GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
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**
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
BTK	Isolated growth hormone deficiency, Type III, X-linked	NM_000061.3:c.1906G>T, NM_000061.3:c.1889T>A, NM_000061.3:c.1838G>A, NM_000061.3:c.1820C>A, NM_000061.3:c.1773C>A, NM_000061.3:c.1766A>G, NM_000061.3:c.1559G>A, NM_000061.3:c.1558C>T, NM_000061.3:c.1516T>C, NM_000061.3:c.1506C>A, NM_000061.3:c.1288A>G, NM_000061.3:c.1275C>A, NM_000061.3:c.1223T>C, NM_000061.3:c.1125T>G, NM_000061.3:c.1082A>G, NM_000061.3:c.1001A>C, NM_000061.3:c.919A>G, NM_000061.3:c.862C>T, NM_000061.3:c.763C>T, NM_000061.3:c.755G>A, NM_000061.3:c.718G>T, NM_000061.3:c.338T>A
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, NM_000492.4:c.1089_1090delCT, NM_000492.4:c.1090T>C, NM_000492.4:c.1115delA, NM_000492.4:c.1116G>T,

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List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
		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_1344delGAAAGATATTAATTTTCAAGATA, 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, NM_000492.4:c.1407_1416delGGTGATTATG, NM_000492.4:c.1414_1415insT, NM_000492.4:c.1416delG, NM_000492.4:c.1433_1434delCA, NM_000492.4:c.1435G>T, NM_000492.4:c.1437delG, NM_000492.4:c.1438G>T, NM_000492.4:c.1439G>A, NM_000492.4:c.1444_1445insT, NM_000492.4:c.1456G>T, NM_000492.4:c.1466C>A, NM_000492.4:c.1468delT, NM_000492.4:c.1469_1470delTC, NM_000492.4:c.1475C>T, NM_000492.4:c.1477_1478delCA, NM_000492.4:c.1477C>T, NM_000492.4:c.1478A>T, NM_000492.4:c.1480_1481delTT, NM_000492.4:c.1487G>A, NM_000492.4:c.1505T>C, NM_000492.4:c.1505T>G, NM_000492.4:c.1510G>T, NM_000492.4:c.1516_1518delATC, NM_000492.4:c.1516A>G, NM_000492.4:c.1517T>C, NM_000492.4:c.1518C>G, NM_000492.4:c.1520_1522delTCT, NM_000492.4:c.1519A>G, NM_000492.4:c.1519A>T, NM_000492.4:c.1523T>G, NM_000492.4:c.1525delG, NM_000492.4:c.1525G>C, NM_000492.4:c.1528delG, NM_000492.4:c.1529_1530delTT, NM_000492.4:c.1538A>G, NM_000492.4:c.1542_1543delAT, NM_000492.4:c.1546_1547delAG, NM_000492.4:c.1550A>G, NM_000492.4:c.1558G>A, NM_000492.4:c.1558G>T, NM_000492.4:c.1559T>A, NM_000492.4:c.1572C>A, NM_000492.4:c.1573C>T, 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NM_000492.4:c.2341C>T, NM_000492.4:c.2353C>T, NM_000492.4:c.2374C>T, NM_000492.4:c.2380delG, NM_000492.4:c.2389_2390insC, NM_000492.4:c.2395C>T, NM_000492.4:c.2416_2417insAT, NM_000492.4:c.2421A>G, NM_000492.4:c.2429delG, NM_000492.4:c.2433_2434insT, NM_000492.4:c.2440C>T, NM_000492.4:c.2443G>T, NM_000492.4:c.2452delT, NM_000492.4:c.2462_2463delGT, NM_000492.4:c.2464G>T, NM_000492.4:c.2467G>T, NM_000492.4:c.2471delT, NM_000492.4:c.2472_2473insAACG, NM_000492.4:c.2476G>T, NM_000492.4:c.2479G>T, NM_000492.4:c.2482_2483insA, NM_000492.4:c.2488A>T, NM_000492.4:c.2490+1G>A, NM_000492.4:c.2490+1G>T, NM_000492.4:c.2490+2T>C, NM_000492.4:c.2491-2A>C, NM_000492.4:c.2491-2A>G, NM_000492.4:c.2491G>T, NM_000492.4:c.2497delT, NM_000492.4:c.2496_2497insT, NM_000492.4:c.2506G>T, NM_000492.4:c.2508delT, NM_000492.4:c.2537G>A, NM_000492.4:c.2538G>A, NM_000492.4:c.2547C>A, NM_000492.4:c.2551C>T, NM_000492.4:c.2552G>T, NM_000492.4:c.2553_2554insT, NM_000492.4:c.2555A>T, NM_000492.4:c.2556T>G, 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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,

GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
		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, 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, 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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

GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
DKC1	Dyskeratosis congenita, X-Linked	NM_001363.5:c.91C>A, NM_001363.5:c.91C>G, NM_001363.5:c.194G>C, NM_001363.5:c.196A>G, NM_001363.5:c.200C>T, NM_001363.5:c.204C>A, NM_001363.5:c.214_215delCTinsTA, NM_001363.5:c.838A>C
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, 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
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
EMD	Emery-Dreifuss Muscular Dystrophy 1, X-Linked	NM_000117.3:c.547C>A, NM_000117.3:c.630_634delCCGTG
F8	Hemophilia A	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, 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,

GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
		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, NM_000132.4:c.3651delA, NM_000132.4:c.3631A>T, NM_000132.4:c.3624delT, NM_000132.4:c.3607G>T, NM_000132.4:c.3565dupA, NM_000132.4:c.3548_3549delAA, NM_000132.4:c.3540delA, NM_000132.4:c.3505delG, NM_000132.4:c.3500dupA, NM_000132.4:c.3496A>T, NM_000132.4:c.3493G>T, NM_000132.4:c.3490delT, NM_000132.4:c.3421C>T, NM_000132.4:c.3417dupT, NM_000132.4:c.3416_3417delCT, NM_000132.4:c.3409_3410delCT, NM_000132.4:c.3402delG, NM_000132.4:c.3385delC, NM_000132.4:c.3371C>A, NM_000132.4:c.3344delT, NM_000132.4:c.3302_3303delAG, NM_000132.4:c.3300delA, NM_000132.4:c.3298A>T, NM_000132.4:c.3289C>T, NM_000132.4:c.3279G>A, NM_000132.4:c.3255_3258delTAAA, NM_000132.4:c.3251C>G, NM_000132.4:c.3224delC, NM_000132.4:c.3203_3204delGA, NM_000132.4:c.3196C>T, NM_000132.4:c.3168_3187delTGAGTTTAAAAAAGTGACAC, NM_000132.4:c.3152delT, NM_000132.4:c.3150_3151insTC, NM_000132.4:c.3053delA, NM_000132.4:c.3034G>C, NM_000132.4:c.3031A>T, NM_000132.4:c.2462_2463delGG, NM_000132.4:c.2412_2421delCTCCTCTAGT, 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, 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,

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
		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, 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
FMR1	Fragile X syndrome	Premutation allele (CGG)n
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
HBA1/2	Thalassemia, alpha	--MED ; --SEA ; --THAI ; - α3.7 ; - α4.2 ; - α20.5 ; --FIL

GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

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, 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
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

GENESeeker

List of variants analysed for the Geneseeker Essential panel version 3.0, however, only variants classified as pathogenic and likely pathogenic at the time of analysis are reported.

ESSENTIAL 3.0

GENE	DISEASE NAME	VAR NAME
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
MECP2	RETT Syndrome	NM_001110792.2:c.1318G>A, NM_001110792.2:c.1084_1086delAGC, NM_001110792.2:c.1001C>T, NM_001110792.2:c.1000C>T, NM_001110792.2:c.952C>T, NM_001110792.2:c.916C>T, NM_001110792.2:c.844C>T, NM_001110792.2:c.842delG, NM_001110792.2:c.799C>T, NM_001110792.2:c.789delC, NM_001110792.2:c.766C>T, NM_001110792.2:c.741G>A, NM_001110792.2:c.719C>G, NM_001110792.2:c.710C>T, NM_001110792.2:c.647C>G, NM_001110792.2:c.538C>T, NM_001110792.2:c.251dupC
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
NR0B1	Congenital Adrenal Hypoplasia, X-linked	NM_000475.5:c.1319A>T, NM_000475.5:c.1316T>G, NM_000475.5:c.1107G>A, NM_000475.5:c.890T>C, NM_000475.5:c.873G>C, NM_000475.5:c.847C>T, NM_000475.5:c.813C>G, NM_000475.5:c.800G>C, NM_000475.5:c.788T>A, NM_000475.5:c.704G>A, NM_000475.5:c.591C>A, NM_000475.5:c.513G>A, NM_000475.5:c.388_389delTA, NM_000475.5:c.315G>C, NM_000475.5:c.273C>A
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
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
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
SLC6A8	Cerebral creatine deficiency syndrome 1	NM_005629.4:c.316_318delTTC, NM_005629.4:c.395G>T, NM_005629.4:c.1011C>G, NM_005629.4:c.1141G>C, NM_005629.4:c.1216_1218delTTC, NM_005629.4:c.1540C>T
SMN1	Spinal Muscular Atrophy	Exon 7del
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