MAYO CLINIC LABORATORIES

Targeted Variants Interrogated by Expanded Carrier Screen Panels

The following list of genes and noted variants are included in ECSP / Expanded Carrier Screen for Select Genetic Disorders, Varies. These genes and variants, except for *FMR1* for Fragile X, are also included in ECWOF / Expanded Carrier Screen Panel for Select Genetic Disorders, Without Fragile X, Varies.

Associated Phenotype	Gene (Transcript)	Variants
17-Alpha-hydroxylase/ 17,20-lyase deficiency	*CYP17A1* (NM_000102)	c.51G>A, c.81C>A, c.160_162del, c.286C>T, c.287G>A, c.316T>C, c.340T>G, c.347A>T, c.601T>A, c.715C>T, c.985T>G, c.1024C>A, c.1039C>T, c.1040G>A, c.1073G>A, c.1084C>T, c.1118A>T, c.1216T>C*, c.1243+5G>A, c.1283C>T, c.1435_1438dup
17-Beta hydroxysteroid dehydrogenase 3 deficiency	*HSD17B3* (NM_000197)	c.239G>A, c.277+4A>T, c.278-1G>C, c.389A>G, c.845C>T
3-Methylglutaconic aciduria type III and autosomal dominant optic atrophy 3 with cataract	*OPA3* (NM_025136)	c.143-1G>C*, c.277G>A, c.415C>T
6-Pyruvoyltetrahydropterin synthase (PTS) deficiency	*PTS* (NM_000317)	c.46C>T, c.74G>A, c.155A>G*, c.259C>T*, c.286G>A, c.347A>G
Abetalipoproteinemia	*MTTP* (NM_000253)	c.419dup, c.1619G>A, c.1769G>T, c.2212del, c.2593G>T*
ACAD9 deficiency	*ACAD9* (NM_014049)	c.130T>A, c.187G>T, c.260T>A, c.380G>A, c.659C>T, c.797G>A, c.976G>C*, c.1237G>A, c.1240C>T, c.1249C>T, c.1405C>T, c.1553G>A, c.1594C>T
Acrodermatitis enteropathica	*SLC39A4* (NM_130849)	c.599C>T, c.989G>A, c.1120G>A, c.1224_1228del, c.1576G>A
Adenosine deaminase deficiency	*ADA* (NM_000022)	c.43C>G, c.58G>A, c.220G>T, c.248C>A, c.301C>T*, c.302G>A*, c.302G>T*, c.320T>C*, c.385G>A, c.419G>A, c.445C>T, c.466C>T, c.467G>A, c.529G>A, c.536C>A, c.596A>C, c.631C>T*, c.632G>A*, c.646G>A, c.872C>T*, c.956_960del*, c.986C>T*
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6* (NM_133647)	c.1478_1485del, c.2023C>T, c.2436+1del, c.3031C>T
Alpha-1-antitrypsin deficiency	*SERPINA1* (NM_000295)	c.863A>T*, c.1096G>A*
Alpha mannosidosis	*MAN2B1* (NM_000528)	c.1780C>T, c.1856dup, c.2248C>T
Alpha-methylacetoacetic aciduria	*ACAT1* (NM_000019)	c.149del, c.433C>G, c.472A>G, c.473A>G, c.623G>A, c.814C>T, c.826+1G>T, c.935T>C, c.1006-2A>C, c.1163+2T>C
Alport syndrome	*COL4A3* (NM_000091)	c.4420_4424del*, c.4441C>T, c.4571C>G
	COL4A4 (NM_000092)	c.81_86del, c.2906C>G*, c.3601G>A, c.3713C>A, c.4129C>T*, c.4715C>T*
	COL4A5 (NM_000495)	c.4691G>C, c.4946T>G, c.5030G>A
Argininosuccinic aciduria	*ASL* (NM_000048)	c.346C>T, c.446+1G>A*, c.532G>A*
Aromatase deficiency	*CYP19A1* (NM_031226)	c.452-1G>A, c.469del, c.628G>A, c.629-3C>A, c.1094G>A, c.1123C>T, c.1303C>T, c.1310G>A
Asparagine synthetase deficiency	*ASNS* (NM_133436)	c.17C>A, c.1084T>G*, c.1193A>G, c.1648C>T
Aspartylglucosaminuria	*AGA* (NM_000027)	c.200_201del, c.214T>C*, c.482G>A, c.488G>C
Ataxia with isolated vitamin E deficiency	*TTPA* (NM_000370)	c.358G>A, c.400C>T, c.487del, c.513_514insTT, c.744del

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Ataxia-telangiectasia	*ATM* (NM_000051)	c.103C>T, c.1564_1565del*, c.3245_3247delinsTGAT, c.3576G>A, c.5712dup, c.5763-1050A>G, c.5908C>T*, c.5932G>T, c.7517_7520del, c.7638_7646del*, Deletion analysis of exon 2–3, exon 4, exon 5–6, exon 7, exon 8–9, exon 10–11, exon 12–16, exon 17–18, exon 19–22, exon 23–24, exon 25–26, exon 27, exon 29–31, exon 32–33, exon 34, exon 35–36, exon 38, exon 40–43, exon 44–45, exon 46–49, exon 50, exon 51–54, exon 55–56, exon 57, exon 58–59, exon 62–63
Autosomal recessive Miyoshi muscular dystrophy, autosomal recessive limb-girdle muscular dystrophy, and autosomal recessive distal myopathy	*DYSF* (NM_003494)	c.1566C>G, c.2372C>G, c.2779del, c.2875C>T, c.2997G>T, c.3373del, c.4497del, c.4872del, c.5057+5G>A, c.5525G>A, c.5713C>T
Autosomal recessive osteopetrosis (ARO)	*TCIRG1* (NM_006019)	c.117+1G>A, c.504-8G>A, c.702del, c.1331G>T, c.1421C>A, c.1554+2T>A, c.1674-1G>A, c.2008C>T
Autosomal recessive polycystic kidney disease	*PKHD1* (NM_138694)	c.107C>T*, c.1342G>C*, c.1486C>T*, c.4870C>T*, c.5895dup*, c.9689del*, c.10412T>G
Bardet-Biedl syndrome 1	*BBS1* (NM_024649)	c.616T>G*, c.830+1G>T, c.1169T>G*
Bardet-Biedl syndrome 10	*BBS10* (NM_024685)	c.271dup*
Bardet-Biedl syndrome 2	*BBS2* (NM_031885)	c.72C>G, c.224T>G, c.311A>C*, c.472-2A>G, c.823C>T, c.940del, c.1895G>C*
Bare lymphocyte syndrome, type II	*CIITA* (NM_000246)	c.1141G>T, c.2063G>A, c.2888+1G>A, c.3080_3082del, c.3229_3233+7del, c.3317+1G>A
Biotin- thiamine-responsive basal ganglia disease (BTBGD)	*SLC19A3* (NM_025243)	c.68G>T, c.74dup, c.958G>C, c.980-14A>G, c.1264A>G
Biotinidase deficiency	*BTD* (NM_000060)	c.98_104delinsTCC*, c.100G>A, c.235C>T*, c.511G>A*, c.528G>T*, c.755A>G*, c.1207T>G*, c.1330G>C*, c.1368A>C*, c.1466A>C, c.1595C>T*, c.1612C>T*
Bloom syndrome	*BLM* (NM_000057)	c.2207_2212delinsTAGATTC*
Canavan disease	*ASPA* (NM_000049)	c.433-2A>G, c.693C>A*, c.854A>C*, c.914C>A*
CAPN3-related limb-girdle muscular dystrophy	*CAPN3* (NM_000070)	c.140_142del, c.328C>T, c.550del*, c.598_612del, c.801+1G>A, c.946-1G>A, c.1469G>A*, c.1507_1511del, c.1525G>T, c.1795dup, c.1992+1G>T, c.2242C>T, c.2306G>A
Carbamoylphosphate synthetase I deficiency	*CPS1* (NM_001875)	c.130C>T, c.1529del, c.1631C>T, c.1760G>A, c.2148T>A, c.2359C>T, c.2945G>A, c.3037_3039del, c.3784C>T, c.4357C>T
Carnitine palmitoyltransferase I (CPT I) deficiency	*CPT1A* (NM_001876)	c.1364A>C, c.1436C>T, c.2129G>A*
Carnitine palmitoyltransferase II (CPT II) deficiency	*CPT2* (NM_000098)	c.110_111dup, c.149C>A*, c.338C>T*, c.359A>G*, c.370C>T*, c.452G>A*, c.520G>A, c.680C>T*, c.1145G>A, c.1239_1240del*, c.1507C>T, c.1646G>A*, c.1737del, c.1763C>G, c.1883A>C, c.1891C>T, c.1925_1937del
Cartilage-hair hypoplasia	*RMRP* (NR_003051)	n.71A>G*, n.263G>T*
CEP290-related retinal dystrophies	*CEP290* (NM_025114)	c.384_387del*, c.2991+1655A>G, c.4723A>T, c.5668G>T
Cerebral creatine deficiency syndrome	*GAMT* (NM_000156)	c.59G>C, c.133T>A, c.148A>C, c.299_311dup, c.220G>C, c.327G>A, c.407C>T

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cerebrotendinous xanthomatosis	*CYP27A1* (NM_000784)	c.379C>T, c.434G>A, c.646G>C, c.819del*, c.844+1G>A, c.845-1G>A, c.1183C>T, c.1214G>A, c.1263+1G>A, c.1420C>T, c.1421G>A, c.1435C>T
Choroideremia	*CHM* (NM_000390)	c.877C>T, c.1584_1587del, c.1609+2dup
CHRNE-related congenital myasthenic syndrome	*CHRNE* (NM_000080)	c.130dup, c.422C>T, c.721C>T, c.865C>T, c.917G>T, c.971del, c.1291G>C, c.1327del, c.1353dup, c.1429del
Chronic granulomatous disease	*CYBA* (NM_000101)	c.7C>T, c.58+4_58+7del, c.70G>A, c.166dup, c.269G>A, c.287+2T>C
Citrin deficiency	*SLC25A13* (NM_014251)	c.74C>A, c.127C>T, c.615+5G>A, c.852_855del, c.1064G>A, c.1078C>T, c.1177+1G>A, c.1399C>T*, c.1420G>A
Citrullinemia	*ASS1* (NM_000050)	c.421-2A>G, c.1168G>A*
CLN3-related neuronal ceroid lipofuscinosis	*CLN3* (NM_000086)	Deletion analysis of exon 7–8
CLN5-related neuronal ceroid lipofuscinosis	*CLN5* (NM_006493)	c.225G>A, c.1175_1176del*
CLN6-related neuronal ceroid lipofuscinosis	*CLN6* (NM_017882)	c.17G>C, c.139C>T, c.200T>C, c.214G>T, c.268_271dup, c.308G>A, c.368G>A, c.461_463del, c.510_512del, c.663C>G, c.794_796del
CLN8-related neuronal ceroid lipofuscinosis	*CLN8* (NM_018941)	c.70C>G, c.88G>C, c.610C>T, c.789G>C
Cohen syndrome	*VPS13B* (NM_017890)	c.3348_3349del
Combined malonic and methylmalonic aciduria	*ACSF3* (NM_174917)	c.593T>G, c.728C>T, c.1073C>T, c.1075G>A*, c.1394_1411del, c.1411C>T*, c.1412G>A, c.1567C>T, c.1672C>T*
Combined pituitary hormone deficiency 2	*PROP1* (NM_006261)	c.150del, c.217C>T, c.301_302del, c.349T>A, c.358C>T
Combined pituitary hormone deficiency 3	*LHX3* (NM_014564)	c.111del, c.148A>T, c.229C>T, c.347A>G, c.644C>T, c.687G>A
Congenital amegakaryocytic thrombocytopenia	*MPL* (NM_005373)	c.79+2T>A*, c.127C>T*, c.305G>C, c.556C>T, c.769C>T
Congenital disorder of glycosylation, type Ia	*PMM2* (NM_000303)	c.193G>T, c.338C>T, c.357C>A*, c.395T>C, c.415G>A*, c.422G>A*, c.470T>C*, c.484C>T, c.677C>G, c.691G>A, c.710C>T, c.722G>C
Congenital disorder of glycosylation, type Ib	*MPI* (NM_002435)	c.152T>C, c.305C>T*, c.413T>C, c.656G>A, c.884G>A
Congenital disorder of glycosylation, type Ic	*ALG6* (NM_013339)	c.257+5G>A, c.680G>A, c.897_899del, c.998C>T, c.1432T>C
Congenital hyperinsulinism	*ABCC8* (NM_000352)	c.560T>A, c.3989-9G>A*, c.4160_4162del
Congenital nephrotic syndrome, type 1	*NPHS1* (NM_004646)	c.121_122del, c.1096A>C, c.1481del, c.2335-1G>A, c.3325C>T, c.3478C>T
Congenital nephrotic syndrome, type 2	*NPHS2* (NM_014625)	c.353C>T, c.412C>T, c.413G>A*, c.467del, c.503G>A, c.868G>A
Corticosterone methyloxidase deficiency	*CYP11B2* (NM_000498)	c.104_108del, c.541C>T, c.554C>T, c.594A>C, c.763G>T*
CRB1-related retinal dystrophies	*CRB1* (NM_201253)	c.498_506del*, c.613_619del, c.1148G>A, c.1576C>T, c.2234C>T, c.2290C>T, c.2688T>A, c.2843G>A*, c.3299T>C, c.3307G>A, c.4121_4130del
CTSD-related neuronal ceroid lipofuscinosis	*CTSD* (NM_001909)	c.685T>A, c.764dup, c.1149G>C

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis	*CFTR* (NM_000492)	c.-9_14del, c.1A>C, c.1A>G, c.2T>A, c.2T>C, c.2T>G, c.3G>A, c.3G>T, c.11C>A*, c.14C>T, c.19G>T, c.40A>T, c.42del, c.43del, c.50del, c.50dup, c.53+1G>T, c.54-2A>G, c.54-1G>A, c.57G>A, c.79G>T*, c.88C>T, c.112_113del, c.114C>G, c.115C>T*, c.137C>A, c.156del, c.164+1G>A, c.164+1G>C, c.164+1G>T, c.164+2T>A, c.164+2T>C, c.164+2T>G, c.164+12T>C*, c.165-2A>G, c.165-1G>A, c.166G>A*, c.168del*, c.170G>A, c.171G>A, c.175dup, c.178G>T*, c.200C>T*, c.206T>A, c.217del, c.220C>T*, c.223C>T*, c.233del, c.233dup, c.234del, c.236G>A, c.248dup, c.254G>A*, c.259T>A*, c.262_263del*, c.263T>A, c.263T>G, c.271G>A, c.273+1G>A, c.273+3A>C*, c.274-2A>C, c.274-2A>G, c.274-1G>A*, c.274-1G>C, c.274-1G>T, c.274G>A*, c.274G>T*, c.292C>T*, c.302T>G, c.307G>T*, c.310del*, c.313del, c.319_326del, c.325_327delinsG, c.326_327del, c.328del*, c.328G>C*, c.330C>A, c.340A>T, c.343G>T, c.349C>T*, c.350G>A*, c.357del, c.366T>A, c.376G>A, c.380T>G, c.409_412del, c.413_415dup, c.415_416insGA, c.415_416insTA, c.416A>T, c.422C>A, c.424del, c.429del, c.433del, c.442del, c.443T>C*, c.445G>T, c.470T>G, c.476dup, c.476T>A, c.487A>G, c.489+1G>T*, c.489+2T>C*, c.489+2T>G, c.489+3A>G, c.490-2A>C, c.490-2A>G, c.490-1G>A, c.494del, c.522_526del, c.526del, c.531del*, c.532G>A*, c.543_546del, c.547C>A, c.550del, c.567C>A, c.575A>G, c.577G>A, c.577G>T*, c.578_579+5del*, c.579+1G>T*, c.579+3A>G*, c.579+5G>A*, c.580-1G>T*, c.595C>T, c.601del, c.606G>A, c.613C>T*, c.617T>G*, c.619C>T, c.647G>A, c.650_659del, c.653T>A, c.658C>T*, c.675T>A, c.680T>G, c.708del, c.714del, c.722_743del*, c.741C>G, c.743+1G>A*, c.743+1G>C, c.744-2A>G, c.761del, c.773del, c.803del*, c.805_806del, c.825C>G, c.828C>A, c.850dup, c.860dup, c.861_865del*, c.868C>T, c.870-2A>G, c.881_882del, c.912C>G, c.927del, c.935_937del*, c.938C>A, c.948del*, c.959T>A, c.980del, c.987del, c.988G>T*, c.1000C>T*, c.1005_1006insG, c.1007T>A*, c.1013C>T*, c.1021T>C*, c.1021_1022dup*, c.1029del*, c.1040G>A*, c.1040G>C*, c.1053_1054del, c.1055G>A*, c.1057C>T, c.1068G>A, c.1069del, c.1075_1079delinsAAAAA*, c.1075C>A*, c.1081del, c.1083del, c.1086T>A, c.1090T>C, c.1093_1094del, c.1116+1G>A*, c.1116+1G>C*, c.1117-1G>A, c.1130dup, c.1135G>T*, c.1152del*, c.1155_1156dup*, c.1155_1156insTA*, c.1159_1160del, c.1162_1168del, c.1177del, c.1192dup, c.1202G>A, c.1203G>A, c.1209+1G>A, 5T (c.1210-7_1210-6del)*, 9T (c.1210-13G>T)*, c.1240C>T, c.1327_1330dup*, c.1340del*, c.1364C>A*, c.1365_1366del*, c.1393-2A>G, c.1393-1G>A*, c.1397C>A, c.1397C>G, c.1400T>C, c.1418del*, c.1433_1434del, c.1435G>T, c.1438G>T*, c.1439del, c.1446dup, c.1456G>T, c.1466C>A*, c.1470_1471del, c.1475C>T*, c.1477C>T*, c.1477_1478del, c.1478A>T, c.1482_1483del, c.1487G>A, c.1510G>T, c.1519_1521del*, c.1521_1523del*, c.1528del, c.1545_1546del*, c.1550A>G*, c.1558G>A*, c.1558G>T*, c.1572C>A, c.1573C>T, c.1584+1G>A*, c.1584+2T>C, c.1585-8G>A*, c.1585-2A>G, c.1585-1G>A*, c.1588A>C, c.1606A>T, c.1611_1612del, c.1624G>T*, c.1628A>C, c.1642_1643del, c.1645A>C, c.1645_1648del, c.1646G>A*, c.1646G>T, c.1647T>G*, c.1648G>T, c.1650del, c.1651G>A, c.1652del, c.1652G>A*, c.1654C>T, c.1656del, c.1657C>T*, c.1670del, c.1673T>C, c.1674del, c.1675G>A*,

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492) (continued)	c.1679G>A*, c.1679G>C*, c.1679+1G>A*, c.1679+1G>C*, c.1679+2T>C, c.1680-886A>G, c.1680-877G>T*, c.1680-1G>A*, c.1682C>A*, c.1692del, c.1705T>G, c.1707T>A, c.1714_1715del, c.1721C>A*, c.1731C>A, c.1736A>G, c.1738del, c.1742dup, c.1753G>T*, c.1766+1G>A*, c.1766+1G>C*, c.1766+1G>T*, c.1766+2T>C, c.1766+3A>G*, c.1766+5G>T*, c.1786_1787del, c.1792A>T, c.1792_1798del, c.1800del, c.1820_1903del*, c.1826A>G, c.1865G>A*, c.1871_1878del, c.1900C>T, c.1909C>T, c.1911del, c.1919_1920del, c.1923_1931delinsA*, c.1966G>T*, c.1973_1985delinsAGAAA*, c.1976del, c.1981del, c.1986_1989del*, c.1990G>T, c.2012del, c.2017G>T, c.2036G>A, c.2044del, c.2051_2052delinsG*, c.2052del*, c.2052dup*, c.2053C>T, c.2053dup, c.2058_2061dup*, c.2062A>T, c.2065C>T, c.2074G>T, c.2083dup, c.2089dup, c.2108del, c.2125C>T*, c.2128A>T*, c.2143C>T, c.2156T>A, c.2158C>T*, c.2175dup*, c.2188G>T*, c.2195T>G*, c.2203del, c.2215del*, c.2233G>T, c.2249_2256del, c.2276_2277del, c.2277del, c.2324_2325del, c.2327C>G, c.2341C>T, c.2353C>T*, c.2374C>T, c.2380del, c.2393dup, c.2421A>G*, c.2423_2424dup, c.2435dup, c.2440C>T, c.2453del, c.2463_2464del, c.2464G>T, c.2467G>T, c.2472del, c.2479G>T, c.2488A>T, c.2490+1G>A*, c.2490+1G>T, c.2490+2T>C, c.2491-2A>G, c.2491G>T*, c.2502del, c.2502dup, c.2508del, c.2537G>A*, c.2538G>A, c.2547C>A, c.2551C>T, c.2555A>T, c.2566_2567insT, c.2583del, c.2589_2599del, c.2592_2593del, c.2600T>A, c.2601dup, c.2615del, c.2619+1G>A, c.2619+2T>A, c.2619+4dup, c.2620-26A>G*, c.2620-2A>G, c.2620-1G>C, c.2620-1G>T, c.2645G>A, c.2657+2_2657+3insA, c.2657+5G>A*, c.2658-2A>G, c.2658-1G>C, c.2658-1G>T, c.2668C>T*, c.2735C>A*, c.2737_2738insG*, c.2739T>A*, c.2763_2764dup, c.2776_2777del, c.2777del, c.2780T>C, c.2810dup*, c.2825del, c.2834C>T*, c.2836A>T, c.2856G>C, c.2859_2890del, c.2875del*, c.2876del, c.2896del, c.2908G>C, c.2908+2T>C, c.2909-1G>A, c.2909del, c.2924_2925del, c.2930C>T, c.2932A>T*, c.2988G>A*, c.2988+1G>A*, c.2988+2T>C, c.2989-2A>G, c.2989-1G>A, c.2997_3000del, c.2998del, c.3002_3003del, c.3007G>T, c.3017C>A, c.3021del, c.3022del, c.3039del, c.3039dup, c.3041A>G, c.3042_3043del, c.3061C>T, c.3067_3072del*, c.3068_3072del, c.3103C>T, c.3106del, c.3124C>T, c.3139_3139+1del, c.3140-26A>G*, c.3140-1G>A*, c.3154T>G*, c.3160C>G, c.3161del, c.3176T>G, c.3179A>C*, c.3181G>C*, c.3189G>A, c.3194T>C, c.3196C>T*, c.3197G>A, c.3199G>A*, c.3205G>A*, c.3208C>T*, c.3209G>A, c.3211C>T, c.3222T>A, c.3229_3230del, c.3230T>C, c.3263dup, c.3264del, c.3266G>A*, c.3276C>A*, c.3276C>G, c.3287del, c.3291del, c.3292T>C, c.3293G>A, c.3294del, c.3294G>A, c.3299A>C*, c.3302T>A*, c.3304A>T, c.3310G>T*, c.3315del, c.3364del*, c.3368-2A>G, c.3368-1G>A, c.3382A>T*, c.3430C>T, c.3434G>A, c.3435G>A, c.3445del, c.3454G>C*, c.3468+2T>C*, c.3469-2A>G, c.3472C>T*, c.3484C>T*, c.3486_3487del, c.3492dup, c.3497del, c.3528del*, c.3529A>T, c.3530del*, c.3534_3535insTCAA, c.3536_3539del, c.3540del, c.3546C>G, c.3556C>T, c.3587C>G, c.3592del, c.3600del, c.3605del, c.3611G>A, c.3612G>A*, c.3617C>A, c.3618_3619del, c.3659del*, c.3691del*, c.3712C>T, c.3717+1G>A*, c.3717+4A>G, c.3717+40A>G, c.3718-2477C>T*, c.3718-3T>G*, c.3718-1G>A*,

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492) (continued)	c.3731G>A*, c.3744del*, c.3747del, c.3752G>A*, c.3761T>G*, c.3763T>C, c.3764C>A*, c.3764C>G*, c.3764C>T*, c.3773dup*, c.3808del, c.3808G>A, c.3810T>A, c.3816_3817del, c.3822G>A, c.3829del, c.3835_3836del, c.3841C>T, c.3846G>A*, c.3848G>T, c.3855del, c.3871C>T, c.3873+1G>A, c.3873+2T>C*, c.3883del, c.3883_3884insG, c.3883_3886del, c.3908del*, c.3908dup, c.3909C>G*, c.3921T>A, c.3922G>T, c.3929G>A, c.3937C>T*, c.3947G>A, c.3963+2T>A, c.3964-1G>A, c.3976del, c.3985G>C, c.4025_4028dup, c.4039dup, c.4040_4041del, c.4042del, c.4046G>A*, c.4077_4080delinsAA*, c.4086dup, c.4111G>T, c.4139del, c.4140del*, c.4141T>C, c.4143C>A*, c.4144C>T*, c.4147dup, c.4168C>T*, c.4170del*, c.4197_4198del, c.4201dup, c.4201G>T, c.4231C>T, c.4234C>T, c.4242+1G>A, c.4242+1G>T, c.4243-2A>C, c.4243-1G>C, c.4251del, c.4252G>T, c.4297G>A, c.4300_4301insAG, c.4364C>G, c.4417G>T, c.4426C>T*, Deletion and duplication analysis of exon 1, exon 2, exon 3, exon 4, exon 5, exon 6, exon 7, exon 8, exon 9, exon 10, exon 11, exon 12, exon 13, exon 14, exon 15, exon 16, exon 17, exon 18, exon 19-20, exon 21, exon 22, exon 23, exon 24, exon 25, exon 26, exon 27
Cystinosis	*CTNS* (NM_004937)	c.18_21del, c.198_218del, c.283G>T, c.329G>T, c.413G>A, c.414G>A, c.416C>T, c.473T>C, c.506G>A, c.589G>A, c.613G>A*, c.853-3C>G, c.969C>G, c.1015G>A, Deletion analysis of exon 1–10
D-Bifunctional protein deficiency and Perrault syndrome	*HSD17B4* (NM_000414)	c.46G>A, c.101C>T, c.1369A>T*
DCLRE1C-related severe combined immunodeficiency	*DCLRE1C* (NM_001033855)	c.109+1G>T, c.241C>T*, c.362+1G>T
Diastrophic dysplasia	*SLC26A2* (NM_000112)	c.-26+2T>C, c.229A>C, c.532C>T*, c.764G>A, c.835C>T*, c.1020_1022del, c.1957T>A*
Dihydrolipoamide dehydrogenase deficiency	*DLD* (NM_000108)	c.104dup, c.140T>C, c.685G>T*, c.1123G>A*, c.1463C>T
Dihydropyrimidine dehydrogenase deficiency	*DPYD* (NM_000110)	c.299_302del*, c.557A>G*, c.1905+1G>A*, c.2846A>T
Duchenne muscular dystrophy	*DMD* (NM_004006)	Deletion and duplication analysis of exon 1, exon 2, exon 3, exon 4, exon 5, exon 6, exon 7, exon 8, exon 9, exon 10, exon 11, exon 12, exon 13, exon 14-15, exon 16, exon 17, exon 18, exon 19, exon 20, exon 21, exon 22, exon 23, exon 24, exon 25, exon 26, exon 27, exon 28, exon 29, exon 30, exon 31, exon 32, exon 33, exon 34, exon 35, exon 36, exon 37, exon 38, exon 39, exon 40, exon 41, exon 42, exon 43, exon 44, exon 45, exon 46, exon 47, exon 48, exon 49, exon 50, exon 51, exon 52, exon 53, exon 54, exon 55, exon 56, exon 57, exon 58, exon 59, exon 60, exon 61, exon 62, exon 63, exon 64, exon 65, exon 66, exon 67, exon 68, exon 69, exon 70, exon 71, exon 72, exon 73, exon 74, exon 75, exon 76, exon 77, exon 78, exon 79
Dyskeratosis congenita	*RTEL1* (NM_032957)	c.1548G>T, c.2288G>T, c.2941C>T, c.2992C>T, c.3724+139G>A*
Dystrophic epidermolysis bullosa	*COL7A1* (NM_000094)	c.425A>G, c.497dup, c.933C>A, c.1732C>T, c.2471dup, c.3840del, c.4039G>C, c.8393T>A, c.8524_8527+10del
Emery-Dreifuss muscular dystrophy	*EMD* (NM_000117)	c.1A>G, c.102C>G, c.123C>G, c.130C>T, c.184dup, c.187+1G>A, c.239_240del, c.251_255del, c.256C>T, c.266-2A>G, c.314_315del, c.315T>G, c.325G>T, c.359_362del, c.441C>A, c.450-2A>G, c.512C>A, c.547C>A, c.617_620del, c.650_654dup, c.677G>A

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Ethylmalonic encephalopathy	*ETHE1* (NM_014297)	c.487C>T, c.488G>A, c.505+1G>T
Fabry disease	*GLA* (NM_000169)	c.124A>C*, c.335G>A*, c.427G>A, c.427G>C, c.679C>T*
Fabry disease: pseudodeficiency allele	*GLA* (NM_000169)	c.937G>T*
Factor XI deficiency	*F11* (NM_000128)	c.166T>C*, c.316C>T, c.403G>T*, c.408C>A, c.438C>A*, c.730C>T, c.841C>T, c.901T>C*, c.1361T>A, c.1613C>T, c.1714_1716+11del, c.1716+1G>A
Familial dysautonomia	*ELP1* (NM_003640)	c.2087G>C*, c.2204+6T>C*, c.2741C>T
Familial Mediterranean fever	*MEFV* (NM_000243)	c.2040G>A*, c.2040G>C*, c.2080A>G*, c.2082G>A*, c.2177T>C*
FANCA-related Fanconi anemia	*FANCA* (NM_000135)	c.65G>A, c.295C>T, c.811C>T, c.862G>T, c.1115_1118del, c.1874G>C*, c.2546del, c.3720_3724del, c.3788_3790del*, Deletion analysis of exon 4–5, exon 10, exon 12–14, exon 23, exon 25–26, exon 28, exon 30, exon 32, exon 37, exon 39–41, exon 41–42, exon 43
FANCC-related Fanconi anemia	*FANCC* (NM_000136)	c.37C>T*, c.67del*, c.456+4A>T*, c.553C>T*, c.692_694del, c.1487T>G, c.1642C>T, c.1661T>C
FANCG-related Fanconi anemia	*FANCG* (NM_004629)	c.307+1G>C*, c.313G>T, c.637_643del, c.1066C>T, c.1077-2A>G, c.1183_1192del, c.1480+1G>C*, c.1795_1804del
FKRP-related muscular dystrophy-dystroglycanopathy	*FKRP* (NM_024301)	c.826C>A*, c.899T>C, c.919T>A, c.1073C>T*, c.1364C>A, c.1387A>G
Fragile X	*FMR1*	Trinucleotide repeat
Fructose intolerance	*ALDOB* (NM_000035)	c.448G>C*, c.524C>A*, c.1005C>G, Deletion analysis of exon 2–6
G6PD deficiency	*G6PD* (NM_000402)	c.221C>G, c.292G>A*, c.556G>A, c.577G>A, c.682C>T, c.1093G>A*, c.1450C>T*, c.1466G>C, c.1466G>T
Galactokinase deficiency	*GALK1* (NM_000154)	c.82C>A, c.94G>A, c.238G>T, c.766C>T, c.1031C>T, c.1144C>T
Galactosemia	*GALT* (NM_000155)	c.-119_-116del*, c.136_140del, c.221T>C*, c.253-2A>G*, c.292G>A*, c.404C>T*, c.413C>T*, c.425T>A*, c.443G>A*, c.505C>A, c.512T>C*, c.563A>G*, c.584T>C*, c.607G>A*, c.626A>G*, c.855G>T*, c.940A>G*, c.958G>A*, c.997C>G*, c.997C>T*, c.1018G>T, c.1030C>A*, c.1138T>C*, Deletion analysis of exon 1–11
Gaucher disease	*GBA* (NM_000157)	c.84dup*, c.115+1G>A*, c.1226A>G*, c.1297G>T*, c.1342G>C*, c.1343A>T*, c.1448T>C*, c.1504C>T*, c.1505G>A, c.1604G>A*, partial exon 9 deletion
Gitelman syndrome	*SLC12A3* (NM_000339)	c.179C>T*, c.1868T>C, c.1925+2T>A, c.2221G>A*, c.2883+1G>T*
GJB2-related nonsyndromic hearing loss and deafness	*GJB2* (NM_004004)	c.35del*, c.71G>A*, c.167del*, c.230G>A, c.235del*, c.269T>C*, c.358_360del, Deletion analysis of exon 2
GJB6-related nonsyndromic hearing loss and deafness	*GJB6* (NM_006783)	Deletion analysis of exon 1–3
Glutaric aciduria type I	*GCDH* (NM_000159)	c.262C>T, c.680G>C, c.877G>A, c.1198G>A*, c.1204C>T*, c.1240G>A*, c.1262C>T*
Glutathione synthetase deficiency	*GSS* (NM_000178)	c.-9+5G>A, c.129+1663A>G, c.656A>G, c.847C>T
Glycine encephalopathy	*AMT* (NM_000481)	c.125A>G, c.878-1G>A, c.959G>A
	GLDC (NM_000170)	c.1545G>C*, c.1691G>T
Glycogen storage disease Ia	*G6PC* (NM_000151)	c.79del, c.247C>T*, c.248G>A*, c.379_380dup*, c.562G>A*, c.562G>C*, c.648G>T*, c.724C>T, c.809G>T, c.980_982del, c.1039C>T*

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Glycogen storage disease Ib	*SLC37A4* (LRG_187)	c.352T>C, c.1243C>T, c.1099G>A, c.1042_1043del, c.1015G>T*
Glycogen storage disease II (Pompe disease)	*GAA* (NM_000152)	c.-32-13T>G*, c.525del*, c.1927G>A, c.1935C>A*, c.2560C>T*, Deletion analysis of exon 18
Glycogen storage disease II (Pompe disease): pseudodeficiency allele	*GAA* (NM_000152)	c.271G>A*, c.1726G>A*, c.2065G>A*
Glycogen storage disease III	*AGL* (NM_000642)	c.16C>T, c.18_19del*, c.1222C>T*, c.2590C>T, c.2681+1G>A, c.3682C>T, c.3965del, c.3980G>A, c.4456del
Glycogen storage disease IV	*GBE1* (NM_000158)	c.288del, c.671T>C, c.691+2T>C*, c.986A>G*, c.1544G>A, c.1571G>A
Glycogen storage disease type V (GSDV, McArdle disease)	*PYGM* (NM_005609)	c.148C>T*, c.613G>A, c.715_717del, c.2262del, c.2392T>C*
GM1 gangliosidosis and mucopolysaccharidosis IVB (MPS-IVB, Morquio syndrome)	*GLB1* (NM_000404)	c.176G>A, c.602G>A, c.1321G>A, c.1325G>A
GM1 gangliosidosis and mucopolysaccharidosis IVB (MPS-IVB, Morquio syndrome): Pseudodeficiency Allele	*GLB1* (NM_000404)	c.1783C>T
Hemoglobinopathy, Alpha thalassemia	*HBA1* (NM_000558)	Deletion and duplication analysis of exon 3
	HBA2 (NM_000517)	c.427T>C*, c.*94A>G*, Deletion and duplication analysis of exon 3
Hemoglobinopathy, Beta thalassemia	*HBB* (NM_000518)	c.-140C>T, c.-138C>T, c.-137C>G, c.-81A>G*, c.-80T>A, c.-79A>G*, c.-78A>C, c.-78A>G*, c.1A>G, c.2T>G*, c.17_18del, c.20del*, c.25_26del, c.26A>G, c.27dup, c.36del, c.45dup*, c.46del, c.48G>A, c.51del*, c.52A>T*, c.59A>G*, c.75T>A*, c.79G>T*, c.85dup*, c.92G>C, c.92+1G>A*, c.92+5G>C*, c.92+6T>C*, c.93-21G>A*, c.112del, c.114G>A, c.118C>T*, c.126_129del*, c.135del*, c.155del, c.203_204del, c.217dup, c.271G>T, c.287dup, c.315+1G>A*, c.316-197C>T*, c.316-106C>G*, c.316-3C>A, c.316-2A>C*, c.316-2A>G, c.316-1G>T, c.364G>A, c.383_385del, c.*113A>G*, Deletion and duplication analysis of exon 1–2, exon 3
Hemoglobinopathy, Hb C		c.19G>A
Hemoglobinopathy, Hb D		c.364G>C
Hemoglobinopathy, Hb E		c.79G>A*
Hemoglobinopathy, Hb S		c.20A>T*
Hemophilia B	*F9* (NM_000133)	c.-49T>A, c.127C>T, c.217G>A, c.223C>T, c.278-3A>G, c.316G>A, c.572G>A, c.677G>A, c.697G>A, c.1025C>T, c.1135C>T*, c.1151G>T, c.1328T>C
Hermansky-Pudlak syndrome 1	*HPS1* (NM_000195)	c.166_168del, c.288del, c.355del, c.391C>T, c.397G>T, c.398+5G>A, c.418del, c.532dup, c.716T>C, c.962del, c.1189del, c.1323dup, c.1472_1487dup, c.1691del, c.1744-2A>C, c.1749G>A, c.1996G>T, c.2003T>C
Hermansky-Pudlak syndrome 3	*HPS3* (NM_032383)	c.1163+1G>A*, c.1189C>T, c.1691+2T>G, c.2482-2A>G, c.2589+1G>C, Deletion analysis of exon 1
HMG-CoA lyase deficiency	*HMGCL* (NM_000191)	c.109G>T, c.122G>A*, c.206_207del
Holocarboxylase synthetase deficiency	*HLCS* (NM_000411)	c.710T>C, c.1522C>T, c.1648G>A, c.1741G>A
Homocystinuria	*CBS* (NM_000071)	c.572C>T*, c.833T>C*, c.919G>A*
Hydrolethalus syndrome	*HYLS1* (NM_145014)	c.632A>G*

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Hypophosphatasia	*ALPL* (NM_000478)	c.407G>A, c.571G>A*, c.979T>C, c.1133A>T*, c.1250A>G, c.1559del
Isovaleric acidemia	*IVD* (NM_002225.3)	c.157C>T, c.465+2T>C, c.941C>T*, c.1241G>A
Joubert syndrome 2	*TMEM216* (NM_001173990)	c.218G>T*, c.230G>C, c.253C>T*, c.341T>G, c.398T>G
Junctional epidermolysis bullosa	*LAMA3* (NM_000227)	c.1981C>T
	LAMB3 (NM_000228)	c.124C>T, c.727C>T, c.956_957ins77, c.1903C>T
	LAMC2 (NM_005562)	c.283C>T
Krabbe disease	*GALC* (NM_000153)	c.246A>G, c.334A>G*, c.683_694delinsCTC, c.857G>A*, c.1472del*, c.1586C>T*, c.2002A>C, Deletion analysis of 30Kb del (exon 11–17)
Krabbe disease: pseudodeficiency allele	*GALC* (NM_000153)	c.550C>T*, c.742G>A*, c.1685T>C*
LCHAD deficiency/mitochondrial trifunctional protein deficiency (MTP deficiency)	*HADHA* (NM_000182)	c.1528G>C*
Leber congenital amaurosis 13/ Retinitis pigmentosa 53	*RDH12* (NM_152443)	c.184C>T*, c.565C>T, c.677A>G, c.806_810del
Leigh syndrome due to mitochondrial complex III deficiency, Bjornstad syndrome, or GRACILE syndrome	*BCS1L* (NM_004328)	c.166C>T, c.232A>G*, c.550C>T
Leigh syndrome, French-Canadian type	*LRPPRC* (NM_133259)	c.1061C>T
Leukoencephalopathy with vanishing white matter	*EIF2B5* (NM_003907)	c.271A>G, c.318A>T, c.338G>A, c.406C>T, c.584G>A, c.925G>C, c.1157G>T
Lipoid adrenal hyperplasia	*STAR* (NM_000349)	c.64+1G>T, c.545G>T, c.559G>A, c.577C>T, c.650G>C, c.653C>T, c.749G>A, c.772C>T
Lipoprotein lipase deficiency	*LPL* (NM_000237)	c.264T>A, c.644G>A*, c.662T>C, c.701C>T, c.755T>C, c.808C>T, c.835C>G
Lysinuric protein intolerance	*SLC7A7* (NM_001126106)	c.161G>T, c.625+1G>C, c.726G>A*, c.895-2A>T*, c.1185_1188del, c.1228C>T*, c.1381_1384dup
Lysosomal acid lipase deficiency	*LIPA* (NM_000235)	c.260G>T, c.599T>C, c.894G>A, c.1024G>A*
Maple syrup urine disease, type Ia	*BCKDHA* (NM_000709)	c.1312T>A*
Maple syrup urine disease, type Ib	*BCKDHB* (NM_183050)	c.548G>C*, c.832G>A*, c.1114G>T
Maple syrup urine disease, type II	*DBT* (NM_001918)	c.670G>T, c.827T>G*, c.901C>T, Deletion analysis of exon 1–11
Meckel syndrome 1	*MKS1* (NM_017777)	c.392_393del, c.1048C>T, c.1408-34_1408-6del*
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	*ACADM* (NM_000016)	c.127G>A*, c.199T>C, c.985A>G*
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1* (NM_015166)	c.135dup, c.176G>A*, c.178-10T>A, c.278C>T
Metachromatic leukodystrophy	*ARSA* (NM_000487.5)	c.257G>A, c.293C>T, c.302G>A, c.465+1G>A*, c.542T>G, c.641C>T, c.769G>C, c.1210+1G>A, c.1232C>T, c.1283C>T*, c.1408_1418del
Metachromatic leukodystrophy: pseudodeficiency allele	*ARSA* (NM_000487.5)	c.1055A>G*, c.1178C>G*, c.*96A>G*
Metachromatic leukodystrophy: pseudodeficiency allele	*ARSA* (NM_000487.5)	c.1055A>G*, c.1178C>G*, c.*96A>G*
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC* (NM_015506)	c.271dup*, c.331C>T, c.347T>C*, c.394C>T*, c.440G>C, c.482G>A*, c.658_660del

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
MFSD8 related neuronal ceroid lipofuscinosis	*MFSD8* (NM_152778)	c.362A>G, c.754+2T>A, c.863+3_863+4insT, c.881C>A, c.929G>A, c.1235C>T
Microcephaly, postnatal progressive, with seizures and brain atrophy (MCPHSBA)	*MED17* (NM_004268)	c.1112T>C
Mitochondrial complex I deficiency	*NDUFAF5* (NM_024120)	c.477A>C, c.686T>C, c.749G>T
	NDUFS6 (NM_004553)	c.186+2T>A, c.344G>A, c.352C>T
MMAA-related methylmalonic acidemia	*MMAA* (NM_172250)	c.283C>T, c.433C>T, c.988C>T
MMAB-related methylmalonic acidemia	*MMAB* (NM_052845)	c.291-1G>A, c.556C>T, c.568C>T, c.571C>T, c.700C>T
Mucolipidosis II alpha/beta	*GNPTAB* (NM_024312)	c.242G>T, c.344_345del, c.569A>T, c.616_619del, c.1001G>A, c.1123C>T, c.1196C>T, c.1208T>C, c.1399del, c.1514G>A, c.1581del, c.1759C>T, c.2533C>T, c.2715+1G>A, c.3335+6T>G*, c.3443_3446del, c.3503_3504del*, c.3565C>T*, c.3613C>T
Mucolipidosis III gamma	*GNPTG* (NM_032520)	c.196C>T, c.316G>A, c.318-1G>C, c.333G>A, c.347_349del, c.379_391del, c.445del, c.610-2A>G, c.611del, c.619_620insT, c.639del, c.857C>T
Mucolipidosis IV	*MCOLN1* (NM_020533)	c.304C>T*, c.406-2A>G*, c.1207C>T, Deletion analysis of exon 1–7
Mucopolysaccharidosis II	*IDS* (NM_000202)	c.262C>T*, c.404A>G, c.514C>T, c.998C>T, c.1122C>T, c.1264T>G, c.1327C>T, c.1402C>T
Mucopolysaccharidosis IVA	*GALNS* (NM_000512)	c.337A>T, c.901G>T, c.1156C>T
Mucopolysaccharidosis type I (MPS-I)	*IDUA* (NM_000203)	c.152G>A, c.192C>A, c.208C>T, c.266G>A, c.613_617dup, c.979G>C, c.1037T>G, c.1205G>A*, c.1598C>G*
Mucopolysaccharidosis type I (MPS-I): pseudodeficiency allele	*IDUA* (NM_000203)	c.235G>A*, c.246C>G*, c.667G>A, c.898G>A, c.965T>A*
Mucopolysaccharidosis type IIIA (Sanfilippo A)	*SGSH* (NM_000199)	c.197C>G*, c.364G>A, c.449G>A, c.734G>A*, c.877C>T, c.892T>C, c.1080del*, c.1272_1282del
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU* (NM_000263)	c.245G>A, c.384-1G>A, c.503G>A, c.507_516del, c.889C>T, c.944dup, c.1558C>T*
Mucopolysaccharidosis type IIIC (Sanfilippo C)	*HGSNAT* (NM_152419)	c.234+1G>A, c.372-2A>G, c.410T>C, c.493+1G>A, c.525dup, c.848C>T, c.1030C>T*, c.1250+1G>A, c.1270G>A, c.1411G>A, c.1553C>T, c.1622C>T
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	*ARSB* (NM_000046)	c.629A>G, c.1143-8T>G, c.1143-1G>C
Mucopolysaccharidosis VII	*GUSB* (NM_000181)	c.526C>T, c.1069C>T, c.1222C>T, c.1244C>T
Mucopolysaccharidosis VII: pseudodeficiency allele	*GUSB* (NM_000181)	c.454G>A*
Multiple sulfatase deficiency	*SUMF1* (NM_182760)	c.463T>C*, c.739G>C, c.788G>T, c.836C>T*, c.1033C>T, c.1045C>T
Muscular dystrophy-dystroglycanopathy and autosomal recessive dilated cardiomyopathy	*FKTN* (NM_001079802)	c.139C>T, c.340G>A, c.346C>T, c.454dup, c.920G>A, c.1167dup*
Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3	*POMGNT1* (NM_017739)	c.1324C>T, c.1478C>G, c.1539+1G>A*, c.1666G>A*
MUT-related methylmalonic acidemia	*MMUT* (NM_000255)	c.91C>T, c.278G>A, c.322C>T, c.349G>T, c.643G>A, c.655A>T*, c.1106G>A*, c.1867G>A, c.2150G>T

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Nemaline myopathy	*NEB* (NM_004543)	c.3255+1G>A, c.13994G>T, c.17041A>C, c.17728C>T, Deletion analysis of exon 55
Niemann-Pick disease type C (NPC)	*NPC1* (NM_000271)	c.1211G>A*, c.1421C>T*, c.1553G>A, c.1628C>T, c.2072C>T, c.2324A>C, c.2621A>T*, c.2819C>A, c.2972_2973del*, c.3019C>G*, c.3104C>T, c.3160G>A, c.3182T>C*, c.3467A>G*, c.3557G>A*
	NPC2 (NM_006432)	c.27del, c.58G>T, c.115G>A*, c.190+5G>A, c.199T>C, c.332del, c.352G>T, c.358C>T, c.436C>T
Niemann-Pick disease, type A/B	*SMPD1* (NM_000543)	c.911T>C*, c.971C>T, c.996del*, c.1267C>T*, c.1426C>T*, c.1493G>A*, c.1493G>T*, c.1829_1831del*
Nijmegen breakage syndrome	*NBN* (NM_002485)	c.657_661del*, c.1089C>A
Nonaka myopathy	*GNE* (NM_005476)	c.527A>T, c.1003C>T, c.1132G>T*, c.1714G>C, c.1892C>T, c.2086G>A, c.2135T>C*
Pendred syndrome	*SLC26A4* (NM_000441)	c.-103T>C*, c.279del, c.412G>T, c.626G>T, c.707T>C*, c.716T>A, c.919-2A>G*, c.1001+1G>A, c.1003T>C, c.1115C>T, c.1151A>G, c.1246A>C, c.1334T>G, c.1540C>A, c.2168A>G*
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1* (NM_004035)	c.167dup, c.191del, c.190_192del, c.372_389del, c.442C>T, c.538+1G>A, c.904C>T, c.926A>G, c.928T>C, c.1276_1277del, c.1789_1792del, c.1872T>A
Peroxisome biogenesis disorders/ Zellweger syndrome spectrum	*PEX1* (NM_000466)	c.2T>C, c.569C>A, c.788_789del, c.2097dup*, c.2528G>A*, c.2614C>T, c.2730del, c.2916del
	PEX2 (NM_000318)	c.279_283del, c.339_345del, c.355C>T*, c.373C>T, c.669G>A, c.834_838del
	PEX6 (NM_000287)	c.402del, c.802_815del, c.1314_1321del, c.1688+1G>A, c.1715C>T*, c.1947del, c.2094+2T>C
Phenylketonuria	*PAH* (NM_000277)	c.117C>G*, c.143T>C, c.194T>C*, c.473G>A*, c.782G>A*, c.814G>T, c.838G>A, c.842C>T*, c.896T>G*, c.1045T>C, c.1066-11G>A*, c.1222C>T*, c.1223G>A*, c.1241A>G*, c.1315+1G>A*
POLG-Related Disorders	*POLG* (NM_002693)	c.830A>T, c.1399G>A*, c.1491G>C, c.2243G>C*, c.2542G>A*, c.3139C>T
Polyglandular autoimmune syndrome type 1	*AIRE* (NM_000383)	c.247A>G, c.254A>G, c.415C>T, c.489dup, c.769C>T*, c.967_979del*
Pontocerebellar hypoplasia type 2 (PCH2)	*SEPSECS* (NM_016955)	c.715G>A*, c.1001A>G*
PPT1 related neuronal ceroid lipofuscinosis	*PPT1* (NM_000310)	c.29T>A, c.223A>C, c.364A>T, c.451C>T, c.656T>A
Primary ciliary dyskinesia	*CFAP298* (NM_021254)	c.735C>G
Primary ciliary dyskinesia 3	*DNAH5* (NM_001369)	c.1730G>C, c.10384C>T, c.10815del, c.13338+5G>A, c.13486C>T
Primary ciliary dyskinesia 9	*DNAI2* (NM_023036)	c.346-3T>G, c.739C>T, c.787C>T, c.1304G>A*, c.1357dup, c.1494+1G>A
Primary hyperoxaluria type 1 (PH1)	*AGXT* (NM_000030)	c.454T>A*, c.508G>A*, c.731T>C*, c.822G>C
Primary hyperoxaluria type 2 (PH2)	*GRHPR* (NM_012203)	c.103del*, c.404+3_404+6del
Primary hyperoxaluria type 3 (PH3)	*HOGA1* (NM_138413)	c.700+5G>T*, c.860G>T, c.944_946del*
Primary systemic carnitine deficiency	*SLC22A5* (NM_003060)	c.67_69del, c.95A>G, c.136C>T*, c.632A>G, c.760C>T*, c.844C>T*, c.865C>T, c.1009del, c.1400C>G*

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Progressive familial intrahepatic cholestasis	*ABCB11* (NM_003742)	c.379del, c.611+1G>A, c.890A>G, c.1146del, c.1295G>C, c.1445A>G, c.1558A>T, c.1723C>T, c.1941del, c.2012-8T>G, c.2178+1G>A, c.2343+1G>T, c.2783_2787dup, c.3169C>T, c.3213+1del, c.3268C>T, c.3767dup, c.3904G>T
Propionic acidemia	*PCCA* (NM_000282)	c.491T>C, c.862A>G, c.937C>T, c.1196G>A, c.1676G>T*, c.1685C>G, Deletion analysis of exon 3–4
	PCCB (NM_000532)	c.280G>T, c.335G>A, c.457G>C, c.502G>A, c.872G>A, c.1173dup, c.1218_1231delinsTAGAGCACAGGA*, c.1228C>T, c.1283C>T, c.1304A>G, c.1352C>T, c.1495C>T, c.1534C>T, c.1556T>C
Pseudocholinesterase deficiency	*BCHE* (NM_000055)	c.293A>G*, c.884A>G, c.1004T>C, c.1253G>T*
Pycnodysostosis	*CTSK* (NM_000396)	c.136C>T, c.236G>A, c.721C>T, c.926T>C, c.990A>G
RAPSN-related congenital myasthenic syndrome	*RAPSN* (NM_005055)	c.41T>C*, c.133G>A, c.264C>A*, c.490C>T, c.807C>A, c.848T>C
Retinitis pigmentosa 25	*EYS* (NM_001142800)	c.408_423del, c.4350_4356del, c.4957dup, c.7919G>A, c.8805C>A, c.9405T>A
Retinitis pigmentosa 28	*FAM161A* (NM_001201543)	c.1309A>T, c.1355_1356del*, c.1567C>T*, c.1786C>T
Retinitis pigmentosa 59 and congenital disorder of glycosylation, type 1bb	*DHDDS* (NM_024887)	c.124A>G*
Retinoschisis	*RS1* (NM_000330)	c.216G>C, c.221G>T, c.325G>C
Rhizomelic chondrodysplasia punctata type 3 (RCDP3)	*AGPS* (NM_003659)	c.545G>A, c.926C>T, c.1256G>A, c.1406T>C, c.1411G>A, c.1703C>T
Rhizomelic chondrodysplasia punctata, type 1	*PEX7* (NM_000288)	c.40A>C, c.120C>G, c.345T>G, c.370_396del, c.649G>A, c.653C>T*, c.875T>A*, c.903+1G>C
Roberts syndrome	*ESCO2* (NM_001017420)	c.252_253del, c.294_297del, c.308_309del, c.417dup, c.505C>T, c.604C>T, c.745_746del, c.751dup, c.760dup, c.764_765del, c.879_880del, c.955+2_955+5del, c.1111dup, c.1131+1G>A, c.1132-7A>G, c.1263+1G>C, c.1269G>A, c.1354-18G>A, c.1461_1462del, c.1597dup, c.1615T>G, c.1674-2A>G
RPE65-related retinal dystrophies	*RPE65* (NM_000329)	c.65T>C, c.95-2A>T*, c.271C>T, c.304G>T, c.394G>A*, c.615_616del, c.700C>T, c.907A>T, c.1067del, c.1102T>C, c.1543C>T
Salla disease and infantile free sialic acid storage disease (ISSD)	*SLC17A5* (NM_012434)	c.115C>T, c.406A>G, c.802_816del, c.1007_1008del
Sandhoff disease	*HEXB* (NM_000521)	c.115del, c.445+1G>A, c.850C>T, c.1509-26G>A, c.1514G>A, c.1597C>T, Deletion analysis of exon 1–5
Segawa syndrome	*TH* (NM_000360)	c.-70G>A, c.364C>T, c.605G>A*, c.614T>C, c.646G>A, c.1141C>A, c.1388C>T, c.1400A>G
SGCA-related limb girdle muscular dystrophy	*SGCA* (NM_000023)	c.229C>T*, c.739G>A
SGCB-related limb-girdle muscular dystrophy	*SGCB* (NM_000232)	c.272G>C, c.341C>T, c.452C>G
SGCG-related limb girdle muscular dystrophy	*SGCG* (NM_000231)	c.87dup, c.525del, c.787G>A, c.848G>A
Sjogren-Larsson syndrome	*ALDH3A2* (NM_000382)	c.943C>T*, c.1094C>T, c.1108-1G>C, c.1297_1298del*
Smith-Lemli-Opitz syndrome	*DHCR7* (NM_001360)	c.1A>G, c.278C>T*, c.356A>T, c.452G>A, c.470T>C*, c.506C>T, c.724C>T*, c.725G>A*, c.744G>T, c.906C>G*, c.964-1G>C*, c.976G>T*, c.1055G>A, c.1210C>T*, c.1228G>A*, c.1342G>A

Targeted Variants Interrogated by Expanded Carrier Screen Panels (continued)

Associated Phenotype	Gene (Transcript)	Variants
Spastic ataxia, Charlevoix-Saguenay type	*SACS* (NM_014363)	c.7504C>T, c.8393C>A*, c.8844del
Spinal muscular atrophy	*SMN1*	Deletion analysis of exon 7–8
	SMN1 (NM_000344)	c.*3+80T>G*
	SMN2	Deletion and duplication analysis of exon 7–8 in cases with 0 copies of SMN1
Spondylothoracic dysostosis	*MESP2* (NM_001039958)	c.307G>T, c.373C>G, c.500_503dup
STRC-related nonsyndromic hearing loss	*STRC* (NM_153700)	Deletion analysis of exon 1–29
Tay-Sachs disease	*HEXA* (NM_000520)	c.508C>T*, c.533G>A*, c.749G>A, c.805G>A*, c.805+1G>A, c.1073+1G>A*, c.1274_1277dup*, c.1421+1G>C*, Deletion analysis of exon 1
Tay-Sachs disease: pseudodeficiency allele	*HEXA* (NM_000520)	c.508C>T*, c.533G>A*, c.749G>A, c.805G>A*, c.805+1G>A, c.1073+1G>A*, c.1274_1277dup*, c.1421+1G>C*, Deletion analysis of exon 1
Tay-Sachs disease: pseudodeficiency allele	*HEXA* (NM_000520)	c.739C>T*, c.745C>T
TPP1-related neuronal ceroid lipofuscinosis	*TPP1* (NM_000391)	c.229G>A, c.311T>A, c.509-1G>C*, c.622C>T*, c.851G>T, c.1027G>A, c.1093T>C, c.1266G>C, c.1340G>A, c.1678_1679del
Tyrosinemia, type I	*FAH* (NM_000137)	c.192G>T, c.456G>A, c.554-1G>T, c.698A>T, c.707-1G>A, c.782C>T*, c.786G>A, c.1062+5G>A*, c.1069G>T*
Usher syndrome, type 1B	*MYO7A* (NM_000260)	c.93C>A*, c.448C>T, c.494C>T, c.635G>A, c.700C>T, c.1797G>A, c.1996C>T, c.2005C>T, c.2476G>A, c.3508G>A, c.3719G>A, c.3764del, c.5573T>C, c.5648G>A, c.5886_5888del, c.5945G>A, c.6070C>T
Usher syndrome, type 1C	*USH1C* (NM_005709)	c.91C>T, c.308G>A, c.496+1G>A
Usher syndrome, type 1D	*CDH23* (NM_022124)	c.336+1G>A, c.902G>A, c.2289+1G>A, c.3880C>T, c.4488G>C, c.5237G>A*, c.6050-9G>A, c.8230G>A
Usher syndrome, type 1F	*PCDH15* (NM_033056)	c.7C>T, c.733C>T*
Usher syndrome, type 2A	*USH2A* (NM_206933)	c.920_923dup, c.1256G>T, c.2209C>T, c.2299del*, c.2898del, c.4338_4339del, c.12067-2A>G*
Usher syndrome, type 3A	*CLRN1* (NM_174878)	c.144T>G*, c.359T>A, c.449T>C, c.528T>G*
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	*ACADVL* (NM_000018)	c.848T>C*
Wilson disease	*ATP7B* (NM_000053)	c.1934T>G*, c.2333G>A*, c.2333G>T*, c.2972C>T*, c.3207C>A*, c.3402del*

* Previously detected in a known positive sample