Supplemental Table S2. List of IMDs with immunological phenotypes, laboratory investigations, treatment options (if aplicable), OMIM references and IEMbase IDs.

Inherited metabolic disorder (n=171)	Gene	Autoimmunity	Immunodeficiency & Infections	Innate immune defects	Inflammation	Diagnostic laboratory markers	Specific treatment	OMIM Disease No.	IEMbase ID (hyperlinked)
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS (n=35)									
Disorders of pyrimidine metabolism									
Uridine monophosphate synthase deficiency (Orotic aciduria type I)	*UMPS*		X			Purines and pyrimidines (U)		258900	IEM0003
Pyrimidine 5'-nucleotidase superactivity	*NT5C3A*		X			Uric acid (U)	Uridine, possibly ribose	266120;197720	IEM0004
Activation-induced cytidine deaminase deficiency	*AICDA*		X			IgE (S), IgG (S), IgA (S), IgM (S)		605258	IEM0005
Uracil-DNA glycosylase deficiency	*UNG*		X			IgE (S), IgG (S), IgA (S), IgM (S)		608106	IEM0006
dUTP pyrophosphatase deficiency	*DUT*	X				DNA		601266	IEM1084
CTP synthase 1 deficiency	*CTPS1*		X			DNA		615897	IEM1272
Disorders of purine metabolism									
Phosphoribosyl pyrophosphate synthetase 1 deficiency	*PRPS1*		X			Uric acid (U, P), Purines and pyrimidines (U)	Dietary reduction of red and organ meets, fish; Allopurinol 5-10 mg/kg/day; S-adenosylmethionine 30	300661	IEM0008
Adenosine deaminase deficiency (SCID)	*ADA*		X			Purines and pyrimidines (U)	ERT, HSCT, gene therapy	102700	IEM0013
Purine nucleoside phosphorylase deficiency	*PNP*		X			Purines and pyrimidines (U)	Bone marrow transplant; possible phase I/II clinical trial for gene therapy (doi.org/10.14785/lymphosign-2018-	613179;164050	IEM0015
Hypoxanthine guanine phosphoribosyltransferase deficiency (Lesch-Nyhan syndrome)	*HPRT1*		X			Uric acid (U, P), Purines and pyrimidines (U)	Allopurinol	300322;308000	IEM0017
Adenylate kinase 2 deficiency	*AK2*		X	X		DNA		267500	IEM0020
FAMIN deficiency	*LACC1*		X		X	Blood count		618795	IEM1275
NUDT15 deficiency	*NUDT15*			X		DNA		615792	IEM1277
Disorders of nucleotide metabolism									
3' Repair exonuclease 1 deficiency	*TREX1*	X				ASAT/ALAT (P), Pterins (CSF)	JAK inhibitors (baricitinib, ruxolitinib)	192315	IEM0026
Ribonuclease H2 subunit A, B or C deficiency (Aicardi-Goutières syndrome type 2, 3 or 4)	*RNASEH2A*	X				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	610333	IEM0029
SAMHD1 deficiency (Aicardi-Goutières syndrome type 5)	*SAMHD1*	X				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	612952	IEM0031
RNA-specific adenosine deaminase deficiency (Aicardi-Goutières syndrome type 6)	*ADAR*	X				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF)	JAK inhibitors	127400	IEM0032
MDA5 superactivity	*IFIH1*	X	X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	182250	IEM0033
STING superactivity	*TMEM173*		X		X	CRP (P), Interferon-stimulated genes or interferon signature (PBMC), Erythrocyte sedimentation rate, IgG (S)	JAK inhibitors (tofacitinib)	615934	IEM0034
2',5'-Oligoadenylate synthetase 1 deficiency	*OAS1*		X	X		Immunoglobulins (B), Blood count		222100	IEM0035
Equilibrative nucleoside transporter 3 deficiency	*SLC29A3*			X	X	Erythrocyte sedimentation rate; IgG (S)		602782	IEM0041
Disorders of glutathione metabolism									
Glutathione synthetase deficiency, sever	*GSS*		X			Hemoglobin (B), Reticulocytes (B), Glutathione (RBC), 5-Oxoproline (U)	Na bicarbonate to treat metabolic acidosis, antioxidants (vitamin C, E), avoid drugs like acetylsalycylic acid, phenopbarbital, sulfonamides)	266130	IEM0050
NRF2 superactivity	*NFE2L2*		X			Amino acids (P), Creatinine (P)		617744	IEM0055
Disorders of amino acid transport									
Cystinuria type A	*SLC3A1*		X			Amino acids (P, U)		220100	IEM0068
Cystinuria type B	*SLC7A9*		X			Amino acids (P, U)		220100	IEM0069
Lysinuric protein intolerance	*SLC7A7*			X		Amino acids (P, U), Ammonia (P), Ferritin (S), Lipid panel (S), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline	222700	IEM0070
Disorders of branched-chain amino acid metabolism									
Isovaleric acidemia	*IVD*	X	X	X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P),	Protein restricted diet; Carnitine 50-100 mg/kg/day; Glycine 150-259 mg/kg/day	243500	IEM0113
Methylcrotonylglycinuria type 1	*MCCC1*		X			Organic acids (U), Acylcarnitines (DBS, P)	In symptomatic patients +/- protein restriction; +/- carnitine	210200	IEM0116
Methylcrotonylglycinuria type 2	*MCCC2*		X			Organic acids (U), Acylcarnitines (DBS, P)	In symptomatic patients +/- protein restriction; +/- biotin	210200	IEM0117
3-Hydroxyisobutyryl-CoA deacylase deficiency	*HIBCH*		X			Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P)	Moderate protein restricted diet	250620	IEM0120
Propionyl-CoA-carboxylase deficiency (Propionic acidemia PCCA (PCCB)	*PCCA; PCCB*		X			Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P)	Low protein diet, L-Carnitine 100-200 mg/kg/day, Metronidazole 10-20 mg/kg/day alternating 1-2 weeks, +/-NH3 scavengers (Carbaglu, Na benzoate), acute management of hyerammonemic crises	232000	IEM0124 IEM0125
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*		X			Amino acids (P), Organic acids (U), MMA (S), Acylcarnitines (DBS, P)	Low protein diet, L-Carnitine 100-200 mg/kg/day, vitamin B12, acute management of hyerammonemic crises, orthotopic liver transplant	251000	IEM0127
Malonic aciduria	*MLYCD*		X			Organic acids (U), Acylcarnitines (DBS, P), Lactate (P), Glucose (S), Lipid panel (S)	low fat, high carbohydrate diet, MCT oil, low dose L-carnitine	248360	IEM0129

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Inherited metabolic disorder (n=171)	Gene	Autoimmunity	Immunodeficiency & Infections	Innate immune defects	Inflammation	Diagnostic laboratory markers	Specific treatment	OMIM Disease No.	IEMbase ID (hyperlinked)
Disorders of lysine metabolism									
5-phosphohydroxylysine phospholyase	*PHYKPL*		X			5-Phosphohydroxylysine (U)		615011	IEM0136
Disorders of proline and ornithine metabolism									
Prolidase deficiency	*PEPD*		X			Amino acids (U)		170100;613230	IEM0144
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS (n=28)									
Disorders of cobalamin metabolism									
Intrinsic factor deficiency (TCN3 deficiency)	*CBLIF*	X		X		Homocysteine (P), Organic acids (U), Vitamin B12 (S)		261000	IEM0205
Cubilin deficiency	*CUBN*	X		X		Homocysteine (P), Organic acids (U), Vitamin B12 (S)		261100	IEM0206
Amnionless deficiency	*AMN*	X		X		Homocysteine (P), Organic acids (U), Vitamin B12 (S)	Hydroxycobalamin IM	261100	IEM0207
Transcobalamin (TC 2) deficiency	*TCN2*	X		X		Homocysteine (P), Organic acids (U), Vitamin B12 (S)		275350	IEM0209
Adenosylcobalamin and methylcobalamin synthesis defect - cblF	*LMBRD1*			X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Homocysteine (P), SAM/SAH (P), Blood count	Hydroxycobalamin 1 mg IM daily	277380	IEM0211
Adenosylcobalamin and methylcobalamin synthesis defect - cblJ	*ABCD4*			X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Homocysteine (P), SAM/SAH (P)	Hydroxycobalamin 1 mg IM daily	614857	IEM0212
Adenosylcobalamin and methylcobalamin synthesis defect - cblC	*MMACHC*			X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), SAM/SAH (P), Blood count	Hydroxycobalamin 1 mg IM daily, betaine 250 mg/kg/day; folinic acid 5-30 mg/day; L-carnitine 50-200 mg/kg/day; no protein restriction; maintain normal methionine levels (+/- supplementation)	277400	IEM0213
Methylmalonic aciduria and homocystinuria, cblC type, digenic	*MMACHC; PRDX1*			X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), SAM/SAH (P), Blood count	Hydroxycobalamin 1 mg IM daily	609831;176763	IEM0214
Adenosylcobalamin and methylcobalamin synthesis defect - cblD-MMA-HC	*MMADHC*			X		Homocysteine (P), Organic acids (U), Vitamin B12 (S)	Hydroxycobalamin, betaine	277410	IEM0215
Adenosylcobalamin synthesis defect - cblD-MMA	*MMADHC*	X				Homocysteine (P), Organic acids (U), Vitamin B12 (S)	Hydroxycobalamin, betaine	277410	IEM0215
Adenosylcobalamin synthesis defect - cbl A	*MMAA*	X				Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Ammonia (B), Anion gap	Hydroxycobalamin injections	251100	IEM0218
Adenosylcobalamin synthesis defect - cbl B	*MMAB*	X				Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Ammonia (B), Anion gap	Hydroxycobalamin injections	251110	IEM0219
Sodium-dependent multivitamin transporter deficiency	*SLC5A6*		X			ALP (P), IgG(S), 1,25-Dihydroxy vitamin D (S)		604024	IEM1124
Disorders of folate metabolism									
Proton-coupled folate transporter (PCFT) deficiency (Hereditary folate malabsorption)	*SLC46A1*	X	X			Folate (S), 5-Methyl-THF (CSF), Blood count, Immunoglobulins (S)		229050	IEM0221
5,10-Methylene-tetrahydrofolate dehydrogenase deficiency	*MTHFD1*		X			Homocysteine (P), Organic acids (U), Folate (S), 5-Methyl-THF (CSF)		172460	IEM0224
Dihydrofolate reductase deficiency	*DHFR*	X				5-Methyl-THF (CSF), Blood count	Folinic acid	126060	IEM0225
5,10-Methenyltetrahydrofolate synthetase deficiency	*MTHFS*		X			5-Methyl-THF (CSF)		604197	IEM1035
Disorders of thiamine metabolism									
Mitochondrial thiamine pyrophosphate carrier deficiency (Bilateral striatal necrosis)	*SLC25A19*		X			Organic acids (U), Lactate (CSF)		606521	IEM0232
Disorders of niacin and NAD metabolism									
NAD(P)HX dehydratase deficiency	*NAXD*	X				DNA		615910	IEM0244
Disorders of copper metabolism									
Wilson disease (ATPase copper transporting beta deficiency)	*ATP7B*			X		Copper (S, U), Ceruloplasmin (S)	Penicillamine 750-1500 mg/day in adults, (10 mg/kg/day in children) divided in 2-3 doses together with 25 mg pyridoxine ; trientine 900-2500 mg/day in adults divided in 2-3 doses; zinc sulphate 600 mg/day divided in 3 doses in adults	277900	IEM0279
Menkes disease (ATPase copper transporting alpha deficiency)	*ATP7A*		X			Copper (S, U), Ceruloplasmin (S)	Copper chloride or L-histidine 350-500 ug/day IV or SC	304150	IEM0280
Disorders of iron metabolism									
Atransferrinemia	*TF*		X			Transferrin (S), Iron (liver)		209300	IEM0294
Transferrin receptor deficiency	*TFRC*		X			B cells, circulating (B), IgG(S)		616740	IEM0295
Disorders of zinc metabolism									
Acrodermatitis enteropathica	*SLC39A4*		X			Zinc (S); ALP (P)	Zinc po 150-400 Zn sulfate/day (35-90 mg elemental Zn)	201100	IEM0297
Hyperzincemia and hypercalprotectinemia	*PSTPIP1*		X		X	DNA		604416	IEM1258
Disorders of manganese metabolism									
Solute carrier family 39 (Zn transporter) deficciency CDG2N-CDG	*SLC39A8*		X			Sialotransferins (S), Manganese (B, U)	Uridine + galactose, manganese	616721	IEM0303
Disorders of magnesium metabolism									

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Inherited metabolic disorder (n=171)	Gene	Autoimmunity	Immunodeficiency & Infections	Innate immune defects	Inflammation	Diagnostic laboratory markers	Specific treatment	OMIM Disease No.	IEMbase ID (hyperlinked)
Claudin 16 deficiency	*CLDN16*		X			Citric acid (U), Calcium (P, U), Magnesium (P, U), Uric acid (P), Parathyroid hormone, PTH (S)	Magnesium replacement	248250	IEM0309
Claudin 19 deficiency	*CLDN19*		X			Calcium (P, U), Magnesium (P, U)	Magnesium replacement	248190	IEM0310
DISORDERS OF CARBOHYDRATES (n=9)									
Disorders of galactose metabolism									
Galactose-1-phosphate uridyltransferase deficiency	*GALT*		X			AST/ALT (P), Galactose-1-P (RBC), GALT enzyme activity (rbc)	Galactose restriction	230400	IEM0322
Disorders of the pentose phosphate pathway and polyol metabolism									
Transaldolase deficiency	*TALDO1*			X		ASAT/ALAT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Ferritin (S), Polyol (U)		606003	IEM0329
Lysine specific methyltransferase 2D deficiency (Kabuki syndrome)	*KMT2D*		X			DNA		147920	IEM1492
Glycogen storage diseases									
Glucose-6-phosphate translocase deficiency	*SLC37A4*		X	X	X	ASAT/ALAT (P), Biotinidase (P), Triglyceride (S), Uric acid (P), Lactate, fasted (P, U), Blood count, 1,5-Anhydroglucitol-6-phosphate (P, U), Glycogen (liver)		232220	IEM0355
Lysosomal alpha-1,4-glucosidase deficiency (Pompe disease)	*GAA*			X		ASAT/ALAT (P), CK (P), Glycogen (muscle)	Alglucosidase alpha	232300	IEM0356
HOIL1 deficiency	*RBCK1*		X			CK (P); DNA		615895	IEM0365
LAMP2 deficiency (Danon disease)	*LAMP2*			X		ASAT/ALAT (P), CK (P), Glycogen (muscle)		300257	IEM0367
HOIL1 interacting protein deficiency	*RNF31*		X		X	DNA		612487	IEM1135
Disorders of glycolysis									
Triosephosphate isomerase deficiency	*TPI1*		X			Dihydroxyacetone phosphate (rbc)		615512	IEM0382
MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM (n=13)									
Disorders of the Krebs cycle									
Fumarase deficiency, tumoral phenotype	*FH*		X			Organic acids (U)		606812	IEM0402
Isocitrate dehydrogenase 1 deficiency	*IDH1*		X			Organic acids (U)		147700	IEM1090
Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication									
FBXL4 deficiency	*FBXL4*		X			Lactate (P), ASAT/ALAT (P), CK (P), Blood count		615471	IEM0502
Disorders of mitochondrial transcription and RNA transcript processing									
Mitochondrial RNA-processing endoribonuclease deficiency	*RMRP*		X			DNA		607095;250250;250460	IEM0515
Disorders of mitochondrial tRNA incorporation and recycling									
Mitochondrial seryl-tRNA synthetase deficiency	*SARS2*	X				Lactate (P), Uric acid (S)		613845	IEM0563
Mitochondrial valyl-tRNA synthetase deficiency	*VARS2*		X			ASAT/ALAT (P)		615917	IEM0565
Disorders of mitochondrial fission									
STAT2 deficiency	*STAT2*		X			Lactate (P, CSF)		616636	IEM0575
Disorders of mitochondrial fusion									
SPATA5 deficiency	*SPATA5*		X			DNA		616577	IEM1380
Disorders of mitochondrial phospholipid metabolism									
MEGDEL Syndrome	*SERAC1*		X			Lactate (P); Organic acids (U); Filipin staining		614739	IEM0582
Tafazzin deficiency (Barth syndrome)	*TAZ*		X			Organic acids (U), Lipid panel (S), Urinalysis, 3-Methylglutaconic acid (U), 3-Methylglutaric acid (U),	Elamipretide	302060	IEM0583
MICOS complex subunit MIC26 deficiency	*APOO*		X			Acylcarnitines (DBS, P), Lactate (P), Pyruvate (P)		300753	IEM1376
Disorders of mitochondrial protein quality control									
CLPB deficiency	*CLPB*		X			Organic acids (U), Blood count		616271	IEM0594
Disorders associated with single large-scale mtDNA deletions									
Pearson Syndrome	*multiple*			X		DNA		557000	IEM1501
DISORDERS OF LIPIDS (n=18)									
Disorders of fatty acid oxidation and transport									

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Fatty acid transport protein 4 deficiency	*SLC27A4*		X			DNA		608649	IEM0638
Disorders of cytoplasmic triglyceride metabolism									
Lipin 2 deficiency (Majeed syndrome)	*LPIN2*		X			DNA		609628	IEM0658
Diacylglycerol acyltransferase deficicncy	*DGAT1*		X			AST/ALT (P), Albumin (S), IgG(S)		615863	IEM0659
Abhydrolase D5 deficiency (Chanarin-Dorfman syndrome)	*ABHD5*			X		ASAT/ALAT (P)		275630	IEM0660
Adipose triglyceride lipase deficiency	*PNPLA2*			X		CK (P)		610717	IEM0661
Disorders of non-lysosomal sphingolipid metabolism									
CYP4F22 omega hydroxylase deficiency	*CYP4F22*		X			DNA		604777	IEM0679
Disorders of eicosanoid metabolism									
Thromboxane synthase deficiency	*TBXAS1*			X		DNA		231095	IEM0684
Disorders of phosphoinositide metabolism									
Catalytic phosphatidylinositol 3-kinase δ subunit superactivity	*PIK3CD*		X		X	IgG (S), IgM (S)		615513	IEM0699
Phosphatidylinositol 3-kinase regulatory subunit 1 deficiency (SHORT syndrome)	*PIK3R1*	X	X	X		IgG (S), IgM (S), IgA (S)		269880;616005	IEM0700
Phosphatidylinositol 3,4,5-trisphosphate 5-phosphatase deficiency	*INPPL1*		X			Phosphate (P)		258480	IEM0705
Phosphatidylinositol 4,5-bisphosphate phospholipase C γ2 deficiency	*PLCG2*	X	X	X		IgE (S)		614878	IEM0709
Disorders of lipoprotein metabolism									
Tangier disease (ABCA1)	*ABCA1*		X			Lipid panel (S)		600046;205400	IEM0735
Disorders of cholesterol biosynthesis									
Mevalonate kinase deficiency (Hyper Ig D syndrome)	*MVK*			X		DNA	Inflammatory control (non-steroidal anti-inflammatory drugs, corticosteroids, IL-1 targeting biologic agents, TNF-alpha blockade, allogenic stem cell	610377	IEM0740
Disorders of steroid metabolism									
ACTH receptor deficiencyMelanocortin-2 receptor deficiency	*MC2R*		X			ACTH, Steroids (P); Glucose (P)		202200	IEM0766
MIRAGE syndrome	*SAMD9*		X			Corticotropin (P), Cortisol (P), Renin Activity		617053	IEM1497
Disorders of bile acid synthesis									
3β-Hydroxy-Δ5-C27-steroid dehydrogenase-isomerase deficiency	*HSD3B7*				X	Bile acids (P,U), Bilirubin, conjugated (P), Vitamins D/E		607764	IEM0779
Δ4-3-Oxosteroid-5β-reductase deficiency	*AKR1D1*				X	Bile acids (P,U), Bilirubin, conjugated (P), ASAT/ALAT (P)		604741	IEM0780
Oxysterol 7α-hydroxylase deficiency	*CYP7B1*				X	Glucose (P), Bile acids (P,U), Bilirubin, total/direct (P), Vitamins E (P), ASAT/ALAT (P), AP (P), 27-	Chenodeoxycholic acid, liver transplant	603711	IEM0781
DISORDERS OF TETRAPYRROLES (n=2)									
Disorders of heme metabolism									
GATA1 deficiency	*GATA1*		X			Reticulocytes (B)			IEM0796
Disorders of bilirubin metabolism and biliary transport									
ABCB4 deficiency	*ABCB4*				X	AST/ALT (P), GGT (S), Bile acids (S), Vitamins A, D, E, K (S)		602347	IEM0807
STORAGE DISORDERS (n=15)									
Disorders of autophagy									
EPG5 deficiency (Vici syndrome)	*EPG5*		X			DNA		242840	IEM0811
Neuronal ceroid lipofuscinosis									
Lysosomal transmembrane CLN3 protein deficiency (Batten Spielmeyer-Vogt disease)	*CLN3*			X		Peripheral smear	In trial gene therapy (NCT03770572)	204200	IEM0821
Sphingolipidoses									
Acid beta-glucosidase deficiency (Gaucher disease)	*GBA*	X				Enzyme activity (WBC)	Enzyme replacement therapy, substrate reduction, bone marrow transplantation	230800	IEM0832
Saposin A and/or C deficiency	*PSAP*				X	Sulfatides (U); Protein (CSF)		610539	IEM0833
Sphingomyelinase deficiency (Niemann-Pick disease type A or B)	*SMPD1*	X	X			Enzyme activity (WBC)		257200;607616	IEM0834
Beta-galactosidase-1 deficiency (GM1-gangliosidosis)	*GLB1*			X		Oligosaccharide (U), Enzyme activity (WBC)	In trial gene therapy (NCT03952637)	230500	IEM0835

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Hexosaminidase A and B deficiency (Sandhoff disease)	*HEXB*			X		Enzyme activity (WBC)		268800	IEM0836
Galactocerebrosidase deficiency (Krabbe disease)	*GALC*				X	Enzyme (DBS, L, F), Protein (CSF)		245200	IEM0837
Acid ceramidase deficiency, inflammatory phenotype (Farber disease)	*ASAH1*				X	DNA		228000	IEM0845
Oligosaccharidoses									
Neuraminidase deficiency (Sialidosis)	*NEU1*			X		Enzyme activity (WBC), Oligosaccharides (U)		256550	IEM0848
Protective protein/cathepsin A deficiency (Goldberg syndrome)	*CTSA*			X		Enzyme activity (WBC), Oligosaccharides (U)		256540	IEM0849
Alpha-mannosidase B deficiency	*MAN2B1*		X	X		Oligosaccharides (U); Enzyme assay (DBS, L, F)	Recombinant enzyme replacement therapy (velmanase alfa); HCT	248500	IEM0850
Alpha-L-fucosidase deficiency	*FUCA1*			X		Enzyme activity (DBS,WBC), Fucose (U)	Bone marrow transplant	230000	IEM0853
Aspartylglucosaminidase deficiency	*AGA*			X		Enzyme activity (DBS,WBC), Aspartylglucosamine (U)		208400	IEM0854
Mucopolysaccharidoses									
Mucopolysaccharidosis-plus	*VPS33A*		X			Total GAGs (U), sHeparan sulfate (U)		617303	IEM0869
DISORDERS OF PEROXISOMES AND OXALATE (n=7)									
Disorders of plasmalogen synthesis									
Peroxisomal biogenesis factor 7 deficiency (RCDP Type 1)	*PEX7*		X			Plasmalogens (RBC)		215100	IEM0878
Dihydroxyacetone phosphate acyltransferase deficiency (RCDP Type 2)	*GNPAT*		X			Plasmalogens (RBC)		602744	IEM0879
Alkyldihydroxyacetonephosphate synthase deficiency (RCDP Type 3)	*AGPS*		X			Plasmalogens (RBC)		600121	IEM0880
Peroxisomal disorders not involving lipid metabolism									
Catalase deficiency (Acatalasemia)	*CAT*		X			DNA		614097	IEM0902
Disorders of oxalate metabolism									
D-glycerate dehydrogenase deficency (Primary hyperoxaluria type II)	*GRHPR*	X	X			Oxalic acid (U, P), Glyceric acid (U), Creatinine (P), Urea (P)		260000	IEM0905
4-hydroxy-2-oxoglutarate aldolase deficiency (Primary hyperoxaluria type III)	*HOGA1*	X	X			Oxalic acid (U, P), Organic acids (U)		613616	IEM0906
Alanine-glyoxylate aminotransferase deficiency deficiency (Primary hyperoxaluria type I)	*AGXT*	X	X			Oxalic acid (U, P), Glycolic acid (P, U), Creatinine (P), Urea (P)	Liver or liver/kidney transplant	259900	IEM0903
CONGENITAL DISORDERS OF GLYCOSYLATION (n=31)									
Disorders of N-linked glycosylation									
Mannosyltransferase 4-5 deficiency ALG11-CDG	*ALG11*			X		Albumin (S), Lactate (P), Factor XI (B), Sialotransferrins (S)		613661	IEM0916
Mannosyltransferase 8 deficiency ALG12-CDG	*ALG12*		X			ASAT/ALAT (P), Glucose (S), Lipid panel (S), Sialotransferrins (S), IGF (P)		607143	IEM0920
Magnesium transporter 1 deficiency MAGT1-CDG	*MAGT1*		X			Sialotransferrins (S)		300716;300853	IEM0925
Glucosidase 1 deficiency GCS1-CDG	*MOGS*		X			AST/ALT (P), Oligosaccharide (U), Sialotransferins (S)		606056	IEM0927
N-acetylglucosaminyltransferase 2 deficiency MGAT2-CDG	*MGAT2*		X	X		ASAT/ALAT (P), CK (P), Sialotransferrins (S), Factor IX and XII (B), Antithrombin III (P)		212066	IEM0931
Congenital disorder of glycosylation DDOST-CDG	*DDOST*		X			Sialotransferrins (S), Antithrombin (B), Factor XI (B), Proteins C and S (S)		614507;602202	IEM1192
Translocon associated protein complex subunit SSR3 deficiency SSR3-CDG	*SSR3*		X			DNA		606213	IEM1548
Core-specific lysosomal alpha-1,6-mannosidase deficieincy MAN2B2-CDG	*MAN2B2*		X			CRP (P), IgE (S), Erythrocyte sedimentation rate		618899	IEM1549
Disorders of O-xylosylation and glycosaminoglycan synthesis									
Exostosin-like glycosyltransferase 3 deficiency	*EXTL3*		X			DNA		617425	IEM0952
Disorders of O-GalNAcylation									
Core 1 β-1,3-galactosyltransferase chaperone deficiency	*C1GALT1C1*	X				Hemoglobin (B), Blood count		300622	IEM0964
Disorders of glycosylphosphatidylinositol biosynthesis									
Phosphatidylinositol glycan anchor biosynthesis class T deficiency PIGT-CDG	*PIGT*				X	ALP (P), GPI-anchored proteins (WBC, F)		615398	IEM0982
Disorders of glycolipid glycosylation									
Alpha-1,4-Galactosyltransferase deficiency A4GALT-CDG	*A4GALT*	X				DNA		111400	IEM0990
Disorders of dolichol metabolism									
Dolichol-P-mannose synthase-2 deficiency DPM2-CDG	*DPM2*		X			AST/ALT (P), CK (P), Sialotransferrins (S)		615042	IEM0996

Supplemental Table S2. List of IMDs with immunological phenotypes, laboratory investigations, treatment options (if aplicable), OMIM references and IEMbase IDs.

Inherited metabolic disorder (n=171)	Gene	Autoimmunity	Immunodeficiency & Infections	Innate immune defects	Inflammation	Diagnostic laboratory markers	Specific treatment	OMIM Disease No.	IEMbase ID (hyperlinked)
Disorders of monosaccharide synthesis and interconversion									
Phosphoglucomutase 3 deficiency PGM3-CDG	*PGM3*		X			Sialotransferins (S), IgE (S)		615816;172100	IEM1004
Glucose-6-phosphatase catalytic subunit 3 deficiency (Dursun syndrome)	*G6PC3*		X		X	Blood count		612541	IEM1005
Disorders of nucleotide-sugar synthesis									
UGP2-CDG	*UGP2*		X			DNA		618744	IEM1557
Disorders of Golgi transport									
Congenital disorder of glycosylation SLC35A2-CDG	*SLC35A2*		X			Sialotransferrins (S)		314375	IEM1009
GDP-fucose transporter deficiency SLC35C1-CDG	*SLC35C1*	X	X	X		Neutrophil motility/rolling (B)		266265	IEM1011
Disorders of vesicular trafficking									
Component of COG complex 6 deficiency COG6-CDG	*COG6*		X			AST/ALT (P), CK (P), Lactate (P), Sialotransferins (S), Vitamins A, D, E, K (S)		606977;614576	IEM1016
Component of COG complex 7 deficiency COG7-CDG	*COG7*		X			AST/ALT (P), CK (P), Glucose (S), Total/direct bilirubin (S), Sialotransferins (S)		608779	IEM1017
Component of COG complex 8 deficiency COG8-CDG	*COG8*		X			Sialotransferins (S), Apo-CIII (S)		611182	IEM1018
Jagunal 1 deficiency JAGN1-CDG	*JAGN1*		X			DNA		616022	IEM1019
Vacuolar protein sorting 13B deficiency VPS13B-CDG	*VPS13B*		X			Sialotransferrins (S)		216550	IEM1023
Coatomer protein complex subunit alpha deficiency	*COPA*	X				DNA		616414	IEM1403
Vacuolar protein sorting 45 homolog deficiency	*VPS45*		X			DNA		615285	IEM1413
Unc-13 homolog D deficiency (FHL Type 3)	*UNC13D*		X	X		AST/ALT (P), Pterins (U)		608898	IEM1419
Syntaxin 11 deficiency (FHL Type 4)	*STX11*			X		DNA		603552	IEM1420
Syntaxin-binding protein 2 deficiency (FHL Type 5)	*STXBP2*			X		DNA		613101	IEM1421
Disorders of Golgi homeostasis									
Accessory subunit 1 of the vacuolar-ATPase protein deficiency ATP6AP1-CDG	*ATP6AP1*		X	X		Sialotransferrins (S); Ceruloplasmin (S); Copper (S); IgG (S)		300972	IEM1027
ATPase H(+)-transporting lysosomal accessory protein 2 deficiency ATP6AP2-CDG	*ATP6AP2*		X			Sialotransferins (S), ASAT/ALAT (P); IgG (S); Factor XI (B)		300423	IEM1028
Transmembrane protein 165 deficiency TMEM165-CDG	*TMEM165*		X			AST/ALT (P), CK (P),Sialotransferins (S), Insulin (S)		614727;614726	IEM1029
OTHER (n=13)									
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases									
Threonyl-tRNA synthetase 1 deficiency	*TARS1*		X			DNA		618546	IEM1316
Autosomal recessive dyskeratosis congenita type 1	*NOLA3*	X				DNA		224230	IEM1335
Autosomal recessive dyskeratosis congenita type 2	*NOLA2*	X				DNA		613987	IEM1336
Cytosolic ribosomal SA deficiency	*RPSA*		X			DNA		271400	IEM1367
Shwachman-Diamond syndrome, DNAJC21 type	*DNAJC21*	X				DNA		617052	IEM1371
Shwachman-Diamond syndrome, EIF6 type	*EIF6*		X			AST/ALT (P), Vitamin A (P)		602912	IEM1372
Disorders of lysosome-related organelle biogenesis									
Adaptor-related protein complex 3 subunit beta 1 deficiency (Hermansky-Pudlak syndrome type 2)	*AP3B1*		X			DNA		608233	IEM1384
Biogenesis of lysosomal organelles complex 2 subunit 2 deficiency (Hermansky-Pudlak syndrome type 5)	*HPS5*		X			DNA		614074	IEM1387
Pallidin deficiency (Hermansky-Pudlak syndrome type 9)	*BLOC1S6*			X		DNA		614171	IEM1391
Adaptor-related protein complex 3 subunit delta 1 deficiency (Hermansky-Pudlak syndrome type 10)	*AP3D1*		X			DNA		617050	IEM1392
Lysosomal trafficking regulator deficiency (Chediak-Higashi syndrome)	*LYST*	X	X	X	X	DNA		214500	IEM1393
Myosin VA deficiency (Griscelli syndrome type 1)	*MYO5A*		X			DNA		214450	IEM1394
Ras-related protein Rab27A deficiency (Griscelli syndrome type 2)	*RAB27A*		X			DNA		607624	IEM1395