Name	Hepatomegaly	Elevated transaminases	ALF	Cholestasis	Steatosis/fatty liver	Fibrosis/cirrhosis	Liver tumor	Other	Diagnostic markers	Specific treatment	Representative references (PMID or DOI)
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS											
Disorders of ammonia detoxification											
N-acetylglutamate synthase deficiency					X				Ammonia (B), Urea (P), Amino acids (P)	Protein restriction, ammonia scavengers (carglumic acid), citrulline	28900784, 11131349
Carbamoylphosphate synthetase I deficiency		X	X		X	X			Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P)	Protein restriction, ammonia scavengers, citrulline, liver transplant	28900784
Ornithine transcarbamylase deficiency	X	X	X	X		X	HCC		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	28900784, 22129577, 24485820, 21884343, 27070778, 24485820, 28887792
Argininosuccinate synthetase deficiency	X		X		X	X	HCC		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, arginine, liver transplant	28900784, 29209134, 15334737
Argininosuccinate lyase deficiency	X	**X**	**X**			X	HCC		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P, U), Orotic acid (U)	Protein restriction, ammonia scavengers, arginine, liver transplant	28900784
Arginase deficiency	X		X	X	X	X	HCC		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers	28900784, 22964440
Mitochondrial ornithine transporter deficiency		X	X	X	X	X			Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P, U), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline	28900784
Citrin deficiency		X		**X**	**X**	**X**	**HCC**		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U), Galactose (RBC)	MCT oil, lactose-free formula, fat-soluble vitamins, arginine, sodium pyruvate	28900784, 30591617, 18385606, 14606711
Disorders of amino acid transport											
Lysinuric protein intolerance	X	X			X	X			Ammonia (B), Amino acids (P, U)	Protein restriction, ammonia scavengers, citrulline	28057010
Disorders of tyrosine metabolism											
Hawkinsinuria		X							Organic acids (U)		10412819
Tyrosinemia type I	X	X	**X**		X	X	**HCC**		AST/ALT (P), Amino acids (P), Organic acids (U), Succinylacetone (P, U), AFP (S)	Tyrosine restriction, NTBC, liver transplant	6188953
Disorders of sulfur amino acid and sulfide metabolism											
Glycine N-methyltransferase deficiency	X	X							AST/ALT (P), Amino acids (P), Homocysteine (P), SAH/SAM (P)		11596649, 14739680
S-adenosylhomocysteine hydrolase deficiency		X					HCC		AST/ALT (P), CK (P), Coagulation factors (P), Amino acids (P), Homocysteine (P), SAH/SAM (P)	Methionine restriction, creatine, phosphatidylcholine, liver transplant	26527160, 26095522
Adenosine kinase deficiency		X		X	X	X			AST/ALT (P), Glucose (S), Amino acids (P), Purines (U), Total/direct bilirubin (S)	Methionine restriction	26642971
Disorders of branched-chain amino acid metabolism											
Dihydrolipoamide dehydrogenase deficiency	X		**X**						AST/ALT (P), NH3 (P), Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)	Protein restriction?, carnitine supplementation	9161958, 9764998, 23478190
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	X	X			X				AST/ALT (P), NH3 (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)	Protein restriction, carnitine supplementation	2443756, 9658458, 11593134, 27284350
Methylmalonate semialdehyde dehydrogenase deficiency		X	X						AST/ALT (P), Organic acids (U), Lactate (P)		DOI:10.32598/jpr.7.1.55, 21863277
Propionic acidemia	X								Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P)	Low protein diet, carnitine	29433791
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	X					X			Amino acids (P), Organic acids (U), MMA (S), Acylcarnitines (DBS, P)	Low protein diet, carnitine, hydroxycobalamin	29433791
Combined malonic and methylmalonic aciduria	X	X							Organic acids (U), MMA (S)		21841779, 26915364
Disorders of proline and ornithine metabolism											
Prolidase deficiency	X	X							Amino acids (U)		27385964, 16470701
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS											
Disorders of cobalamin metabolism											
cblC disease	X	X		X					Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)	Hydroxycobalamin, betaine	30178268
Disorders of riboflavin metabolism											
Multiple acyl-CoA dehydrogenase deficiency type 1	X				**X**				AST/ALT (P), CK (P), Glucose (S), Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P)		10052728, 7173260
Disorder of pyridoxine metabolism											
PNPO deficiency		X				X			5HIAA/HVA/3OMD (CSF), Amino acids (P)	PLP	25256445, 26108646
Disorders of copper metabolism											
Wilson disease		X	X	X	X	X	HCC, ICC		Copper (S, U), Ceruloplasmin (S), ASAT/ALAT (P), Blood count	Zinc, penicillamine, trientine	26897948, 25369181
MEDNIK syndrome		X		X					AST/ALT (P), Copper (S, U), Ceruloplasmin (S), VLCFA (P)	Copper histidine	23423674
Disorders of iron metabolism											
Hereditary hemochromatosis type 1						X	HCC		AST/ALT (P), Glucose (S), Total/direct bilirubin (S), Iron (S), Ferritin (S)	Phlebotomy	29404472
Hemojuvelin deficiency						X			AST/ALT (P), Glucose (S), Total/direct bilirubin (S), Iron (S), Ferritin (S)	Phlebotomy	12060140
Hepcidin deficiency						X			AST/ALT (P), Glucose (S), Total/direct bilirubin (S), Iron (S), Ferritin (S)	Phlebotomy	12469120
Transferrin receptor 2 deficiency						X			AST/ALT (P), Glucose (S), Total/direct bilirubin (S), Iron (S), Ferritin (S)	Phlebotomy	12060140
Ferroportin deficiency						X			AST/ALT (P), Glucose (S), Total/direct bilirubin (S), Iron (S), Ferritin (S)	Phlebotomy	20691492
Aceruloplasminemia		X							Iron (brain, S), copper (S), Ceruloplasmin (S), Ferritin (S), Blood count	Iron chelation	24002824
Disorders of manganese metabolism											
SLC30A10 deficiency	X	X		X	X	**X**			ASAT/ALAT (P), Blood count, Manganese (B)	Chelation (EDTA), iron supplementation	22341972, 22341971
DISORDERS OF CARBOHYDRATES											
Disorders of carbohydrate transport and absorption											
Fanconi-Bickel syndrome	**X**				X	X	HCC		AST/ALT (P), Glucose (S), Amino acids (U), Urinalysis, Lipid panel (S), Oligosaccharide (U), Galactose (RBC)	Corn starch, electrolyte replacement	9809815, 28382841
Disorders of galactose metabolism											
Classic galactosemia	X		X	X	X	X	HA		AST/ALT (P), Galactose-1-P (RBC), GALT enzyme activity (RBC)	Galactose restriction	14843794, 13914022, 24273939, 12983108
Uridine diphosphate galactose-4-epimerase deficiency	X	X	X	X					AST/ALT (P), Glucose (S), Amino acids (P), Total/direct bilirubin (S), Oligosaccharide (U)	Galactose restriction	10086948, 28247339
Disorders of fructose metabolism											
Hereditary fructose intolerance	X		X	X	X	X			AST/ALT (P), Glucose (S), Coagulation factors (P), Total/direct bilirubin (S), Urinalysis, Lipid panel (S), Oligosaccharide (U), Sialotransferins (S)	Fructose restriction	14304516, 655145, 738900
Disorders of the pentose phosphate pathway and polyol metabolism											
Transaldolase deficiency	**X**	**X**	X		X	X	HCC		AST/ALT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Feritin (S), Polyol (U)		24097415, 21119539, 15877206, 23315216, 29721915
Glycogen storage diseases											
Glycogen storage disease type Ib	**X**				X	X	HA, HCC		AST/ALT (P), Glucose (S), Blood count	Corn starch, G-CSF	12373567, 21481415
Glycogen storage disease type III	**X**	**X**				X	HA, HCC		AST/ALT (P), CK (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S), Oligosaccharide (U), Enzyme (DBS, L, F)	Corn starch, high-protein diet	17196294, 10914784, 26697579
Glycogen storage disease type IV	**X**	X				**X**	HCC		AST/ALT (P), Coagulation factors (P), Total/direct bilirubin (S), Enzyme (DBS, L, F)	Corn starch, liver transplant	13279125
Glycogen storage disease type VI	**X**	X			X	X	HA		AST/ALT (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S)	Corn starch	25266922
Glycogen storage disease type IXa	**X**	X			X				AST/ALT (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S)	Corn starch	25266922
Glycogen storage disease type IXb	**X**	X			X				AST/ALT (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S)	Corn starch	25266922
Glycogen storage disease type IXc	**X**	X			X	**X**	HA		AST/ALT (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S)	Corn starch	25266922, 28627441
Disorders of gluconeogenesis											
Glycogen storage disease type I a	X				X	X	HA, HCC		AST/ALT (P), Glucose (S), Urinalysis, Lactate (P), Lipid panel (S), Uric acid (P)	Corn starch	12373567, 21481415
Fructose-1,6-bisphosphatase deficiency		X	X						AST/ALT (P), Glucose (S), Amino acids (P), Organic acids (U), Urinalysis, Lactate (P)		29203193
Cytosolic phosphoenolpyruvate carboxykinase deficiency									AST/ALT (P), Glucose (S), Amino acids (P), Organic acids (U)		26971250, 28216384
MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM											
Disorders of the Krebs cycle											
GTP-specific succinyl-CoA ligase α subunit deficiency	X	X			X				AST/ALT (P), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)		26475597
Disorders of mitochondrial carriers											
Cytosolic glycerol-3-phosphate dehydrogenase deficiency	X	X			X	X			AST/ALT (P), Organic acids (U), Lipid panel (S)		24549054, 22226083, 27368975
Disorders of complex I assembly											
Acyl-CoA dehydrogenase 9 deficiency		X	X						AST/ALT (P), CK (P), Glucose (S), Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)		17564966
Disorders of complex III assembly											
BCS1L deficiency		X		X					Lactate (P), Iron (S)		11528392, 12215968
Disorders of complex IV assembly and ancillary proteins											
SCO1 deficiency	X		X		X				AST/ALT (P), Lactate (P)		11013136
Disorders of complex V assembly											
Transmembrane protein 70 deficiency	X								AST/ALT (P), CK (P), Amino acids (P), Organic acids (U), Lactate (P)		25326274
Disorders of mitochondrial DNA depletion											
DNA polymerase γ catalytic subunit (POLG1) deficiency		X	X		X	X			AST/ALT (P), Organic acids (U), Lactate (P)		23419467
DNA polymerase γ accessory subunit (POLG2) deficiency			X		X				AST/ALT (P), Organic acids (U), Lactate (P)		27592148
Mitochondrial deoxyguanosine kinase deficiency	X	X	X	X	X	X	HCC		AST/ALT (P), Glucose (S), AFP (S), Total/direct bilirubin (S), Lactate (P)		18205204, 16908739
MPV17 deficiency	X	**X**	**X**	**X**	X	X	HCC		AST/ALT (P), Glucose (S), AFP (S), Total/direct bilirubin (S), Lactate (P)		29282788
TWINKLE mitochondrial DNA helicase deficiency		X	X	X	X	X			AST/ALT (P), Glucose (S), AFP (S), Total/direct bilirubin (S), Lactate (P)		21681116, 30391088
Disorders of mitochondrial transcription and RNA transcript processing											
TRMU deficiency	X		**X**						AST/ALT (P), Coagulation factors (P)		19732863
Disorders of mitochondrial translation factors											
Mitochondrial elongation factor G1 (GFM1) deficiency		X	X						AST/ALT (P), Lactate (P)		15537906
Mitochondrial elongation factor Tu deficiency		X							AST/ALT (P), Lactate (P)		17160893
Mitochondrial elongation factor Ts deficiency			X	X							21741925
Disorders of mitochondrial tRNA											
Mitochondrial tRNA(Glu) deficiency		X							AST/ALT (P), CK (P), Lactate (P)		

Name	Hepatomegaly	Elevated transaminases	ALF	Cholestasis	Steatosis/fatty liver	Fibrosis/cirrhosis	Liver tumor	Other	Diagnostic markers	Specific treatment	Representative references (PMID or DOI)
Disorders of mitochondrial tRNA incorporation and recycling											
Mitochondrial glutamyl-tRNA synthetase deficiency	X	X			X				AST/ALT (P), Lactate (P)		23008233
Mitochondrial phenylalanyl-tRNA synthetase deficiency		X							AST/ALT (P), Lactate (P)		28043061
Mitochondrial tryptophanyl-tRNA synthetase deficiency			X						No laboratoty tests --> DNA		29783990
Disorders of mitochondrial phospholipid metabolism											
MEGDEL Syndrome	X	X	**X**	X	X	X			Organic acids (U), Lactate (P)		23918762, 29205472
DISORDERS OF LIPIDS											
Disorders of carnitine metabolism											
Carnitine transporter deficiency		X							AST/ALT (P), CK (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Carnitine supplementation	9916797
Carnitine palmitoyltransferase 1 deficiency	X	X			X				AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Frequent feeds,low-fat diet, MCT supplementation	11441142
Carnitine palmitoyltransferase 2 deficiency	X	X			X				AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Frequent feeds, low-fat diet, MCT supplementation	8682496, 1961225, 1999498
Carnitine acylcarnitine translocase deficiency	X	X			X				AST/ALT (P), CK (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)	Frequent feeds, low-fat diet, MCT supplementation	9544911, 7564255, 7807931
Disorders of fatty acid oxidation and transport											
Medium-chain acyl CoA dehydrogenase deficiency	X	X			X				AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Avoidance of fasting	18045290, 2279505, 3793003
Very long-chain acyl CoA dehydrogenase deficiency	X	X			X				AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Frequent feeds, low-fat diet, MCT supplementation	3793003, 10077518
Long-chain hydroxyacyl-CoA dehydrogenase deficiency	X	X		X	X				AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Frequent feeds, low-fat diet, MCT supplementation	10518281, 9003853, 2308028
Mitochondrial trifunctional protein deficiency	X	X			X				AST/ALT (P), NH3 (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)	Frequent feeds, low-fat diet, MCT supplementation	18045290
Disorders of ketone body metabolism											
3-Hydroxy-3-methylglutaryl-CoA synthase deficiency		X							AST/ALT (P), Glucose (S), Organic acids (U), Acylcarnitines (DBS, P)	Avoidance of fasting	11479731
Mitochondrial acetoacetyl-CoA thiolase deficiency	X	X							Organic acids (U), Acylcarnitines (DBS, P), Acetoacetate (P, U), Glucose (B)	Avoidance of fasting	15877211, 28689740
Disorders of intracellular triglyceride metabolism											
Chanarin-Dorfman syndrome		X			X				ASAT/ALAT (P)		8938558, 21994851
Disorders of lipoprotein metabolism											
Apolipoprotein B deficiency (familial hypobetalipoproteinemia)					X	X			Lipid panel (S), Vitamins A and E (P), Apo B (P)		24842304
Abetalipoproteinemia					X	X	HCC		Lipid panel (S), Vitamins A and E (P), Apo B (P)		24842304
Disorders of cholesterol biosynthesis											
Mevalonate kinase deficiency	X	X		X		X			Organic acids (U), Leukotrienes (U)		DOI: 10.2174/1874309900903010045, 9714005, 21548022, 21399979, 25469482, 22405037
Smith-Lemli-Opitz syndrome		X		X		X			Lipid panel (S), Sterols (P)	Cholesterol supplementation	15580635
Lathosterolosis		X		X		X			Lipid panel (S), Sterols (P)	Liver transplant?	15580635, 24621408
Disorders of bile acid synthesis											
3β-Hydroxy-Δ5-C27-steroid dehydrogenase deficiency	X	X		**X**		X			AST/ALT (P), ALP (S), GGT (S), Total/direct bilirubin (S), Lipid panel (S), Bile acids (S, U), Vitamins A, D, E, K (S)	Chenodeoxycholic acid	3470305, 7915305, 17645593
Δ4-3-Oxosteroid-5β-reductase deficiency	X	X		**X**	X				AST/ALT (P), ALP (S), GGT (S), Total/direct bilirubin (S), Bile acids (S, U)	Chenodeoxycholic and cholic acids, liver transplant	3198770, 8301429, 8707100
Cerebrotendinous xanthomatosis				**X**					Sterols (P), Bile acids (S, U)	Chenodeoxycholic acid	12555943, 28937538, 15795599
Oxysterol 7α-hydroxylase deficiency	X	X		X		X			AST/ALT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Lipid panel (S), Bile acids (S, U), Vitamins A, D, E, K (S)	Chenodeoxycholic acid, liver transplant	9802883, 30546280
α-Methylacyl-CoA racemase deficiency				X					Pristanic acid (S)	Cholic acid	12512044
PMP70 deficiency				X		X			AST/ALT (P), Bile acids (S, U)		25168382
Bile acid-CoA ligase deficiency		X		X		X			AST/ALT (P), GGT (S), Total/direct bilirubin (S), Bile acids (S, U)		22089923, 22783059
DISORDERS OF TETRAPYRROLES											
Disorders of heme metabolism											
Acute intermittent porphyria		X				X	**HCC**, CC		AST/ALT (P), Porphobilinogen (P, U), aminolevulinic acid (P, U), porphyrins (P, U, feces), enzyme (RBC)	Hemin	30413387, 8918510, 23344888
Hereditary coproporphyria		X					HCC		AST/ALT (P), Porphyrins (P, U, feces)	Hemin	23344888
Porphyria variegata		X					HCC		AST/ALT (P), Porphobilinogen (P, U), aminolevulinic acid (P, U), porphyrins (P, U, feces), enzyme (B)	Hemin	30413387, 23344888
Ferrochelatase deficiency		X		X		X			AST/ALT (P), Protoporphyrins (B, RBC)	Sun avoidance, afamelanotide, β-carotene, liver transplant	20857522, 17360790
δ-aminolevulinic acid synthase superactivity		X		X		X			AST/ALT (P), Protoporphyrins (B, RBC)	Sun avoidance, iron?, afamelanotide, β-carotene, liver transplant, BMT	18760763, 25856424, 26193873
Heme oxygenase 1 deficiency	X								Blood count, Total/direct bilirubin (S), haptoglobin (S), Ferritin (S), Lipid panel (S), Urinalysis		9884342
Disorders of bilirubin metabolism and biliary transport											
Crigler-Najjar syndrome		X				X			Total/direct bilirubin (S)	Phototherapy, liver transplant	29176474, 28590786
ATP8B1 deficiency	X	X		**X**		X			AST/ALT (P), GGT (S), Bile acids (S), Vitamins A, D, E, K (S)	Ursodeoxycholic acid, partial external biliary diversion, liver transplant	30236549
ABCB11 deficiency	X	X		**X**		X	HCC, HB		AST/ALT (P), GGT (S), Bile acids (S), Vitamins A, D, E, K (S)	Ursodeoxycholic acid, partial external biliary diversion, liver transplant	30236549, 16871584
ABCB4 deficiency	X	X		**X**		X	CC		AST/ALT (P), GGT (S), Bile acids (S), Vitamins A, D, E, K (S)	Ursodeoxycholic acid, partial external biliary diversion, liver transplant	30236549, 26473142
Bile acid receptor deficiency		X		**X**		X			AST/ALT (P), GGT (S), Bile acids (S), Vitamins A, D, E, K (S)	Liver transplant	26888176
STORAGE DISORDERS											
Sphingolipidoses											
Gaucher disease	X					X	HCC		Blood count, Enzyme (DBS, L, F)	Enzyme replacement therapy	7450398, 27842801, 10787452, 29423829
Acid sphingomyelinase deficiency	X	X				X			AST/ALT (P), Blood count, Enzyme (DBS, L, F)		24718843, 1919942, 28228103
Metachromatic leukodystrophy							*	**	Enzyme (DBS, L, F), Sulfatide (U), Protein (CSF)	Bone marrow transplant	27261095, 9732500, 20446223, 26160127
Combined saposin deficiency	X								Enzyme (DBS, L, F)		2514102, 11309366, 19267410
Mucopolysaccharidoses											
Hurler syndrome	X					X			GAG (U), Enzyme (DBS, L, F)	Enzyme replacement therapy, bone marrow transplant	3096134
Hunter syndrome	X					X			GAG (U), Enzyme (DBS, L, F)	Enzyme replacement therapy	3096134
Sanfilippo disease	X	X				X			GAG (U), Enzyme (DBS, L, F)		3096134, 25439061
Disorders of lysosomal cholesterol metabolism											
Niemann-Pick disease type C	X	X		**X**		X	HCC		Enzyme (DBS, L, F), Filipin test (F)		12142809, 30737051, 2602232, 8599014, 14581809
Lysosomal acid lipase deficiency	X	X		X	X	X			AST/ALP (P), Lipid panel (S), Enzyme (DBS, L)		23403440
Disorders of lysosomal transport or sorting											
Nephropathic cystinosis				X				NRH	Glucose (S), Amino acids (P), Urinalysis		18466290, 16527704
DISORDERS OF PEROXISOMES											
Disorders of peroxisomal β-oxidation											
Peroxisomal acyl-CoA oxidase 1 deficiency	X								VLCFA (P)		17458872
D-bifunctional protein deficiency	X			X	X	X			VLCFA (P), Pristanic acid (S), Pristanic acid (S)		16278854
Disorders of peroxisomal biogenesis											
Zellweger spectrum disorders	X	X		X	X	X	HCC		VLCFA (P), Pristanic acid (S), Phytanic acid (S), ASAT/ALAT (P), Plasmalogens (RBC), Pipecolic acid (S, U)	Cholic acid	26453805, 27469511, 30057520
CONGENITAL DISORDERS OF GLYCOSYLATION											
Disorders of N-linked glycosylation											
Phosphomannomutase 2 deficiency	X	X				X			AST/ALT (P), Coagulation factors (P), Lipid panel (S), Sialotransferins (S), Enzyme (DBS, L, F)		28108845
Phosphomannose isomerase deficiency	X	X			X	X			AST/ALT (P), Glucose (S), Coagulation factors (P), Lipid panel (S), Sialotransferins (S), Enzyme (DBS, L, F)		28108845
ALG13-CDG	X								AST/ALT (P), Sialotransferins (S)		28777499
ALG1-CDG	X	X							AST/ALT (P), Sialotransferins (S)		28108845
RFT1-CDG	X								AST/ALT (P), CK (P), Coagulation factors (P), Sialotransferins (S)		18313027
ALG3-CDG	X	X			X	X			AST/ALT (P), Sialotransferins (S)		28108845
ALG6-CDG	X								Sialotransferins (S)		28108845
ALG8-CDG	X	X		X	X	X		***	Sialotransferins (S)		28108845
ALG9-CDG	X								AST/ALT (P), Coagulation factors (P), Lipid panel (S), Sialotransferins (S)		28108845
ALG8-CDG	X			X					AST/ALT (P), Sialotransferins (S)		28108845
MOGS-CDG	X								AST/ALT (P), Oligosaccharide (U), Sialotransferins (S)		10788335
GANAB-CDG								***	No laboratoty tests --> DNA		27259053
Disorders of dolichol metabolism											
DPM1-CDG	X								AST/ALT (P), CK (P), Sialotransferins (S)		10642602
Disorders of monosaccharide synthesis and interconversion											
Sialuria							ICC		Sialic acid (U)		27142465
Phosphoglucomutase 1 deficiency		**X**			X	X			AST/ALT (P), NH3 (P), CK (P), Coagulation factors (P), Sialotransferins (S)		28108845
Glycosylation disorders of vesicular trafficking											
COG2-CDG		X							AST/ALT (P), Sialotransferins (S)		24784932
COG4-CDG	X	X									28108845
COG5-CDG	X	X				X			AST/ALT (P), Sialotransferins (S)		28108845
COG6-CDG	X	X		X		X			AST/ALT (P), CK (P), Lactate (P), Sialotransferins (S), Vitamins A, D, E, K (S)		28108845
COG7-CDG	X	X		X		X			AST/ALT (P), CK (P), Glucose (S), Total/direct bilirubin (S), Sialotransferins (S)		28108845
TRAPPC11-CDG		X		X	X	X			AST/ALT (P), CK (P)		28827486, 26322222, 27862579, 29855340
Disorders of Golgi homeostasis											
ATP6AP1-CDG	X			X		X			AST/ALT (P), Total/direct bilirubin (S), Copper (S, U)		28108845
ATP6AP2-CDG	X	X			X	X			AST/ALT (P)		29127204
TMEM199-CDG		X				X			AST/ALT (P), ALP (S), Lipid panel (S), Sialotransferins (S)		28108845
CCDC115-CDG	X	X		X		X			AST/ALT (P), ALP (S), Lipid panel (S), Copper (S, U), Sialotransferins (S)		28108845
TMEM165-CDG	X								AST/ALT (P), CK (P), Sialotransferins (S)		22683087
Disorder of deglycosylation											
N-glycanase 1 deficiency		X									27388694