Supplementary Table 1. 259 genes contained on an NGS panel for NewbornDiscovery

Gene_symbol	Gene_MIM#	Phenotype	Phenotype_MIM#	Inheritance	Category B
AASS	605113	Hyperlysinemia | Saccharopinuria	238700 | 268700	AR | AR	
ABCC2	601107	Dubin-Johnson syndrome	237500	AR	
ABCD1	300371	Adrenoleukodystrophy; Adrenomyeloneuropathy, adult	300100	XLR	
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	614857	AR	Y
ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	611283	N/A	
ACAD9	611103	Mitochondrial complex I deficiency due to ACAD9 deficiency	611126	AR	
ACADL	609576	N/A	N/A	N/A	
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	AR	Y
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of	201470	AR	
ACADSB	600301	2-methylbutyrylglycinuria	610006	AR	
ACADVL	609575	VLCAD deficiency	201475	AR	Y
ACAT1	607809	Alpha-methylacetoacetic aciduria	203750	AR	Y
ACSF3	614245	Combined malonic and methylmalonic aciduria	614265	N/A	Y
ADA	608958	Adenosine deaminase deficiency, partial; Severe combined immunodeficiency due to ADA deficiency	102700	SMo, AR	
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	614300	AR	
AGL	610860	Glycogen storage disease IIIa; Glycogen storage disease IIIb	232400	AR	
AGXT	604285	Hyperoxaluria, primary, type 1	259900	AR	
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752	AR	
ALDH4A1	606811	Hyperprolinemia, type II	239510	AR	
ALDOA	103850	Glycogen storage disease XII	611881	AR	
ALDOB	612724	Fructose intolerance	229600	AR	
ALPL	171760	Hypophosphatasia, adult | Hypophosphatasia, childhood | Hypophosphatasia, infantile | Odontohypophosphatasia	146300 | 241510 | 241500 | 146300	AR, AD | AR | AR | AR, AD	
AMT	238310	Glycine encephalopathy	605899	AR	
APOB	107730	Hypercholesterolemia, due to ligand-defective apo B | Hypobetalipoproteinemia	144010 | 615558	AD | AR	
ARG1	608313	Argininemia	207800	AR	
ARSA	607574	Metachromatic leukodystrophy	250100	AR	
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200	AR	
ASL	608310	Argininosuccinic aciduria	207900	AR	Y
ASS1	603470	Citrullinemia	215700	AR	Y
ATP7A	300011	Menkes disease | Occipital horn syndrome | Spinal muscular atrophy, distal, X-linked 3	309400 | 304150 | 300489	XLR | XLR | XLR	
ATP7B	606882	Wilson disease	277900	AR	
ATP8B1	602397	Cholestasis, benign recurrent intrahepatic | Cholestasis, intrahepatic, of pregnancy, 1 | Cholestasis, progressive familial intrahepatic 1	243300 | 147480 | 211600	AR | AD | AR	
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic | Alpha-thalassemia/mental retardation syndrome | Mental retardation-hypotonic	300448 | 301040 | 309580	N/A | XLD | XLR	

		facies syndrome, X-linked			
AUH	600529	3-methylglutaconic aciduria, type I	250950	AR	
BCKDHA	608348	Maple syrup urine disease, type Ia	248600	AR	Y
BCKDHB	248611	Maple syrup urine disease, type Ib	248600	AR	Y
BCKDK	614901	Branched-chain ketoacid dehydrogenase kinase deficiency	614923	N/A	
BTD	609019	Biotinidase deficiency	253260	AR	
CASR	601199	Hypercalciuric hypercalcemia | Hyperparathyroidism, neonatal | Hypocalcemia, autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome | Hypocalciuric hypercalcemia, type I	N/A | 239200 | 601198 | 145980	N/A | AR | AD | AD	
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types; Thrombosis, hyperhomocysteinemic	236200	AR	
CD320	606475	Methylmalonic aciduria due to transcobalamin receptor defect	613646	N/A	Y
CD3D	186790	Immunodeficiency 19	615617	AR	
CD3E	186830	Immunodeficiency 18; Immunodeficiency 18, SCID variant	615615	AR	
CDH23	605516	Deafness, autosomal recessive 12 | Usher syndrome, type 1D; Usher syndrome, type 1D/F digenic	601386 | 601067	AR | AR, DR	
CFTR	602421	Congenital bilateral absence of vas deferens | Cystic fibrosis | Sweat chloride elevation without CF	277180 | 219700 | N/A	AR | AR | N/A	
COCH	603196	Deafness, autosomal dominant 9	601369	AD	
CPS1	608307	Carbamoylphosphate synthetase I deficiency	237300	AR	
CPT1A	600528	CPT deficiency, hepatic, type IA	255120	AR	Y
CPT2	600650	CPT deficiency, hepatic, type II | CPT II deficiency, lethal neonatal | Myopathy due to CPT II deficiency	600649 | 608836 | 255110	AR | AR | AR	Y
CTNS	606272	Cystinosis, atypical nephropathic | Cystinosis, late-onset juvenile or adolescent nephropathic | Cystinosis, nephropathic | Cystinosis, ocular nonnephropathic	219800 | 219900 | 219800 | 219750	AR | AR | AR | AR	
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency | Aldosteronism, glucocorticoid-remediable	202010 | 103900	AR | AD	
CYP17A1	609300	17,20-lyase deficiency, isolated; 17-alpha-hydroxylase/17,20-lyase deficiency	202110	AR	
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	201910	AR	
DBT	248610	Maple syrup urine disease, type II	248600	AR	Y
DCLRE1C	605988	Omenn syndrome | Severe combined immunodeficiency, Athabascan type	603554 | 602450	AR | AR	
DDC	107930	Aromatic L-amino acid decarboxylase deficiency	608643	AR	
DECR1	222745	N/A	N/A	N/A	
DFNB59	610219	Deafness, autosomal recessive 59	610220	AR	
DLAT	608770	Pyruvate dehydrogenase E2 deficiency	245348	AR	
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	246900	AR	
DNAJC19	608977	3-methylglutaconic aciduria, type V	610198	AR	
DUOX2	606759	Thryoid dyshormonogenesis 6	607200	AR	
DUOXA2	612772	Thyroid dyshormonogenesis 5	274900	AR	
ETFA	608053	Glutaric acidemia IIA	231680	AR	Y
ETFB	130410	Glutaric acidemia IIB	231680	AR	Y
ETFDH	231675	Glutaric acidemia IIC	231680	AR	Y

ETHE1	608451	Ethylmalonic encephalopathy	602473	AR	
FAH	613871	Tyrosinemia, type I	276700	AR	
FOXE1	602617	Bamforth-Lazarus syndrome	241850	AR	
FOXN1	600838	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	601705	AR	
FTCD	606806	Glutamate formiminotransferase deficiency	229100	AR	
G6PC	613742	Glycogen storage disease Ia	232200	AR	
G6PD	305900	Favism | Hemolytic anemia due to G6PD deficiency	134700 | 300908	AD | N/A	
GAA	606800	Glycogen storage disease II	232300	AR	
GALC	606890	Krabbe disease	245200	AR	
GALE	606953	Galactose epimerase deficiency	230350	AR	
GALK1	604313	Galactokinase deficiency with cataracts	230200	AR	
GALNS	612222	Mucopolysaccharidosis IVA	253000	AR	
GALT	606999	Galactosemia	230400	AR	Y
GAMT	601240	Cerebral creatine deficiency syndrome 2	612736	AR	
GATA1	305371	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities | Leukemia, megakaryoblastic, with or without Down syndrome, somatic | Thrombocytopenia with beta-thalassemia, X-linked | Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	300835 | 190685 | 314050 | 300367	XLR | N/A | XLR | XLR	
GATM	602360	Cerebral creatine deficiency syndrome 3	612718	AR	
GBA	606463	Gaucher disease, perinatal lethal | Gaucher disease, type I | Gaucher disease, type II | Gaucher disease, type III | Gaucher disease, type IIIC	608013 | 230800 | 230900 | 231000 | 231005	AR | AR | AR | AR | AR	
GBE1	607839	Glycogen storage disease IV | Polyglucosan body disease, adult form	232500 | 263570	AR | AR	
GCDH	608801	Glutaric aciduria, type I	231670	AR	Y
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia | Hyperphenylalaninemia, BH4-deficient, B	128230 | 233910	AR, AD | AR	
GCSH	238330	Glycine encephalopathy	605899	AR	
GJB2	121011	Bart-Pumphrey syndrome | Deafness, autosomal dominant 3A | Deafness, autosomal recessive 1A | Hystrix-like ichthyosis with deafness | Keratitis-ichthyosis-deafness syndrome | Keratoderma, palmoplantar, with deafness | Vohwinkel syndrome	149200 | 601544 | 220290 | 602540 | 148210 | 148350 | 124500	AD | AD | AR, DD | AD | AD | AD | AD	
GJB3	603324	Deafness, autosomal dominant 2B | Deafness, autosomal dominant, with peripheral neuropathy | Deafness, autosomal recessive | Deafness, digenic, GJB2/GJB3 | Erythrokeratodermia variabilis et progressiva	612644 | N/A | N/A | 220290 | 133200	AD | N/A | N/A | AR, DD | AR, AD	
GJB6	604418	Deafness, autosomal dominant 3B | Deafness, autosomal recessive 1B | Deafness, digenic GJB2/GJB6 | Ectodermal dysplasia 2, Clouston type	612643 | 612645 | 220290 | 129500	AD | AR | AR, DD | AD	
GLA	300644	Fabry disease; Fabry disease, cardiac variant	301500	XL	
GLB1	611458	GM1-gangliosidosis, type I | GM1-gangliosidosis, type II | GM1-gangliosidosis, type III | Mucopolysaccharidosis type IVB (Morquio)	230500 | 230600 | 230650 | 253010	AR | AR | AR | AR	
GLDC	238300	Glycine encephalopathy	605899	AR	
GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism	610199	AR	
GLYCTK	610516	D-glyceric aciduria	220120	AR	
GNA11	139313	Hypocalcemia, autosomal dominant 2 | Hypocalciuric hypercalcemia, type II	615361 | 145981	AD | AD	
GNAS	139320	Acromegaly, somatic | ACTH-independent macronodular adrenal hyperplasia | McCune-Albright syndrome, somatic, mosaic | Osseous heteroplasia, progressive | Pseudohypoparathyroidism Ia | Pseudohypoparathyroidism Ib | Pseudohypoparathyroidism Ic | Pseudopseudohypoparathyroidism	102200 | 219080 | 174800 | 166350 | 103580 | 603233 | 612462 | 612463	N/A | IC | N/A | AD | AD | AD | AD | AD	

GNMT	606628	Glycine N-methyltransferase deficiency	606664	AR	
GNS	607664	Mucopolysaccharidosis type IIID	252940	AR	
GSS	601002	Glutathione synthetase deficiency | Hemolytic anemia due to glutathione synthetase deficiency	266130 | 231900	AR | AR	
GUSB	611499	Mucopolysaccharidosis VII	253220	AR	
GYS1	138570	Glycogen storage disease 0, muscle	611556	AR	
GYS2	138571	Glycogen storage disease 0, liver	240600	AR	
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency | Hyperinsulinemic hypoglycemia, familial, 4	231530 | 609975	AR | AR	
HADHA	600890	Fatty liver, acute, of pregnancy; HELLP syndrome, maternal, of pregnancy; LCHAD deficiency | Trifunctional protein deficiency	609016 | 609015	AR | AR	Y
HADHB	143450	Trifunctional protein deficiency	609015	AR	Y
HAL	609457	Histidinemia	235800	AD	
HBA1	141800	Erythremias, alpha- | Heinz body anemias, alpha- | Hemoglobin H disease, nondeletional | Methemoglobinemias, alpha- | Thalassemias, alpha-	N/A | 140700 | 613978 | N/A | 604131	N/A | AD | N/A | N/A | N/A	
HBA2	141850	Erythrocytosis | Heinz body anemia | Hemoglobin H disease, nondeletional | Hypochromic microcytic anemia | Thalassemia, alpha-	N/A | 140700 | 613978 | N/A | 604131	N/A | AD | N/A | N/A | N/A	
HBB	141900	Delta-beta thalassemia; Hereditary persistence of fetal hemoglobin | Erythremias, beta- | Heinz body anemias, beta- | Methemoglobinemias, beta- | Sickle cell anemia | Thalassemia-beta, dominant inclusion-body | Thalassemias, beta-	141749 | N/A | 140700 | N/A | 603903 | 603902 | 613985	AD | N/A | AD | N/A | AR | N/A | N/A	
HBG1	142200	Fetal hemoglobin quantitative trait locus 1	141749	AD	
HBG2	142250	Cyanosis, transient neonatal | Fetal hemoglobin quantitative trait locus 1	613977 | 141749	AD | AD	
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	309541	XLR	Y
HESX1	601802	Growth hormone deficiency with pituitary anomalies; Pituitary hormone deficiency, combined, 5; Septooptic dysplasia	182230	AR, AD	
HGD	607474	Alkaptonuria	203500	AR	
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C) | Retinitis pigmentosa 73	252930 | 616544	AR | AR	
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency	250620	AR	
HLCS	609018	Holocarboxylase synthetase deficiency	253270	AR	Y
HMBS	609806	Porphyria, acute intermittent; Porphyria, acute intermittent, nonerythroid variant	176000	AD	
HMGCL	613898	HMG-CoA lyase deficiency	246450	AR	Y
HMGCS2	600234	HMG-CoA synthase-2 deficiency	605911	N/A	
HPD	609695	Hawkinsinuria | Tyrosinemia, type III	140350 | 276710	AD | AR	
HPRT1	308000	HPRT-related gout | Lesch-Nyhan syndrome	300323 | 300322	XLR | XLR	
HSD17B10	300256	17-beta-hydroxysteroid dehydrogenase X deficiency | ?Mental retardation, X-linked syndromic 10	300438 | 300220	XLD | XLR	
HSD17B4	601860	D-bifunctional protein deficiency | Perrault syndrome 1	261515 | 233400	AR | AR	
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	201810	AR	
HYAL1	607071	?Mucopolysaccharidosis type IX	601492	AR	
IDS	300823	Mucopolysaccharidosis II	309900	XLR	
IDUA	252800	Mucopolysaccharidosis Ih | Mucopolysaccharidosis Ih/s | Mucopolysaccharidosis Is	607014 | 607015 | 607016	AR | AR | AR	
IL2RA	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity	606367	AR	

IL2RG	308380	Combined immunodeficiency, X-linked, moderate | Severe combined immunodeficiency, X-linked	312863 | 300400	XLR | XLR	
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	608971	AR	
ILDR1	609739	Deafness, autosomal recessive 42	609646	AR	
IVD	607036	Isovaleric acidemia	243500	AR	Y
IYD	612025	Thyroid dyshormonogenesis 4	274800	AR	
JAK3	600173	SCID, autosomal recessive, T-negative/B-positive type	600802	AR	
LCK	153390	?Immunodeficiency 22	615758	AR	
LDHA	150000	Glycogen storage disease XI	612933	AR	
LHX3	600577	Pituitary hormone deficiency, combined, 3	221750	AR	
LHX4	602146	Pituitary hormone deficiency, combined, 4	262700	AD	
LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures	614462	AR	
LIG4	601837	LIG4 syndrome	606593	N/A	
LIPA	613497	Cholesteryl ester storage disease; Wolman disease	278000	AR	
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	277380	AR	Y
MAT1A	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency; Methionine adenosyltransferase deficiency, autosomal recessive	250850	AR, AD	
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200	AR	
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210	AR	
MCEE	608419	Methylmalonyl-CoA epimerase deficiency	251120	AR	Y
MLYCD	606761	Malonyl-CoA decarboxylase deficiency	248360	AR	
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	251100	AR	Y
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	AR	Y
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	277400	AR	Y
MMADHC	611935	Homocystinuria, cblD type, variant 1; Methylmalonic aciduria and homocystinuria, cblD type; Methylmalonic aciduria, cblD type, variant 2	277410	AR	Y
MTHFD1	172460	N/A	N/A	N/A	
MTHFR	607093	Homocystinuria due to MTHFR deficiency	236250	AR	
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	250940	AR	
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	236270	AR	
MUT	609058	Methylmalonic aciduria, mut(0) type	251000	AR	Y
MVK	251170	Hyper-IgD syndrome | Mevalonic aciduria | Porokeratosis 3, multiple types	260920 | 610377 | 175900	AR | AR | AD	
MYH9	160775	Deafness, autosomal dominant 17 | Epstein syndrome | Fechtner syndrome | Macrothrombocytopenia and progressive sensorineural deafness | May-Hegglin anomaly | Sebastian syndrome	603622 | 153650 | 153640 | 600208 | 155100 | 605249	AD | AD | AD | AD | AD | AD	
MYO15A	602666	Deafness, autosomal recessive 3	600316	AR	
MYO7A	276903	Deafness, autosomal dominant 11 | Deafness, autosomal recessive 2 | Usher syndrome, type 1B	601317 | 600060 | 276900	AD | AR | AR	
NADK2	615787	?2,4-dienoyl-CoA reductase deficiency	616034	AR	
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V | Mucopolysaccharidosis type IIIB (Sanfilippo B)	616491 | 252920	AD | AR	

NAGS	608300	N-acetylglutamate synthase deficiency	237310	AR	
NFKBIA	164008	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency	612132	AD	
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	N/A	
NKX2-1	600635	Chorea, hereditary benign | Choreoathetosis, hypothyroidism, and neonatal respiratory distress	118700 | 610978	AD | AD	
NKX2-5	600584	Atrial septal defect 7, with or without AV conduction defects | Conotruncal heart malformations, variable | Hypoplastic left heart syndrome 2 | Hypothyroidism, congenital nongoitrous, 5 | Tetrology of Fallot | Ventricular septal defect 3	108900 | 217095 | 614435 | 225250 | 187500 | 614432	AD | N/A | AD | AD | AD | AD	
NPC1	607623	Niemann-Pick disease, type C1; Niemann-Pick disease, type D	257220	AR	
NPC2	601015	Niemann-pick disease, type C2	607625	AR	
NR0B1	300473	46XY sex reversal 2, dosage-sensitive | Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism	300018 | 300200	XL | XLR	
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia	258870	AR	
OPA3	606580	3-methylglutaconic aciduria, type III | Optic atrophy 3 with cataract	258501 | 165300	AR | AD	
OPLAH	614243	5-oxoprolinase deficiency	260005	AR	
OTC	300461	Ornithine transcarbamylase deficiency	311250	XLR	
OTOF	603681	Auditory neuropathy, autosomal recessive, 1; Deafness, autosomal recessive 9	601071	AR	
OXCT1	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	N/A	
PAH	612349	Phenylketonuria; Hyperphenylalaninemia, non-PKU mild	261600	AR	
PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	AR	
PC	608786	Pyruvate carboxylase deficiency	266150	AR	
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, D	264070	AR	
PCCA	232000	Propionicacidemia	606054	AR	Y
PCCB	232050	Propionicacidemia	606054	AR	Y
PCDH15	605514	Deafness, autosomal recessive 23 | Usher syndrome, type 1D/F digenic | Usher syndrome, type 1F	609533 | 601067 | 602083	AR | AR, DR | AR	
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	312170	XLD	
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	614111	N/A	
PDP1	605993	Pyruvate dehydrogenase phosphatase deficiency	608782	AR	
PEPD	613230	Prolidase deficiency	170100	AR	
PFKM	610681	Glycogen storage disease VII	232800	AR	
PGAM2	612931	Glycogen storage disease X	261670	AR	
PHGDH	606879	Neu-Laxova syndrome 1 | Phosphoglycerate dehydrogenase deficiency	256520 | 601815	AR | AR	
PHKA1	311870	Muscle glycogenosis	300559	XLR	
PHKA2	300798	Glycogen storage disease, type IXa1; Glycogen storage disease, type IXa2	306000	XLR	
PHKB	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive	261750	AR	
PHKG2	172471	Cirrhosis due to liver phosphorylase kinase deficiency | Glycogen storage disease IXc	N/A | 613027	N/A | AR	
PKLR	609712	Adenosine triphosphate, elevated, of erythrocytes | Pyruvate kinase deficiency	102900 | 266200	AD | AR	
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency	613179	AR	

PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	610090	AR	
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis | Disordered steroidogenesis due to cytochrome P450 oxidoreductase	201750 | 613571	AR | N/A	
POU1F1	173110	Pituitary hormone deficiency, combined, 1	613038	AR, AD	
PPM1K	611065	?Maple syrup urine disease, mild variant	615135	N/R	Y
PRODH	606810	Hyperprolinemia, type I	239500	AR	
PROP1	601538	Pituitary hormone deficiency, combined, 2	262600	AR	
PSAP	176801	Combined SAP deficiency | Gaucher disease, atypical | Krabbe disease, atypical | Metachromatic leukodystrophy due to SAP-b deficiency	611721 | 610539 | 611722 | 249900	AR | N/A | AR | AR	
PSAT1	610936	?Phosphoserine aminotransferase deficiency | Neu-Laxova syndrome 2	610992 | 616038	AR | AR	
PSPH	172480	Phosphoserine phosphatase deficiency	614023	AR	
PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	608971	AR	
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	261640	AR	
PYGL	613741	Glycogen storage disease VI	232700	AR	
PYGM	608455	McArdle disease	232600	AR	
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C	261630	AR	
RAG1	179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity | Combined cellular and humoral immune defects with granulomas | Omenn syndrome | Severe combined immunodeficiency, B cell-negative	609889 | 233650 | 603554 | 601457	N/A | AR | AR | AR	
RAG2	179616	Combined cellular and humoral immune defects with granulomas | Omenn syndrome | Severe combined immunodeficiency, B cell-negative	233650 | 603554 | 601457	AR | AR | AR	
SERPINA1	107400	Emphysema due to AAT deficiency; Emphysema-cirrhosis, due to AAT deficiency; Hemorrhagic diathesis due to antithrombin Pittsburgh	613490	AR	
SERPINA7	314200	[Thyroxine-binding globulin QTL]	300932	N/A	
SGSH	605270	Mucopolysaccharidisis type IIIA (Sanfilippo A)	252900	AR	
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	607483	AR	
SLC1A1	133550	Dicarboxylic aminoaciduria	222730	AR	
SLC22A5	603377	Carnitine deficiency, systemic primary	212140	AR	
SLC25A13	603859	Citrullinemia, adult-onset type II | Citrullinemia, type II, neonatal-onset	603471 | 605814	AR | AR	Y
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970	AR	
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	212138	AR	Y
SLC26A4	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct | Pendred syndrome	600791 | 274600	AR | AR	
SLC2A1	138140	Dystonia 9 | GLUT1 deficiency syndrome 1, infantile onset, severe | GLUT1 deficiency syndrome 2, childhood onset | Stomatin-deficient cryohydrocytosis with neurologic defects	601042 | 606777 | 612126 | 608885	AD | AR, AD | AD | AD	
SLC2A2	138160	Fanconi-Bickel syndrome	227810	AR	
SLC37A4	602671	Glycogen storage disease Ib | Glycogen storage disease Ic	232220 | 232240	AR | AR	
SLC39A4	607059	Acrodermatitis enteropathica	201100	AR	
SLC3A1	104614	Cystinuria	220100	AR, AD	
SLC46A1	611672	Folate malabsorption, hereditary	229050	AR	
SLC5A5	601843	Thyroid dyshormonogenesis 1	274400	AR	

SLC6A19	608893	Hartnup disorder | Hyperglycinuria | Iminoglycinuria, digenic	234500 | 138500 | 242600	AR | AD | AR	
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	300352	XLR	
SLC7A7	603593	Lysinuric protein intolerance	222700	AR	
SLC7A9	604144	Cystinuria	220100	AR, AD	
SLCO1B1	604843	Hyperbilirubinemia, Rotor type, digenic	237450	DR	
SLCO1B3	605495	Hyperbilirubinemia, Rotor type, digenic	237450	DR	
SMPD1	607608	Niemann-Pick disease, type A | Niemann-Pick disease, type B	257200 | 607616	AR | AR	
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	612716	?AD, AR	
STAR	600617	Lipoid adrenal hyperplasia	201710	AR	
STK4	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	614868	N/A	
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	AR	Y
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	245400	AR	Y
SUGCT	609187	Glutaric aciduria III	231690	AR	
TAT	613018	Tyrosinemia, type II	276600	AR	
TAZ	300394	Barth syndrome	302060	XLR	
TCN2	613441	Transcobalamin II deficiency	275350	AR	
TG	188450	Thyroid dyshormonogenesis 3	274700	AR	
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6	614450	AD	
THRB	190160	Thyroid hormone resistance | Thyroid hormone resistance, autosomal recessive | Thyroid hormone resistance, selective pituitary	188570 | 274300 | 145650	AD | AR | AD	
TMC1	606706	Deafness, autosomal dominant 36 | Deafness, autosomal recessive 7	606705 | 600974	AD | AR	
TMPRSS3	605511	Deafness, autosomal recessive 8/10	601072	AR	
TPO	606765	Thyroid dyshormonogenesis 2A	274500	AR	
TRHR	188545	Thyrotropin-releasing hormone resistance, generalized	N/A	N/A	
TSHB	188540	Hypothryoidism, congenital, nongoitrous 4	275100	AR	
TSHR	603372	Hyperthyroidism, familial gestational | Hyperthyroidism, nonautoimmune | Hypothyroidism, congenital, nongoitrous, 1 | Thyroid adenoma, hyperfunctioning, somatic | Thyroid carcinoma with thyrotoxicosis	603373 | 609152 | 275200 | N/A | N/A	N/A | IC, AD | AR | N/A | N/A	
UGT1A1	191740	Crigler-Najjar syndrome, type I | Crigler-Najjar syndrome, type II | Hyperbilirubinemia, familial transient neonatal | [Bilirubin, serum level of, QTL1] | [Gilbert syndrome]	218800 | 606785 | 237900 | 601816 | 143500	AR | AR | AR | N/A | AR	
UROS	606938	Porphyria, congenital erythropoietic	263700	AR	
ZAP70	176947	Selective T-cell defect	269840	AR	