# CarrierSeq ECS Kit: 420-gene panel menu

## Increase variant detection rates by targeting a broader range of recessive and inherited disorders

Using next-generation sequencing (NGS), the Ion Torrent™ CarrierSeq™ ECS Kit 420-gene panel targets the full coding region of all genes, enabling the analysis of >36,000 nonbenign ClinVar variants for single-nucleotide variants (SNVs), insertions and deletions (indels), and copy number variants (CNVs).

| Variant | Gene | CNV target |
|---|---|---|
| Achalasia-addisonianism-alacrima syndrome | *AAAS* | CNV |
| Harlequin ichthyosis | *ABCA12* | CNV |
| Stargardt disease, type 1 | *ABCA4* | CNV |
| Progressive familial intrahepatic cholestasis, type 2 | *ABCB11* | CNV |
| Progressive familial intrahepatic cholestasis, type 3 | *ABCB4* | CNV |
| Pseudoxanthoma elasticum | *ABCC6* | CNV |
| Familial hyperinsulinism, *ABCC8*-related | *ABCC8* | CNV |
| Adrenoleukodystrophy, X-linked | *ABCD1* | CNV |
| Mitochondrial complex I deficiency, *ACAD9*-related | *ACAD9* | CNV |
| Medium chain acyl-CoA dehydrogenase deficiency | *ACADM* | CNV |
| Short chain acyl-CoA dehydrogenase deficiency | *ACADS* | CNV |
| Short/branched chain acyl-CoA dehydrogenase deficiency | *ACADSB* | CNV |
| Very long chain acyl-CoA dehydrogenase deficiency | *ACADVL* | CNV |
| Beta-ketothiolase deficiency | *ACAT1* | CNV |
| Acyl-CoA oxidase I deficiency | *ACOX1* | CNV |
| Combined malonic and methylmalonic aciduria | *ACSF3* | CNV |
| Severe combined immunodeficiency, ADA-related | *ADA* | CNV |

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| Variant | Gene | CNV target |
|---|---|---|
| Ehlers-Danlos syndrome, type VIIC | *ADAMTS2* | CNV |
| Bilateral frontoparietal polymicrogyria | *ADGRG1* | CNV |
| Aspartylglucosaminuria | *AGA* | CNV |
| Glycogen storage disease, type III (Cori/Forbes) | *AGL* | CNV |
| Rhizomelic chondrodysplasia punctata, type 3 | *AGPS* | CNV |
| Hyperoxaluria, primary, type 1 | *AGXT* | CNV |
| Autoimmune polyendocrinopathy syndrome, type I | *AIRE* | CNV |
| Sjögren-Larsson syndrome | *ALDH3A2* | CNV |
| Pyridoxine-dependent epilepsy | *ALDH7A1* | CNV |
| Hereditary fructose intolerance | *ALDOB* | CNV |
| Congenital disorder of glycosylation, type 1C | *ALG6* | CNV |
| Alström syndrome | *ALMS1* | CNV |
| Hypophosphatasia, *ALPL*-related | *ALPL* | CNV |
| Persistent Müllerian duct syndrome, type 1 | *AMH* | CNV |
| Persistent Müllerian duct syndrome, type 2 | *AMHR2* | CNV |
| Glycine encephalopathy, *AMT*-related | *AMT* | CNV |
| Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK) | *AP1S1* | CNV |
| Familial nephrogenic diabetes insipidus, *AQP2*-related | *AQP2* | CNV |
| Androgen insensitivity syndrome, X-linked | *AR* | CNV |
| Argininemia | *ARG1* | CNV |
| Metachromatic leukodystrophy, *ARSA*-related | *ARSA* | CNV |
| Mucopolysaccharidosis, type VI (Maroteaux-Lamy) | *ARSB* | CNV |
| Argininosuccinate lyase deficiency | *ASL* | CNV |
| Asparagine synthetase deficiency | *ASNS* | CNV |
| Canavan disease | *ASPA* | CNV |
| Citrullinemia, type 1 | *ASS1* | CNV |
| Ataxia-telangiectasia | *ATM* | CNV+ |
| Renal tubular acidosis and deafness, *ATP6V1B1*-related | *ATP6V1B1* | CNV |
| Menkes syndrome, X-linked | *ATP7A* | CNV |
| Wilson disease | *ATP7B* | CNV |
| Progressive familial intrahepatic cholestasis, type 1 | *ATP8B1* | CNV |
| Alpha-thalassemia intellectual disability syndrome, X-linked | *ATRX* | CNV |
| Bardet-Biedl syndrome 1 | *BBS1* | CNV |
| Bardet-Biedl syndrome 10 | *BBS10* | CNV |
| Bardet-Biedl syndrome 12 | *BBS12* | CNV |
| Bardet-Biedl syndrome 2 | *BBS2* | CNV |
| Bardet-Biedl syndrome 4 | *BBS4* | CNV+ |
| Bardet-Biedl syndrome 9 | *BBS9* | CNV |
| Pseudocholinesterase deficiency | *BCHE* | CNV |
| Maple syrup urine disease, type 1A | *BCKDHA* | CNV |
| Maple syrup urine disease, type 1B | *BCKDHB* | CNV |
| GRACILE syndrome | *BCS1L* | CNV |
| Bloom syndrome | *BLM* | CNV |
| Fanconi anemia, group J | *BRIP1* | CNV |
| Bartter syndrome, type 4a | *BSND* | CNV |
| Biotinidase deficiency | *BTD* | CNV |
| Isolated growth hormone deficiency, type III, X-linked | *BTK* | CNV |
| Desbuquois dysplasia 1 | *CANT1* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Limb-girdle muscular dystrophy, type 2A | *CAPN3* | CNV |
| Catecholaminergic polymorphic ventricular tachycardia | *CASQ2* | CNV |
| Homocystinuria, *CBS*-related | *CBS* | CNV |
| Mental retardation, autosomal recessive 3 | *CC2D1A* | CNV+ |
| Usher syndrome, type 1D | *CDH23* | CNV |
| Leber congenital amaurosis, type *CEP290* | *CEP290* | CNV |
| Retinitis pigmentosa 26 | *CERKL* | CNV |
| Cystic fibrosis | *CFTR* | CNV+ |
| Choroideremia, X-linked | *CHM* | CNV |
| Congenital myasthenic syndrome, *CHRNE*-related | *CHRNE* | CNV |
| Escobar syndrome | *CHRNG* | CNV |
| Bare lymphocyte syndrome, *CIITA*-related | *CIITA* | CNV |
| Ceroid lipofuscinosis, neuronal, 3 | *CLN3* | CNV+ |
| Ceroid lipofuscinosis, neuronal, *5* | *CLN5* | CNV |
| Ceroid lipofuscinosis, neuronal, 6 | *CLN6* | CNV |
| Ceroid lipofuscinosis, neuronal, 8 (a.k.a. Northern epilepsy) | *CLN8* | CNV |
| Usher syndrome, type 3 | *CLRN1* | CNV |
| Achromatopsia, *CNGA3*-related | *CNGA3* | CNV |
| Achromatopsia, *CNGB3*-related | *CNGB3* | CNV |
| Fibrochondrogenesis, type 2 | *COL11A2* | CNV |
| Alport syndrome, *COL4A3*-related | *COL4A3* | CNV |
| Alport syndrome, *COL4A4*-related | *COL4A4* | CNV |
| Alport syndrome, X-linked | *COL4A5* | CNV |
| Dystrophic epidermolysis bullosa, *COL7A1*-related | *COL7A1* | CNV |
| Carbamoyl phosphate synthetase I deficiency | *CPS1* | CNV |
| Carnitine palmitoyltransferase IA deficiency | *CPT1A* | CNV |
| Carnitine palmitoyltransferase II deficiency | *CPT2* | CNV |
| Leber congenital amaurosis 8 | *CRB1* | CNV |
| Cystinosis | *CTNS* | CNV+ |
| Papillon-Lefevre syndrome | *CTSC* | CNV |
| Ceroid lipofuscinosis, neuronal, 10 (CLN10 disease) | *CTSD* | CNV |
| Pycnodysostosis | *CTSK* | CNV |
| Chronic granulomatous disease, *CYBA*-related | *CYBA* | CNV |
| Chronic granulomatous disease, X-linked | *CYBB* | CNV |
| Congenital adrenal hyperplasia, 11-beta-hydroxylase-deficient | *CYP11B1* | CNV |
| Corticosterone methyloxidase deficiency | *CYP11B2* | CNV |
| Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency | *CYP17A1* | CNV |
| Aromatase deficiency | *CYP19A1* | CNV |
| Primary congenital glaucoma | *CYP1B1* | CNV |
| Congenital adrenal hyperplasia, 21-hydroxylase-deficient | *CYP21A2* | SC |
| Cerebrotendinous xanthomatosis | *CYP27A1* | CNV |
| Vitamin D–dependent rickets, type 1A | *CYP27B1* | CNV |
| Maple syrup urine disease, type 2 | *DBT* | CNV |
| Severe combined immunodeficiency, type athabaskan | *DCLRE1C* | CNV |
| Xeroderma pigmentosum group E | *DDB2* | CNV |
| Smith-Lemli-Opitz syndrome | *DHCR7* | CNV |
| Retinitis pigmentosa 59 | *DHDDS* | CNV |
| Dyskeratosis congenita, X-linked | *DKC1* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Dihydrolipoamide dehydrogenase deficiency | *DLD* | CNV |
| Duchenne/Becker muscular dystrophy | *DMD* | CNV+ |
| Ciliary dyskinesia, primary 3 | *DNAH5* | CNV |
| Ciliary dyskinesia, primary 1 | *DNAI1* | CNV |
| Ciliary dyskinesia, primary 9 | *DNAI2* | CNV |
| Ciliary dyskinesia, primary, 16 | *DNAL1* | CNV |
| Congenital myasthenic syndrome, *DOK7*-related | *DOK7* | CNV |
| Dihydropyrimidine dehydrogenase deficiency | *DPYD* | CNV |
| Limb-girdle muscular dystrophy, type 2B | *DYSF* | CNV |
| Hypohidrotic ectodermal dysplasia, X-linked | *EDA* | CNV |
| Hypohidrotic ectodermal dysplasia | *EDAR* | CNV |
| Wolcott-Rallison syndrome | *EIF2AK3* | CNV |
| Leukoencephalopathy with vanishing white matter | *EIF2B5* | CNV |
| Emery-Dreifuss muscular dystrophy 1, X-linked | *EMD* | CNV |
| Xeroderma pigmentosum, group D | *ERCC2* | CNV |
| Xeroderma pigmentosum, group B | *ERCC3* | CNV |
| Xeroderma pigmentosum, group F | *ERCC4* | CNV |
| Xeroderma pigmentosum, group G | *ERCC5* | CNV |
| Cockayne syndrome, type B | *ERCC6* | CNV |
| Cockayne syndrome, type A | *ERCC8* | CNV |
| Roberts syndrome | *ESCO2* | CNV |
| Glutaric acidemia, type 2A | *ETFA* | CNV |
| Glutaric acidemia, type 2B | *ETFB* | CNV |
| Glutaric acidemia, type 2C | *ETFDH* | CNV |
| Ethylmalonic encephalopathy | *ETHE1* | CNV |
| Ellis-van Creveld syndrome, *EVC*-related | *EVC* | CNV |
| Ellis-van Creveld syndrome, *EVC2*-related | *EVC2* | CNV |
| Pontocerebellar hypoplasia, type 1B | *EXOSC3* | CNV |
| Retinitis pigmentosa 25 | *EYS* | CNV |
| Factor XI deficiency | *F11* | CNV |
| Prothrombin deficiency | *F2* | CNV |
| Hemophilia A | *F8* | CNV |
| Hemophilia B | *F9* | CNV |
| Tyrosinemia, type I | *FAH* | CNV |
| Retinitis pigmentosa 28 | *FAM161A* | CNV |
| Fanconi anemia, group A | *FANCA* | CNV+ |
| Fanconi anemia, group C | *FANCC* | CNV+ |
| Fanconi anemia, group G | *FANCG* | CNV |
| Fumarase deficiency | *FH* | CNV |
| Limb-girdle muscular dystrophy, type 2I | *FKRP* | CNV |
| Walker-Warburg syndrome, *FKTN*-related | *FKTN* | CNV |
| Glycogen storage disease, type IA | *G6PC* | CNV |
| Glucose-6-phosphate dehydrogenase deficiency | *G6PD* | CNV |
| Glycogen storage disease, type II (Pompe disease) | *GAA* | CNV+ |
| Krabbe disease | *GALC* | CNV+ |
| Galactose epimerase deficiency | *GALE* | CNV |
| Galactokinase deficiency (galactosemia, type II) | *GALK1* | CNV |
| Mucopolysaccharidosis, type IVA | *GALNS* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Hyperphosphatemic familial tumoral calcinosis | *GALNT3* | CNV |
| Galactosemia | *GALT* | CNV+ |
| Guanidinoacetate methyltransferase deficiency | *GAMT* | CNV |
| Gaucher disease | *GBA* | CNV |
| Glycogen storage disease, type IV | *GBE1* | CNV |
| Glutaric acidemia, type 1 | *GCDH* | CNV |
| Dopa-responsive dystonia | *GCH1* | CNV |
| Grebe syndrome | *GDF5* | CNV |
| Combined oxidative phosphorylation deficiency 1 | *GFM1* | CNV |
| Isolated growth hormone deficiency, type IA/II | *GH1* | CNV+ |
| Isolated growth hormone deficiency, type IB | *GHRHR* | CNV |
| Charcot-Marie-Tooth disease with deafness, X-linked | *GJB1* | CNV |
| Non-syndromic hearing loss (a.k.a. connexin 26) | *GJB2* | CNV+ |
| Erythrokeratodermia variabilis et progressiva | *GJB3* | CNV |
| Non-syndromic hearing loss (a.k.a. connexin 30) | *GJB6* | CNV+ |
| Fabry disease | *GLA* | CNV+ |
| Mucopolysaccharidosis, type IVB / GM1 gangliosidosis | *GLB1* | CNV |
| Glycine encephalopathy, *GLDC*-related | *GLDC* | CNV |
| Lethal congenital contracture syndrome 1 | *GLE1* | CNV |
| Inclusion body myopathy 2 | *GNE* | CNV |
| Mucolipidosis II/IIIA | *GNPTAB* | CNV |
| Mucolipidosis III gamma | *GNPTG* | CNV |
| Mucopolysaccharidosis, type IIID (Sanfilippo D) | *GNS* | CNV |
| Geroderma osteodysplastica | *GORAB* | CNV |
| Bernard-Soulier syndrome, type A2 | *GP1BA* | CNV |
| Bernard-Soulier syndrome, type B | *GP1BB* | CNV |
| Bernard-Soulier syndrome, type C | *GP9* | CNV |
| Primary hyperoxaluria, type 2 | *GRHPR* | CNV |
| Leber congenital amaurosis 1 | *GUCY2D* | CNV |
| Mucopolysaccharidosis, type VII | *GUSB* | CNV |
| Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency | *HADHA* | CNV |
| Trifunctional protein deficiency | *HADHB* | CNV |
| Congenital neutropenia, *HAX1*-related | *HAX1* | CNV |
| Alpha-thalassemia | *HBA1* | SC |
| Alpha-thalassemia | *HBA2* | SC |
| Beta-hemoglobinopathies | *HBB* | CNV+ |
| Tay-Sachs disease | *HEXA* | CNV+ |
| Sandhoff disease | *HEXB* | CNV |
| Hemochromatosis, type 1 | *HFE* | CNV |
| Hemochromatosis, type 2A | *HFE2* | CNV |
| Alkaptonuria | *HGD* | CNV |
| Mucopolysaccharidosis, type IIIC (Sanfilippo C) | *HGSNAT* | CNV |
| Holocarboxylase synthetase deficiency | *HLCS* | CNV |
| 3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency | *HMGCL* | CNV |
| Heme oxygenase-1 deficiency | *HMOX1* | CNV |
| Primary hyperoxaluria, type 3 | *HOGA1* | CNV |
| Tyrosinemia, type 3 | *HPD* | CNV |
| Hermansky-Pudlak syndrome 1 | *HPS1* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Hermansky-Pudlak syndrome 3 | *HPS3* | CNV |
| Hermansky-Pudlak syndrome 4 | *HPS4* | CNV |
| 17-beta hydroxysteroid dehydrogenase 3 deficiency | *HSD17B3* | CNV |
| D-bifunctional protein deficiency | *HSD17B4* | CNV |
| 3-beta-hydroxysteroid dehydrogenase type II deficiency | *HSD3B2* | CNV |
| Hydrolethalus syndrome | *HYLS1* | CNV |
| Mucopolysaccharidosis, type II (Hunter syndrome) | *IDS* | CNV |
| Mucopolysaccharidosis, type I (Hurler syndrome) | *IDUA* | CNV |
| Dysautonomia, familial (*IKBKAP* or *ELP1*) | *IKBKAP* | CNV |
| Severe combined immunodeficiency, X-linked | *IL2RG* | CNV |
| Glanzmann thrombasthenia | *ITGB3* | CNV+ |
| Isovaleric acidemia | *IVD* | CNV |
| Congenital hyperinsulinism, *KCNJ11*-related | *KCNJ11* | CNV |
| *LAMA2*-related muscular dystrophy | *LAMA2* | CNV |
| Herlitz junctional epidermolysis bullosa, *LAMA3*-related | *LAMA3* | CNV |
| Herlitz junctional epidermolysis bullosa, *LAMB3*-related | *LAMB3* | CNV |
| Herlitz junctional epidermolysis bullosa, *LAMC2*-related | *LAMC2* | CNV |
| Leber congenital amaurosis, type *LCA5* | *LCA5* | CNV |
| Familial hypercholesterolemia, *LDLR*-related | *LDLR* | CNV |
| Familial hypercholesterolemia, *LDLRAP1*-related | *LDLRAP1* | CNV |
| Leydig cell hypoplasia | *LHCGR* | CNV |
| Stuve-Wiedemann syndrome | *LIFR* | CNV |
| Lysosomal acid lipase deficiency | *LIPA* | CNV |
| Woolly hair/hypotrichosis syndrome | *LIPH* | CNV |
| Deafness, autosomal recessive 77 | *LOXHD1* | CNV |
| Lipoprotein lipase deficiency | *LPL* | CNV |
| Leigh syndrome, French-Canadian type | *LRPPRC* | CNV |
| Chediak-Higashi syndrome | *LYST* | CNV |
| Alpha-mannosidosis | *MAN2B1* | CNV |
| Hypermethioninemia | *MAT1A* | CNV |
| 3-methylcrotonyl-CoA carboxylase 1 deficiency | *MCCC1* | CNV |
| 3-methylcrotonyl-CoA carboxylase 2 deficiency | *MCCC2* | CNV |
| Mucolipidosis, type IV | *MCOLN1* | CNV+ |
| RETT syndrome | *MECP2* | CNV |
| Microcephaly, postnatal progressive, with seizures and brain atrophy | *MED17* | CNV |
| Familial Mediterranean fever | *MEFV* | CNV |
| Spondylothoracic dysostosis, *MESP2*-related | *MESP2* | CNV |
| Ceroid lipofuscinosis, neuronal, 7 | *MFSD8* | CNV |
| Bardet-Biedl syndrome 6 | *MKKS* | CNV |
| Meckel-Gruber syndrome, type 1 | *MKS1* | CNV |
| Megalencephalic leukoencephalopathy with subcortical cysts | *MLC1* | CNV |
| Malonyl-CoA decarboxylase deficiency | *MLYCD* | CNV |
| Methylmalonic aciduria, *MMAA*-related | *MMAA* | CNV |
| Methylmalonic aciduria, *MMAB*-related | *MMAB* | CNV |
| Methylmalonic aciduria and homocystinuria, type cblC | *MMACHC* | CNV |
| Methylmalonic aciduria and homocystinuria, type cblD | *MMADHC* | CNV |
| Molybdenum cofactor deficiency | *MOCS1* | CNV |
| Congenital disorder of glycosylation, type 1B | *MPI* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Congenital amegakaryocytic thrombocytopenia | *MPL* | CNV |
| Hepatocerebral mitochondrial DNA depletion syndrome, *MPV17*-related | *MPV17* | CNV |
| Ataxia-telangiectasia-like disorder 1 | *MRE11* | CNV |
| Homocystinuria due to deficiency of *MTHFR* | *MTHFR* | CNV |
| Myotubular myopathy, X-linked | *MTM1* | CNV |
| Homocystinuria, type cblE | *MTRR* | CNV |
| Abetalipoproteinemia | *MTTP* | CNV |
| Methylmalonic aciduria, type mut(0) | *MUT* | CNV |
| Deafness, autosomal recessive, 3 | *MYO15A* | CNV |
| Usher syndrome, type 1B | *MYO7A* | CNV |
| Mucopolysaccharidosis, type IIIB (Sanfilippo B) | *NAGLU* | CNV |
| N-acetylglutamate synthase deficiency | *NAGS* | CNV |
| Nijmegen breakage syndrome | *NBN* | CNV |
| Charcot-Marie-Tooth disease type 4D | *NDRG1* | CNV |
| Mitochondrial complex I deficiency, *NDUFAF5*-related | *NDUFAF5* | CNV |
| Mitochondrial complex I deficiency | *NDUFS4* | CNV |
| Mitochondrial complex I deficiency, *NDUFS6*-related | *NDUFS6* | CNV |
| Nemaline myopathy, *NEB*-related | *NEB* | CNV+ |
| Sialidosis | *NEU1* | CNV |
| Hydatidiform mole, recurrent | *NLRP7* | CNV |
| Niemann-Pick disease, type C1/D | *NPC1* | CNV |
| Niemann-Pick disease, type C2 | *NPC2* | CNV |
| Juvenile nephronophthisis | *NPHP1* | CNV |
| Congenital Finnish nephrosis | *NPHS1* | CNV |
| Steroid-resistant nephrotic syndrome | *NPHS2* | CNV |
| Congenital adrenal hypoplasia, X-linked | *NR0B1* | CNV |
| Enhanced S-cone syndrome | *NR2E3* | CNV |
| Congenital insensitivity to pain with anhidrosis (CIPA) | *NTRK1* | CNV |
| Ornithine aminotransferase deficiency | *OAT* | CNV |
| Lowe syndrome, X-linked | *OCRL* | CNV |
| Costeff syndrome (3-methylglutaconic aciduria, type 3) | *OPA3* | CNV |
| Ornithine transcarbamylase deficiency | *OTC* | CNV |
| Phenylketonuria | *PAH* | CNV+ |
| Pantothenate kinase-associated neurodegeneration | *PANK2* | CNV |
| Pyruvate carboxylase deficiency | *PC* | CNV |
| Propionic acidemia, *PCCA*-related | *PCCA* | CNV |
| Propionic acidemia, *PCCB*-related | *PCCB* | CNV |
| Usher syndrome, type 1F | *PCDH15* | CNV+ |
| Pyruvate dehydrogenase deficiency, X-linked | *PDHA1* | CNV |
| Pyruvate dehydrogenase deficiency, *PDHB*-related | *PDHB* | CNV |
| Prolidase deficiency | *PEPD* | CNV |
| Cytochrome-c oxidase deficiency | *PET100* | CNV |
| Peroxisome biogenesis disorder 1A (Zellweger) | *PEX1* | CNV |
| Peroxisome biogenesis disorder 6A (Zellweger) | *PEX10* | CNV |
| Peroxisome biogenesis disorder 3A (Zellweger) | *PEX12* | CNV |
| Peroxisome biogenesis disorder 5A (Zellweger) | *PEX2* | CNV |
| Peroxisome biogenesis disorder 4A (Zellweger) | *PEX6* | CNV |
| Rhizomelic chondrodysplasia punctata, type 1 | *PEX7* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Glycogen storage disease, type VII | *PFKM* | CNV |
| Phosphoglycerate dehydrogenase deficiency | *PHGDH* | CNV |
| Multiple congenital anomalies-hypotonia-seizures syndrome 1 | *PIGN* | CNV |
| Polycystic kidney disease, autosomal recessive | *PKHD1* | CNV |
| Infantile neuroaxonal dystrophy 1 | *PLA2G6* | CNV |
| Congenital disorder of glycosylation, type 1A, *PMM2*-related | *PMM2* | CNV |
| Pyridoxal 5′-phosphate-dependent epilepsy | *PNPO* | CNV |
| *POLG*-related disorders | *POLG* | CNV |
| Xeroderma pigmentosum variant | *POLH* | CNV |
| Muscle-eye-brain disease, *POMGNT1*-related | *POMGNT1* | CNV |
| Cytochrome P450 oxidoreductase deficiency | *POR* | CNV |
| Ceroid lipofuscinosis, neuronal, 1 | *PPT1* | CNV |
| Myasthenic syndrome, congenital, 22 | *PREPL* | CNV+ |
| Combined pituitary hormone deficiency 2 | *PROP1* | CNV |
| Arts syndrome, X-linked | *PRPS1* | CNV |
| Metachromatic leukodystrophy, *PSAP*-related | *PSAP* | CNV |
| 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency | *PTS* | CNV |
| Mitochondrial myopathy and sideroblastic anemia (MLASA1) | *PUS1* | CNV |
| Glycogen storage disease, type V (McArdle disease) | *PYGM* | CNV |
| Carpenter syndrome | *RAB23* | CNV |
| Omenn syndrome, *RAG1*-related | *RAG1* | CNV |
| Omenn syndrome, *RAG2*-related | *RAG2* | CNV |
| Congenital myasthenic syndrome, *RAPSN*-related | *RAPSN* | CNV |
| Pontocerebellar hypoplasia, type 1 and 6, *RARS2*-related | *RARS2* | CNV |
| Leber congenital amaurosis, type *RDH12* | *RDH12* | CNV |
| Retinal dystrophies, *RLBP1*-associated | *RLBP1* | CNV |
| Cartilage-hair hypoplasia | *RMRP* | CNV |
| Aicardi-Goutieres syndrome, *RNASEH2C*-related | *RNASEH2C* | CNV |
| Leber congenital amaurosis 2 | *RPE65* | CNV |
| Ciliopathies, *RPGRIP1L*-related | *RPGRIP1L* | CNV |
| Juvenile retinoschisis, X-linked | *RS1* | CNV |
| Dyskeratosis congenita, *RTEL1*-related | *RTEL1* | CNV |
| Autosomal recessive spastic ataxia of Charlevoix-Saguenay | *SACS* | CNV |
| MIRAGE syndrome | *SAMD9* | CNV |
| Aicardi-Goutires syndrome | *SAMHD1* | CNV+ |
| Shwachman-Diamond syndrome | *SBDS* | CNV |
| Pontocerebellar hypoplasia, type 2D | *SEPSECS* | CNV |
| Alpha-1-antitrypsin deficiency | *SERPINA1* | CNV |
| Limb-girdle muscular dystrophy, type 2D | *SGCA* | CNV |
| Limb-girdle muscular dystrophy, type 2E | *SGCB* | CNV |
| Limb-girdle muscular dystrophy, type 2F | *SGCD* | CNV |
| Limb-girdle muscular dystrophy, type 2C | *SGCG* | CNV |
| Mucopolysaccharidosis, type IIIA (Sanfilippo A) | *SGSH* | CNV |
| Gitelman syndrome | *SLC12A3* | CNV |
| Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome) | *SLC12A6* | CNV |
| Salla disease | *SLC17A5* | CNV |
| Megaloblastic anemia syndrome | *SLC19A2* | CNV |
| Carnitine deficiency | *SLC22A5* | CNV |

| Variant | Gene | CNV target |
|---|---|---|
| Citrullinemia, type II | *SLC25A13* | CNV |
| Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome | *SLC25A15* | CNV |
| Carnitine-acylcarnitine translocase deficiency | *SLC25A20* | CNV |
| Achondrogenesis, type 1B | *SLC26A2* | CNV |
| Congenital chloride diarrhea | *SLC26A3* | CNV |
| Pendred syndrome | *SLC26A4* | CNV |
| Autism spectrum, epilepsy, and arthrogryposis | *SLC35A3* | CNV |
| Glycogen storage disease, type IB | *SLC37A4* | CNV |
| Acrodermatitis enteropathica | *SLC39A4* | CNV |
| Cystinuria, type A | *SLC3A1* | CNV+ |
| Oculocutaneous albinism, type 4 | *SLC45A2* | CNV |
| Corneal dystrophy and perceptive deafness | *SLC4A11* | CNV |
| Creatine transporter defect (cerebral creatine deficiency syndrome 1, X-linked) | *SLC6A8* | CNV |
| Lysinuric protein intolerance | *SLC7A7* | CNV |
| Cystinuria, type B | *SLC7A9* | CNV |
| Schimke immunoosseous dysplasia | *SMARCAL1* | CNV |
| Spinal muscular atrophy | *SMN1* | SC |
| Niemann-Pick disease, types A/B | *SMPD1* | CNV |
| 5-alpha reductase deficiency | *SRD5A2* | CNV |
| GM3 synthase deficiency | *ST3GAL5* | CNV |
| Lipoid congenital adrenal hyperplasia | *STAR* | CNV |
| Deafness, autosomal recessive 16 | *STRC* | CNV+ |
| Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) | *SUCLA2* | CNV |
| Multiple sulfatase deficiency | *SUMF1* | CNV |
| Leigh syndrome | *SURF1* | CNV |
| Tyrosinemia, type II | *TAT* | CNV |
| Osteopetrosis, infantile malignant, *TCIRG1*-related | *TCIRG1* | CNV |
| Hereditary spastic paraparesis, type 49 | *TECPR2* | CNV |
| Hemochromatosis, type 3, *TFR2*-related | *TFR2* | CNV |
| Lamellar ichthyosis, type 1 | *TGM1* | CNV |
| Segawa syndrome, *TH*-related | *TH* | CNV |
| Deafness, autosomal dominant 36, autosomal recessive 7 | *TMC1* | CNV |
| Joubert syndrome 2/Meckel syndrome 2 | *TMEM216* | CNV |
| Congenital hypothyroidism | *TPO* | CNV |
| Ceroid lipofuscinosis, neuronal, 2 | *TPP1* | CNV |
| Aicardi-Goutieres syndrome, *TREX1*-related | *TREX1* | CNV |
| Bardet-Biedl syndrome 11 | *TRIM32* | CNV |
| Mulibrey nanism syndrome | *TRIM37* | CNV |
| Acute infantile liver failure, *TRMU*-related | *TRMU* | CNV |
| Pontocerebellar hypoplasia | *TSEN54* | CNV |
| Combined oxidative phosphorylation deficiency 3 | *TSFM* | CNV |
| Congenital hypothyroidism | *TSHB* | CNV |
| Hypothyroidism, congenital, nongoitrous, 1 | *TSHR* | CNV |
| Tricho-hepato-enteric syndrome | *TTC37* | CNV |
| Familial dilated cardiomyopathy | *TTN* | CNV |
| Ataxia with vitamin E deficiency | *TTPA* | CNV |
| Myoneurogastrointestinal encephalopathy (MNGIE) | *TYMP* | CNV |
| Oculocutaneous albinism, type 1 | *TYR* | CNV |

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| Variant | Gene | CNV target |
|---|---|---|
| Oculocutaneous albinism, type 3 | *TYRP1* | CNV |
| Crigler-Najjar syndrome | *UGT1A1* | CNV |
| Beta-ureidopropionase deficiency | *UPB1* | CNV |
| Usher syndrome, type 1C | *USH1C* | CNV |
| Usher syndrome, type 2A | *USH2A* | CNV+ |
| Choreo-acanthocytosis | *VPS13A* | CNV+ |
| Cohen syndrome | *VPS13B* | CNV |
| Congenital neutropenia, *VPS45*-related | *VPS45* | CNV |
| Pontocerebellar hypoplasia, type 2E | *VPS53* | CNV |
| Pontocerebellar hypoplasia, type 1A | *VRK1* | CNV |
| Microphthalmia/Anophthalmia, *VSX2*-related | *VSX2* | CNV |
| Von Willebrand disease | *VWF* | CNV |
| Wiskott-Aldrich syndrome, X-linked | *WAS* | CNV |
| Progressive pseudorheumatoid dysplasia | *WISP3* | CNV |
| Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome | *WNT10A* | CNV |
| Werner syndrome | *WRN* | CNV |
| Xeroderma pigmentosum group A | *XPA* | CNV |
| Xeroderma pigmentosum group C | *XPC* | CNV |
| Spastic paraplegia type 15 | *ZFYVE26* | CNV |

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