GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
ABCA4	Stargardt disease 1 including Cone-rod dystrophy 3	NM_000350.3:c.6449G>A, NM_000350.3:c.6394G>T, NM_000350.3:c.6320G>A, NM_000350.3:c.6179T>G, NM_000350.3:c.6148G>C, NM_000350.3:c.6118C>T, NM_000350.3:c.6089G>A, NM_000350.3:c.5912T>G, NM_000350.3:c.5882G>A, NM_000350.3:c.5881G>A, NM_000350.3:c.5819T>C, NM_000350.3:c.5714+5G>A, NM_000350.3:c.5512delC, NM_000350.3:c.5461-10T>C, NM_000350.3:c.5338C>G, NM_000350.3:c.4793C>A, NM_000350.3:c.4469G>A, NM_000350.3:c.4457C>T, NM_000350.3:c.4429C>T, NM_000350.3:c.4139C>T, NM_000350.3:c.3540_3555delGTCTAAGGGTTTCTCC, NM_000350.3:c.3364G>A, NM_000350.3:c.3322C>T, NM_000350.3:c.3210_3211dupGT, NM_000350.3:c.3106G>A, NM_000350.3:c.3083C>T, NM_000350.3:c.2971G>C, NM_000350.3:c.2791G>A, NM_000350.3:c.2690C>T, NM_000350.3:c.2616_2617delCT, NM_000350.3:c.2588G>C, NM_000350.3:c.2300T>A, NM_000350.3:c.2160+1G>T, NM_000350.3:c.1964T>G, NM_000350.3:c.1938-1G>A, NM_000350.3:c.1848delA, NM_000350.3:c.1804C>T, NM_000350.3:c.1771delT, NM_000350.3:c.1755delA, NM_000350.3:c.1715G>A, NM_000350.3:c.1622T>C, NM_000350.3:c.1225delA, NM_000350.3:c.1222C>T, NM_000350.3:c.1018T>G, NM_000350.3:c.763C>T, NM_000350.3:c.661G>A, NM_000350.3:c.634C>T, NM_000350.3:c.286A>G, NM_000350.3:c.67-2A>G, NM_000350.3:c.52C>T
ACAD9	Mitochondrial complex I deficiency	NM_014049.5:c.23delT, NM_014049.5:c.130T>A, NM_014049.5:c.358delT, NM_014049.5:c.453+1G>A, NM_014049.5:c.797G>A, NM_014049.5:c.976G>C, NM_014049.5:c.1240C>T, NM_014049.5:c.1249C>T, NM_014049.5:c.1594C>T
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	NM_000016.6:c.127G>A, NM_000016.6:c.250C>T, NM_000016.6:c.287-2A>G, NM_000016.6:c.362C>T, NM_000016.6:c.446_449delTGAC, NM_000016.6:c.447G>A, NM_000016.6:c.447G>T, NM_000016.6:c.616C>T, NM_000016.6:c.617G>A, NM_000016.6:c.683C>A, NM_000016.6:c.734C>T, NM_000016.6:c.797A>G, NM_000016.6:c.799G>A, NM_000016.6:c.815_827delTTGCAATGGGAGC, NM_000016.6:c.890A>G, NM_000016.6:c.984delG, NM_000016.6:c.985A>G, NM_000016.6:c.1100_1103delAGTT
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	NM_000017.4:c.136C>T, NM_000017.4:c.314T>A, NM_000017.4:c.319C>T, NM_000017.4:c.417G>C, NM_000017.4:c.529T>C, NM_000017.4:c.561_568delCAATGCCT, NM_000017.4:c.826G>A, NM_000017.4:c.1095G>T, NM_000017.4:c.1108A>G, NM_000017.4:c.1147C>T
ACADSB	2-methylbutyrylglycinuria	NM_001609.4:c.303+1G>A, NM_001609.4:c.443C>T, NM_001609.4:c.621G>A, NM_001609.4:c.763C>T, NM_001609.4:c.1159G>A
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	NM_000018.4:c.278-1G>A, NM_000018.4:c.295_296delAC, NM_000018.4:c.343-1delG, NM_000018.4:c.400C>T, NM_000018.4:c.477+1G>C, NM_000018.4:c.520G>A, NM_000018.4:c.685C>T, NM_000018.4:c.739A>C, NM_000018.4:c.753-2A>C, NM_000018.4:c.848T>C, NM_000018.4:c.890_892delAGA, NM_000018.4:c.1096C>T, NM_000018.4:c.1097G>A, NM_000018.4:c.1106T>C, NM_000018.4:c.1139_1141delAGG, NM_000018.4:c.1182+1G>A, NM_000018.4:c.1357C>T, NM_000018.4:c.1360G>A, NM_000018.4:c.1373_1374insC, NM_000018.4:c.1385_1386insG, NM_000018.4:c.1406G>A, NM_000018.4:c.1468G>C, NM_000018.4:c.1532+1G>A, NM_000018.4:c.1837C>T, NM_000018.4:c.1843C>T, NM_000018.4:c.1844G>A, NM_000018.4:c.1882delC
ACAT1	Beta-Ketothiolase Deficiency	NM_000019.4:c.2T>A, NM_000019.4:c.409_416delTCTCAAAG, NM_000019.4:c.547G>A, NM_000019.4:c.622C>T, NM_000019.4:c.904delA, NM_000019.4:c.1031_1033delAAG, NM_000019.4:c.1082_1083insA, NM_000019.4:c.1136G>T, NM_000019.4:c.1138G>A
ACOX1	Acyl-CoA Oxidase I Deficiency	NM_004035.7:c.832A>G, NM_004035.7:c.591delG, NM_004035.7:c.532G>T
ADA	Adenosine deaminase deficiency	NM_000022.4:c.986C>T, NM_000022.4:c.956_960delAAGAG, NM_000022.4:c.890C>A, NM_000022.4:c.872C>T, NM_000022.4:c.632G>A, NM_000022.4:c.320T>C, NM_000022.4:c.247G>A, NM_000022.4:c.226C>T
ADAMTS2	Ehlers-Danlos syndrome, type VII-C	NM_014244.5:c.2384G>A
AGA	Aspartylglucosaminuria	NM_000027.4:c.904G>A, NM_000027.4:c.800dupT, NM_000027.4:c.755G>A, NM_000027.4:c.488G>C, NM_000027.4:c.302C>T, NM_000027.4:c.214T>C
AGL	Glycogen storage disease IIIa/IIIb (Cori or Forbes disease)	NM_000642.3:c.17_18delAG, NM_000642.3:c.16C>T, NM_000642.3:c.112A>G, NM_000642.3:c.294-2A>T, NM_000642.3:c.1222C>T, NM_000642.3:c.1481G>A, NM_000642.3:c.1485delT, NM_000642.3:c.1783C>T, NM_000642.3:c.1999delC, NM_000642.3:c.2039G>A, NM_000642.3:c.2590C>T, NM_000642.3:c.3214_3215delGA, NM_000642.3:c.3980G>A, NM_000642.3:c.4260-12A>G, NM_000642.3:c.4260-1G>T, NM_000642.3:c.4342G>C, NM_000642.3:c.4454delT, NM_000642.3:c.4528_4529insA
AGPS	Rhizomelic Chondrodysplasia Punctata, Type 3	NM_003659.4:c.926C>T, NM_003659.4:c.1256G>A, NM_003659.4:c.1406T>C, NM_003659.4:c.1703C>T
AGXT	Hyperoxaluria, primary, type 1	NM_000030.3:c.25_26insC, NM_000030.3:c.32C>A, NM_000030.3:c.121G>A, NM_000030.3:c.166-2A>G, NM_000030.3:c.245G>A, NM_000030.3:c.248A>G, NM_000030.3:c.322T>C, NM_000030.3:c.454T>A, NM_000030.3:c.466G>A, NM_000030.3:c.508G>A, NM_000030.3:c.560C>T, NM_000030.3:c.590G>A, NM_000030.3:c.613T>C, NM_000030.3:c.697C>T, NM_000030.3:c.698G>A, NM_000030.3:c.731T>C, NM_000030.3:c.738G>A, NM_000030.3:c.836T>C, NM_000030.3:c.860G>A

GENESeeker

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
ALDOB	Hereditary fructose Intolerance	NM_000035.4:c.1067C>A, NM_000035.4:c.1027T>C, NM_000035.4:c.1013C>T, NM_000035.4:c.1005C>G, NM_000035.4:c.720C>A, NM_000035.4:c.612T>A, NM_000035.4:c.524C>A, NM_000035.4:c.448G>C, NM_000035.4:c.442T>C, NM_000035.4:c.360_363delCAAA, NM_000035.4:c.178C>T, NM_000035.4:c.136A>T, NM_000035.4:c.113-1_115delGGTA, NM_000035.4:c.10C>T, NM_000035.4:c.2T>C
ALG6	Congenital Disorder of Glycosylation, Type 1C	NM_013339.4:c.53G>A, NM_013339.4:c.316C>T, NM_013339.4:c.482A>G, NM_013339.4:c.495-3C>G, NM_013339.4:c.893_895delTAA, NM_013339.4:c.998C>T, NM_013339.4:c.1432T>C
ALMS1	Alstrom syndrome	NM_001378454.1:c.888_904delTCAGCACCCGCTTATAG, NM_001378454.1:c.1571_1573delCTCinsT, NM_001378454.1:c.8161C>T, NM_001378454.1:c.8380C>T, NM_001378454.1:c.9908-1G>A, NM_001378454.1:c.10576_10577delAT, NM_001378454.1:c.10772delC, NM_001378454.1:c.11311_11314delAGAG, NM_001378454.1:c.11446C>T, NM_001378454.1:c.11448_11449insA, NM_001378454.1:c.11607_11608delCT, NM_001378454.1:c.11613_11614delCT, NM_001378454.1:c.12436C>T, NM_001378454.1:c.12442C>T
ALPL	Hypophosphatasia, infantile	NM_000478.6:c.98C>T, NM_000478.6:c.211C>T, NM_000478.6:c.212G>C, NM_000478.6:c.323C>T, NM_000478.6:c.346G>A, NM_000478.6:c.407G>A, NM_000478.6:c.526G>A, NM_000478.6:c.535G>A, NM_000478.6:c.571G>A, NM_000478.6:c.620A>C, NM_000478.6:c.814C>T, NM_000478.6:c.881A>C, NM_000478.6:c.892G>A, NM_000478.6:c.1001G>A, NM_000478.6:c.1133A>T, NM_000478.6:c.1250A>G, NM_000478.6:c.1306T>C, NM_000478.6:c.1366G>A, NM_000478.6:c.1574delG
AMT	Glycine encephalopathy (AMT-related)	NM_000481.4:c.959G>A, NM_000481.4:c.826G>C, NM_000481.4:c.806G>A, NM_000481.4:c.574C>T, NM_000481.4:c.259-1G>C, NM_000481.4:c.139G>A, NM_000481.4:c.125A>G
AR	Androgen insensitivity syndrome, X-Linked	NM_000044.6:c.340C>T, NM_000044.6:c.1769-11T>A, NM_000044.6:c.1771A>T, NM_000044.6:c.1937C>A, NM_000044.6:c.2323C>T, NM_000044.6:c.2391G>A, NM_000044.6:c.2395C>G, NM_000044.6:c.2567G>A, NM_000044.6:c.2650A>T
ARG1	Argininemia	NM_000045.4:c.32T>C, NM_000045.4:c.61C>T, NM_000045.4:c.365G>A, NM_000045.4:c.413G>T, NM_000045.4:c.703G>C, NM_000045.4:c.869C>G, NM_000045.4:c.871C>T
ARSA	Metachromatic leukodystrophy	NM_000487.6:c.1408_1418delGCAGCTGTGAC, NM_000487.6:c.1401_1411delGTTAGACGCAG, NM_000487.6:c.1283C>T, NM_000487.6:c.1241delC, NM_000487.6:c.1232C>T, NM_000487.6:c.1210+1G>A, NM_000487.6:c.1175G>A, NM_000487.6:c.1174C>T, NM_000487.6:c.1150G>A, NM_000487.6:c.1125_1126delCT, NM_000487.6:c.1108-2A>G, NM_000487.6:c.991G>T, NM_000487.6:c.986C>T, NM_000487.6:c.979G>A, NM_000487.6:c.938G>A, NM_000487.6:c.937C>T, NM_000487.6:c.931G>A, NM_000487.6:c.899T>C, NM_000487.6:c.883G>A, NM_000487.6:c.869G>T, NM_000487.6:c.869G>A, NM_000487.6:c.854+1G>A, NM_000487.6:c.827C>T, NM_000487.6:c.763G>A, NM_000487.6:c.739G>A, NM_000487.6:c.737G>A, NM_000487.6:c.641C>T, NM_000487.6:c.583delT, NM_000487.6:c.582delC, NM_000487.6:c.542dupT, NM_000487.6:c.542T>G, NM_000487.6:c.465+1G>A, NM_000487.6:c.346C>T, NM_000487.6:c.302G>A, NM_000487.6:c.293C>T, NM_000487.6:c.257G>A, NM_000487.6:c.195delC, NM_000487.6:c.34delG
ARSB	Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	NM_000046.5:c.1562G>A, NM_000046.5:c.1438dupG, NM_000046.5:c.1366C>T, NM_000046.5:c.1214G>A, NM_000046.5:c.1178A>C, NM_000046.5:c.1161dupC, NM_000046.5:c.1143-1G>C, NM_000046.5:c.1143-8T>G, NM_000046.5:c.979C>T, NM_000046.5:c.971G>T, NM_000046.5:c.944G>A, NM_000046.5:c.937C>G, NM_000046.5:c.921delA, NM_000046.5:c.753C>G, NM_000046.5:c.707T>C, NM_000046.5:c.629A>G, NM_000046.5:c.589C>T, NM_000046.5:c.571C>T, NM_000046.5:c.427delG, NM_000046.5:c.410G>T, NM_000046.5:c.389C>T, NM_000046.5:c.349T>C
ASL	Argininosuccinic aciduria	NM_000048.4:c.35G>A, NM_000048.4:c.337C>T, NM_000048.4:c.346C>T, NM_000048.4:c.392C>T, NM_000048.4:c.437G>A, NM_000048.4:c.446+1G>A, NM_000048.4:c.505T>C, NM_000048.4:c.525-2A>T, NM_000048.4:c.532G>A, NM_000048.4:c.539T>G, NM_000048.4:c.544C>T, NM_000048.4:c.557G>A, NM_000048.4:c.578G>A, NM_000048.4:c.602+1G>A, NM_000048.4:c.857A>G, NM_000048.4:c.925G>A, NM_000048.4:c.1043_1055delGCGTCATCTCTAC, NM_000048.4:c.1060C>T, NM_000048.4:c.1135C>T, NM_000048.4:c.1144-2A>G, NM_000048.4:c.1153C>T, NM_000048.4:c.1255_1256delCT, NM_000048.4:c.1366_1367insG, NM_000048.4:c.1366C>T
ASPA	Canavan disease	NM_000049.4:c.212G>A, NM_000049.4:c.433-2A>G, NM_000049.4:c.654C>A, NM_000049.4:c.693C>A, NM_000049.4:c.838C>T, NM_000049.4:c.854A>C, NM_000049.4:c.863A>G, NM_000049.4:c.914C>A
ASS1	Citrullinemia type I	NM_054012.4:c.40G>A, NM_054012.4:c.53C>T, NM_054012.4:c.256C>T, NM_054012.4:c.257G>A, NM_054012.4:c.323G>T, NM_054012.4:c.349G>A, NM_054012.4:c.380G>A, NM_054012.4:c.421-2A>G, NM_054012.4:c.470G>A, NM_054012.4:c.496-2A>G, NM_054012.4:c.535T>C, NM_054012.4:c.539G>A, NM_054012.4:c.571G>A, NM_054012.4:c.787G>A, NM_054012.4:c.793C>T, NM_054012.4:c.794G>A, NM_054012.4:c.805G>A, NM_054012.4:c.814C>T, NM_054012.4:c.835C>T, NM_054012.4:c.836G>A, NM_054012.4:c.910C>T, NM_054012.4:c.919C>T, NM_054012.4:c.928A>C, NM_054012.4:c.970G>A,

GENESeeker

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_054012.4:c.970+5G>A, NM_054012.4:c.1085G>T, NM_054012.4:c.1087C>T, NM_054012.4:c.1088G>A, NM_054012.4:c.1168G>A, NM_054012.4:c.1194-1G>C
ATP7A	Menkes Syndrome, X-Linked	NM_000052.7:c.1639C>T, NM_000052.7:c.1972_1973insTGTT, NM_000052.7:c.2531G>A, NM_000052.7:c.2938C>T, NM_000052.7:c.2981C>T, NM_000052.7:c.3255_3256delAC, NM_000052.7:c.3294+2T>G, NM_000052.7:c.3911A>G, NM_000052.7:c.3914_3920delACTCCCC, NM_000052.7:c.3931A>G
ATP7B	Wilson disease	NM_000053.4:c.4301C>T, NM_000053.4:c.4135C>T, NM_000053.4:c.4088C>T, NM_000053.4:c.4058G>A, NM_000053.4:c.3990_3993delTTAT, NM_000053.4:c.3955C>T, NM_000053.4:c.3809A>G, NM_000053.4:c.3796G>A, NM_000053.4:c.3694A>C, NM_000053.4:c.3688A>G, NM_000053.4:c.3359T>A, NM_000053.4:c.3207C>A, NM_000053.4:c.3101A>G, NM_000053.4:c.3083delA, NM_000053.4:c.2975C>T, NM_000053.4:c.2972C>T, NM_000053.4:c.2930C>T, NM_000053.4:c.2906G>A, NM_000053.4:c.2807T>A, NM_000053.4:c.2804C>T, NM_000053.4:c.2795C>A, NM_000053.4:c.2762G>A, NM_000053.4:c.2755C>T, NM_000053.4:c.2755C>G, NM_000053.4:c.2621C>T, NM_000053.4:c.2605G>A, NM_000053.4:c.2532delA, NM_000053.4:c.2356-2A>G, NM_000053.4:c.2305A>G, NM_000053.4:c.2297C>G, NM_000053.4:c.2267C>T, NM_000053.4:c.2123T>C, NM_000053.4:c.2071G>A, NM_000053.4:c.1934T>G, NM_000053.4:c.1922T>C, NM_000053.4:c.1846C>T, NM_000053.4:c.1745_1746delTA, NM_000053.4:c.1512dupT, NM_000053.4:c.1285+5G>T, NM_000053.4:c.1145_1151delCCCAACT, NM_000053.4:c.915T>A, NM_000053.4:c.562C>T, NM_000053.4:c.98T>C, NM_000053.4:c.19_20delCA
BCKDHA	Maple syrup urine disease, type Ia	NM_000709.4:c.14delT, NM_000709.4:c.632C>T, NM_000709.4:c.659C>T, NM_000709.4:c.740_741insT, NM_000709.4:c.761C>A, NM_000709.4:c.796delA, NM_000709.4:c.853G>C, NM_000709.4:c.868G>A, NM_000709.4:c.906_907delTG, NM_000709.4:c.905A>C, NM_000709.4:c.917delT, NM_000709.4:c.929C>G, NM_000709.4:c.964C>T, NM_000709.4:c.979G>A, NM_000709.4:c.1036C>T, NM_000709.4:c.1037G>A, NM_000709.4:c.1234G>A
BCKDHB	Maple syrup urine disease, type Ib	NM_183050.4:c.302G>A, NM_183050.4:c.342T>G, NM_183050.4:c.344-1G>A, NM_183050.4:c.356T>G, NM_183050.4:c.479T>G, NM_183050.4:c.488A>T, NM_183050.4:c.508C>A, NM_183050.4:c.508C>G, NM_183050.4:c.508C>T, NM_183050.4:c.509G>A, NM_183050.4:c.526A>T, NM_183050.4:c.547C>T, NM_183050.4:c.548G>C, NM_183050.4:c.748G>T, NM_183050.4:c.752T>C, NM_183050.4:c.799C>T, NM_183050.4:c.832G>A, NM_183050.4:c.952-1G>A, NM_183050.4:c.970C>T, NM_183050.4:c.1046G>A, NM_183050.4:c.1114G>T
BCS1L	GRACILE syndrome	NM_001079866.2:c.103G>C, NM_001079866.2:c.133C>T, NM_001079866.2:c.148A>G, NM_001079866.2:c.166C>T, NM_001079866.2:c.232A>G, NM_001079866.2:c.547C>T, NM_001079866.2:c.548G>A, NM_001079866.2:c.550C>T, NM_001079866.2:c.696delT, NM_001079866.2:c.830G>A, NM_001079866.2:c.1057G>A
BRIP1	Fanconi anemia, Group J	NM_032043.3:c.3209C>A, NM_032043.3:c.2990_2993delCAAA, NM_032043.3:c.2392C>T, NM_032043.3:c.2237_2240delTCAA, NM_032043.3:c.1702_1703delAA, NM_032043.3:c.1045G>C, NM_032043.3:c.502C>T, NM_032043.3:c.139C>G
BSND	Bartter syndrome, type 4a	NM_057176.3:c.1A>T, NM_057176.3:c.3G>A, NM_057176.3:c.10G>T, NM_057176.3:c.22C>T, NM_057176.3:c.23G>A, NM_057176.3:c.23G>T, NM_057176.3:c.35T>C, NM_057176.3:c.139G>A
BTD	Biotinidase Deficiency	NM_001370658.1:c.124G>A, NM_001370658.1:c.175C>T, NM_001370658.1:c.274G>C, NM_001370658.1:c.383G>A, NM_001370658.1:c.451G>A, NM_001370658.1:c.468G>T, NM_001370658.1:c.497G>A, NM_001370658.1:c.523A>G, NM_001370658.1:c.535G>A, NM_001370658.1:c.569A>G, NM_001370658.1:c.571C>T, NM_001370658.1:c.583C>T, NM_001370658.1:c.604G>A, NM_001370658.1:c.695A>G, NM_001370658.1:c.697C>T, NM_001370658.1:c.734A>T, NM_001370658.1:c.873delT, NM_001370658.1:c.908A>G, NM_001370658.1:c.1046C>T, NM_001370658.1:c.1260delG, NM_001370658.1:c.1279C>T, NM_001370658.1:c.1292G>A, NM_001370658.1:c.1308A>C, NM_001370658.1:c.1429C>T, NM_001370658.1:c.1447_1451delGGGAT, NM_001370658.1:c.1471C>G, NM_001370658.1:c.1535C>T, NM_001370658.1:c.1552C>T
CAPN3	Muscular dystrophy, limb-girdle, autosomal recessive 1	NM_000070.3:c.133G>A, NM_000070.3:c.220_221insT, NM_000070.3:c.246G>A, NM_000070.3:c.257C>T, NM_000070.3:c.328C>T, NM_000070.3:c.549delA, NM_000070.3:c.551C>T, NM_000070.3:c.580delT, NM_000070.3:c.597_611delGTTCTGGAGTGCTCT, NM_000070.3:c.662G>T, NM_000070.3:c.676G>A, NM_000070.3:c.853_854insAGTTGATTGC, NM_000070.3:c.956C>T, NM_000070.3:c.1319delG, NM_000070.3:c.1466G>A, NM_000070.3:c.1468C>T, NM_000070.3:c.1469G>A, NM_000070.3:c.1599_1602delGAGC, NM_000070.3:c.1610A>G, NM_000070.3:c.1715G>A, NM_000070.3:c.1743_1745+1delTGAG, NM_000070.3:c.1788_1789insA, NM_000070.3:c.1837delA, NM_000070.3:c.2120A>G, NM_000070.3:c.2212C>T, NM_000070.3:c.2243G>A, NM_000070.3:c.2248_2249insCAGT, NM_000070.3:c.2257G>A, NM_000070.3:c.2306G>A, NM_000070.3:c.2362_2363delAGinsTCATCT, NM_000070.3:c.2362_2363delAGinsTCATCT
CBS	Homocystinuria	NM_000071.3:c.1616T>C, NM_000071.3:c.1471C>T, NM_000071.3:c.1397C>T, NM_000071.3:c.1379C>T, NM_000071.3:c.1330G>A, NM_000071.3:c.1316G>A, NM_000071.3:c.1265C>T, NM_000071.3:c.1150A>G, NM_000071.3:c.1136G>A, NM_000071.3:c.1058C>T, NM_000071.3:c.1006C>T, NM_000071.3:c.992C>A, NM_000071.3:c.969G>A, NM_000071.3:c.959T>C, NM_000071.3:c.919G>A, NM_000071.3:c.904G>A, NM_000071.3:c.797G>A, NM_000071.3:c.572C>T, NM_000071.3:c.526G>T, NM_000071.3:c.502G>A, NM_000071.3:c.415G>A, NM_000071.3:c.393G>C, NM_000071.3:c.374G>A, NM_000071.3:c.341C>T, NM_000071.3:c.325T>C, NM_000071.3:c.304A>C, NM_000071.3:c.162G>A, NM_000071.3:c.146C>T

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
CDH23	Deafness, autosomal recessive 12	NM_022124.6:c.146-2A>G, NM_022124.6:c.189delC, NM_022124.6:c.288+1G>A, NM_022124.6:c.902G>A, NM_022124.6:c.1858+2T>G, NM_022124.6:c.3141C>A, NM_022124.6:c.3293A>G, NM_022124.6:c.3515_3518delCATC, NM_022124.6:c.3579+2T>C, NM_022124.6:c.4504C>T, NM_022124.6:c.5237G>A, NM_022124.6:c.5663T>C, NM_022124.6:c.6050-9G>A, NM_022124.6:c.6392delC, NM_022124.6:c.6442G>A, NM_022124.6:c.7660G>A, NM_022124.6:c.7823G>A, NM_022124.6:c.9319+1_9319+4delGTAA, NM_022124.6:c.9565C>T
CEP290	Ciliopathy	NM_025114.4:c.7394_7395delAG, NM_025114.4:c.7341dupA, NM_025114.4:c.7324G>T, NM_025114.4:c.6798G>A, NM_025114.4:c.6645+1G>A, NM_025114.4:c.6624delG, NM_025114.4:c.6448_6455delCAGTTGAA, NM_025114.4:c.5668G>T, NM_025114.4:c.5611_5614delCAAA, NM_025114.4:c.4962_4963delAA, NM_025114.4:c.4916C>A, NM_025114.4:c.4723A>T, NM_025114.4:c.4705-1G>T, NM_025114.4:c.4656delA, NM_025114.4:c.4393C>T, NM_025114.4:c.3185delT, NM_025114.4:c.2249T>G, NM_025114.4:c.1681C>T, NM_025114.4:c.1665_1666delAA, NM_025114.4:c.1501G>T, NM_025114.4:c.613C>T, NM_025114.4:c.384_387delTAGA, NM_025114.4:c.164_167delCTCA, NM_025114.4:c.21G>T
CERKL	Retinitis pigmentosa 26	NM_201548.5:c.1553_1569dupTTATCAGTCTTTATGGA, NM_201548.5:c.1012C>T, NM_201548.5:c.847C>T, NM_201548.5:c.780delT, NM_201548.5:c.769C>T, NM_201548.5:c.312delA
CFTR	Cystic fibrosis	NM_000492.3:c.*8753C-T, NM_000492.4:c.-165G>A, NM_000492.4:c.-13_10delCGCCCGAGAGACCATGCAGAGGT, NM_000492.4:c.1A>C, NM_000492.4:c.1A>G, NM_000492.4:c.2T>A, NM_000492.4:c.2T>C, NM_000492.4:c.2T>G, NM_000492.4:c.3G>A, NM_000492.4:c.4delC, NM_000492.4:c.3G>T, NM_000492.4:c.4C>T, NM_000492.4:c.11C>A, NM_000492.4:c.14C>T, NM_000492.4:c.19G>T, NM_000492.4:c.35_36insTATCA, NM_000492.4:c.38C>T, NM_000492.4:c.40delA, NM_000492.4:c.40A>T, NM_000492.4:c.43delC, NM_000492.4:c.44delT, NM_000492.4:c.43_44insT, NM_000492.4:c.43_44insTT, NM_000492.4:c.44T>C, NM_000492.4:c.51delC, NM_000492.4:c.53+1G>T, NM_000492.4:c.54-2A>G, NM_000492.4:c.54-1G>A, NM_000492.4:c.56G>A, NM_000492.4:c.57G>A, NM_000492.4:c.79delG, NM_000492.4:c.79G>A, NM_000492.4:c.79G>T, NM_000492.4:c.88C>T, NM_000492.4:c.98_115delAATTGTCAGACATATACC, NM_000492.4:c.104_105insA, NM_000492.4:c.109_110delAT, NM_000492.4:c.114C>G, NM_000492.4:c.115C>T, NM_000492.4:c.125C>T, NM_000492.4:c.137C>A, NM_000492.4:c.137C>T, NM_000492.4:c.143_146delATCT, NM_000492.4:c.148T>C, NM_000492.4:c.152delA, NM_000492.4:c.152_153insAAAATTGG, NM_000492.4:c.164+1G>A, NM_000492.4:c.164+1G>C, NM_000492.4:c.164+1_164+2insT, NM_000492.4:c.164+1G>T, NM_000492.4:c.164+2T>A, NM_000492.4:c.164+2T>C, NM_000492.4:c.164+2T>G, NM_000492.4:c.164+3_164+4insT, NM_000492.4:c.164+12T>C, NM_000492.4:c.165-3C>T, NM_000492.4:c.165-2A>G, NM_000492.4:c.165-1G>A, NM_000492.4:c.166G>A, NM_000492.4:c.167delA, NM_000492.4:c.166G>T, NM_000492.4:c.169T>G, NM_000492.4:c.170G>A, NM_000492.4:c.171G>A, NM_000492.4:c.172_175delGATA, NM_000492.4:c.174_175insA, NM_000492.4:c.178G>A, NM_000492.4:c.178G>T, NM_000492.4:c.200C>T, NM_000492.4:c.206T>A, NM_000492.4:c.215delC, NM_000492.4:c.220C>T, NM_000492.4:c.221G>A, NM_000492.4:c.223C>T, NM_000492.4:c.224G>A, NM_000492.4:c.228delT, NM_000492.4:c.227_228insT, NM_000492.4:c.234delC, NM_000492.4:c.236G>A, NM_000492.4:c.241delT, NM_000492.4:c.246_247insT, NM_000492.4:c.254G>A, NM_000492.4:c.254G>T, NM_000492.4:c.259_260delTT, NM_000492.4:c.259T>A, NM_000492.4:c.263T>A, NM_000492.4:c.263T>G, NM_000492.4:c.271G>A, NM_000492.4:c.273G>C, NM_000492.4:c.273+1G>A, NM_000492.4:c.273+3A>C, NM_000492.4:c.273+4A>G, NM_000492.4:c.274-2A>C, NM_000492.4:c.274-2A>G, NM_000492.4:c.274-1G>A, NM_000492.4:c.274-1G>C, NM_000492.4:c.274-1G>T, NM_000492.4:c.274G>A, NM_000492.4:c.274G>T, NM_000492.4:c.292C>T, NM_000492.4:c.293A>G, NM_000492.4:c.296C>T, NM_000492.4:c.302T>G, NM_000492.4:c.302_303insA, NM_000492.4:c.305T>G, NM_000492.4:c.307G>T, NM_000492.4:c.309delA, NM_000492.4:c.312delA, NM_000492.4:c.317_324delTAGCTTCC, NM_000492.4:c.325_326delTA, NM_000492.4:c.325T>C, NM_000492.4:c.325_327delTATinsG, NM_000492.4:c.325T>G, NM_000492.4:c.326A>G, NM_000492.4:c.327T>A, NM_000492.4:c.328delG, NM_000492.4:c.328G>C, NM_000492.4:c.328G>T, NM_000492.4:c.330C>A, NM_000492.4:c.340A>T, NM_000492.4:c.343G>T, NM_000492.4:c.346G>A, NM_000492.4:c.349C>T, NM_000492.4:c.350G>A, NM_000492.4:c.350G>T, NM_000492.4:c.355A>G, NM_000492.4:c.357delC, NM_000492.4:c.358G>A, NM_000492.4:c.366T>A, NM_000492.4:c.370G>C, NM_000492.4:c.376G>A, NM_000492.4:c.377G>A, NM_000492.4:c.378_379insTTA, NM_000492.4:c.380T>G, NM_000492.4:c.386delT, NM_000492.4:c.391delT, NM_000492.4:c.402_403insAC, NM_000492.4:c.409delC, NM_000492.4:c.409_412delCTCC, NM_000492.4:c.410T>C, NM_000492.4:c.411_412insCTA, NM_000492.4:c.415_416insGA, NM_000492.4:c.415_416insTA, NM_000492.4:c.416A>G, NM_000492.4:c.416A>T, NM_000492.4:c.419_420insA, NM_000492.4:c.422C>A, NM_000492.4:c.424delA, NM_000492.4:c.425delT, NM_000492.4:c.432delC, NM_000492.4:c.442delA, NM_000492.4:c.443T>A, NM_000492.4:c.443T>C, NM_000492.4:c.445G>A, NM_000492.4:c.445G>T, NM_000492.4:c.446G>T, NM_000492.4:c.454A>G, NM_000492.4:c.455T>G, NM_000492.4:c.459_476delAATAGCTATGTTTAGTTT, NM_000492.4:c.469_482delTTTAGTTTGATTTA, NM_000492.4:c.470T>G, NM_000492.4:c.473_474insT, NM_000492.4:c.476T>A, NM_000492.4:c.476T>C, NM_000492.4:c.481T>G, NM_000492.4:c.483_484insCC, NM_000492.4:c.487delA, NM_000492.4:c.487A>G, NM_000492.4:c.489+1G>T, NM_000492.4:c.489+2T>C, NM_000492.4:c.489+2T>G, NM_000492.4:c.489+3A>G, NM_000492.4:c.490-2A>C, NM_000492.4:c.490-2A>G, NM_000492.4:c.490-1G>A, NM_000492.4:c.490-1G>T, NM_000492.4:c.492delT, NM_000492.4:c.494T>C, NM_000492.4:c.496A>G, NM_000492.4:c.505_506insG, NM_000492.4:c.509G>A, NM_000492.4:c.518_522delATAAA, NM_000492.4:c.523A>G, NM_000492.4:c.525delA, NM_000492.4:c.530delT, NM_000492.4:c.529_530insT, NM_000492.4:c.532G>A, NM_000492.4:c.535C>A, NM_000492.4:c.541_544delGTTA, NM_000492.4:c.547C>A, NM_000492.4:c.549delC, NM_000492.4:c.559delA, NM_000492.4:c.567C>A, NM_000492.4:c.571T>G, NM_000492.4:c.575_579+2delATGAAGT, NM_000492.4:c.575A>G, NM_000492.4:c.577G>A, NM_000492.4:c.577G>T, NM_000492.4:c.579+1G>T, NM_000492.4:c.579+3A>G, NM_000492.4:c.579+5G>A, NM_000492.4:c.580-2A>C, NM_000492.4:c.580-1G>T, NM_000492.4:c.592G>A, NM_000492.4:c.595C>T, NM_000492.4:c.601delG, NM_000492.4:c.601G>A, NM_000492.4:c.606G>A, NM_000492.4:c.613C>T, NM_000492.4:c.617T>G, NM_000492.4:c.619C>T, NM_000492.4:c.647G>A, NM_000492.4:c.650_659delAGTTGTTACA, NM_000492.4:c.650A>G, NM_000492.4:c.653T>A, NM_000492.4:c.656_657insA, NM_000492.4:c.658C>T, NM_000492.4:c.675T>A, NM_000492.4:c.680T>G, NM_000492.4:c.695T>A, NM_000492.4:c.704delT, NM_000492.4:c.714delT, NM_000492.4:c.715delG, NM_000492.4:c.720_741delAGGGAGAATGATGATGAAGTAC, NM_000492.4:c.738_739insTACA, NM_000492.4:c.741C>G, NM_000492.4:c.743+1G>A, NM_000492.4:c.743+1G>C, NM_000492.4:c.743+2T>C, NM_000492.4:c.744-14_744-3delTGATTGATTTAC, NM_000492.4:c.744-2A>G, NM_000492.4:c.760delA, NM_000492.4:c.773delG, NM_000492.4:c.772A>G, NM_000492.4:c.794T>G, NM_000492.4:c.800delA, NM_000492.4:c.803_804delAT, NM_000492.4:c.811delT, NM_000492.4:c.825C>G, NM_000492.4:c.828C>A, NM_000492.4:c.830G>A, NM_000492.4:c.835G>T, NM_000492.4:c.844_845insA, NM_000492.4:c.846A>T, NM_000492.4:c.848_860delAAATGATTGAAAAinsTG, NM_000492.4:c.856_857insA, NM_000492.4:c.859_863delAACTT, NM_000492.4:c.864_868delAAGAC, NM_000492.4:c.865A>T, NM_000492.4:c.868C>T, NM_000492.4:c.869+1G>C, NM_000492.4:c.869+1G>T, NM_000492.4:c.869+3A>T, NM_000492.4:c.869+5G>A, NM_000492.4:c.870-2A>G, NM_000492.4:c.870-1G>C, NM_000492.4:c.880_881delAA, NM_000492.4:c.886_887insCT, NM_000492.4:c.912C>G, NM_000492.4:c.926delC, NM_000492.4:c.927_929delCTT, NM_000492.4:c.933C>G, NM_000492.4:c.938C>A, NM_000492.4:c.941G>A, NM_000492.4:c.946delT, NM_000492.4:c.950T>A, NM_000492.4:c.959T>A, NM_000492.4:c.980delT, NM_000492.4:c.985delA, NM_000492.4:c.988G>T, NM_000492.4:c.997C>T, NM_000492.4:c.1000C>T, NM_000492.4:c.1001G>A, NM_000492.4:c.1001G>T, NM_000492.4:c.1006_1007insG, NM_000492.4:c.1007T>A, NM_000492.4:c.1013C>T, NM_000492.4:c.1018delA, NM_000492.4:c.1018_1019insTC, NM_000492.4:c.1021T>C, NM_000492.4:c.1029delC, NM_000492.4:c.1029_1030insG, NM_000492.4:c.1037T>C, NM_000492.4:c.1040G>A, NM_000492.4:c.1040G>C, NM_000492.4:c.1040G>T, NM_000492.4:c.1045G>C, NM_000492.4:c.1046C>T, NM_000492.4:c.1052_1053delCT, NM_000492.4:c.1052C>G, NM_000492.4:c.1054C>T, NM_000492.4:c.1055G>A, NM_000492.4:c.1057C>T, NM_000492.4:c.1067delG, NM_000492.4:c.1068G>A, NM_000492.4:c.1075_1079delCAAACinsAAAAA, NM_000492.4:c.1075C>A, NM_000492.4:c.1079C>A, NM_000492.4:c.1081delT, NM_000492.4:c.1082delG, NM_000492.4:c.1083_1084insTATGA, NM_000492.4:c.1086T>A,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000492.4:c.1089_1090delCT, NM_000492.4:c.1090T>C, NM_000492.4:c.1115delA, NM_000492.4:c.1116G>T, NM_000492.4:c.1116+1G>A, NM_000492.4:c.1116+1G>C, NM_000492.4:c.1116+1G>T, NM_000492.4:c.1117-2A>G, NM_000492.4:c.1117-1G>A, NM_000492.4:c.1117G>A, NM_000492.4:c.1126_1127insA, NM_000492.4:c.1126C>T, NM_000492.4:c.1135G>T, NM_000492.4:c.1141A>T, NM_000492.4:c.1151delA, NM_000492.4:c.1151_1152insAT, NM_000492.4:c.1154_1155insT, NM_000492.4:c.1155_1156insTA, NM_000492.4:c.1159_1160delTT, NM_000492.4:c.1160_1166delTAACGAC, NM_000492.4:c.1175T>G, NM_000492.4:c.1177delG, NM_000492.4:c.1190_1191insA, NM_000492.4:c.1196delC, NM_000492.4:c.1202G>A, NM_000492.4:c.1203G>A, NM_000492.4:c.1209+1G>A, NM_000492.4:c.1209+1G>T, NM_000492.4:c.1210-34_1210-33delTG, NM_000492.4:c.1210-34_1210-31delTGTG, NM_000492.4:c.1210-35_1210-34insTG, NM_000492.4:c.1210-35_1210-34insTGTG, NM_000492.4:c.1210-14_1210-11delTGTT, NM_000492.4:c.1210-15_1210-13delGTGinsT, NM_000492.4:c.1210-12_1210-11delTT, NM_000492.4:c.1210-13_1210-12insTGTGTT, NM_000492.4:c.1210-13_1210-12insTGTT, NM_000492.4:c.1210-13_1210-12insTT, NM_000492.4:c.1210-13G>T, NM_000492.4:c.1210-11delTinsGTGTG, NM_000492.4:c.1210-11delTinsGTG, NM_000492.4:c.1210-11T>G, NM_000492.4:c.1210-2_1210-1delAG, NM_000492.4:c.1210-1delG, NM_000492.4:c.1219G>T, NM_000492.4:c.1231_1235delAAAGC, NM_000492.4:c.1235_1236insA, NM_000492.4:c.1240C>T, NM_000492.4:c.1297_1303delTTCTCAC, NM_000492.4:c.1301C>A, NM_000492.4:c.1301C>G, NM_000492.4:c.1315C>T, NM_000492.4:c.1323_1344delGAAAGATATTAATTTCAAGATA, NM_000492.4:c.1325_1326insAGAT, NM_000492.4:c.1328_1329delAT, NM_000492.4:c.1327G>T, NM_000492.4:c.1339delA, NM_000492.4:c.1344_1347delAGAA, NM_000492.4:c.1364C>A, NM_000492.4:c.1365_1366delGG, NM_000492.4:c.1367T>C, NM_000492.4:c.1370_1378delCTGGATCCA, NM_000492.4:c.1372delG, NM_000492.4:c.1373G>T, NM_000492.4:c.1393-2A>G, NM_000492.4:c.1393-1G>A, NM_000492.4:c.1397C>A, NM_000492.4:c.1397C>G, NM_000492.4:c.1399C>T, NM_000492.4:c.1400T>C, 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NM_000492.4:c.1558G>T, NM_000492.4:c.1559T>A, NM_000492.4:c.1572C>A, NM_000492.4:c.1573C>T, NM_000492.4:c.1584+1G>A, NM_000492.4:c.1584+1G>T, NM_000492.4:c.1584+2T>C, NM_000492.4:c.1585-8G>A, NM_000492.4:c.1585-2A>G, NM_000492.4:c.1585-1G>A, NM_000492.4:c.1588A>C, NM_000492.4:c.1601C>A, NM_000492.4:c.1606A>T, NM_000492.4:c.1610_1611delAC, NM_000492.4:c.1612_1613insAT, NM_000492.4:c.1624G>T, NM_000492.4:c.1625G>A, NM_000492.4:c.1628A>C, NM_000492.4:c.1642_1643delCT, NM_000492.4:c.1643_1646delTGAG, NM_000492.4:c.1645A>C, NM_000492.4:c.1646G>A, NM_000492.4:c.1646G>T, NM_000492.4:c.1647T>A, NM_000492.4:c.1647T>G, NM_000492.4:c.1648G>T, NM_000492.4:c.1650delA, NM_000492.4:c.1651delG, NM_000492.4:c.1651_1652delGGinsAA, NM_000492.4:c.1651G>A, NM_000492.4:c.1652G>A, NM_000492.4:c.1654C>A, NM_000492.4:c.1655delA, NM_000492.4:c.1654C>T, NM_000492.4:c.1657C>T, NM_000492.4:c.1658G>A, NM_000492.4:c.1660_1661insA, NM_000492.4:c.1670delC, NM_000492.4:c.1673T>C, NM_000492.4:c.1674delA, NM_000492.4:c.1675G>A, NM_000492.4:c.1679G>A, NM_000492.4:c.1679G>C, NM_000492.4:c.1679+1G>A, NM_000492.4:c.1679+1G>C, NM_000492.4:c.1679+2T>C, NM_000492.4:c.1680-886A>G, NM_000492.4:c.1680-883A>G, NM_000492.4:c.1680-877G>T, NM_000492.4:c.1680-1G>A, NM_000492.4:c.1680-1G>C, NM_000492.4:c.1680A>C, NM_000492.4:c.1681_1682insC, NM_000492.4:c.1682C>A, NM_000492.4:c.1684G>C, NM_000492.4:c.1687T>A, NM_000492.4:c.1687T>C, NM_000492.4:c.1687T>G, NM_000492.4:c.1690delA, NM_000492.4:c.1701delT, NM_000492.4:c.1700A>C, NM_000492.4:c.1703T>A, NM_000492.4:c.1705T>G, NM_000492.4:c.1706A>G, NM_000492.4:c.1707T>A, NM_000492.4:c.1713_1714delAG, NM_000492.4:c.1721C>A, NM_000492.4:c.1726G>T, NM_000492.4:c.1727G>C, NM_000492.4:c.1731C>A, NM_000492.4:c.1733_1734delTA, NM_000492.4:c.1736A>G, NM_000492.4:c.1738delG, NM_000492.4:c.1738_1739insT, NM_000492.4:c.1753G>T, NM_000492.4:c.1760T>C, NM_000492.4:c.1766G>A, NM_000492.4:c.1766+1G>A, NM_000492.4:c.1766+1G>C, NM_000492.4:c.1766+1G>T, NM_000492.4:c.1766+2T>A, NM_000492.4:c.1766+2T>C, NM_000492.4:c.1766+3A>C, NM_000492.4:c.1766+3A>G, NM_000492.4:c.1766+5G>T, NM_000492.4:c.1767-1G>A, NM_000492.4:c.1786_1787delGC, NM_000492.4:c.1792_1798delAAAACTA, NM_000492.4:c.1792A>T, NM_000492.4:c.1799delG, NM_000492.4:c.1826A>G, NM_000492.4:c.1826A>T, NM_000492.4:c.1837G>A, NM_000492.4:c.1841A>G, NM_000492.4:c.1853T>C, NM_000492.4:c.1865G>A, NM_000492.4:c.1871_1878delGCTATTTT, NM_000492.4:c.1882G>A, NM_000492.4:c.1882G>C, NM_000492.4:c.1900C>T, NM_000492.4:c.1909C>T, NM_000492.4:c.1911delG, NM_000492.4:c.1918_1919delTT, NM_000492.4:c.1919_1920insTA, NM_000492.4:c.1923C>A, NM_000492.4:c.1923_1931delCTCAAAACTinsA, NM_000492.4:c.1936G>T, NM_000492.4:c.1943delA, NM_000492.4:c.1943A>T, NM_000492.4:c.1966G>T, NM_000492.4:c.1970delG, NM_000492.4:c.1973_1985delGAAATTCAATCCTinsAGAAA, NM_000492.4:c.1973delGinsAGAAA, NM_000492.4:c.1974delA, NM_000492.4:c.1980delA, NM_000492.4:c.1979C>G, NM_000492.4:c.1984_1987delCTAA, NM_000492.4:c.1990G>T, NM_000492.4:c.2009_2010insA, NM_000492.4:c.2011delT, NM_000492.4:c.2017G>T, NM_000492.4:c.2036G>A, NM_000492.4:c.2042delA, NM_000492.4:c.2044_2045insC, NM_000492.4:c.2045_2046insA, NM_000492.4:c.2046_2047delAA, NM_000492.4:c.2051_2052insT, NM_000492.4:c.2051_2052delAAinsG, NM_000492.4:c.2052_2053insC, NM_000492.4:c.2053C>T, NM_000492.4:c.2057_2058insTTTT, NM_000492.4:c.2062A>T, NM_000492.4:c.2065C>T, NM_000492.4:c.2074G>T, NM_000492.4:c.2079_2080insG, NM_000492.4:c.2084delA, NM_000492.4:c.2107delA, NM_000492.4:c.2125C>T, NM_000492.4:c.2128A>T, NM_000492.4:c.2143C>T, NM_000492.4:c.2146A>T, NM_000492.4:c.2156T>A, NM_000492.4:c.2158C>T, NM_000492.4:c.2173_2174insA, NM_000492.4:c.2184_2185insA, NM_000492.4:c.2188G>T, NM_000492.4:c.2195T>G, NM_000492.4:c.2202delA, NM_000492.4:c.2215delG, NM_000492.4:c.2233G>T, NM_000492.4:c.2239_2246delGCGATACT, NM_000492.4:c.2248_2255delCCTCGCAT, NM_000492.4:c.2249C>T, NM_000492.4:c.2252G>T, NM_000492.4:c.2274_2275delCC, NM_000492.4:c.2274delC, 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NM_000492.4:c.2552G>T, NM_000492.4:c.2553_2554insT, NM_000492.4:c.2555A>T, NM_000492.4:c.2556T>G, NM_000492.4:c.2562delT, NM_000492.4:c.2566_2567insT, NM_000492.4:c.2573delG, NM_000492.4:c.2579delT, NM_000492.4:c.2584_2594delGTGCTAATTTG, NM_000492.4:c.2591_2592delTT, NM_000492.4:c.2600T>A, NM_000492.4:c.2600_2601insA, NM_000492.4:c.2615delC, NM_000492.4:c.2619+1G>A, NM_000492.4:c.2619+1_2619+2insT, NM_000492.4:c.2619+2T>A, NM_000492.4:c.2620-26A>G, NM_000492.4:c.2620-2A>G, NM_000492.4:c.2620-1G>C, NM_000492.4:c.2620-1G>T, NM_000492.4:c.2641_2642insTG, NM_000492.4:c.2645G>A, NM_000492.4:c.2657+1_2657+2insA, NM_000492.4:c.2657+5G>A, NM_000492.4:c.2658-2A>G, NM_000492.4:c.2658-1G>C, NM_000492.4:c.2658-1G>T, NM_000492.4:c.2668C>T, NM_000492.4:c.2686_2687insT, NM_000492.4:c.2700T>A, NM_000492.4:c.2735C>A, NM_000492.4:c.2737_2738insG, NM_000492.4:c.2738A>G, NM_000492.4:c.2739T>A, NM_000492.4:c.2757C>G, NM_000492.4:c.2758G>A, NM_000492.4:c.2761_2762insGA, NM_000492.4:c.2768C>A, NM_000492.4:c.2775delT, NM_000492.4:c.2775_2776delTT, NM_000492.4:c.2780T>C, NM_000492.4:c.2807_2810delCACTinsAGA, NM_000492.4:c.2809_2810insT, NM_000492.4:c.2810_2811insG, NM_000492.4:c.2813T>G, NM_000492.4:c.2822delT, NM_000492.4:c.2825delT, NM_000492.4:c.2834C>T, NM_000492.4:c.2836A>T, NM_000492.4:c.2845C>T, NM_000492.4:c.2851delA, NM_000492.4:c.2855T>C, NM_000492.4:c.2856G>C, NM_000492.4:c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC, NM_000492.4:c.2869_2870insG, NM_000492.4:c.2875delG, NM_000492.4:c.2876delC, NM_000492.4:c.2879_2882delCTAT, NM_000492.4:c.2896delA, NM_000492.4:c.2900T>C, NM_000492.4:c.2908G>C, NM_000492.4:c.2908+1G>A, NM_000492.4:c.2908+2T>C,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000492.4:c.2909-15T>G, NM_000492.4:c.2909-5_2909-4insATAGGTGGGATTCTTA, NM_000492.4:c.2909-1delG, NM_000492.4:c.2909-1G>A, NM_000492.4:c.2909G>A, NM_000492.4:c.2923_2924delAG, NM_000492.4:c.2930C>T, NM_000492.4:c.2932A>T, NM_000492.4:c.2936A>C, NM_000492.4:c.2936A>T, NM_000492.4:c.2939T>A, NM_000492.4:c.2945_2946delTT, NM_000492.4:c.2967_2968insA, NM_000492.4:c.2981_2988+1delTCATCCAGG, NM_000492.4:c.2984_2985insC, NM_000492.4:c.2988G>A, NM_000492.4:c.2988+1G>A, NM_000492.4:c.2988+1G>C, NM_000492.4:c.2988+1G>T, NM_000492.4:c.2988+2T>C, NM_000492.4:c.2989-2A>G, NM_000492.4:c.2989-2A>T, NM_000492.4:c.2989-1G>A, NM_000492.4:c.2991G>C, NM_000492.4:c.2992delT, NM_000492.4:c.2994_2997delATTA, NM_000492.4:c.2997delA, NM_000492.4:c.3000_3001delTG, NM_000492.4:c.3007G>T, NM_000492.4:c.3011delC, NM_000492.4:c.3014T>G, NM_000492.4:c.3017C>A, NM_000492.4:c.3020delT, NM_000492.4:c.3022delG, NM_000492.4:c.3032T>G, NM_000492.4:c.3037delC, NM_000492.4:c.3036_3037insC, NM_000492.4:c.3038C>A, NM_000492.4:c.3041_3042delAC, NM_000492.4:c.3041A>G, NM_000492.4:c.3061C>T, NM_000492.4:c.3063_3068delAGTGAT, NM_000492.4:c.3068_3072delTAGTG, NM_000492.4:c.3095A>G, NM_000492.4:c.3104delA, NM_000492.4:c.3103C>T, NM_000492.4:c.3107C>A, NM_000492.4:c.3110C>A, NM_000492.4:c.3124C>T, NM_000492.4:c.3139delG, NM_000492.4:c.3139_3139+1delGG, NM_000492.4:c.3139+1G>A, NM_000492.4:c.3139+1G>T, NM_000492.4:c.3140-26A>G, NM_000492.4:c.3140-1G>A, NM_000492.4:c.3154T>G, NM_000492.4:c.3161delA, NM_000492.4:c.3160C>G, NM_000492.4:c.3176T>G, NM_000492.4:c.3179A>C, NM_000492.4:c.3181G>C, NM_000492.4:c.3183_3184insCTATG, NM_000492.4:c.3188G>A, NM_000492.4:c.3189G>A, NM_000492.4:c.3194T>C, NM_000492.4:c.3196C>T, NM_000492.4:c.3197G>A, NM_000492.4:c.3199G>A, NM_000492.4:c.3200C>T, NM_000492.4:c.3205G>A, NM_000492.4:c.3208C>T, NM_000492.4:c.3209G>A, NM_000492.4:c.3211C>T, NM_000492.4:c.3212A>C, NM_000492.4:c.3215_3216insT, NM_000492.4:c.3222T>A, NM_000492.4:c.3227_3228delCT, NM_000492.4:c.3230_3231delTG, NM_000492.4:c.3230T>C, NM_000492.4:c.3254A>G, NM_000492.4:c.3261_3262insA, NM_000492.4:c.3264delC, NM_000492.4:c.3266G>A, NM_000492.4:c.3276C>A, NM_000492.4:c.3276C>G, NM_000492.4:c.3287delT, NM_000492.4:c.3291delC, NM_000492.4:c.3292T>C, NM_000492.4:c.3293delG, NM_000492.4:c.3293G>A, NM_000492.4:c.3294G>A, NM_000492.4:c.3294G>C, NM_000492.4:c.3294G>T, NM_000492.4:c.3299A>C, NM_000492.4:c.3302T>A, NM_000492.4:c.3302T>G, NM_000492.4:c.3304A>T, NM_000492.4:c.3310G>T, NM_000492.4:c.3315delG, NM_000492.4:c.3324delC, NM_000492.4:c.3342_3343insA, NM_000492.4:c.3353C>T, NM_000492.4:c.3363delA, NM_000492.4:c.3367+1G>A, NM_000492.4:c.3367+2T>A, NM_000492.4:c.3367+2T>C, NM_000492.4:c.3368-2A>G, NM_000492.4:c.3368-2A>T, NM_000492.4:c.3368-1G>A, NM_000492.4:c.3380G>A, NM_000492.4:c.3382A>T, NM_000492.4:c.3409A>G, NM_000492.4:c.3420_3421insAGTA, NM_000492.4:c.3430C>T, NM_000492.4:c.3434G>A, NM_000492.4:c.3435G>A, NM_000492.4:c.3445delT, NM_000492.4:c.3454G>C, NM_000492.4:c.3468G>A, NM_000492.4:c.3468+1G>A, NM_000492.4:c.3468+1_3468+2insT, NM_000492.4:c.3468+2T>C, NM_000492.4:c.3468+3_3468+4insT, NM_000492.4:c.3468+5G>A, NM_000492.4:c.3469-20T>C, NM_000492.4:c.3469-2A>G, NM_000492.4:c.3472C>T, NM_000492.4:c.3475T>C, NM_000492.4:c.3476C>T, NM_000492.4:c.3485_3486delGA, NM_000492.4:c.3484C>T, NM_000492.4:c.3485G>T, NM_000492.4:c.3495delG, NM_000492.4:c.3496delT, NM_000492.4:c.3527delC, NM_000492.4:c.3529delA, NM_000492.4:c.3529A>T, NM_000492.4:c.3531_3532insTCAA, NM_000492.4:c.3533_3536delCAAC, NM_000492.4:c.3533C>A, NM_000492.4:c.3538delA, NM_000492.4:c.3546C>G, NM_000492.4:c.3556C>T, NM_000492.4:c.3569_3570delTT, NM_000492.4:c.3587C>G, NM_000492.4:c.3592delG, NM_000492.4:c.3598delA, NM_000492.4:c.3605delA, NM_000492.4:c.3611G>A, NM_000492.4:c.3612G>A, NM_000492.4:c.3617C>A, NM_000492.4:c.3618_3619delAG, NM_000492.4:c.3617C>G, NM_000492.4:c.3619delG, NM_000492.4:c.3657_3658insA, NM_000492.4:c.3659delC, NM_000492.4:c.3659C>T, NM_000492.4:c.3664_3665insTCAA, NM_000492.4:c.3689delT, NM_000492.4:c.3700A>G, NM_000492.4:c.3712C>T, NM_000492.4:c.3717G>A, NM_000492.4:c.3717+1G>A, NM_000492.4:c.3717+4A>G, NM_000492.4:c.3717+5G>A, NM_000492.4:c.3717+40A>G, NM_000492.4:c.3718-2477C>T, NM_000492.4:c.3718-3T>G, NM_000492.4:c.3718-1G>A, NM_000492.4:c.3719T>G, NM_000492.4:c.3728T>A, NM_000492.4:c.3731G>A, NM_000492.4:c.3737C>T, NM_000492.4:c.3739G>A, NM_000492.4:c.3744delA, NM_000492.4:c.3745delG, NM_000492.4:c.3745G>A, NM_000492.4:c.3745G>C, NM_000492.4:c.3746G>A, NM_000492.4:c.3752G>A, NM_000492.4:c.3761T>G, NM_000492.4:c.3763T>C, NM_000492.4:c.3764C>A, NM_000492.4:c.3764C>G, NM_000492.4:c.3764C>T, NM_000492.4:c.3766_3767insC, NM_000492.4:c.3767_3768insT, NM_000492.4:c.3773_3774insG, NM_000492.4:c.3806T>A, NM_000492.4:c.3808delG, NM_000492.4:c.3808G>A, NM_000492.4:c.3810T>A, NM_000492.4:c.3812_3813delGT, NM_000492.4:c.3822G>A, NM_000492.4:c.3828delA, NM_000492.4:c.3834_3835delTT, NM_000492.4:c.3841C>T, NM_000492.4:c.3844T>G, NM_000492.4:c.3846G>A, NM_000492.4:c.3848G>T, NM_000492.4:c.3849_3850insAA, NM_000492.4:c.3854delC, NM_000492.4:c.3857T>C, NM_000492.4:c.3868C>A, NM_000492.4:c.3871C>T, NM_000492.4:c.3873G>C, NM_000492.4:c.3873+1G>A, NM_000492.4:c.3873+1G>T, NM_000492.4:c.3873+2T>C, NM_000492.4:c.3874-2A>G, NM_000492.4:c.3874-1G>A, NM_000492.4:c.3874delA, NM_000492.4:c.3878_3881delTATT, NM_000492.4:c.3883delA, NM_000492.4:c.3883_3884insG, NM_000492.4:c.3883_3884insT, NM_000492.4:c.3890_3891insT, NM_000492.4:c.3897_3898insT, NM_000492.4:c.3902_3903insA, NM_000492.4:c.3903delA, NM_000492.4:c.3907A>C, NM_000492.4:c.3909C>G, NM_000492.4:c.3915_3916insCC, NM_000492.4:c.3921T>A, NM_000492.4:c.3922G>T, NM_000492.4:c.3925C>T, NM_000492.4:c.3929G>A, NM_000492.4:c.3937C>T, NM_000492.4:c.3940_3947delGAAATATG, NM_000492.4:c.3947G>A, NM_000492.4:c.3957_3958insAGGG, NM_000492.4:c.3963+1G>A, NM_000492.4:c.3963+1G>C, NM_000492.4:c.3963+1G>T, NM_000492.4:c.3963+2T>A, NM_000492.4:c.3963+2T>G, NM_000492.4:c.3964-1G>A, NM_000492.4:c.3971T>C, NM_000492.4:c.3976delT, NM_000492.4:c.3985G>C, NM_000492.4:c.3987_3988delAC, NM_000492.4:c.3988C>T, NM_000492.4:c.3997delG, NM_000492.4:c.3997G>T, NM_000492.4:c.4003C>T, NM_000492.4:c.4004T>C, NM_000492.4:c.4023_4024insGGGG, NM_000492.4:c.4024delG, NM_000492.4:c.4034_4035insCCTA, NM_000492.4:c.4035_4041delCCTAAGC, NM_000492.4:c.4037_4038insA, NM_000492.4:c.4040_4041delGC, NM_000492.4:c.4041delC, NM_000492.4:c.4046G>A, NM_000492.4:c.4057delT, NM_000492.4:c.4056G>T, NM_000492.4:c.4077_4080delTGTTinsAA, NM_000492.4:c.4078delG, NM_000492.4:c.4085_4086insT, NM_000492.4:c.4111G>T, NM_000492.4:c.4124A>C, NM_000492.4:c.4126_4130delTTGGA, NM_000492.4:c.4136+1G>A, NM_000492.4:c.4139delC, NM_000492.4:c.4140delA, NM_000492.4:c.4141T>C, NM_000492.4:c.4143C>A, NM_000492.4:c.4143C>G, NM_000492.4:c.4144_4145insA, NM_000492.4:c.4144C>T, NM_000492.4:c.4169delA, NM_000492.4:c.4168C>T, NM_000492.4:c.4194_4195delTC, NM_000492.4:c.4198_4199delTG, NM_000492.4:c.4199delG, NM_000492.4:c.4200_4201insG, NM_000492.4:c.4201G>T, NM_000492.4:c.4231C>T, NM_000492.4:c.4234C>T, NM_000492.4:c.4242+1G>A, NM_000492.4:c.4242+1G>T, NM_000492.4:c.4242+2T>C, NM_000492.4:c.4242+5G>A, NM_000492.4:c.4243-2A>C, NM_000492.4:c.4243-2A>G, NM_000492.4:c.4243-1G>C, NM_000492.4:c.4250delA, NM_000492.4:c.4252delG, NM_000492.4:c.4252G>T, NM_000492.4:c.4272C>A, NM_000492.4:c.4272C>G, NM_000492.4:c.4280T>C, NM_000492.4:c.4296_4297insGA, NM_000492.4:c.4297G>A, NM_000492.4:c.4297G>T, NM_000492.4:c.4303_4316delAGCCTCTTCCGGCA, NM_000492.4:c.4326delC, NM_000492.4:c.4333G>A, NM_000492.4:c.4337delG, NM_000492.4:c.4357C>T, NM_000492.4:c.4364C>A, NM_000492.4:c.4364C>G, NM_000492.4:c.4400delT, NM_000492.4:c.4417G>T, NM_000492.4:c.4426C>T, NM_000492.4:c.-102delA, NM_000492.4:c.1210-35_1210-34insTGTGTG, NM_000492.4:c.1210-17_1210-13delGTGTGinsT, NM_000492.4:c.1210-11delTinsGTGTGTG
CLN3	Neuronal ceroid-lipofuscinosis	NM_001042432.2:c.1272delG, NM_001042432.2:c.1210C>A, NM_001042432.2:c.883G>A, NM_001042432.2:c.622dupT, NM_001042432.2:c.597C>A
CLN5	Ceroid lipofuscinosis, neuronal, 5	NM_006493.4:c.188G>A, NM_006493.4:c.188G>C, NM_006493.4:c.230G>A, NM_006493.4:c.286C>T, NM_006493.4:c.377T>G, NM_006493.4:c.377_378insA, NM_006493.4:c.418C>T, NM_006493.4:c.428A>G, NM_006493.4:c.446T>C, NM_006493.4:c.448C>T, NM_006493.4:c.466C>T, NM_006493.4:c.472T>C, NM_006493.4:c.473G>C, NM_006493.4:c.521_522insC, NM_006493.4:c.688G>A, NM_006493.4:c.771delA, NM_006493.4:c.775_776delAT, NM_006493.4:c.803_818delATCTGGGAAATGAAAC, NM_006493.4:c.879C>A
CLN6	Ceroid Lipofuscinosis, Neuronal, 6	NM_017882.3:c.663C>G, NM_017882.3:c.307C>T, NM_017882.3:c.214G>T, NM_017882.3:c.214G>C, NM_017882.3:c.200T>C, NM_017882.3:c.139C>T

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
CLN8	Ceroid Lipofuscinosis, Neuronal, 8 (a.ka. Northern Epilepsy)	NM_018941.4:c.88delG, NM_018941.4:c.88G>C, NM_018941.4:c.610C>T, NM_018941.4:c.789G>C
CLRN1	Usher syndrome, type 3A	NM_174878.3:c.633dupT, NM_174878.3:c.433+1061A>T, NM_174878.3:c.189C>A, NM_174878.3:c.144T>G, NM_174878.3:c.118T>G, NM_174878.3:c.92C>T
CNGB3	Achromatopsia 3/Macular degeneration, juvenile	NM_019098.5:c.2048_2049delCA, NM_019098.5:c.2011G>T, NM_019098.5:c.1672G>T, NM_019098.5:c.1405T>G, NM_019098.5:c.1208G>A, NM_019098.5:c.1148delC, NM_019098.5:c.1063C>T, NM_019098.5:c.893_897delCAAAA, NM_019098.5:c.887_896delCTTCTACAAA, NM_019098.5:c.886_890delACTTC, NM_019098.5:c.819_826delCAGACTCC, NM_019098.5:c.446_447insT
COL4A3	Alport syndrome	NM_000091.5:c.343delG, NM_000091.5:c.346C>A, NM_000091.5:c.898G>A, NM_000091.5:c.2083G>A, NM_000091.5:c.2110delC, NM_000091.5:c.2954G>T, NM_000091.5:c.4411_4415delTTTTC, NM_000091.5:c.4421T>C, NM_000091.5:c.4441C>T, NM_000091.5:c.4484A>G, NM_000091.5:c.4571C>G, NM_000091.5:c.5002_*6delAAAAGACACTGAAGCTAA
COL4A4	Alport Syndrome, COL4A4-Related	NM_000092.5:c.4923C>A, NM_000092.5:c.4129C>T, NM_000092.5:c.3713C>A, NM_000092.5:c.3601G>A, NM_000092.5:c.2312delG, NM_000092.5:c.71+1G>A
COL7A1	Dystrophic Epidermolysis Bullosa, COL7A1-Related	NM_000094.4:c.8524_8527+10delGAAGGTGAGGACAG, NM_000094.4:c.8479C>T, NM_000094.4:c.8440C>T, NM_000094.4:c.8393T>A, NM_000094.4:c.8371C>T, NM_000094.4:c.8245G>A, NM_000094.4:c.7957G>A, NM_000094.4:c.7930-1G>C, NM_000094.4:c.7912G>T, NM_000094.4:c.7440+4delC, NM_000094.4:c.7411C>T, NM_000094.4:c.7345-1G>A, NM_000094.4:c.6946G>A, NM_000094.4:c.6859G>A, NM_000094.4:c.6752G>A, NM_000094.4:c.6670G>T, NM_000094.4:c.6573+1G>T, NM_000094.4:c.6527dupC, NM_000094.4:c.6205C>T, NM_000094.4:c.6187C>T, NM_000094.4:c.6091G>A, NM_000094.4:c.5821-1G>A, NM_000094.4:c.5532+1G>A, NM_000094.4:c.5443G>C, NM_000094.4:c.5287C>T, NM_000094.4:c.5096C>T, NM_000094.4:c.5052+1G>A, NM_000094.4:c.4888C>T, NM_000094.4:c.4783G>C, NM_000094.4:c.4373C>T, NM_000094.4:c.4119+1G>T, NM_000094.4:c.4039G>C, NM_000094.4:c.3831+1G>T, NM_000094.4:c.3809C>T, NM_000094.4:c.2471dupG, NM_000094.4:c.1907G>T, NM_000094.4:c.933C>A, NM_000094.4:c.887delG, NM_000094.4:c.706C>T, NM_000094.4:c.592G>A, NM_000094.4:c.425A>G, NM_000094.4:c.336C>G, NM_000094.4:c.238G>T
CPS1	Carbamoylphosphate synthetase I deficiency	NM_001875.5:c.697C>T, NM_001875.5:c.1631C>T, NM_001875.5:c.1912C>T, NM_001875.5:c.3555delA
CPT1A	Carnitine palmitoyltransferase deficiency, hepatic, type IA	NM_001876.4:c.1538C>T, NM_001876.4:c.1493A>G, NM_001876.4:c.1436C>T, NM_001876.4:c.1393G>T, NM_001876.4:c.1361A>G, NM_001876.4:c.1241C>T, NM_001876.4:c.1216C>T, NM_001876.4:c.1079A>G, NM_001876.4:c.335_336delCC, NM_001876.4:c.298C>T, NM_001876.4:c.281+1G>A, NM_001876.4:c.222C>A
CPT2	Carnitine palmitoyltransferase deficiency, hepatic, type II, infantile,lethal neonathal	NM_000098.3:c.149C>A, NM_000098.3:c.338C>T, NM_000098.3:c.359A>G, NM_000098.3:c.370C>T, NM_000098.3:c.452G>A, NM_000098.3:c.463_464insT, NM_000098.3:c.481C>T, NM_000098.3:c.520G>A, NM_000098.3:c.638A>G, NM_000098.3:c.680C>T, NM_000098.3:c.723_724delAC, NM_000098.3:c.886C>T, NM_000098.3:c.1148T>A, NM_000098.3:c.1238_1239delAG, NM_000098.3:c.1237C>T, NM_000098.3:c.1369A>T, NM_000098.3:c.1437C>G, NM_000098.3:c.1763C>G, NM_000098.3:c.1782delC, NM_000098.3:c.1883A>C, NM_000098.3:c.1891C>T
CRB1	Leber congenital amaurosis 8	NM_201253.3:c.484G>A, NM_201253.3:c.493_501delGATGGAATT, NM_201253.3:c.610_616delGAAATAG, NM_201253.3:c.936T>G, NM_201253.3:c.2290C>T, NM_201253.3:c.2401A>T, NM_201253.3:c.2416G>T, NM_201253.3:c.2688T>A, NM_201253.3:c.2843G>A, NM_201253.3:c.2983G>T, NM_201253.3:c.3053_3054insTTATA, NM_201253.3:c.3094G>A, NM_201253.3:c.3122T>C, NM_201253.3:c.3299T>C, NM_201253.3:c.3299T>G, NM_201253.3:c.3383delT, NM_201253.3:c.3419T>A, NM_201253.3:c.3997G>T
CTNS	Cystinosis, nephropathic	NM_004937.3:c.124G>A, NM_004937.3:c.283G>T, NM_004937.3:c.329G>T, NM_004937.3:c.357_360delCAGC, NM_004937.3:c.397_398delAT, NM_004937.3:c.414G>A, NM_004937.3:c.416C>T, NM_004937.3:c.506G>A, NM_004937.3:c.589G>A, NM_004937.3:c.645_646insA, NM_004937.3:c.853-3C>G, NM_004937.3:c.1015G>A
CTSD	Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)	NM_001909.5:c.1149G>C, NM_001909.5:c.685T>A
CTSK	Pycnodysostosis	NM_000396.4:c.926T>C, NM_000396.4:c.721C>T, NM_000396.4:c.436G>C, NM_000396.4:c.236G>A, NM_000396.4:c.154A>T
CYP21A2	Adrenal hyperplasia, congenital/Hyperandrogenism, nonclassic type due to 21-hydroxylase deficiency	NM_000500.9:c.293-13C/A>G, NM_000500.9:c.332_339del, NM_000500.9:c.518T>A, NM_000500.9:c.710T>A, NM_000500.9:c.713T>A, NM_000500.9:c.719T>A, NM_000500.9:c.923dup, NM_000500.9:c.955C>T, NM_000500.9:c.1069C>T, 30kb deletion, Large gene conversion

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
DBT	Maple syrup urine disease, type II	NM_001918.5:c.1281+1G>A, NM_001918.5:c.939G>C, NM_001918.5:c.901C>T, NM_001918.5:c.871C>T, NM_001918.5:c.827T>G, NM_001918.5:c.772+1G>A, NM_001918.5:c.670G>T, NM_001918.5:c.581C>G, NM_001918.5:c.294C>G, NM_001918.5:c.272_275delCAGT, NM_001918.5:c.126T>G
DCLRE1C	Omenn syndrome/Severe combined immunodeficiency, Athabascan type	NM_001033855.3:c.1903dupA, NM_001033855.3:c.1639G>T, NM_001033855.3:c.1559dupA, NM_001033855.3:c.780+1delG, NM_001033855.3:c.597C>A, NM_001033855.3:c.457G>A, NM_001033855.3:c.2T>C
DDB2	Xeroderma Pigmentosum Group E	NM_000107.3:c.730A>G, NM_000107.3:c.818G>A, NM_000107.3:c.919G>T, NM_000107.3:c.937C>T
DHCR7	Smith-Lemli-Opitz syndrome	NM_001360.3:c.1342G>A, NM_001360.3:c.1337G>A, NM_001360.3:c.1228G>A, NM_001360.3:c.1210C>T, NM_001360.3:c.1055G>A, NM_001360.3:c.1054C>T, NM_001360.3:c.1031G>A, NM_001360.3:c.976G>T, NM_001360.3:c.964-1G>C, NM_001360.3:c.907G>A, NM_001360.3:c.904T>C, NM_001360.3:c.866C>T, NM_001360.3:c.841G>A, NM_001360.3:c.839A>G, NM_001360.3:c.832-1G>C, NM_001360.3:c.744G>T, NM_001360.3:c.730G>A, NM_001360.3:c.725G>A, NM_001360.3:c.724C>T, NM_001360.3:c.682C>T, NM_001360.3:c.506C>T, NM_001360.3:c.461C>G, NM_001360.3:c.453G>A, NM_001360.3:c.452G>A, NM_001360.3:c.356A>T, NM_001360.3:c.292C>T, NM_001360.3:c.278C>T, NM_001360.3:c.151C>T, NM_001360.3:c.1A>G
DHDDS	Retinitis pigmentosa 59	NM_205861.3:c.124A>G, NM_205861.3:c.328delA, NM_205861.3:c.995C>G
DLD	Dihydrolipoamide dehydrogenase deficiency	NM_000108.5:c.105_106insA, NM_000108.5:c.913_923delACTTGTGATGT, NM_000108.5:c.1483A>G
DMD	Duchenne muscular dystrophy	NM_004006.3:c.10774delA, NM_004006.3:c.10454delT, NM_004006.3:c.10453_10454delCT, NM_004006.3:c.10447_10448delTC, NM_004006.3:c.10141C>T, NM_004006.3:c.10086+1G>A, NM_004006.3:c.10033C>T, NM_004006.3:c.9854_9863delTGAGACTGGA, NM_004006.3:c.9862G>T, NM_004006.3:c.9851G>A, NM_004006.3:c.9650-2A>G, NM_004006.3:c.9568C>T, NM_004006.3:c.9564-1G>A, NM_004006.3:c.9380C>G, NM_004006.3:c.9361+1G>C, NM_004006.3:c.9361+1G>A, NM_004006.3:c.9346C>T, NM_004006.3:c.9337C>T, NM_004006.3:c.9164-1G>T, NM_004006.3:c.9164-1G>C, NM_004006.3:c.8944C>T, NM_004006.3:c.8713C>T, NM_004006.3:c.8668G>A, NM_004006.3:c.8656C>T, NM_004006.3:c.8652_8653delCT, NM_004006.3:c.8608C>T, NM_004006.3:c.8464C>T, NM_004006.3:c.8443C>T, NM_004006.3:c.8374_8375delAA, NM_004006.3:c.8358G>A, NM_004006.3:c.8086delC, NM_004006.3:c.8069T>G, NM_004006.3:c.8064_8065delTA, NM_004006.3:c.7922delA, NM_004006.3:c.7894C>T, NM_004006.3:c.7771G>T, NM_004006.3:c.7764dupT, NM_004006.3:c.7683G>A, NM_004006.3:c.7682G>A, NM_004006.3:c.6986dupA, NM_004006.3:c.6982A>T, NM_004006.3:c.6964delG, NM_004006.3:c.6943G>T, NM_004006.3:c.6936delA, NM_004006.3:c.6906G>A, NM_004006.3:c.6834delT, NM_004006.3:c.6763-2A>G, NM_004006.3:c.6391_6392dupCA, NM_004006.3:c.6391_6392delCA, NM_004006.3:c.6373C>T, NM_004006.3:c.6340A>T, NM_004006.3:c.6292C>T, NM_004006.3:c.6238delC, NM_004006.3:c.6226G>T, NM_004006.3:c.6182delC, NM_004006.3:c.6014_6017delCTCA, NM_004006.3:c.6000T>A, NM_004006.3:c.5922+3G>C, NM_004006.3:c.5899C>T, NM_004006.3:c.5807T>A, NM_004006.3:c.5773G>T, NM_004006.3:c.5697delA, NM_004006.3:c.5671A>T, NM_004006.3:c.5640T>A, NM_004006.3:c.5570_5571dupAA, NM_004006.3:c.5554C>T, NM_004006.3:c.5530C>T, NM_004006.3:c.5363C>G, NM_004006.3:c.5353C>T, NM_004006.3:c.5313dupT, NM_004006.3:c.5287C>T, NM_004006.3:c.4843A>T, NM_004006.3:c.4806A>T, NM_004006.3:c.4735G>T, NM_004006.3:c.4518+5G>A, NM_004006.3:c.4500delA, NM_004006.3:c.4486delG, NM_004006.3:c.4471_4472delAA, NM_004006.3:c.4409_4412dupGTCT, NM_004006.3:c.4405C>T, NM_004006.3:c.4375C>T, NM_004006.3:c.4117C>T, NM_004006.3:c.4071G>C, NM_004006.3:c.3779_3783delCTTTG, NM_004006.3:c.3747delG, NM_004006.3:c.3697delC, NM_004006.3:c.3639dupA, NM_004006.3:c.3432+3A>G, NM_004006.3:c.3432+1G>A, NM_004006.3:c.3295C>T, NM_004006.3:c.3276+1G>A, NM_004006.3:c.3246_3247insTTTCTAAAAA, NM_004006.3:c.3124A>T, NM_004006.3:c.3121C>T, NM_004006.3:c.3087G>A, NM_004006.3:c.3076G>T, NM_004006.3:c.3022A>T, NM_004006.3:c.2929dupC, NM_004006.3:c.2866C>T, NM_004006.3:c.2815_2816delTT, NM_004006.3:c.2816T>A, NM_004006.3:c.2804-1G>A, NM_004006.3:c.2804-2A>T, NM_004006.3:c.2803+1G>T, NM_004006.3:c.2803+1G>A, NM_004006.3:c.2758C>T, NM_004006.3:c.2755A>T, NM_004006.3:c.2650C>T, NM_004006.3:c.2547delT, NM_004006.3:c.2523delA, NM_004006.3:c.2484T>G, NM_004006.3:c.2482T>G, NM_004006.3:c.2479delG, NM_004006.3:c.2380+2T>C, NM_004006.3:c.2380+1G>C, NM_004006.3:c.2332C>T, NM_004006.3:c.2302C>T, NM_004006.3:c.2294_2297delCCAT, NM_004006.3:c.2281_2285delGAAAA, NM_004006.3:c.2169-3delT, NM_004006.3:c.2137C>T, NM_004006.3:c.2125delC, NM_004006.3:c.1900_1903dupAAGT, NM_004006.3:c.1900A>T, NM_004006.3:c.1886C>A, NM_004006.3:c.1734delA, NM_004006.3:c.1529_1530delTC, NM_004006.3:c.1489C>T, NM_004006.3:c.1371delG, NM_004006.3:c.1341_1342dupAG, NM_004006.3:c.1332-9A>G, NM_004006.3:c.1306dupG, NM_004006.3:c.1286C>A, NM_004006.3:c.1261C>T, NM_004006.3:c.1070delC, NM_004006.3:c.1048G>T, NM_004006.3:c.1012G>T,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_004006.3:c.676_678delAAG, NM_004006.3:c.627delA, NM_004006.3:c.615T>A, NM_004006.3:c.583C>T, NM_004006.3:c.530+1delG, NM_004006.3:c.489G>A, NM_004006.3:c.433C>T, NM_004006.3:c.412_413delAA, NM_004006.3:c.220delC, NM_004006.3:c.204dupC, NM_004006.3:c.199G>T, NM_004006.3:c.160_162delCTC, NM_004006.3:c.137_138dupAT, NM_004006.3:c.137A>T, NM_004006.3:c.133C>T, CNV deletions/ duplications
DPYD	Dihydropyrimidine Dehydrogenase Deficiency	NM_000110.4:c.1905+1G>A, NM_000110.4:c.1679T>G, NM_000110.4:c.1109_1110delTA, NM_000110.4:c.775A>G, NM_000110.4:c.703C>T, NM_000110.4:c.299_302delTCAT, NM_000110.4:c.257C>T
DYSF	Muscular dystrophy, limb-girdle, type 2B	NM_001130987.2:c.203_204delTGinsAT, NM_001130987.2:c.289A>C, NM_001130987.2:c.394_395delCC, NM_001130987.2:c.547C>T, NM_001130987.2:c.605C>A, NM_001130987.2:c.661C>G, NM_001130987.2:c.706C>T, NM_001130987.2:c.759+1G>C, NM_001130987.2:c.797G>A, NM_001130987.2:c.853C>T, NM_001130987.2:c.991G>A, NM_001130987.2:c.991G>T, NM_001130987.2:c.1033+1G>A, NM_001130987.2:c.1149+1G>A, NM_001130987.2:c.1216G>C, NM_001130987.2:c.1312T>C, NM_001130987.2:c.1372G>A, NM_001130987.2:c.1380+2T>C, NM_001130987.2:c.1464C>A, NM_001130987.2:c.1487_1488insA, NM_001130987.2:c.1494-2A>G, NM_001130987.2:c.1494-1G>A, NM_003494.4:c.1481-1G>A, NM_001130987.2:c.1609G>A, NM_001130987.2:c.1674delA, NM_001130987.2:c.1692+2T>A, NM_001130987.2:c.1717C>T, NM_001130987.2:c.1867C>T, NM_001130987.2:c.1888C>T, NM_001130987.2:c.1915G>C, NM_001130987.2:c.1927G>T, NM_001130987.2:c.2921_2925delACCAG, NM_001130987.2:c.2923C>T, NM_001130987.2:c.3020C>T, NM_001130987.2:c.3051G>T, NM_001130987.2:c.3095A>G, NM_001130987.2:c.3119G>A, NM_001130987.2:c.3166C>T, NM_001130987.2:c.3167G>C, NM_001130987.2:c.3175C>T, NM_001130987.2:c.3184C>T, NM_001130987.2:c.3212T>G, NM_001130987.2:c.3229-2A>T, NM_001130987.2:c.3235_3236insAGGCGG, NM_001130987.2:c.3498_3499delTGinsAA, NM_001130987.2:c.3531C>A, NM_001130987.2:c.3532C>T, NM_001130987.2:c.3694delC, NM_001130987.2:c.3762delA, NM_001130987.2:c.3778C>T, NM_001130987.2:c.3859G>T, NM_001130987.2:c.3913A>T, NM_001130987.2:c.3946A>G, NM_001130987.2:c.3957delG, NM_001130987.2:c.4011delC, NM_001130987.2:c.4039C>G, NM_001130987.2:c.4057G>A, NM_001130987.2:c.4144C>T, NM_001130987.2:c.4159_4160delTG, NM_001130987.2:c.4253C>A, NM_001130987.2:c.4307G>A, NM_001130987.2:c.4873C>T, NM_001130987.2:c.5102C>T, NM_001130987.2:c.5194C>T, NM_001130987.2:c.5318A>G, NM_001130987.2:c.5383C>T, NM_001130987.2:c.5458-2A>C, NM_001130987.2:c.5546G>A, NM_001130987.2:c.5546+1G>T, NM_001130987.2:c.5614G>T, NM_001130987.2:c.5626G>A, NM_001130987.2:c.5642+1G>A, NM_001130987.2:c.5710delG, NM_001130987.2:c.5761C>T, NM_001130987.2:c.5812_5813delGA, NM_001130987.2:c.5830C>T, NM_001130987.2:c.5952_5955delCCAG, NM_001130987.2:c.6095_6096insA, NM_001130987.2:c.6109G>T, NM_001130987.2:c.6115C>T, NM_001130987.2:c.6116G>A, NM_001130987.2:c.6241C>T, NM_001130987.2:c.6320C>T, NM_001130987.2:c.4993delG
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked	NM_001399.5:c.181T>C, NM_001399.5:c.183C>G, NM_001399.5:c.187G>A, NM_001399.5:c.206G>T, NM_001399.5:c.463C>T, NM_001399.5:c.466C>T, NM_001399.5:c.467G>A, NM_001399.5:c.573_574insT, NM_001399.5:c.671G>C, NM_001399.5:c.826C>T, NM_001399.5:c.1045G>A
EIF2AK3	Wolcott-Rallison Syndrome	NM_004836.7:c.1763G>A, NM_004836.7:c.994G>T
EMD	Emery-Dreifuss Muscular Dystrophy 1, X-Linked	NM_000117.3:c.547C>A, NM_000117.3:c.630_634delCCGTG
ERCC2	Xeroderma Pigmentosum Group D	NM_000400.4:c.2230_2233dupCTAG, NM_000400.4:c.2176C>T, NM_000400.4:c.2047C>T, NM_000400.4:c.1972C>T, NM_000400.4:c.1703_1704delTT, NM_000400.4:c.1621A>C, NM_000400.4:c.1454T>C, NM_000400.4:c.1381C>G, NM_000400.4:c.1354C>T, NM_000400.4:c.1308-1G>A, NM_000400.4:c.1304T>G, NM_000400.4:c.950-2A>G, NM_000400.4:c.949+1G>A, NM_000400.4:c.719-1G>A, NM_000400.4:c.567G>A, NM_000400.4:c.183+2T>A
ERCC3	Xeroderma Pigmentosum Group B	NM_000122.2:c.1858delG, NM_000122.2:c.1757_1758delAG, NM_000122.2:c.1757delA, NM_000122.2:c.1633C>T, NM_000122.2:c.1273C>T, NM_000122.2:c.296T>C
ERCC4	Xeroderma Pigmentosum Group F	NM_005236.3:c.2T>C, NM_005236.3:c.49G>T, NM_005236.3:c.538_539delAG, NM_005236.3:c.706T>C, NM_005236.3:c.1461_1462insA, NM_005236.3:c.2280_2283delGTTT, NM_005236.3:c.2395C>T
ERCC5	Xeroderma pigmentosum, group G/Cockayne syndrome	NM_000123.4:c.88+2T>C, NM_000123.4:c.215C>A, NM_000123.4:c.381-2A>G, NM_000123.4:c.406C>T, NM_000123.4:c.463_464insA, NM_000123.4:c.526C>T, NM_000123.4:c.787C>T, NM_000123.4:c.2143_2144insA, NM_000123.4:c.2375C>T, NM_000123.4:c.2573T>C, NM_000123.4:c.2620G>A, NM_000123.4:c.2743delA
ERCC6	Cockayne syndrome	NM_000124.4:c.3862C>T, NM_000124.4:c.3591_3592dupGA, NM_000124.4:c.3284C>G, NM_000124.4:c.2587C>T, NM_000124.4:c.2203C>T, NM_000124.4:c.2047C>T, NM_000124.4:c.1550G>A, NM_000124.4:c.1357C>T, NM_000124.4:c.422+1G>A, NM_000124.4:c.207dupG, NM_000124.4:c.48_49delCT

GENESeeker

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
ERCC8	Cockayne, type A	NM_000082.4:c.966C>A, NM_000082.4:c.618-1G>A, NM_000082.4:c.613G>C, NM_000082.4:c.593_594dupAT, NM_000082.4:c.37G>T
ESCO2	Roberts Syndrome	NM_001017420.3:c.291_292insGA, NM_001017420.3:c.306_307delAA, NM_001017420.3:c.505C>T, NM_001017420.3:c.604C>T, NM_001017420.3:c.874_877delGACA, NM_001017420.3:c.877_878delAG, NM_001017420.3:c.1269G>A, NM_001017420.3:c.1596_1597insT, NM_001017420.3:c.1615T>G
ETFA	Glutaric acidemia IIA	NM_000126.4:c.797C>T, NM_000126.4:c.470T>G
ETFB	Glutaric Acidemia, Type 2B	NM_001985.3:c.614_616delAGA, NM_001985.3:c.491G>A, NM_001985.3:c.490C>T, NM_001985.3:c.382G>A, NM_001985.3:c.278_279insG, NM_001985.3:c.61C>T, NM_001985.3:c.58-53_58-52insG
ETFDH	Glutaric aciduria type IIC	NM_004453.4:c.2T>C, NM_004453.4:c.250G>A, NM_004453.4:c.413T>G, NM_004453.4:c.508G>T, NM_004453.4:c.524G>T, NM_004453.4:c.643G>A, NM_004453.4:c.1001T>C, NM_004453.4:c.1234G>T, NM_004453.4:c.1351G>C, NM_004453.4:c.1367C>T, NM_004453.4:c.1568_1569delCT, NM_004453.4:c.1773_1774delAT, NM_004453.4:c.1822delG, NM_004453.4:c.1832G>A
ETHE1	Ethylmalonic Encephalopathy	NM_014297.5:c.604dupG, NM_014297.5:c.554T>G, NM_014297.5:c.488G>A, NM_014297.5:c.487C>T, NM_014297.5:c.440_450delACAGCATGGCC, NM_014297.5:c.221dupA
EYS	Retinitis pigmentosa 25	NM_001142800.2:c.9405T>A, NM_001142800.2:c.9299_9302delCTCA, NM_001142800.2:c.9036delT, NM_001142800.2:c.8834G>A, NM_001142800.2:c.8648_8655delCATGCAGA, NM_001142800.2:c.8629_8632dupACAG, NM_001142800.2:c.8569G>T, NM_001142800.2:c.8408dupA, NM_001142800.2:c.7822C>T, NM_001142800.2:c.7095T>G, NM_001142800.2:c.6170delA, NM_001142800.2:c.6102dupT, NM_001142800.2:c.5928-2A>G, NM_001142800.2:c.5857G>T, NM_001142800.2:c.5757dupT, NM_001142800.2:c.5044G>T, NM_001142800.2:c.4597_4613delTCAAGCAACCAGAGACT, NM_001142800.2:c.4462_4469dupAGCCCCTC, NM_001142800.2:c.4350_4356delTATAGCT, NM_001142800.2:c.4120C>T, NM_001142800.2:c.4045C>T, NM_001142800.2:c.3329C>G, NM_001142800.2:c.2826_2827delAT, NM_001142800.2:c.1211dupA, NM_001142800.2:c.863-4_863-3insT, NM_001142800.2:c.571dupA, NM_001142800.2:c.490C>T, NM_001142800.2:c.232delT, NM_001142800.2:c.103C>T
F11	Factor XI deficiency, autosomal recessive	NM_000128.4:c.166T>C, NM_000128.4:c.403G>T, NM_000128.4:c.438C>A, NM_000128.4:c.595+3A>G, NM_000128.4:c.731A>G, NM_000128.4:c.809A>T, NM_000128.4:c.901T>C, NM_000128.4:c.1211C>A, NM_000128.4:c.1613C>T, NM_000128.4:c.1693G>A
F8	Hemophilia A	Intron 22 inversion, NM_000132.4:c.7033_7040delTGCGAGGC, NM_000132.4:c.7034G>A, NM_000132.4:c.7031G>A, NM_000132.4:c.7030G>T, NM_000132.4:c.7030G>A, NM_000132.4:c.7021G>T, NM_000132.4:c.7016G>T, NM_000132.4:c.7012delC, NM_000132.4:c.6996G>A, NM_000132.4:c.6995G>C, NM_000132.4:c.6988delC, NM_000132.4:c.6986C>T, NM_000132.4:c.6969_6977delCTACCTTCG, NM_000132.4:c.6976C>G, NM_000132.4:c.6921delC, NM_000132.4:c.6919_6920delGA, NM_000132.4:c.6914_6918delATCAA, NM_000132.4:c.6915delT, NM_000132.4:c.6905T>C, NM_000132.4:c.6904T>G, NM_000132.4:c.6901-2A>G, NM_000132.4:c.6900+1G>A, NM_000132.4:c.6887delA, NM_000132.4:c.6870G>A, NM_000132.4:c.6869G>T, NM_000132.4:c.6857_6867delATGGCCATCAG, NM_000132.4:c.6865C>T, NM_000132.4:c.6842T>C, NM_000132.4:c.6839T>C, NM_000132.4:c.6836T>G, NM_000132.4:c.6836T>C, NM_000132.4:c.6827T>G, NM_000132.4:c.6825T>A, NM_000132.4:c.6797delG, NM_000132.4:c.6797G>A, NM_000132.4:c.6796G>A, NM_000132.4:c.6780_6788delAGGAGTAAC, NM_000132.4:c.6786_6787insCAA, NM_000132.4:c.6760delC, NM_000132.4:c.6760C>T, NM_000132.4:c.6752T>A, NM_000132.4:c.6746T>G, NM_000132.4:c.6743G>C, NM_000132.4:c.6740_6741delAG, NM_000132.4:c.6739G>T, NM_000132.4:c.6738delA, NM_000132.4:c.6574+5G>C, NM_000132.4:c.6574+3A>C, NM_000132.4:c.6574+1G>T, NM_000132.4:c.6574+1G>A, NM_000132.4:c.6565_6566delGA, NM_000132.4:c.6551A>T, NM_000132.4:c.6548T>G, NM_000132.4:c.6544C>G, NM_000132.4:c.6537C>G, NM_000132.4:c.6533G>A, NM_000132.4:c.6520C>G, NM_000132.4:c.6517_6519dupACT, NM_000132.4:c.6515C>G, NM_000132.4:c.6501delC, NM_000132.4:c.6497delG, NM_000132.4:c.6494delC, NM_000132.4:c.6488T>G, NM_000132.4:c.6482C>T, NM_000132.4:c.6482C>A, NM_000132.4:c.6477delT, NM_000132.4:c.6469_6470delAA, NM_000132.4:c.6468_6469delCA, NM_000132.4:c.6465delA, NM_000132.4:c.6464_6465delAA, NM_000132.4:c.6449A>T, NM_000132.4:c.6430-3C>G, NM_000132.4:c.6273+1G>A, NM_000132.4:c.6269T>A, NM_000132.4:c.6263C>T, NM_000132.4:c.6253G>T, NM_000132.4:c.6250A>T, NM_000132.4:c.6243G>C, NM_000132.4:c.6242G>C, NM_000132.4:c.6239C>T, NM_000132.4:c.6213A>T, NM_000132.4:c.6194G>A, NM_000132.4:c.6136dupA, NM_000132.4:c.6120_6135delTCAGACTCCCCTGGGA, NM_000132.4:c.6134G>T, NM_000132.4:c.6130delC, NM_000132.4:c.6120T>A, NM_000132.4:c.6116_6117delAG, NM_000132.4:c.6116-2A>G, NM_000132.4:c.6115+6T>A, NM_000132.4:c.6115+4A>G, NM_000132.4:c.6115+3G>T, NM_000132.4:c.6115+2T>C, NM_000132.4:c.6115+1G>A, NM_000132.4:c.6107A>G, NM_000132.4:c.6099delT, NM_000132.4:c.6094C>T, NM_000132.4:c.6089dupG, NM_000132.4:c.6084delG, NM_000132.4:c.6078_6079delTG, NM_000132.4:c.6070dupC, NM_000132.4:c.6046C>G, NM_000132.4:c.6037G>A, NM_000132.4:c.6016G>T, NM_000132.4:c.5999G>C, NM_000132.4:c.5964_5967dupGGAG, NM_000132.4:c.5960_5961delAA,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000132.4:c.5953delC, NM_000132.4:c.5954G>C, NM_000132.4:c.5939A>C, NM_000132.4:c.5934T>G, NM_000132.4:c.5923dupA, NM_000132.4:c.5924T>A, NM_000132.4:c.5914_5915delAT, NM_000132.4:c.5894G>T, NM_000132.4:c.5891T>C, NM_000132.4:c.5888T>C, NM_000132.4:c.5884T>G, NM_000132.4:c.5881T>A, NM_000132.4:c.5879G>T, NM_000132.4:c.5869C>T, NM_000132.4:c.5861_5866delCTCAGG, NM_000132.4:c.5853A>C, NM_000132.4:c.5833A>G, NM_000132.4:c.5825G>T, NM_000132.4:c.5816C>T, NM_000132.4:c.5816C>A, NM_000132.4:c.5766C>A, NM_000132.4:c.5752delT, NM_000132.4:c.5721C>G, NM_000132.4:c.5719dupA, NM_000132.4:c.5719A>T, NM_000132.4:c.5712G>C, NM_000132.4:c.5697delC, NM_000132.4:c.5696dupT, NM_000132.4:c.5689_5690delCT, NM_000132.4:c.5686G>C, NM_000132.4:c.5680G>A, NM_000132.4:c.5675dupT, NM_000132.4:c.5674G>A, NM_000132.4:c.5348_5357delGAGCAGAAGT, NM_000132.4:c.5345T>G, NM_000132.4:c.5343T>A, NM_000132.4:c.5339C>T, NM_000132.4:c.5337delG, NM_000132.4:c.5330T>C, NM_000132.4:c.5321A>T, NM_000132.4:c.5308G>A, NM_000132.4:c.5301C>A, NM_000132.4:c.5291A>G, NM_000132.4:c.5271delT, NM_000132.4:c.5269T>C, NM_000132.4:c.5254delG, NM_000132.4:c.5251A>T, NM_000132.4:c.5243delA, NM_000132.4:c.5227_5228delAG, NM_000132.4:c.5220-1G>A, NM_000132.4:c.5012G>A, NM_000132.4:c.5010delT, NM_000132.4:c.4999delC, NM_000132.4:c.4996C>T, NM_000132.4:c.4987A>T, NM_000132.4:c.4979C>T, NM_000132.4:c.4969C>T, NM_000132.4:c.4942C>T, NM_000132.4:c.4935G>A, NM_000132.4:c.4934G>A, NM_000132.4:c.4926delA, NM_000132.4:c.4925A>G, NM_000132.4:c.4922dupT, NM_000132.4:c.4918G>T, NM_000132.4:c.4899delT, NM_000132.4:c.4895dupT, NM_000132.4:c.4895delT, NM_000132.4:c.4864G>A, NM_000132.4:c.4858delC, NM_000132.4:c.4848delC, NM_000132.4:c.4841delA, NM_000132.4:c.4828G>T, NM_000132.4:c.4814C>A, NM_000132.4:c.4806delA, NM_000132.4:c.4805_4806delAA, NM_000132.4:c.4798A>T, NM_000132.4:c.4794G>T, NM_000132.4:c.4770T>A, NM_000132.4:c.4719_4729delTGCAAAGACTC, NM_000132.4:c.4720delG, NM_000132.4:c.4712_4715delAAAG, NM_000132.4:c.4697_4701dupTGAGA, NM_000132.4:c.4694_4697delTTCT, NM_000132.4:c.4687delG, NM_000132.4:c.4686delA, NM_000132.4:c.4672_4675delAACA, NM_000132.4:c.4662_4663delGA, NM_000132.4:c.4658delA, NM_000132.4:c.4619delT, NM_000132.4:c.4561C>T, NM_000132.4:c.4549_4550delGT, NM_000132.4:c.4543_4544delCCinsA, NM_000132.4:c.4542delT, NM_000132.4:c.4531G>A, NM_000132.4:c.4519delA, NM_000132.4:c.4512delG, NM_000132.4:c.4492_4496delGTTCT, NM_000132.4:c.4492_4493delGT, NM_000132.4:c.4492delG, NM_000132.4:c.4483delG, NM_000132.4:c.4483G>T, NM_000132.4:c.4474A>T, NM_000132.4:c.4473C>G, NM_000132.4:c.4473C>A, NM_000132.4:c.4460delA, NM_000132.4:c.4450delA, NM_000132.4:c.4446dupG, NM_000132.4:c.4430_4431delAG, NM_000132.4:c.4425_4426delAA, NM_000132.4:c.4423C>T, NM_000132.4:c.4408G>T, NM_000132.4:c.4382_4383delAC, NM_000132.4:c.4363C>T, NM_000132.4:c.4345delG, NM_000132.4:c.4345G>T, NM_000132.4:c.4339delG, NM_000132.4:c.4339dupG, NM_000132.4:c.4318delT, NM_000132.4:c.4296_4300delTTCTC, NM_000132.4:c.4280delT, NM_000132.4:c.4272delC, NM_000132.4:c.4265_4266delAT, NM_000132.4:c.4242dupA, NM_000132.4:c.4241C>A, NM_000132.4:c.4201C>T, NM_000132.4:c.4199delC, NM_000132.4:c.4156C>T, NM_000132.4:c.4103delC, NM_000132.4:c.4094_4100delATTTGAC, NM_000132.4:c.4072C>T, NM_000132.4:c.4035delA, NM_000132.4:c.4006C>T, NM_000132.4:c.3994_3997delAGAG, NM_000132.4:c.3991_3992delAA, NM_000132.4:c.3984dupA, NM_000132.4:c.3982C>T, NM_000132.4:c.3967C>T, NM_000132.4:c.3964C>T, NM_000132.4:c.3940A>C, NM_000132.4:c.3922G>T, NM_000132.4:c.3913C>T, NM_000132.4:c.3907_3911delACCAA, NM_000132.4:c.3902delA, NM_000132.4:c.3887delT, NM_000132.4:c.3863dupC, NM_000132.4:c.3860delT, NM_000132.4:c.3851_3852delCA, NM_000132.4:c.3844A>T, NM_000132.4:c.3833delA, NM_000132.4:c.3830delC, NM_000132.4:c.3827C>G, NM_000132.4:c.3772delT, NM_000132.4:c.3766G>T, NM_000132.4:c.3756delG, NM_000132.4:c.3736delC, NM_000132.4:c.3735_3736delCCinsATTTCT, NM_000132.4:c.3710delC, NM_000132.4:c.3652delG, 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NM_000132.4:c.2419dupA, NM_000132.4:c.2409delT, NM_000132.4:c.2404C>T, NM_000132.4:c.2397delT, NM_000132.4:c.2384_2388delGAACA, NM_000132.4:c.2383A>G, NM_000132.4:c.2373dupG, NM_000132.4:c.2360delA, NM_000132.4:c.2338delA, NM_000132.4:c.2102_2106delTGGAA, NM_000132.4:c.2097G>A,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000132.4:c.2096T>A, NM_000132.4:c.2095A>T, NM_000132.4:c.2095A>G, NM_000132.4:c.2095A>C, NM_000132.4:c.2089_2090delGT, NM_000132.4:c.2090T>A, NM_000132.4:c.2072C>T, NM_000132.4:c.2071C>A, NM_000132.4:c.2066T>G, NM_000132.4:c.2058_2059delAC, NM_000132.4:c.2060T>C, NM_000132.4:c.2057C>G, NM_000132.4:c.2032A>T, NM_000132.4:c.2029T>C, NM_000132.4:c.2015_2017delTCT, NM_000132.4:c.2000delT, NM_000132.4:c.1996_1999delGACT, NM_000132.4:c.1996_1999dupGACT, NM_000132.4:c.1990_1991delCA, NM_000132.4:c.1991A>C, NM_000132.4:c.1988C>T, NM_000132.4:c.1985G>C, NM_000132.4:c.1952A>C, NM_000132.4:c.1947_1950delTTTG, NM_000132.4:c.1941_1944delAGTT, NM_000132.4:c.1934A>C, NM_000132.4:c.1925_1928delATAG, NM_000132.4:c.1913G>A, NM_000132.4:c.1912G>A, NM_000132.4:c.1904-1G>A, NM_000132.4:c.1904-37G>A, NM_000132.4:c.1752+5G>C, NM_000132.4:c.1736A>T, NM_000132.4:c.1726G>T, NM_000132.4:c.1703G>T, NM_000132.4:c.1688C>G, NM_000132.4:c.1682A>C, NM_000132.4:c.1681G>A, NM_000132.4:c.1675G>T, NM_000132.4:c.1667T>A, NM_000132.4:c.1661G>A, NM_000132.4:c.1653T>G, NM_000132.4:c.1640G>A, NM_000132.4:c.1639T>C, NM_000132.4:c.1630G>A, NM_000132.4:c.1619C>G, NM_000132.4:c.1618C>A, NM_000132.4:c.1596dupG, NM_000132.4:c.1596G>A, NM_000132.4:c.1595G>A, NM_000132.4:c.1594T>G, NM_000132.4:c.1585A>G, NM_000132.4:c.1560delT, NM_000132.4:c.1538-1G>T, NM_000132.4:c.1538-2A>T, NM_000132.4:c.1477A>G, NM_000132.4:c.1467_1472dupCAGACC, NM_000132.4:c.1463C>T, NM_000132.4:c.1463C>G, NM_000132.4:c.1443+3A>C, NM_000132.4:c.1443+2T>C, NM_000132.4:c.1442_1443dupTG, NM_000132.4:c.1443+1G>A, NM_000132.4:c.1440_1441insA, NM_000132.4:c.1438_1439delCT, NM_000132.4:c.1432G>A, NM_000132.4:c.1420G>T, NM_000132.4:c.1410_1413delTTTA, NM_000132.4:c.1406G>C, NM_000132.4:c.1400T>G, NM_000132.4:c.1397G>A, NM_000132.4:c.1394C>G, NM_000132.4:c.1390G>T, NM_000132.4:c.1357G>T, NM_000132.4:c.1348T>G, NM_000132.4:c.1338delA, NM_000132.4:c.1336dupC, NM_000132.4:c.1337G>C, NM_000132.4:c.1337G>A, NM_000132.4:c.1331_1332delAA, NM_000132.4:c.1331A>C, NM_000132.4:c.1325A>G, NM_000132.4:c.1324T>C, NM_000132.4:c.1324T>A, NM_000132.4:c.1316G>A, NM_000132.4:c.1311delG, NM_000132.4:c.1301G>A, NM_000132.4:c.1293delG, NM_000132.4:c.1234T>C, NM_000132.4:c.1214T>G, NM_000132.4:c.1207C>G, NM_000132.4:c.1203G>A, NM_000132.4:c.1200_1201delTT, NM_000132.4:c.1202G>A, NM_000132.4:c.1189dupC, NM_000132.4:c.1187A>T, NM_000132.4:c.1175C>G, NM_000132.4:c.1175C>A, NM_000132.4:c.1172G>C, NM_000132.4:c.1165delC, NM_000132.4:c.1090G>A, NM_000132.4:c.1086G>A, NM_000132.4:c.1077_1080delTGAA, NM_000132.4:c.1078_1079delGA, NM_000132.4:c.1042T>C, NM_000132.4:c.986G>T, NM_000132.4:c.985dupT, NM_000132.4:c.985delT, NM_000132.4:c.986G>C, NM_000132.4:c.986G>A, NM_000132.4:c.974_975delTT, NM_000132.4:c.967G>A, NM_000132.4:c.948_951delAACA, NM_000132.4:c.943delG, NM_000132.4:c.941C>T, NM_000132.4:c.935delT, NM_000132.4:c.919delA, NM_000132.4:c.920T>G, NM_000132.4:c.912C>T, NM_000132.4:c.907delG, NM_000132.4:c.902G>C, NM_000132.4:c.899A>T, NM_000132.4:c.899A>C, NM_000132.4:c.889delG, NM_000132.4:c.886C>T, NM_000132.4:c.883T>C, NM_000132.4:c.872A>G, NM_000132.4:c.871G>T, NM_000132.4:c.850G>T, NM_000132.4:c.849delT, NM_000132.4:c.850G>A, NM_000132.4:c.836T>A, NM_000132.4:c.832G>A, NM_000132.4:c.824A>G, NM_000132.4:c.822G>A, NM_000132.4:c.820T>C, NM_000132.4:c.796G>T, NM_000132.4:c.788-1G>T, NM_000132.4:c.788-1G>C, NM_000132.4:c.788-1G>A, NM_000132.4:c.788-2A>T, NM_000132.4:c.787+2T>C, NM_000132.4:c.787G>C, NM_000132.4:c.779C>G, NM_000132.4:c.775A>T, NM_000132.4:c.770_771insCC, NM_000132.4:c.764G>A, NM_000132.4:c.760A>T, NM_000132.4:c.755C>A, NM_000132.4:c.729delT, NM_000132.4:c.709C>T, NM_000132.4:c.695_698delAGAA, NM_000132.4:c.688_689delGA, NM_000132.4:c.685_686delTC, NM_000132.4:c.680G>A, NM_000132.4:c.676A>T, NM_000132.4:c.589_591delGTA, NM_000132.4:c.577G>A, NM_000132.4:c.571C>T, NM_000132.4:c.566C>A, NM_000132.4:c.557_559delACT, NM_000132.4:c.560T>A, NM_000132.4:c.557A>G, NM_000132.4:c.556G>A, NM_000132.4:c.553A>G, NM_000132.4:c.545A>T, NM_000132.4:c.535T>C, NM_000132.4:c.532C>G, NM_000132.4:c.525C>A, NM_000132.4:c.519_523delTACCT, NM_000132.4:c.514_515insTCAAGATA, NM_000132.4:c.515G>A, NM_000132.4:c.514T>C, NM_000132.4:c.493C>T, NM_000132.4:c.489T>A, NM_000132.4:c.476T>C, NM_000132.4:c.472C>T, NM_000132.4:c.471G>A, NM_000132.4:c.440_448dupTCTTCCCTG, NM_000132.4:c.446delC, NM_000132.4:c.440T>A, NM_000132.4:c.435_436insTTT, NM_000132.4:c.433G>C, NM_000132.4:c.430G>T, NM_000132.4:c.421G>T, NM_000132.4:c.415C>T, NM_000132.4:c.407A>C, NM_000132.4:c.405T>A, NM_000132.4:c.404A>G, NM_000132.4:c.403G>A, NM_000132.4:c.265+1G>T, NM_000132.4:c.265G>A, NM_000132.4:c.255_257delACC, NM_000132.4:c.250_255delAGGCCA, NM_000132.4:c.250A>G, NM_000132.4:c.230T>C, NM_000132.4:c.224delA, NM_000132.4:c.225T>A, NM_000132.4:c.223G>T, NM_000132.4:c.217T>C, NM_000132.4:c.214G>A, NM_000132.4:c.209T>C, NM_000132.4:c.202_207delACTCTG, NM_000132.4:c.201_202dupGA, NM_000132.4:c.203C>A, NM_000132.4:c.201G>T, NM_000132.4:c.199_200delAA, NM_000132.4:c.200A>C, NM_000132.4:c.199A>G, NM_000132.4:c.195C>A, NM_000132.4:c.185C>G, NM_000132.4:c.173delC, NM_000132.4:c.144-5C>G, NM_000132.4:c.144-11T>G, NM_000132.4:c.144-26A>T, NM_000132.4:c.143+1G>A, NM_000132.4:c.128dupT, NM_000132.4:c.120delC,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000132.4:c.98G>A, NM_000132.4:c.97T>G, NM_000132.4:c.88G>A, NM_000132.4:c.86T>G, NM_000132.4:c.77T>C, NM_000132.4:c.73delT, NM_000132.4:c.65G>C, NM_000132.4:c.1A>G
F9	Hemophilia B	NM_000133.4:c.19A>T, NM_000133.4:c.52T>C, NM_000133.4:c.79G>A, NM_000133.4:c.80A>T, NM_000133.4:c.82T>C, NM_000133.4:c.1031T>C, NM_000133.4:c.1136G>A, NM_000133.4:c.1150C>T
FAH	Tyrosinemia, type I	NM_000137.4:c.47A>T, NM_000137.4:c.103G>A, NM_000137.4:c.192G>T, NM_000137.4:c.401C>A, NM_000137.4:c.456G>A, NM_000137.4:c.554-1G>T, NM_000137.4:c.607-6T>G, NM_000137.4:c.707-1G>A, NM_000137.4:c.782C>T, NM_000137.4:c.786G>A, NM_000137.4:c.837+1G>A, NM_000137.4:c.938delC, NM_000137.4:c.982C>T, NM_000137.4:c.1009G>A, NM_000137.4:c.1021C>T, NM_000137.4:c.1027G>T, NM_000137.4:c.1062+5G>A, NM_000137.4:c.1069G>T, NM_000137.4:c.1090G>T, NM_000137.4:c.1141A>G
FANCA	Fanconi anemia, complementation group A	NM_000135.4:c.4130C>G, NM_000135.4:c.3788_3790delTCT, NM_000135.4:c.3763G>T, NM_000135.4:c.3558dupG, NM_000135.4:c.2303T>C, NM_000135.4:c.1115_1118delTTGG, NM_000135.4:c.233_236delTTGA
FANCC	Fanconi anemia, complementation group C	NM_000136.3:c.1642C>T, NM_000136.3:c.1487T>G, NM_000136.3:c.1103_1104delTG, NM_000136.3:c.1015delA, NM_000136.3:c.996+1G>T, NM_000136.3:c.416G>A, NM_000136.3:c.67delG, NM_000136.3:c.37C>T
FANCG	Fanconi anemia, complementation group G	NM_004629.2:c.1852_1853delAA, NM_004629.2:c.1795_1804delTGGATCCGTC, NM_004629.2:c.1480+1G>C, NM_004629.2:c.1077-2A>G, NM_004629.2:c.907_908dupCT, NM_004629.2:c.637_643delTACCGCC, NM_004629.2:c.510+1G>A, NM_004629.2:c.313G>T
FH	Fumarase Deficiency	NM_000143.4:c.1446_1449delAAAG, NM_000143.4:c.1431_1433dupAAA, NM_000143.4:c.1394A>G, NM_000143.4:c.1293delA, NM_000143.4:c.1255T>C, NM_000143.4:c.1236+1G>C, NM_000143.4:c.1200delT, NM_000143.4:c.1189G>A, NM_000143.4:c.1093A>G, NM_000143.4:c.1067T>A, NM_000143.4:c.901dupA, NM_000143.4:c.793G>A, NM_000143.4:c.760C>T, NM_000143.4:c.698G>A, NM_000143.4:c.697C>T, NM_000143.4:c.521C>G, NM_000143.4:c.320A>C, NM_000143.4:c.40dupC
FKRP	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	NM_024301.5:c.160C>T, NM_024301.5:c.235G>A, NM_024301.5:c.1154C>A, NM_024301.5:c.1343C>T, NM_024301.5:c.1387A>G
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	NM_001079802.2:c.411C>A, NM_001079802.2:c.509C>A, NM_001079802.2:c.1112A>G
FMR1	Fragile X syndrome	Premutation allele (CGG)n
G6PC1/ G6PC	Glycogen storage disease Ia (von Gierke disease)	NM_000151.4:c.47C>G, NM_000151.4:c.113A>T, NM_000151.4:c.229T>C, NM_000151.4:c.230+1G>C, NM_000151.4:c.247C>T, NM_000151.4:c.248G>A, NM_000151.4:c.370G>A, NM_000151.4:c.376_377insTA, NM_000151.4:c.447-1G>A, NM_000151.4:c.497T>G, NM_000151.4:c.508C>T, NM_000151.4:c.551G>A, NM_000151.4:c.562G>C, NM_000151.4:c.626A>G, NM_000151.4:c.883C>T, NM_000151.4:c.1039C>T
GAA	Glycogen storage disease II (Pompe disease)	NM_000152.5:c.118C>T, NM_000152.5:c.307T>G, NM_000152.5:c.525delT, NM_000152.5:c.546_546+3delGGTG, NM_000152.5:c.546G>A, NM_000152.5:c.546G>C, NM_000152.5:c.655G>A, NM_000152.5:c.697delA, NM_000152.5:c.710C>T, NM_000152.5:c.767_768insT, NM_000152.5:c.853C>T, NM_000152.5:c.877G>A, NM_000152.5:c.925G>A, NM_000152.5:c.953T>C, NM_000152.5:c.1064T>C, NM_000152.5:c.1115A>T, NM_000152.5:c.1316T>A, NM_000152.5:c.1327-2A>G, NM_000152.5:c.1407_1409delCAA, NM_000152.5:c.1430delT, NM_000152.5:c.1445C>T, NM_000152.5:c.1465G>A, NM_000152.5:c.1548G>A, NM_000152.5:c.1552-3C>G, NM_000152.5:c.1561G>A, NM_000152.5:c.1585_1586delTCinsGT, NM_000152.5:c.1634C>T, NM_000152.5:c.1644_1645insG, NM_000152.5:c.1799G>A, NM_000152.5:c.1827_1828insA, NM_000152.5:c.1846_1847insA, NM_000152.5:c.1912G>T, NM_000152.5:c.1927G>A, NM_000152.5:c.1933G>T, NM_000152.5:c.1935C>A, NM_000152.5:c.2012T>G, NM_000152.5:c.2015G>A, NM_000152.5:c.2041-1G>A, NM_000152.5:c.2063_2064insCGAGC, NM_000152.5:c.2105G>T, NM_000152.5:c.2237G>A, NM_000152.5:c.2238G>A, NM_000152.5:c.2238G>C, NM_000152.5:c.2512C>T, NM_000152.5:c.2543delC, NM_000152.5:c.2560C>T
GALC	Krabbe disease	NM_000153.4:c.2056T>C, NM_000153.4:c.1964delC, NM_000153.4:c.1814dupA, NM_000153.4:c.1796T>G, NM_000153.4:c.1723_1724insT, NM_000153.4:c.1700A>C, NM_000153.4:c.1695delT, NM_000153.4:c.1592G>A, NM_000153.4:c.1591C>T, NM_000153.4:c.1586C>T, NM_000153.4:c.1543G>A, NM_000153.4:c.1489+2_1489+3delTG, NM_000153.4:c.1488_1489+2delTGGT, NM_000153.4:c.1488_1489delTG, NM_000153.4:c.1472delA, NM_000153.4:c.1161+2T>G, NM_000153.4:c.1153G>T, NM_000153.4:c.1004A>G, NM_000153.4:c.953C>G,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000153.4:c.658C>T, NM_000153.4:c.655C>T, NM_000153.4:c.628A>T, NM_000153.4:c.582+1G>A, NM_000153.4:c.453G>A, NM_000153.4:c.430delA, NM_000153.4:c.388G>A, NM_000153.4:c.334A>G, NM_000153.4:c.332G>A, NM_000153.4:c.236G>A, NM_000153.4:c.205C>T
GALT	Galactosemia	NM_000155.4:c.17delC, NM_000155.4:c.41delCinsTT, NM_000155.4:c.71_72insA, NM_000155.4:c.113A>C, NM_000155.4:c.118G>T, NM_000155.4:c.130G>A, NM_000155.4:c.132delG, NM_000155.4:c.143G>C, NM_000155.4:c.152G>A, NM_000155.4:c.158G>A, NM_000155.4:c.199C>T, NM_000155.4:c.200G>A, NM_000155.4:c.203A>C, NM_000155.4:c.218_219delCT, NM_000155.4:c.221T>C, NM_000155.4:c.253-2A>G, NM_000155.4:c.265T>G, NM_000155.4:c.287_289delACA, NM_000155.4:c.290A>G, NM_000155.4:c.292G>A, NM_000155.4:c.329-2A>C, NM_000155.4:c.367C>T, NM_000155.4:c.377+7A>C, NM_000155.4:c.386T>C, NM_000155.4:c.400delT, NM_000155.4:c.404C>T, NM_000155.4:c.413C>T, NM_000155.4:c.416T>G, NM_000155.4:c.425T>A, NM_000155.4:c.428C>T, NM_000155.4:c.442C>T, NM_000155.4:c.443G>A, NM_000155.4:c.443G>C, NM_000155.4:c.501_503delGGT, NM_000155.4:c.505C>A, NM_000155.4:c.508-1G>C, NM_000155.4:c.510C>A, NM_000155.4:c.512T>C, NM_000155.4:c.547C>A, NM_000155.4:c.552C>A, NM_000155.4:c.563A>G, NM_000155.4:c.565-3_575delCAGGTATGGGCCAG, NM_000155.4:c.568T>C, NM_000155.4:c.580T>C, NM_000155.4:c.584T>C, NM_000155.4:c.596delC, NM_000155.4:c.601C>T, NM_000155.4:c.602G>A, NM_000155.4:c.607G>A, NM_000155.4:c.610C>T, NM_000155.4:c.617A>G, NM_000155.4:c.619C>T, NM_000155.4:c.626A>G, NM_000155.4:c.634C>T, NM_000155.4:c.688-2A>C, NM_000155.4:c.692G>A, NM_000155.4:c.713_722delACTGGTTAGT, NM_000155.4:c.772C>T, NM_000155.4:c.775C>T, NM_000155.4:c.790delC, NM_000155.4:c.790_792delCTAinsTAG, NM_000155.4:c.793C>G, NM_000155.4:c.823C>G, NM_000155.4:c.844C>G, NM_000155.4:c.855G>T, NM_000155.4:c.904+1G>T, NM_000155.4:c.905-2A>G, NM_000155.4:c.907G>A, NM_000155.4:c.939G>A, NM_000155.4:c.947G>A, NM_000155.4:c.957C>A, NM_000155.4:c.985T>C, NM_000155.4:c.997C>G, NM_000155.4:c.997C>T, NM_000155.4:c.998G>A, NM_000155.4:c.1006A>T, NM_000155.4:c.1030C>A, NM_000155.4:c.1048delA, NM_000155.4:c.1049delC, NM_000155.4:c.1138T>C
GAMT	Guanidinoacetate methyltransferase deficiency	NM_000156.6:c.590T>C, NM_000156.6:c.506G>A
GBE1	Glycogen storage disease IV	NM_000158.4:c.2052+1G>A, NM_000158.4:c.1883A>G, NM_000158.4:c.1774G>T, NM_000158.4:c.1604A>G, NM_000158.4:c.1571G>A, NM_000158.4:c.1570C>T, NM_000158.4:c.1543C>T, NM_000158.4:c.986A>G, NM_000158.4:c.986A>C, NM_000158.4:c.771T>A, NM_000158.4:c.466_470delCGTAT
GCDH	Glutaricaciduria, type I	NM_000159.4:c.74C>A, NM_000159.4:c.271+1G>A, NM_000159.4:c.383G>A, NM_000159.4:c.416C>T, NM_000159.4:c.442G>A, NM_000159.4:c.542A>G, NM_000159.4:c.572T>C, NM_000159.4:c.636-1G>A, NM_000159.4:c.680G>C, NM_000159.4:c.743C>T, NM_000159.4:c.751C>T, NM_000159.4:c.764C>T, NM_000159.4:c.769C>T, NM_000159.4:c.877G>A, NM_000159.4:c.883T>C, NM_000159.4:c.914C>T, NM_000159.4:c.947C>A, NM_000159.4:c.1001_1002delAG, NM_000159.4:c.1015A>G, NM_000159.4:c.1060G>A, NM_000159.4:c.1060G>C, NM_000159.4:c.1093G>A, NM_000159.4:c.1148G>A, NM_000159.4:c.1168G>C, NM_000159.4:c.1198G>A, NM_000159.4:c.1198_1199insT, NM_000159.4:c.1204C>T, NM_000159.4:c.1244-2A>C, NM_000159.4:c.1247C>T, NM_000159.4:c.1262C>T
GENE	Disease name	varname
GFM1	Combined Oxidative Phosphorylation Deficiency 1	NM_024996.7:c.139C>T, NM_024996.7:c.521A>G, NM_024996.7:c.748C>T, NM_024996.7:c.1294_1297delACAG, NM_024996.7:c.1528_1529delAG
GJB2	Deafness, autosomal recessive 1	NM_004004.6:c.617A>G, NM_004004.6:c.557C>T, NM_004004.6:c.551G>C, NM_004004.6:c.550C>T, NM_004004.6:c.516G>A, NM_004004.6:c.503A>G, NM_004004.6:c.465T>A, NM_004004.6:c.439G>A, NM_004004.6:c.427C>T, NM_004004.6:c.416G>A, NM_004004.6:c.413G>A, NM_004004.6:c.402delG, NM_004004.6:c.380G>A, NM_004004.6:c.365A>T, NM_004004.6:c.358_360delGAG, NM_004004.6:c.334_335delAA, NM_004004.6:c.313_326delAAGTTCATCAAGGG, NM_004004.6:c.310_323delAGGAAGTTCATCAA, NM_004004.6:c.299_300delAT, NM_004004.6:c.299A>T, NM_004004.6:c.270dupA, NM_004004.6:c.269dupT, NM_004004.6:c.269T>C, NM_004004.6:c.250G>T, NM_004004.6:c.250G>C, NM_004004.6:c.249C>G, NM_004004.6:c.241C>G, NM_004004.6:c.239A>C, NM_004004.6:c.238C>T, NM_004004.6:c.235delC, NM_004004.6:c.231G>A, NM_004004.6:c.230G>A, NM_004004.6:c.229T>C, NM_004004.6:c.227T>C, NM_004004.6:c.176_191delGCTGCAAGAACGTGTG, NM_004004.6:c.169C>T, NM_004004.6:c.139G>T, NM_004004.6:c.132G>A, NM_004004.6:c.35delG
GJB3	Deafness, autosomal recessive	NM_024009.3:c.94C>T, NM_024009.3:c.529T>G, NM_024009.3:c.580G>A
GJB6	Deafness, autosomal recessive 1	NM_001110219.3:c.689dupA, NM_001110219.3:c.485dupA, NM_001110219.3:c.443delC, NM_001110219.3:c.383_384delTA, NM_001110219.3:c.261dupA, NM_001110219.3:c.169C>T, NM_001110219.3:c.14C>T, CNV deletions

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
GLB1	GM1 gangliosidosis and mucopolysaccharidosis type IVB	NM_000404.4:c.1733A>G, NM_000404.4:c.1646C>T, NM_000404.4:c.1577dupG, NM_000404.4:c.1549G>T, NM_000404.4:c.1456_1466dupGGTGCATATAT, NM_000404.4:c.1452C>G, NM_000404.4:c.1445G>A, NM_000404.4:c.1444C>T, NM_000404.4:c.1370G>A, NM_000404.4:c.1369C>T, NM_000404.4:c.1355dupA, NM_000404.4:c.1325G>A, NM_000404.4:c.1321G>A, NM_000404.4:c.1313G>A, NM_000404.4:c.1310A>T, NM_000404.4:c.1223A>C, NM_000404.4:c.1174_1175delCT, NM_000404.4:c.1068+1G>T, NM_000404.4:c.1051C>T, NM_000404.4:c.1004C>T, NM_000404.4:c.947A>G, NM_000404.4:c.901G>A, NM_000404.4:c.818G>T, NM_000404.4:c.817T>C, NM_000404.4:c.622C>T, NM_000404.4:c.602G>A, NM_000404.4:c.601C>T, NM_000404.4:c.591dupT, NM_000404.4:c.457+2T>C, NM_000404.4:c.442C>T, NM_000404.4:c.442C>A, NM_000404.4:c.438_440delTCT, NM_000404.4:c.276G>A, NM_000404.4:c.202C>T, NM_000404.4:c.176G>A, NM_000404.4:c.175C>T, NM_000404.4:c.171C>G, NM_000404.4:c.152T>C, NM_000404.4:c.145C>T
GLDC	Glycine encephalopathy (GLDC-related)	NM_000170.3:c.2405C>T, NM_000170.3:c.2284G>A, NM_000170.3:c.2216G>A, NM_000170.3:c.2113G>A, NM_000170.3:c.1705G>A, NM_000170.3:c.1691G>T, NM_000170.3:c.1545G>C, NM_000170.3:c.1229G>A, NM_000170.3:c.1166C>T, NM_000170.3:c.322G>T
GLE1	Lethal Congenital Contracture Syndrome 1	NM_001003722.2:c.898-2A>G, NM_001003722.2:c.1412_1413delAG, NM_001003722.2:c.1807C>T, NM_001003722.2:c.2051T>C, NM_001003722.2:c.2067_2070delCTTT
GNE	Inclusion body myopathy, autosomal recessive	NM_001128227.3:c.2228T>C, NM_001128227.3:c.2179G>T, NM_001128227.3:c.2179G>A, NM_001128227.3:c.2116T>C, NM_001128227.3:c.2086G>A, NM_001128227.3:c.1937C>G, NM_001128227.3:c.1891G>A, NM_001128227.3:c.1820G>A, NM_001128227.3:c.1807G>T, NM_001128227.3:c.1002T>A, NM_001128227.3:c.880C>T, NM_001128227.3:c.830G>A, NM_001128227.3:c.766G>A, NM_001128227.3:c.478C>T
GNPTAB	Mucolipidosis type II and III	NM_024312.5:c.3663delG, NM_024312.5:c.3598G>A, NM_024312.5:c.3565C>T, NM_024312.5:c.3560_3561delAG, NM_024312.5:c.3503_3504delTC, NM_024312.5:c.3410T>A, NM_024312.5:c.3326dupA, NM_024312.5:c.3173C>G, NM_024312.5:c.2896delA, NM_024312.5:c.2383delG, NM_024312.5:c.1931C>T, NM_024312.5:c.1906dupA, NM_024312.5:c.1759C>T, NM_024312.5:c.1196C>T, NM_024312.5:c.1000C>T, NM_024312.5:c.749dupA, NM_024312.5:c.732_733delAA, NM_024312.5:c.648_651delAGAA, NM_024312.5:c.616_619delACAG, NM_024312.5:c.99delC, NM_024312.5:c.25C>T
GNS	Mucopolysaccharidosis, Type IIID (Sanfilippo D)	NM_002076.4:c.1226dupG, NM_002076.4:c.1169delA, NM_002076.4:c.1168C>T, NM_002076.4:c.1063C>T, NM_002076.4:c.413C>G
GRHPR	Primary Hyperoxaluria, Type 2	NM_012203.2:c.101delG, NM_012203.2:c.295C>T, NM_012203.2:c.435G>A, NM_012203.2:c.622C>T
GUCY2D	Leber congenital amaurosis 1	NM_000180.4:c.456C>A, NM_000180.4:c.620delC, NM_000180.4:c.1694T>C, NM_000180.4:c.2734_2735delTT, NM_000180.4:c.2945-1delG
GUSB	Mucopolysaccharidosis, Type VII	NM_000181.4:c.1881G>T, NM_000181.4:c.1856C>T, NM_000181.4:c.1831C>T, NM_000181.4:c.1730G>T, NM_000181.4:c.1618G>T, NM_000181.4:c.1534G>A, NM_000181.4:c.1521G>A, NM_000181.4:c.1429C>T, NM_000181.4:c.1338G>A, NM_000181.4:c.1337G>A, NM_000181.4:c.1244+1G>A, NM_000181.4:c.1244C>T, NM_000181.4:c.1222C>T, NM_000181.4:c.1219_1220insC, NM_000181.4:c.1144C>T, NM_000181.4:c.1084G>A, NM_000181.4:c.1065+1G>T, NM_000181.4:c.1061C>T, NM_000181.4:c.1050G>C, NM_000181.4:c.866G>A, NM_000181.4:c.820_821delAC, NM_000181.4:c.646C>T, NM_000181.4:c.526C>T, NM_000181.4:c.499C>T, NM_000181.4:c.442C>T
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	NM_000182.5:c.2146+1G>A, NM_000182.5:c.2132dupC, NM_000182.5:c.2131C>A, NM_000182.5:c.2027G>A, NM_000182.5:c.1918C>T, NM_000182.5:c.1793_1794delAT, NM_000182.5:c.1678C>T, NM_000182.5:c.1644delC, NM_000182.5:c.1620+2_1620+6delTAAGG, NM_000182.5:c.1528G>C, NM_000182.5:c.1422dupT, NM_000182.5:c.1132C>T, NM_000182.5:c.919-2A>G, NM_000182.5:c.845T>A, NM_000182.5:c.499delA, NM_000182.5:c.274_278delTCATC
HADHB	Trifunctional protein deficiency	NM_000183.3:c.788A>G, NM_000183.3:c.1331G>A, NM_000183.3:c.1364T>G
HBA1/2	Thalassemia, alpha	--MED ; --SEA ; --THAI ; - α3.7 ; - α4.2 ; - α20.5 ; --FIL

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
HBB	Thalassemias, beta	NM_000518.5:c.*110_*114delTAAAA, NM_000518.5:c.*113A>G, NM_000518.5:c.*112A>G, NM_000518.5:c.*110_*111delTA, NM_000518.5:c.*111A>G, NM_000518.5:c.*110T>C, NM_000518.5:c.*6C>G, NM_000518.5:c.440_441dupAC, NM_000518.5:c.440A>T, NM_000518.5:c.440A>C, NM_000518.5:c.439C>G, NM_000518.5:c.438T>A, NM_000518.5:c.437A>G, NM_000518.5:c.436T>C, NM_000518.5:c.436T>A, NM_000518.5:c.435G>C, NM_000518.5:c.431A>C, NM_000518.5:c.428C>A, NM_000518.5:c.421G>A, NM_000518.5:c.415G>C, NM_000518.5:c.410G>A, NM_000518.5:c.404T>A, NM_000518.5:c.397A>G, NM_000518.5:c.380_396delTGCAGGCTGCCTATCAG, NM_000518.5:c.385_388delGCTGinsCCACA, NM_000518.5:c.383_385delAGG, NM_000518.5:c.385G>C, NM_000518.5:c.383A>C, NM_000518.5:c.380T>G, NM_000518.5:c.380T>A, NM_000518.5:c.371_378delCCCCACCA, NM_000518.5:c.370_378delACCCCACCA, NM_000518.5:c.374C>G, NM_000518.5:c.374C>A, NM_000518.5:c.364G>T, NM_000518.5:c.364G>C, NM_000518.5:c.364G>A, NM_000518.5:c.363A>C, NM_000518.5:c.353A>G, NM_000518.5:c.347C>A, NM_000518.5:c.343_344delCTinsG, NM_000518.5:c.344T>C, NM_000518.5:c.341T>A, NM_000518.5:c.337T>C, NM_000518.5:c.332T>C, NM_000518.5:c.328delG, NM_000518.5:c.328G>A, NM_000518.5:c.323dupG, NM_000518.5:c.320T>G, NM_000518.5:c.316-1G>T, NM_000518.5:c.316-1G>C, NM_000518.5:c.316-1G>A, NM_000518.5:c.316-2A>G, NM_000518.5:c.316-2A>C, NM_000518.5:c.316-3C>G, NM_000518.5:c.316-3C>A, NM_000518.5:c.316-7C>G, NM_000518.5:c.316-7C>A, NM_000518.5:c.316-14T>G, NM_000518.5:c.316-90A>G, NM_000518.5:c.316-106C>T, NM_000518.5:c.316-106C>G, NM_000518.5:c.316-125A>G, NM_000518.5:c.316-146T>G, NM_000518.5:c.316-197C>T, NM_000518.5:c.316-238C>T, NM_000518.5:c.315+4_315+5delAG, NM_000518.5:c.315+5G>C, NM_000518.5:c.315+2delT, NM_000518.5:c.315+1delG, NM_000518.5:c.315+2T>G, NM_000518.5:c.315+2T>C, NM_000518.5:c.315+2T>A, NM_000518.5:c.315+1G>T, NM_000518.5:c.315+1G>C, NM_000518.5:c.315+1G>A, NM_000518.5:c.315G>C, NM_000518.5:c.312C>G, NM_000518.5:c.306G>C, NM_000518.5:c.305A>G, NM_000518.5:c.304G>A, NM_000518.5:c.302C>T, NM_000518.5:c.300_301insTCTGAGAA, NM_000518.5:c.299A>T, NM_000518.5:c.299A>G, NM_000518.5:c.299A>C, NM_000518.5:c.298G>T, NM_000518.5:c.298G>C, NM_000518.5:c.298G>A, NM_000518.5:c.292_295dupCACG, NM_000518.5:c.295G>A, NM_000518.5:c.293A>T, NM_000518.5:c.287dupA, NM_000518.5:c.286A>G, NM_000518.5:c.282_283dupTG, NM_000518.5:c.283G>C, NM_000518.5:c.277C>T, NM_000518.5:c.277C>A, NM_000518.5:c.275T>C, NM_000518.5:c.271G>T, NM_000518.5:c.271G>A, NM_000518.5:c.269G>A, NM_000518.5:c.268A>C, NM_000518.5:c.244_265delCTCAAGGGCACCTTTGCCACAC, NM_000518.5:c.266T>C, NM_000518.5:c.263C>T, NM_000518.5:c.258dupT, NM_000518.5:c.257T>C, NM_000518.5:c.253delA, NM_000518.5:c.251delG, NM_000518.5:c.249G>C, NM_000518.5:c.248A>T, NM_000518.5:c.248A>C, NM_000518.5:c.247A>G, NM_000518.5:c.246C>A, NM_000518.5:c.235delC, NM_000518.5:c.230delC, NM_000518.5:c.230C>A, NM_000518.5:c.226delC, NM_000518.5:c.221_224dupATGG, NM_000518.5:c.217_221delAGTGAinsT, NM_000518.5:c.219_220delTG, NM_000518.5:c.220G>A, NM_000518.5:c.217dupA, NM_000518.5:c.216dupT, NM_000518.5:c.216delT, NM_000518.5:c.209G>A, NM_000518.5:c.208G>A, NM_000518.5:c.206T>A, NM_000518.5:c.203_204delTG, NM_000518.5:c.203T>A, NM_000518.5:c.201delA, NM_000518.5:c.199A>G, NM_000518.5:c.196A>C, NM_000518.5:c.189_195delTCATGGC, NM_000518.5:c.194delG, NM_000518.5:c.190C>T, NM_000518.5:c.184A>T, NM_000518.5:c.182T>A, NM_000518.5:c.179A>C, NM_000518.5:c.176delC, NM_000518.5:c.176C>G, NM_000518.5:c.170G>A, NM_000518.5:c.169G>C, NM_000518.5:c.164_168delTTATGinsGGCATCA, NM_000518.5:c.166dupA, NM_000518.5:c.162delT, NM_000518.5:c.155delC, NM_000518.5:c.155C>T, NM_000518.5:c.153delT, NM_000518.5:c.147delG, NM_000518.5:c.143_146dupATCT, NM_000518.5:c.143dupA, NM_000518.5:c.137T>C, NM_000518.5:c.135delC, NM_000518.5:c.134C>G, NM_000518.5:c.130G>T, NM_000518.5:c.127_129delTTT, NM_000518.5:c.126_129delCTTT, NM_000518.5:c.130G>A, NM_000518.5:c.128T>C, NM_000518.5:c.126delC, NM_000518.5:c.127T>G, NM_000518.5:c.127T>C, NM_000518.5:c.114_120delGACCCAG, NM_000518.5:c.117_118delCC, NM_000518.5:c.118C>T, NM_000518.5:c.115delA, NM_000518.5:c.114G>A, NM_000518.5:c.112delT, NM_000518.5:c.113G>A, NM_000518.5:c.110delC, NM_000518.5:c.109C>T, NM_000518.5:c.108C>A, NM_000518.5:c.102_104delGGT, NM_000518.5:c.103G>T, NM_000518.5:c.93-21_96delGGTCTATTTTCCCACCCTTAGGCTG, NM_000518.5:c.93-22_95delTGGTCTATTTTCCCACCCTTAGGCT, NM_000518.5:c.94_95insGGC, NM_000518.5:c.93G>T, NM_000518.5:c.93-17_93-1delTATTTTCCCACCCTTAG, NM_000518.5:c.93-1G>T, NM_000518.5:c.93-1G>C, NM_000518.5:c.93-1G>A, NM_000518.5:c.93-2A>G, NM_000518.5:c.93-2A>C, NM_000518.5:c.93-3T>G, NM_000518.5:c.93-15T>G, NM_000518.5:c.93-21G>A, NM_000518.5:c.93-23T>C, NM_000518.5:c.92+6T>C, NM_000518.5:c.92+5G>T, NM_000518.5:c.92+5G>C, NM_000518.5:c.92+5G>A, NM_000518.5:c.92+2T>G, NM_000518.5:c.92+2T>C, NM_000518.5:c.92+2T>A, NM_000518.5:c.92+1G>T, NM_000518.5:c.92+1G>C, NM_000518.5:c.92+1G>A, NM_000518.5:c.92G>C, NM_000518.5:c.92G>A, NM_000518.5:c.91A>G, NM_000518.5:c.91A>C, NM_000518.5:c.90C>T, NM_000518.5:c.85dupC, NM_000518.5:c.86T>A, NM_000518.5:c.82G>T, NM_000518.5:c.81G>C, NM_000518.5:c.80A>G, NM_000518.5:c.79_80insT, NM_000518.5:c.79G>T, NM_000518.5:c.79G>A, NM_000518.5:c.68_74delAAGTTGG,

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000518.5:c.75T>A, NM_000518.5:c.71_73delTTG, NM_000518.5:c.70G>T, NM_000518.5:c.70G>A, NM_000518.5:c.68A>C, NM_000518.5:c.64_67delGATG, NM_000518.5:c.67G>T, NM_000518.5:c.67G>C, NM_000518.5:c.64dupG, NM_000518.5:c.61G>A, NM_000518.5:c.59A>G, NM_000518.5:c.55G>A, NM_000518.5:c.52A>T, NM_000518.5:c.51delC, NM_000518.5:c.51C>T, NM_000518.5:c.48G>A, NM_000518.5:c.46delT, NM_000518.5:c.47G>A, NM_000518.5:c.45dupG, NM_000518.5:c.36delT, NM_000518.5:c.33dupC, NM_000518.5:c.34G>A, NM_000518.5:c.32C>A, NM_000518.5:c.27dupG, NM_000518.5:c.25_26delAA, NM_000518.5:c.26A>G, NM_000518.5:c.22_24delGAG, NM_000518.5:c.22G>A, NM_000518.5:c.20delA, NM_000518.5:c.20A>T, NM_000518.5:c.19_20delGAinsAT, NM_000518.5:c.20A>C, NM_000518.5:c.18_19delTG, NM_000518.5:c.15_19delTCCTGinsATCTT, NM_000518.5:c.17_18delCT, NM_000518.5:c.19G>A, NM_000518.5:c.8A>C, NM_000518.5:c.4delG, NM_000518.5:c.4G>T, NM_000518.5:c.3G>A, NM_000518.5:c.2T>G, NM_000518.5:c.2T>C, NM_000518.5:c.2T>A, NM_000518.5:c.1A>G, NM_000518.5:c.-18C>G, NM_000518.5:c.-29G>A, NM_000518.5:c.-41delT, NM_000518.5:c.-50A>C, NM_000518.5:c.-77_-76delAA, NM_000518.5:c.-75G>C, NM_000518.5:c.-78A>G, NM_000518.5:c.-78A>C, NM_000518.5:c.-79A>G, NM_000518.5:c.-80T>C, NM_000518.5:c.-80T>A, NM_000518.5:c.-81A>G, NM_000518.5:c.-82C>T, NM_000518.5:c.-82C>A, NM_000518.5:c.-136C>T, NM_000518.5:c.-136C>G, NM_000518.5:c.-137C>T, NM_000518.5:c.-137C>G, NM_000518.5:c.-137C>A, NM_000518.5:c.-138C>T, NM_000518.5:c.-138C>G, NM_000518.5:c.-138C>A, NM_000518.5:c.-140C>T, NM_000518.5:c.-142C>T, NM_000518.5:c.-151C>T, NM_000518.5:c.*185_*209delGGTCAGTGCATTTAAAACATAAAGA, NM_000518.5:c.-50A>G, NM_000518.5:c.-142C>A
HEXA	Tay-Sachs disease	NM_000520.6:c.1537C>T, NM_000520.6:c.1528C>T, NM_000520.6:c.1510delC, NM_000520.6:c.1511G>A, NM_000520.6:c.1510C>T, NM_000520.6:c.1499delT, NM_000520.6:c.1496G>A, NM_000520.6:c.1495C>T, NM_000520.6:c.1490A>G, NM_000520.6:c.1453T>C, NM_000520.6:c.1444G>A, NM_000520.6:c.1422G>C, NM_000520.6:c.1351C>G, NM_000520.6:c.1278_1281dupCTAT, NM_000520.6:c.1277_1278insTAT, NM_000520.6:c.1274_1277dupTATC, NM_000520.6:c.1260G>C, NM_000520.6:c.1214_1215delAAinsG, NM_000520.6:c.1177C>T, NM_000520.6:c.1176G>A, NM_000520.6:c.987G>A, NM_000520.6:c.986+3A>G, NM_000520.6:c.915_917delCTT, NM_000520.6:c.805+1G>C, NM_000520.6:c.805+1G>A, NM_000520.6:c.805G>A, NM_000520.6:c.759_774dupGCTTGCAGAGTTTGAC, NM_000520.6:c.772G>C, NM_000520.6:c.749G>A, NM_000520.6:c.745C>T, NM_000520.6:c.739C>T, NM_000520.6:c.672+1G>A, NM_000520.6:c.632T>C, NM_000520.6:c.629C>T, NM_000520.6:c.611A>G, NM_000520.6:c.540C>G, NM_000520.6:c.538T>C, NM_000520.6:c.533G>T, NM_000520.6:c.533G>A, NM_000520.6:c.532C>T, NM_000520.6:c.509G>A, NM_000520.6:c.508C>T, NM_000520.6:c.459+5G>A, NM_000520.6:c.380T>G, NM_000520.6:c.254-1G>C, NM_000520.6:c.173G>A, NM_000520.6:c.116T>G, NM_000520.6:c.78G>A, NM_000520.6:c.77G>A, NM_000520.6:c.2T>C, NM_000520.6:c.1A>T, NM_000520.6:c.1A>G
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	NM_000521.4:c.114delG, NM_000521.4:c.170delG, NM_000521.4:c.202_203insGG, NM_000521.4:c.298delC, NM_000521.4:c.508C>T, NM_000521.4:c.797A>G, NM_000521.4:c.841C>T, NM_000521.4:c.850C>T, NM_000521.4:c.1234_1238delAAAGC, NM_000521.4:c.1250C>T, NM_000521.4:c.1308_1309delCA, NM_000521.4:c.1344delT, NM_000521.4:c.1367A>C, NM_000521.4:c.1375G>T, NM_000521.4:c.1380G>A, NM_000521.4:c.1514_1515insGGCAAGTGCTGTT, NM_000521.4:c.1537_1538delCT, NM_000521.4:c.1618_1619insGTTCATGTTATCTACAGACGT
HFE	Hemochromatosis, Type 1	NM_000410.4:c.18G>C, NM_000410.4:c.193A>T, NM_000410.4:c.252G>A, NM_000410.4:c.277G>C, NM_000410.4:c.314T>C, NM_000410.4:c.829G>A, NM_000410.4:c.989G>T
HGD	Alkaptonuria	NM_000187.4:c.1189-2A>G, NM_000187.4:c.1111dupC, NM_000187.4:c.1102A>G, NM_000187.4:c.899T>G, NM_000187.4:c.873C>A, NM_000187.4:c.808G>A, NM_000187.4:c.688C>T, NM_000187.4:c.674G>A, NM_000187.4:c.481G>A, NM_000187.4:c.469+2T>C, NM_000187.4:c.342+1G>A, NM_000187.4:c.283-4C>T, NM_000187.4:c.283-5delT, NM_000187.4:c.175delA, NM_000187.4:c.172A>T, NM_000187.4:c.140C>T, NM_000187.4:c.16-1G>A
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)/Retinitis pigmentosa 73	NM_152419.3:c.493+1G>A, NM_152419.3:c.607C>T, NM_152419.3:c.848C>T, NM_152419.3:c.1030C>T, NM_152419.3:c.1250+1G>A, NM_152419.3:c.1378-1G>A, NM_152419.3:c.1464+1G>A, NM_152419.3:c.1501delA, NM_152419.3:c.1553C>T, NM_152419.3:c.1622C>T, NM_152419.3:c.1843G>A
HMGCL	HMG-CoA lyase deficiency	NM_000191.3:c.835G>A, NM_000191.3:c.698A>G, NM_000191.3:c.505_506delTC, NM_000191.3:c.230delT, NM_000191.3:c.206_207delCT, NM_000191.3:c.122G>A
HPD	Tyrosinemia, type III	NM_002150.3:c.1005C>G, NM_002150.3:c.987delA, NM_002150.3:c.774T>G, NM_002150.3:c.600C>G
HPS1	Hermansky-Pudlak Syndrome 1	NM_000195.5:c.1996G>T, NM_000195.5:c.1472_1487dupCCAGCAGGGGAGGCCC, NM_000195.5:c.972delC, NM_000195.5:c.398+5G>A, NM_000195.5:c.397G>T
HSD17B4	D-bifunctional protein deficiency	NM_000414.4:c.46G>A, NM_000414.4:c.317G>C, NM_000414.4:c.650A>G, NM_000414.4:c.972+1G>T, NM_000414.4:c.1369A>T

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
however, only variants classified as pathogenic and likely pathogenic
at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
HYLS1	Hydrolethalus Syndrome	NM_001134793.2:c.632A>G, NM_001134793.2:c.669G>A, NM_001134793.2:c.724C>T
IDS	Mucopolysaccharidosis, Type II (Hunter Syndrome)	NM_000202.8:c.1508T>A, NM_000202.8:c.1505G>C, NM_000202.8:c.1466G>C, NM_000202.8:c.1464G>T, NM_000202.8:c.1148delC, NM_000202.8:c.1122C>T, NM_000202.8:c.998C>T, NM_000202.8:c.937C>T, NM_000202.8:c.880-8A>G, NM_000202.8:c.690_691insT, NM_000202.8:c.683C>T, NM_000202.8:c.596_599delAACA, NM_000202.8:c.597delA, NM_000202.8:c.587T>C, NM_000202.8:c.514C>T, NM_000202.8:c.404A>G, NM_000202.8:c.388_389insG, NM_000202.8:c.314_317dupTCAA, NM_000202.8:c.283A>G, NM_000202.8:c.278delC, NM_000202.8:c.240+1G>A, NM_000202.8:c.208dupC
IKBAP/ ELP1	Dysautonomia, familial	NM_003640.5:c.3332delC, NM_003640.5:c.2741C>T, NM_003640.5:c.2328delT, NM_003640.5:c.2204+6T>C, NM_003640.5:c.2087G>C, NM_003640.5:c.2087G>A, NM_003640.5:c.1460+2T>C
IL11RA	Crigler-Najjar Syndrome	NM_001142784.3:c.-3327A>G
IL2RG	Severe Combined Immunodeficiency, X-Linked	NM_000206.3:c.854G>A, NM_000206.3:c.664C>T, NM_000206.3:c.454+1G>A, NM_000206.3:c.452T>C, NM_000206.3:c.355A>T, NM_000206.3:c.343T>C, NM_000206.3:c.341G>A, NM_000206.3:c.186T>A
IVD	Isovaleric acidemia	NM_002225.5:c.-8T>G, NM_002225.5:c.125T>C, NM_002225.5:c.148C>T, NM_002225.5:c.149G>A, NM_002225.5:c.149G>C, NM_002225.5:c.234+1G>A, NM_002225.5:c.358G>A, NM_002225.5:c.381delT, NM_002225.5:c.397_398delTG, NM_002225.5:c.422_423insGAAT, NM_002225.5:c.456+2T>C, NM_002225.5:c.469_470insGT, NM_002225.5:c.498delG, NM_002225.5:c.550+1G>A, NM_002225.5:c.584G>A, NM_002225.5:c.596G>T, NM_002225.5:c.617delT, NM_002225.5:c.784+1G>A, NM_002225.5:c.851G>A, NM_002225.5:c.932C>T, NM_002225.5:c.985_986delAT, NM_002225.5:c.1132T>C, NM_002225.5:c.1136_1138+4delTTGGTGA, NM_002225.5:c.1138+1_1138+4delGTGA, NM_002225.5:c.1174C>T, NM_002225.5:c.1177delT, NM_002225.5:c.1183C>T, NM_002225.5:c.1199A>G
LAMA2	LAMA2-related Muscular Dystrophy	NM_000426.4:c.112+1G>A, NM_000426.4:c.184G>T, NM_000426.4:c.725G>A, NM_000426.4:c.825delC, NM_000426.4:c.1050delT, NM_000426.4:c.1612C>T, NM_000426.4:c.1634T>A, NM_000426.4:c.2045_2046delAG, NM_000426.4:c.2098_2099delTT, NM_000426.4:c.2323-2A>T, NM_000426.4:c.2451-2A>G, NM_000426.4:c.2750-1G>C, NM_000426.4:c.2901C>A, NM_000426.4:c.2962C>T, NM_000426.4:c.3215delG, NM_000426.4:c.3237C>A, NM_000426.4:c.3620_3642delCCAAGGGCATTGTTTTTCAACAT, NM_000426.4:c.3629delT, NM_000426.4:c.3718C>T, NM_000426.4:c.3976C>T, NM_000426.4:c.4437-5T>A, NM_000426.4:c.4645C>T, NM_000426.4:c.5050G>T, NM_000426.4:c.5227G>T, NM_000426.4:c.6008delA, NM_000426.4:c.6037delT, NM_000426.4:c.6334A>T, NM_000426.4:c.6429+1G>A, NM_000426.4:c.6616delT, NM_000426.4:c.6955C>T, NM_000426.4:c.7147C>T, NM_000426.4:c.7277_7278delCT, NM_000426.4:c.7534delC, NM_000426.4:c.7732C>T, NM_000426.4:c.7810C>T, NM_000426.4:c.7888C>T, NM_000426.4:c.8314delA, NM_000426.4:c.8684C>G, NM_000426.4:c.8705delT, NM_000426.4:c.8748delA, NM_000426.4:c.9098_9099insCAAA, NM_000426.4:c.9220delA, NM_000426.4:c.9253C>T
LAMA3	Epidermolysis bullosa, junctional, Herlitz type (LAMA3-related)	NM_198129.4:c.5160delG, NM_198129.4:c.6009delG, NM_198129.4:c.6808C>T, NM_198129.4:c.6943A>T, NM_198129.4:c.7489C>T, NM_198129.4:c.8177+2T>G, NM_198129.4:c.8962C>T, NM_198129.4:c.9156_9157insA, NM_198129.4:c.9704_9705insT
LAMB3	Epidermolysis bullosa, junctional, Herlitz type (LAMB3-related)	NM_000228.3:c.3228+1G>T, NM_000228.3:c.3228+1G>A, NM_000228.3:c.2806C>T, NM_000228.3:c.1903C>T, NM_000228.3:c.1830G>A, NM_000228.3:c.1587_1588delAG, NM_000228.3:c.1438_1442delCCGTG, NM_000228.3:c.1357delT, NM_000228.3:c.904delT, NM_000228.3:c.727C>T, NM_000228.3:c.628+1delG, NM_000228.3:c.628G>A, NM_000228.3:c.565-2A>G, NM_000228.3:c.496C>T, NM_000228.3:c.124C>T
LAMC2	Epidermolysis bullosa, junctional, Herlitz type (LAMC2-related)	NM_005562.3:c.283C>T, NM_005562.3:c.343C>T, NM_005562.3:c.405-1G>A, NM_005562.3:c.1659C>A, NM_005562.3:c.1782_1783delGC, NM_005562.3:c.2136_2142delCCAGAAC, NM_005562.3:c.3069+1G>A, NM_005562.3:c.3120_3121insA, NM_005562.3:c.3510_3511insA
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	NM_001127671.2:c.2503G>T, NM_001127671.2:c.2013dupT, NM_001127671.2:c.1789C>T, NM_001127671.2:c.1018_1022delAATTG, NM_001127671.2:c.653dupT, NM_001127671.2:c.171_174delTAAC
LOXHD1	Deafness, autosomal recessive 77	NM_001384474.1:c.4714C>T, NM_001384474.1:c.4524_4525delAG, NM_001384474.1:c.4526G>A, NM_001384474.1:c.3924C>A, NM_001384474.1:c.3874C>T, NM_001384474.1:c.2008C>T, NM_001384474.1:c.512-1G>A, NM_001384474.1:c.457_461dupCGCCA, NM_001384474.1:c.2T>A
LRPPRC	Leigh Syndrome, French-Canadian Type	NM_133259.4:c.3830_3837delGTGGTGCA, NM_133259.4:c.1061C>T
MAN2B1	Alpha-Mannosidosis	NM_000528.4:c.2686_2687delCTinsG, NM_000528.4:c.2436+2T>C, NM_000528.4:c.2426T>C, NM_000528.4:c.2401G>T, NM_000528.4:c.2398G>A,

GENESeeker

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_000528.4:c.2368C>T, NM_000528.4:c.2278C>T, NM_000528.4:c.2119C>T, NM_000528.4:c.2013delT, NM_000528.4:c.1929G>A, NM_000528.4:c.1915C>T, NM_000528.4:c.1830+1G>C, NM_000528.4:c.1780C>T, NM_000528.4:c.1259G>T, NM_000528.4:c.1067C>G, NM_000528.4:c.384G>A, NM_000528.4:c.215A>T, NM_000528.4:c.1A>G
MAT1A	Hypermethioninemia	NM_000429.3:c.1070C>T, NM_000429.3:c.1043_1044delTG, NM_000429.3:c.1006G>A, NM_000429.3:c.966T>G, NM_000429.3:c.914T>C, NM_000429.3:c.827_828insG, NM_000429.3:c.791G>A, NM_000429.3:c.790C>T, NM_000429.3:c.595C>T, NM_000429.3:c.538_539insTG
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	NM_020166.5:c.2079delA, NM_020166.5:c.1942G>A, NM_020166.5:c.1930G>T, NM_020166.5:c.1905delA, NM_020166.5:c.1526delG, NM_020166.5:c.1380T>G, NM_020166.5:c.1310T>C, NM_020166.5:c.1277T>C, NM_020166.5:c.1155A>C, NM_020166.5:c.1114C>T, NM_020166.5:c.1074delG, NM_020166.5:c.640-1G>A, NM_020166.5:c.640-2A>G, NM_020166.5:c.558delA, NM_020166.5:c.559T>C, NM_020166.5:c.388G>A, NM_020166.5:c.343C>T, NM_020166.5:c.310C>T
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	NM_022132.5:c.295G>C, NM_022132.5:c.380C>G, NM_022132.5:c.464G>A, NM_022132.5:c.499T>C, NM_022132.5:c.515_516insT, NM_022132.5:c.639delG, NM_022132.5:c.729_730insC, NM_022132.5:c.838G>T, NM_022132.5:c.929C>G, NM_022132.5:c.994C>T, NM_022132.5:c.1015G>A, NM_022132.5:c.1065A>T, NM_022132.5:c.1072+1G>A, NM_022132.5:c.1309A>G, NM_022132.5:c.1367C>T, NM_022132.5:c.1576_1577insT, NM_022132.5:c.1580G>A
MCOLN1	Mucolipidosis IV	NM_020533.3:c.304C>T, NM_020533.3:c.964C>T, NM_020533.3:c.1084G>T, NM_020533.3:c.1207C>T
MEFV	Familial Mediterranean fever	NM_000243.3:c.2282G>A, NM_000243.3:c.2230G>T, NM_000243.3:c.2177T>C, NM_000243.3:c.2084A>G, NM_000243.3:c.2082G>A, NM_000243.3:c.2080A>G, NM_000243.3:c.2076_2078delAAT, NM_000243.3:c.2040G>C, NM_000243.3:c.2040G>A, NM_000243.3:c.1958G>A, NM_000243.3:c.1772T>C, NM_000243.3:c.1437C>G, NM_000243.3:c.1223G>A, NM_000243.3:c.1141C>T, NM_000243.3:c.1016C>T, NM_000243.3:c.800C>T, NM_000243.3:c.688G>A, NM_000243.3:c.656dupG, NM_000243.3:c.501G>C, NM_000243.3:c.443A>T, NM_000243.3:c.163dupA
MFSD8	Ceroid Lipofuscinosis, Neuronal, 7	NM_001371596.2:c.1525_1526delCT, NM_001371596.2:c.1286G>A, NM_001371596.2:c.1235C>T, NM_001371596.2:c.1090delA, NM_001371596.2:c.999-2A>G, NM_001371596.2:c.929G>A, NM_001371596.2:c.894T>G, NM_001371596.2:c.881C>A, NM_001371596.2:c.362A>G
MKKS	Bardet-Biedl Syndrome 6	NM_170784.3:c.1436C>G, NM_170784.3:c.1225_1226delGG, NM_170784.3:c.830T>C, NM_170784.3:c.724G>T, NM_170784.3:c.353delG, NM_170784.3:c.250C>T
MKS1	Bardet-Biedl syndrome 13/Joubert syndrome 28/Meckel syndrome 1	NM_017777.4:c.1349T>C, NM_017777.4:c.1319G>C, NM_017777.4:c.1024+1G>A, NM_017777.4:c.857A>G, NM_017777.4:c.814G>C, NM_017777.4:c.508C>T
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	NM_015166.4:c.839C>T, NM_015166.4:c.424-2A>C, NM_015166.4:c.423C>A, NM_015166.4:c.422A>G, NM_015166.4:c.278C>T, NM_015166.4:c.274C>T, NM_015166.4:c.206C>T, NM_015166.4:c.135dupC, NM_015166.4:c.33dupC
MLYCD	Malonyl-CoA decarboxylase deficiency	NM_012213.3:c.560C>G, NM_012213.3:c.679_680insTGAAGC, NM_012213.3:c.755delT
MMAA	Methylmalonic Aciduria, MMAA-Related	NM_172250.3:c.283C>T, NM_172250.3:c.387C>A, NM_172250.3:c.440G>A, NM_172250.3:c.447_448insG, NM_172250.3:c.451delC, NM_172250.3:c.503delC, NM_172250.3:c.586C>T, NM_172250.3:c.620A>G, NM_172250.3:c.811G>T, NM_172250.3:c.1032delT
MMAB	Methylmalonic Aciduria, MMAB-Related	NM_052845.4:c.700C>T, NM_052845.4:c.577G>A, NM_052845.4:c.569G>A, NM_052845.4:c.568C>T, NM_052845.4:c.557G>A, NM_052845.4:c.556C>T, NM_052845.4:c.548A>T, NM_052845.4:c.220G>T, NM_052845.4:c.197-1G>T, NM_052845.4:c.197-1G>A
MMACHC	Cobalamin C disease	NM_015506.3:c.270_271insA, NM_015506.3:c.331C>T, NM_015506.3:c.347T>C, NM_015506.3:c.382_384delTAC, NM_015506.3:c.388T>C, NM_015506.3:c.389A>G, NM_015506.3:c.394C>T, NM_015506.3:c.440G>C, NM_015506.3:c.481C>T, NM_015506.3:c.482G>A, NM_015506.3:c.544_545delTG, NM_015506.3:c.608G>A, NM_015506.3:c.609G>A, NM_015506.3:c.615C>A, NM_015506.3:c.615C>G, NM_015506.3:c.616_617insG, NM_015506.3:c.616C>T, NM_015506.3:c.656_658delAGA, NM_015506.3:c.688C>T
MMADHC	Homocystinuria, cblD type, variant 1/Methylmalonic aciduria and homocystinuria, cblD type/Methylmalonic aciduria, cblD type, variant 2	NM_015702.3:c.795dupT, NM_015702.3:c.776T>C, NM_015702.3:c.748C>T, NM_015702.3:c.746A>G, NM_015702.3:c.737A>G, NM_015702.3:c.545C>A, NM_015702.3:c.478+1G>T, NM_015702.3:c.419dupA, NM_015702.3:c.57_64delCTCTTTAG

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
MMUT	Methylmalonic acidemia	NM_000255.4:c.2150G>T, NM_000255.4:c.2080C>T, NM_000255.4:c.1924G>C, NM_000255.4:c.1871A>G, NM_000255.4:c.1867G>A, NM_000255.4:c.1741C>T, NM_000255.4:c.1658delT, NM_000255.4:c.1445-2A>G, NM_000255.4:c.1420C>T, NM_000255.4:c.1399C>T, NM_000255.4:c.1280G>A, NM_000255.4:c.1207C>T, NM_000255.4:c.1181T>A, NM_000255.4:c.1106G>A, NM_000255.4:c.914T>C, NM_000255.4:c.682C>T, NM_000255.4:c.671_678dupAATTTATG, NM_000255.4:c.655A>T, NM_000255.4:c.643G>A, NM_000255.4:c.607G>A, NM_000255.4:c.572C>A, NM_000255.4:c.313T>C, NM_000255.4:c.280G>A, NM_000255.4:c.278G>A, NM_000255.4:c.91C>T
MOCS1	Molybdenum cofactor deficiency A	NM_001358530.2:c.1027C>T, NM_001358530.2:c.956G>A, NM_001358530.2:c.397_406delCCGGACGTGG, NM_001358530.2:c.218G>A, NM_001358530.2:c.217C>T
MPI	Congenital disorder of glycosylation, type Ib	NM_002435.3:c.305C>T, NM_002435.3:c.413T>C, NM_002435.3:c.656G>A, NM_002435.3:c.884G>A, NM_002435.3:c.982C>T, NM_002435.3:c.1016_1019delACCC
MPV17	Mitochondrial DNA depletion syndrome	NM_002437.5:c.498C>A, NM_002437.5:c.462-2A>C, NM_002437.5:c.359G>A, NM_002437.5:c.284dupG, NM_002437.5:c.263_265delAGA, NM_002437.5:c.263A>T, NM_002437.5:c.149G>A, NM_002437.5:c.148C>T, NM_002437.5:c.70G>T
MTHFR	Homocystinuria due to MTHFR deficiency	NM_005957.5:c.1768delC, NM_005957.5:c.1743G>A, NM_005957.5:c.1129C>T, NM_005957.5:c.971A>G, NM_005957.5:c.968T>C, NM_005957.5:c.547C>T, NM_005957.5:c.439C>T, NM_005957.5:c.3G>A
MTM1	Myotubular Myopathy, X-Linked	NM_000252.3:c.70C>T, NM_000252.3:c.420C>G, NM_000252.3:c.461T>G, NM_000252.3:c.594_598delCCCTG, NM_000252.3:c.670C>T, NM_000252.3:c.721C>T, NM_000252.3:c.780T>A, NM_000252.3:c.962_963insA, NM_000252.3:c.963delA, NM_000252.3:c.1261-10A>G, NM_000252.3:c.1304_1305insTCCTA, NM_000252.3:c.1356_1357delCC, NM_000252.3:c.1415_1416delGT
MTTP	Abetalipoproteinemia	NM_001386140.1:c.703_704delAC, NM_001386140.1:c.1619G>A, NM_001386140.1:c.1769G>T, NM_001386140.1:c.1867+1G>A, NM_001386140.1:c.2030delC, NM_001386140.1:c.2593G>T
MYO15A	Deafness, autosomal recessive 3	NM_016239.4:c.625G>T, NM_016239.4:c.754_755insA, NM_016239.4:c.3313G>T, NM_016239.4:c.3334delG, NM_016239.4:c.3385C>T, NM_016239.4:c.3693-2A>G, NM_016239.4:c.3756+1G>T, NM_016239.4:c.4351G>A, NM_016239.4:c.4750_4751insTC, NM_016239.4:c.5326C>T, NM_016239.4:c.5492G>T, NM_016239.4:c.6003delG, NM_016239.4:c.6046+2T>G, NM_016239.4:c.6614C>T, NM_016239.4:c.6743C>T, NM_016239.4:c.6863_6873delCGGACCTGGAG, NM_016239.4:c.8148G>T, NM_016239.4:c.8410A>T, NM_016239.4:c.8429_8447delGCGGGCAGCTGCGGGTCCT, NM_016239.4:c.8548C>T, NM_016239.4:c.9956_9959delCTGA, NM_016239.4:c.10573delA
MYO7A	Usher syndrome, type 1B	NM_000260.4:c.3G>A, NM_000260.4:c.133-2A>G, NM_000260.4:c.448C>T, NM_000260.4:c.494C>T, NM_000260.4:c.634C>T, NM_000260.4:c.635G>A, NM_000260.4:c.640G>A, NM_000260.4:c.731G>C, NM_000260.4:c.1184G>A, NM_000260.4:c.1344-1G>A, NM_000260.4:c.1797G>A, NM_000260.4:c.1884C>A, NM_000260.4:c.1996C>T, NM_000260.4:c.2023C>T, NM_000260.4:c.2476G>A, NM_000260.4:c.2617C>T, NM_000260.4:c.3134T>C, NM_000260.4:c.3504-1G>C, NM_000260.4:c.3508G>A, NM_000260.4:c.3595_3596insT, NM_000260.4:c.3719G>A, NM_000260.4:c.3763delA, NM_000260.4:c.4018G>A, NM_000260.4:c.4024delT, NM_000260.4:c.5227C>T, NM_000260.4:c.5392C>T, NM_000260.4:c.5507T>G, NM_000260.4:c.5618G>A, NM_000260.4:c.5824G>T, NM_000260.4:c.5884_5887delTTCT, NM_000260.4:c.5967C>G, NM_000260.4:c.6024delG
NAGS	N-acetylglutamate Synthase Deficiency	NM_153006.3:c.916-2A>T, NM_153006.3:c.971G>A, NM_153006.3:c.1025delG, NM_153006.3:c.1289T>C, NM_153006.3:c.1299G>C, NM_153006.3:c.1306_1307insT
NDRG1	Charcot-Marie-Tooth Disease type 4D	NM_006096.4:c.928C>T, NM_006096.4:c.538-1G>A, NM_006096.4:c.442C>T, NM_006096.4:c.16C>T, NM_006096.4:c.-18-2_-18-1delAG
NEB	Nemaline myopathy 2, autosomal recessive	NM_001164507.2:c.25404+1_25404+2insATGGA, NM_001164507.2:c.25174G>T, NM_001164507.2:c.24874-1G>A, NM_001164507.2:c.24687_24688delGA, NM_001164507.2:c.24665_24666delTT, NM_001164507.2:c.23989C>T, NM_001164507.2:c.23421_23422delAG, NM_001164507.2:c.21945+1G>A, NM_001164507.2:c.21076C>T, NM_001164507.2:c.19285_19286delGCinsAA, NM_001164507.2:c.12203_12204delTG, NM_001164507.2:c.8031_8041delAAATAAACGAG, NM_001164507.2:c.6105dupT, NM_001164507.2:c.5567G>A, NM_001164507.2:c.3191A>G, NM_001164507.2:c.2173G>T, NM_001164507.2:c.843T>G
NPC1	Niemann-Pick disease, type C1	NM_000271.5:c.3662delT, NM_000271.5:c.3611_3614delTTAC, NM_000271.5:c.3467A>G, NM_000271.5:c.3425T>C, NM_000271.5:c.3182T>C, NM_000271.5:c.3175C>T, NM_000271.5:c.3107C>T, NM_000271.5:c.3104C>T, NM_000271.5:c.3019C>G, NM_000271.5:c.2974G>T, NM_000271.5:c.2972_2973delAG, NM_000271.5:c.2974G>A, NM_000271.5:c.2932C>T, NM_000271.5:c.2873G>A, NM_000271.5:c.2861C>T, NM_000271.5:c.2848G>A, NM_000271.5:c.2842G>A, NM_000271.5:c.2761C>T, NM_000271.5:c.2324A>C, NM_000271.5:c.2072C>T, NM_000271.5:c.1628C>T, NM_000271.5:c.1211G>A, NM_000271.5:c.1042C>T, NM_000271.5:c.813_815delCAT, NM_000271.5:c.743G>T, NM_000271.5:c.530G>A, NM_000271.5:c.352_353delAG, NM_000271.5:c.337T>C

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
NPC2	Niemann-pick disease, type C2	NM_006432.5:c.441+1G>A, NM_006432.5:c.436C>T, NM_006432.5:c.358C>T, NM_006432.5:c.352G>T, NM_006432.5:c.295T>C, NM_006432.5:c.190+5G>A, NM_006432.5:c.115G>A, NM_006432.5:c.58G>T, NM_006432.5:c.27delG
NPHP1	Juvenile Nephronophthisis	NM_001128178.3:c.1716+1G>T, NM_001128178.3:c.1016dupC, NM_001128178.3:c.771+58C>T, NM_001128178.3:c.555dupA, NM_001128178.3:c.455C>G, NM_001128178.3:c.80T>A, NM_001128178.3:c.1delA
NPHS1	Nephrotic syndrome, type 1	NM_004646.4:c.3478C>T, NM_004646.4:c.3325C>T, NM_004646.4:c.3109+1G>A, NM_004646.4:c.2928G>T, NM_004646.4:c.2746G>T, NM_004646.4:c.2491C>T, NM_004646.4:c.2464G>A, NM_004646.4:c.2456A>T, NM_004646.4:c.1715G>A, NM_004646.4:c.1481delC, NM_004646.4:c.1307_1308dupAC, NM_004646.4:c.121_122delCT, NM_004646.4:c.59-5C>G
NR2E3	Enhanced S-cone syndrome	NM_014249.4:c.119-2A>C, NM_014249.4:c.226C>T, NM_014249.4:c.227G>A, NM_014249.4:c.297_298delGT, NM_014249.4:c.361G>A, NM_014249.4:c.932G>A, NM_014249.4:c.1034_1038delTGCAG
NTRK1	Insensitivity to pain, congenital, with anhidrosis	NM_002529.4:c.1076A>G, NM_002529.4:c.1474G>A, NM_002529.4:c.1726delT, NM_002529.4:c.1729G>C, NM_002529.4:c.1759A>G, NM_002529.4:c.1870C>T, NM_002529.4:c.1926_1927insT, NM_002529.4:c.1960C>T, NM_002529.4:c.2084C>T, NM_002529.4:c.2339G>C
OAT	Ornithine Aminotransferase Deficiency	NM_000274.4:c.1276C>T, NM_000274.4:c.1250C>T, NM_000274.4:c.1205T>C, NM_000274.4:c.994G>A, NM_000274.4:c.955C>T, NM_000274.4:c.952delG, NM_000274.4:c.952G>A, NM_000274.4:c.901-2A>G, NM_000274.4:c.824G>A, NM_000274.4:c.812G>A, NM_000274.4:c.677C>T, NM_000274.4:c.627T>A, NM_000274.4:c.596C>A, NM_000274.4:c.539G>C, NM_000274.4:c.533G>A, NM_000274.4:c.278G>T, NM_000274.4:c.268C>G, NM_000274.4:c.159delC
OCRL	Lowe syndrome, X-Linked	NM_000276.4:c.903_904delAG, NM_000276.4:c.1499G>A, NM_000276.4:c.2299C>T, NM_000276.4:c.2402_2403insA, NM_000276.4:c.2530C>T, NM_000276.4:c.2534delA
OTC	Ornithine transcarbamylase deficiency	NM_000531.6:c.77G>A, NM_000531.6:c.118C>T, NM_000531.6:c.119G>A, NM_000531.6:c.134T>C, NM_000531.6:c.148G>T, NM_000531.6:c.238A>G, NM_000531.6:c.245T>G, NM_000531.6:c.259G>A, NM_000531.6:c.275G>A, NM_000531.6:c.332T>C, NM_000531.6:c.421C>T, NM_000531.6:c.460G>T, NM_000531.6:c.563G>T, NM_000531.6:c.589G>T, NM_000531.6:c.617T>G, NM_000531.6:c.646C>G, NM_000531.6:c.674C>T, NM_000531.6:c.717+2T>C, NM_000531.6:c.829C>T
PAH	Phenylalanine hydroxylase deficiency (including phenylketonuria)	NM_000277.3:c.1315+1G>A, NM_000277.3:c.1243G>A, NM_000277.3:c.1241A>G, NM_000277.3:c.1238G>C, NM_000277.3:c.1222C>T, NM_000277.3:c.1217T>C, NM_000277.3:c.1208C>T, NM_000277.3:c.1199+17G>A, NM_000277.3:c.1199+1G>A, NM_000277.3:c.1197A>T, NM_000277.3:c.1184C>A, NM_000277.3:c.1169A>G, NM_000277.3:c.1166delC, NM_000277.3:c.1162G>A, NM_000277.3:c.1139C>T, NM_000277.3:c.1068C>A, NM_000277.3:c.1066-3C>T, NM_000277.3:c.1066-11G>A, NM_000277.3:c.1045T>C, NM_000277.3:c.1042C>G, NM_000277.3:c.1033G>T, NM_000277.3:c.1030G>A, NM_000277.3:c.955G>T, NM_000277.3:c.926C>T, NM_000277.3:c.926C>A, NM_000277.3:c.912+1G>A, NM_000277.3:c.898G>T, NM_000277.3:c.896T>G, NM_000277.3:c.842+5G>A, NM_000277.3:c.838G>A, NM_000277.3:c.829T>G, NM_000277.3:c.823C>T, NM_000277.3:c.818C>T, NM_000277.3:c.814G>T, NM_000277.3:c.809G>A, NM_000277.3:c.806delT, NM_000277.3:c.782G>A, NM_000277.3:c.764T>C, NM_000277.3:c.755G>A, NM_000277.3:c.754C>T, NM_000277.3:c.745C>T, NM_000277.3:c.737C>A, NM_000277.3:c.734T>C, NM_000277.3:c.733G>C, NM_000277.3:c.728G>A, NM_000277.3:c.727C>T, NM_000277.3:c.722delG, NM_000277.3:c.722G>A, NM_000277.3:c.721C>T, NM_000277.3:c.688G>A, NM_000277.3:c.673C>G, NM_000277.3:c.665A>G, NM_000277.3:c.638T>C, NM_000277.3:c.611A>G, NM_000277.3:c.569T>C, NM_000277.3:c.533A>G, NM_000277.3:c.529G>A, NM_000277.3:c.527G>T, NM_000277.3:c.509+1G>A, NM_000277.3:c.508C>G, NM_000277.3:c.503delA, NM_000277.3:c.490A>G, NM_000277.3:c.482T>C, NM_000277.3:c.473G>A, NM_000277.3:c.472C>T, NM_000277.3:c.450dupA, NM_000277.3:c.442-1G>A, NM_000277.3:c.442-5C>G, NM_000277.3:c.441+5G>T, NM_000277.3:c.441+1G>A, NM_000277.3:c.357delC, NM_000277.3:c.331C>T, NM_000277.3:c.320A>G, NM_000277.3:c.311C>A, NM_000277.3:c.284_286delTCA, NM_000277.3:c.261C>A, NM_000277.3:c.250G>T, NM_000277.3:c.232G>A, NM_000277.3:c.204A>T, NM_000277.3:c.194T>C, NM_000277.3:c.165T>G, NM_000277.3:c.158G>A, NM_000277.3:c.157C>T, NM_000277.3:c.143T>C, NM_000277.3:c.136G>A, NM_000277.3:c.117C>G, NM_000277.3:c.47_48delCT
PANK2	Pantothenate Kinase-Associated Neurodegeneration	NM_001386393.1:c.460C>T, NM_001386393.1:c.491_492delCT, NM_001386393.1:c.881A>T, NM_001386393.1:c.1231G>A, NM_001386393.1:c.1253C>T
PC	Pyruvate Carboxylase Deficiency	NM_001040716.2:c.1748G>T, NM_001040716.2:c.496G>A, NM_001040716.2:c.434T>C
PCCA	Propionic acidemia	NM_000282.4:c.229C>T, NM_000282.4:c.259_260insT, NM_000282.4:c.412G>A, NM_000282.4:c.600+1G>A, NM_000282.4:c.862A>T, NM_000282.4:c.1022_1023insT, NM_000282.4:c.1118T>A, NM_000282.4:c.1224_1225delTT, NM_000282.4:c.1284+1G>A, NM_000282.4:c.1594_1597delTTGT, NM_000282.4:c.1891G>C, NM_000282.4:c.1899+1_1899+4delGTAA

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1,
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at the time of analysis are reported.

PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
PCCB	Propionicacidemia	NM_000532.5:c.331C>T, NM_000532.5:c.337C>T, NM_000532.5:c.502G>A, NM_000532.5:c.562G>A, NM_000532.5:c.683C>T, NM_000532.5:c.737G>T, NM_000532.5:c.984_985insT, NM_000532.5:c.1169_1170insT, NM_000532.5:c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA, NM_000532.5:c.1219_1224delGGCATCinsAA, NM_000532.5:c.1223_1226delTCAT, NM_000532.5:c.1228C>T, NM_000532.5:c.1229_1230insT, NM_000532.5:c.1283C>T, NM_000532.5:c.1304A>G, NM_000532.5:c.1490C>T, NM_000532.5:c.1534C>T, NM_000532.5:c.1537_1538insCCC, NM_000532.5:c.1606A>G
PCDH15	Usher syndrome, type 1F	NM_033056.4:c.5724_5755delACGCACAAATGTTTCAGAACTTCAAACTATGT, NM_033056.4:c.5659A>T, NM_033056.4:c.4961_4962insTGAT, NM_033056.4:c.4937_4940dupTGAT, NM_033056.4:c.4864delA, NM_033056.4:c.4548_4551dupATCT, NM_001384140.1:c.3718-2A>G, NM_001384140.1:c.2645_2646delAT, NM_001384140.1:c.1940C>G, NM_001384140.1:c.1737C>G, NM_001384140.1:c.1583T>A, NM_001384140.1:c.1088delT, NM_001384140.1:c.1021C>T, NM_001384140.1:c.1006C>T, NM_001384140.1:c.785G>A, NM_001384140.1:c.400C>T, NM_001384140.1:c.400C>G, NM_001384140.1:c.7C>T
PDHA1	Pyruvate Dehydrogenase Deficiency, X-Linked	NM_000284.4:c.262C>T, NM_000284.4:c.773A>C, NM_000284.4:c.787C>G, NM_000284.4:c.871G>A
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	NM_000466.3:c.3505_3517delCAGTTGTTTTCAC, NM_000466.3:c.2916delA, NM_000466.3:c.2528G>A, NM_000466.3:c.2097dupT, NM_000466.3:c.1991T>C, NM_000466.3:c.1952_1960dupCAGTGTGGA, NM_000466.3:c.1842delA, NM_000466.3:c.1239+1G>T, NM_000466.3:c.877C>T
PEX12	Peroxisome Biogenesis Disorder 3A (Zellweger)	NM_000286.3:c.959C>T, NM_000286.3:c.894delC, NM_000286.3:c.888_889delCT, NM_000286.3:c.771delC, NM_000286.3:c.538C>T, NM_000286.3:c.455_459dupGGAAA
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	NM_000318.3:c.789_790delCT, NM_000318.3:c.163G>A
PEX7	Rhizomelic chondrodysplasia punctata type 1	NM_000288.4:c.532C>T, NM_000288.4:c.618G>A, NM_000288.4:c.649G>A, NM_000288.4:c.653C>T, NM_000288.4:c.694C>T, NM_000288.4:c.722A>T, NM_000288.4:c.854A>G, NM_000288.4:c.875T>A, NM_000288.4:c.903+1G>C
PKHD1	Polycystic kidney disease, autosomal recessive	NM_138694.4:c.12027C>G, NM_138694.4:c.11611T>C, NM_138694.4:c.11363_11372delCTTCCCTGGA, NM_138694.4:c.11284C>A, NM_138694.4:c.10585G>C, NM_138694.4:c.10515C>A, NM_138694.4:c.10452dupT, NM_138694.4:c.10412T>G, NM_138694.4:c.10219C>T, NM_138694.4:c.10036T>C, NM_138694.4:c.9866G>T, NM_138694.4:c.9719G>A, NM_138694.4:c.9689delA, NM_138694.4:c.9530T>C, NM_138694.4:c.9370C>T, NM_138694.4:c.9107T>G, NM_138694.4:c.8870T>C, NM_138694.4:c.8824C>T, NM_138694.4:c.8518C>T, NM_138694.4:c.8408G>A, NM_138694.4:c.8407T>C, NM_138694.4:c.8317G>T, NM_138694.4:c.6854G>A, NM_138694.4:c.6499C>T, NM_138694.4:c.5895dupA, NM_138694.4:c.5498C>T, NM_138694.4:c.5325_5326delAG, NM_138694.4:c.4870C>T, NM_138694.4:c.4165C>A, NM_138694.4:c.3940delA, NM_138694.4:c.3766delC, NM_138694.4:c.3761_3762delCCinsG, NM_138694.4:c.3367G>A, NM_138694.4:c.3229-2A>C, NM_138694.4:c.2854G>A, NM_138694.4:c.2827_2828delGA, NM_138694.4:c.2747A>C, NM_138694.4:c.2452C>T, NM_138694.4:c.2414C>T, NM_138694.4:c.2341C>T, NM_138694.4:c.1486C>T, NM_138694.4:c.1409G>A, NM_138694.4:c.1342G>C, NM_138694.4:c.982C>T, NM_138694.4:c.930delC, NM_138694.4:c.682A>G, NM_138694.4:c.664A>G, NM_138694.4:c.370C>T, NM_138694.4:c.353delG, NM_138694.4:c.107C>T, NM_138694.4:c.85G>T
PLA2G6	Infantile neuroaxonal dystrophy 1	NM_003560.4:c.2370T>G, NM_003560.4:c.2239C>T, NM_003560.4:c.1903C>T, NM_003560.4:c.1894C>T, NM_003560.4:c.1634A>C, NM_003560.4:c.1612C>T, NM_003560.4:c.929T>A, NM_003560.4:c.238G>A, NM_003560.4:c.109C>T
PMM2	Congenital disorder of glycosylation type Ia	NM_000303.3:c.26G>A, NM_000303.3:c.53C>G, NM_000303.3:c.95_96delTAinsGC, NM_000303.3:c.95T>G, NM_000303.3:c.97C>T, NM_000303.3:c.109C>T, NM_000303.3:c.127G>C, NM_000303.3:c.131T>C, NM_000303.3:c.190delT, NM_000303.3:c.193G>T, NM_000303.3:c.255+2T>C, NM_000303.3:c.256-1G>C, NM_000303.3:c.317A>T, NM_000303.3:c.323C>T, NM_000303.3:c.338C>T, NM_000303.3:c.349G>C, NM_000303.3:c.357C>A, NM_000303.3:c.368G>A, NM_000303.3:c.385G>A, NM_000303.3:c.395T>C, NM_000303.3:c.415G>A, NM_000303.3:c.422G>A, NM_000303.3:c.442G>A, NM_000303.3:c.470T>C, NM_000303.3:c.484C>T, NM_000303.3:c.563A>G, NM_000303.3:c.620T>C, NM_000303.3:c.623G>C, NM_000303.3:c.647A>T, NM_000303.3:c.652C>G, NM_000303.3:c.669C>G, NM_000303.3:c.677C>G, NM_000303.3:c.691G>A, NM_000303.3:c.710C>G, NM_000303.3:c.710C>T
PNPO	Pyridoxal 5'-phosphate-dependent epilepsy	NM_018129.4:c.674G>A, NM_018129.4:c.685C>T
POLG	POLG-Related Disorders	NM_002693.3:c.3644-1G>A, NM_002693.3:c.3630dupC, NM_002693.3:c.3286C>T, NM_002693.3:c.3218C>T, NM_002693.3:c.3151G>C, NM_002693.3:c.2794C>T, NM_002693.3:c.2617G>T, NM_002693.3:c.2605C>T, NM_002693.3:c.2591A>G, NM_002693.3:c.2557C>T, NM_002693.3:c.2542G>A, NM_002693.3:c.2243G>C, NM_002693.3:c.2209G>C, NM_002693.3:c.2207A>G, NM_002693.3:c.1879C>T,

GENESeeker

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_002693.3:c.1760C>T, NM_002693.3:c.1754G>A, NM_002693.3:c.1550G>T, NM_002693.3:c.1491G>C, NM_002693.3:c.1437C>G, NM_002693.3:c.1399G>A, NM_002693.3:c.1120C>T, NM_002693.3:c.911T>G, NM_002693.3:c.830A>T, NM_002693.3:c.803G>C, NM_002693.3:c.752C>T
POMGNT1	Muscle-Eye-Brain Disease, POMGNT1-Related	NM_017739.4:c.1864delC, NM_017739.4:c.1814G>C, NM_017739.4:c.1666G>A, NM_017739.4:c.1545delC, NM_017739.4:c.1539+1G>T, NM_017739.4:c.1539+1G>A, NM_017739.4:c.1469G>A, NM_017739.4:c.1425G>A, NM_017739.4:c.1411A>T, NM_017739.4:c.1274G>C, NM_017739.4:c.932G>A, NM_017739.4:c.931C>T, NM_017739.4:c.880-1G>A, NM_017739.4:c.794G>A, NM_017739.4:c.652+1G>A, NM_017739.4:c.636C>T, NM_017739.4:c.187C>T, NM_017739.4:c.92dupA
PPT1	Ceroid Lipofuscinosis, Neuronal, 1	NM_000310.4:c.840dupA, NM_000310.4:c.627+1G>T, NM_000310.4:c.541G>T, NM_000310.4:c.451C>T, NM_000310.4:c.223A>C, NM_000310.4:c.29T>A
PROP1	Combined Pituitary Hormone Deficiency 2	NM_006261.5:c.469dupT, NM_006261.5:c.373C>T, NM_006261.5:c.358C>T, NM_006261.5:c.349T>A, NM_006261.5:c.310delC, NM_006261.5:c.301_302delAG, NM_006261.5:c.295C>T, NM_006261.5:c.263T>C, NM_006261.5:c.247C>T, NM_006261.5:c.218G>A, NM_006261.5:c.217C>T, NM_006261.5:c.157delA, NM_006261.5:c.150delA, NM_006261.5:c.112_124delTCGAGTGCTCCAC, NM_006261.5:c.4delG, NM_006261.5:c.2T>C, NM_006261.5:c.343-11C>G
PRPS1	Arts syndrome, X-Linked	NM_002764.4:c.193G>A, NM_002764.4:c.344T>C, NM_002764.4:c.398A>C, NM_002764.4:c.455T>C, NM_002764.4:c.869T>C, NM_002764.4:c.916G>A
PSAP	Metachromatic Leukodystrophy, PSAP-Related	NM_002778.4:c.1288C>T, NM_002778.4:c.1046T>C, NM_002778.4:c.643A>C, NM_002778.4:c.607C>T, NM_002778.4:c.1A>T
PYGM	McArdle disease	NM_005609.4:c.2392T>C, NM_005609.4:c.2262delA, NM_005609.4:c.2128_2130delTTC, NM_005609.4:c.2009C>T, NM_005609.4:c.1963G>A, NM_005609.4:c.1827G>A, NM_005609.4:c.1768+1G>A, NM_005609.4:c.1726C>T, NM_005609.4:c.1722T>G, NM_005609.4:c.1628A>C, NM_005609.4:c.1621G>T, NM_005609.4:c.1466C>G, NM_005609.4:c.1094C>T, NM_005609.4:c.613G>A, NM_005609.4:c.501dupT, NM_005609.4:c.481C>T, NM_005609.4:c.393delG, NM_005609.4:c.280C>T, NM_005609.4:c.255C>A, NM_005609.4:c.148C>T, NM_005609.4:c.13_14delCT, NM_005609.4:c.1A>G
RAB23	Carpenter Syndrome	NM_016277.5:c.434T>A, NM_016277.5:c.407dupC
RAG1	Omenn syndrome / T- B- severe combined immunodeficiency	NM_000448.3:c.256_257delAA, NM_000448.3:c.940C>T, NM_000448.3:c.983G>A, NM_000448.3:c.1681C>T, NM_000448.3:c.1682G>A, NM_000448.3:c.2164G>A, NM_000448.3:c.2320G>T, NM_000448.3:c.2326C>T, NM_000448.3:c.2333G>A, NM_000448.3:c.2814T>G, NM_000448.3:c.2923C>T, NM_000448.3:c.3016A>G
RAG2	Omenn syndrome / T- B- severe combined immunodeficiency	NM_000536.4:c.1504A>G, NM_000536.4:c.1352G>C, NM_000536.4:c.686G>A, NM_000536.4:c.685C>T, NM_000536.4:c.601C>T, NM_000536.4:c.283G>A, NM_000536.4:c.230C>A, NM_000536.4:c.115A>G
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	NM_005055.5:c.848T>C, NM_005055.5:c.807C>A, NM_005055.5:c.603C>A, NM_005055.5:c.566C>T, NM_005055.5:c.490C>T, NM_005055.5:c.484G>A, NM_005055.5:c.416T>C, NM_005055.5:c.264C>A
RDH12	Leber congenital amaurosis 13	NM_152443.3:c.146C>T, NM_152443.3:c.152T>A, NM_152443.3:c.164C>T, NM_152443.3:c.184C>T, NM_152443.3:c.209_210insC, NM_152443.3:c.295C>A, NM_152443.3:c.377C>T, NM_152443.3:c.379G>T, NM_152443.3:c.448+1_448+4delGTAA, NM_152443.3:c.451C>A, NM_152443.3:c.451C>G, NM_152443.3:c.464C>T, NM_152443.3:c.523T>C, NM_152443.3:c.565C>T, NM_152443.3:c.677A>G, NM_152443.3:c.805_809delGCCCT
RLBP1	Bothnia retinal dystrophy/Fundus albipunctatus/Retinitis punctata albescens	NM_000326.5:c.875C>T, NM_000326.5:c.700C>T, NM_000326.5:c.452G>A, NM_000326.5:c.333T>G
RPE65	Leber congenital amaurosis 2	NM_000329.3:c.1543C>T, NM_000329.3:c.1355T>G, NM_000329.3:c.1301C>T, NM_000329.3:c.1292A>G, NM_000329.3:c.1102T>C, NM_000329.3:c.1087C>A, NM_000329.3:c.1022T>C, NM_000329.3:c.907A>T, NM_000329.3:c.881A>C, NM_000329.3:c.514_515delGT, NM_000329.3:c.394G>A, NM_000329.3:c.272G>A, NM_000329.3:c.271C>T
RPGRIP1L	Ciliopathies, RPGRIP1L-Related	NM_015272.5:c.3706C>T, NM_015272.5:c.3634_3637delGAAA, NM_015272.5:c.3548C>G, NM_015272.5:c.2794_2795delTT, NM_015272.5:c.2614C>T, NM_015272.5:c.2413C>T, NM_015272.5:c.2050C>T, NM_015272.5:c.2030C>T, NM_015272.5:c.1975T>C, NM_015272.5:c.1843A>C, NM_015272.5:c.1326_1329delAAAA, NM_015272.5:c.1329dupA, NM_015272.5:c.1177G>A, NM_015272.5:c.776+1G>A,

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
		NM_015272.5:c.757C>T, NM_015272.5:c.697A>T, NM_015272.5:c.394A>T
SACS	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	NM_014363.6:c.13237C>T, NM_014363.6:c.12160C>T, NM_014363.6:c.11624G>A, NM_014363.6:c.10954C>A, NM_014363.6:c.10907G>A, NM_014363.6:c.8844delT, NM_014363.6:c.8107C>T, NM_014363.6:c.7504C>T, NM_014363.6:c.6781C>A, NM_014363.6:c.6563T>A, NM_014363.6:c.6355C>T, NM_014363.6:c.5618_5619delAT, NM_014363.6:c.4933C>T, NM_014363.6:c.3198T>A, NM_014363.6:c.994A>T, NM_014363.6:c.517C>T
SBDS	Shwachman-Diamond syndrome	NM_016038.4:c.652C>T, NM_016038.4:c.505C>T, NM_016038.4:c.377G>C, NM_016038.4:c.258+2T>C, NM_016038.4:c.183_184delTAinsCT, NM_016038.4:c.184A>T, NM_016038.4:c.127G>T, NM_016038.4:c.120delG
SERPINA1	Alpha-1-antitrypsin deficiency	NM_000295.5:c.1177C>T, NM_000295.5:c.1093G>A, NM_000295.5:c.848A>T, NM_000295.5:c.839A>T, NM_000295.5:c.739C>T, NM_000295.5:c.514G>T, NM_000295.5:c.514G>A, NM_000295.5:c.415G>A, NM_000295.5:c.347T>A, NM_000295.5:c.272G>A, NM_000295.5:c.250G>A, NM_000295.5:c.230C>T, NM_000295.5:c.194T>C, NM_000295.5:c.187C>T
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3	NM_000023.4:c.101G>A, NM_000023.4:c.229C>T, NM_000023.4:c.371T>C, NM_000023.4:c.518T>C, NM_000023.4:c.574C>T, NM_000023.4:c.662G>A, NM_000023.4:c.739G>A, NM_000023.4:c.850C>T, NM_000023.4:c.901_902insCC
SGCB	Limb-Girdle Muscular Dystrophy, Type 2E	NM_000232.5:c.552T>G, NM_000232.5:c.452C>G, NM_000232.5:c.341C>T, NM_000232.5:c.323T>G, NM_000232.5:c.299T>A, NM_000232.5:c.272G>T, NM_000232.5:c.272G>C
SGCG	Muscular dystrophy, limb-girdle, type 2C	NM_000231.3:c.88delG, NM_000231.3:c.195_195+3delAGTA, NM_000231.3:c.505+1G>A, NM_000231.3:c.521delT, NM_000231.3:c.787G>A, NM_000231.3:c.848G>A
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A)	NM_000199.5:c.1380delT, NM_000199.5:c.1339G>A, NM_000199.5:c.1298G>A, NM_000199.5:c.1167C>A, NM_000199.5:c.892T>C, NM_000199.5:c.877C>T, NM_000199.5:c.757delG, NM_000199.5:c.752G>C, NM_000199.5:c.617G>C, NM_000199.5:c.466A>T, NM_000199.5:c.449G>A, NM_000199.5:c.416C>T, NM_000199.5:c.383C>T, NM_000199.5:c.364G>A, NM_000199.5:c.337_345delCAAGCTGGTinsGCACAGGTGAG, NM_000199.5:c.320delT, NM_000199.5:c.235A>C, NM_000199.5:c.220C>T, NM_000199.5:c.197C>G, NM_000199.5:c.130G>A
SLC12A6	Agenesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)	NM_001365088.1:c.3031C>T, NM_001365088.1:c.2023C>T, NM_001365088.1:c.1584_1585delCTinsG, NM_001365088.1:c.619C>T, NM_001365088.1:c.366T>G, NM_001365088.1:c.316+1G>A
SLC17A5	Sialic acid storage disorder, infantile	NM_012434.5:c.1259+1G>A, NM_012434.5:c.918T>G, NM_012434.5:c.500T>C, NM_012434.5:c.406A>G, NM_012434.5:c.115C>T, NM_012434.5:c.43G>T
SLC25A13	Citrullinemia, Type II	NM_014251.3:c.1813C>T, NM_014251.3:c.1801G>T, NM_014251.3:c.1801G>A, NM_014251.3:c.1799dupA, NM_014251.3:c.1592G>A, NM_014251.3:c.1411_1412delCT, NM_014251.3:c.1311+1G>A, NM_014251.3:c.1231-1G>A, NM_014251.3:c.1177+1G>A, NM_014251.3:c.1078C>T, NM_014251.3:c.852_855delTATG, NM_014251.3:c.674C>T, NM_014251.3:c.674C>A, NM_014251.3:c.615+5G>A, NM_014251.3:c.615+1G>C
SLC25A15	Ornithine translocase deficiency	NM_014252.4:c.44C>T, NM_014252.4:c.110T>G, NM_014252.4:c.212T>A, NM_014252.4:c.535C>T, NM_014252.4:c.538G>A, NM_014252.4:c.553_555delTTC, NM_014252.4:c.569G>A, NM_014252.4:c.658G>A, NM_014252.4:c.815C>T, NM_014252.4:c.824G>A
SLC26A2	Sulfate transporter-related osteochondrodysplasias, includes achondrogenesis type 1B, atelosteogenesis type 2, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia	NM_000112.4:c.496G>A, NM_000112.4:c.532C>T, NM_000112.4:c.767T>C, NM_000112.4:c.832delC, NM_000112.4:c.835C>T, NM_000112.4:c.1010_1012delTTG, NM_000112.4:c.1273A>G, NM_000112.4:c.1361A>C, NM_000112.4:c.1535C>A, NM_000112.4:c.1723delA, NM_000112.4:c.1878delG, NM_000112.4:c.1957T>A, NM_000112.4:c.2033G>T
SLC26A4	Pendred syndrome	NM_000441.2:c.269C>T, NM_000441.2:c.281C>T, NM_000441.2:c.412G>T, NM_000441.2:c.554G>C, NM_000441.2:c.563T>C, NM_000441.2:c.626G>T, NM_000441.2:c.707T>C, NM_000441.2:c.898A>C, NM_000441.2:c.915_916insG, NM_000441.2:c.918+2T>C, NM_000441.2:c.919-2A>G, NM_000441.2:c.961A>T, NM_000441.2:c.970A>T, NM_000441.2:c.1001G>T, NM_000441.2:c.1001+1G>T, NM_000441.2:c.1003T>C, NM_000441.2:c.1034T>A, NM_000441.2:c.1061T>C, NM_000441.2:c.1151A>G, NM_000441.2:c.1174A>T, NM_000441.2:c.1197delT, NM_000441.2:c.1226G>A, NM_000441.2:c.1229C>T, NM_000441.2:c.1246A>C,

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		NM_000441.2:c.1263+1G>A, NM_000441.2:c.1334T>G, NM_000441.2:c.1454C>T, NM_000441.2:c.1468A>C, NM_000441.2:c.1489G>A, NM_000441.2:c.1634T>G, NM_000441.2:c.1707+5G>A, NM_000441.2:c.1790T>C, NM_000441.2:c.1826T>G, NM_000441.2:c.1975G>C, NM_000441.2:c.2048T>C, NM_000441.2:c.2131G>A, NM_000441.2:c.2162C>T, NM_000441.2:c.2168A>G, NM_000441.2:c.2211G>C
SLC37A4	Glycogen storage disease Ib	NM_001467.6:c.1243C>T, NM_001467.6:c.1063G>T, NM_001467.6:c.1042_1043delCT, NM_001467.6:c.1016G>A, NM_001467.6:c.1015G>T, NM_001467.6:c.899G>A, NM_001467.6:c.706_708delGTG, NM_001467.6:c.593A>T, NM_001467.6:c.352T>C, NM_001467.6:c.287G>A, NM_001467.6:c.1124-2_1124-1delAG, NM_001467.6:c.83G>A
SLC45A2	Oculocutaneous albinism, Type 4	NM_016180.5:c.1121delT, NM_016180.5:c.986delC, NM_016180.5:c.469G>A
SLC4A11	Corneal Dystrophy and Perceptive Deafness	NM_001174089.2:c.2558G>A, NM_001174089.2:c.2557C>T, NM_001174089.2:c.2518A>G, NM_001174089.2:c.2480T>C, NM_001174089.2:c.2270C>T, NM_001174089.2:c.2216G>A, NM_001174089.2:c.2185_2192dupTATGACAC, NM_001174089.2:c.2176G>A, NM_001174089.2:c.1765C>T, NM_001174089.2:c.1418C>T, NM_001174089.2:c.1415G>A, NM_001174089.2:c.1343G>A, NM_001174089.2:c.990_991insA, NM_001174089.2:c.589T>C, NM_001174089.2:c.577C>T, NM_001174089.2:c.425_432delGCTTCGCC
SMN1	Spinal Muscular Atrophy	Exon 7del
SMPD1	Niemann-Pick disease, types A/B	NM_000543.5:c.96G>A, NM_000543.5:c.100_116delGGCCTGGTGCTGGCGCT, NM_000543.5:c.103_107delCTGGT, NM_000543.5:c.103_118delCTGGTGCTGGCGCTGG, NM_000543.5:c.105delG, NM_000543.5:c.353delC, NM_000543.5:c.475T>C, NM_000543.5:c.551C>T, NM_000543.5:c.558_574delGCCCCCCAAACCCCCTA, NM_000543.5:c.557C>T, NM_000543.5:c.558_559insC, NM_000543.5:c.559delC, NM_000543.5:c.573delT, NM_000543.5:c.688C>T, NM_000543.5:c.689G>A, NM_000543.5:c.730G>A, NM_000543.5:c.737delG, NM_000543.5:c.739G>A, NM_000543.5:c.742G>A, NM_000543.5:c.757G>C, NM_000543.5:c.781_803delAGCCTGTTGAGTGGGCTGGGCCC, NM_000543.5:c.788T>A, NM_000543.5:c.839_840insCATCCCCG, NM_000543.5:c.911T>C, NM_000543.5:c.940G>A, NM_000543.5:c.991delC, NM_000543.5:c.995C>G, NM_000543.5:c.1092-1G>C, NM_000543.5:c.1117C>T, NM_000543.5:c.1152G>A, NM_000543.5:c.1264-1G>T, NM_000543.5:c.1267C>T, NM_000543.5:c.1299T>G, NM_000543.5:c.1327C>T, NM_000543.5:c.1418_1419delCT, NM_000543.5:c.1426C>T, NM_000543.5:c.1624C>T, NM_000543.5:c.1628delA, NM_000543.5:c.1805G>A, NM_000543.5:c.1826_1828delGCC
STAR	Lipoid Congenital Adrenal Hyperplasia	NM_000349.3:c.772C>T, NM_000349.3:c.749G>A, NM_000349.3:c.577C>T, NM_000349.3:c.562C>T, NM_000349.3:c.559G>A, NM_000349.3:c.545G>T, NM_000349.3:c.545G>A
STRC	Deafness, autosomal recessive 16	NM_153700.2:c.5188C>T, NM_153700.2:c.5185C>T, NM_153700.2:c.5168_5171delTTCT, NM_153700.2:c.4545+1G>C, NM_153700.2:c.3556C>T
TAT	Tyrosinemia, Type II	NM_000353.3:c.1297C>T, NM_000353.3:c.1249C>T, NM_000353.3:c.668C>G, NM_000353.3:c.236-5A>G, NM_000353.3:c.169C>T
TCIRG1	Osteopetrosis, autosomal recessive 1	NM_006019.4:c.112_113delAG, NM_006019.4:c.179A>G, NM_006019.4:c.1213G>A, NM_006019.4:c.1331G>T, NM_006019.4:c.1674-1G>A, NM_006019.4:c.2236+1G>A, NM_006019.4:c.2415-3C>G
TFR2	Hemochromatosis, Type 3, TFR2-Related	NM_003227.4:c.2374G>A, NM_003227.4:c.2343G>A, NM_003227.4:c.2014C>T, NM_003227.4:c.1861_1872delGCCGTGGCCCAG, NM_003227.4:c.1665delC, NM_003227.4:c.1632_1633delGA, NM_003227.4:c.1473+1G>A, NM_003227.4:c.1469T>G, NM_003227.4:c.1403G>A, NM_003227.4:c.1330G>A, NM_003227.4:c.1235_1237delACA, NM_003227.4:c.1186C>T, NM_003227.4:c.949C>T, NM_003227.4:c.840C>G, NM_003227.4:c.750C>G, NM_003227.4:c.515T>A, NM_003227.4:c.313C>T
TH	Tyrosine hydroxylase deficiency	NM_000360.4:c.1388C>T, NM_000360.4:c.1141C>A, NM_000360.4:c.917G>A, NM_000360.4:c.733A>C, NM_000360.4:c.614T>C, NM_000360.4:c.605G>A
TMC1	Deafness, autosomal recessive 7	NM_138691.3:c.100C>T, NM_138691.3:c.425G>A, NM_138691.3:c.454-1G>C, NM_138691.3:c.1165C>T, NM_138691.3:c.1763+3A>G, NM_138691.3:c.1842G>A, NM_138691.3:c.1960A>G
TMEM216	Joubert syndrome 2	NM_001173990.3:c.78_81delGAAC, NM_001173990.3:c.218G>A, NM_001173990.3:c.218G>T, NM_001173990.3:c.230G>C, NM_001173990.3:c.253C>T, NM_001173990.3:c.341T>G
TPP1	Ceroid lipofuscinosis, neuronal, 2/Spinocerebellar ataxia, autosomal recessive 7	NM_000391.4:c.1340G>A, NM_000391.4:c.1093T>C, NM_000391.4:c.851G>T, NM_000391.4:c.827A>T, NM_000391.4:c.622C>T, NM_000391.4:c.616C>T, NM_000391.4:c.509-1G>C, NM_000391.4:c.141_144delGAGT
TREX1	Aicardi-Goutieres syndrome 1	NM_033629.6:c.341G>A, NM_033629.6:c.490C>T
TRIM32	Bardet-Biedl syndrome 11	NM_012210.4:c.1459G>A, NM_012210.4:c.1559delC

GENESeeker

List of variants analysed for the Geneseeker panel version 3.1, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

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GENE	DISEASE NAME	VAR NAME
TRIM37	Mulibrey nanism syndrome	NM_015294.6:c.2212delG, NM_015294.6:c.2056C>T, NM_015294.6:c.1668-1G>C, NM_015294.6:c.1478_1479delAG, NM_015294.6:c.1411C>T, NM_015294.6:c.1346dupA, NM_015294.6:c.1037_1040dupAGAT, NM_015294.6:c.965G>T, NM_015294.6:c.745C>T, NM_015294.6:c.496_500delAGGAA, NM_015294.6:c.326G>C, NM_015294.6:c.227T>C
TSEN54	Pontocerebellar hypoplasia	NM_207346.3:c.670_671delAA, NM_207346.3:c.736C>T, NM_207346.3:c.887G>A, NM_207346.3:c.919G>T, NM_207346.3:c.1027C>T, NM_207346.3:c.1039A>T
TSFM	Combined Oxidative Phosphorylation Deficiency 3	NM_005726.6:c.1_2delAT, NM_005726.6:c.21_22delGC, NM_005726.6:c.517delC, NM_005726.6:c.856C>T
TSHB	Congenital hypothyroidism	NM_000549.5:c.94G>T, NM_000549.5:c.145G>A, NM_000549.5:c.205C>T
TSHR	Hypothyroidism, congenital, nongoitrous, 1	NM_000369.5:c.100G>A, NM_000369.5:c.122G>C, NM_000369.5:c.202C>T, NM_000369.5:c.326G>A, NM_000369.5:c.484C>G, NM_000369.5:c.500T>A, NM_000369.5:c.1170T>G, NM_000369.5:c.1741_1742insC
TTN	Familial dilated cardiomyopathy	NM_001267550.2:c.107889delA, NM_001267550.2:c.106070_106071delAT, NM_001267550.2:c.104092delC, NM_001267550.2:c.104092C>T, NM_001267550.2:c.102271C>T, NM_001267550.2:c.98818_98821delTCCA, NM_001267550.2:c.92373_92379delTGAATTC, NM_001267550.2:c.69344C>G, NM_001267550.2:c.60681dupT, NM_001267550.2:c.56648-1G>A, NM_001267550.2:c.52372delG, NM_001267550.2:c.48253delA, NM_001267550.2:c.47915dupT, NM_001267550.2:c.39082G>A, NM_001267550.2:c.32471-1G>A, NM_001267550.2:c.28300_28303delAGCA, NM_001267550.2:c.25978G>A, NM_001267550.2:c.16881C>A, NM_001267550.2:c.15796C>T, NM_001267550.2:c.4724_4728delTGAAA, NM_001267550.2:c.3165-1G>T
TTPA	Ataxia with Vitamin E Deficiency	NM_000370.3:c.744delA, NM_000370.3:c.661C>T, NM_000370.3:c.575G>A
TYR	Albinism, oculocutaneous, type IA	NM_000372.5:c.1A>G, NM_000372.5:c.115T>G, NM_000372.5:c.140G>A, NM_000372.5:c.164G>A, NM_000372.5:c.230G>A, NM_000372.5:c.242C>T, NM_000372.5:c.265T>C, NM_000372.5:c.272G>A, NM_000372.5:c.285_286insA, NM_000372.5:c.325G>A, NM_000372.5:c.533G>A, NM_000372.5:c.568delG, NM_000372.5:c.616G>A, NM_000372.5:c.646T>A, NM_000372.5:c.650G>A, NM_000372.5:c.823G>T, NM_000372.5:c.896G>A, NM_000372.5:c.1012_1013insC, NM_000372.5:c.1111A>G, NM_000372.5:c.1118C>A, NM_000372.5:c.1146C>A, NM_000372.5:c.1147G>A, NM_000372.5:c.1163delT, NM_000372.5:c.1177delG, NM_000372.5:c.1209G>T, NM_000372.5:c.1217C>T, NM_000372.5:c.1255G>A, NM_000372.5:c.1265G>A, NM_000372.5:c.1336G>A, NM_000372.5:c.1342G>A, NM_000372.5:c.1466_1467insT, NM_000372.5:c.1500_1501insC
TYRP1	Oculocutaneous albinism, Type 3	NM_000550.3:c.105delT, NM_000550.3:c.176C>G, NM_000550.3:c.497C>G, NM_000550.3:c.1054_1057delACAA, NM_000550.3:c.1067G>A, NM_000550.3:c.1101delA, NM_000550.3:c.1120C>T, NM_000550.3:c.1369_1370insCAGA, NM_000550.3:c.1557T>G
UGT1A1	Crigler-Najjar Syndrome	NM_000463.3:c.44T>G, NM_000463.3:c.524T>A, NM_000463.3:c.674T>G
UGT1A8	Crigler-Najjar Syndrome	NM_019076.5:c.1012C>T, NM_019076.5:c.1061A>G, NM_019076.5:c.1121G>T, NM_019076.5:c.1198C>T, NM_019076.5:c.1202T>C, NM_019076.5:c.1447T>G
USH1C	Usher syndrome, type 1C	NM_153676.4:c.2688_2695dupAATTCACC, NM_153676.4:c.2622_2623delCA, NM_153676.4:c.2547-1G>T, NM_153676.4:c.2362G>A, NM_153676.4:c.388G>A, NM_153676.4:c.238delC, NM_153676.4:c.238dupC, NM_153676.4:c.216G>A
USH2A	Usher syndrome, type 2A	NM_206933.4:c.15520-1G>A, NM_206933.4:c.15371delT, NM_206933.4:c.15089C>A, NM_206933.4:c.14926G>A, NM_206933.4:c.14803C>T, NM_206933.4:c.14519T>C, NM_206933.4:c.14442C>A, NM_206933.4:c.13709delG, NM_206933.4:c.12574C>T, NM_206933.4:c.12234_12235delGA, NM_206933.4:c.11864G>A, NM_206933.4:c.11549-5_11549-4insT, NM_206933.4:c.10636G>A, NM_206933.4:c.10561T>C, NM_206933.4:c.10073G>A, NM_206933.4:c.9799T>C, NM_206933.4:c.8981G>A, NM_206933.4:c.8431C>A, NM_206933.4:c.7364G>A, NM_206933.4:c.6862G>T, NM_206933.4:c.6670G>T, NM_206933.4:c.5975A>G, NM_206933.4:c.5743_5744delAG, NM_206933.4:c.5573-2A>G, NM_206933.4:c.4338_4339delCT, NM_206933.4:c.3491_3492delCT, NM_206933.4:c.2898delG, NM_206933.4:c.2299delG, NM_206933.4:c.2296T>C, NM_206933.4:c.2276G>T, NM_206933.4:c.2167+5G>A, NM_206933.4:c.2135delC, NM_206933.4:c.956G>A, NM_206933.4:c.920_923dupGCCA, NM_206933.4:c.820C>T, NM_206933.4:c.779T>G
VPS13A	Choreoacanthocytosis	NM_033305.3:c.622C>T, NM_033305.3:c.2898T>G, NM_033305.3:c.3091delG, NM_033305.3:c.9109C>T, NM_033305.3:c.9275+1G>T
WAS	Wiskott-Aldrich syndrome	NM_000377.3:c.134C>T, NM_000377.3:c.173C>G, NM_000377.3:c.809T>C, NM_000377.3:c.814T>C, NM_000377.3:c.881T>C, NM_000377.3:c.1442T>A
WNT10A	Odontoonychodermal dysplasia/Schopf-Schulz-Passarge syndrome/Tooth agenesis, selective, 4	NM_025216.3:c.321C>A, NM_025216.3:c.347T>C, NM_025216.3:c.383G>A, NM_025216.3:c.697G>T

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PATIENT 3.1

GENE	DISEASE NAME	VAR NAME
XPA	Xeroderma pigmentosum Group A	NM_000380.4:c.731A>G, NM_000380.4:c.727C>T, NM_000380.4:c.619C>T, NM_000380.4:c.501delG, NM_000380.4:c.348T>A, NM_000380.4:c.323G>T
ZFYVE26	Spastic Paraplegia Type 15	NM_015346.4:c.5485-1G>A, NM_015346.4:c.5422C>T, NM_015346.4:c.4936C>T, NM_015346.4:c.4312C>T, NM_015346.4:c.3642_3643insCCACACTTAG, NM_015346.4:c.3206G>A, NM_015346.4:c.3182delT, NM_015346.4:c.2887G>C, NM_015346.4:c.2114dupC, NM_015346.4:c.1477C>T