Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
INTERTMEDIARY METABOLISM: NUTRIENTS (n=29)											
Disorders of glycine and serine metabolism											
3-Phosphoglycerate dehydrogenase deficiency	*PHGDH*	AR	601815						Adducted thumbs		S-Methyl-THF (CSF) Glycine (CSF) Serine (CSF)
Mitochondrial serine hydroxymethyltransferase deficiency	*SHMT2*	AR	138450	Arched eyebrows		Thin upper lip	Eyelashes, long Long palpebral fissures				S-Methyl-THF (CSF)
Other disorders of amino acid metabolism											
Aminoacylase 1 deficiency	*ACY1*	AR	609924				Hypertelorism				
Organic acidurias											
3-hydroxyisobutyryl-CoA hydrolase deficiency	*HIBCH*	AR	250620	Dysmorphic facies			Strabismus		Syndactyly of toes		Organic acids (U), S-(2-carboxypropyl)-cysteine (U), S-2-carboxyvorooyl-cysteamine (U)
3-Hydroxyisobutyrate dehydrogenase deficiency	*HIBADH*	AR	608475	Triangular face	Long philtrum		Long eyelashes Prominent eye brows	Low set ears	Clinodactyly of the fifth finger and toe Syndactyly of the second and third toes		C4-OH 3-Hydroxyisobutyryl-carnitine (DBS, P), Organic acids (U)
Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide											
Adenosine kinase deficiency	*ADK*	AR	614300	Frontal bossing						Short stature	Amino acids (P) Purines and pyrimidines (U)
Disorders of ornithine, proline and hydroxyproline metabolism											
Delta-1-pyrroline-5-carboxylate synthase deficiency	*ALDH18A1*	AD, AR	219150	Progeroid appearance						Short stature	Amino acids (P)
Pyrroline-5-carboxylate reductase 1 deficiency	*PYCR1*	AR	612940	Triangular face Wide forehead Midface hypoplasia	Bulbous nose		Deeply set eyes Hypotelorism	Prominent ears			DNA
Pyrroline-5-carboxylate reductase 2 deficiency	*PYCR2*	AR	616420	Bitemporal narrowing Malar hypoplasia Triangular face	Bulbous nose Long philtrum Short nose Upturned nose		Upslanting palpebral fissures Down-slanting palpebral fissures	Large malformed ears Low-set ears			DNA
Disorders of glutamate/glutamine and aspartate/asparagine metabolism											
Asparagine synthetase deficiency	*ASNS*	AR	615574	Micrognathia				Large malformed ears			Amino acids (P, CSF)
Other disorders of peptide metabolism											
Prolidase deficiency	*PEPD*	AR	170100 613230	Low posterior hairline Facial hirsutism Micrognathia	Saddle nose Small nose Beaked nose	High arched palate	Widely spaced eyes Exophthalmos				Amino acids (U)
Disorders of polyamine metabolism											
Spermidine-spermine N(1)-acetyltransferase overactivity	*SAT1*	XL	313020	Facial dysmorphism Micrognathia		Thin upper lip					Apolipoprotein A-I level Cholestanol (P)
Ornithine decarboxylase 1 superactivity	*ODC1*	AD	165640	High forehead	Tubular nose		Mild ptosis Widely spaced eyes Deeply set eyes	Macrotia			N-Acetylputrescine (P)
Disorders of galactose and fructose metabolism											
Triokinase/FMN cyclase deficiency	*TKFC*	AR	618805				Microphthalmia				Lactate (P)
Disorders of glycolysis											
Aldolase A deficiency GSD 12	*ALDOA*	AR	611881	Triangular face		Wide mouth Thin lips		Low set ears			Bilirubin (P), Reticulocytes (B), Aldolase A (Ec)
Disorders of glycogen metabolism											
Liver glycogen phosphorylase deficiency	*PYGL*	AR	232700	Adiposity (doll-like facies)						Short stature	Biotinidase (P) Glycogen (U) Ketones, fasted (P, U)
Hepatic phosphorylase kinase γ2 subunit deficiency	*PHKG2*	AR	613027	Adiposity (doll-like facies)						Short stature	Biotinidase (P) Glycogen (U) Ketones, fasted (P, U)
Phosphorylase kinase β subunit deficiency	*PHKB*	AR	261750	Adiposity (doll-like facies)						Short stature	Biotinidase (P) Glycogen (U) Ketones, fasted (P, U)
Hepatic phosphorylase kinase α2 subunit deficiency	*PHKA2*	XL	306000	Adiposity (doll-like facies)						Short stature	Biotinidase (P) Glycogen (U) Ketones, fasted (P, U)
Amylo-1,6-glucosidase (debrancher) deficiency	*AGL*	AR	232400	Adiposity (doll-like facies)						Short stature	Biotinidase (P) Glycogen (U) Ketones, fasted (P, U)
Disorders of pentose metabolism											
Ribose-5-phosphate isomerase deficiency	*RPIA*	AR	608611	Coarse facies Dolichocephaly Micrognathia	Depressied nasal bridge	Tented upper lip High arched narrow palate	Deep-set eyes	Protruding ears Cupped ears Large ears			Polyols (U, P, CSF); Sugar phosphates (U)
Transaldolase deficiency	*TALDO1*	AR	606003	Riangular face Infraorbital creases	Prominent philtrum	Wide mouth Thin lips		Low set ears			ASAT/ALAT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Ferritin (S), Polyol (U)
Transketolase deficiency	*TKT*	AR	617044		Depressied nasal bridge		Widely spaced eyes Broad eyebrows Full eyebrows	Posteriorly rotated ears			Polyols (U, P, CSF); Sugar phosphates (U)
Sedoheptulose kinase deficiency	*SHPK*	AR	617713	High forehead with large fontanels	High nasal bridge	Small mouth	Hypotelorism Round asymmetrical eyes Small orbit	Dysplastic ears Low set ears	Adducted thumbs Small feet Arthrogrypposis		Polyols (U)
Disorders of carbohydrate transmembrane transport and absorption											
Sialin deficiency	*SLC17A5*	AR	269920	Coarse facial features							Oligosaccharides (U)
Neuronal glucose transporter deficiency	*SLC45A1*	AR	617532	Maxillary prognathism	Broad nasal bridge	Thick lips Semi-open mouth	Thick eyebrows		Short limbs		DNA
Other disorders of carbohydrate metabolism											
Phosphoglucomutase 2-like 1 deficiency	*PGM2L1*	AR	611610	Pointed chin	Depressed nasal bridge Prominent nasal tip Long philtrum	Thin upper lip High-arched and narrow palate	Downslanting palpebral fissures Epicanthal folds	Large ears Prominent ears			DNA
Disorders of carnitine metabolism											

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
γ-Butyrobetaine hydroxylase deficiency	*BBOX1*	AR	603312	Short forehead Retrognathia	High nasal bridge Flat nasal root		Epicanthal folds Strabismus Long eye lashes Down-slanting palpebral fissures	Large ears			DNA
Disorders of mitochondrial fatty acid oxidation											
Short-chain acyl CoA dehydrogenase deficiency Ethylmalonic aciduria	*ACADS*	AR	201470		Short nose	Small mouth					Organic acids (U), Acylcarnitines (DBS, P)
INTERMEDIARY METABOLISM: ENERGY (n=42)											
Disorders of pyruvate metabolism											
Mitochondrial pyruvate carrier deficiency	*MPC1*	AR	614741			Thin upper lip	Epicanthal folds				Organic acids (U), Lactate (P), Pyruvate (P)
Pyruvate dehydrogenase E1 alpha deficiency	*PDHA1*	XL	312170	Retrognathia Narrow head Frontal bossing	Broad nasal bridge Flared nostrils Broad philtrum	Thin upper lip	Downslanting palpebral fissures	Low set ears			Lactate (P), Pyruvate (P), Amino acids (P)
Disorders of the Krebs cycle											
Alpha-ketoglutarate dehydrogenase deficiency	*OGDH*	AR	203740		Depressed nasal bridge Long philtrum Short nose		Epicanthus	Low-set ears			2-Ketoglutarate (U) Ketones (P, U) Lactate (P) Glucose (P)
Cytosolic NADP+-dependent isocitrate dehydrogenase 1 superactivity	*IDH1*	AR	147700	Retrognathia Wide forehead	Long philtrum	Thin upper lip	Telecanthus Short palpebral fissures			Widely spaced nipples	
Fumarate hydratase deficiency	*FH*	AR	606812	Coarse facial features			Hypertelorism				Lactate (P), Organic acids (U)
OGDHL deficiency	*OGDHL*	AR	617513	Scaphocephaly		Large mouth High arched palate	Downslanting palpebral fissures				DNA
Disorders of mtDNA-encoded tRNA and rRNA											
Mitochondrial tRNA(Glu) deficiency	*MT-TE*	MT	500009			Macroglossia					DNA
Disorders of complex I-V subunits and assembly factors											
Complex I assembly factor 5 deficiency	*NDUFAF5*	AR	618238	Facial dysmorphism Micrognathia	Long philtrum	Small mouth					Lactate (P,CSF)
NADH dehydrogenase subunit C2 deficiency	*NDUFC2*	AR	619170	Midface hypoplasia							Lactate (P)
LRPPRC deficiency	*LRPPRC*	AR	220111	Prominent forehead Midface hypoplasia Large anterior fontanelle	Broad nasal bridge		Widely spaced eyes Synophrys Arched eyebrows				Lactate (P)
Cytochrome c oxidase subunit 5A deficiency	*COX5A*	AR	603773	Large fontanelles Frontal bossing			Long eyelashes Synophrys Deep-set eyes		Short hands and feet		DNA
Transmembrane protein 70 deficiency	*TMEM70*	AR	614052	Flat occiput	Long philtrum			Low set ears			CK (P), Lactate (P), Ammonia (P), 3-Methylglutaconic acid (U)
NADH dehydrogenase alpha subcomplex subunit 8 deficiency	*NDUFA8*	AR	603359			High palate					Lactate (P), Organic acids (U)
NADH dehydrogenase beta subcomplex subunit 11 deficiency	*NDUFB11*	XL	252010				Microphthalmia				Lactate (P), Organic acids (U)
Holocytochrome c synthase deficiency	*HCCS*	XL	309801				Microphthalmia			Short stature	DNA
Cytochrome c oxidase subunit 4I1 deficiency	*COX4I1*	AR	123864		Broad nasal bridge						DNA
Cytochrome c oxidase subunit 6A2 deficiency	*COX6A2*	AR	619062			Arched palate, high					DNA
Skeletal anomalies and mental retardation syndrome	*RAFSIF*	AR	616994	Craniofacial dysmorphism Flat oval face		Cleft lip Cleft palate	Hypertelorism	Low set ears			DNA
Mitochondrial ATP synthase F1 assembly factor 2 deficiency	*ATPAF2*	AR	604273	Micrognathia Frontal bossing	High nasal bridge Prominent nasal bridge	Large mouth		Low set ears	Rocker-bottom feet	Hypospadias	Lactate (P), Organic acids (U)
Disorders of mitochondrial transcript processing and modification											
Pseudouridine synthase 1 deficiency	*PUS1*	AR	600462	Micrognathia Retrognathia	Short nose Short philtrum	Gum hypertrophy	Ptosis Exophthalmus				Lactate (P)
Mitochondrial RNA polymerase deficiency	*POLRMT*	AD, AR	619743				Hypotelorism			Short stature	
Combined oxidative phosphorylation deficiency 58	*TEFM*	AR	620451		Broad nasal bridge Columella, low-hanging		Epicanthus	Low-set ears			Lactate (P)
Disorders of mitochondrial aminoacyl-tRNA synthetases											
Mitochondrial tyrosyl-tRNA synthetase deficiency	*YARS2*	AR	613561	Triangular shaped head			Wide spaced eyes.				Lactate (P), Blood count
Mitochondrial valyl-tRNA synthetase deficiency	*VARS2*	AR	615917	Micrognathia							DNA
Mitochondrial prolyl-tRNA synthetase deficiency	*PARS2*	AR	612036	Sloping forehead	Short nose Broad nasal bridge Anteverted nares Short philtrum	Open mouth	Widely spaced eyes Upslanting palpebral fissures				Lactate (P)
Disorders of the mitoribosome											
Mitochondrial ribosomal small subunit 2 deficiency	*MRPS2*	AR	617950				Strabismus Upslanting palpebral fissures	Low set ears			Lactate (P), Organic acids (U)
Combined Oxidative Phosphorylation Defect 2	*MRPS16*	AR	610498					Low set ears			
Mitochondrial ribosomal small subunit 14 deficiency	*MRPS14*	AR	611978	Midface hypoplasia	Depressed nasal bridge		Widely spaced eyes	Low set ears Posteriorly rotated ears			Lactate (P)
Mitochondrial ribosomal small subunit 28 deficiency	*MRPS28*	AR	611990	Round face	Long philtrum			Low set ears Posteriorly rotated ears		Short neck	Lactate (P), Organic acids (U)
Mitochondrial ribosomal large subunit 50 deficiency	*MRPL50*	AR	611854						Long tapering fingers and toes		DNA
Disorders of mitochondrial shuttles and carriers											
Cytosolic malate dehydrogenase deficiency	*MDH1*	AR	618959	High forehead	Depressed nasal bridge						DNA
Mitochondrial ATP-Mg-phosphate transporter deficiency	*SLC25A24*	AR	612289	Micrognathia Prognathism Triangular face Midface hypoplasia Progeroid appearance	Long philtrum Small nose	Thin upper lip Arched palate, high Protruding tongue	Deeply set eyes Down-slanting palpebral fissures Hypertelorism Short palpebral fissures	Dysplastic ears Low-set ears Posteriorly rotated ears		Short stature Small nipples Widely spaced nipples	DNA
Disorders of mitochondrial protein import and quality control											

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
MAGMAS deficiency	*PAM16*	AR	613320	Broad forehead with frontal bossing Sparse hair	Depressed nasal bridge Short nose Anteverted nares Wide nares	Open mouth Protruding tongue				Narrow chest Short neck Short limbs	DNA
HSPE1 deficiency	*HSPE1*	AD	600141			Thin upper lip					Pyridoxal 5'-phosphate, PLP (CSF)
Garg-Mishra progeroid syndrome	*TOMM7*	AR	620601	Broad forehead Triangular face	Broad nasal bridge Bulbous nose		Microphthalmia			Short stature	DNA
deficiency	*FBXL4*	AR	615471	Craniofacial dysmorphism							DNA
HSPA9 deficiency	*HSPA9*	AR	182170		Hypoplastic nose			Microtia		Short stature	DNA
LONP1 deficiency	*LONP1*	AR	600373	Craniofacial dysmorphism						Short stature	DNA
Miscellaneous disorders associated with mitochondrial dysfunction											
Peptidyl-tRNA hydrolase 2 deficiency	*PTRH2*	AR	616263	Midface hypoplasia		Thin upper lip	Widely spaced eyes Exotropia		Proximally placed thumbs Deformities of the fingers and toes		DNA
LETM1 deficiency	*LETM1*	AR	620089	Long face Micrognathia	Prominent nose	High arched palate		Low set ears			DNA, Organic acids (U)
Disorders of mitochondrial and non-mitochondrial metabolite repair											
Acyl-CoA-synthase 3 deficiency Combined malonic and methylmalonic aciduria	*ACSF3*	AR	614245	Facial dysmorphism							Organic acids (U), Acylcarnitines (DBS, P)
Ubiquitous glucose-6-phosphatase deficiency	*G6PC3*	AR	612541	Midface hypoplasia Plagiocephaly Sparse hair		Tented mouth				Pectus carinatum	DNA, Abnormal neutrophil N-glycans
LIPID METABOLISM AND TRANSPORT (n=40)											
Disorders of peroxisomal fatty acid oxidation											
Peroxisomal straight-chain acyl-CoA oxidase deficiency	*ACOX1*	AR	264470	Facial dysmorphism	Depressed nasal bridge		Widely spaced eyes Epicanthal fold	Low set ears			VLCFA (P), Plasmalogens (P)
D-bifunctional protein deficiency	*HSD17B4*	AR	261515	Frontal bossing	Depressed nasal bridge		Upslanting palpebral fissures				VLCFA (P), Plasmalogens (P), Organic acids (U)
Phytanoyl-CoA hydroxylase deficiency Refsum disease	*PHYH*	AR	266500	Midface hypoplasia			Epicanthic folds	Low set ears			Pipecolic acid (P, U), Phytanic acid (S, U), Protein (CSF)
Disorders of eicosanoid metabolism											
Leukotriene C4 synthase deficiency	*LTC4S*	AR	246530	Mild facial dysmorphism							Leucotriens (P, CSF)
Prostaglandin transporter deficiency	*SLCO2A1*	AR	119900	Coarse facial features							Prostaglandins (U)
15-hydroxy-prostaglandin dehydrogenase deficiency	*HPGD*	AR	259100	Coarse facial features							Prostaglandins (U)
Disorders of glycerophospholipid metabolism											
Peroxisomal targeting signal 2 receptor deficiency	*PEX7*	AR	215100	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Plasmalogens (P)
Glycerone 3-phosphate acyltransferase deficiency	*GNPAT*	AR	602744	Micrognathia	Broad nasal bridge Anteverted nares	High arched palate	Epicanthal folds	Dysplastic ears			VLCFA (P), Plasmalogens (P)
PLCH1 deficiency	*PLCH1*	AR	612835		Defects of the nose	Cleft lip	Widely spaced eyes				DNA
Alkylglycerone 3-phosphate synthase deficiency	*AGPS*	AR	600121	Full cheeks Midface hypoplasia	Small nose Anteverted nares		Widely spaced eyes				VLCFA (P), Plasmalogens (P)
Peroxin 5 deficiency	*PEX5*	AR	214110	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Plasmalogens (P)
Phosphatidylinositol 4,5-bisphosphate phospholipase C β3 deficiency	*PLCB3*	AR	600230		Depressed nasal bridge Short nose Anteverted nares		Widely spaced eyes Prominent eyes				DNA
Lysophosphatidylcholine-esterified long-chain fatty acid transporter deficiency	*MFSD2A*	AR	616486				Upslanting palpebral fissures				Lysophosphatidylcholines (P)
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency	*OCRL*	XL	300555				Microphthalmia				Amino acids (P, U), DNA
Disorders of phosphatidylcholine, phosphatidylserine and phosphatidylethanolamine metabolism											
Ethanolaminephosphotransferase 1 deficiency	*SELENOI*	AR	607915			Bifid uvula Cleft palate					DNA
Phosphatidylserine synthase 1 superactivity	*PTDSS1*	AD	151050	Wide forehead Micrognathia	Choanal atresia Choanal stenosis	Small tongue	Hypertelorism	Large malformed ears		Short stature	DNA
Disorders of palmitoylation											
Porcupine palmitoyltransferase deficiency	*PORCN*	XL	305600			Lip papillomas	Strabismus Microphthalmia		Syndactyly Polydactyly Camptodactyly		DNA
ZDHHC15 palmitoyltransferase deficiency	*ZDHHC15*	XL	300577	Long face Brachycephaly			Down-slanting palpebral fissures	Large ears			DNA
Disorders of phosphatidylinositol metabolism											
VAC14 deficiency type 1	*VAC14*	AR	216340						Thumb hypolasia Arbormal halluces		DNA
Phosphatidylinositol-4-phosphate 3-kinase catalytic subunit type 2 alpha deficiency	*PIK3C2A*	AR	618440	Coarse face	Broad nasal bridge		Epicanthal folds			Short stature Retrognathia	Mucopolysaccharides (U)
Phosphatidylinositol 4-kinase type 2-alpha deficiency	*PI4K2A*	AR	609763	High forehead	Bulbous nose		Epicanthal folds Hypertelorism	Long ears			Sialotransferrins (S), Lactate (P)
Phosphatidylinositol 4,5-bisphosphate phospholipase C β4 deficiency	*PLCB4*	AD, AR	614669	Micrognathia		Cleft palate Glossoptosis Microstomia		Cupped pinna Low-set ears Auricular clefts Dysplastic ears Posteriorly rotated ears			DNA
Phosphatidylinositol 3,4,5-trisphosphate 3-phosphatase deficiency	*PTEN*	AD	601728			Arched palate, high Microstomia Scrotal tongue					DNA
Phosphatidylinositol 3-kinase regulatory subunit 2 superactivity	*PIK3R2*	AD	603387	Frontal bossing	Depressed nasal bridge Prominent philtral groove	Tent-shaped mouth	Large eyes				DNA
Myotubularin 1 deficiency	*MTM1*	XL	310400	Narrow, elongated face		Arched palate, high					DNA
Disorders of ether lipid metabolism											
Fatty Acyl-CoA reductase 1deficiency	*FAR1*	AR	616154	Coarse facial features Facial dysmorphism	Long philtrum			Long ears			Plasmalogens (Ec)
Disorders of sphingolipid synthesis and recycling											
Hedgehog acyltransferase deficiency	*HHAT*	AR	608116			Large mouth	Puffy eyelids Upslantine palpebral fissures				DNA
Alkaline ceramidase 3 deficiency	*ACER3*	AR	617762	Coarse facial features	Flat philtrum			Low-set ears		Short stature	DNA
Arachidonate lipoxygenase 3 deficiency	*ALOXE3*	AR	242100			Eclabium					DNA
Arachinodate 12R-lipoxygenase deficiency	*ALOX12B*	AR	242100			Eclabium					DNA
Fatty acid 2-hydroxylase deficiency	*FA2H*	AR	612319				Exotropia				DNA

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
Sphingomyelin synthase 2 deficiency	*SGMS2*	AR	611574	Midface hypoplasia	Depressed nasal bridge						DNA
Disorders of sterol biosynthesis											
Mevalonate kinase deficiency	*MVK*	AR	610377	Triangular-shaped face Dolichocephaly			Down-slanting palpebral fissures	Dysplastic ears			Leucotriens (P), Organic acids (U)
NSDHL deficiency CK syndrome	*NSDHL*	XL	300891	Long, narrow face Micrognathia Retrognathia	High nasal bridge	High arched palate	Almond-shaped eyes Epicanthal folds Upslanting palpebral fissures	Posteriorly rotated ears			Sterols (P)
7-dehydrocholesterol reductase deficiency Smith-Lemli-Opitz syndrome	*DHCR7*	AR	270400	Bitemporal narrowing Micrognathia	Anteverted nares Broad nasal tip	Epicanthic folds	Strabismus	Low-set ears			Sterols (P)
Desmosterolosis	*DHCR24*	AR	602398	Micrognathia		Cleft palate					Sterols (P)
Lathosterolosis	*SC5D*	AR	607330	Micrognathia Bitemporal narrowing	Anteverted nares	Cleft palate			Toe syndactyly, 2-3 Toe syndactyly, 2-5		Sterols (P)
Chondrodysplasia punctata 2	*EBP*	XL	302960	Micrognathia Midface hypoplasia	Anteverted nares Nasal hypoplasia	Cleft palate			Toe syndactyly, 2-3 Toe syndactyly, 2-5		Sterols (P)
SC4MOL deficiency	*MSMO1*	AR	607545	Frontal bossing		Arched palate, high				Short stature	Sterols (P)
Squalene synthase deficiency	*FDFT1*	AR	184420	Micrognathia	Depressed nasal bridge Square nasal tip		Epicanthal folds	Large ears Posteriorly rotated ears	Dorsal foot fat pads Syndactyly of the second and third toes		Sterols (P), LDL/HDL cholesterol (P), Triglycerides (P), Organic acids (U)
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=38)											
Disorders of pyrimidine and purine metabolism											
Dihydroorotate dehydrogenase deficiency	*DHODH*	AR	263750	Malar hypoplasia Micrognathia	Choanal atresia	Cleft lip Cleft palate		Cup-shaped ears Low-set ears		Accessory nipples	Purines and pyrimidines (U, P)
Dihydropyrimidinase deficiency	*DPYS*	AR	222748 613326	Coarse facies	Broad nasal bridge	Thin upper lip vermillion	Widely spaced eyes	Prominent ears	Underdeveloped distal phalanges of 3rd fingers Clinodactyly of the fifth finger		Purines and pyrimidines (U, P)
Beta-ureidopropionase deficiency	*UPB1*	AR	613161;606673				Widely spaced eyes Strabismus				
Phosphoribosyl pyrophosphate synthetase 1 superactivity	*PRPS1*	XL	300661	Facial dysmorphism							Lactate (P), DNA
Adenosine monophosphate deaminase 3 deficiency	*AMPD2*	AR	615809 615686	Midface hypoplasia	Broad nasal bridge	Short upper lip Macroglossia		Dysplastic ears			DNA
Phosphoribosylaminoimidazole carboxylase deficiency	*PAICS*	AR	172439	Craniofacial dysmorphism	Anteverted nares Choanal atresia Depressed nasal bridge Nasal hypoplasia		Hypertelorism			Short stature	DNA
AICAR transformylase-IMP cyclohydrolase deficiency	*ATIC*	AR	608688 601731	Coarse facies	High nasal bridge Anteverted nares	Wide mouth Thin upper lip		Low set ears			Purines and pyrimidines (U)
Disorders of ectonucleotide and nucleic acid metabolism											
PRUNE1 deficiency	*PRUNE1*	AR	617481	Dysmorphic features		Narrow palate					DNA
RNA-specific adenosine deaminase 2 deficiency	*ADARB1*	AR	618862	Prominent chin	Depressed nasal bridge Short philtrum	Tented upper lip	Epicanthal folds with telecanthus Upslanting palpebral fissures	Indented and pointed helices			DNA
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases											
CLP1 deficiency	*CLP1*	AR	615803		Broad nasal roots Short nose with upturned nostrils		Long palpebral fissures and eyelashes with prominent eyes				DNA
tRNA methyltransferase 1 deficiency	*TRMT1*	AR	618302	Hypoplastic maxilla	Broad nasal bridge		Synophrys Narrow palpebral fissures Broad eyebrows	Protruding ears			DNA
tRNA-specific adenosine deaminase 3 deficiency	*ADAT3*	AR	615286	Prominent forehead Micrognathia	Prominent nose	Wide mouth Full lipss High arched palate	Strabismus Epicanthal fold Widely spaced eyes Upslanting palpebral fissures Synophrys	Low-set ears Large ears			DNA
Galloway-Mowat syndrome, GON7 type	*GON7*	AR	617436	Narrow forehead	Depressed nasal bridge Pinched nose		Almond-shaped eyes Hypotelorism	Large ears Low set ears			DNA
Galloway-Mowat syndrome type 2	*LAGE3*	XL	301006	Micrognathia			Hypertelorism				DNA
Galloway-Mowat syndrome type 3	*OSGEP*	AR	617729	Micrognathia			Hypertelorism				DNA
Galloway-Mowat syndrome type 4	*TP53RK*	AR	617730				Widely spaced eyes	Large ears	Tapered fingers	Short stature	DNA
Galloway-Mowat syndrome type 5	*TPRKB*	AR	617731	Elongated face Prominent chin			Epicanthal folds Widely spaced eyes Deep-set eyes	Fleshy lobes			DNA
Glutaminyl-tRNA synthetase 1 deficiency	*QARS1*	AR	615760				Hypotelorism				DNA
Pseudouridine synthase 3 deficiency	*PUS3*	AR	617051	Coarse face							DNA
NSUN2 deficiency	*NSUN2*	AR	611091	Narrow, elongated face			Esotropia			Short stature	DNA
Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome	*CTU2*	AR	618142	Micrognathia							DNA
AIMP2/p38 deficiency	*AIMP2*	AR	618006	Triangular face	Broad nose Short philtrum	Thin upper lip			Long fingers		DNA
Disorders of ribosomal biogenesis											
NEPRO-related skeletal dysplasia	*NEPRO*	AR	618853	Midface hypoplasia Retrognathia Hypoplastic maxilla Wide anterior fontanel	Depressed nasal bridge Bulbous nose		Broad eyebrows Medially sparse eyebrows		Short hands and feet Hypoplastic nails	Sparse scalp hair	DNA
POLR3GL deficiency	*POLR3GL*	AR	619234	Retrognathia Midface hypoplasia	Beaked nose Long columella High nasal bridge	Thin lips Downturned mouth	Upslanting palpebral fissures Widely spaced eyes	Posteriorly rotated ears Low set ears			DNA
Treacher Collins syndrome type 1	*TCOF1*	AD	154500	Malar hypoplasia Micrognathia			Down-slanting palpebral fissures				DNA
Treacher Collins syndrome type 2	*POLR1D*	AD, AR	613717	Malar hypoplasia			Down-slanting palpebral fissures				
Treacher Collins syndrome type 3	*POLR1C*	AR	248390			Cleft palate	Down-slanting palpebral fissures				DNA

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
Treacher Collins syndrome type 4	*POLR1B*	AD	618939	Craniofacial dysmorphism Malar hypoplasia				Microtia			DNA
Acrofacial dysostosis, Cincinnati type	*POLR1A*	AD	616462	Craniofacial dysmorphism Micrognathia							DNA
Wiedemann-Rautenstrauch syndrome	*POLR3A*	AR	264090	Progeroid appearance						Short stature	DNA
POP1 deficiency	*POP1*	AR	617396	Midface hypoplasia							DNA
Bowen-Conradi syndrome	*EMG1*	AR	211180	Micrognathia							DNA
Diamond-Blackfan anemia type 14	*TSR2*	XL	300946	Micrognathia		Cleft palate					DNA
Diamond-Blackfan anemia type 15	*RPS28*	AD	606164	Malar hypoplasia Micrognathia		Cleft palate					DNA
Cytosolic large ribosomal subunit 10 deficiency	*RPL10*	XL	300998	Prognathism	Flat philtrum Long philtrum						DNA
Cytosolic small ribosomal subunit 23 deficiency	*RPS23*	AD	617412		Depressed nasal bridge		Epicanthal folds	Large malformed ears	Single transverse palmar crease	Short stature	DNA
Poly(A)-specific ribonuclease deficiency	*PARN*	AR	616353	Midface hypoplasia							
Exoribonuclease 1 deficiency	*ERI1*	AR	608739	Plagiocephaly			Ptosis	Posteriorly rotated ears	Syndactyly Short metacarpals Nail hypoplasia Clinodactyly Brachydactyly	Mongolian spot	DNA
COMPLEX MOLECULE AND ORGANELLE METABOLISM (n=183)											
Disorders of N-linked protein glycosylation											
ALG13-CDG UDP-N-acetylglucosamine transferase catalytic subunit deficiency	*ALG13*	XL	300884	Broad coarse facies Retrognathia			Widely spaced eyes	Low set ears	Small feet Small hands		DNA
ALG1-CDG Mannosyltransferase 1 deficiency	*ALG1*	AR	608540		Anteverted nares Broad nasal bridge	Thin lips Small mouth	Widely spaced eyes Short palpebral fissures	Large ears Cupped ears			Sialotransferrins (S), Dolichol-linked GlcNAc2 (S), Protein C (S), Xeno-tetrasaccharide (S, FB)
ALG11-CDG Mannosyltransferase 1 deficiency	*ALG11*	AR	613661	High forehead Retrognathia Small head	Long philtrum					Inverted nipples	Sialotransferrins (S), Dolichol-linked Man3/4GlcNAc2 (S), Prolactin (P), Factor XI (B), Antithrombin III (P)
ALG9-CDG Mannosyltransferase 7-9 deficiency	*ALG9*	AR	608776	Frontal bossing	Depressed nasal bridge	Large mouth	Widely spaced eyes	Low set ears	Broad thumbs	Inverted widely spaced nipples	
ALG12-CDG Mannosyltransferase 8 deficiency	*ALG12*	AR	607143	Midface hypoplasia Dolichocephaly Retrognathia	Prominent nose		Deep-set eyes		Short metacarpals		Sialotransferrins (S), ASAT/ALAT (P), Glucose (S), Lipid panel (S),IGF BP3, Antithrombin III (P)
ALG8-CDG Glucosyltransferase 2 deficiency	*ALG8*	AD, AR	608104	Asymmetric skull	Long philtrum		Widely spaced eyes	Low set ears Posteriorly rotated ears	Brachydactyly Clubfoot Camptodactyly	Short neck Inverted nipples	Sialotransferrins (S), Albumin (S), Factor IX and XI (B), Antithrombin III (P), CK (P)
TUSC3-CDG Oligosaccharyltransferase subunit tusc 3 deficiency	*TUSC3*	AR	611093	Long face Pointed chin			Widely spaced eyes Deep-set eyes		Camptodactyly Syndactyly		DNA
MOGS-CDG Glucosidase 1 deficiency	*MOGS*	AR	606056	Subtle facial dysmorphia Retrognathia Prominent occiput	Broad nose	High arched palate	Short palpebral fissures Long eyelashes				Sialotransferrins (S), N-glycans (S), ASAT/ALAT (P), Oligosaccharide (U)
MGAT2-CDG N-acetylglucosaminyltransferase 2 deficiency	*MGAT2*	AR	212066	Retrognathia	Beaked nose High nasal bridge Long philtrum	Thin upper lip Open mouth Gum hypertrophy	Long eyelashes Thick eyebrows	Large ears		short neck	Sialotransferrins (S), ASAT/ALAT (P), CK (P), Factor IX and XII (B), Antithrombin III (P), N-glycans (S, FB)
FUT8-CDG α-1,6-fucosyltransferase deficiency	*FUT8*	AR	618005	Broad forehead Retrognathia Bitemporal narrowing Hirsutism	Broad nasal bridge Short nose	High palate	Buphthalmos				N-glycans missing core fucose (S, FB), Glucose (P), Blood count, DNA
B4GALT1-CDG Beta-1,4-galactosyltransferase 1 deficiency	*B4GALT1*	AR	607091	Prominent forehead	Long philtrum	Thin upper lip	Widely spaced eyes	Low set ears			Sialotransferrins (S), Antithrombin (B), Factor XI (B), N-glycans (S, FB)
OSTC-CDG Oligosaccharyltransferase complex-CDG	*OSTC*	AR	619023	Micrognathia Retrognathia	High nasal bridge	Full lips V-shaped cleft palate	Widely spaced eyes Downward palpebral fissures Exotropia	Low set ears	Transverse palmar crease Camptodactyly		Sialotransferrins (S)
SSR3-CDG Translocon associated protein complex subunit SSR3 deficiency	*SSR3*	AR	606213	Micrognathia					Clinodactyly of the fourth and fifth toes		Sialotransferrins (S)
PMM2-CDG	*PMM2*	AR	601785	Prominent forehead Prominent jaw	Prominent nose Long philtrum	Thin upper lip High arched palate	Almond-shaped eye	Large ears Large lobes Dysplastic ears	Long fingers and toes Slender fingers and toes	Cryptorchidism	Sialotransferrins (S)
STT3A-CDG	*STT3A*	AD	615596 601134	Large fontanelles Prognathism High anterior hairline		Thin upper lip	Short palpebral fissures			Inverted nipples Short stature	Sialotransferrins (S)
LYSET-CDG TMEM251 deficiency	*LYSET*	AR	619345	Coarse facies	Broad nose	Full lips				Short stature	Alpha-iduronidase, Iduronate-2-sulfatase (WBC)
MAN2B2-CDG	*MAN2B2*	AR	618899	Micrognathia		Cleft palate	Widely spaced eyes			Hypospadias	Sialotransferrins (S), C-reactive protein (P), CRP (P), IgE (S)
DPAGT1-CDG UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency	*DPAGT1*	AR	191350, 608093	Micrognathia		High arched palate	Strabismus Exotropia				Sialotransferrins (S), Antithrombin III (P)
GNPTAB-CDG UDP-N-acetylglucosamine-1-phosphotransferase subunit alpha/beta deficiency	*GNPTAB*	AR	252500	Coarse facial features		Macroglossia					N-glycans (S), Enzyme activity (S), Oligosaccharides (U)
GNPTG-CDG UDP-N-acetylglucosamine-1-phosphotransferase subunit gamma deficiency	*GNPTG*	AR	252605	Coarse facial features							N-glycans (S), Enzyme activity (S), Oligosaccharides (U)
RFT1-CDG Flippase of Man5GlcNAc2-PP-Dol deficiency	*RFT1*	AR	611908, 612015							Inverted nipples	Sialotransferrins (S), Dolichol-linked Man5GlcNAc2 (S), Prolactin (P), Factor XI (B), Antithrombin II (P)
ALG3-CDG Mannosyltransferase 6 deficiency	*ALG3*	AR	608750, 601110	Micrognathia			Strabismus		Clubfoot Long fingers		Sialotransferrins (S), Antithrombin III (P), Apolipoprotein B (S). Protein S (S). Glucose (P)
SSR4-CDG Signal sequence receptor 4 protein (TRAP complex) deficiency	*SSR4*	XL	300090	Prominent forehead			Strabismus		Clinodactyly	Hypospadias	Sialotransferrins (S)
EDEM3-CDG Endoplasmic reticulum degradation-enhancing alpha-mannosidase-like protein 3 deficiency	*EDEM3*	AR	610214 619491	Retrognathia	Bulbous nose	Thin upper lip	Epicanthal folds Strabismus				DNA, N-glycans (S, FB)
MAN1B1-CDG Mannosyl-oligosaccharide alpha-1,2-mannosidase deficiency	*MAN1B1*	AR	604346, 614202	Flat oval face	Bulbous nose	Thin upper lip	Strabismus			Inverted nipples	Sialotransferrins (S), N-glycans (S, FB)
MAN2A2-CDG Alpha-mannosidase IIA deficiency	*MAN2A2*	AR	600988	Brachycephaly	Broad nose with thick overlying skin	Thick lips	Broad eyebrows Epicanthal fold		Tapering fingers		DNA
Disorders of O-linked protein glycosylation											
OGT-CDG O-linked N-acetylglucosamine transferase deficiency	*OGT*	XL	300997	Retrognathia Dolichocephaly Prominent jaw	Broad nose	Full lipss	Widely spaced eyes Amblyopia	Low set ears	Hypoplastic toe Long fingers Thin fingers Clinodactyly	Supernumerary nipple	DNA
EOGT-CDG EGF domain-specific O-linked N-acetylglucosamine transferase deficiency	*EOGT*	AR	615297	Aplasia cutis Bony defect of skull					Terminal transverse defect of toes Nail hypoplasia Tapered digits		DNA
EXT2-CDG Exostosin glycosyltransferase 2 deficiency	*EXT2*	AR	616682	Coarse facies Prominent forehead Sparse hair	Prominent nasal tip Long philtrum	Decayed teeth	Hypotelorism	Large ears			DNA
TMTC3-CDG	*TMTC3*	AR	617255				Microphthalmia				DNA

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
B3GALTL-CDG O-Fucose-specific beta-1,3-N-glucosyltransferase deficiency	*B3GLCT*	AR	261540	Prominent forehead		Long filtrum Cleft lip Cleft palate	Short palpebral fissures			Broad neck	DNA
POMT1-CDG O-Mannosyltransferase 1 deficiency	*POMT1*	AR	607423				Buphthalmos Microphthalmia Exophthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
POMT2-CDG O-Mannosyltransferase 2 deficiency	*POMT2*	AR	607439,				Buphthalmos Microphthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
POMGNT1-CDG O-Mannose beta-1,2-N-acetylglucosaminyltransferase deficiency	*POMGNT1*	AR	606822				Buphthalmos Microphthalmia Exophthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
POMGNT2-CDG O-mannose β-1,4-N-acetylglucosaminyltransferase deficiency	*POMGNT2*	AR	614828				Macrophthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
B3GALNT2-CDG β-1,3-galactosaminyltransferase 2 deficiency	*B3GALNT2*	AR	610194				Microphthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
CRPPA-CDG 2-C-methyl-D-arythritol 4-phosphate cytidyltransferase deficiency	*CRPPA*	AR	614631				Microphthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
FKTN-CDG Fukutin deficiency	*FKTN*	AR	607440/ 611588				Microphthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
FKRP-CDG Fukutin-related protein deficiency	*FKRP*	AR	606596				Microphthalmia				Matriglycan-specific monoclonal antibody, CK (P), DNA
GALNT2-CDG UDP-N-acetyl-alpha-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 2 deficiency	*GALNT2*	AR	602274/ 618885	Long face Prominent jaw	Short philtrum						HDL cholesterol (P), DNA
XYLT1-CDG Xylosyltransferase 1 deficiency	*XYLT1*	AR	608124/ 615777	Flat oval face	Depressed nasal bridge	Cleft palate			Brachydactyly Clubfoot Short metacarpals	Short stature	DNA
B4GALT7-CDG Beta-1,4-galactosyltransferase 7 deficiency	*B4GALT7*	AR	604327/ 130070	Frontal bossing Flat midface					Long fingers Fingers, overlapping	Short stature	DNA
FAM20B-CDG Glycosaminoglycan xylosylkinase deficiency	*FAM20B*	AR	611063	Flat oval face Midface hypoplasia	Depressed nasal bridge				Short limbs Thumb hypoplasia	Short neck Short stature	DNA
B3GALT6-CDG β-1,3-galactosyltransferase 6 deficiency	*B3GALT6*	AR	615291/ 271640;61534 9	Flat midface		Cleft palate				Pectus carinatum Short stature	DNA
B3GAT3-CDG β-1,3-glucuronyltransferase 3 deficiency	*B3GAT3*	AR	606374/ 245600	Flat midface		Cleft palate			Clubfoot Fingers, overlapping	Short stature	DNA
HS2ST1-CDG Heparan sulfate 2-O-sulfotransferase 1 deficiency	*HS2ST1*	AR	619194		Prominent nasal tip	Wide mouth	Upslanting palpebral fissures				DNA
HS6ST1-CDG Heparan sulfate 6-O-sulfate transferase 1 deficiency	*HS6ST1*	AD	604846;6148 80			Cleft palate					DNA
HS6ST2-CDG Heparan sulfate 6-O-sulfotransferase 2 deficiency	*HS6ST2*	XL	301025	Flat midface Triangular face Bitemporal narrowing	Broad nose tip	Thin upper lip	Deeply set eyes	Low set ears			DNA
CSGALNACT1-CDG Chondroitin sulfate N-acetylgalactosaminyltransferase 1 deficiency	*CSGALNACT1*	AR	616615	Flat midface						Short stature	DNA
DSE-CDG Dermatan sulfate epimerase deficiency	*DSE*	AR	605942/ 615539				Down-slanting palpebral fissures		Long digits Tapered digits		DNA
CHST14-CDG Dermatan 4-sulfotransferase 1 deficiency	*CHST14*	AR	608429/ 601776				Down-slanting palpebral fissures		Long tapered fingers and toes		DNA
CANT1-CDG UDP-galactose nucleotidase deficiency	*CANT1*	AR	613165/ 617719	Microretrognathia Midface hypoplasia Round face	Short nose	Microstomia	Prominent eyes				DNA
SLC26A2-CDG Sulfate transporter deficiency	*SLC26A2*	AR	606718			Cleft palate		Cauliflower ear			DNA
BPNT2-CDG Golgi-resident phosphoadenosine phosphate phosphatase deficiency	*BPNT2*	AR	614010/ 614078	Facial dysmorphism Micrognathia		Cleft palate					DNA
SLC35B2-CDG Phosphoadenosine 5'-phosphosulfate transporter deficiency	*SLC35B2*	AR	620269			Cleft palate					DNA
Disorders of lipid glycosylation											
C18orf32-CDG Chromosome 18 open reading frame 32 deficiency	*C18orf32*	AR	619985							Inverted nipples	Alkaline phosphatase (P), DNA
PIGA-CDG Phosphatidylinositol glycan anchor biosynthesis class A protein deficiency	*PIGA*	XL	311770, 300868;30081 3	Coarse face	Broad nasal bridge						GPI-anchored proteins (WBC,FB), ALP (P),
PIGC-CDG Glycosylphosphatidylinositol glycan anchor biosynthesis C protein deficiency	*PIGC*	AR	601730, 615716	Coarse face							Flow cytometry of GPI markers (PLT)
PIGN-CDG Multiple congenital anomalies-hypotonia-seizures syndrome type 1	*PIGN*	AR	614080	Coarse facies Bitemporal narrowing	Depressed nasal bridge	Open mouth Thin lips		Large ears with overfolded helices			GPI-anchored proteins (WBC,FB), ALP (P)
PIGO-CDG	*PIGO*	AR	614749		Short nose Broad nasal bridge	Tented mouth	Wide spaced eyes		Nail hypoplasia Absent nails of the fifth digits Brachytelephalangy		GPI-anchored proteins (WBC,FB), ALP (P)
PIGU-CDG Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis	*PIGU*	AR	618590	Long face High forehead Bitemporal narrowing	Depressed nasal bridge	Thin upper lip Large mouth High arched palate	Strabismus Epicanthal folds	Low-set or posteriorly rotated ears			Flow cytometry of GPI markers (PLT)
PIGK-CDG	*PIGK*	AR	618879	Long face Triangular face Myopathic face	Anteverted nose Short nose Broad nose Long philtrum	Tented lip Thin upper lip	Epicanthal fold Synophrys Hypotelorism Strabismus Esotropia High-arched eyebrows Sparse lateral eyebrows	Dysplastic ears Low set ears			Flow cytometry of GPI markers (PLT)
PGAP1-CDG	*PGAP1*	AR	615802	Prominent forehead		Large mouth Abnormal teeth	High-arched eyebrows		Abnormal hand morphology	Short neck	DNA
PIGV-CDG Phosphatidylinositol glycan anchor biosynthesis class V protein deficiency	*PIGV*	AR	610274, 239300		Broad nasal bridge		Hypertelorism Upslanting palpebral fissures				GPI-anchored proteins (WBC,FB), ALP (P)
PIGY-CDG Phosphatidylinositol glycan anchor biosynthesis class Y, deficiency	*PIGY*	AR	610662, 239300					Large lobes			DNA, ALP (P)
PIGT-CDG Phosphatidylinositol glycan anchor biosynthesis class T protein deficiency	*PIGT*	AR	610272, 615398		Depressed nasal bridge Long philtrum					Inverted nipples	GPI-anchored proteins (WBC,FB), ALP (P)
PIGS-CDG Phosphatidylinositol glycan anchor biosynthesis class S protein deficiency	*PIGS*	AR	610271, 618143	Coarse facial features		Macroglossia					Flow cytometry of GPI markers (PLT)
PIGF-CDG Phosphatidylinositol glycan anchor biosynthesis class F protein deficiency	*PIGF*	AR	600153, 619356	Coarse face Micrognathia	Short nose	Microstomia	Upslanting palpebral fissures				Flow cytometry of GPI markers (PLT)
PIGB-CDG	*PIGB*	AR	604122	Coarse facies Pointed chin Full cheeks Micrognathia	Broad nasal bridge	Tented upper lip Protruding tongue	Widely spaced eyes	Upturned earlobes			GPI-anchored proteins (WBC,FB), ALP (P)

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
PGAP3-CDG PER1-like domain-containing protein 1 deficiency	*PGAP3*	AR	611801	Micrognathia		Cleft palate					GPI-anchored proteins (WBC,FB), ALP (P)
Disorders of multiple glycosylation pathways											
GMPPA-CDG GDP-mannose pyrophosphorylase B deficiency	*GMPPA*	AR	615510	Triangular face Prominent jaw Prominent forehead	Short philtrum	Downturned mouth					DNA
NPL-CDG	*NPL*	AR	611412							Retrognathia	Sialic acid, free (U)
SLC35A2-CDG	*SLC35A2*	XL	314375	Coarse facies							Sialotransferrins (S), N-glycans (S)
ATP6V1A-CDG	*ATP6V1A*	AR	617403	Triangular face Retrognathia Recessed forehead Short pointed chin	Beaked nose Broad nasal tip Narrow base and nares		Strabismus Widely spaced eyes Entropion Small palpebral fissures	Prominent ears Low set ears			Sialotransferrins (S), Lipid panel (S)
SRD5A3-CDG Steroid 5 alpha-reductase 3 deficiency	*SRD5A3*	AR	612379	Facial dysmorphism							Sialotransferrins (S), Antithrombin III (P), Protein S (S)
NANS-CDG N-acetylneuraminic acid synthase deficiency	*NANS*	AR	610442	Prominent forehead Coarse facies Brachycephaly	Broad nose Depressed nasal bridge	Full lipss	Synophrys	Flat ears			N-acetyl-D-mannosamine (P,U)
ATP6V1E1-CDG	*ATP6V1E1*	AR	617402	Short forehead Prominent jaw Saggy cheeks	Saddle nose Anteverted nares	Crowding of the teeth Midline cleft palate	Blepharophimosis Strabismus Entropion Infraorbital puffiness		Mild camptodactyly Overriding toes		Sialotransferrins (S)
SLC35A3-CDG UDP-N-acetylglucosamine transporter deficiency	*SLC35A3*	AR	615553	Micrognathia		Cleft palate	Proptosis	Cupped ears	Overlapping fingers Rocker bottom feet	Short limbs	N-glycans (S)
SLC35C1-CDG GDP-fucose transporter deficiency	*SLC35C1*	AR	266265	Oval face	Short nose Depressed nasal bridge					Short stature	N-glycans (S), Neutrophil motility and rolling, Sialyl-Lewis on neutrophils
DOLK-CDG Dolichol kinase deficiency	*DOLK*	AR	610768	Broad forehead	Flat nose		Widely spaced eyes	Low set ears Large lobes			Sialotransferrins (S), Lipid-linked oligosaccharide (F)
ATP6AP2-CDG	*ATP6AP2*	XL	300423	Micrognathia				Low set ears		Laterally facing nipples Hypospadias	Sialotransferrins (S), ASAT/ALAT (P), IgG (S), Factor XI (B), N and O-glycans (S)
PGM3-CDG Phosphoglucomutase 3 deficiency	*PGM3*	AR	615816 172100	Midface hypoplasia Micrognathia		Downturned corners of mouth				Short neck	N-and O-glycans (S), IgE (S)
DPM2-CDG Dolichol-P-mannose synthase-2 deficiency	*DPM2*	AR	615042				Strabismus				Sialotransferrins (S), ASAT/ALAT (P), CK (P)
CCDC115-CDG	*CCDC115*	AR	616828	Long face			Ptosis				Sialotransferrins (S), ALP (P), Ceruloplasmin (S), Cholesterol (S), N and O-glycans (S)
ATP6V0A2-CDG Lysosomal H(+)-transporting ATPase V0 subunit A2 deficiency	*ATP6V0A2*	AR	611716	Facial dysmorphism						Aged appearance	Sialotransferrins (S), ASAT/ALAT (P), N and O-glycans (S)
TMEM165-CDG TMEM165 (TPARL) protein deficiency	*TMEM165*	AR	614726 614727	Midface hypoplasia							Sialotransferrins (S), ASAT/ALAT (P), CK (P), N-glycans (S)
MPDU1-CDG Dol-P-Man utilization 1 deficiency	*MPDU1*	AR	609180	Retrognathia	Broad-based nose Smooth philtrum	Thin lips	Widely spaced eyes	Low-set, posterior-rotated ears			Sialotransferrins (S), Antithrombin III (P)
UGDH-CDG	*UGDH*	AR	618792	Pointed chin	Short flat philtrum	Full lower lip	Blepharophimosis Synophrys Deep-set eyes Epicanthal folds	Protruding earlobes			DNA
Glucose-6-phosphate transporter deficiency (SLC37A4-CDG)	*SLC37A4*	AD	602671/ 619525	Narrow, elongated face							Sialotransferrins (S), N-glycans (S), Coagulation factors (P), GGT (P)
Glucosamine-6-phosphate N-acetyltransferase 1 deficiency (GNPNAT1-CDG)	*GNPNAT1*	AR	616510						Large hands	Short stature	DNA
Phosphoglucomutase 1 deficiency (PGM1-CDG)	*PGM1*	AR	171900, 614921			Bifid uvula Cleft palate				Short stature	Sialotransferrins (S), N-glycans (S), Creatine kinase (P), Fatty acids and ketones (P,U)
UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase superactivity (GNE-CDG)	*GNE*	AD	603824	Coarse facies							N-Acetylneuraminic acid (U), Sialic acid, free (U)
UDP-glucose pyrophosphorylase 2 deficiency (UGP2-CDG)	*UGP2*	AR	191760, 618744	Facial dysmorphism	Depressed nasal bridge		Arched eyebrows				DNA
UDP-glucuronic acid-UDP-N-acetylgalactosamine dual transporter deficiency (SLC35D1-CDG)	*SLC35D1*	AR	610804/ 269250		Nasal hypoplasia				Clubfoot	Dwarfism	DNA
TDP-D-glucose 4,6-dehydrogenase deficiency (TGDS-CDG)	*TGDS*	AR	616145	Micrognathia		Cleft palate			Hyperphalangy of index finger Radial deviation of the index finger	Short stature	DNA
Disorders of deglycosylation											
N-glycanase 1 deficiency	*NGLY1*	AR	615273		Depressed nasal bridge	Small mouth High arched palate	Strabismus Long palpebral fissures Widely spaced eyes	Low set ears	Small hands Small feet Clasped thumb Clinodactyly		Oligosaccharides (U), GlcNAc-Asn (U)
MAN2C1 deficiency	*MAN2C1*	AR	619775	Facial dysmorphism							DNA
Disorders of mitochondrial membrane biogenesis and remodeling											
Phosphatidylserine decarboxylase deficiency	*PISD*	AR	612770	Midface hypoplasia	Depressed nasal bridge				Brachydactyly		DNA
Tafazzin deficiency	*TAZ*	XL	302060	Cherubic face							Organic acids (U), Acylcarnitines (P)
PTPMT1 deficiency	*PTPMT1*	AR	609538	Sloping forehead	Bulbous nose High-arched palate Long philtrum		Epicanthal folds Exotropia Hypertelorism Upslanting palpebral fissures	Large ears Low-set ears			DNA
Peroxisomal biogenesis disorders											
Peroxin 1 deficiency Zellweger	*PEX1*	AR	234580 214100 601539	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 2 deficiency Zellweger	*PEX2*	AR	614866 614867	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 3 deficiency Zellweger	*PEX3*	AR	617370 614882	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 6 deficiency Zellweger	*PEX6*	AR	614862 614863 616617	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 10 deficiency Zellweger	*PEX10*	AR	614870 614871	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 14B deficiency Zellweger	*PEX14*	AR	614920	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 12 deficiency Zellweger	*PEX12*	AR	614859 266510	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 13 deficiency Zellweger	*PEX13*	AR	614883 614885	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 14 deficiency Zellweger	*PEX14*	AR	614887	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 16 deficiency Zellweger	*PEX16*	AR	614876 614877	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Peroxin 19 deficiency Zellweger	*PEX19*	AR	614886	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
Peroxin 26 deficiency Zellweger	*PEX26*	AR	614872 614873	High forehead	Broad nasal bridge	Slender upper lip High arched palate	Widely spaced eyes Epicanthal fold	Square ears Low set ears	Clubfoot		VLCFA (P), Pipecolic acid (P)
Disorders of vesicular trafficking											DNA, N and O-glycans
VPS51-CDG	*VPS51*	AR	618606			Open mouth Thick upper lip vermilion High arched palate	Hypotelorism Strabismus		Flat feet		DNA, N and O-glycans
STX16-CDG	*STX16*	AD	603233	Round face						Short stature	DNA, Calcium (P), Phosphate (P), Parathyroid hormone, PTH (S)
GM130-CDG	*GM130*	AR	620240	Prominent forehead		Tender upper lip					CK (P), DNA
COG2-CDG Conserved oligomeric Golgi complex subunit 2 deficiency	*COG2*	AR	606974	Facial dysmorphism							Sialotransferrins (S), Ceruloplasmin (S), Copper (S)
COG3-CDG Conserved oligomeric Golgi complex subunit 3 deficiency	*COG3*	AR	606975	Long face	Long philtrum Prominent nose	Thin lips	Widely spaced eyes	Low set ears			Sialotransferrins (S)
COG4-CDG Conserved oligomeric Golgi complex subunit 4 deficiency	*COG4*	AR	606976 613489	Micrognathia					Clubfoot	Short stature	Sialotransferrins (S)
TRAPPC9-CDG TRAPPC9Trafficking protein particle complex 9 deficiency	*TRAPPC9*	AR	613192		Broad nasal bridge		Hypertelorism				DNA
Archain 1 deficiency	*ARCN1*	AD	617164	Micrognathia		Cleft palate				Short stature	DNA
Craniolenticulosutural dysplasia	*SEC23A*	AR	607812	High forehead	Anteverted nares Prominent nose	Bifid uvula Cleft palate	Esotropia Hypertelorism				DNA
SCYL1-binding protein deficiency	*GORAB*	AR	231070	Malar hypoplasia				Long ears			DNA
AP1S2 deficiency	*AP1S2*	XL	304340	Coarse facial features							DNA
AP4B1 deficiency	*AP4B1*	AR	607245	Bitemporal narrowing Coarse face	Bulbous nose Short philtrum Wide nasal bridge	Wide mouth					DNA
AP4E1deficiency	*AP4E1*	AR	607244	Bitemporal narrowing Coarse facial features Narrow, elongated face	Bulbous nose Short philtrum Wide nasal bridge	Wide mouth	Down-slanting palpebral fissures			Short stature	DNA
AP4M1 deficiency	*AP4M1*	AR	602296	Bitemporal narrowing Coarse face	Broad nasal bridge Bulbous nose Short philtrum	Arched palate, high Wide mouth					DNA
AP4S1 deficiency	*AP4S1*	AR	607243	Coarse facial features	Broad nasal bridge Bulbous nose Short philtrum	Big open mouth	Hypertelorism				DNA
RAB18 deficiency	*RAB18*	AR	614222				Microphthalmia				DNA
RAB3GAP1 deficiency	*RAB3GAP1*	AR	600118				Microphthalmia				DNA
RAB3GAP2 deficiency	*RAB3GAP2*	AR	614225				Microphthalmia				DNA
VPS4A deficiency	*VPS4A*	AR	600982				Exotropia				DNA
ATP9A deficiency	*ATP9A*	AR	609126		Smooth philtrum	Thin vermillion of the upper lip High palate					DNA
RAB5C deficiency	*RAB5C*	AR	604037	Coarse facial features							DNA
Golgin A2 deficiency	*GOLGA2*	AR	620240	Micrognathia		Tented upper lip					DNA
Golgin A2 deficiency	*DEDHMB*	AR	620240	Micrognathia		Tented upper lip					Creatine kinase (P), DNA
Disorders of lysosome-related organelle biogenesis											
Arthrogryposis-renal dysfunction-cholestasis syndrome type 1	*VPS33B*	AR	208085	Sloping forehead Micrognathia		High arched palate		Low set ears	Transverse palmar crease Clubfeet Large hands Proximally placed thumbs	Barrel-shaped chest Congenital hip dislocation	DNA
Hermansky-Pudlak syndrome type 2	*AP3B1*	AR	608233	Retrognathia	Broad nasal root Long philtrum	Thin upper lip	Epicanthal folds	Low-set and posteriorly rotated ears			DNA
Hermansky-Pudlak syndrome type 7	*DTNBP1*	AR	614075				Exotropia				DNA
Hermansky-Pudlak syndrome type 10	*AP3D1*	AR	617050				Hypotelorism				DNA
Mucopolysaccharidosis-plus syndrome	*VPS33A*	AR	617303	Coarse facial features		Macroglossia				Hernias	Mucopolysaccharides (U)
Griscelli syndrome type 1	*MYO5A*	AR	214450				Exotropia				DNA
GFAP deficiency Alexander disease	*GFAP*	AD	203450			High arched palate				Short neck Kyphosis	DNA
Disorders of organelle interplay											
BAP31 deficiency	*BCAP31*	XL	300475	Long face	Short nose		Congenital strabismus Hypotelorism	Large ears			DNA
EMC1 deficiency	*EMC1*	AR	616875		Short philtrum		Esotropia			Retrognathia	DNA
Acyl-CoA-binding domain-containing protein 5 deficiency	*ACBD5*	AR	616618		Tubular nose		Hypotelorism Ptosis	Prominent ears			VLCFA (P), Pipecolic acid (P)
COG1-CDG Conserved oligomeric Golgi complex subunit 1 deficiency	*COG1*	AR	611209	Micrognathia	Broad nasal bridge Long philtrum	Small mouth High palate	Downslanting palpebral fissures Widely spaced eyes	Posteriorly rotated ears Low set ears Microtia			Sialotransferrins (S)
COG6-CDG	*COG6*	AR	606977 614576	Retrognathia			Long palpebral fissures		Postaxial polydactyly		Sialotransferrins (S), ASAT/ALAT (P), CK (P), Lactate (P), Vitamins A, D, E, K (S)
COG7-CDG Conserved oligomeric Golgi complex subunit 7 deficiency	*COG7*	AR	608779	Flat face Micrognathia		Full lipss		Low set ears Dysplastic ears	Adducted thumbs	Short neck Loose, wrinkled skin Inverted nipples	Sialotransferrins (S), ASAT/ALAT (P), CK (P), Glucose (S), Total/direct bilirubin (S)
TRAPPC11-CDG	*TRAPPC11*	AR	615356	Full cheeks Retrognathia		Thin upper lip Everted lower lip			High arches Camptodactyly		CK (P), DNA
TRAPPC4 deficiency	*TRAPPC4*	AR	618741	Pointed chin Bitemporal narrowing	Prominent nasal tip Long philtrum	Wide mouth Open mouth Thin upper lip Tented upper lip					DNA
SCYL2 deficiency	*SCYL2*	AR	618766	Prominent forehead High anterior hairline	Depressed nasal bridge with bulbous nose Long philtrum	Small mouth Thin lips		Low set ears Dysplastic ears		Short neck	DNA
GET4-CDG	*GET4*	AR	612056	Bitemporal narrowing		Open mouth	Deep-set eyes Synophrys		Hindfoot valgus Clinodactyly toes	Brachycephaly	Sialotransferrins (S), N-Acetyl aspartate (CNS)
Disorders of autophagy											
ATG7 deficiency	*ATG7*	AR	619422	Long face Retrognathia	Smooth philtrum	Gum hypertrophy High arched palate					DNA

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
WIPI2 deficiency	*WIPI2*	AR	618453						Thumb hypoplasia Overriding toes Clinodactyly Camptodactyly	Short stature Kyphosis	DNA
EPG5 deficiency	*EPG5*	AR	242840	Micrognathia		Cleft lip Cleft palate	Hypertelorism	Low-set ears			IgG(S), DNA
SNX14 deficiency	*SNX14*	AR	616354	Coarse facial features Facial dysmorphism							DNA
TECPR2 deficiency	*TECPR2*	AR	615031	Round face						Short stature	DNA
TBCK deficiency	*TBCK*	AR	616899	Bitemporal narrowing Coarse face	Anteverted nares Bulbous nose High nasal bridge	Macroglossia Thick lips	Deeply set eyes				DNA
Other disorders of complex molecule degradation											
Alpha-iduronidase deficiency Hurler disease	*IDUA*	AR	607014	Coarse facies						Microdontia	Mucopolysaccharides (U), Enzyme testing
Alpha-galactosidase A deficiency Fabry disease	*GLA*	XL	301500	Recessed forehead Coarse facies	Prominent nose Bulbous nasal tip	Thick lips	Thick eyebrows	Prominent lobules of the ears			Globotriaosylceramide, Globotriaosylsphingosine
Alpha-glucosidase deficiency Pompe disease	*GAA*	AR	232300			Macroglossia					Oligosaccharides (U), Glycogen, ASAT/ALAT (P), CK (P)
Formyl-glycine generating enzyme deficiency	*SUMF1*	AR	268800	Coarse facial features							DNA
Beta-hexosaminidase subunit beta deficiency	*HEXB*	AR	268800	Doll-like face							Oligosaccharides (U), Glycogen, ASAT/ALAT (P), CK (P)
Beta-galactosidase 1 deficiency, Morquio B	*GLB1*	AR	230500	Coarse facial features							DNA
Neutral sphingomyelinase 3 deficiency	*SMPD4*	AR	618622		Depressed nasal bridge	Cleft lip Thin upper lip	Epicanthal folds Short palpebral fissures				DNA
Site-1 protease deficiency	*MBTPS1*	AR	618392	Prominent forehead and cheekbones				Large ears Posteriorly rotated ears			Glycosaminoglycans (U), Lysosomal enzymes (DBS)
Iduronate 2-sulfatase deficiency	*IDS*	XL	309900	Coarse facial features						Hernias	Mucopolysaccharides (U)
Heparan N-sulfatase deficiency	*SGSH*	AR	252900	Coarse facial features							Mucopolysaccharides (U)
N-acetylglucosaminidase deficiency	*NAGLU*	AR	252920	Coarse facial features							Mucopolysaccharides (U)
Heparan-alpha-glucosaminide N-acetyltransferase deficiency	*HGSNAT*	AR	252930	Coarse facial features							Mucopolysaccharides (U)
N-acetylglucosamine 6-sulfatase deficiency	*GNS*	AR	252940	Coarse facial features							Mucopolysaccharides (U)
N-acetylgalactosamine 6-sulfatase deficiency	*GALNS*	AR	253000	Coarse facial features							Mucopolysaccharides (U)
N-acetylgalactosamine 4-sulfatase deficiency	*ARSB*	AR	253200	Coarse facial features						Hernias	Mucopolysaccharides (U)
Beta-glucuronidase deficiency	*GUSB*	AR	253220	Coarse facial features						Short stature Hernias	Mucopolysaccharides (U)
Arylsulfatase K deficiency	*ARSK*	AR	610011	Coarse facial features							Mucopolysaccharides (U)
NEU1-CDG	*NEU1*	AR	256550	Coarse facial features						Hernias Short stature	Oligosaccharides (U)
Cathepsin A deficiency	*CTSA*	AR	256540	Coarse facial features						Hernias	Oligosaccharides (U)
Alpha-mannosidase B deficiency	*MAN2B1*	AR	248500	Coarse facial features Prognathism		Macroglossia				Hernias	Oligosaccharides (U)
Beta-mannosidase deficiency	*MANBA*	AR	248510	Coarse facial features						Short stature	Enzyme testing
Alpha-N-acetylgalactosaminidase deficiency	*NAGA*	AR	609242	Coarse facial features							Enzyme testing
Alpha-L-fucosidase deficiency	*FUCA1*	AR	230000	Coarse facial features						Short stature	Oligosaccharides (U)
Aspartylglucosaminidase deficiency	*AGA*	AR	208400	Coarse facial features						Hernias Short stature	Oligosaccharides (U)
Cathepsin K deficiency	*CTSK*	AR	265800		Aquiline nose					Short stature	DNA
COFACTOR AND MINERAL METABOLISM (n=29)											
Disorders of molybdenum cofactor metabolism											
Cyclic pyranopterin monophosphate synthase deficiency MoCo deficiency A	*MOCS1*	AR	603707	Frontal bossing Long face Prominent cheeks	Small nose Long philtrum	Thick lips	Enophthalmos Wide spaced eyes Long palpebral fissures Deep-set eyes				Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Molybdopterin synthase deficiency MoCo deficiency B	*MOCS2*	AR	603708	Median facial dysplasia Frontal bossing							Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Gephyrin deficiency MoCo deficiency C	*GPHN*	AR	603930	Coarse facies Long face Prominent cheeks Bitemporal narrowing Frontal bossing	Small nose Depressed nasal bridge		Enophthalmos Wide spaced eyes Long palpebral fissuress Deep-set eyes				Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Molybdopterin synthase sulfurase deficiency	*MOCS3*	AR	609277	Median facial dysplasia							S-Sulfocysteine (U), Purines and pyrimidines (U), Uric acid (P, U)
Disorders of copper metabolism											
Copper-transporting ATPase subunit alpha deficiency (Menkes)	*ATP7A*	AR	309400	Peculiar facies						Hernias	Ceruloplasmin (S), Copper (S)
Disorders of zinc metabolism											
SLC30A5 deficiency	*SLC30A5*	AR	607819	Flat facial profile				Small deep-set ears	Dystrophic nails		DNA
Spondylocheirodysplastic Ehlers-Danlos syndrome	*SLC39A13*	AR	612350			Bifid uvula High palate	Down-slanting palpebral fissures Protuberant eyes			Short stature	Lysyl pyridinoline/hydroxylysyl pyridinoline (LP/HP) ratio (U)
Disorders of pantothenate and CoA metabolism											
Phosphopantothenoylcysteine synthetase deficiency	*PPCS*	AR	609853						Abnormally placed thumbs Abnormal dermatoglyphics Hypoplastic toenails	Loose, wrinkled skin	DNA, Lactate (P)
Phosphopantothenoylcysteine decarboxylase deficiency	*PPCDC*	AR	609854	Triangular face Micrognathia							Carnitine and acylcarnitines (P) Organic acids (U Glucose (P)

Name n=374	Gene	Inheritance	OMIM	Head and face	Nose and philtrum	Mouth and tongue	Periorbital region	Ear	Hands and feet	Other	Laboratory tests
Disorders of pyridoxine metabolism											
Pyridoxal 5'-phosphate binding protein deficiency	*PLPBP*	AR	617290	Prominent forehead			Upslanting palpebral fissures		Syndactyly		B6 vitaminers (CSF, P), Biogenic amines (CSF)
Disorders of folate metabolism											
Mitochondrial 10-formyltetrahydrofolate dehydrogenase deficiency	*ALDH1L2*	AR	613584		Broad nasal root Anteverted nares Long philtrum	Thin upper lip	Widely spaced eyes Epicanthal fold				DNA
Disorders of cobalamin metabolism											
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR	277380	Micrognathia Retrognathia		Pegged teeth Bifid incisors Cleft palate	Upslanting palpebral fissures	Cupped ears	Pes equinovarus		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P), Blood count
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR	614857	Mild facial dysmorphism							Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P)
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR	277400	Long face High forehead	Smooth philtrum			Large ears Low set ears			Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)
Epi-cblC	*MMACHC,PRDX1*	AR	609831 176763	Long face High forehead	Smooth philtrum			Large ears Low set ears			Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR	277410	Long face High forehead	Smooth philtrum			Large ears Low set ears			SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*	XL	309541		Long nose	Thick lips		Large ears	Long fingers		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria and homocystinuria due to Ronin deficiency	*THAP11*	AR	609119	Craniofacial dysmorphysm							Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria and homocystinuria due to ZNF143 deficiency	*ZNF143*	AR	603433	Dolichocephaly Microcephaly	Depressed nasal bridge	Thick lips	Prominent eyes		Long digits Slender digits	Supernumerary nipple	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Other disorders of vitamin metabolism											
L-dehydroascorbate transporter deficiency	*SLC2A10*	AR	208050	Facial stigmata						Hernia, diaphragmatic	DNA
γ-Glutamyl carboxylase deficiency	*GGCX*	AR	277450		Nasal hypoplasia						Coagulation factors
Vitamin D 1-α-hydroxylase deficiency	*CYP27B1*	AR	264700	Frontal bossing							Vitamin D metabolites
Plasma retinol-binding protein deficiency	*RBP4*	AD	616428				Microphthalmia				Vitamin A (P)
Vitamin D 25-hydroxylase deficiency	*CYP2R1*	AR	600081	Frontal bossing							Vitamin D metabolites
Vitamin A receptor deficiency	*STRA6*	AR	601186	Micrognathia	Broad nasal bridge		Microphthalmia Blepharophimosis			Hernia, diaphragmatic Short stature	DNA
Vitamin D receptor deficiency	*VDR*	AR	277440	Frontal bossing							Vitamin D metabolites, PTH (S)
Retinaldehyde dehydrogenase 3 deficiency	*ALDH1A3*	AR	615113				Microphthalmia Entrooion				DNA
Retinoic acid receptor β deficiency	*RARB*	AD, AR	615524	Facial dysmorphism			Microphthalmia			Hernia, diaphragmatic	DNA
Retinol dehydrogenase 11 deficiency	*RDH11*	AR	616108	Facial dysmorphism Malar hypoplasia	Prominent alae nasi					Short stature	DNA
METABOLIC CELL SIGNALING (n=13)											
Neurotransmitter disorders											
Glycine receptor subunit beta deficiency	*GLRB*	AR	614619				Esotropia				DNA
Ionotropic glutamate receptor NMDA type subunit 2D superactivity	*GRIN2D*	AD	617162	Full cheeks		Tented upper lip	Deep-set eyes Long eyelashes Thick eyebrows		Flat feet		DNA
Ionotropic glutamate receptor AMPA type subunit 3 deficiency	*GRIA3*	XL	300699				Deeply set eyes			Short stature	DNA
Metabotropic glutamate receptor 1 deficiency	*GRM1*	AR	614831				Esotropia			Short stature	DNA
Intellectual developmental disorder (GRIK2)	*GRIK2*	AR	611092	Facial dysmorphism							DNA
GABA type A receptor subunit alpha 3 deficiency	*GABRA3*	XL	301091	Micrognathia		Cleft palate Small mouth		Large ears		Short stature	DNA
Disorders of the synaptic vesicle cycle											
CEDNIK syndrome	*SNAP29*	AR	609528	Elongated face Micrognathia	Flat, broad nasal root Long pointed nose		Downward-slanting palpebral fissures Widely spaced eyes Deep-set eyes Thick eyebrows	Low set ears			DNA
Synaptotagmin 1 deficiency	*SYT1*	AD	618218	Prominent high forehead	Short nose with prominent nasal tipsmooth philtrum	Thin upper lip	Mild Epicanthal fold Almond-shaped eyes Horizontal low-set eyebrows				DNA
Torsin 1A deficiency	*TOR1A*	AR	618947	Retrognathia Prominent occiput	Short nose Anteverted nares	Wide mouth	Deep-set eyes Upslanting palpebral fissures	Large, low-set and posteriorly rotated ears		Short neck	DNA
Clathrin heavy chain deficiency	*CLTC*	AD	617854		Depressed nasal bridge Anteverted nares Long philtrum	Thin upper lip Open mouth		Low set ears Prominent ears		Inverted nipples	DNA
DLG4 deficiency	*DLG4*	AD	618793	Facial dysmorphism Long face		Arched palate, high			Long and thin fingers		
Disorders of insulin metabolism											
AKT2 deficiency	*AKT2*	AD	164731		Depressed nasal bridge		Widely spaced eyes Proptosis				DNA, Free fatty acids (S), Glucose (P), Insulin (P)
Disorders of steroid metabolism											
Cytochrome P450 oxidoreductase deficincy	*POR*	AR	201750	Midface hypoplasia	Choanal atresia Choanal stenosis			Dysplastic ears			Steroids (P, U)