Supplementary Table S1. List of inherited metabolical diseases with hematologic phenotypes, laboratory investigation, treatment options (if applicable), OMIM references and IEMbase ID.

Disorder (n=264)	Gene	Inheritance	Abormal blood cell morphology	Coagulation abnormalities	Anemias	Abnormal blood count	Hypercoagulability	Marrow abnormality	Other	Diagnostic markers	Specific treatmnt	OMIM	IEMbase code
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS (n=25)													
Disorders of pyrimidine metabolism													
CAD trifunctional protein deficiency	*CAD*	AR	Anisocytosis Poikilocytosis		Anemia, dyserythropoietic					Purines and pyrimidines (U), Sialotransferrins (S)	Possibly uridine	616457	IEM0001
Uridine monophosphate synthase deficiency	*UMPS*	AR	Anisocytosis Poikilocytosis		Anemia, megaloblastic Hypochromia					Purines and pyrimidines (U)	Uridine	258900	IEM0003
Pyrimidine-5'-nucleotidase I deficiency	*NT5C3A*	AR	Basophilic stippling		Anemia, non-spherocytic, hemolytic with basophilic stippling					Uric acid (U), Glutathione (RBC)	Uridine, possibly ribose	266130;606224,191720	IEM0004
dUTP pyrophosphatase deficiency	*DUT*	AR				Pancytopenia		Bone marrow aplasia	Macrocytosis	DNA		601266	IEM1084
Disorders of purine metabolism													
Adenylate kinase 1 deficiency	*AK1*	AR	Basophilic stippling		Anemia, non-spherocytic, hemolytic with basophilic stippling					Adenylate kinase activity (Ec), DNA		612631	IEM0019
Adenylate kinase 2 deficiency	*AK2*	AR							Leukocytosis	DNA		267500	IEM0020
Adenosine deaminase superactivity	*ADA*	AR			Anemia, hemolytic	Lymphopenia				Purines (U)	ERT, HSCT, gene therapy	608958	IEM1274
NUDT15 deficiency	*NUDT15*	AD							Leukocytosis	DNA		615792	IEM1277
Disorders of glutathione metabolism													
Gamma-glutamylcysteine synthetase deficiency	*GCLC*	AR			Anemia, hemolytic					Hemoglobin (B), Reticulocytes (B), Glutathione (RBC)	Avoid drugs that precipitate hemolytic crisis in G6PD deficiency (phenobarbital, acetylsalyciclic acid, sulfonamides)	230450	IEM0049
Glutathione synthetase deficiency	*GSS*	AR			Anemia, hemolytic					5-Oxoproline (U), Hemoglobin (B), Reticulocytes (B), Glutathione (RBC), Lactate (P), blood gas	Na bicarbonate to treat metabolic acidosis, antioxidants (vitamin C, E), N-acetlycysteine, avoid drugs like acetylsalycylic acid, phenopbarbital, sulfonamides)	266130	IEM0050
Glutathione reductase deficiency	*GSR*	AR			Anemia, hemolytic					Glutathione (P), Bilirubin (P)		138300	IEM0053
Disorders of ammonia detoxification													
Ornithine transcarbamylase deficiency	*OTC*	LX		Coagulopathy						Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	311250	IEM0058
Arginase 1 deficiency	*ARG1*	AR		Coagulopathy						Ammonia (P); Amino acids (P); Orotic acid (U)	Protein restriction, ammonia scavengers	207800	IEM0061
Citrin deficiency	*SLC25A13*	AR		Coagulation, impaired	Anemia					Ammonia (P); Amino acids (P)	Low carbohydrate diet, MCT oil, ammonia scavengers, liver transplant	605814;603471	IEM0063
Disorders of amino acid transport													
Lysinuric protein intolerance	*SLC7A7*	AR						Hemophagocytosis		Amino acids (P, U), Ammonia (P), Ferritin (S), Lipid panel (S), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline	222700	IEM0070
Disorders of sulfur amino acid and sulfide metabolism													
S-adenosylhomocysteine hydrolase deficiency	*AHCY*	AR		Coagulopathy						SAM & SAH (P), Amino acids (P)	Methionine or protein restricted diet, phosphatidylcholine and creatine supplements, liver transplant	613752	IEM0100
Cystathionine beta-synthase deficiency	*CBS*	AR					Thromboembolism			SAM & SAH (P), Amino acids (P)	Pyridoxine; vitamin B12, folate, low protein diet, +/- cysteine; betaine; enzyme therapy in clinical trials	236200	IEM0102
Disorders of branched-chain amino acid metabolism													
Isovaleryl-CoA dehydrogenase deficiency	*IVD*	AR				Pancytopenia Thrombocytopenia Neutropenia			Leukocytosis	Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Urinalysis	Protein restricted diet;L- Carnitine; Glycine	243500	IEM0113
3-Methylglutaconyl-CoA hydratase deficiency	*AUH*	AR				Thrombocytopenia				Organic acids (U), Acylcamitines (P,DBS)	Protein restriction, L-carnitine	250950	IEM0118
Propionic acidemia due to propionyl-CoA carboxylase subunit alpha deficiency	*PCCA*	AR			Anemia	Neutropenia Thrombocytopenia		Hemophagocytosis		Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, L-Carnitine , acute management of hyerammonemic crises, orthotopic liver transplant	232000	IEM0124
Propionic acidemia due to propionyl-CoA carboxylase subunit beta deficiency	*PCCB*	AR			Anemia	Neutropenia Thrombocytopenia			Hemophagocytosis	Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, L-Carnitine , acute management of hyerammonemic crises, orthotopic liver transplant	232000	IEM0125
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*	AR			Anemia	Neutropenia Thrombocytopenia				Amino acids (P); Organic acids (U); Acylcamitines (U, P, DBS); Anion gap	Low protein diet, L-Carnitine , vitamin B12, acute management of hyerammonemic crises, orthotopic liver transplant	251000	IEM0127
Disorders of proline and ornithine metabolism													
Prolidase deficiency	*PEPD*	AR			Anemia	Thrombocytopenia				Amino acids (U)		170100;613230	IEM0144
Disorders of serine metabolism													
3-phosphoglycerate dehydrogenase deficiency	*PHGDH*	AR			Anemia, megaloblastic					5-Methyl-THF (CSF), Amino acids (P)	L-serine; Glycine	606879;601851	IEM0181
Disorders of glycine metabolism													
Mitochondrial glycine transporter deficiency	*SLC25A38*	AR			Anemia, sideroblastic Anemia, microcytic, hypochromic Anemia, microcytic					Ferritin (S)		205950	IEM0191
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS (n=41)													
Disorders of lipoic acid and iron-sulfur metabolism													
Glutaredoxin 5 deficiency	*GLRX5*	AR			Anemia, sideroblastic					Ferritin (S), Transferrin (S), Lactate (P), Amino acids (P)		205950	IEM0197
ABCB7 deficiency	*ABCB7*	XL						Ringed sideroblasts on bone marrow		Protoporphyrin (Ec)		301310	IEM0202
NFS1 deficiency	*NFS1*	AR		Disseminated intravascular coagulation						Lactate (P)		603485	IEM1123
Ferredoxin 2 deficiency	*FDX2*	AR			Anemia, microcytic Anemia, microcytic, hypochromic					Myoglobin (U), Lactate (P), Organic acids (U)		614585	IEM1121
Disorders of cobalamin metabolism													
Intrinsic factor deficiency	*CBLIF*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic	Pancytopenia				Homocysteine (P), Amino acids (P), Organic acids (U), Holotranscobalamin (P), Vitamin B12 (S)		261000	IEM0205
Cubilin deficiency	*CUBN*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic	Pancytopenia				Homocysteine (P), Amino acids (P), Organic acids (U), Holotranscobalamin (P), Vitamin B12 (S)		261100	IEM0206
Amnionless deficiency	*AMN*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic	Pancytopenia				Homocysteine (P); Organic acids (U); Vitamin B12 (S)	Hydroxycobalamin IM	261100	IEM0207
Transcobalamin 2 deficiency	*TCN2*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic	Pancytopenia				Amino acids (P), Organic acids (U), Holotranscobalamin (P)		275350	IEM0209
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P), Blood count	Hydroxycobalamin	277380	IEM0211
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P)	Hydroxycobalamin	614857	IEM0212
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic					Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)	Hydroxycobalamin, betaine; folinic acid; L-carnitine; no protein restriction; maintain normal methionine levels (+/- supplementation)	277400	IEM0213
Epi-cblC	*MMACHC; PRDX1*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic					SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin	609831;176763	IEM0214
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR	Neutrophils, hypersegmented		Anemia, megaloblastic	Pancytopenia				SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin, betaine	277410	IEM0215
Methionine synthase reductase deficiency-cblE	*MTRR*	AR			Anemia, megaloblastic					SAM (P,CSF), Amino acids (P), Organic acids (U)	Hydroxycobalamin, Betaine; Folinic acid	236270	IEM0216
Methionine synthase deficiency	*MTR*	AR			Anemia, megaloblastic					SAM (P,CSF), Amino acids (P), Organic acids (U)	Hydroxycobalamin, Betaine; Folinic acid	250940	IEM0217
Methylmalonic aciduria, cblA type	*MMAA*	AR				Pancytopenia				Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin injections, low protein diet, L-carnitine	251100	IEM0218
Methylmalonic aciduria, cblB type	*MMAB*	AR				Pancytopenia				Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine, hydroxycobalamin, ammonia scavengers	251110	IEM0219

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Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*	XL							Macrocytosis	Amino acids (P), Organic acids (U), Acylcamitines (P,DBS)		309541	IEM0220
Methylmalonic aciduria and homocystinuria due to Ronin deficiency	*THAP11*	AR							Macrocytosis	Amino acids (P), Organic acids (U), Acylcamitines (P,DBS)		609119	IEM1125
Methylmalonic aciduria and homocystinuria due to ZNF143 deficiency	*ZNF143*	AR							Macrocytosis	Amino acids (P), Organic acids (U), Acylcamitines (P,DBS)		603433	IEM1256
Disorders of folate metabolism													
Proton-coupled folate transporter deficiency	*SLC46A1*	AR			Anemia, megaloblastic	Pancytopenia				Folate (S), 5-Methyl-THF (CSF), Blood count (B), Immunoglobulins (S)		229050	IEM0221
5,10-methylenetetrahydrofolate reductase deficiency	*MTHFR*	AR					Thromboembolic episodes			Amino acids (P); Homocysteine (P); 5-Methyltetrahydrofolate (CSF), Folate (S)	Betaine, folinic acid or mefolinate, vitamin B12 , vitamin B6	236250	IEM0223
5,10-Methylene-tetrahydrofolate dehydrogenase deficiency	*MTHFD1*	AR			Anemia, megaloblastic Hemolytic uremic syndrome (atypical)	Thrombocytopenia				Amino acids (P); Homocysteine (P); 5-Methyltetrahydrofolate (CSF), Folate (S)		172460	IEM0224
Dihydrofolate reductase deficiency	*DHFR*	AR			Anemia, megaloblastic	Pancytopenia				LDH (P), Hb (B), Folates (B, CSF)	Folinic acid	126060	IEM0225
Folate transporter 1 deficiency	*SLC19A1*	AR			Anemia, megaloblastic					Homocysteine (P), Hemoglobin (B)		600424	IEM1254
Disorders of thiamine metabolism													
Thiamine transporter 1 deficiency (Rogers syndrome)	*SLC19A2*	AR			Anemia, sideroblastic	Thrombocytopenia				Lactate (P), Glucose (S)	Thiamine	603941	IEM0229
Disorders of niacin and NAD metabolism													
NAD(P)HX dehydratase deficiency	*NAXD*	AR				Pancytopenia				DNA		615910	IEM0244
Disorders of pantothenate metabolism													
Pantothenate kinase 2 deficiency (Hallervorden-Spatz disease)	*PANK2*	AR	Spiculated red cells							Iron (brain), DNA	Possible iron chelation, possible panthotenate	234200	IEM0246
Disorders of vitamin K metabolism													
γ-Glutamyl carboxylase deficiency	*GGCX*	AR		Bleeding tendency Epistaxis						Coagulation factors (Blood)		277450	IEM0271
Vitamin K epoxide reductase deficiency	*VKORC1*	AR		Bleeding tendency						Coagulation factors (Blood)		607473	IEM0272
Microsomal epoxide hydrolase deficiency	*EPHX1*	AR			Anemia					Bile acids (S)		607748	IEM0273
Disorders of copper metabolism													
Copper-transporting ATPase subunit beta deficiency (Wilson disease)	*ATP7B*	AR		Coagulopathy	Anemia, hemolytic Hemolysis	Thrombocytopenia			Leukocytosis	ASAT/ALAT (P); Copper (S, U); Ceruloplasmin (S)	Penicillamine with pyridoxine ; Trientine ; Zinc sulphate	277900	IEM0279
Copper-transporting ATPase subunit alpha deficiency (Menkes disease)	*ATP7A*	AR			Anemia	Neutropenia				Copper (S, U), Ceruloplasmin (S)	Copper chloride or L-histidine IV or SC	309400	IEM0280
Disorders of iron metabolism													
Hereditary ceruloplasmin deficiency	*CP*	AR			Anemia, microcytic					Ceruloplasmin (S), Ferritin (S)		604290	IEM0292
Matriptrase 2 deficiency	*TMPRSS6*	AR			Anemia, microcytic					Ferritin (S), Transferrin saturation, Hepcidin (P)		206200	IEM0293
Hereditary transferrin deficiency	*TF*	AR			Anemia, hypochromic					Iron (LI)		209300	IEM0294
Transferrin receptor deficiency	*TFRC*	AR			Anemia	Neutropenia Thrombocytopenia				B cells, circulating (B), IgG(S)		616740	IEM0295
Divalent metal transporter 1 deficiency	*SLC11A2*	AR			Anemia, microcytic					Iron (S, LI), Transferrin saturation, Ferritin (S)		206100	IEM0296
BMP6 deficiency	*BMP6*							Iron overload		Ferritin (S)		112266	IEM1444
Endosomal ferrireductase deficiency	*STEAP3*	AD			Anemia, microcytic, hypochromic			Iron overload		DNA		615234	IEM1445
Disorders of zinc metabolism													
Hyperzincemia and hypercalprotectinemia	*PSTPIP1*	AD			Anemia					DNA		604416	IEM1258
DISORDERS OF CARBOHYDRATES (n=18)													
Disorders of carbohydrate transport and absorption													
Glucose transporter 1 deficiency	*SLC2A1*	AD, AR			Anemia, hemolytic					Glucose (P, CSF)	Ketogenic diet, clinical trials with triheptanoin	606777;612126;601042;612	IEM0314
Galactose-1-phosphate uridyltransferase deficiency	*GALT*	AR			Anemia, hemolytic					Chemistry (P,U), Oligosaccharides (U), Amino acids (P)	Galactose restriction	230400	IEM0322
Disorders of the pentose phosphate pathway and polyol metabolism													
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	XL			Anemia, hemolytic					LDH (P), Bilirubin (P), Reticulocytes (B)		300908	IEM0327
Transaldolase deficiency	*TALDO1*	AR			Anemia	Thrombocytopenia			Leukocytosis	ASAT/ALAT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Ferritin (S), Polyol (U)	Liver transplant	606003	IEM0329
Glycogen storage diseases													
Glucose-6-phosphate transporter deficiency	*SLC37A4*	AR		Bleeding tendency		Neutropenia				ASAT/ALAT (P), CK (P), Lactate (P), Glucose (P), Biotinidase (P)	Frequent meals, uncooked cornstarch, filgrastim (G-CSF), neutropenia responds to empagliflozin	232220	IEM0355
Alpha-glucosidase deficiency (Pompe disease)	*GAA*	AR	Vacuolated lymphocytes							ASAT/ALAT (P), CK (P), Glycogen (M)	Alglucosidase alpha	232300	IEM0356
Lysosome-associated membrane protein 2 deficiency (Danon disease)	*LAMP2*	XL	Vacuolated lymphocytes							ASAT/ALAT (P), CK (P), Glycogen (M)		300257	IEM0367
HOIL1 interacting protein deficiency	*RNF31*	AR			Anemia					DNA		612487	IEM1135
Disorders of gluconeogenesis													
Glucose-6-phosphatase deficiency (von Gierke disease, GSD1a)	*G6PC*	AR		Bleeding tendency						ASAT/ALAT (P), Lactate (P), Glucose (S), Glycogen (L), Biotinidase (P)	Frequent meals, uncooked cornstarch	232200	IEM0370
Disorders of glycolysis													
Hemolytic anemia due to hexokinase 1 deficiency	*HK1*	AR			Anemia, hemolytic					Bilirubin (P), Blood count (B), DNA		235700	IEM0374
Glucose-6-phosphate isomerase deficiency	*GPI*	AR			Anemia, hemolytic Hemolytic crisis					Bilirubin, unconjugated (P), Hemoglobin (B), Reticulocytes (B), Enzyme activity		615802	IEM0379
Muscle phosphofructokinase deficiency (Tarui disease)	*PFKM*	AR			Anemia, hemolytic					CK (P), Glycogen (M), Uric acid (P)		232800	IEM0380
Aldolase A deficiency	*ALDOA*	AR			Anemia, hemolytic					Bilirubin (P), Reticulocytes (B), Aldolase A (Ec)		611881	IEM0381
Triosephosphate isomerase deficiency	*TPI1*	AR			Anemia, hemolytic					Dihydroxyacetone phosphate (Ec)		615512	IEM0382
Triosephosphate isomerase deficiency	*TPI1*	AR			Anemia					Dihydroxyacetone phosphate (Ec)		615512	IEM0382
Phosphoglycerate kinase deficiency	*PGK1*	XL			Anemia, hemolytic					Reticulocytes (B)		300653	IEM0383
Pyruvate kinase deficiency	*PKLR*	AR	Spiculated red cells		Anemia, hemolytic Hemolytic crisis		Thrombosis			Bilirubin, unconjugated (P), Ferritin (S), Hemoglobin (B), Reticulocytes (B)		266200	IEM0386
Bisphosphoglycerate mutase deficiency	*BPGM*	AR			Anemia, hemolytic					Hemoglobin (B)		222800	IEM1661

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MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM (n=31)													
Disorders of the Krebs cycle													
Fumarate hydratase deficiency	*FH*	AR				Neutropenia				Lactate (P), Organic acids (U), Catecholamines (P, U)	Alpha-adrenergic receptor blocker	606812	IEM0401
Disorders of mitochondrial carriers													
Mitochondrial dicarboxylate transporter deficiency	*SLC25A10*	AR			Anemia, microcytic, hypochromic					DNA		606794	IEM1139
Disorders of complex I subunits													
NADH dehydrogenase beta subcomplex subunit 11 deficiency	*NDUFB11*	XL			Anemia, sideroblastic					Lactate (P,CSF)		252010	IEM0429
Disorders of complex III subunits													
UQCRFS1 deficiency	*UQCRFS1*	AR			Anemia					DNA		618775	IEM1483
Disorders of complex IV subunits													
Cytochrome c oxidase subunit 4I1 deficiency	*COX4I1*	AR							Macrocytosis	DNA		123864	IEM1216
Disorders of complex IV assembly and ancillary proteins													
Cytochrome c oxidase assembly factor 10 deficiency	*COX10*	AR			Anemia					Lactate (P), Hemoglobin (B)		220110;256000	IEM0470
Cytochrome c oxidase assembly factor 16 deficiency	*COX16*	AR		Coagulopathy						Lactate (P), Glucose (P), Organic acids (U), Acylcarnitines (P,DBS)		618064	IEM1514
Disorders of complex V subunits													
Mitochondrial ATP synthase F0 subunit 6 deficiency	*MT-ATP6*	MT			Anemia, sideroblastic					Lactate (P)		516060	IEM0484
Disorders of mitochondrial cytochrome synthesis and incorporation													
Mitochondrial cytochrome c deficiency	*CYCS*	AD				Thrombocytopenia				Thrombocytes (B)		612004	IEM0489
Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication													
Mitochondrial deoxyguanosine kinase deficiency	*DGUOK*	AR						Thrombocytopenia		a-fetoprotein (S), Lactate (P), Amino acids (P), Sialotrasferins (S)		251880;601465	IEM0493
FBXL4 deficiency	*FBXL4*	AR				Neutropenia				Lactate (P)		615471	IEM0502
Disorders of mitochondrial transcription and RNA transcript processing													
CCA-adding tRNA-nucleotidyltransferase deficiency	*TRNT1*	AR			Anemia, sideroblastic Microcytosis					IgG (serum), Amino acids (P)		616959	IEM0507
Pseudouridine synthase 1 deficiency	*PUS1*	AR			Anemia, sideroblastic					Lactate (P)		600462	IEM0511
tRNA 5-methylaminomethyl-2-thiouridylate-methyltransferase deficiency	*TRMU*	AR		Coagulopathy						Lactate (P)		613070	IEM0514
Mitochondrial RNA-processing endoribonuclease deficiency	*RMRP*	AR				Lymphopenia				DNA		607095;250250;250460	IEM0515
Mitochondrial RNA polymerase deficiency	*POLRMT*					Thrombocytopenia				Lactate (P), 5-Methyl-THF (CSF)		601778	IEM1534
Mitochondrial ribosomopathies													
Mitochondrial oxodicarboxylate carrier deficiency	*SLC25A21*	AR			Anemia, microcytic, hypochromic					Organic acids (U), Quinolinic acid (U)		607571	IEM1209
Disorders of mitochondrial tRNA incorporation and recycling													
Mitochondrial leucyl-tRNA synthetase deficiency	*LARS2*	AR			Anemia, sideroblastic					Lactate (P)		615300	IEM0560
Mitochondrial seryl-tRNA synthetase deficiency	*SARS2*	AR				Pancytopenia				Lactate (P), Uric acid (S)		613845	IEM0563
Mitochondrial tyrosyl-tRNA synthetase deficiency	*YARS2*	AR			Anemia, sideroblastic					Lactate (P), Blood count		613561	IEM0564
Mitochondrial tyrosyl-tRNA synthetase deficiency	*YARS2*	AR			Anemia					Lactate (P), Blood count		613561	IEM0564
Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit A deficiency	*QRSL1*	AR			Anemia					Lactate (P)		617209	IEM0569
Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit C deficiency	*GATC*	AR			Anemia					Lactate (P)		617210	IEM0570
Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit B deficiency	*GATB*	AR			Anemia					Lactate (P)		603645	IEM1159
Disorders of mitochondrial phospholipid metabolism													
Tafazzin deficiency (Barth syndrome)	*TAZ*	XL				Neutropenia				Organic acids (U), Lipid panel (S), Urinalysis, 3-Methylglutaconic acid (U), 3-Methylglutaric acid (U), Carnitine, free (P), Blood count, Elamipretide		302060	IEM0583
MICOS complex subunit MIC13 deficiency	*MICOS13*	AR		Coagulopathy						Lactate (P), Organic acids (U)		616658	IEM1375
Disorders of mitochondrial protein import													
TIMM14 deficiency	*DNAJC19*	AR			Anemia, microcytic, hypochromic					Organic acids (U), CK (P), Lactate (P), 3-Methylglutaconic acid (U), Blood count		610198	IEM0585
Disorders of mitochondrial protein quality control													
CLPB deficiency	*CLPB*	AR				Neutropenia				DNA		616271	IEM0594
HSPA9 deficiency	*HSPA9*	AR			Anemia, sideroblastic			Ringed sideroblasts on bone marrow		Bone marrow stain; DNA		182170	IEM0597
Oxa1-like deficiency	*OXA1L*				Anemia					Amino acids (P), Iron (S)		601066	IEM1543
Other disorders of mitochondrial homeostasis													
Sideroflexin 4 deficiency	*SFXN4*	AR			Anemia, macrocytic					Lactate (P)		615578	IEM0609
DISORDERS OF LIPIDS (n=21)													
Disorders of fatty acid oxidation and transport													
Isolated deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase	*HADHA*	AD						Hemophagocytosis		Organic acids (U), Acylcamitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	600890	IEM0636
Fatty acid transport protein 4 deficiency	*SLC27A4*	AR							Eosinophilia	DNA		608649	IEM0638
Disorders of cytoplasmic triglyceride metabolism													
Lipin 2 deficiency (Majeed syndrome)	*LPIN2*	AR			Anemia, dyserythropoietic Anemia, microcytic, hypochromic	Neutropenia				DNA		609628	IEM0658
Diacylglycerol acyltransferase deficicncy	*DGAT1*	AR			Anemia					ASAT/ALAT (P), Albumin (S), IgG(S)		615863	IEM0659
CGI-58 deficiency	*ABHD5*	AR	Vacuolated lymphocytes							ASAT/ALAT (P)		275630	IEM0660

Supplementary Table S1. List of inherited metabolical diseases with hematologic phenotypes, laboratory investigation, treatment options (if applicable), OMIM references and IEMbase ID.

Disorder (n=264)	Gene	Inheritance	Abormal blood cell morphology	Coagulation abnormalities	Anemias	Abnormal blood count	Hypercoagulability	Marrow abnormality	Other	Diagnostic markers	Specific treatmnt	OMIM	IEMbase code
Adipose triglyceride lipase deficiency	*PNPLA2*	AR	Vacuolated granulocytes with lipid droplets (Jordans' anomaly)							CK (P)		610717	IEM0661
Disorders of non-mitochondrial phospholipid metabolism													
Diacylglycerol kinase ε deficiency	*DGKE*	AR			Hemolytic uremic syndrome (atypical)					DNA		615008	IEM0675
Disorders of eicosanoid metabolism													
Thromboxane synthase deficiency	*TBXAS1*	AR			Anemia	Thrombocytopenia			Leukocytosis	DNA		231095	IEM0684
Prostaglandin transporter deficiency	*SLCO2A1*	AR			Anemia					DNA		259100;119900;259100	IEM0686
Cytosolic phospholipase A2α deficiency	*PLA2G4A*	AR			Anemia					DNA		600522	IEM1072
Disorders of phosphoinositide metabolism													
Phosphatidylinositol 3-kinase regulatory subunit 1 deficiency (SHORT syndrome)	*PIK3R1*	AD				Lymphopenia				IgG; IgA; IgM (S)		269880;616005	IEM0700
Scavenger receptor B1 deficiency (SCARB1)	*SCARB1*	AD, AR		Platelet function, abnormal						HDL cholesterol (P)		601040;610762	IEM1039
Disorders of lipoprotein metabolism													
Microsomal triglyceride transfer protein deficiency	*MTTP*	AR	Acanthocytosis Spiculated red cells	Bleeding tendency						LDL/HDL cholesterol (P), Apo B (P), Vitamins A/E (P)		200100;157147	IEM0723
Apolipoprotein E deficiency	*APOE*	AR	Cytoplasmic granules (histiocytes) Sea blue histiocytes			Thrombocytopenia				Lipid panel (S)	Statins, niacin, fibrates	617347	IEM0725
Disorders of cholesterol biosynthesis													
Mevalonate kinase deficiency	*MVK*	AR			Anemia	Thrombocytopenia			Leukocytosis	Leucotriens (P), Organic acids (U)	Inflammatory control (non-steroidal anti-inflammatory drugs, corticosteroids, IL-1 targeting biologic agents, TNF-alpha blockade, allogenic stem cell transplantation)	610377	IEM0740
Disorders of steroid metabolism													
MIRAGE syndrome	*SAMD9*	AD				Thrombocytopenia				Steroids (P), Corticotropin (P)		617053	IEM1497
Disorders of bile acid synthesis													
3β-Hydroxy-Δ5-C27-steroid dehydrogenase-isomerase deficiency	*HSD3B7*	AR		Vit K responsive bleeding						Bile acids (P,U); Bilirubin, conjugated (P); Vitamins D/E		607764	IEM0779
Oxysterol 7α-hydroxylase deficiency	*CYP7B1*	AR		Vit K responsive bleeding						Bile acids (U), Sterols (P), Vitamin E (P)	Chenodeoxycholic acid, liver transplant	603711	IEM0781
Sterol 27-hydroxylase deficiency	*CYP27A1*	AR						Foam cells		Lipid panel (S); Sterols (P); Cholestane pentol glucuronide (U); 25-Hydroxy-Vitamin D (P)	Chenodeoxycholic acid, HMG-CoA reductase inhibitors, low density lipoprotein apheresis	213700	IEM0782
α-Methylacyl-CoA racemase deficiency	*AMACR*	AR		Vit K responsive bleeding						Bile acids (U), VLCFA (P), Vitamins D/E (P)	Cholic acid	604489	IEM0783
Congenital bile acid synthesis defect ABCD3	*ABCD3*	AR			Anemia					Bile acids (P), ASAT/ALAT (P), Iron (S)		616278	IEM1187
STORAGE DISORDERS (n=29)													
Neuronal ceroid lipofuscinosis													
CLN3 disease	*CLN3*	AR	Vacuolated lymphocytes Sea blue histiocytes							Peripheral smear, DNA	In trial gene therapy (NCT03770572)	204200	IEM0821
Sphingolipidoses													
Glucocerebrosidase deficiency (Gaucher disease)	*GBA*	AR			Anemia	Thrombocytopenia Pancytopenia		Foam cells Hemophagocytosis		Glucosylsphingosine (S); Chitotriosidase (B)	Enzyme replacement therapy, substrate reduction, bone marrow transplantation	230800	IEM0832
Gaucher disease-like disorder due to saposin C deficiency	*PSAP*	AR			Anemia	Thrombocytopenia		Foam cells		Sulfatides (U), Protein (CSF), Lysosomal enzymes (DBS)		610539	IEM0833
Acid sphingomyelinase deficiency	*SMPD1*	AR				Pancytopenia Thrombocytopenia		Foam cells		Enzyme activity (WBC)		257200;607616	IEM0834
Beta-galactosidase-1 deficiency (Morquio syndrome type B)	*GLB1*	AR	Vacuolated lymphocytes (Alder) Reilly bodies					Foam cells		Oligosaccharides (U), Lysosomal enzymes (DBS)	In trial gene therapy (NCT03952637)	230500	IEM0835
Beta-hexosaminidase subunit beta deficiency (Sandhoff disease)	*HEXB*	AR	Vacuolated lymphocytes					Foam cells		Oligosaccharides (U), Lysosomal enzymes (DBS)		268800	IEM0837
Acid ceramidase deficiency, inflammatory phenotype (Farber disease)	*ASAH1*	AR						Foam cells		Lysosomal enzymes (DBS)		228000	IEM0845
Combinded saposin deficiency	*PSAP*	AR						Foam cells		Sulfatides (U), Protein (CSF), Lysosomal enzymes (DBS)		611721	IEM0847
Oligosaccharidoses													
Alpha-neuraminidase deficiency	*NEU1*	AR	Vacuolated lymphocytes					Foam cells		Oligosaccharides (U), Lysosomal enzymes (DBS)		256550	IEM0848
Cathepsin A deficiency	*CTSA*	AR	Vacuolated lymphocytes					Foam cells		Oligosaccharides (U), Lysosomal enzymes (DBS)		256540	IEM0849
Alpha-mannosidase B deficiency	*MAN2B1*	AR	Vacuolated lymphocytes					Foam cells		Oligosaccharides (U), Lysosomal enzymes (DBS)	Recombinant enzyme replacement therapy (velmanase alfa); HCT	248500	IEM0850
Beta-mannosidase deficiency	*MANBA*	AR						Foam cells		Enzyme activity (DBS,WBC), Oligosaccharides (U)		248510	IEM0851
Alpha-L-fucosidase deficiency	*FUCA1*	AR	Vacuolated lymphocytes					Foam cells		Enzyme activity (DBS,WBC), Fucose (U)	Bone marrow transplant	230000	IEM0853
Aspartylglucosaminidase deficiency	*AGA*	AR	Vacuolated lymphocytes							Enzyme activity (DBS,WBC), Aspartylglucosamine (U)		208400	IEM0854
Mucolipidoses													
UDP-N-acetylglucosamine-1-phosphotransferase subunit alpha/beta deficiency	*GNPTAB*	AR						Foam cells		Oligosaccharide (U), Glycosaminoglycans, Enzyme activity (S)		252600	IEM0855
Mucolipin 1 deficiency	*MCOLN1*	AR						Foam cells		G ASATrin (S)		252650	IEM0857
Mucopolysaccharidoses													
Alpha-iduronidase deficiency	*IDUA*	AR	(Alder) Reilly bodies							Total GAGs (U), Dermatan sulfate (U), Heparan sulfate (U), Enzyme activity (WBC)	Hematopoietic cell transplantation (HCT), enzyme replacement therapy (laronidase)	607014;607015;607016	IEM0858
Iduronate 2-sulfatase deficiency (Hunter disease)	*IDS*	XL	(Alder) Reilly bodies							Total GAGs (U), Dermatan sulfate (U), Heparan sulfate (U), Enzyme activity (WBC)	Enzyme replacement therapy (idursulfase)	309900	IEM0859
Heparan N-sulfatase deficiency (Sanfilippo A)	*SGSH*	AR	(Alder) Reilly bodies							Mucopolysaccharides (U); Enzyme assay (DBS, L, F)	Clinical trial with intracerebroventricular infusion of chimeric fusion of recombinant enzyme +IGF2	252900	IEM0860
N-acetylglucosaminidase deficiency (Sanfilippo B)	*NAGLU*	AR	(Alder) Reilly bodies							Mucopolysaccharides (U); Enzyme assay (DBS, S, F)	Clinical trial with intracerebral adenovirus associated viral vector containing human NAGLU cDNA	252920	IEM0861
Heparan-alpha-glucosaminide N-acetyltransferase deficiency (Sanfilippo C)	*HGSNAT*	AR	(Alder) Reilly bodies							Mucopolysaccharides (U); Enzyme assay (DBS, L, F)		252930	IEM0862
N-acetylglucosamine 6-sulfatase deficiency (Sanfilippo D)	*GNS*	AR	(Alder) Reilly bodies							Mucopolysaccharides (U); Enzyme assay (DBS, L, F)		252940	IEM0863
N-acetylgalactosamine 6-sulfatase deficiency (Morquio A)	*GALNS*	AR	(Alder) Reilly bodies							Total GAGs (U), Keratan sulfate (U), Enzyme activity (WBC)	Elosulfase	253000	IEM0864
N-acetylgalactosamine 4-sulfatase deficiency	*ARSB*	AR	(Alder) Reilly bodies							Total GAGs (U), Dermatan sulfate (U), Enzyme activity (WBC)	Galsulfase	253200	IEM0866
Beta-glucuronidase deficiency (Sly disease)	*GUSB*	AR	(Alder) Reilly bodies							Total GAGs (U), Dermatan sulfate (U), Heparan sulfate (U), Enzyme activity (S, WBC)	Vestronidase	253220	IEM0867

Supplementary Table S1. List of inherited metabolical diseases with hematologic phenotypes, laboratory investigation, treatment options (if applicable), OMIM references and IEMbase ID.

Disorder (n=264)	Gene	Inheritance	Abormal blood cell morphology	Coagulation abnormalities	Anemias	Abnormal blood count	Hypercoagulability	Marrow abnormality	Other	Diagnostic markers	Specific treatmnt	OMIM	IEMbase code
Mucopolysaccharidosis-plus syndrome	*VPS33A*	AR	Granulation in lymphocytes		Anemia	Thrombocytopenia		Foam cells Bone marrow hypoplasia		Mucopolysaccharides (U), Oligosaccharides (U)		617303	IEM0869
Disorders of lysosomal cholesterol metabolism													
Niemann-Pick disease type C1	*NPC1*	AR	Sea blue histiocytes					Foam cells Blue histiocytes Hemophagocytosis		Oxysterols (P); Filipin staining (F)	Miglustat; experimental intrathecal or intravenous 2-hydroxypropyl-beta-cyclodextrin; oral Arimoclomol	257220	IEM0870
Niemann-Pick disease type C2	*NPC2*	AR	Sea blue histiocytes					Foam cells Blue histiocytes		Oxyterols (P), filipin test (F)		607625	IEM0871
Lysosomal acid lipase deficiency (Wolman disease)	*LIPA*	AR	Spiculated red cells		Anemia	Thrombocytopenia		Hemophagocytosis		Lipid panel (S), , Enzyme activity (S)	Enzyme replacement therapy	278000	IEM0872
DISORDERS OF TETRAPYRROLES (n=8)													
Disorders of heme metabolism													
Erythroid 5-aminolevulinate synthase deficiency	*ALAS2*	XL			Anemia, microcytic, hypochromic			Syderoblasts (bone marrow)		Porphyrins (U,RBC)		300751	IEM0786
Uroporphyrinogen III synthase deficiency	*UROS*	AR			Anemia, microcytic, hypochromic					Porphyrins (U, RBC, stools)	Bone marrow transplant	263700	IEM0790
Ferrochelatase deficiency	*FECH*	AD			Anemia Microcytosis					Free protorphyrin (RBC); Fluorescence scanning (P)	Afamelanotide	177000	IEM0795
GATA1 deficiency	*GATA1*	XL			Anemia, macrocytic	Thrombocytopenia Neutropenia				Reticulocytes (B)			IEM0796
NADH-cytochrome b5 reductase deficiency	*CYB5R3*	AR							Methemoglobin	DNA		250800	IEM0798
Cytochrome b5 deficiency	*CYB5A*	AR							Methemoglobin	Steroids (P)		250790	IEM0799
Heme oxygenase 1 deficiency	*HMOX1*	AR			Anemia, hemolytic					DNA		614034	IEM0800
CLPX deficiency	*CLPX*	AD			Anemia, microcytic, hypochromic					Protoorphyrin (RBC)		618015	IEM1189
CONGENITAL DISORDERS OF GLYCOSYLATION (n=45)													
Disorders of N-linked glycosylation													
MPI-CDG	*MPI*	AR		Coagulopathy			Thrombosis			Sialotransferins (S), Coagulation factors (P), Albumin (S)		602579	IEM0909
DPAGT1-CDG	*DPAGT1*	AR		Coagulopathy						Sialotrasferrins (S), Antithrombin III (P)		608093	IEM0910
ALG1-CDG	*ALG1*	AR		Coagulopathy		Thrombocytopenia				Sialotransferins (S), IGG (P), B cells, circulating (blood), Protein C (S)		608540	IEM0914
ALG2-CDG	*ALG2*	AR		Coagulopathy						Factor XI (B), Sialotrasferrins (S)		607906	IEM0915
ALG11-CDG	*ALG11*	AR		Coagulopathy					Leukocytosis	Factor XI (B), Antithrombin III (P), Sialotrasferrins (S)		613661	IEM0916
RFT1-CDG	*RFT1*	AR		Coagulopathy			Thrombosis			Sialotransferins (S), ASAT/ALAT (P), Coagulation factors (P)		612015	IEM0917
ALG3-CDG	*ALG3*	AR		Coagulopathy						Antithrombin III (P), Protein S (S), Sialotrasferrins (S)		601110	IEM0918
ALG9-CDG	*ALG9*	AR		Coagulopathy						Lipid panel (S), Sialotransferrins (S), Albumin (S), Factor XI (B)		608776	IEM0919
ALG12-CDG	*ALG12*	AR		Coagulopathy		Thrombocytopenia				ASAT/ALAT (P), Glucose (S), Lipid panel (S), Sialotransferrins (S), IGF BP3, Antithrombin III (P)		607143	IEM0920
ALG6-CDG	*ALG6*	AR		Coagulopathy						Factor XI (B), Sialotrasferrins (S), ASAT/ALAT (P)		603147	IEM0921
ALG8-CDG	*ALG8*	AD, AR		Coagulopathy	Anemia	Thrombocytopenia				CK (P), Sialotransferrins (S), Albumin (S), Coagulation factors (P)		608104	IEM0922
MGAT2-CDG	*MGAT2*	AR		Bleeding tendency Coagulopathy						ASAT/ALAT (P), CK (P), Sialotransferrins (S), Coagulation factors (P)		212066	IEM0931
STT3B-CDG	*STT3B*	AR				Thrombocytopenia				Sialotrasferrins (S)		615597	IEM1193
DDOST-CDG	*DDOST*	AR		Coagulopathy						Factor XI (B), Proteins C/S (S), Sialotrasferrins (S)		614507;602202	IEM1192
B4GALT1-CDG	*B4GALT1*	AR		Perinatal bleeding diathesis Coagulopathy						Antithrombin (B), Factor XI (B), Sialotrasferrins (S)		607091	IEM1194
MAN2B2-CDG	*MAN2B2*	AR			Anemia	Lymphopenia Thrombocytopenia				Sialotransferrins (S), C-reaktive protein, CRP (P), IgE (S)		618899	IEM1549
Disorders of glycosylphosphatidylinositol biosynthesis													
PIGM-CDG	*PIGM*	AR					Thrombosis			Flow cytometry of GPI markers (Gr)		610293	IEM0977
PIGT-CDG	*PIGT*	AR			Hemolysis					ALP (P), GPI-anchored proteins (WBC, F)		615398	IEM0982
Disorders of dolichol metabolism													
SRD5A3-CDG	*SRD5A3*	AR		Coagulopathy						Antithrombin III (P), Protein C (S), Sialotrasferrins (S)		612379	IEM0993
DPM1-CDG	*DPM1*	AR		Coagulopathy						Factor XI (B), Sialotrasferrins (S)		608799	IEM0995
DPM3-CDG	*DPM3*	AR		Coagulopathy						ASAT/ALAT (P), CK (P), Sialotransferrins (S), Dolichol-P-mannose (S), Factor XI (B)		612937	IEM0997
MPDU1-CDG	*MPDU1*	AR		Coagulopathy						Sialotrasferrins (S), Antithrombin III (P)		609180	IEM0998
Disorders of monosaccharide synthesis and interconversion													
PGM1-CDG	*PGM1*	AR		Coagulopathy			Thrombosis			ASAT/ALAT (P), CK (P), Ammonia (P), Sialotransferrins (S), Antithrombin III (P)		614921	IEM1003
PGM3-CDG	*PGM3*	AR				Neutropenia				Sialotransferrins (S), Normal B-cell (CD19+) count		615816;172100	IEM1004
Ubiquitous glucose-6-phosphatase deficiency	*G6PC3*	AR			Anemia	Neutropenia Thrombocytopenia				Blood count; DNA		612541	IEM1005
Disorders of Golgi transport													
SLC35A1-CDG	*SLC35A1*	AR	Macrothrombocytopenia	Bleeding tendency		Thrombocytopenia				Sialotrasferrins (S)		603585	IEM1008
SLC35C1-CDG	*SLC35C1*	AR							Neutrophilia Bombay phenotype	Neutrophil motility/rolling (B)		266265	IEM1011
Disorders of vesicular trafficking													
Conserved oligomeric Golgi complex subunit 1 deficiency	*COG1*	AR				Thrombocytopenia				Sialotransferrins (S)		611209	IEM1013
Component of COG complex 6 deficiency	*COG6*	AR						Hemophagocytosis		ASAT/ALAT (P), CK (P), Lactate (P), Sialotransferins (S), Vitamins A, D, E, K (S)		606977;614576	IEM1016
Conserved oligomeric Golgi complex subunit 8 deficiency	*COG8*	AR		Coagulopathy						Factor XI (B), Protein C (S), Sialotrasferrins (S)		611182	IEM1018

Supplementary Table S1. List of inherited metabolical diseases with hematologic phenotypes, laboratory investigation, treatment options (if applicable), OMIM references and IEMbase ID.

Disorder (n=264)	Gene	Inheritance	Abormal blood cell morphology	Coagulation abnormalities	Anemias	Abnormal blood count	Hypercoagulability	Marrow abnormality	Other	Diagnostic markers	Specific treatmnt	OMIM	IEMbase code
Jagunal 1 deficiency	*JAGN1*	AR				Neutropenia				DNA		616022	IEM1019
Congenital dyserythropoietic anemia type 2	*SEC23B*	AR			Anemia, dyserythropoietic					Bilirubin (P), Acidified serum test, HEMPAS test		224100	IEM1020
Cohen syndrome VPS13B-CDG	*VPS13B*	AR				Neutropenia				Sialotrasferrins (S)		216550	IEM1023
Conserved oligomeric Golgi complex subunit 2 deficiency	*COG2*	AR		Coagulopathy						Sialotrasferrins (S), Ceruloplasmin (S), Copper (S)		606974	IEM1199
VPS45 deficiency	*VPS45*	AR				Neutropenia				DNA		615285	IEM1413
NBAS deficiency	*NBAS*	AR	Pelger Huet bodies	Coagulopathy						ASAT/ALAT (P), DNA		608025	IEM1415
Familial hemophagocytic lymphohistiocytosis type 3	*UNC13D*	AR				Pancytopenia				ASAT/ALAT (P), Neopterin (U), DNA		608898	IEM1419
Grey platelet syndrome	*NBEAL2*	AR		Bleeding tendency		Thrombocytopenia				DNA		139090	IEM1432
Combined factor V and factor VIII deficiency type 1	*LMAN1*	AR		Bleeding tendency						Coagulation factors (P)		227300	IEM1433
Combined factor V and factor VIII deficiency type 2	*MCFD2*	AR		Bleeding tendency						DNA		613625	IEM1434
RINT1-CDG	*RINT1*	AR		Coagulopathy						ASAT/ALAT (P), DNA		618641	IEM1675
VPS4A deficiency	*VPS4A*	AR			Anemia					DNA		609982	IEM1518
Disorders of Golgi homeostasis													
ATP6AP1-CDG	*ATP6AP1*	XL							Leukocytosis	Sialotransferrins (S); Ceruloplasmin (S); Copper (S); IgG (S)		300972	IEM1027
SLC37A4-CDG	*SLC37A4*	AD		Coagulopathy	Anemia	Thrombocytopenia				ASAT/ALAT (P), CK (P), Lactate (P), Glucose (P), Biotinidase (P)	Frequent meals, uncooked cornstarch, filgrastim		IEM1670
ATP6AP2-CDG	*ATP6AP2*	XL		Coagulopathy						Sialotransferins (S), ASAT/ALAT (P); IgG (S); Factor XI (B)		300423	IEM1028
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=31)													
Disorders of nucleotide and nucleic acid metabolism													
STING superactivity	*TMEM173*	AD				Lymphopenia				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*	AD							Leukocytosis	IgG(S), Leukocytes (B)		222100	IEM0035
Equilibrative nucleoside transporter 1 deficiency	*SLC29A1*	AR							Augustine-null blood type	DNA			IEM0040
Equilibrative nucleoside transporter 3 deficiency	*SLC29A3*	AR							Leukocytosis	Erythrocyte sedimentation rate; IgG (S)		602782	IEM0041
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases													
Leucyl-tRNA synthetase 1 deficiency	*LARS1*	AR			Anemia, microcytic					ASAT/ALAT (P), Lactate (P), DNA		615438	IEM1310
Disorders of ribosomal biogenesis													
X-linked dyskeratosis congenita	*DKC1*	XLR				Thrombocytopenia				DNA		305000	IEM1334
Autosomal recessive dyskeratosis congenita type 1	*NOLA3*	AR				Pancytopenia				DNA		224230	IEM1335
Autosomal recessive dyskeratosis congenita type 2	*NOLA2*	AR				Thrombocytopenia Pancytopenia				DNA		613987	IEM1336
Nucleophosmin 1 deficiency	*NPM1*	AD				Thrombocytopenia				DNA		164040	IEM1337
Diamond-Blackfan anemia type 1	*RPS19*	AD			Anemia, macrocytic					Reticulocytes (B)		105650	IEM1343
Diamond-Blackfan anemia type 3	*RPS24*	AD			Anemia, macrocytic					Reticulocytes (B)		610629	IEM1344
Diamond-Blackfan anemia type 4	*RPS17*	AD			Anemia, macrocytic					Reticulocytes (B)		612527	IEM1345
Diamond-Blackfan anemia type 5	*RPL35A*	AD			Anemia, macrocytic					Reticulocytes (B)		612528	IEM1346
Diamond-Blackfan anemia type 6	*RPL5*	AD			Anemia, macrocytic					Reticulocytes (B)		612561	IEM1347
Diamond-Blackfan anemia type 7	*RPL11*	AD			Anemia, macrocytic					Reticulocytes (B)		612562	IEM1348
Diamond-Blackfan anemia type 8	*RPS7*	AD			Anemia, macrocytic					Reticulocytes (B)		612563	IEM1349
Diamond-Blackfan anemia type 9	*RPS10*	AD			Anemia, macrocytic					Reticulocytes (B)		613308	IEM1350
Diamond-Blackfan anemia type 10	*RPS26*	AD			Anemia, macrocytic					Reticulocytes (B)		613309	IEM1351
Diamond-Blackfan anemia type 11	*RPL26*	AD			Anemia, macrocytic					Reticulocytes (B)		614900	IEM1352
Diamond-Blackfan anemia type 12	*RPL15*	AD			Anemia, macrocytic					Reticulocytes (B)		615550	IEM1353
Diamond-Blackfan anemia type 13	*RPS29*	AD			Anemia, megaloblastic					Reticulocytes (B)		615909	IEM1354
Diamond-Blackfan anemia type 14	*TSR2*	XLR			Anemia, macrocytic					Reticulocytes (B)		300946	IEM1355
Diamond-Blackfan anemia type 15	*RPS28*	AD			Anemia, macrocytic					Reticulocytes (B)		606164	IEM1356
Diamond-Blackfan anemia type 16	*RPL27*	AD			Anemia, macrocytic					Reticulocytes (B)		617408	IEM1357
Diamond-Blackfan anemia type 17	*RPS27*	AD			Anemia, macrocytic					Reticulocytes (B)		617409	IEM1358
Diamond-Blackfan anemia type 18	*RPL18*	AD			Anemia, hypoplastic, macrocytic					Reticulocytes (B)		618310	IEM1359
Diamond-Blackfan anemia type 19	*RPL35*	AD			Anemia, macrocytic					Reticulocytes (B)		618312	IEM1360
Diamond-Blackfan anemia type 20	*RPS15A*	AD			Anemia, macrocytic					Reticulocytes (B)		618313	IEM1361
Cytosolic small ribosomal subunit 20 deficiency	*RPS20*	AD			Anemia, macrocytic					DNA			IEM1365
Shwachman-Diamond syndrome, DNAJC21 type	*DNAJC21*	AR				Pancytopenia				DNA		617052	IEM1371
Shwachman-Diamond syndrome, EIF6 type	*EIF6*	AR				Neutropenia Thrombocytopenia				DNA		602912	IEM1372
OTHER (n=15)													
Disorders of lysosome-related organelle biogenesis													
Hermansky-Pudlak syndrome type 1	*HPS1*	AR		Bleeding tendency						DNA		203300	IEM1383
Hermansky-Pudlak syndrome type 2	*AP3B1*	AR				Neutropenia Thrombocytopenia				DNA		608233	IEM1384

Supplementary Table S1. List of inherited metabolical diseases with hematologic phenotypes, laboratory investigation, treatment options (if applicable), OMIM references and IEMbase ID.

Disorder (n=264)	Gene	Inheritance	Abormal blood cell morphology	Coagulation abnormalities	Anemias	Abnormal blood count	Hypercoagulability	Marrow abnormality	Other	Diagnostic markers	Specific treatmnt	OMIM	IEMbase code
Hermansky-Pudlak syndrome type 5	*HPS5*	AR				Neutropenia				DNA		614074	IEM1387
Hermansky-Pudlak syndrome type 6	*HPS6*	AR		Platelet function, abnormal						DNA		614075	IEM1388
Hermansky-Pudlak syndrome type 7	*DTNBP1*	AR		Platelet function, abnormal						DNA		614075	IEM1389
Hermansky-Pudlak syndrome type 8	*BLOC1S3*	AR		Platelet function, abnormal						DNA		614077	IEM1390
Hermansky-Pudlak syndrome type 9	*BLOC1S6*	AR				Thrombocytopenia			Leukocytosis	DNA		614171	IEM1391
Hermansky-Pudlak syndrome type 10	*AP3D1*	AR				Neutropenia				DNA		617050	IEM1392
Chediak-Higashi syndrome	*LYST*	AR		Bleeding tendency		Neutropenia Pancytopenia				DNA		214500	IEM1393
Griscelli syndrome type 2	*RAB27A*	AR				Neutropenia Thrombocytopenia				DNA		607624	IEM1395
Disorders of choline neurotransmission													
Choreoacanthocytosis	*VPS13A*	AR	Acanthocytosis							DNA		200150	IEM1400
Disorders of the synaptic vesicle cycle													
Rabenosyn 5 deficiency	*RBSN*	AR							Macrocytosis	Microalbumine (U)		609511	IEM1462
Disorders of oxalate metabolism													
Glyoxylate reductase/hydroxypyruvate reductase deficiency	*GRHPR*	AR				Pancytopenia				Oxalic acid (U, P), Glyceric acid (U), Creatinine (P), Urea (P)		260000	IEM0905
Mitochondrial 4-hydroxy-2-oxoglutarate aldolase 1 deficiency deficiency	*HOGA1*	AR				Pancytopenia				Oxalic acid (P,U), Organic acids (U)		613616	IEM0906
Alanine-glyoxylate aminotransferase deficiency (peroxisomal)	*AGXT*	AR				Pancytopenia				Oxalic acid (U, P), Glycolic acid (P, U), Creatinine (P), Urea (P)	Liver or liver/kidney transplant	259900	IEM0903