Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS (n=60)											
Disorders of pyrimidine metabolism											
Dihydroorotate dehydrogenase deficiency	*DHODH*							Coloboma	N-Carbamyl asparate (U), Purines and pyrimidines (U),		263750;126064
Deoxythymidylate kinase deficiency	*DTYMK*							Blindness, cortical	Amino acids (P), Lactate (P, CSF)		188345
Disorders of purine metabolism											
Phosphoribosyl pyrophosphate synthetase 1 deficiency	*PRPS1*			X				Vision, progressive loss	Lactate (P), DNA	Dietary reduction of red and organ meets, fish; Allopurinol 5-10 mg/kg/day; S-adenosylmethionine 30 mg/kg/day	300661
Inosine-5'-monophosphate dehydrogenase deficiency	*IMPDH1*				X	X		Vision loss	Lactate (P), DNA		146690
Inosine-5'-triphosphate pyrophosphohydrolase deficiency	*ITPA*		X						ITP (RBC)		147520
AICAR transformylase/IMP cyclohydrolase deficiency	*ATIC*							Blindness	Purines and pyrimidines (U)		608688;601731
Phosphoribosylaminoimidazole carboxylase deficiency	*PAICS*							Hypertelorism	DNA		172439
Disorders of glutathione metabolism											
Glutathione synthetase deficiency	*GSS*	X			X				5-Oxoproline (U), Glutathione (rbc), Hemoglobin (B)	Na bicarbonate to treat metabolic acidosis, antioxidants (vitamin C, E), avoid drugs like acetylsalycylic acid, phenopbarbital, sulfonamides)	266130
Glutathione reductase deficiency	*GSR*		X						Glutathione (P), Bilirubin (P)		138300
Disorders of ammonia detoxification											
Ornithine transcarbamylase deficiency	*OTC*							Vision, impaired	Orotic acid (U), Ammonia (B), Amino acids (P)	Dietary protein restriction, citrulline po administration 170 mg/kg/day or 3.8g/m2/day (in acute phase Arginine IV infusion 200 mg/kg or 4000 mg/m2); NH3 scavengers (in acute phase Na phenylacetate and Na benzoate, for chronic management Na phenylbutyrate <20 kg: ≤250mg/kg/dc,d >20 kg: 5g/m2/d maximum: 12g/day); liver transplantation; gene therapy in clinical trials	311250
Mitochondrial ornithine transporter deficiency, HHH	*SLC25A15*							Vision, impaired	Orotic acid (U), Ammonia (B), Homocitrulline (U), Amino acids (P)	Dietary protein restriction, essential amino acid supplementation; Na benzoate 250mg/kg/d maximum: 12g/d; Na phenylbutyrate 250mg/kg/d maximum: 12g/d; L-Arginine <20 kg: 100-200mg/kg/d >20 kg: 2.5-6g/m2/d maximum: 6g/d; L-Citrulline 100-200mg/kg/d maximum: 6g/d	238970
Disorders of amino acid transport											
Neuronal system A amino acid transporter deficiency	*SLC38A8*	X	X	X	X	X	X	Diminished visual activity	Lactate (P), DNA		609218
Lysosomal cationic amino acid transporter deficiency	*SLC7A14*				X			Blindness	DNA		615725
Taurine transporter deficiency	*SLC6A6*				X				Amino acids (P, U)		186854
Aminoacylase deficiencies											
Aspartoacylase deficiency	*ASPA*			X		X		Blindness	Organic acids (U)		271900
Disorders of monoamine metabolism											
Tyrosine hydroxylase deficiency	*TH*					X			Prolactin (P); Biogenic amines (CSF)	L-dopa/Carbidopa	191290
Aromatic L-amino acid decarboxylase deficiency	*DDC*					X			3-O-Methylodopa (DBS,P), Organic acids (U), Biogenic amines (CSF)	Pyridoxine, dopa agonist, MAO inhibitor, central anticholinergic	608643
Dopamine beta-hydroxylase deficiency	*DBH*					X		Eyes opening, delayed	Catecholamines (U, P), Biogenic amines (CSF)	Droxidopa	223360
Dopamine transporter deficiency	*SLC6A3*					X			Organic acids (U), Biogenic amines (CSF)	Dopamine agonists	613135;126455
Dopamin-serotonin vesicular transport defect	*SLC18A2*					X			Catecholamines (U, P), Biogenic amines (CSF)	Dopamine agonists	193001
Disorders of tetrahydrobiopterin metabolism											
Autosomal dominant GTP cyclohydrolase 1 deficiency	*GCH1*					X			Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	L-dopa/Carbidopa	600225
6-Pyruvoyl-tetrahydropterin synthase deficiency	*PTS*					X			Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	Tetrahydrobiopterin, L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; +/- folinic acid	261640
Sepiapterin reductase deficiency	*SPR*					X			Pterins (U, CSF), Biogenic amines (CSF)	L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; MAO inhibitor, serotonin reuptake inhibitor, dopamine agonist, anticholinergics, melatonin	182125
Disorders of tyrosine metabolism											

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Tyrosinase deficiency	*TYR*				X	X		Translucent iris, Absent iris pigment, Absent retinal pigment, Albinotic optic disc, Blue-gray irides, Photophobia, Pink irides	DNA		203100
Tyrosine aminotransferase deficiency	*TAT*	X						Absent iris pigment, Lacrimation	Amino acids (P), Organic acids (U)	Tyr-restricted diet	276600
Homogentisate 1,2 - dioxygenase deficiency, Alkaptonuria	*HGD*							Scleral pigmentation	Homogentisate (U)	Nitisinone	203500
4-hydroxyphenylpyruvate dioxygenase-like protein deficiency	*HPDL*							Vision, decreased	DNA		619026
Disorders of sulfur amino acid and sulfide metabolism											
Methionine adenosyltransferase I/III deficiency	*MAT1A*					X			SAM & SAH (P), Amino acids (P)		250850
S-adenosylhomocysteine hydrolase deficiency	*AHCY*					X			SAM & SAH (P), Amino acids (P)	Methionine or protein restricted diet, phosphatidylcholine and creatine supplements, liver transplant	613752
Cystathionine beta-synthase deficiency	*CBS*		X				X	Iridodonesis	SAM & SAH (P), Amino acids (P)	Pyridoxine 10 mg/kg/day (max 500 mg/day); vitamin B12, folate, low protein diet, +/- cysteine; betaine 50 mg/kg twice daily up to 150-200 mg/kg/day; enzyme therapy in clinical trials	236200
Isolated sulfite oxidase deficiency	*SUOX*		X						a-aminosemialdehyde (CSF), PLP (CSF), Sulfite (U), Amino acids (P)	Trials of dietary therapy (no proven benefit) low-protein diet restricted in cysteine and methionine; experimental (with minimal or no benefits): betaine, thiamine, cysteamine, penicillamin	272300
Disorders of branched-chain amino acid metabolism											
Branched-chain aminotransferase 2 deficiency	*BCAT2*					X			Amino acids (P)		238340;113530
3-Methylglutaconyl-CoA hydratase deficiency	*AUH*			X		X			Organic acids (U), Acylcarnitines (P,DBS)	Protein restriction, L-carnitine	250950
17-beta-hydroxysteroid dehydrogenase type 10 deficiency	*HSD17B10*							Vision, decreased	Organic acids (U), Acylcarnitines (P,DBS), Lactate (P), Glucose (S)	Protein restricted diet, L-Carnitine	300438
Propionic acidemia due to propionyl-CoA carboxylase subunit alpha deficiency	*PCCA*			X					Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine	232000
Propionic acidemia due to propionyl-CoA carboxylase subunit beta deficiency	*PCCB*			X					Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine	232000
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*			X					Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, L-Carnitine 100-200 mg/kg/day, vitamin B12, acute management of hyerammonemic crises, orthotopic liver transplant	251000
Disorders of proline and ornithine metabolism											
Delta-1-pyrroline-5-carboxylate synthase deficiency, spastic paraplegia phenotype	*ALDH18A1*	X	X						Amino acids (P), Ammonia (P)	Arginine 150 mg/kg/day	219150
Pyrroline-5-carboxylate reductase 1 deficiency	*PYCR1*							Hypotelorism, Deeply set eyes, Blue sclerae	DNA		612940;614438
Pyrroline-5-carboxylate reductase 2 deficiency	*PYCR2*					X		Upslanting palpebral fissures, Downslanting palpebral fissures	DNA		616420
X-prolyl aminopeptidase 3 deficiency	*XPNPEP3*				X				DNA		613159
Ornithine aminotransferase deficiency	*OAT*		X		X		X	Blindness	Guanidino compounds (U,P,CSF), Amino acids (P)	Low protein diet	258870
Disorders of β- and γ-amino acids											
Dihydropyrimidine dehydrogenase deficiency	*DPYD*			X		X		Coloboma	Purines and pyrimidines (U)	No treatment in pediatrics, in adults discontinue fluorouracil treatment	274270;612779
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*					X			Organic acids (U)	Vigabatrin (no proven benefit)	271980;610045
Disorders of glutamate metabolism											
Glutamate aspartate transporter deficiency	*SLC1A3*							Photophobia	DNA		612656
Ionotropic glutamate receptor NMDA type subunit 1 dysregulation	*GRIN1*					X		Blindness, cortical	DNA		614254;617820
Ionotropic glutamate receptor NMDA type subunit 2B dysregulation	*GRIN2B*					X			DNA		616139;613970
Ionotropic glutamate receptor NMDA type subunit 2D superactivity	*GRIN2D*							Visual impairment	DNA		617162
Ionotropic glutamate receptor delta type subunit 2 deficiency	*GRID2*					X			DNA		616204
Ionotropic glutamate receptor AMPA type subunit 3 deficiency	*GRIA3*							Deeply set eyes	DNA		300699
Ionotropic glutamate receptor AMPA type subunit 4 dysregulation	*GRIA4*					X			DNA		617864
Metabotropic glutamate receptor 1 superactivity	*GRM1*					X			DNA		617691

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Metabotropic glutamate receptor 6 deficiency	*GRM6*				X		X	Hemeralopia	DNA		257270
Disorder of glutamine metabolism											
Glutaminase deficiency	*GLS*		X						Amino acids (P)		618328
Disorder of asparagine metabolism											
Asparagine synthetase deficiency	*ASNS*							Blindness, cortical	Amino acids (P)		615574
Disorders of serine metabolism											
3-phosphoglycerate dehydrogenase deficiency	*PHGDH*		X			X			5-Methyl-THF (CSF), Amino acids (P)	L-serine (500-700 mg/kg/day); glycine (200 mg/kg/day)	606879;601851
Disorders of glycine metabolism											
Nonketotic hyperglycinemia due to glycine decarboxylase deficiency	*GLDC*			X					Amino acids (P)	Na benzoate 200-550 mg/kg/day to max 750 mg/kg/day or 5.5 g/m2 BSA in adults up to 16.5 g/m2/day in severe cases; dextrometorphan 3 to 15 mg/kg/day or ketamine	238300
Nonketotic hyperglycinemia due to aminomethyltransferase deficiency	*AMT*			X					Amino acids (P)	Na benzoate 200-550 mg/kg/day to max 750 mg/kg/day or 5.5 g/m2 BSA in adults up to 16.5 g/m2/day in severe cases; dextrometorphan 3 to 15 mg/kg/day or ketamine	605899
Glycine receptor subunit beta deficiency	*GLRB*					X	X		DNA		614619
Glycine encephalopathy due to H protein deficiency	*GCSH*			X					Amino acids (P)		605899
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS (n=59)											
Disorders of lipoic acid and iron-sulfur metabolism											
BOLA3 deficiency	*BOLA3*			X					Lactate (P), Amino acids (P)		614299
Glutaredoxin 5 deficiency	*GLRX5*			X		X			Ferritin (S), Transferrin (S), Lactate (P), Amino acids (P)		205950
IBA57 deficiency	*IBA57*			X				Visual impairment	Lactate (P), Amino acids (P)		615330
ISCA1 deficiency	*ISCA1*				X	X			Lactate (P), Amino acids (P)		617613
ISCA2 deficiency	*ISCA2*			X		X			5-Methyl-THF (CSF), Lactate (P), Amino acids (P)		616370
Ferredoxin reductase deficiency	*FDXR*			X	X	X		Visual impairment	Complexes I - IV activity (muscle); DNA		617717
Ferredoxin 2 deficiency	*FDX2*			X		X			Myoglobin (U), Lactate (P), Organic acids (U)		614585
Disorders of cobalamin metabolism											
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*				X	X		Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin 1 mg IM daily	277380
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*				X	X		Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin 1 mg IM daily	614857
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*				X	X		Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	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
Epi-cblC	*MMACHC; PRDX1*				X			Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin 1 mg IM daily	609831;176763
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*				X				SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin, betaine	277410
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*			X					SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin, betaine	277410
Methionine synthase reductase deficiency-cblE	*MTRR*					X		Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U)	Hydroxycobalamin 1 mg IM daily, betaine 250 mg/kg/day; folinic acid 5-30 mg/day	236270
Methionine synthase deficiency	*MTR*					X		Vision, impaired	SAM (P,CSF), Amino acids (P), Organic acids (U)	Hydroxycobalamin 1 mg IM daily, betaine 250 mg/kg/day; folinic acid 5-30 mg/day	250940
Methylmalonic aciduria, cblA type	*MMAA*			X					Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin injections	251100
Methylmalonic aciduria, cblB type	*MMAB*			X					Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin injections	251110
Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*							Vision, impaired	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)		309541
Methylmalonic aciduria and homocystinuria due to ZNF143 deficiency	*ZNF143*							Vision, impaired	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)		603433

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Methylmalonic aciduria and homocystinuria due to Ronin deficiency	*THAP11*							Vision, impaired	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)		609119
Disorders of folate metabolism											
Dihydrofolate reductase deficiency	*DHFR*					X			LDH (P), Hb (B), Folates (B, CSF)	Folinic acid	126060
Disorders of pyridoxine metabolism											
Pyridoxal kinase deficiency	*PDXK*			X					DNA		618511
Disorders of biotin metabolism											
Biotinidase deficiency	*BTD*	X		X					Acyglycines (U), Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)	Biotin 5-10 mg/day	253260
Disorders of thiamine metabolism											
Thiamine transporter 1 deficiency	*SLC19A2*			X					Lactate (P), Glucose (S)	Thiamine	603941
Thiamine transporter 2 deficiency	*SLC19A3*					X			Lactate (P), Organic acids (U)	Thiamine, biotin	606152
Disorders of riboflavin metabolism											
Riboflavin transporter 3 deficiency	*SLC52A3*							Vision loss	Flavins (B), Organic acids (U), Acylcarnitines (P,DBS)		211500
Riboflavin transporter 2 deficiency	*SLC52A2*							Vision loss	Flavins (B), Organic acids (U), Acylcarnitines (P,DBS)		614707
Flavin adenine dinucleotide synthetase deficiency (early onset)	*FLAD1*		X			X			Flavins (B), Acyglycines (U), Organic acids (U), Acylcarnitines (P,DBS)		255100
Disorders of niacin and NAD metabolism											
Nicotinamide mononucleotide adenylyl transferase 1 deficiency	*NMNAT1*				X	X	X	Oculo-digital sign, Coloboma, Blindness, Slow or near-absent pupillary reactions	DNA		608553
Mitochondrial NAD kinase 2 deficiency	*NADK2*			X					Lactate (P), Amino acids (P), Acylcarnitines (P,DBS)		616034
Disorders of pantothenate metabolism											
Pantothenate kinase 2 deficiency	*PANK2*			X	X	X		Vision, impaired	Iron (brain), DNA	Possible iron chelation, possible panthotenate	234200
Disorders of vitamin A metabolism											
Plasma retinol-binding protein deficiency (recessive)	*RBP4*	X	X	X	X			Visual loss, Microphthalmia, Microcoria, Displacement of pupil, inferior, Coloboma of the iris, inferior, Coloboma, Anophthalmia, uni- or bilateral	Vitamin A (P)		616428;615147
Vitamin A receptor deficiency	*STRA6*							Microphthalmia, Coloboma, Blepharophimosis, Anophthalmia, uni- or bilateral	DNA		601186
Lecithin retinol acyltransferase deficiency	*LRAT*			X	X	X		Slow or near-absent pupillary reactions, Photophobia, Diminished visual activity	DNA		613341
Retinal isomerase deficiency	*RPE65*			X	X	X	X	Visual impairment, Hypopigmented fundus, Blindness	DNA	Voretigene neparvovec-rzyl	204100;613794
Retinol dehydrogenase 5 deficiency	*RDH5*				X				DNA		136880
Retinol dehydrogenase 12 deficiency	*RDH12*				X			Blindness	DNA		612712
Interphotoreceptor retinol-binding protein deficiency	*RBP3*		X	X	X			Diminished visual activity	DNA		615233
Retinaldehyde dehydrogenase 3 deficiency	*ALDH1A3*			X	X			Microphthalmia, Entropion, Coloboma	DNA		615113
Cellular retinaldehyde-binding protein deficiency	*RLBP1*				X				DNA		180090
Retinoic acid receptor β deficiency	*RARB*							Microphthalmia	DNA		615524
Retinol dehydrogenase 11 deficiency	*RDH11*		X		X			Diminished visual activity	DNA		616108
Disorders of vitamin K metabolism											
Menaquinone-4 synthetase deficiency	*UBIAD1*	X							DNA		121800
Disorders of molybdenum metabolism											
Cyclic pyranopterin monophosphate synthase deficiency	*MOCS1*		X					Blindness, cortical	Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)	cPMP (fosdenopterin)	603707

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Molybdopterin synthase deficiency	*MOCS2*		X					Blindness, cortical	Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)		603708
Gephyrin deficiency	*GPHN*		X					Blindness, cortical	Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)		603930
Disorders of copper metabolism											
Copper-transporting ATPase subunit alpha deficiency (Menkes disease)	*ATP7A*		X	X	X			Ptosis	Copper (S, U), Ceruloplasmin (S)	Copper chloride or L-histidine 350-500 ug/day IV or SC	309400
Copper-transporting ATPase subunit beta deficiency (Wilson disease)	*ATP7B*	X	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
Acetyl-CoA transporter deficiency	*SLC33A1*		X						Copper (S, U), Ceruloplasmin (S)		614482
MEDNIK-like syndrome	*AP1B1*							Photophobia	Copper (S, U), Ceruloplasmin (S)		242150
Deficiency of copper chaperone for superoxide dismutase	*CCS*		X						DNA		603864
Disorders of iron metabolism											
Ferroportin 1 deficiency	*SLC40A1*		X						Ferritin (S), Transferrin saturation		606069
Ferritin light chain dysregulation	*FTL*		X						Ferritin (S), Transferrin saturation		600886
Disorders of manganese metabolism											
Spondylocheirodysplastic Ehlers-Danlos syndrome	*SLC39A13*							Protuberant eyes, Downslanting palpebral fissures, Blue sclerae	Lysyl pyridinoline (U), Hydroxylysyl pyridinoline (U)		612350
Birk-Landau-Perez syndrome	*SLC30A9*					X			Potassium (P)		617595
Disorders of zinc metabolism											
SLC39A8 deficiency	*SLC39A8*					X			Zinc (S, U), Manganese (B), Lactate (P), Sialotrasferrins (S)	Uridine + galactose, manganese	616721
Disorders of magnesium metabolism											
Claudin 10 deficiency	*CLDN10*							Alacrima	Calcium (P,U), Magnesium (B, U), Renin (P), 25-Hydroxy-Vitamin D (P)		617671
Claudin 16 deficiency	*CLDN16*					X	X		Magnesium (P,U), Calcium (P,U), Uric acid (P), Organic acids (U)	Magnesium replacement	248250
Claudin 19 deficiency	*CLDN19*				X	X	X		Magnesium (P,U), Calcium (P,U)	Magnesium replacement	248190
DISORDERS OF CARBOHYDRATES (n=18)											
Disorders of carbohydrate transport and absorption											
Glucose transporter 2 deficiency (Fanconi-Bickel syndrome)	*SLC2A2*		X						Chemistry (P,U), Oligosaccharides (U), Amino acids (P)	Corn starch, electrolyte replacement	227810
Disorders of galactose and fructose metabolism											
Galactose-1-phosphate uridyltransferase deficiency	*GALT*		X						Chemistry (P,U), Oligosaccharides (U), Amino acids (P)	Galactose restriction	230400
Galactose epimerase deficiency	*GALE*		X						Chemistry (P,U), Oligosaccharides (U), Amino acids (P)		230350
Galactokinase deficiency	*GALK1*		X						Oligosaccharides (U)		230200
Triokinase/FMN cyclase deficiency	*TKFC*		X					Microphthalmia	Lactate (P)		618805
Disorders of the pentose phosphate pathway and polyol metabolism											
Ribose-5-phosphate isomerase deficiency	*RPIA*			X	X	X		Visual impairment	Polyols (P,U,CSF)		608611
Transketolase deficiency	*TKT*		X			X		Uveitis, Conjunctivitis	Polyols (P,U), Sugar phosphates (U)		617044
Sorbitol dehydrogenase deficiency	*SORD*		X						Polyols (P,U)		182500
Disorders of insulin secretion and signaling											
Kabuki syndrome	*KDM6A*					X			DNA		300867

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Disorders of glycogen metabolism											
HOIL1 deficiency	*RBCK1*					X			DNA		615895
Lysosome-associated membrane protein 2 deficiency (Danon disease)	*LAMP2*		X		X		X		Glycogen (muscle), Myocytes, vacuolated		300257
Laforin deficiency	*EPM2A*							Visual hallucinations, Vision loss	DNA		254780
Malin deficiency	*NHLRC1*							Visual hallucinations, Vision, progressive loss	DNA		254780
Liver glycogenin 2 deficiency	*GYG2*					X			DNA		300198
Disorders of gluconeogenesis											
Cytosolic phosphoenolpyruvate carboxykinase deficiency	*PCK1*			X					Lactate (P), Amino acids (P), Organic acids (U)		261680
Disorders of glycolysis											
Retinitis pigmentosa type 79	*HK1*			X	X			Photophobia, Diminished visual activity, Color vision deficit, Bull's-eye maculopathy	DNA		617460
Neurodevelopmental disorder with visual defects and brain anomalies (NEDVIBA)	*HK1*			X	X				DNA		617460
Phosphoglycerate kinase deficiency	*PKG1*				X				Reticulocytes (B)		300653
MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM (n=136)											
Disorders of pyruvate metabolism											
Mitochondrial pyruvate carrier deficiency	*MPC1*					X			Pyruvate (P), Glucose (P), Lactate (P), Organic acids (U)		614741
Disorders of the Krebs cycle											
Mitochondrial aconitase deficiency	*ACO2*			X	X	X			Organic acids (U)		614559
Mitochondrial NADPH-dependent isocitrate dehydrogenase 3 β subunit deficiency	*IDH3B*			X	X			Diminished visual activity	DNA		612572
Fumarase deficiency	*FH*			X				Vision, impaired	Lactate (P), Organic acids (U)		150800
Mitochondrial malate dehydrogenase deficiency	*MDH2*				X				Lactate (P), Organic acids (U)		617339
Isocitrate dehydrogenase 1 deficiency	*IDH1*					X		Telecanthus, Short palpebral fissures			147700
Mitochondrial NAD+-dependent isocitrate dehydrogenase 3 subunit alpha deficiency	*IDH3A*	X			X	X		Visual impairment	DNA		619007
Disorders of mitochondrial carriers											
Adenine nucleotide translocator deficiency AR	*SLC25A4*		X			X			Lactate (P)		609283
Mitochondrial citrate carrier deficiency	*SLC25A1*					X		Blindness, cortical	Organic acids (U)		615182
Mitochondrial ATP-Mg/phosphate transporter deficiency	*SLC25A24*						X	Short palpebral fissures, Downslanting palpebral fissures, Deeply set eyes	DNA		612289
Disorders of complex I subunits											
NADH dehydrogenase flavoprotein 1 deficiency	*NDUFV1*					X			Lactate (P)		618225
NADH dehydrogenase flavoprotein 2 deficiency	*NDUFV2*			X					Lactate (P)		618229
NADH dehydrogenase iron-sulfur protein 1 deficiency	*NDUFS1*			X		X			Lactate (P)		618229
NADH dehydrogenase iron-sulfur protein 2 deficiency	*NDUFS2*					X			Lactate (P)		618228
NADH dehydrogenase iron-sulfur protein 8 deficiency	*NDUFS8*					X			Lactate (P)		618222
NADH dehydrogenase alpha subcomplex subunit 1 deficiency	*NDUFA1*					X			Lactate (P)		301020
NADH dehydrogenase alpha subcomplex subunit 9 deficiency	*NDUFA9*				X				Lactate (P)		256000
NADH dehydrogenase beta subcomplex subunit 11 deficiency	*NDUFB11*							Microphthalmia	Lactate (P)		252010

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
NADH dehydrogenase core subunit 1 deficiency	*MT-ND1*			X					Lactate (P)		252010
NADH dehydrogenase core subunit 2 deficiency	*MT-ND2*			X					Lactate (P)		252010
NADH dehydrogenase core subunit 3 deficiency	*MT-ND3*			X		X			Lactate (P)		252010
NADH dehydrogenase core subunit 4 deficiency	*MT-ND4*			X					Lactate (P)		252010
NADH dehydrogenase core subunit 4L deficiency	*MT-ND4L*			X					Lactate (P)		252010
NADH dehydrogenase core subunit 5 deficiency	*MT-ND5*			X					Lactate (P)		252010
NADH dehydrogenase core subunit 6 deficiency	*MT-ND6*			X					Lactate (P)		252010
NADH dehydrogenase alpha subcomplex subunit 13 deficiency	*NDUFA13*			X	X	X			Lactate (P)		252010
Complex I assembly factor 8 deficiency	*NDUFAF8*			X					Lactate (P)		618776
Disorders of complex I assembly											
Complex I assembly factor 2 deficiency	*NDUFAF2*			X		X			Lactate (P)		618233
Complex I assembly factor 3 deficiency	*NDUFAF3*			X					Lactate (P)		618240
FOXRED1 deficiency	*FOXRED1*							Blindness, cortical	Lactate (P)		618241
NUBPL deficiency	*NUBPL*					X			Lactate (P)		618242
TIMMDC1 deficiency	*TIMMDC1*					X			Lactate (P)		615534
Complex I assembly factor 7 deficiency	*NDUFAF7*			X				Vision, progressive loss, Posterior staphyloma	DNA		615898
VAC14 deficiency type 1	*VAC14*					X			DNA		216340
Dynamin 2 deficiency	*DNM2*					X			DNA		602378
DNAJC30 deficiency	*DNAJC30*				X				DNA	Idebenone	619382
Disorders of complex II subunits											
Succinate dehydrogenase subunit D deficiency	*SDHD*				X	X			Lactate (P), Organic acids (U)		168000
Disorders of complex II assembly											
Succinate dehydrogenase complex assembly factor 1 deficiency	*SDHAF1*				X	X			Lactate (P), Organic acids (U)		252011
Disorders of complex IV subunits											
Cytochrome c oxidase subunit 2 deficiency	*MT-CO2*				X				Lactate (P)		516040
Disorders of complex IV assembly and ancillary proteins											
SCO2 deficiency	*SCO2*					X			Lactate (P)		604377;608908
SURF1 deficiency	*SURF1*			X		X			Lactate (P)		256000;616684
TACO1 deficiency	*TACO1*			X					Lactate (P)		220110
PET100 deficiency	*PET100*		X					Visual impairment	Lactate (P)		220110
FASTKD2 deficiency	*FASTKD2*			X					Lactate (P)		220110
APOPT1 deficiency	*APOPT1*					X		Visual loss	DNA		220110
CEP89 deficiency	*CEP89*		X						Lactate (P), Amino acids (P)		615470
NDUFA4 deficiency	*NDUFA4*			X		X			Lactate (P)		619065
Disorders of complex V subunits											

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Mitochondrial ATP synthase F1 subunit alpha deficiency	*ATP5F1A*					X			Lactate (P), Amino acids (P)		616045;615228
Mitochondrial ATP synthase F0 subunit 6 deficiency	*MT-ATP6*			X	X	X		Blindness	Lactate (P)		516060
DAPIT deficiency	*ATP5MD*					X			Lactate (P), Amino acids (P)		615204
Disorders of complex V assembly											
Transmembrane protein 70 deficiency	*TMEM70*		X						Ammonia (B), Lactate (P), Organic acids (U)		614052
Disorders of mitochondrial cytochrome synthesis and incorporation											
Mitochondrial cytochrome b deficiency	*MT-CYB*		X			X		Visual impairment	Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)		516020
Holocytochrome c synthase deficiency	*HCCS*	X	X					Microphthalmia, Anophthalmia, uni- or bilateral	DNA		309801
Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication											
Mitochondrial DNA polymerase gamma catalytic subunit deficiency	*POLG*					X			Lactate (P), Organic acids (U)		607459
Mitochondrial DNA polymerase gamma accessory subunit deficiency	*POLG2*					X			Lactate (P)		610131
Mitochondrial deoxyguanosine kinase deficiency	*DGUOK*					X			a-fetoprotein (S), Lactate (P), Amino acids (P), Sialotrasferrins (S)		251880;601465
MPV17 deficiency	*MPV17*	X			X	X			Lactate (P)		256810
TWINKLE mitochondrial DNA helicase deficiency	*TWNK*					X			DNA		271245;616138
Mitochondrial thymidine kinase 2 deficiency	*TK2*					X			CK (P), Lactate (P)		609560;188250
Mitochondrial ribonucelotide reductase subunit 2 deficiency	*RRM2B*					X			Lactate (P), Amino acids (P)		604712
DNA2 helicase deficiency	*DNA2*					X			DNA		615156
Mitochondrial ribonuclease H1 deficiency	*RNASEH1*					X			Lactate (P)		615156
Mitochondrial genome maintenance exonuclease 1 deficiency	*MGME1*					X			DNA		615084
FBXL4 deficiency	*FBXL4*		X						Lactate (P)		615471
Topoisomerase 3α deficiency	*TOP3A*					X		Vision, impaired	DNA		618098
Single-stranded DNA-binding protein 1 deficiency	*SSBP1*			X	X			Visual loss	DNA		165510
Disorders of mitochondrial transcription and RNA transcript processing											
Mitochondrial RNA import protein deficiency	*PNPT1*					X			Lactate (P)		614932;614934
Mitochondrial poly(A) polymerase deficiency	*MTPAP*			X		X			DNA		613672
CCA-adding tRNA-nucleotidyltransferase deficiency	*TRNT1*				X				IgG (serum), Amino acids (P)		616959
Mitochondrial methionyl-tRNA formyltransferase deficiency	*MTFMT*			X		X			Lactate (P)		614947
tRNA 5-taurinomethyluridine modifier deficiency	*GTPBP3*							Visual impairment	Lactate (P)		616198
tRNA 5-carboxymethylaminomethyl transferase deficiency	*MTO1*			X					Lactate (P)		614702
Mitochondrial RNA-processing endoribonuclease deficiency	*RMRP*							Hypotelorism	DNA		607095;250250;250460
Mitochondrial RNA polymerase deficiency	*POLRMT*					X		Hypotelorism	Lactate (P), 5-Methyl-THF (CSF)		601778
Mitochondrial ribosomopathies											
Mitochondrial ribosomal large subunit 44 deficiency	*MRPL44*				X				DNA		615395
Mitochondrial ribosomal small subunit 2 deficiency	*MRPS2*					X		Short palpebral fissures	Lactate (P), Organic acids (U)		617950
Mitochondrial ribosomal small subunit 28 deficiency	*MRPS28*		X			X			Lactate (P), Organic acids (U)		611990

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Disorders of mitochondrial translation factors											
Mitochondrial elongation factor G1 deficiency	*GFM1*					X			Lactate (P)		609060
Mitochondrial elongation factor Ts deficiency	*TSFM*			X					CK (P), Lactate (P)		610505
C12orf65 release factor deficiency	*C12ORF65*			X		X			Lactate (P)		613559
Disorders of mitochondrial tRNA											
Mitochondrial tRNA(Arg) deficiency	*MT-TR*					X			Lactate (P)		590005
Mitochondrial tRNA(Asn) deficiency	*MT-TN*					X			Lactate (P)		590010
Mitochondrial tRNA(Cys) deficiency	*MT-TC*					X			Lactate (P)		590020
Mitochondrial tRNA(His) deficiency	*MT-TH*			X	X				Lactate (P)		590040
Mitochondrial tRNA(Leu) 1 deficiency	*MT-TL1*				X				Lactate (P)		590050
Mitochondrial tRNA(Leu) 2 deficiency	*MT-TL2*					X			Lactate (P)		590055
Mitochondrial tRNA(Phe) deficiency	*MT-TF*				X			Photophobia	Lactate (P)		590070
Mitochondrial tRNA(Ser) 2 deficiency	*MT-TS2*		X						Lactate (P)		590085
Mitochondrial tRNA(Trp) deficiency	*MT-TW*			X		X			Lactate (P)		590095
Mitochondrial tRNA(Tyr) deficiency	*MT-TY*			X		X			Lactate (P)		590100
Mitochondrial tRNA(Val) deficiency	*MT-TV*		X		X				Lactate (P)		590105
Disorders of mitochondrial tRNA incorporation and recycling											
Mitochondrial alanyl-tRNA synthetase deficiency	*AARS2*					X			Lactate (P)		614096;615889
Mitochondrial arginyl-tRNA synthetase deficiency	*RARS2*							Visual impairment	Lactate (P)		611523
Mitochondrial asparaginyl-tRNA synthetase deficiency	*NARS2*					X			Lactate (P)		616239
Mitochondrial glutamyl-tRNA synthetase deficiency	*EARS2*					X		Visual impairment	Lactate (P)		614924
Mitochondrial isoleucyl-tRNA synthetase deficiency	*IARS2*		X			X			Lactate (P)		616007
Mitochondrial methionyl-tRNA synthetase deficiency	*MARS2*					X			Lactate (P)		611390
Mitochondrial phenylalanyl-tRNA synthetase deficiency	*FARS2*					X		Visual impairment	Lactate (P)		614946
Mitochondrial valyl-tRNA synthetase deficiency	*VARS2*					X	X		Lactate (P)		615917
Mitochondrial tryptophanyl-tRNA synthetase deficiency	*WARS2*			X	X	X			Lactate (P)		617710
Peptidyl-tRNA hydrolase 2 deficiency	*PTRH2*					X			DNA		616263
Mitochondrial prolyl-tRNA synthetase deficiency	*PARS2*			X				Visual impairment	Lactate (P)		612036
Disorders of mitochondrial fission											
Dynamin-like protein 1 deficiency	*DNM1L*			X		X			VLCFA (P), Lactate (P)		614388
Mitochondrial fission factor deficiency	*MFF*			X		X		Vision loss	Lactate (P), DNA		617086
UGO-1 like protein deficiency	*SLC25A46*			X		X		Vision, progressive loss	Lactate (P), Organic acids (U)		616505
Disorders of mitochondrial fusion											
OPA1 deficiency	*OPA1*			X		X		Vision loss, onset, Scotomata, Glaucoma, Color vision deficit	Lactate (P)		125250
OPA3 deficiency	*OPA3*		X	X	X	X		Visual impairment	Organic acids (U)		258501

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Mitofusin 2 deficiency	*MFN2*			X					DNA		609260;617087
MSTO1 deficiency	*MSTO1*			X	X				DNA		617675
Disorders of mitochondrial phospholipid metabolism											
Acylglycerol kinase deficiency	*AGK*		X						Lactate (P), Organic acids (U)		212350;614691
Phosphatidylserine decarboxylase deficiency	*PISD*		X						DNA		612770
Disorders of mitochondrial protein import											
TIMM14 deficiency	*DNAJC19*			X					Organic acids (U)		610198
Mohr-Tranebjaerg syndrome	*TIMM8A*			X				Vision, impaired, Blindness	DNA		304700
TIMM50 deficiency	*TIMM50*			X					Lactate (P), Organic acids (U)		607381
GFER deficiency	*GFER*		X		X				Lactate (P)		613076
Disorders of mitochondrial protein quality control											
Mitochondrial processing peptidase alpha deficiency	*PMPCA*		X			X			DNA		213200
Mitochondrial processing peptidase β deficiency	*PMPCB*			X					Lactate (P)		617954
Mitochondrial intermediate peptidase deficiency	*MIPEP*		X						Lactate (P), Organic acids (U)		617228
CLPB deficiency	*CLPB*		X						DNA		616271
LONP1 deficiency	*LONP1*		X			X			DNA		600373
HSP60 deficiency	*HSPD1*			X		X			Lactate (P), Organic acids (U)		612233;605280
Sacsin deficiency	*SACS*				X				DNA		270550
m-AAA protease AFG3L2 subunit deficiency	*AFG3L2*					X			DNA		614487;610246
Paraplegin deficiency	*SPG7*			X		X			DNA		607259
YME1L1 deficiency	*YME1L1*			X			X	Amblyopia	Lactate (P)		617302
Other disorders of mitochondrial homeostasis											
Sideroflexin 4 deficiency	*SFXN4*							Visual impairment	Lactate (P)		615578
ATAD3A deficiency	*ATAD3A*		X	X		X	X		DNA		617183
Transmembrane protein 126A deficiency	*TMEM126A*			X				Vision, progressive loss, Scotomata, Diminished visual activity	DNA		612989
C1q binding protein deficiency	*C1QBP*					X			Lactate (P)		617713
Nogo-interacting mitochondrial protein deficiency	*RTN4IP1*			X		X		Vision, impaired	DNA		616732
MICOS complex subunit MIC13 deficiency	*MICOS13*			X	X			Vision loss	Lactate (P), Organic acids (U)		616658
Mitochondrial thioredoxin 2 deficiency	*TXN2*			X	X				Lactate (P)		616811
Mitochondrial calcium uniporter 2 deficiency	*MICU2*					X			DNA		610632
Primary CoQ10 deficiencies											
Prenyl diphosphate synthase subunit 1 deficiency	*PDSS1*			X					DNA		607429;607426
Coenzyme Q2 polyprenyltranferase deficiency	*COQ2*			X					DNA		609825;607426
Coenzyme Q7 hydroxylase deficiency	*COQ7*							Visual impairment	Lactate (P), Organic acids (U)		616733
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=32)											

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Disorders of nucleotide and nucleic acid metabolism											
3' Repair exonuclease 1 deficiency	*TREX1*				X			Glaucoma	Interferon-α (CSF), Pterins (CSF)		192315
Ribonuclease H2 subunit B deficiency, AGS2	*RNASEH2B*							Glaucoma	Interferon-α (CSF), Pterins (CSF)	JAK inhibitors	610181
Ribonuclease H2 subunit C deficiency, AGS3	*RNASEH2C*							Glaucoma	Interferon-α (CSF), Pterins (CSF)	JAK inhibitors	610329
Ribonuclease H2 subunit A deficiency, AGS4	*RNASEH2A*							Glaucoma	Interferon-α (CSF), Pterins (CSF)	JAK inhibitors	610333
Ribonuclease T2 deficiency	*RNASET2*					X			Interferon-α (CSF)		612951
RNA-specific adenosine deaminase deficiency, AGS6	*ADAR*							Glaucoma	Interferon-α (CSF), Pterins (CSF)	JAK inhibitors	127400
MDA5 superactivity, AGS7	*IFIH1*							Glaucoma	Interferon-α (CSF), Pterins (CSF)		182250
ABCC6 deficiency	*ABCC6*				X			Visual impairment	Lactate (P), DNA		264800
Ectonucleotide pyrophosphatase/phosphodiesterase 1 deficiency	*ENPP1*				X				Lactate (P), DNA		613312
DDX58 superactivity	*DDX58*							Glaucoma	Phosphate (P,U)		616298
PRUNE1 deficiency	*PRUNE1*		X					Blindness, cortical	DNA		617481
RNA-specific adenosine deaminase 2 deficiency	*ADARB1*							Blindness, cortical	DNA		618862
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases											
tRNA splicing endonuclease subunit 2 deficiency	*TSEN2*							Visual impairment	DNA		612389
CLP1 deficiency	*CLP1*					X			DNA		615803
NSUN2 deficiency	*NSUN2*					X			DNA		611091
tRNA-specific adenosine deaminase 3 deficiency	*ADAT3*					X			DNA		615286
Elongator complex protein 1 deficiency	*ELP1*							Lacrimation	DNA		223900
Galloway-Mowat syndrome, YRDC type	*YRDC*							Blindness	DNA		612276
Galloway-Mowat syndrome, GON7 type	*GON7*							Blindness	DNA		617436
Galloway-Mowat syndrome type 2	*LAGE3*							Visual impairment	DNA		301006
Galloway-Mowat syndrome type 3	*OSGEP*							Hypertelorism	DNA		617729
Pseudouridine synthase 3 deficiency	*PUS3*					X			DNA		617051
Pseudouridine synthase 3 deficiency	*PUS3*					X			DNA		617051
Arginyl-tRNA synthetase 1 deficiency	*RARS1*					X			DNA		616140
Glutaminyl-tRNA synthetase 1 deficiency	*QARS1*							Hypotelorism	DNA		615760
Histidyl-tRNA synthetase 1 deficiency	*HARS1*				X	X		Visual loss	DNA		614504
Disorders of ribosomal biogenesis											
Treacher Collins syndrome type 1	*TCOF1*							Coloboma	DNA		154500
POLR3-related leukodystrophy	*POLR1C*						X		DNA		248390;616494
Treacher Collins syndrome type 4	*POLR1B*							Microtia	DNA		618939
POLR3K deficiency	*POLR3K*			X		X			DNA		619310
Disorders of choline neurotransmission											
Choline transporter deficiency	*SLC5A7*					X			DNA		617143

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Choline acetyltransferase deficiency	*CHAT*					X			DNA		254210
DISORDERS OF LIPIDS (n=77)											
Disorders of carnitine metabolism											
γ-Butyrobetaine hydroxylase deficiency	*BBOX1*					X		Long eye lashes	Acylcarnitines (P,DBS)	L-Carnitine	603312
Disorders of fatty acid oxidation and transport											
Long-chain hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*				X				Myoglobin (U), Acylcarnitines (P,DBS)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	609015
Trifunctional protein subunit α deficiency	*HADHA*				X				Organic acids (U), Acylcarnitines (P,DBS)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	609015
Trifunctional protein subunit β deficiency	*HADHB*				X				Organic acids (U), Acylcarnitines (P,DBS)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	609015
Lysophosphatidylcholine-esterified long-chain fatty acid transporter deficiency	*MFSD2A*							Upslanting palpebral fissures	Lysophosphatidylcholines (P)		616486
TANGO2 deficiency	*TANGO2*			X					Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)		616878
Disorders of fatty acid synthesis and elongation											
Mitochondrial enoyl-CoA reductase deficiency	*MECR*			X					Lactate (MRS), DNA		617282
Very long-chain fatty acid elongase 1 deficiency	*ELOVL1*			X	X	X	X	Visual loss, Photophobia	DNA		611813
Very long-chain fatty acid elongase 4 deficiency, retinal phenotype	*ELOVL4*				X				DNA		614457
Very long-chain fatty acid elongase 5 deficiency	*ELOVL5*					X			VLCFA (P)		615957
Mitochondrial malonyltransferase deficiency	*MCAT*			X		X		Visual loss	DNA		614479
Disorders of the fatty alcohol cycle											
Fatty aldehyde dehydrogenase deficiency (Sjogren-Larsson syndrome)	*ALDH3A2*				X				DNA		270200
Disorders of intracellular triglyceride metabolism											
Abhydrolase D5 deficiency	*ABHD5*		X						DNA		275630
Disorders of non-mitochondrial phospholipid metabolism											
Phosphocholine cytidylyltransferase 1α deficiency	*PCYT1A*				X			Diminished visual activity	Acetylcholine (P), Phosphocholine (P)		608940
Phosphatidylserine synthase 1 superactivity	*PTDSS1*							Ocular abnormalities	DNA		151050
DDHD2 deficiency	*DDHD2*			X		X		Telecanthus	DNA		609340
ABHD12 deficiency	*ABHD12*		X		X				VLCFA (P)		612674
Ethanolaminephosphotransferase 1 deficiency	*SELENOI*				X				DNA		607915
CTP-phosphoethanolamine cytidylyltransferase 2 deficiency	*PCYT2*							Visual impairment	DNA		618770
Disorders of non-lysosomal sphingolipid metabolism											
Serine palmitoyltransferase subunit 1 deficiency	*SPTLC1*		X		X				DNA		162400
Serine palmitoyltransferase subunit 2 deficiency	*SPTLC2*				X			Vision, impaired, Scotomata	DNA		613640
Nonlysosomal glucosylceramidase deficiency	*GBA2*		X			X			DNA		614409
Fatty acid 2-hydroxylase deficiency	*FA2H*			X		X			Iron (brain), DNA		612319
Sphingosine-1-phosphate lyase deficiency	*SGPL1*					X			ACTH (P), Sphingolipids (P)		617575
Ceramide transfer protein superactivity	*COL4A3BP*					X		Vision, impaired, Epicanthus, Upslanting palpebral fissures	DNA		616351
Sphingomyelin synthase 2 deficiency	*SGMS2*							Glaucoma	DNA		611574

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Alkaline ceramidase 3 deficiency	*ACER3*			X					DNA		617762
ABCA12 lipid transporter deficiency	*ABCA12*							Ectropion	DNA		601277
Arachinodate 12R-lipoxygenase deficiency	*ALOX12B*							Ectropion	DNA		242100
Arachidonate lipoxygenase 3 deficiency	*ALOXE3*							Ectropion	DNA		242100
Sphingolipid-1-delta (4)-desaturase deficiency	*DEGS1*					X			Dihydroceramide (P)		618404
Disorders of palmitoylation											
ZDHHC9 palmitoyltransferase deficiency	*ZDHHC9*					X			DNA		300799
Porcupine palmitoyltransferase deficiency	*PORCN*		X	X		X		Microphthalmia, Diminished visual activity, Coloboma, Aniridia, Anophthalmia, uni- or bilateral	DNA		305600
Ceroid lipofuscinosis, neuronal, 1	*PPT1*			X	X			Vision loss	Lysosomal enzymes (DBS)		256730
Hedgehog acyltransferase deficiency	*HHAT*						X		DNA		608116
Disorders of phosphoinositide metabolism											
Phosphatidylinositol 3,5-bisphosphate-5-phosphatase deficiency, neuroskeletal phenotype	*FIG4*	X	X					Visual hallucinations, Protruding eyes	DNA		611228
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency	*OCRL*	X	X					Microphthalmia, Glaucoma, Diminished visual activity	Amino acids (P)		300555
Synaptojanin 1 deficiency	*SYNJ1*							Vision, impaired, Staring gaze	Lactate (P)		615530
Myotubularin 1 deficiency	*MTM1*					X			DNA		310400
Myotubularin-related protein 2 regulatory protein deficiency	*SBF2*							Glaucoma	DNA		604563
Myotubularin-related protein 2 activator deficiency	*SBF1*					X			DNA		615284
Phosphatidylinositol 3-kinase regulatory subunit 2 superactivity	*PIK3R2*			X		X		Large eyes	DNA		603387
Phosphatidylinositol 3,4,5-trisphosphate 3-phosphatase deficiency	*PTEN*		X		X		X		DNA		158350
Inositol polyphosphate 5-phosphatase deficiency	*INPP5E*		X		X			Diminished visual activity, Microphthalmia	DNA		213300;610156
Phosphatidylinositol 4,5-bisphosphate phospholipase C γ2 deficiency	*PLCG2*	X	X						IgA, IgM, IgG (S)		614878
Inositol 1,4,5-triphosphate receptor type 1 deficiency	*ITPR1*					X			DNA		606658
Inositol polyphosphate 5-phosphatase K deficiency	*INPP5K*		X						DNA		617404
Phosphatidylinositol 4,5-bisphosphate 3-kinase regulatory subunit deficiency	*PIK3R5*					X			Alpha-fetoprotein (S)		615217
Phosphatidylinositol 4,5-bisphosphate phospholipase C β3 deficiency	*PLCB3*				X				DNA		600230
Phosphatidylinositol-4-phosphate 3-kinase catalytic subunit type 2 alpha deficiency	*PIK3C2A*		X						Calcium (P), Mucopolysaccharides (U)		618440
Disorders of lipoprotein metabolism											
Familial hypercholesterolemia heterozygous (LDLR)	*LDLR*	X							LDL/HDL cholesterol (P), Apo B (P), Triglycerides (P)	Statins, ezetimibe, PCSK9 inhibitors	143890;606945
Autosomal recessive hypercholesterolemia (ARH)	*LDLRAP1*	X							LDL/HDL cholesterol (P), Apo B (P), Triglycerides (P)	Statins, ezetimibe, PCSK9 inhibitors	603813;605747
Familial defective apolipoprotein B (APOB)	*APOB*	X							LDL/HDL cholesterol (P), Apo B (P)	Statins, PCSK9 inhibitors	144010;605019
Apolipoprotein B deficiency	*APOB*				X				LDL/HDL cholesterol (P), Apo B (P), Vitamins A/E (P)	Statins, PCSK9 inhibitors	144010;605019
PCSK9 superactivity	*PCSK9*	X							LDL/HDL cholesterol (P), Apo B (P), Triglycerides (P)	PCSK9 inhibitors	607786;613589
Microsomal triglyceride transfer protein deficiency	*MTTP*				X				LDL/HDL cholesterol (P), Apo B (P), Vitamins A/E (P)		200100;157147
Lipoprotein lipase deficiency (LPL)	*LPL*				X				LDL/HDL cholesterol (P), Triglycerides (P)		609708;238600
Apolipoprotein C-II deficiency (APOC2)	*APOC2*				X				LDL/HDL cholesterol (P), Triglycerides (P)		608083;207750

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
GPIHBP1 deficiency	*GPIHBP1*				X				LDL/HDL cholesterol (P), Triglycerides (P)		615947
Familial LCAT deficiency	*LCAT*	X							LDL/HDL cholesterol (P), Triglycerides (P), Apolipoprotein A-I		136120;606967
Tangier disease (ABCA1)	*ABCA1*	X							LDL/HDL cholesterol (P), Triglycerides (P), Apolipoprotein A-I		600046;205400
Hereditary apolipoprotein A1-related amyloidosis	*APOA1*							Glaucoma	Albumin (S)		105200
Familial combined hyperlipidemia	*USF1*	X							LDL/HDL cholesterol (P), Apo B (P), Triglycerides (P)		144250;602491
Very low-density lipoprotein receptor deficiency	*VLDLR*					X			DNA		224050
Disorders of cholesterol biosynthesis											
Mevalonate kinase deficiency (severe)	*MVK*		X						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
SC4MOL deficiency	*MSMO1*		X						Sterols (P)		607545
Chondrodysplasia punctata 2, recessive	*EBP*		X					Microphthalmia	Sterols (P)		302960
Squalene synthase deficiency	*FDFT1*							Diminished visual activity	Sterols (P), LDL/HDL cholesterol (P), Triglycerides (P), Organic acids (U)		184420
Chondrodysplasia punctata 2, recessive	*EBP*				X	X			Sterols (P)		302960
Lathosterolosis	*SC5D*	X	X			X			Sterols (P)	Liver transplant?	607330
Smith-Lemli-Opitz syndrome	*DHCR7*		X			X			Sterols (P)	Cholesterol supplementation	270400
Lanostherol synthase deficiency	*LSS*		X						Sterols (P)		616509
Lanosterol demethylase deficiency	*CYP51A1*		X						DNA		601637
Disorders of steroid metabolism											
Steroid sulfatase deficiency	*STS*	X							DNA		308100
Disorders of bile acid synthesis											
Oxysterol 7α-hydroxylase deficiency	*CYP7B1*		X	X					Bile acids (U), Sterols (P), Vitamin E (P)	Chenodeoxycholic acid, liver transplant	603711
Sterol 27-hydroxylase deficiency	*CYP27A1*		X						Sterols (P)	Chenodeoxycholic acid	213700
α-Methylacyl-CoA racemase deficiency	*AMACR*				X			Vision, progressive loss	Bile acids (U), VLCFA (P), Vitamins D/E (P)	Cholic acid	604489
STORAGE DISORDERS (n=57)											
Disorders of autophagy											
EPG5 deficiency, Vici syndrome	*EPG5*		X			X		Ocular albinism, Hypopigmentation, retinal	DNA		242840
Spatacsin deficiency	*SPG11*				X	X			DNA		616668
AP5Z1 deficiency	*AP5Z1*				X				DNA		613647
SQSTM1 deficiency	*SQSTM1*					X			DNA		617158
AP4M1 deficiency	*AP4M1*					X			DNA		602296
AP4E1deficiency	*AP4E1*					X		Downslanting palpebral fissures	DNA		607244
TBCK deficiency	*TBCK*							Visual loss, Deeply set eyes	DNA		616899
ALS2 deficiency	*ALS2*							Ocular abnormalities, Eyes opening, delayed	DNA		606353
MTMR14 deficiency	*MTMR14*					X			DNA		611089
ATG5 deficiency	*ATG5*					X			DNA		617584

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Neuronal ceroid lipofuscinosis											
Tripeptidyl-peptidase 1 deficiency	*TPP1*			X	X			Vision loss	Lysosomal enzymes (DBS)		204500
CLN3 disease	*CLN3*			X	X			Vision loss	DNA		204200
CLN5 disease	*CLN5*			X	X			Vision loss	DNA		256731
CLN6 disease	*CLN6*			X	X			Vision loss	DNA		601780
CLN7 disease	*MFSD8*			X	X			Vision loss	DNA	Antisense oligonucleotide milasen (N=1)	610951
CLN8 disease Northern epilepsy variant	*CLN8*			X	X			Vision loss	DNA		610003
Cathepsin D deficiency	*CTSD*			X	X			Vision loss	Lysosomal enzymes (DBS)		610127
Progranulin deficiency	*GRN*					X		Vision loss	DNA		614706
CLN14 disease	*KCTD7*			X				Vision loss	DNA		611726
Sphingolipidoses											
Glucocerebrosidase deficiency, Gaucher disease	*GBA*					X			Lysosomal enzymes (DBS)	Enzyme replacement therapy, substrate reduction, bone marrow transplantation	230800
Gaucher disease-like disorder due to saposin C deficiency	*PSAP*					X			Lysosomal enzymes (DBS)		610539
Acid sphingomyelinase deficiency, Niemann-Pick disease type A or B	*SMPD1*				X			Vision loss	Lysosomal enzymes (DBS)		257200;607616
Beta-galactosidase-1 deficiency, GM1-gangliosidosis	*GLB1*	X		X	X				Oligosaccharides (U), Lysosomal enzymes (DBS)		230500
Beta-hexosaminidase subunit alpha deficiency, Tay-Sachs disease	*HEXA*				X			Vision, impaired	Oligosaccharides (U), Lysosomal enzymes (DBS)		272800
Beta-hexosaminidase subunit beta deficiency, Sandhoff disease	*HEXB*				X			Vision, impaired	Oligosaccharides (U), Lysosomal enzymes (DBS)		268800
GM2 activator deficiency, GM2-gangliosidosis AB variant	*GM2A*				X			Vision, impaired	Oligosaccharides (U), Lysosomal enzymes (DBS)		272750
Beta-galactosylceramidase deficiency, Krabbe disease	*GALC*							Blindness	DNA		245200
Gaucher disease-like disorder due to saposin C deficiency	*PSAP*							Blindness	DNA		610539
Alpha-galactosidase A deficiency, Fabry disease	*GLA*	X							Globotriaosylsphingosine	Enzyme replacement therapy	301500
Acid ceramidase deficiency, primary neurologic phenotype, Farber disease	*ASAH1*				X				Lysosomal enzymes (DBS)		228000
Oligosaccharidoses											
Alpha-neuraminidase deficiency	*NEU1*	X			X				Oligosaccharides (U), Lysosomal enzymes (DBS)		256550
Cathepsin A deficiency	*CTSA*	X			X			Vision, impaired	Oligosaccharides (U), Lysosomal enzymes (DBS)		256540
Alpha-mannosidase deficiency	*MAN2B1*	X							Oligosaccharides (U), Lysosomal enzymes (DBS)	Recombinant enzyme replacement therapy (velmanase alfa); HCT	248500
Beta-mannosidase deficiency	*MANBA*					X			Lysosomal enzymes (DBS)		248510
Alpha-N-acetylgalactosaminidase deficiency type II, Kanzaki disease	*NAGA*	X							Lysosomal enzymes (DBS)		609241
Alpha-N-acetylgalactosaminidase deficiency type I, Schindler disease	*NAGA*		X	X					Oligosaccharides (U), Lysosomal enzymes (DBS)		609241
Alpha-L-fucosidase deficiency, Fucosidosis	*FUCA1*	X							Oligosaccharides (U), Lysosomal enzymes (DBS)	Bone marrow transplant	230000
Mucolipidoses											
UDP-N-acetylglucosamine-1-phosphotransferase subunit alpha/beta deficiency	*GNPTAB*	X							Oligosaccharides (U), Lysosomal enzymes (DBS)		252600
Mucolipin 1 deficiency	*MCOLN1*	X		X	X	X		Photophobia	Gastrin (S)		252650
Site-1 protease deficiency	*MBTPS1*		X						Glycosaminoglycans (U), Lysosomal enzymes (DBS)		618392
Mucopolysaccharidoses											

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Mucopolysaccharidosis-plus syndrome	*VPS33A*			X					Mucopolysaccharides (U), Oligosaccharides (U)		617303
Heparan N-sulfatase deficiency	*SGSH*				X				Mucopolysaccharides (U)	Clinical trial with intracerebroventricular infusion of chimeric fusion of recombinant enzyme +IGF2	252900
N-acetylglucosaminidase deficiency	*NAGLU*				X				Mucopolysaccharides (U)	Clinical trial with intracerebral adenovirus associated viral vector containing human NAGLU cDNA	
Heparan-alpha-glucosaminide N-acetyltransferase deficiency	*HGSNAT*				X				Mucopolysaccharides (U)		252930
Disorders of lysosomal cholesterol metabolism											
Niemann-Pick disease type C1	*NPC1*					X			Oxyterols (P), filipin test (F)	Miglustat; experimental intrathecal or intravenous 2-hydroxypropyl-beta-cyclodextrin; oral Arimoclomol	257220
Niemann-Pick disease type C2	*NPC2*					X			Oxyterols (P), filipin test (F)		607625
Disorders of glycosaminoglycan degradation											
Alpha-iduronidase deficiency, Hurler, Scheie disease	*IDUA*	X		X				Glaucoma	Mucopolysaccharides (U), Lysosomal enzymes (DBS)	Laronidase; hematopoietic stem cell transplant	607014;607015;607016
Iduronate 2-sulfatase deficiency, Hunter disease	*IDS*				X				Mucopolysaccharides (U), Lysosomal enzymes (DBS)	Idursulfase	309900
N-acetylgalactosamine-6-sulfatase deficiency, Morquio A disease	*GALNS*	X							Mucopolysaccharides (U), Lysosomal enzymes (DBS)	Elosulfase	253000
Beta-galactosidase deficiency, Morquio B disease	*GLB1*	X							Mucopolysaccharides (U), Lysosomal enzymes (DBS)		230500
N-acetylgalactosamine - 4 - sulfatase deficiency, Maroteaux - Lamy disease	*ARSB*	X						Glaucoma	Mucopolysaccharides (U), Lysosomal enzymes (DBS)	Galsulfase	253200
Beta-glucuronidase deficiency, Sly disease	*GUSB*	X							Mucopolysaccharides (U), Lysosomal enzymes (DBS)	Vestronidase	253220
Arylsulfatase G deficiency	*ARSG*				X				DNA		618144
Disorders of lysosomal transport or sorting											
Nephropathic cystinosis	*CTNS*	X			X			Vision, impaired, Photophobia	Amino acids (P)	Cysteamine	219800;219900;219750
Sialin deficiency (milder)	*SLC17A5*			X		X			Oligosaccharides (U)		604369
Glucocerebrosidase receptor deficiency	*SCARB2*					X			Lysosomal enzymes (DBS)		254900
Disorders of lysosomal protein degradation											
Hypomyelinating leukodystrophy - 12	*VPS11*							Blindness	Sulfatide (U), Ceramides (U)		616683
DISORDERS OF PEROXISOMES AND OXALATE (n=27)											
Disorders of plasmalogen synthesis											
Peroxisomal targeting signal 2 receptor deficiency	*PEX7*		X						VLCFA (P), Plasmalogens (P)		215100
Glycerone 3-phosphate acyltransferase deficiency	*GNPAT*		X						VLCFA (P), Plasmalogens (P)		602744
Alkylglycerone 3-phosphate synthase deficiency	*AGPS*		X						VLCFA (P), Plasmalogens (P)		600121
Fatty Acyl-CoA reductase superactivity	*FAR1*		X						Plasmalogens (RBC)		616107
Peroxin 5 deficiency	*PEX5*		X			X			DNA		214110
Fatty Acyl-CoA reductase superactivity	*FAR1*		X						Plasmalogens (RBC)		616107
Disorders of peroxisomal β-oxidation											
X-linked adrenoleukodystrophy and adrenomyeloneuropathy	*ABCD1*							Vision loss	VLCFA (P)	HCT at early stages of cerebral X-ALD; HSC gene therapy with lentiviral vector	300100
Peroxisomal straight-chain acyl-CoA oxidase deficiency	*ACOX1*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Plasmalogens (P)		264470
D-bifunctional protein deficiency	*HSD17B4*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Plasmalogens (P), Organic acids (U)		261515
Phytanoyl-CoA hydroxylase deficiency	*PHYH*				X				VLCFA (P), Pipecolic acid (P)	Phytanic acid restriction	266500
Disorders of peroxisomal biogenesis											

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Zellweger spectrum disorders - Peroxin 1 deficiency	*PEX1*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		234580;214100;601539
Zellweger spectrum disorders - Peroxin 2 deficiency	*PEX2*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614866;614867
Zellweger spectrum disorders - Peroxin 3 deficiency	*PEX3*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		617370;614882
Zellweger spectrum disorders - Peroxin 5 deficiency	*PEX5*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		214110
Zellweger spectrum disorders - Peroxin 6 deficiency	*PEX6*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614862;614863;616617
Zellweger spectrum disorders - Peroxin 10 deficiency	*PEX10*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614870;614871
Zellweger spectrum disorders - Peroxin 14B deficiency	*PEX11B*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614920
Zellweger spectrum disorders - Peroxin 12 deficiency	*PEX12*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614859;266510
Zellweger spectrum disorders - Peroxin 13 deficiency	*PEX13*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614883;614885
Zellweger spectrum disorders - Peroxin 14 deficiency	*PEX14*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614887
Zellweger spectrum disorders - Peroxin 16 deficiency	*PEX16*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614876;614877
Zellweger spectrum disorders - Peroxin 19 deficiency	*PEX19*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614886
Zellweger spectrum disorders - Peroxin 26 deficiency	*PEX26*		X		X			Glaucoma, Diminished visual activity	VLCFA (P), Pipecolic acid (P)		614872;614873
Disorders of oxalate metabolism											
Alanine-glyoxylate aminotransferase deficiency, Primary hyperoxaluria type I	*AGXT*			X	X					Liver or liver/kidney transplant; lumasiran	259900
Hydroxyacid oxidase 1 deficiency	*HAO1*							Anisocoria, Alacrima	Oxalic acid (P,U), Glycolic acid (P,U)		605023
Glyoxylate reductase deficiency, Primary hyperoxaluria type II	*GRHPR*			X	X				Oxalic acid (P,U), Glyceric acid (U)		260000
4-hydroxy-2-oxoglutarate aldolase deficiency, Primary hyperoxaluria type III	*HOGA1*			X	X				Oxalic acid (P,U), Organic acids (U)		613616
CONGENITAL DISORDERS OF GLYCOSYLATION (n=92)											
Disorders of N-linked glycosylation											
PMM2-CDG	*PMM2*				X	X			Factor XI (B), Arylsulfatase A (P), Sialotrasferrins (S)		601785
DPAGT1-CDG	*DPAGT1*		X			X			Sialotrasferrins (S)		608093
ALG13-CDG	*ALG13*			X		X			Sialotrasferrins (S)		300884
ALG1-CDG	*ALG1*					X		Ocular abnormalities	Sialotrasferrins (S)		608540
ALG2-CDG	*ALG2*		X					Coloboma	Factor XI (B), Sialotrasferrins (S)		607906
ALG11-CDG	*ALG11*			X	X	X			Factor XI (B), Sialotrasferrins (S)		613661
RFT1-CDG	*RFT1*							Vision, impaired	Factor XI (B), Sialotrasferrins (S)		612015
ALG3-CDG	*ALG3*			X		X			Antithrombin III (P), Protein S (S), Sialotrasferrins (S)		601110
ALG9-CDG	*ALG9*					X			Factor XI (B), Sialotrasferrins (S)		608776
ALG12-CDG	*ALG12*				X	X			Sialotrasferrins (S)		607143
ALG6-CDG	*ALG6*				6				Factor XI (B), Sialotrasferrins (S)		603147
ALG8-CDG	*ALG8*		X	X					Albumin (S), Sialotrasferrins (S)		608104
SSR4-CDG	*SSR4*					X			Sialotrasferrins (S)		300934
GCS1-CDG	*MOGS*							Short palpebral fissures, Long eye lashes	Oligosaccharides (U), Sialotrasferrins (S)		606056
MAN1B1-CDG	*MAN1B1*					X			Sialotrasferrins (S)		614202;604346

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
MGAT2-CDG	*MGAT2*					X	X	Delayed visual maturation	Factor IX/XI (B), Sialotrasferrins (S)		212066
FUT8-CDG	*FUT8*							Glaucoma, Buphthalmos	DNA		618005
STT3B-CDG	*STT3B*			X					Sialotrasferrins (S)		615597
DDOST-CDG	*DDOST*					X			Factor XI (B), Proteins C/S (S), Sialotrasferrins (S)		614507;602202
B4GALT1-CDG	*B4GALT1*						X		Antithrombin (B), Factor XI (B), Sialotrasferrins (S)		607091
SSR3-CDG	*SSR3*					X			DNA		606213
MAN2B2-CDG	*MAN2B2*					X			Sialotransferrins (S), C-reaktive protein, CRP (P), IgE (S)		618899
Disorders of O-mannosylation											
POMT1-CDG	*POMT1*	X	X		X			Microphthalmia, Glaucoma, Exophthalmia, Buphthalmos	CK (P), Sialotrasferrins (S)		236670;613555;609308
POMT2-CDG	*POMT2*	X	X		X			Microphthalmia, Glaucoma, Exophthalmia, Buphthalmos	CK (P), DNA		613150;613156;613158
POMGNT1-CDG	*POMGNT1*	X	X		X		X	Microphthalmia, Glaucoma, Exophthalmia, Buphthalmos	CK (P), DNA		253280;613151;613157
POMGNT2-CDG	*POMGNT2*					X			CK (P), DNA		614830;618135
B3GALNT2-CDG	*B3GALNT2*		X				X	Microphthalmia	CK (P), DNA		615181
POMK-CDG	*POMK*		X		X		X	Microphthalmia, Glaucoma	CK (P), DNA		616094;615249
CRPPA-CDG	*CRPPA*	X	X	X	X			Vitreous, persistent hyperplastic primary			614643;616052
FKTN-CDG A	*FKTN*		X	X	X			Microphthalmia	CK (P), DNA		611588
FKRP-CDG A	*FKRP*	X	X		X	X		Microphthalmia,Coloboma	CK (P), DNA		606596
TMEM5-CDG	*RXYLT1*				X				CK (P), DNA		615041
B4GAT1-CDG	*B4GAT1*	X		X	X			Blindness	CK (P), DNA		615287
LARGE1-CDG	*LARGE1*		X	X	X	X			CK (P), DNA		613154;608840
Disorders of O-xylosylation and glycosaminoglycan synthesis											
XYLT1-CDG	*XYLT1*						X		DNA		615777
XYLT2-CDG	*XYLT2*		X		X				DNA		605822
B4GALT7-CDG	*B4GALT7*							Exophthalmia	DNA		130070
CHSY1-CDG	*CHSY1*			X					DNA		605282
CHST14-CDG	*CHST14*	X	X		X	X	X	Glaucoma, Blue sclerae, Downslanting palpebral fissures	DNA		601776
DSE-CDG	*DSE*							Telecanthus, Blue sclerae, Downslanting palpebral fissures	DNA		615539
CHST6-CDG	*CHST6*	X						Photophobia	DNA		217800
CANT1-CDG	*CANT1*						X	Prominent eyes, Glaucoma	DNA		251450
Golgi-resident phosphoadenosine phosphate phosphatase deficiency	*IMPAD1*							Prominent eyes	DNA		614078
Plasma membrane citrate transporter deficiency	*SLC13A5*					X			DNA		615905
HS6ST2-CDG	*HS6ST2*						X	Deeply set eyes	DNA		301025
Disorders of O-GlcNAcylation											
OGT-CDG	*OGT*					X		Amblyopia	DNA		300997
GALNT14-CDG	*GALNT14*	X					X	Keratoconus	DNA		608225

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Disorders of O-fucosylation											
POFUT1-CDG	*POFUT1*							Hypopigmentation, retinal	DNA		615327
B3GALTL-CDG	*B3GLCT*	X	X						DNA		261540
Disorders of glycosylphosphatidylinositol biosynthesis											
PIGQ-CDG	*PIGQ*			X		X	X	Visual impairment, Telecanthus, Alacrima	ALP (P), DNA		605754
PIGL-CDG	*PIGL*							Coloboma	DNA		280000
PIGV-CDG	*PIGV*							Upslanting palpebral fissures, Blindness	ALP (P), DNA		239300
PIGN-CDG	*PIGN*					X			DNA		614080
PIGT-CDG	*PIGT*					X	X	Vision, decreased	ALP (P), DNA		615398
GPAA1-CDG	*GPAA1*					X			DNA		617810
PIGS-CDG	*PIGS*							Blindness, cortical	DNA		618143
PIGB-CDG	*PIGB*							Ophthalmological anomalies	ALP (P), DNA		604122
PIGP-CDG	*PIGP*							Visual impairment	DNA		617599
PIGU-CDG	*PIGU*							Visual impairment	DNA		618590
Disorders of glycolipid glycosylation											
ST3GAL5-CDG	*ST3GAL5*			X					Lactosylceramide (P), GM3 ganglioside (P)		609056
Disorders of dolichol metabolism											
DHDDS-CDG	*DHDDS*				X			Vision loss	DNA		613861;608172
NUS1-CDG	*NUS1*				X				DNA		617082
SRD5A3-CDG	*SRD5A3*		X	X		X		Microphthalmia, Glaucoma, Coloboma	Antithrombin III (P), Protein S (S), Sialotrasferrins (S)		612379
DOLK-CDG	*DOLK*					X			Sialotrasferrins (S)		610768
DPM1-CDG	*DPM1*				X	X			Factor XI (B), Sialotrasferrins (S)		608799
MPDU1-CDG	*MPDU1*			X		X		Vision, impaired	Sialotrasferrins (S)		609180
Disorders of nucleotide-sugar synthesis											
GMPPA-CDG	*GMPPA*							Ocular abnormalities, Alacrima	DNA		615510
GMPPB-CDG	*GMPPB*		X						CK (P), Sialotrasferrins (S)		615350;615351;615352
UGP2-CDG	*UGP2*			X		X			DNA		618744
Disorders of Golgi transport											
SLC35A2-CDG	*SLC35A2*				X	X		Visual impairment, Ocular abnormalities	Sialotrasferrins (S)		314375
Disorders of vesicular trafficking											
Conserved oligomeric Golgi complex subunit 5 deficiency	*COG5*					X		Blindness	Sialotrasferrins (S)		613612
Conserved oligomeric Golgi complex subunit 8 deficiency	*COG8*					X			Factor XI (B), Sialotrasferrins (S)		611182
Cohen syndrome VPS13B-CDG	*VPS13B*				X		X		Sialotrasferrins (S)		216550
Conserved oligomeric Golgi complex subunit 4 superactivity	*COG4*		X						DNA		618150
COPB2 deficiency	*COPB2*							Blindness, cortical	DNA		617800

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Archain 1 deficiency	*ARCN1*	X							DNA		617164
Craniolenticulosutural dysplasia	*SEC23A*	X		X		X		Hypertelorism	DNA		607812
TRAPPC4 deficiency	*TRAPPC4*							Visual impairment	DNA		618741
TRAPPC6B deficiency	*TRAPPC6B*					X			DNA		617862
TRAPPC9 deficiency	*TRAPPC9*							Hypertelorism	DNA		613192
TRAPPC12 deficiency	*TRAPPC12*			X					DNA		617669
SCYL2 deficiency	*SCYL2*			X					DNA		618766
Rubicon deficiency	*RUBCN*					X			DNA		615705
RAB18 deficiency	*RAB18*	X	X	X				Microphthalmia	DNA		614222
RAB3GAP1 deficiency	*RAB3GAP1*	X	X	X				Microphthalmia	DNA		600118
RAB3GAP2 deficiency	*RAB3GAP2*		X					Microphthalmia	DNA		614225;212720
VPS4A deficiency	*VPS4A*		X			X			DNA		609982
VPS41 deficiency	*VPS41*			X					DNA		605485
Disorders of Golgi homeostasis											
ATP6VOA2-CDG	*ATP6V0A2*					X	X	Amblyopia	Sialotrasferrins (S)		219200;278250
ATP6V1A-CDG	*ATP6V1A*							Entropion	Sialotrasferrins (S)		617403
ATP6V1E1-CDG	*ATP6V1E1*							Entropion	Sialotrasferrins (S)		617402
Disorder of deglycosylation											
N-glycanase 1 deficiency	*NGLY1*	X				X		Hypolacrima, Alacrima	Oligosaccharides (U)		615273
OTHER (n=25)											
Disorders of the synaptic vesicle cycle											
KIF5A deficiency	*KIF5A*					X			DNA		602821
Dynamin 2 deficiency	*DNM2*				X				DNA		602378
Synaptic vesicle glycoprotein 2A deficiency	*SV2A*			X					N-acetylaspartate (MRS), Lactate (MRS)		185860
Synaptobrevin 1 deficiency	*VAMP1*					X			DNA		108600
IL1RAPL1 deficiency	*IL1RAPL1*					X			DNA		300143
Synaptotagmin 1 deficiency	*SYT1*					X			DNA		618218
Torsin 1A deficiency	*TOR1A*					X			DNA		618947
Disorders of lysosome-related organelle biogenesis											
Hermansky-Pudlak syndrome type 1	*HPS1*							Ocular albinism	DNA		203300
Hermansky-Pudlak syndrome type 2	*AP3B1*							Ocular albinism	DNA		608233
Hermansky-Pudlak syndrome type 3	*HPS3*							Ocular albinism	DNA		614072
Hermansky-Pudlak syndrome type 4	*HPS4*							Ocular albinism	DNA		614073
Hermansky-Pudlak syndrome type 5	*HPS5*							Ocular albinism	DNA		614074
Hermansky-Pudlak syndrome type 6	*HPS6*							Ocular albinism	DNA		614075

Name (n=583)		Corneal involvement	Lens involvement	Optic nerve involvement	Retinal involvement	Oculomotor involvement	Refractive errors	Other	Laboratory investigations	Specific treatment	Disorder OMIM No.
Hermansky-Pudlak syndrome type 7	*DTNBP1*							Ocular albinism	DNA		614075
Hermansky-Pudlak syndrome type 8	*BLOC1S3*							Ocular albinism	DNA		614077
Hermansky-Pudlak syndrome type 9	*BLOC1S6*							Ocular albinism	DNA		614171
Hermansky-Pudlak syndrome type 10	*AP3D1*					X		Ocular albinism; Hypotelorism	DNA		617050
Chediak-Higashi syndrome	*LYST*					X			DNA		214500
Griscelli syndrome type 1	*MYO5A*					X			DNA		214450
Disorders of organelle interplay											
EMC1 deficiency	*EMC1*					X			DNA		616875
BAP31 deficiency	*BCAP31*			X		X			DNA		300475
Miscellaneous											
PGM2L1 deficiency	*PGM2L1*					X			DNA		611610
Kearns-Sayre Syndrome					X	X			Protein (CSF), 5-Methyl-THF (CSF)		530000
Leber Hereditary Optic Neuropathy, LHON	*PRICKLE3*			X				Loss of central vision	DNA		535000
Ataxia oculomotor apraxia 1 (AOA1)	*APTX*					X			Albumin (S)		606350