Лабораторія "Ультрагеном" Тестування CarrierSeq 420 генів

Condition	Gene	CNV Target
Achalasia-Addisonianism-Alacrima Syndrome	AAAS	
Harlequin ichthyosis	ABCA12	
Stargardt Disease, Type 1	ABCA4	
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	
Progressive Familial Intrahepatic Cholestasis, Type 3	ABCB4	
Pseudoxanthoma elasticum	ABCC6	
Familial Hyperinsulinism, ABCC8-Related	ABCC8	
Adrenoleukodystrophy, X-Linked	ABCD1	
Mitochondrial Complex I Deficiency, ACAD9-Related	ACAD9	
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	
Short Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	
Short/branched chain acyl-CoA dehydrogenase	ACADSB	
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	
Beta-Ketothiolase Deficiency	ACAT1	
Acyl-CoA Oxidase I Deficiency	ACOX1	
Combined Malonic and Methylmalonic Aciduria	ACSF3	
Severe Combined Immunodeficiency, ADA-Related	ADA	
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	
Bilateral Frontoparietal Polymicrogyria	ADGRG1	
Aspartylglucosaminuria	AGA	
Glycogen Storage Disease, Type III (Cori/Forbes)	AGL	
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	
Hyperoxaluria, Primary, Type 1	AGXT	
Autoimmune polyendocrinopathy syndrome, type I	AIRE	
Sjogren-Larsson Syndrome	ALDH3A2	
Pyridoxine-dependent epilepsy	ALDH7A1	
Hereditary Fructose Intolerance	ALDOB	
Congenital Disorder of Glycosylation, Type 1C	ALG6	
Alstrom Syndrome	ALMS1	
Hypophosphatasia, ALPL-Related	ALPL	
Persistent Müllerian duct syndrome type 1	AMH	
Persistent Müllerian duct syndrome type 2	AMHR2	
Glycine Encephalopathy, AMT-Related	AMT	
Mental retardation, enteropathy, deafness, peripheral neuropathy, ich	AP1S1	
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	AQP2	
Androgen insensitivity syndrome, X-Linked	AR	
Argininemia	ARG1	
Metachromatic Leukodystrophy, ARSA-Related	ARSA	
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	ARSB	
Argininosuccinate Lyase Deficiency	ASL	
Asparagine Synthetase Deficiency	ASNS	
Canavan Disease	ASPA	
Citrullinemia, Type 1	ASS1	
Ataxia-Telangiectasia	ATM	CNV

Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	ATP6V1B1	
Menkes Syndrome, X-Linked	ATP7A	
Wilson Disease	ATP7B	
Progressive Familial Intrahepatic Cholestasis, Type 1	ATP8B1	
Alpha-Thalassemia Intellectual Disability Syndrome, X-Linked	ATRX	
Bardet-Biedl Syndrome 1	BBS1	
Bardet-Biedl Syndrome 10	BBS10	
Bardet-Biedl Syndrome 12	BBS12	
Bardet-Biedl Syndrome 2	BBS2	
Bardet-Biedl Syndrome 4	BBS4	CNV
Bardet-Biedl Syndrome 9	BBS9	
Pseudocholinesterase Deficiency	BCHE	
Maple Syrup Urine Disease, Type 1A	BCKDHA	
Maple Syrup Urine Disease, Type 1B	BCKDHB	
GRACILE Syndrome	BCS1L	
Bloom Syndrome	BLM	
Fanconi anemia, Group J	BRIP1	
Bartter syndrome, Type 4a	BSND	
Biotinidase Deficiency	BTD	
Isolated growth hormone deficiency, Type III, X-linked	BTK	
Desbuquois dysplasia 1	CANT1	
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	
Catecholaminergic polymorphic ventricular tachycardia	CASQ2	
Homocystinuria, CBS-Related	CBS	
Mental retardation, autosomal recessive 3	CC2D1A	CNV
Usher Syndrome, Type 1D	CDH23	
Leber Congenital Amaurosis, Type CEP290	CEP290	
Retinitis Pigmentosa 26	CERKL	
Cystic Fibrosis	CFTR	CNV
Choroideremia, X-Linked	CHM	
Congenital Myasthenic Syndrome, CHRNE-Related	CHRNE	
Escobar Syndrome	CHRNG	
Bare Lymphocyte Syndrome, CIITA-Related	CIITA	
Ceroid Lipofuscinosis, Neuronal, 3	CLN3	CNV
Ceroid Lipofuscinosis, Neuronal, 5	CLN5	
Ceroid Lipofuscinosis, Neuronal, 6	CLN6	
Ceroid Lipofuscinosis, Neuronal, 8 (a.ka. Northern Epilepsy)	CLN8	
Usher Syndrome, Type 3	CLRN1	
Achromatopsia, CNGA3-Related	CNGA3	
Achromatopsia, CNGB3-Related	CNGB3	
Fibrochondrogenesis type 2	COL11A2	
Alport Syndrome, COL4A3-Related	COL4A3	
Alport Syndrome, COL4A4-Related	COL4A4	
Alport Syndrome, X-Linked	COL4A5	
Dystrophic Epidermolysis Bullosa, COL7A1-Related	COL7A1	
Carbamoyl Phosphate Synthetase I Deficiency	CPS1	
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	
Carnitine Palmitoyltransferase II Deficiency	CPT2	
Leber congenital amaurosis 8	CRB1	
Cystinosis	CTNS	CNV
Papillon-Lefevre Syndrome	CTSC	
Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)	CTSD	
Pycnodysostosis	CTSK	
Chronic Granulomatous Disease, CYBA-Related	CYBA	

Chronic Granulomatous Disease, X-Linked	CYBB	
Congenital Adrenal Hyperplasia, 11-beta-hydroxylase-deficient	CYP11B1	
Corticosterone Methyloxidase Deficiency	CYP11B2	
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	CYP17A1	
Aromatase Deficiency	CYP19A1	
Primary Congenital Glaucoma	CYP1B1	
Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient	CYP21A2	SC
Cerebrotendinous Xanthomatosis	CYP27A1	
Vitamin D-dependent rickets type 1A	CYP27B1	
Maple Syrup Urine Disease, Type 2	DBT	
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	
Xeroderma Pigmentosum Group E	DDB2	
Smith-Lemli-Opitz Syndrome	DHCR7	
Retinitis Pigmentosa 59	DHDDS	
Dyskeratosis congenita, X-Linked	DKC1	
Dihydrolipoamide Dehydrogenase Deficiency	DLD	
Duchenne/Becker Muscular Dystrophy	DMD	CNV
Ciliary Dyskinesia, Primary 3	DNAH5	
Ciliary Dyskinesia, Primary 1	DNAI1	
Ciliary Dyskinesia, Primary 9	DNAI2	
Ciliary Dyskinesia, Primary, 16	DNAL1	
Congenital Myasthenic Syndrome, DOK7-Related	DOK7	
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF	
Hypohidrotic Ectodermal Dysplasia, X-Linked	EDA	
Hypohidrotic Ectodermal Dysplasia	EDAR	
Wolcott-Rallison Syndrome	EIF2AK3	
Leukoencephalopathy with Vanishing White Matter	EIF2B5	
Dysautonomia, familial (IKBKAP or ELP1)	IKBKAP	
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	EMD	
Xeroderma Pigmentosum Group D	ERCC2	
Xeroderma Pigmentosum Group B	ERCC3	
Xeroderma Pigmentosum Group F	ERCC4	
Xeroderma pigmentosum Group G	ERCC5	
Cockayne syndrome, type B	ERCC6	
Cockayne syndrome, type A	ERCC8	
Roberts Syndrome	ESCO2	
Glutaric Acidemia, Type 2A	ETFA	
Glutaric Acidemia, Type 2B	ETFB	
Glutaric Acidemia, Type 2C	ETFDH	
Ethylmalonic Encephalopathy	ETHE1	
Ellis-van Creveld Syndrome, EVC-Related	EVC	
Ellis-van Creveld Syndrome, EVC2-related	EVC2	
Pontocerebellar Hypoplasia, Type 1B	EXOSC3	
Retinitis Pigmentosa 25	EYS	
Factor XI deficiency	F11	
Prothrombin deficiency	F2	
Hemophilia A	F8	
Hemophilia B	F9	
Tyrosinemia, Type I	FAH	
Retinitis Pigmentosa 28	FAM161A	
Fanconi Anemia, Group A	FANCA	CNV
Fanconi Anemia, Group C	FANCC	CNV
Fanconi Anemia, Group G	FANCG	

Fumarase Deficiency	FH	
Limb-Girdle Muscular Dystrophy, Type 2I	FKRP	
Walker-Warburg Syndrome, FKTN-Related	FKTN	
Glycogen Storage Disease, Type IA	G6PC	
Glucose-6-Phosphate Dehydrogenase Deficiency*	G6PD	
Glycogen Storage Disease, Type II (Pompe Disease)	GAA	CNV
Krabbe Disease	GALC	CNV
Galactose epimerase deficiency	GALE	
Galactokinase Deficiency (Galactosemia, Type II)	GALK1	
Mucopolysaccharidosis, Type IVA	GALNS	
Hyperphosphatemic familial tumoral calcinosis	GALNT3	
Galactosemia	GALT	CNV
Guanidinoacetate Methyltransferase Deficiency	GAMT	
Gaucher Disease	GBA	
Glycogen Storage Disease, Type IV	GBE1	
Glutaric Acidemia, Type 1	GCDH	
Dopa-responsive dystonia	GCH1	
Grebe syndrome	GDF5	
Combined Oxidative Phosphorylation Deficiency 1	GFM1	
Isolated growth hormone deficiency, Type IA/II	GH1	CNV
Isolated growth hormone deficiency, Type IB	GHRHR	
Charcot-Marie-Tooth Disease with Deafness, X-Linked	GJB1	
Non-Syndromic Hearing Loss (a.k.a. Connexin 26)	GJB2	CNV
Erythrokeratodermia variabilis et progressiva	GJB3	
Non-Syndromic Hearing Loss (a.k.a. Connexin 30)	GJB6	CNV
Fabry Disease	GLA	CNV
Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	GLB1	
Glycine Encephalopathy, GLDC-Related	GLDC	
Lethal Congenital Contracture Syndrome 1	GLE1	
Inclusion Body Myopathy 2	GNE	
Mucolipidosis II/IIIA	GNPTAB	
Mucolipidosis III gamma	GNPTG	
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	GNS	
Geroderma osteodysplastica	GORAB	
Bernard-Soulier Syndrome, Type A2	GP1BA	
Bernard-Soulier Syndrome, Type B	GP1BB	
Bernard-Soulier Syndrome, Type C	GP9	
Primary Hyperoxaluria, Type 2	GRHPR	
Leber congenital amaurosis 1	GUCY2D	
Mucopolysaccharidosis, Type VII	GUSB	
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	
Trifunctional protein deficiency	HADHB	
Congenital Neutropenia, HAX1-Related	HAX1	
Alpha-Thalassemia	HBA1	SC
Alpha-Thalassemia	HBA2	SC
Beta-Hemoglobinopathies	HBB	CNV
Tay-Sachs Disease	HEXA	CNV
Sandhoff Disease	HEXB	
Hemochromatosis, Type 1	HFE	
Hemochromatosis, Type 2A	HFE2	
Alkaptonuria	HGD	
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	HGSNAT	
Holocarboxylase Synthetase Deficiency	HLCS	
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	

Heme Oxygenase-1 Deficiency	HMOX1	
Primary Hyperoxaluria, Type 3	HOGA1	
Tyrosinemia, Type III	HPD	
Hermansky-Pudlak Syndrome 1	HPS1	
Hermansky-Pudlak Syndrome 3	HPS3	
Hermansky-Pudlak syndrome 4	HPS4	
17-beta hydroxysteroid dehydrogenase 3 deficiency	HSD17B3	
D-Bifunctional Protein Deficiency	HSD17B4	
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	HSD3B2	
Hydrolethalus Syndrome	HYLS1	
Mucopolysaccharidosis, Type II (Hunter Syndrome)	IDS	
Mucopolysaccharidosis, Type I (Hurler Syndrome)	IDUA	
Severe Combined Immunodeficiency, X-Linked	IL2RG	
Glanzmann thrombasthenia	ITGB3	CNV
Isovaleric Acidemia	IVD	
Congenital Hyperinsulinism, KCNJ11-Related	KCNJ11	
LAMA2-related Muscular Dystrophy	LAMA2	
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	LAMA3	
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	LAMB3	
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	LAMC2	
Leber Congenital Amaurosis, Type LCA5	LCA5	
Familial Hypercholesterolemia, LDLR-Related	LDLR	
Familial Hypercholesterolemia, LDLRAP1-Related	LDLRAP1	
Leydig cell hypoplasia	LHCGR	
Stuve-Wiedemann Syndrome	LIFR	
Lysosomal Acid Lipase Deficiency	LIPA	
Woolly Hair/Hypotrichosis Syndrome	LIPH	
Deafness, Autosomal Recessive 77	LOXHD1	
Lipoprotein Lipase Deficiency	LPL	
Leigh Syndrome, French-Canadian Type	LRPPRC	
Chediak-Higashi syndrome	LYST	
Alpha-Mannosidosis	MAN2B1	
Hypermethioninemia	MAT1A	
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1	
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2	
Mucolipidosis, Type IV	MCOLN1	CNV
RETT Syndrome	MECP2	
Microcephaly, postnatal progressive, with seizures and brain atrophy	MED17	
Familial Mediterranean Fever	MEFV	
Spondylothoracic Dysostosis, MESP2-Related	MESP2	
Ceroid Lipofuscinosis, Neuronal, 7	MFSD8	
Bardet-Biedl Syndrome 6	MKKS	
Meckel-Gruber Syndrome, Type 1	MKS1	
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	
Malonyl-CoA decarboxylase deficiency	MLYCD	
Methylmalonic Aciduria, MMAA-Related	MMAA	
Methylmalonic Aciduria, MMAB-Related	MMAB	
Methylmalonic Aciduria and Homocystinuria, Type cblC	MMACHC	
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC	
Molybdenum cofactor deficiency	MOCS1	
Congenital Disorder of Glycosylation, Type 1B	MPI	
Congenital Amegakaryocytic Thrombocytopenia	MPL	
Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Relat	MPV17	
Ataxia-telangiectasia-like disorder 1	MRE11	

Homocystinuria due to Deficiency of MTHFR	MTHFR	
Myotubular Myopathy, X-Linked	MTM1	
Homocystinuria, Type cblE	MTRR	
Abetalipoproteinemia	MTTP	
Methylmalonic Aciduria, Type mut(0)	MUT	
Deafness, autosomal recessive, 3	MYO15A	
Usher Syndrome, Type 1B	MYO7A	
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	NAGLU	
N-acetylglutamate Synthase Deficiency	NAGS	
Nijmegen Breakage Syndrome	NBN	
Charcot-Marie-Tooth Disease type 4D	NDRG1	
Mitochondrial Complex I Deficiency, NDUFAF5-Related	NDUFAF5	
Mitochondrial complex I deficiency	NDUFS4	
Mitochondrial Complex I Deficiency, NDUFS6-Related	NDUFS6	
Nemaline Myopathy, NEB-Related	NEB	CNV
Sialidosis	NEU1	
Hydatidiform Mole, Recurrent	NLRP7	
Niemann-Pick Disease, Type C1/D	NPC1	
Niemann-Pick Disease, Type C2	NPC2	
Juvenile Nephronophthisis	NPHP1	
Congenital Finnish Nephrosis	NPHS1	
Steroid-Resistant Nephrotic Syndrome	NPHS2	
Congenital Adrenal Hypoplasia, X-linked	NR0B1	
Enhanced S-Cone Syndrome	NR2E3	
Congenital Insensitivity to Pain with Anhidrosis (CIPA)	NTRK1	
Ornithine Aminotransferase Deficiency	OAT	
Lowe syndrome, X-Linked	OCRL	
Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)	OPA3	
Ornithine Transcarbamylase Deficiency	OTC	
Phenylketonuria	PAH	CNV
Pantothenate Kinase-Associated Neurodegeneration	PANK2	
Pyruvate Carboxylase Deficiency	PC	
Propionic Acidemia, PCCA-Related	PCCA	
Propionic Acidemia, PCCB-Related	PCCB	
Usher Syndrome, Type 1F	PCDH15	CNV
Pyruvate Dehydrogenase Deficiency, X-Linked	PDHA1	
Pyruvate Dehydrogenase Deficiency, PDHB-Related	PDHB	
Prolidase deficiency	PEPD	
Cytochrome-c oxidase deficiency	PET100	
Peroxisome Biogenesis Disorder 1A (Zellweger)	PEX1	
Peroxisome Biogenesis Disorder 6A (Zellweger)	PEX10	
Peroxisome Biogenesis Disorder 3A (Zellweger)	PEX12	
Peroxisome Biogenesis Disorder 5A (Zellweger)	PEX2	
Peroxisome Biogenesis Disorder 4A (Zellweger)	PEX6	
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	
Glycogen Storage Disease, Type VII	PFKM	
Phosphoglycerate Dehydrogenase Deficiency	PHGDH	
Multiple congenital anomalies-hypotonia-seizures syndrome 1	PIGN	
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	
Infantile neuroaxonal dystrophy 1	PLA2G6	
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	PMM2	
Pyridoxal 5'-phosphate-dependent epilepsy	PNPO	
POLG-Related Disorders	POLG	
Xeroderma pigmentosum Variant	POLH	

Muscle-Eye-Brain Disease, POMGNT1-Related	POMGNT1	
Cytochrome P450 oxidoreductase deficiency	POR	
Ceroid Lipofuscinosis, Neuronal, 1	PPT1	
Myasthenic syndrome, congenital, 22	PREPL	CNV
Combined Pituitary Hormone Deficiency 2	PROP1	
Arts syndrome, X-Linked	PRPS1	
Metachromatic Leukodystrophy, PSAP-Related	PSAP	
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	PTS	
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	PUS1	
Glycogen Storage Disease, Type V (McArdle Disease)	PYGM	
Carpenter Syndrome	RAB23	
Omenn Syndrome, RAG1-Related	RAG1	
Omenn Syndrome, RAG2-Related	RAG2	
Congenital Myasthenic Syndrome, RAPSN-Related	RAPSN	
Pontocerebellar Hypoplasia, Type 1 and 6, RARS2-Related	RARS2	
Leber Congenital Amaurosis, Type RDH12	RDH12	
Retinal Dystrophies, RLBP1-Associated	RLBP1	
Cartilage-Hair Hypoplasia	RMRP	
Aicardi-Goutieres syndrome, RNASEH2C-related	RNASEH2C	
Leber Congenital Amaurosis 2	RPE65	
Ciliopathies, RPGRIP1L-Related	RPGRIP1L	
Juvenile Retinoschisis, X-Linked	RS1	
Dyskeratosis Congenita, RTEL1-Related	RTEL1	
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	
MIRAGE syndrome	SAMD9	
Aicardi-Goutières Syndrome	SAMHD1	CNV
Shwachman-Diamond syndrome	SBDS	
Pontocerebellar Hypoplasia, Type 2D	SEPSECS	
Alpha-1-Antitrypsin Deficiency	SERPINA1	
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	
Limb-Girdle Muscular Dystrophy, Type 2F	SGCD	
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	SGSH	
Gitelman Syndrome	SLC12A3	
Agenesis of the Corpus Callosum with Peripheral Neuropathy (Anderm	SLC12A6	
Salla Disease	SLC17A5	
Megaloblastic Anemia Syndrome	SLC19A2	
Carnitine Deficiency	SLC22A5	
Citrullinemia, Type II	SLC25A13	
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrc	SLC25A15	
Carnitine-acylcarnitine translocase deficiency	SLC25A20	
Achondrogenesis, Type 1B	SLC26A2	
Congenital Chloride Diarrhea	SLC26A3	
Pendred Syndrome	SLC26A4	
Autism Spectrum, Epilepsy and Arthrogryposis	SLC35A3	
Glycogen Storage Disease, Type IB	SLC37A4	
Acrodermatitis Enteropathica	SLC39A4	
Cystinuria, Type A	SLC3A1	CNV
Oculocutaneous albinism, Type 4	SLC45A2	
Corneal Dystrophy and Perceptive Deafness	SLC4A11	
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1	SLC6A8	
Lysinuric Protein Intolerance	SLC7A7	
Cystinuria, Type B	SLC7A9	

Schimke Immunoosseous Dysplasia	SMARCAL1	
Spinal Muscular Atrophy	SMN1	SC
Niemann-Pick Disease, Types A/B	SMPD1	
5-alpha reductase deficiency	SRD5A2	
GM3 synthase deficiency	ST3GAL5	
Lipoid Congenital Adrenal Hyperplasia	STAR	
Deafness, autosomal recessive 16	STRC	CNV
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with c	SUCLA2	
Multiple Sulfatase Deficiency	SUMF1	
Leigh Syndrome	SURF1	
Tyrosinemia, Type II	TAT	
Osteopetrosis, Infantile Malignant, TCIRG1-Related	TCIRG1	
Hereditary Spastic Paraparesis, Type 49	TECPR2	
Hemochromatosis, Type 3, TFR2-Related	TFR2	
Lamellar Ichthyosis, Type 1	TGM1	
Segawa Syndrome, TH-Related	TH	
Deafness, autosomal dominant 36, autosomal recessive 7	TMC1	
Joubert Syndrome 2 / Meckel Syndrome 2	TMEM216	
Congenital hypothyroidism	TPO	
Ceroid Lipofuscinosis, Neuronal, 2	TPP1	
Aicardi-Goutieres syndrome, TREX1-related	TREX1	
Bardet-Biedl syndrome 11	TRIM32	
Mulibrey nanism syndrome	TRIM37	
Acute Infantile Liver Failure, TRMU-Related	TRMU	
Pontocerebellar hypoplasia	TSEN54	
Combined Oxidative Phosphorylation Deficiency 3	TSFM	
Congenital hypothyroidism	TSHB	
Hypothyroidism, congenital, nongoitrous, 1	TSHR	
Tricho-Hepato-Enteric Syndrome	TTC37	
Familial dilated cardiomyopathy	TTN	
Ataxia with Vitamin E Deficiency	TTPA	
Myoneurogastrointestinal Encephalopathy (MNGIE)	TYMP	
Oculocutaneous Albinism, Type 1	TYR	
Oculocutaneous albinism, Type 3	TYRP1	
Crigler-Najjar Syndrome	UGT1A1	
Beta-ureidopropionase deficiency	UPB1	
Usher Syndrome, Type 1C	USH1C	
Usher Syndrome, Type 2A	USH2A	CNV
Choreo-acanthocytosis	VPS13A	CNV
Cohen Syndrome	VPS13B	
Congenital Neutropenia, VPS45-Related	VPS45	
Pontocerebellar Hypoplasia, Type 2E	VPS53	
Pontocerebellar Hypoplasia, Type 1A	VRK1	
Microphthalmia/Anophthalmia, VSX2-Related	VSX2	
Von Willebrand disease	VWF	
Wiskott-Aldrich syndrome, X-Linked	WAS	
Progressive Pseudorheumatoid Dysplasia	WISP3	
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	WNT10A	
Werner Syndrome	WRN	
Xeroderma pigmentosum Group A	XPA	
Xeroderma Pigmentosum Group C	XPC	
Spastic Paraplegia Type 15	ZFYVE26	