehydrogenase deficiency
ETFB	AA_Energy_metabolism	multiple acyl-CoA dehydrogenase deficiency, severe neonatal type
ETFDH	AA_Energy_metabolism	multiple acyl-CoA dehydrogenase deficiency
EXT1	Carbohydrate Metabolism	exostoses, multiple, type 1
FAH	AA_metabolism	tyrosinemia type I
FBP1	Carbohydrate Metabolism	fructose-1,6-bisphosphatase deficiency
FGF23	Carbohydrate Metabolism	hyperphosphatemic familial tumoral calcinosis
FH	AA_metabolism	fumaric aciduria
FKRP	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A5
FKTN	Carbohydrate Metabolism	congenital muscular dystrophy without intellectual disability
FLAD1	AA_Energy_metabolism	multiple acyl-CoA dehydrogenase deficiency
FMO3	AA_metabolism	severe primary trimethylaminuria
FTCD	AA_metabolism	formiminoglutamic aciduria
FUCA1	Carbohydrate Metabolism	fucosidosis
FUT8	Carbohydrate Metabolism	congenital disorder of glycosylation with defective fucosylation
G6PC	Carbohydrate Metabolism	glycogen storage disease due to glucose-6-phosphatase deficiency type IA
G6PD	AA_Carbohydrate_Metabolism	class I glucose-6-phosphate dehydrogenase deficiency
GAA	Carbohydrate Metabolism	glycogen storage disease II
GALE	Carbohydrate Metabolism	galactose epimerase deficiency
GALK1	Carbohydrate Metabolism	galactokinase deficiency
GALNS	Carbohydrate Metabolism	mucopolysaccharidosis type 4
GALNT3	Carbohydrate Metabolism	hyperphosphatemic familial tumoral calcinosis
GALT	Carbohydrate Metabolism	classic galactosemia
GAMT	AA_metabolism	guanidinoacetate methyltransferase deficiency
GATM	AA_metabolism	AGAT deficiency
GBE1	Carbohydrate Metabolism	glycogen storage disease due to glycogen branching enzyme deficiency
GCDH	AA_metabolism	glutaryl-CoA dehydrogenase deficiency
GCH1	AA_metabolism	GTP-cyclohydrolase I deficiency

GCK	AA_Carbohydrate_Metabolism	hyperinsulinism due to glucokinase deficiency
GCLC	AA_metabolism	gamma-glutamylcysteine synthetase deficiency
GCSH	AA_metabolism	glycine encephalopathy
GFPT1	Carbohydrate Metabolism	congenital myasthenic syndrome 12
GGT1	AA_metabolism	gamma-glutamyl transpeptidase deficiency
GK	Carbohydrate Metabolism	inborn glycerol kinase deficiency
GLB1	Carbohydrate Metabolism	mucopolysaccharidosis type 4B
GLDC	AA_metabolism	glycine encephalopathy
GLRX5	AA_metabolism	spasticity-ataxia-gait anomalies syndrome
GLUD1	AA_Carbohydrate_Metabolism	hyperinsulinism-hyperammonemia syndrome
GLUL	AA_metabolism	congenital brain dysgenesis due to glutamine synthetase deficiency
GLYCTK	Carbohydrate Metabolism	D-glyceric aciduria
GMPPB	Carbohydrate Metabolism	congenital muscular dystrophy with cerebellar involvement
GNE	Carbohydrate Metabolism	GNE myopathy
GNMT	AA_metabolism	glycine N-methyltransferase deficiency
GNPTAB	Carbohydrate Metabolism	Sanfilippo syndrome type A
GNS	Carbohydrate Metabolism	Sanfilippo syndrome type D
GPHN	AA_metabolism	sulfite oxidase deficiency due to molybdenum cofactor deficiency type C
GPI	AA_Carbohydrate_Metabolism	hemolytic anemia due to glucophosphate isomerase deficiency
GPR143	AA_metabolism	X-linked recessive ocular albinism
GRHPR	Carbohydrate Metabolism	primary hyperoxaluria type 2
GRID2	AA_metabolism	autosomal recessive spinocerebellar ataxia 18
GRM1	AA_metabolism	autosomal recessive spinocerebellar ataxia 13
GSR	AA_metabolism	hemolytic anemia due to glutathione reductase deficiency
GSS	AA_metabolism	glutathione synthetase deficiency with 5-oxoprolinuria
GUSB	Carbohydrate Metabolism	mucopolysaccharidosis type 6,7
GYG1	Carbohydrate Metabolism	glycogen storage disease XV
GYS1	Carbohydrate Metabolism	glycogen storage disease due to muscle and heart glycogen synthase deficiency
GYS2	Carbohydrate Metabolism	glycogen storage disease due to hepatic glycogen synthase deficiency
HADH	AA_Carbohydrate_Energy_Metabolism	hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency

HADHA	AA_Energy_metabolism	long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHB	AA_Energy_metabolism	mitochondrial trifunctional protein deficiency
HCFC1	AA_metabolism	methylmalonic acidemia with homocystinuria, type cblX
HES7	Carbohydrate Metabolism	autosomal recessive spondylocostal dysostosis
HGD	AA_metabolism	alkaptonuria
HGSNAT	Carbohydrate Metabolism	Sanfilippo syndrome type C
HIBCH	AA_metabolism	neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
HK1	AA_Carbohydrate_Metabolism	non-spherocytic hemolytic anemia due to hexokinase deficiency
HLCS	AA_Carbohydrate_Metabolism	holocarboxylase synthetase deficiency
HMGCL	AA_Energy_metabolism	3-hydroxy-3-methylglutaric aciduria
HMGCS2	AA_Energy_metabolism	3-hydroxy-3-methylglutaryl-CoA synthase deficiency
HOGA1	Carbohydrate Metabolism	primary hyperoxaluria type 3
HPD	AA_metabolism	hawkinsinuria
HPS1	AA_metabolism	Hermansky-Pudlak syndrome 1,3
HPS3	AA_metabolism	Hermansky-Pudlak syndrome 3
HPS4	AA_metabolism	Hermansky-Pudlak syndrome 4
HPS5	AA_metabolism	Hermansky-Pudlak syndrome 5
HPS6	AA_metabolism	Hermansky-Pudlak syndrome 6
HSD17B10	AA_metabolism	HSD10 disease
HSD3B7	AA_metabolism	congenital bile acid synthesis defect 1
HTRA2	AA_metabolism	3-methylglutaconic aciduria, type VIII; MGCA8
HYAL1	Carbohydrate Metabolism	mucopolysaccharidosis type 9
IBA57	AA_metabolism	multiple mitochondrial dysfunctions syndrome 3
IDH2	AA_metabolism	D-2-hydroxyglutaric aciduria
IDS	Carbohydrate Metabolism	mucopolysaccharidosis type 2
IDUA	Carbohydrate Metabolism	Hurler syndrome
INPPL1	Carbohydrate Metabolism	schneckenbecken dysplasia
INSR	Carbohydrate Metabolism	hyperinsulinism due to INSR deficiency
ISCA1	AA_metabolism	multiple mitochondrial dysfunctions syndrome 5
ISCA2	AA_metabolism	multiple mitochondrial dysfunctions syndrome 4
IVD	AA_metabolism	isovaleric acidemia
IYD	Thyroid metabolism	familial thyroid dyshormonogenesis
KCNJ11	Carbohydrate Metabolism	hyperinsulinemic hypoglycemia, familial, 2
KCTD7	Carbohydrate Metabolism	progressive myoclonic epilepsy type 3

KHK	Carbohydrate Metabolism	essential fructosuria
KIF22	Carbohydrate Metabolism	spondyloepimetaphyseal dysplasia with multiple dislocations
KL	Carbohydrate Metabolism	tumoral calcinosis, hyperphosphatemic, familial, 3
KRT5	Carbohydrate Metabolism	Dowling-Degos disease
KYNU	AA_metabolism	encephalopathy due to hydroxykynureninuria
L2HGDH	AA_metabolism	L-2-hydroxyglutaric aciduria
LAMP2	Carbohydrate Metabolism	Danon disease
LARGE1	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A6
LCT	Carbohydrate Metabolism	congenital lactase deficiency
LDHA	Carbohydrate Metabolism	glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
LDHB	Carbohydrate Metabolism	glycogen storage disease due to lactate dehydrogenase H-subunit deficiency
LFNG	Carbohydrate Metabolism	autosomal recessive spondylocostal dysostosis
LIAS	AA_Carbohydrate_Metabolism	lipoic acid synthetase deficiency
LIPT1	AA_metabolism	lipoyl transferase 1 deficiency
LMBRD1	AA_metabolism	methylmalonic aciduria and homocystinuria type cblF
LRMDA	AA_metabolism	oculocutaneous albinism type 7
LYST	AA_metabolism	Chediak-Higashi syndrome
MAN1B1	Carbohydrate Metabolism	MAN1B1-CDG
MAN2B1	Carbohydrate Metabolism	alpha-mannosidosis
MANBA	Carbohydrate Metabolism	beta-mannosidosis
MAOA	AA_metabolism	Brunner syndrome
MAT1A	AA_metabolism	brain demyelination due to methionine adenosyltransferase deficiency
MCCC1	AA_metabolism	3-methylcrotonyl-CoA carboxylase deficiency
MCCC2	AA_metabolism	3-methylcrotonyl-CoA carboxylase deficiency
MCEE	AA_metabolism	methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
MCM6	Carbohydrate Metabolism	lactose intolerance (disease),spondylocostal dysostosis 2, autosomal recessive
MESP2	Carbohydrate Metabolism	spondylocostal dysostosis 2, autosomal recessive
MGAT2	Carbohydrate Metabolism	MGAT2-CDG
MICOS13	AA_metabolism	3-methylglutaconic aciduria type 3

MITF	AA_metabolism	ocular albinism with congenital sensorineural deafness
MLPH	AA_metabolism	Griscelli syndrome type 3
MLYCD	Energy_metabolism	malonic aciduria
MMAA	AA_metabolism	vitamin B12-responsive methylmalonic acidemia type cblA
MMAB	AA_metabolism	vitamin B12-responsive methylmalonic acidemia type cblB
MMACHC	AA_metabolism	methylmalonic aciduria and homocystinuria type cblC
MMUT	AA_metabolism	vitamin B12-unresponsive methylmalonic acidemia type mut0
MOCS1	AA_metabolism	sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
MOCS2	AA_metabolism	sulfite oxidase deficiency due to molybdenum cofactor deficiency type A,B
MOGS	Carbohydrate Metabolism	MOGS-CDG
MPC1	AA_metabolism	mitochondrial pyruvate carrier deficiency
MPDU1	Carbohydrate Metabolism	MPDU1-CDG
MPI	Carbohydrate Metabolism	MPI-CDG
MTHFR	AA_metabolism	homocystinuria due to methylene tetrahydrofolate reductase deficiency
MTR	AA_metabolism	methylcobalamin deficiency type cblG
MTRR	AA_metabolism	methylcobalamin deficiency type cblE
MT-TL1	AA_metabolism	3-methylglutaconic aciduria type 1
MYO5A	AA_metabolism	Griscelli syndrome type 1,3
NAGA	Carbohydrate Metabolism	alpha-N-acetylgalactosaminidase deficiency type 1,2,3
NAGLU	Carbohydrate Metabolism	Sanfilippo syndrome type B
NAGS	AA_metabolism	hyperammonemia due to N-acetylglutamate synthase deficiency
NEU1	Carbohydrate Metabolism	sialidosis type 2
NFU1	AA_metabolism	multiple mitochondrial dysfunctions syndrome 1
NGLY1	Carbohydrate Metabolism	NGLY1-deficiency
NKX2-5	Thyroid metabolism	hypothyroidism, congenital, nongoitrous, 5
NUS1	Carbohydrate Metabolism	congenital disorder of glycosylation, type IAA; CDG1AA
OAT	AA_metabolism	gyrate atrophy
OCA2	AA_metabolism	oculocutaneous albinism type 2
OCRL	AA_metabolism	oculocerebrorenal syndrome
OGDH	AA_metabolism	oxoglutaricaciduria
OPA3	AA_metabolism	3-methylglutaconic aciduria type 3
OPLAH	AA_metabolism	5-oxoprolinase deficiency (disease)

OTC	AA_metabolism	ornithine carbamoyltransferase deficiency
OXCT1	Energy_metabolism	succinyl-CoA:3-ketoacid CoA transferase deficiency
P4HA1	Carbohydrate Metabolism	Ehlers-Danlos syndrome, musculocontractural type
PAH	AA_metabolism	phenylketonuria
PAX8	Thyroid metabolism	thyroid hypoplasia
PC	Carbohydrate Metabolism	pyruvate carboxylase deficiency disease
PCBD1	AA_metabolism	dehydratase deficiency
PCCA	AA_metabolism	propionic acidemia
PCCB	AA_metabolism	propionic acidemia
PCK1	Carbohydrate Metabolism	phosphoenolpyruvate carboxykinase deficiency
PCK2	Carbohydrate Metabolism	phosphoenolpyruvate carboxykinase deficiency
PDHA1	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase E1-alpha deficiency
PDHB	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase E1-beta deficiency
PDHX	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase E3-binding protein deficiency
PDP1	AA_Carbohydrate_Metabolism	pyruvate dehydrogenase phosphatase deficiency
PEPD	AA_metabolism	prolidase deficiency
PFKM	AA_Carbohydrate_Metabolism	glycogen storage disease VII
PGAM2	Carbohydrate Metabolism	glycogen storage disease due to phosphoglycerate mutase deficiency
PGAP2	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PGAP3	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PGK1	AA_Carbohydrate_Metabolism	glycogen storage disease due to phosphoglycerate kinase 1 deficiency
PGM1	Carbohydrate Metabolism	PGM1-CDG
PGM3	Carbohydrate Metabolism	PGM3-CDG
PHGDH	AA_metabolism	PHGDH deficiency
PHKA1	Carbohydrate Metabolism	glycogen storage disease IXd
PHKA2	Carbohydrate Metabolism	glycogen storage disease IXa
PHKB	Carbohydrate Metabolism	glycogen storage disease IXb
PHKG2	Carbohydrate Metabolism	glycogen storage disease IXc
PIGA	Carbohydrate Metabolism	multiple congenital anomalies-hypotonia-seizures syndrome 2
PIGG	Carbohydrate Metabolism	intellectual disability, autosomal recessive 53
PIGL	Carbohydrate Metabolism	CHIME syndrome,hyperphosphatasia-intellectual disability syndrome

PIGM	Carbohydrate Metabolism	hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
PIGN	Carbohydrate Metabolism	multiple congenital anomalies-hypotonia-seizures syndrome 1
PIGO	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PIGT	Carbohydrate Metabolism	multiple congenital anomalies-hypotonia-seizures syndrome 3
PIGV	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PIGW	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PIGY	Carbohydrate Metabolism	hyperphosphatasia-intellectual disability syndrome
PKLR	AA_Carbohydrate_Metabolism	pyruvate kinase deficiency of red cells
PMM2	Carbohydrate Metabolism	PMM2-CDG
PMPCB	AA_metabolism	multiple mitochondrial dysfunctions syndrome 6
POFUT1	Carbohydrate Metabolism	Dowling-Degos disease 2, autosomal recessive limb-girdle muscular dystrophy type 2Z
POMGNT1	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3
POMGNT2	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 8
POMK	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 12
POMT1	Carbohydrate Metabolism	congenital muscular dystrophy without intellectual disability
POMT2	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2
PPM1K	AA_metabolism	intermediate maple syrup urine disease
PRDX1	AA_metabolism	methylmalonic aciduria and homocystinuria type cblC
PRKAG2	Carbohydrate Metabolism	lethal congenital glycogen storage disease of heart
PRODH	AA_metabolism	hyperprolinemia type 1
PRRT2	Carbohydrate Metabolism	childhood onset GLUT1 deficiency syndrome 2
PSAT1	AA_metabolism	PSAT deficiency
PSENEN	Carbohydrate Metabolism	Dowling-Degos disease
PSPH	AA_metabolism	PSPH deficiency
PTS	AA_metabolism	BH4-deficient hyperphenylalaninemia A
PYCR1	AA_metabolism	autosomal recessive cutis laxa type 2B

PYGL	Carbohydrate Metabolism	glycogen storage disease VI
PYGM	Carbohydrate Metabolism	glycogen storage disease V
QDPR	AA_metabolism	dihydropteridine reductase deficiency
RAB27A	AA_Carbohydrate_Metabolism	Griscelli syndrome type 2
RBCK1	Carbohydrate Metabolism	polyglucosan body myopathy type 1,glycogen storage disease due to glycogen branching enzyme deficiency
RFT1	Carbohydrate Metabolism	RFT1-CDG
RIPPLY2	Carbohydrate Metabolism	autosomal recessive spondylocostal dysostosis
RPIA	Carbohydrate Metabolism	ribose-5-P isomerase deficiency
RXYLT1	Carbohydrate Metabolism	muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 10
SARDH	AA_metabolism	sarcosinemia
SEC23B	Carbohydrate Metabolism	congenital dyserythropoietic anemia type 2
SERAC1	AA_metabolism	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SGSH	Carbohydrate Metabolism	Sanfilippo syndrome type A
SHPK	Carbohydrate Metabolism	isolated sedoheptulokinase deficiency
SI	Carbohydrate Metabolism	congenital sucrase-isomaltase deficiency
SLC16A1	Energy_Carbohydrate_metabolism	ketoacidosis due to monocarboxylate transporter-1 deficiency,exercise-induced hyperinsulinism
SLC16A12	Carbohydrate Metabolism	juvenile cataract-microcornea-renal glucosuria syndrome
SLC1A1	AA_metabolism	dicarboxylic aminoaciduria
SLC1A4	AA_metabolism	spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
SLC22A5	AA_Energy_metabolism	systemic primary carnitine deficiency disease
SLC24A5	AA_metabolism	oculocutaneous albinism type 6
SLC25A1	AA_metabolism	D,L-2-hydroxyglutaric aciduria
SLC25A13	AA_metabolism	citrullinemia, type II, adult-onset
SLC25A15	AA_metabolism	ornithine translocase deficiency
SLC25A20	AA_Energy_metabolism	carnitine-acylcarnitine translocase deficiency
SLC25A32	AA_Energy_metabolism	multiple acyl-CoA dehydrogenase deficiency, mild type
SLC26A4	Thyroid metabolism	thyroid hypoplasia
SLC2A1	Carbohydrate Metabolism	encephalopathy due to GLUT1 deficiency

SLC2A2	Carbohydrate Metabolism	glycogen storage disease due to GLUT2 deficiency
SLC35A1	Carbohydrate Metabolism	SLC35A1-CDG
SLC35A2	Carbohydrate Metabolism	SLC35A2-CDG
SLC35A3	Carbohydrate Metabolism	autism spectrum disorder - epilepsy - arthrogryposis syndrome
SLC35C1	Carbohydrate Metabolism	leukocyte adhesion deficiency type II
SLC35D1	Carbohydrate Metabolism	schneckenbecken dysplasia
SLC36A2	AA_metabolism	iminoglycinuria
SLC37A4	Carbohydrate Metabolism	glycogen storage disease IC
SLC39A8	Carbohydrate Metabolism	SLC39A8-CDG
SLC3A1	AA_metabolism	cystinuria (disease)
SLC45A2	AA_metabolism	oculocutaneous albinism type 4
SLC46A1	AA_metabolism	hereditary folate malabsorption
SLC5A1	Carbohydrate Metabolism	glucose-galactose malabsorption
SLC5A2	Carbohydrate Metabolism	familial renal glucosuria
SLC5A5	Thyroid metabolism	familial thyroid dyshormonogenesis
SLC6A19	AA_metabolism	iminoglycinuria
SLC6A20	AA_metabolism	iminoglycinuria
SLC6A8	AA_metabolism	creatine transporter deficiency
SLC6A9	AA_metabolism	atypical glycine encephalopathy
SLC7A7	AA_metabolism	lysinuric protein intolerance
SLC7A9	AA_metabolism	cystinuria (disease)
SPR	AA_metabolism	dopa-responsive dystonia due to sepiapterin reductase deficiency
SQSTM1	Carbohydrate Metabolism	myopathy, distal, with rimmed vacuoles; DMRV
SRD5A3	Carbohydrate Metabolism	SRD5A3-CDG
SSR4	Carbohydrate Metabolism	SSR4-CDG
ST3GAL5	Carbohydrate Metabolism	salt and pepper syndrome
STT3A	Carbohydrate Metabolism	STT3A-CDG
STT3B	Carbohydrate Metabolism	STT3B-CDG
SUOX	AA_metabolism	isolated sulfite oxidase deficiency
TALDO1	Carbohydrate Metabolism	transaldolase deficiency
TAT	AA_metabolism	tyrosinemia type II
TAZ	AA_metabolism	Barth syndrome
TDO2	AA_metabolism	familial hypertryptophanemia
TG	Thyroid metabolism	familial thyroid dyshormonogenesis
TH	AA_metabolism	TH-deficient dopa-responsive dystonia
THRB	Thyroid metabolism	thyroid hormone resistance, generalized, autosomal dominant, recessive
TIMM50	AA_metabolism	3-methylglutaconic aciduria type 9
TJP2	AA_metabolism	familial hypercholanemia
TKT	Carbohydrate Metabolism	transketolase deficiency
TMEM165	Carbohydrate Metabolism	TMEM165-CDG
TMEM199	Carbohydrate Metabolism	TMEM199-CDG
TPI1	AA_Carbohydrate_Metabolism	triosephosphate isomerase deficiency

TPO	Thyroid metabolism	familial thyroid dyshormonogenesis
TRAPPC2	Carbohydrate Metabolism	spondyloepiphyseal dysplasia tarda, X-linked
TREH	Carbohydrate Metabolism	diarrhea-vomiting due to trehalase deficiency
TRPV4	Carbohydrate Metabolism	spondyloepiphyseal dysplasia, Maroteaux type
TSHR	Thyroid metabolism	thyroid hypoplasia
TYR	AA_metabolism	oculocutaneous albinism type 1A,B
TYRP1	AA_metabolism	oculocutaneous albinism type 3
UROC1	AA_metabolism	urocanic aciduria (disease)
VPS33A	Carbohydrate Metabolism	mucopolysaccharidosis-like syndrome with congenital heart defects and hematopoietic disorders
XYLT1	Carbohydrate Metabolism	XYLT1-CDG
POGLUT1	Carbohydrate Metabolism	autosomal recessive limb-girdle muscular dystrophy type 2Z
EXT2	Carbohydrate Metabolism	hereditary multiple osteochondromas
MMADHC	AA_metabolism	methylcobalamin deficiency type cblDv1