Amplexa Genetics®

Condition	Gene	Autosomal recessive	X-linked	G2L	G4L
17-Beta-Hydroxysteroid Dehydrogenase Deficiency, Type III	HSD17B3	•		•	•
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasi	CYP21A2	•		•	•
2-Methylbutyryl-CoA Dehydrogenase Deficiency	ACADSB	•			•
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMGCL	•			•
3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADH	•			•
3-Hydroxyisobutryl-CoA Hydrolase Deficiency	HIBCH	•			•
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1	•			•
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2	•			•
Abetalipoproteinemia	MTTP	•			•
Achalasia-Addisonianism-Alacrimia Syndrome	AAAS	•			•
Achondrogenesis, Type 1B	SLC26A2	•		•	•
Achromatopsia CNGB3-Related	CNGB3	•		•	•
Acrodermatitis Enteropathica	SLC39A4	•			•
Acute Infantile Liver Failure, TRMU-Related	TRMU	•			•
Adenosine Deaminase Deficiency	ADA	•			•
Adrenal Hyperplasia	HSD3B2	•			•
Adrenal Hyperplasia V	CYP17A1	•			•
AdrenoleukoDystrophy	ABCD1		•		•
Aicardi-Goutieres syndrom (AGS)	RNASEH2A	•			•
Aicardi-Goutieres syndrom (AGS)	RNASEH2B	•			•
Aicardi-Goutieres syndrom (AGS)	RNASEH2C	•			•
Aicardi-Goutieres syndrom (AGS)	SAMHD1	•			•
Aicardi-Goutieres Syndrome 1	TREX1	•			•
AICA-Ribosiduria	ATIC	•			•
Albinism, Oculocutaneous, Type I	TYR	•		•	•
Albinism, Oculocutaneous, Type II	OCA2	•			•
Albinism, Oculocutaneous, Type III	TYRP1	•			•
Albinism, Oculocutaneous, Type IV	SLC45A2	•			•
Albinism, Oculocutaneous, Type VII	LRMDA	•			•
Alkaptonuria	HGD	•		•	•
Alpers Syndrome	POLG	•		•	•
Alpha thalassemia	HBA1	•		•	•
Alpha thalassemia	HBA2	•		•	•
Alpha-Mannosidosis	MAN2B1	•			•
Alpha-Methylacetoacetic Aciduria	ACAT1	•			•
Alpha-N-Acetylgalactosaminidase Deficiency, Type 1 (Schindler Disease)	NAGA	•			•
Alport Syndrome, COL4A3-Related	COL4A3	•			•
Alport Syndrome, COL4A4-Related	COL4A4	•			•
Alport Syndrome, COL4A5-Related	COL4A5		•		•
Alstrom Syndrome	ALMS1	•			•
Amish Infantile Epilepsy Syndrome	ST3GAL5	•		•	•
Argininosuccinic Aciduria	ASL	•			•
Aromatic L-amino acid Decarboxylase Deficiency	DDC	•			•
Arthrogryposis, Mental Retardation and Seizures	SLC35A3	•			•
Arthrogryposis, Renal Dysfunction and Cholestasis 1	VPS33B	•			•
Asparagine Synthetase Deficiency	ASNS	•			•
Aspartylglycosaminuria	AGA	•		•	•
Ataxia	TTPA	•			•
Ataxia Telangiectasia	ATM	•		•	•
Bardet-Biedl Syndrome 1	BBS1	•		•	•
Bardet-Biedl Syndrome 10	BBS10	•			•
Bardet-Biedl Syndrome 11	TRIM32	•			•
Bardet-Biedl Syndrome 12	BBS12	•			•
Bardet-Biedl Syndrome 13 / Meckel-Gruber Syndrome 1 / Joubert Syndrome 28	MKS1	•		•	•
Bardet-Biedl Syndrome 16	SDCCAG8	•			•
Bardet-Biedl Syndrome 17	LZTFL1	•			•
Bardet-Biedl Syndrome 18	BBIP1	•			•
Bardet-Biedl Syndrome 19	IFT27	•			•
Bardet-Biedl Syndrome 2	BBS2	•		•	•
Bardet-Biedl Syndrome 20	IFT172	•			•
Bardet-Biedl syndrome 21 / Cone-Rod Dystrophy 16 / Retinitis Pigmentosa 64	C8orf37	•			•
Bardet-Biedl Syndrome 3	ARL6	•			•
Bardet-Biedl Syndrome 4	BBS4	•			•
Bardet-Biedl Syndrome 5	BBS5	•			•
Bardet-Biedl Syndrome 6	MKKS	•			•
Bardet-Biedl Syndrome 7	BBS7	•			•
Bardet-Biedl Syndrome 8	TTC8	•			•
Bardet-Biedl Syndrome 9	BBS9	•			•
Bartter Syndrome	BSND	•			•
Bernard-Soulier Syndrome, Type A1	GP1BA	•			•
Bernard-Soulier Syndrome, Type C	GP9	•			•
Beta-Thalassemia	HBB	•		•	•
Bile Acid Synthesis Defect, Type 4	AMACR	•			•
Biotinidase Deficiency	BTD	•		•	•
Blomstrand Chondrodysplasia	PTH1R	•			•
Bloom Syndrome	BLM	•		•	•
Brittle Cornea Syndrome 1	ZNF469	•			•

Canavan Disease	ASPA	•		•	•
Carbamoyl-Phosphate Synthetase 1 Deficiency	CPS1	•			•
Carnitine Deficiency	SLC22A5	•			•
Carnitine Palmitoyltransferase Deficiency, Type 1A	CPT1A	•			•
Carnitine-acylcarnitine Translocase Deficiency	SLC25A20	•			•
Carpenter Syndrome	RAB23	•			•
Cartilage-Hair Hypoplasia	RMRP	•		•	•
Cerebellar Hypoplasia and Mental Retardation	VLDLR		•		•
Cerebellar Hypoplasia, Type 1	EXOSC3	•			•
Cerebral Creatine Deficiency Syndrome 2	GAMT	•			•
Cerebral Dysgenesis, Neuropathy, Ichthyosis and Palmoplantar Keratoderma Syndrome	SNAP29	•			•
Cerebrotendinous Xanthomatosis	CYP27A1	•			•
Ceroid Lipofuscinosis	MFSD8	•			•
Ceroid Lipofuscinosis	CLN8	•			•
Charcot-Marie-Tooth Disease, Type 2B1	LMNA	•			•
Charcot-Marie-Tooth Disease, Type 2EE	MPV17	•			•
Charcot-Marie-Tooth Disease, Type 2S	IGHMBP2	•			•
Charcot-Marie-Tooth Disease, Type 4F	PRX	•			•
Charcot-Marie-Tooth Disease, Type 4H	FGD4	•			•
Choreoacanthocytosis	VPS13A	•			•
Choroideremia	CHM		•		•
Chronic Granulomatous Disease 1	NCF1	•			•
Chronic Granulomatous Disease 4	CYBA	•		•	•
Ciliary Dyskinesia, Type 1	DNAI1	•			•
Ciliary Dyskinesia, Type 9	DNAI2	•			•
Citrin Deficiency	SLC25A13	•		•	•
Citrullinemia, Type 1	ASS1	•			•
Classical homocystinuria	CBS	•		•	•
Coenzyme Q10 Deficiency, Type 2	PDSS1	•			•
Coenzyme Q10 Deficiency, Type 4	COQ8A	•			•
Cohen Syndrome	VPS13B	•			•
Cold-induced Sweating Syndrome 1	CRLF1	•			•
Colobomatous Microphthalmia	STRA6	•			•
Combined Malonic and Methylmalonic Aciduria	ACSF3	•			•
Combined Oxidative Phosphorylation Deficiency 1	GFM1	•			•
Combined Oxidative Phosphorylation Deficiency 3	TSFM	•			•
Cone-Rod Dystrophy and Hearing Loss 2	CEP250	•			•
Congenital Adrenal Hyperplasia, 11-b hydroxylase	CYP11B1	•		•	•
Congenital Amegakaryocytic Thrombocytopenia	MPL	•			•
Congenital Disorder of Glycosylation, Type 1a	PMM2	•		•	•
Congenital Disorder of Glycosylation, Type Ib	MPI	•			•
Congenital Disorder of Glycosylation, Type Ic	ALG6	•			•
Congenital Disorder of Glycosylation, Type Ie	DPM1	•			•
Congenital Disorder of Glycosylation, Type II	B4GALT1	•			•
Congenital Disorder of Glycosylation, Type IIa	MGAT2	•			•
Congenital Disorder of Glycosylation, Type IIb	MOGS	•			•
Congenital Disorder of Glycosylation, Type IIc	SLC35C1	•			•
Congenital Disorder of Glycosylation, Type IIf	SLC35A1	•			•
Congenital Disorder of Glycosylation, Type Ik	ALG1	•			•
Congenital Erythropoietic Porphyria	UROS	•			•
Congenital Hypothyroidism	PAX8	•			•
Congenital Hypothyroidism	TSHR	•			•
Congenital Insensitivity to pain with Anhidrosis	NTRK1	•			•
Congenital Myasthenic Syndrome 4B	CHRNE	•			•
Congenital Non-Bullous Ichthyosiform Erythroderma	ABCA12	•			•
Congenital Thrombotic Thrombocytopenic Purpura	ADAMTS13	•			•
Corneal Endothelial Dystrophy	SLC4A11	•			•
Corpus Callosum Agenesis-Neuronopathy Syndrome	SLC12A6	•			•
Costeff Syndrome	OPA3	•			•
CPT II Deficiency, Infantile	CPT2	•			•
Cutis Laxa Classic, Type 2	ATP6V0A2	•			•
Cutis Laxa, Type IA	FBLN5	•			•
Cutis Laxa, Type IB	EFEMP2	•			•
Cystic Fibrosis	CFTR	•		•	•
Cystinosis	CTNS	•		•	•
Cystinuria	SLC3A1	•			•
Deafness 53	COL11A2	•			•
Deafness 77	LOXHD1	•			•
Dejerine-Sottas Disease	PMP22	•			•
Desmosterolosis	DHCR24	•			•
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	•		•	•
Distal Renal Tubular Acidosis	ATP6V1B1	•			•
Donnai-Barrow Syndrome	LRP2	•			•
Dopa-Responsive Dystonia	TH	•			•
Duchenne Muscular Dystrophy	DMD		•		•
Dysplasminogenemia	PLG	•			•
Ehlers-Danlos Syndrome, Cardiac Valvular Type	COL1A2	•			•
Ehlers-Danlos Syndrome, Dermatosparaxis Type	ADAMTS2	•			•
Ellis-van Creveld Syndrome	EVC2	•		•	•
Ellis-van Creveld Syndrome	EVC	•			•

Emery-Dreifuss Muscular Dystrophy	FHL1		●		●
Emphysema	SERPINA1	●			●
Epidermolysis Bullosa, Type 1	LAMB3	●			●
Epidermolysis Bullosa, Type 2	LAMA3	●			●
Epidermolysis Bullosa, Type 3	LAMC2	●			●
Epidermolysis Bullosa, Type 4	COL17A1	●			●
Epidermolysis Bullosa, Type 5	ITGB4	●			●
Epidermolysis Bullosa, Type 6	ITGA6	●			●
Ethylmalonic Encephalopathy	ETHE1	●			●
Fabry Disease	GLA		●		●
Factor IX Deficiency	F9		●		●
Factor VIII Deficiency	F8		●		●
Factor XI Deficiency	F11	●		●	●
Familial Chloride Diarrhea	SLC26A3	●		●	●
Familial Dysautonomia	ELP1	●		●	●
Familial Hyperinsulinemic Hypoglycemia	ABCC8	●		●	●
Familial Hyperinsulinism	KCNJ11	●		●	●
Familial Mediterranean Fever	MEFV	●		●	●
Fanconi Anemia, Group C	FANCC	●		●	●
Fanconi Anemia, Group G	FANCG	●		●	●
Fanconi Anemia, Troup A	FANCA	●		●	●
Fetal Akinesia Deformation Sequence	RAPSN	●			●
Fragile X Syndrome	FMR1		●		●
French Canadian Type of Leigh Syndrome	LRPPRC	●			●
Fucosidosis	FUCA1	●			●
Fuhrmann Syndrome	WNT7A	●			●
Fumarase Deficiency	FH	●			●
Galactokinase Deficiency, Type II	GALK1	●		●	●
Galactosemia	GALT	●		●	●
Gaucher Disease	GBA	●		●	●
Gitelman Syndrome	SLC12A3	●		●	●
Glutaric Acidemia IIA	ETFA	●			●
Glutaric Acidemia IIC	ETFDH	●			●
Glutaric Acidemia, Type 1	GCDH	●		●	●
Glycine Encephalopathy	GLDC	●			●
Glycogen Storage Disease VII	PFKM	●			●
Glycogen Storage Disease, Type 1A	G6PC1	●		●	●
Glycogen Storage Disease, Type 2 (Pompe Disease)	GAA	●		●	●
Glycogen Storage Disease, Type 3	AGL	●		●	●
Glycogen Storage Disease, Type 4	GBE1	●		●	●
Glycogen Storage Disease, Type B	SLC37A4	●			●
GRACILE Syndrome	BCS1L	●			●
Greenberg Skeletal Dysplasia	LBR	●			●
Griscelli Syndrome, Type 1	MYO5A	●			●
Griscelli Syndrome, Type 2	RAB27A	●			●
Gyrate Atrophy of Choroid and Retina	OAT	●			●
Hemochromatosis, Type 2	HJV	●			●
Hemochromatosis, Type 3	TFR2	●			●
Hepatic Venoocclusive Disease with Immunodeficiency	SP110	●			●
Hereditary Fructose Intolerance	ALDOB	●		●	●
Hermansky-Pudlak Syndrome 1	HPS1	●		●	●
Holocarboxylase Synthetase Deficiency	HLCS	●		●	●
Homocystinuria	MTHFR	●			●
Homocystinuria-Megaloblastic Anemia	MTRR	●			●
Hunter Syndrome	IDS		●		●
Hurler syndrome	IDUA	●			●
Hyaline Fibromatosis Syndrome	ANTXR2	●			●
Hydrolethalus Syndrome	HYLS1	●		●	●
Hyperoxaluria, Type I	AGXT	●			●
Hyperoxaluria, Type II	GRHPR	●			●
Hyperoxaluria, Type III	HOGA1	●			●
Hyperphenylalaninemia	PTS	●			●
Hypoaldosteronism, CMO I Deficiency	CYP11B2	●			●
Hypohidrotic Ectodermal Dysplasia	EDA		●		●
Hypomyelinating leukodystrophy 12	VPS11	●			●
Hypophosphatasia	ALPL	●		●	●
Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 1	DNMT3B	●			●
Infantile Striatonigral Degeneration	NUP62	●			●
Insulin-Like Growth Factor I Deficiency	IGF1	●			●
Isovaleric Acidemia	IVD	●			●
Jervell and Lange-Nielsen Syndrome	KCNQ1	●			●
Joubert Syndrome 1	INPP5E	●			●
Joubert Syndrome 12	KIF7	●			●
Joubert Syndrome 13	TCTN1	●			●
Joubert Syndrome 14	TMEM237	●			●
Joubert Syndrome 15	CEP41	●			●
Joubert Syndrome 16	TMEM138	●			●
Joubert Syndrome 17	CPLANE1	●			●
Joubert Syndrome 18	TCTN3	●			●
Joubert Syndrome 2	TMEM216	●		●	●

Joubert Syndrome 20	TMEM231	•			•
Joubert Syndrome 21	CSPP1	•			•
Joubert Syndrome 22	PDE6D	•			•
Joubert Syndrome 23	KIAA0586	•			•
Joubert Syndrome 24	TCTN2	•			•
Joubert Syndrome 27	B9D1	•			•
Joubert Syndrome 3	AHI1	•			•
Joubert Syndrome 4	NPHP1	•			•
Joubert Syndrome 5	CEP290	•			•
Joubert Syndrome 6	TMEM67	•			•
Joubert Syndrome 7	RPGRIP1L	•			•
Joubert Syndrome 8	ARL13B	•			•
Joubert Syndrome 9	CC2D2A	•			•
Juvenile Amyotrophic Lateral Sclerosis 2	ALS2	•			•
Juvenile Hemochromatosis, Type 2	HAMP	•			•
Juvenile Neuronal Ceroid Lipofuscinose	CLN3	•			•
Juvenile Paget Disease	TNFRSF11B	•			•
Juvenile Retinoschisis	RS1		•		•
Kenny-Caffey Syndrome, Type 1	TBCE	•			•
Krabbe Disease	GALC	•			•
Lamellar Ichthyosis	TGM1	•			•
Late Infantile Neuronal Ceroid Lipofuscinoses	CLN6	•			•
Lathosterolosis	SC5D	•			•
Leber Congenital Amaurosis 16	KCNJ13	•			•
Leber Congenital Amaurosis 2	RPE65	•			•
Leprechaunism	INSR	•			•
Lethal Congenital Contracture Syndrome 1	GLE1	•		•	•
Lethal Congenital Contracture Syndrome 2	ERBB3	•			•
Lethal Osteosclerotic Bone Dysplasia	FAM20C	•			•
Leukoencephalopathy with Vanishing White Matter	EIF2B5	•			•
Limb-Girdle Muscular Dystrophy, Type 1	CAPN3	•			•
Limb-Girdle Muscular Dystrophy, Type 17	PLEC	•			•
Limb-Girdle Muscular Dystrophy, Type 2	DYSF	•			•
Limb-Girdle Muscular Dystrophy, Type 23	LAMA2	•			•
Limb-Girdle Muscular Dystrophy, Type 3	SGCA	•			•
Limb-Girdle Muscular Dystrophy, Type 4	SGCB	•			•
Limb-Girdle Muscular Dystrophy, Type 5	SGCG	•			•
Limb-Girdle Muscular Dystrophy, Type 5C	FKRP	•			•
Limb-Girdle Muscular Dystrophy, Type C1	POMT1	•			•
Limb-Girdle Muscular Dystrophy, Type C3	POMGNT1	•		•	•
Lipoid Adrenal Hyperplasia	STAR	•			•
Lipoprotein Lipase Deficiency	LPL	•			•
Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	•			•
Lysinuric Protein Intolerance	SLC7A7	•			•
Mandibuloacral Dysplasia	ZMPSTE24	•			•
Maple syrup urine Disease, Type 1A	BCKDHA	•		•	•
Maple syrup urine Disease, Type 1B	BCKDHB	•		•	•
Maple syrup urine Disease, Type II	DBT	•			•
Maple Syrup Urine Disease, Type III	DLD	•		•	•
Marinesco-Sjögren Syndrome	SIL1	•			•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	•		•	•
Megalencephalic Leukoencephalopathy 1	MLC1	•			•
Metachromatic Leukodystrophy	ARSA	•		•	•
Methylmalonic Aciduria, MMAB-Related	MMAB	•			•
Methylmalonic Aciduria, MMACHC-Related	MMACHC	•			•
Methylmalonic Aciduria, MMUT-Related	MMUT	•			•
Methylmalonic Aciduria, MMAA-Related	MMAA	•			•
Mitochondrial Complex I Deficiency, ACAD9-Related	ACAD9	•			•
Mitochondrial DNA depletion Syndrome 1, TYMP-Related	TYMP	•			•
Mitochondrial Trifunctional Protein Deficiency 2	HADHB	•			•
Molybdenum Cofactor Deficiency, Type A	MOCS1	•			•
Mucolipidosis II/IIIA	GNPTAB	•		•	•
Mucolipidosis III gamma	GNPTG	•			•
Mucolipidosis, Type IV	MCOLN1	•		•	•
Mucopolysaccharidosis, Type IVB	GLB1	•		•	•
Mucopolysaccharidosis, Type IX	HYAL1	•			•
Mucopolysaccharidosis, Type VI	ARSB	•			•
Mucopolysaccharidosis, Type VII	GUSB	•			•
Mulibrey Nanism	TRIM37	•		•	•
Multiple Pterygium Syndrome	CHRNA1	•			•
Multiple Sulfatase Deficiency	SUMF1	•			•
Myasthenic Syndrome 13	DPAGT1	•			•
Myasthenic Syndrome 22	PREPL	•			•
Myoclonic Epilepsy of Lafora, Type 2A	EPM2A	•			•
Myoclonic Epilepsy of Lafora, Type 2B	NHLRC1	•			•
Myoclonic Epilepsy of Unverricht and Lundborg, Type 1A	CSTB	•			•
Myophosphorylase Deficiency	PYGM	•			•
N-Acetylglutamate Synthase Deficiency	NAGS	•			•
Nemaline Myopathy 2	NEB	•			•
Nemaline Myopathy 5	TNNT1	•			•

Neonatal Glycine Encephalopathy	AMT	•			•
Neonatal Glycine Encephalopathy	GCSH	•			•
Neonatal Ichthyosis-Sclerosing Cholangitis	CLDN1	•			•
Nephronophthisis 3	NPHP3	•			•
Nephrotic Syndrome, Type 1	NPHS1	•		•	•
Nephrotic Syndrome, Type 2	NPHS2	•			•
Neuronal ceroid lipofuscinosis, PPT1-Related	PPT1	•		•	•
Neuronal ceroid lipofuscinosis, TPP1-Related	TPP1	•		•	•
Neuronal ceroid-lipofuscinosis, CLN5-Related	CLN5	•		•	•
Neutropenia, Severe congenital 3	HAX1	•			•
Neutropenia, Severe congenital 4	G6PC3	•			•
Niemann-Pick Disease	SMPD1	•		•	•
Niemann-Pick Disease, Type C1	NPC1	•			•
Niemann-pick Disease, Type C2	NPC2	•			•
Nijmegen Breakage Syndrome	NBN	•			•
Nonaka Myopathy	GNE	•			•
Non-Syndromic Hearing Loss	TMC1	•			•
Non-Syndromic Hearing Loss, GJB2-Related	GJB2	•		•	•
Odontoonychodermal Dysplasia	WNT10A	•			•
Omenn syndrome	RAG2	•			•
Omenn Syndrome	RAG1	•			•
Omenn Syndrome	DCLRE1C	•			•
Osteopetrosis	CA2	•			•
Osteopetrosis, Infantile Malignant	TCIRG1	•		•	•
Pendred Syndrome	SLC26A4	•		•	•
Peroxisomal Acyl-CoA Oxidase Deficiency	ACOX1	•			•
Peroxisome Biogenesis Disorder	PEX7	•			•
Perrault Syndrome 1	HSD17B4	•			•
Perrault Syndrome 5	TWNK	•			•
Phenylketonuria	PAH	•		•	•
Pierson Syndrome	LAMB2	•			•
Pituitary Hormone Deficiency 2	PROP1	•		•	•
Polycystic Kidney Disease 4	PKHD1	•			•
Polyglandular aAutoimmune Syndrome, Type 1	AIRE	•		•	•
Polymicrogyria	ADGRG1	•			•
Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa and Cataract, PHARC Disorder	ABHD12	•			•
Pontocerebellar Hypoplasia, Type 1	RARS2	•			•
Pontocerebellar Hypoplasia, Type 2	TSEN54	•			•
Postnatal Progressive Microcephaly	MED17	•			•
Primary Ciliary Dyskinesia, DNAH5-Related	DNAH5	•			•
Primary congenital glaucoma	CYP1B1	•		•	•
Progressive Cerebellocerebral Atrophy, Type 1	SEPSECS	•			•
Progressive Cerebellocerebral Atrophy, Type 2	VPS53	•			•
Progressive Familial Intrahepatic Cholestasis, Type 1	ATP8B1	•			•
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	•			•
Propionicacidemia	PCCA	•			•
Propionicacidemia	PCCB	•			•
Pseudohypoaldosteronism, Type IB	SCNN1B	•			•
Pseudohypoaldosteronism, Type IB1	SCNN1A	•			•
Pseudohypoaldosteronism, Type IB3	SCNN1G	•			•
Pycnodysostosis	CTSK	•			•
Pyridoxamine 5'-Phosphate Oxidase Deficiency	PNPO	•			•
Pyruvate Kinase Deficiency	PKLR	•			•
Renal Hypomagnesemia 5	CLDN19	•			•
Retinitis Pigmentosa 12	CRB1	•			•
Retinitis Pigmentosa 25	EYS	•			•
Retinitis Pigmentosa 26	CERKL	•			•
Retinitis Pigmentosa 28	FAM161A	•		•	•
Retinitis Pigmentosa 59	DHDDS	•			•
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	•			•
Roberts Syndrome	ESCO2	•			•
Salla Disease	SLC17A5	•		•	•
Sandhoff Disease	HEXB	•		•	•
Sanfilippo Syndrome A	SGSH	•			•
Sanfilippo Syndrome B	NAGLU	•			•
Sanfilippo Syndrome C	HGSNAT	•			•
Schimke Immunoosseous Dysplasia	SMARCAL1	•			•
Schneckenbecken Dysplasia	SLC35D1	•			•
Schwartz-Jampel Syndrome, Type 1	HSPG2	•			•
Seckel Syndrome	ATR	•			•
Senior-Loken Syndrome 4	NPHP4	•			•
Senior-Loken Syndrome 5	IQCB1	•			•
Short-Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	•			•
Short-Rib Thoracic Dysplasia	TTC21B	•			•
Shwachman-Diamond Syndrome	SBDS	•			•
Sialidosis, Type I	NEU1	•			•
Sjogren-Larsson Syndrome	ALDH3A2	•			•
Skin Fragility-Woolly Hair Syndrome	DSP	•			•
Smith-Lemli-Opitz Syndrome	DHCR7	•		•	•
Spastic Ataxia of Charlevoix-Saguenay	SACS	•			•

Spinal Muscular Atrophy	SMN1	•		•	•
Spinal Muscular Atrophy Type 2	SMN2	•			•
Stargardt Disease 1	ABCA4	•			•
Steel Syndrome	COL27A1	•			•
Stuve-Wiedemann Syndrome / Schwartz-Jampel Syndrome, Type 2	LIFR	•			•
Succinate-CoA Ligase Deficiency	SUCLA2	•			•
Sudden infant death with dysgenesis of the testes Syndrome	TSPYL1	•			•
Sulfite Oxidase Deficiency	SUOX	•			•
Tay-Sachs Disease	HEXA	•		•	•
T-cell immunodeficiency, Congenital Alopecia and Nail Dystrophy	FOXN1	•			•
Tetra-Amelia Syndrome 1	WNT3	•			•
Tyrosinemia, Type I	FAH	•		•	•
Usher Syndrome, Type 1	USH1G	•			•
Usher Syndrome, Type 1B	MYO7A	•		•	•
Usher Syndrome, Type 1C	USH1C	•		•	•
Usher Syndrome, Type 1D	CDH23	•		•	•
Usher Syndrome, Type 1F	PCDH15	•		•	•
Usher Syndrome, Type 2A	USH2A	•			•
Usher Syndrome, Type 2C	ADGRV1	•			•
Usher Syndrome, Type 2D	WHRN	•			•
Usher Syndrome, Type 3	CLRN1	•		•	•
Usher Syndrome, Type IJ	CIB2	•			•
Very-long Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	•		•	•
Vitamin D-dependent Rickets, Type I	CYP27B1	•			•
Walker-Warburg Syndrome	FKTN	•		•	•
Warsaw Breakage Syndrome	DDX11	•			•
Wilson Disease	ATP7B	•		•	•
Wolcott-Rallison Syndrome	EIF2AK3	•			•
Wolman Disease	LIPA	•		•	•
Zellweger spectrum Disorders 1A	PEX1	•			•
Zellweger spectrum Disorders 4A	PEX6	•		•	•
Zellweger spectrum Disorders 5A	PEX2	•			•
Zellweger spectrum Disorders 6A	PEX10	•			•