

Monogenic Diseases Variant List

GENE	REFSEQ	VARIANT
ACTB	NM_001101.5	c.1013C>T, c.905G>C, c.802G>T, c.802G>C, c.773C>T, c.629G>A, c.628C>T, c.625G>T, c.625G>A, c.617G>A, c.616C>T, c.611C>G, c.616C>G, c.598T>G, c.587G>T, c.587G>A, c.586C>T, c.547C>G, c.491C>A, c.359C>T, c.351G>T, c.350A>T, c.349G>A, c.224T>C, c.220G>T, c.220G>A, c.217C>T, c.217C>G, c.209C>T, c.193C>T, c.193C>G, c.158A>G, c.1090G>A, c.1066T>C, c.1043C>T, c.548G>A, c.490C>T, c.82C>G, c.890_891del, c.329del, c.559_562del, c.475_476insGG, c.143del, c.547C>T
ACTG1	NM_001614.5	c.1109T>C, c.1013C>G, c.1004G>A, c.848T>C, c.833C>T, c.791C>T, c.766C>T, c.721G>A, c.637A>C, c.628C>T, c.617G>A, c.616C>T, c.608C>A, c.547C>T, c.464C>T, c.404C>T, c.359C>T, c.354G>T, c.353A>T, c.277G>A, c.266C>T, c.209C>T, c.151G>A, c.118C>T, c.94C>T, c.1003C>T, c.629G>A, c.542C>T
ACTG2	NM_001615.4	c.118C>T, c.119G>A, c.134T>C, c.187C>G, c.188G>T, c.188G>A, c.400T>A, c.442C>A, c.442C>T, c.443G>T, c.532C>T, c.533G>A, c.533G>T, c.588G>C, c.588G>T, c.593G>A, c.593G>T, c.632G>T, c.769C>T, c.770G>A, c.806_807delinsAA, c.968C>T, c.613G>A, c.632G>A
ACVR1	NM_001111067.4	c.1124G>C, c.982G>T, c.982G>A, c.774G>C, c.772A>G, c.605G>T, c.587T>C, c.1067G>A, c.983G>A, c.617G>A
ADNP	NM_001282531.3	c.124G>T, c.105_107insG, c.3069_3072del, c.1102C>T, c.2865_2868del, c.2808del, c.2630_2633del, c.2499del, c.2496_2499del, c.2491_2494del, c.2424_2427del, c.2287del, c.2268_2269insG, c.2239G>T, c.2230G>T, c.2213C>A, c.2194_2197del, c.2188C>T, c.2157del, c.2157C>G, c.2156_2157insA, c.2157C>A, c.2155del, c.2129_2130insC, c.1717del, c.1235del, c.1222_1223del, c.1211C>A, c.1106_1108delinsCTGT, c.1046_1047del, c.1033C>T, c.819del, c.673C>T, c.655_656del, c.539_542del, c.517C>T, c.484C>T, c.190_191insA, c.95_96insT, c.70del, c.56_57del, c.67_70del
AKT3	NM_005465.7	c.49G>A, c.1393C>T, c.686A>G, c.548T>A, c.239T>C, c.237G>C
ANKRD11	NM_013275.6	c.7535G>A, c.7825C>T, c.7570_1G>A, c.7569+1G>A, c.7552C>T, c.7519C>T, c.7471-2A>G, c.7470+2T>C, c.7470+1G>C, c.7407C>G, c.7356_7357insC, c.7327C>T, c.7192C>T, c.7189C>T, c.7180C>T, c.7183C>T, c.7083_7084insC, c.7062del, c.7048C>T, c.6836_6837del, c.6793del, c.6792_6793insC, c.6792del, c.6786_6787insA, c.6766C>T, c.6624_6625del, c.6623C>A, c.6569_6597insA, c.6513del, c.6472G>T, c.6469del, c.6430C>T, c.6340C>T, c.6249del, c.6210_6211del, c.6138del, c.6015_6016insA, c.5953_5954del, c.5810_5811insG, c.5777_5778insC, c.5667del, c.5659C>T, c.5651C>G, c.5564_5565del, c.5550C>G, c.5504del, c.5494_5495del, c.5437C>T, c.5317G>T, c.5233_5234del, c.5227C>T, c.5174_5175insC, c.5146G>T, c.5145C>G, c.5123C>A, c.5117del, c.4964_4965del, c.4933del, c.4875del, c.4786G>T, c.4765C>T, c.4750G>T, c.4489_4490del, c.4396_4397del, c.4389_4390del, c.4384_4385insA, c.4372_4375del, c.4334_4335ins, c.4288A>T, c.4261G>T, c.4218C>A, c.4206C>G, c.4205_4206insA, c.4189del, c.4107_4108del, c.4091_4092del, c.4087C>T, c.3948del, c.3931C>T, c.3907del, c.3888_3889insC, c.3885_3886insAGAC, c.3843_3844ins, c.3835_3836insA, c.3774_3775del, c.3770_3773del, c.3768_3769del, c.3704_3707del, c.3702_3705del, c.3632_3633del, c.3591_3594del, c.3562C>T, c.3547_3548del, c.3369_3372del, c.3367_3368del, c.3334_3335insA, c.3319_3322del, c.3309_3310insA, c.3309del, c.3224_3227del, c.3208_3209del, c.3198_3199del, c.3127del, c.3122C>A, c.3084C>A, c.3019C>T, c.2857del, c.2828_2829del, c.2751_2752insT, c.2750_2751del, c.2716C>T, c.2704G>T, c.2647del, c.2647G>T, c.2615_2616del, c.2589_2590insA, c.2523G>A, c.2518del, c.2454_2455insT, c.2409_2412del, c.2404_2407del, c.2398_2401del, c.2368G>T, c.2329_2332del, c.2329G>T, c.2305del, c.2297_2300del, c.2273_2274del, c.2273_2274insT, c.2197C>T, c.2177_2178del, c.2175_2178del, c.2090_2091del, c.2054_2055del, c.1977C>A, c.1977_1978del, c.1896_1897del, c.1893del, c.1801C>T, c.1731_1732insT, c.1679G>C, c.1652G>A, c.1583_1584insA, c.1459G>T, c.1444G>T, c.1388_1389del, c.1385_1388del, c.1381_1384del, c.1372C>T, c.1367_1370del, c.1318C>T, c.1285_1286del, c.1232C>A, c.1201_1204del, c.1018_1019insA, c.915del, c.893-1G>C, c.893-2A>G, c.867C>A, c.769G>T, c.744-1G>T, c.602-1G>A, c.601_601+1insAG, c.505G>T, c.397-1G>A, c.226+1G>A, c.7711C>T, c.7636_7637insG, c.7314_7315insCTAC, c.7303del, c.7290C>G, c.7244_7247del, c.7234C>T, c.7222_7226delinsGGG, c.7225G>T, c.7183C>T, c.7168C>A, c.7144_7145ins, c.7144C>T, c.7114_7115ins, c.7103_7106del, c.7062_7063ins, c.6982C>T, c.6923del, c.6888_6883del, c.6871G>C, c.6847C>T, c.6833_6834del, c.6807_6808del, c.6772_6773ins, c.6760G>T, c.6742C>T, c.6688_6689del, c.6677_6680delinsAGA, c.6670G>T, c.6589_6590insA, c.6580>T, c.6465del, c.6402del, c.6320_6321insG, c.6288_6289insT, c.6218del, c.6212C>G, c.6197_6198delinsA, c.6165del, c.6152_6153ins, c.6101del, c.6045C>A, c.5865del, c.5859del, c.5833G>T, c.5790>C>G, c.5682_5683ins, c.5637_5638insA, c.5560_5563del, c.5537del, c.5469_5470insT, c.5469del, c.5447del, c.5438_5439insT, c.5375_5376del, c.5364C>G, c.5117_5118ins, c.5105_5106ins, c.5089C>T, c.5056del, c.5030_5031del, c.4948_4949ins, c.4902del, c.4742_4743insA, c.4643_4645insC, c.4643del, c.4624_4625del, c.4621_4624del, c.4619_4620del, c.4585A>T, c.4554del, c.4381_4384del, c.4381_4382del, c.4370_4373del, c.4370_4371del, c.4369A>T, c.4345_4346del, c.4333_4336del, c.4306_4307del, c.4306G>T, c.4270del, c.4176G>C, c.4145_4146ins, c.4140_4141del, c.4140C>G, c.4117A>T, c.4114G>T, c.4067del, c.4059del, c.4055_4058del, c.4052_4053insT, c.3936_3937insG, c.3829del, c.3812C>A, c.3787_3788del, c.3779C>A, c.3761del, c.3708_3709del, c.3706C>T, c.3651C>A, c.3603_3604del, c.3603_3604ins, c.3597_3598del, c.3562_3563insA, c.3535_3536insGACA, c.3509_3512del, c.3306_3309del, c.3268del, c.3244C>T, c.3240del, c.3239_3240del, c.3201_3202insA, c.3187A>T, c.3151_3152del, c.3151G>T, c.31387C>A, c.3122C>G, c.3096del, c.3084del, c.3084C>G, c.3066del, c.3025_3026insGA, c.2944G>T, c.2904del, c.2853_2856del, c.2826_2829del, c.2810del, c.2793_2794insT, c.2777del, c.2618_2619del, c.2612_2613insGCGA, c.2607_2608insG, c.2553_2556del, c.2536del, c.2516_2517del, c.2500del, c.2494del, c.2464C>T, c.2434del, c.2413G>T, c.2350G>T, c.2327T>G, c.2299A>T, c.2288_2289del, c.2283C>G, c.2262_2265del, c.2166_2167insA, c.2130G>A, c.2089G>T, c.2088_2089insA, c.2073_2074insC, c.2062_2064insA, c.2047_2048del, c.2022_2023insA, c.1996C>T, c.1977_1978insTTAC, c.1977del, c.1976_1977insA, c.1976_1977insTTAC, c.1976C>T, c.1901_1904del, c.1861A>T, c.1800_1801insG, c.1742_1743del, c.1726G>T, c.1708_1711del, c.1686del, c.1677G>A, c.1621C>T, c.1616_1617del, c.1558del, c.1536_1537insC, c.1523del, c.1477_1478del, c.1462_1463del, c.1422_1423insC, c.1401del, c.1389del, c.1381_1382del, c.1355_1356insA, c.1355del, c.1280_1281del, c.1146_1147insT, c.1119del, c.1063C>T, c.1009C>T, c.998del, c.977del, c.963_964ins, c.641_642ins, c.618del, c.568G>T, c.439C>T, c.424C>T, c.291del, c.249del, c.160C>T, c.99_100insA
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c.2268C>T, c.2269C>T, c.2270C>T, c.2271C>T, c.2272C>T, c.2273C>T, c.2274C>T, c.2275C>T, c.2276C>T, c.2277C>T, c.2278C>T, c.2279C>T, c.2280C>T, c.2281C>T, c.2282C>T, c.2283C>T, c.2284C>T, c.2285C>T, c.2286C>T, c.2287C>T, c.2288C>T, c.2289C>T, c.2290C>T, c.2291C>T, c.2292C>T, c.2293C>T, c.2294C>T, c.2295C>T, c.2296C>T, c.2297C>T, c.2298C>T, c.2299C>T, c.2290del, c.2291del, c.2292del, c.2293del, c.2294del, c.2295del, c.2296del, c.2297del, c.2298del, c.2299del, c.2290C>T, c.2291C>T, c.2292C>T, c.2293C>T, c.2294C>T, c.2295C>T, c.2296C>T, c.2297C>T, c.2298C>T, c.2299C>T, c.2290del, c.2291del, c.2292del, c.2293del, c.2294del, c.2295del, c.2296del, c.2297del, c.2298del, c.2299del, c.2290C>T, c.2291C>T, c.2292C>

ATP1A3	NM_152296.5	c.2840G>A, c.2839G>C, c.2768A>C, c.2767G>T, c.2767G>C, c.2759A>G, c.2570_2572del, c.2552A>C, c.2528C>A, c.2452G>A, c.2417T>G, c.2415C>G, c.2408G>A, c.2407G>A, c.2401G>C, c.2338T>C, c.2318A>G, c.2312C>T, c.2273T>C, c.2270T>C, c.2264G>C, c.2263G>A, c.2222_2229del, c.2225A>T, c.2224G>T, c.2149_2151del, c.2116G>C, c.2051C>T, c.2051C>A, c.1790G>A, c.1109C>T, c.1165G>A, c.1108A>G, c.1088T>C, c.10179C>G, c.1076C>A, c.1073G>A, c.1072G>T, c.1072G>C, c.1036T>C, c.998G>A, c.971A>G, c.958G>A, c.954C>G, c.947G>T, c.946A>G, c.875T>G, c.820A>T, c.460A>G, c.385G>A, c.265G>T, c.265G>A, c.2839G>T, c.2771T>C, c.2677G>A, c.974G>A, c.967C>A, c.410C>T, c.2438C>T, c.2324C>T, c.2144T>C, c.2116G>A, c.1825G>T, c.968C>A, c.410_412del, c.3042A>C, c.2689-1G>T, c.2542+1G>C, c.2512_2516delinsTC, c.2095-1G>A, c.2094+2_2094+3del, c.1807>T>A, c.1630+2>C, c.1630+1G>C, c.1302+1G>A, c.2742+1G>T, c.358-1G>A, c.2763G>A, c.2652_2653insC, c.2631_2632insG, c.1959C>A, c.1930C>T, c.1429A>T, c.1299del, c.1115del, c.562C>T, c.235del, c.2851G>A, c.2767G>A, c.2755_2757del, c.2677G>C, c.2600G>A, c.2479A>T, c.2431T>C, c.2407G>C, c.821T>A, c.410C>A, c.2552A>G, c.2443G>A, c.2401G>A, c.2315G>A, c.2266C>T, c.2263G>T, c.977T>G, c.973G>C, c.2542+1G>C, c.266G>C, c.2267G>A
BCL11A	NM_0022893.4	c.143G>T, c.1831G>T, c.1633del, c.1486G>T, c.1345del, c.1118_1119insC, c.1078_1079insC, c.952A>T, c.794del, c.529C>T, c.295del, c.263C>A, c.193G>T, c.154C>T, c.148C>T, c.370C>T, c.314_315insT, c.310del, c.184del, c.147del
BICD2	NM_001003800.2	c.2321A>G, c.2081G>A, c.1604C>T, c.2320G>A, c.2108C>T, c.2080C>T, c.1636_1638del, c.320C>T
BRAF	NM_004333.6	c.1741A>G, c.1797G>T, c.1796G>C, c.1787G>T, c.1785T>G, c.1781A>T, c.1455G>C, c.1454T>C, c.1406G>A, c.1391G>A, c.793G>C, c.770A>G, c.7471T>G, c.740T>C, c.739T>C, c.739T>G, c.736G>C, c.735A>C, c.735A>T, c.730A>C, c.722C>A, c.1455G>T, c.1914T>G, c.1914T>A, c.1803A>T, c.1802A>G, c.1802A>C, c.1801A>C, c.1801A>T, c.1799_1800delinsC, c.1799T>C, c.1798_1799delinsAG, c.1798G>C, c.1798G>A, c.1796_1797insT, c.1790T>G, c.1790T>A, c.1789_1790delinsTC, c.1789G>C, c.1787G>A, c.1786G>C, c.1786G>T, c.1785T>A, c.1784T>C, c.1783T>C, c.1782T>G, c.1782T>A, c.1781A>C, c.1780G>C, c.1780G>A, c.1779_1780delinsGA, c.1600G>C, c.1593G>T, c.1593G>C, c.1591T>C, c.1591T>A, c.1574T>G, c.1574T>A, c.1502A>T, c.1502A>C, c.1501G>C, c.1501G>A, c.1497A>T, c.1497A>C, c.1495A>G, c.1460T>G, c.1454T>A, c.1411G>T, c.1408A>C, c.1406G>T, c.1406G>C, c.1405_1406delinsT, c.1405_1406delinsTC, c.1405G>C, c.1405G>A, c.1403T>C, c.1402T>C, c.1399T>G, c.1396G>C, c.1396G>A, c.1391G>C, c.1390G>A, c.823G>A, c.785A>G, c.785A>C, c.741T>A, c.722C>T, c.722G>C, c.721A>C, c.1786G>T, c.1781A>G, c.1741A>C, c.1741A>G, c.755G>C, c.784C>A, c.769C>A
CACNA1A	NM_001127222.2	c.5529-12_5529-121del, c.5529-121del, c.5529-121_5529-124del, c.5529-1268C>A, c.5425A>C, c.5426T>C, c.5419G>T, c.5417T>C, c.5326G>A, c.5131C>A, c.5122A>C, c.5120T>C, c.5072T>A, c.5032C>A, c.5015G>C, c.4979G>A, c.4927G>T, c.4549G>A, c.4520C>A, c.4519G>T, c.4514T>C, c.4500_4502del, c.4403C>T, c.4363G>T, c.4363G>C, c.4205T>G, c.4175T>C, c.4174G>C, c.4148A>G, c.4096T>A, c.4067_4069del, c.4067T>G, c.4052G>A, c.4043G>C, c.4042C>T, c.4034G>T, c.3787G>A, c.2142C>A, c.2137G>A, c.2098G>A, c.1839G>A, c.1088T>C, c.1036_1038del, c.877G>A, c.860G>A, c.814T>C, c.757C>T, c.688G>C, c.653C>G, c.652T>C, c.409G>A, c.301G>T, c.184T>G, c.184T>A, c.5422G>A, c.5393C>A, c.5260G>A, c.5123T>C, c.4997G>C, c.4996G>T, c.4174G>A, c.4072C>T, c.4052G>T, c.4043G>A, c.4034T>C, c.2134G>A, c.1999G>A, c.1994C>T, c.1745G>A, c.1594G>A, c.1060C>T, c.904G>A, c.815G>A, c.653C>T, c.439G>A, c.5419G>A, c.5248C>T, c.5014C>T, c.4947G>A, c.4897G>C, c.4897G>T, c.4102C>T, c.4055C>T, c.4028C>A, c.4027T>C, c.3946G>A, c.3692+1G>A, c.1883C>T, c.1067T>C, c.905A>G, c.889G>A, c.835C>T, c.688G>A, c.631+5G>A, c.6684T>G, c.6489_6490insT, c.6340_6410del, c.6189+1G>C, c.6189+1G>A, c.6097T>C, c.6051-1G>A, c.6050-4_6050+2del, c.5941-2A>G, c.5940-1G>A, c.5840-2A>G, c.5840-2A>G, c.5837del, c.5452C>T, c.5400+2T>C, c.5335C>T, c.5250-2A>G, c.5134-2del, c.5134-2A>C, c.5068-1G>A, c.5056C>T, c.4987T>C, c.4905+1G>T, c.4866E-1G>T, c.4842del, c.4829_4829del, c.4633C>T, c.4591-2A>G, c.4389+1G>C, c.3829C>G, c.3829C>T, c.3822+1G>A, c.2764C>T, c.2408G>A, c.3692+1G>T, c.3692_3692+1insC, c.3563del, c.3532del, c.3089+1G>A, c.3039C>G, c.2865del, c.2813_2814insG, c.2812_2813del, c.2764C>T, c.2311A>T, c.2191G>T, c.2105-2A>G, c.1914-1G>A, c.1913+1G>A, c.1912C>T, c.1781del, c.1744C>T, c.1647del, c.1634-15insA, c.1563C>A, c.1555+2del, c.1479_1480del, c.1469G>A, c.1439del, c.1435del, c.1412_1413insA, c.1335_1336insT, c.1248_1249insC, c.1198+1G>A, c.1083-2A>G, c.1082+1G>A, c.978+1G>C, c.978+1G>A, c.841del, c.631+2T>C, c.540-1G>C, c.400-1G>A, c.399+1G>A, c.399+1G>A, c.6435_6436insC, c.6397C>A, c.6371C>A, c.6368del, c.6202C>T, c.6181_6184del, c.6155_6156insC, c.6151_6152insT, c.6114del, c.6043G>T, c.5884del, c.5783_5784insTGGC, c.5477_5478insA, c.5476del, c.5439_5440insT, c.5408_5409insA, c.5342del, c.5151T>G, c.5033_5043insG, c.5044_5045del, c.5015_5016insG, c.4941_4942insT, c.4795del, c.4783_4786delinsC, c.4765del, c.4694_4695insT, c.4663C>T, c.4599C>A, c.4539_4540del, c.4434del, c.4426C>T, c.4383G>A, c.4346del, c.4242C>T, c.4242del, c.4242C>A, c.4203del, c.4085del, c.3925_3926insC, c.3838_3841del, c.3652_3653del, c.3631G>T, c.3590_3593del, c.3578del, c.3532_3533insC, c.3478_3479del, c.3125del, c.3169del, c.3154C>T, c.3138_3139insC, c.3134C>G, c.3122del, c.3105_3106insC, c.3103_3104insT, c.3046del, c.3039C>A, c.3006_3007delinsC, c.2996del, c.2974G>T, c.2959_2960insTC, c.2959+2600insC, c.2903del, c.2899G>T, c.2896del, c.2866C>T, c.2840del, c.2806C>T, c.2785_2786del, c.2755G>T, c.2734C>T, c.2599_2600insC, c.2551C>T, c.2539C>T, c.2524G>T, c.2482C>T, c.2401G>T, c.2357del, c.2350C>T, c.2343_2344insAAG, c.2321del, c.2273_2274insCT, c.2067_2068delinsGGAG, c.2045_2046insG, c.2022C>A, c.2006G>A, c.1984C>T, c.1946del, c.1922del, c.1785C>G, c.1701G>A, c.1586_1587del, c.1577T>C, c.1560T>G, c.1538G>A, c.1434C>G, c.1360C>T, c.1321C>T, c.1282del, c.1144C>T, c.1084G>T, c.1004G>A, c.992C>G, c.876_877insA, c.826G>T, c.816T>A, c.682C>T, c.592C>T, c.526del, c.507G>A, c.471_472del, c.362_363insT, c.304del, c.41del
CAMTA1	NM_015215.4	c.403G>A, c.404A>G, c.180_183del, c.4381+1G>A, c.511-1G>A, c.613G>T, c.627del, c.838del, c.882_883insCG, c.1132up, c.1744G>T, c.1828C>T, c.2446del, c.2915-2A>G, c.1483-1G>A, c.4780C>T, c.4921C>T, c.2429_252del, c.3137C>G, c.2402del, c.501G>A, c.766C>T, c.800del, c.926C>A, c.1156del, c.1455dup, c.1798dup, c.1914_1915del, c.2235C>A, c.2416_2419del, c.2452G>T, c.2460C>A, c.2639G>A, c.2685del, c.2755G>A, c.2740C>T, c.2735C>A, c.2734C>T, c.2733C>A, c.2732C>T, c.2731C>A, c.2730C>T, c.2729C>A, c.2728C>T, c.2727C>A, c.2726C>T, c.2725C>A, c.2724C>T, c.2723C>A, c.2722C>T, c.2721C>A, c.2720C>T, c.2719C>A, c.2718C>T, c.2717C>A, c.2716C>T, c.2715C>A, c.2714C>T, c.2713C>A, c.2712C>T, c.2711C>A, c.2710C>T, c.2709C>A, c.2708C>T, c.2707C>A, c.2706C>T, c.2705C>A, c.2704C>T, c.2703C>A, c.2702C>T, c.2701C>A, c.2700C>T, c.2699C>A, c.2698C>T, c.2697C>A, c.2696C>T, c.2695C>A, c.2694C>T, c.2693C>A, c.2692C>T, c.2691C>A, c.2690C>T, c.2689C>A, c.2688C>T, c.2687C>A, c.2686C>T, c.2685C>A, c.2684C>T, c.2683C>A, c.2682C>T, c.2681C>A, c.2680C>T, c.2679C>A, c.2678C>T, c.2677C>A, c.2676C>T, c.2675C>A, c.2674C>T, c.2673C>A, c.2672C>T, c.2671C>A, c.2670C>T, c.2669C>A, c.2668C>T, c.2667C>A, c.2666C>T, c.2665C>A, c.2664C>T, c.2663C>A, c.2662C>T, c.2661C>A, c.2660C>T, c.2659C>A, c.2658C>T, c.2657C>A, c.2656C>T, c.2655C>A, c.2654C>T, c.2653C>A, c.2652C>T, c.2651C>A, c.2650C>T, c.2649C>A, c.2648C>T, c.2647C>A, c.2646C>T, c.2645C>A, c.2644C>T, c.2643C>A, c.2642C>T, c.2641C>A, c.2640C>T, c.2639C>A, c.2638C>T, c.2637C>A, c.2636C>T, c.2635C>A, c.2634C>T, c.2633C>A, c.2632C>T, c.2631C>A, c.2630C>T, c.2629C>A, c.2628C>T, c.2627C>A, c.2626C>T, c.2625C>A, c.2624C>T, c.2623C>A, c.2622C>T, c.2621C>A, c.2620C>T, c.2619C>A, c.2618C>T, c.2617C>A, c.2616C>T, c.2615C>A, c.2614C>T, c.2613C>A, c.2612C>T, c.2611C>A, c.2610C>T, c.2609C>A, c.2608C>T, c.2607C>A, c.2606C>T, c.2605C>A, c.2604C>T, c.2603C>A, c.2602C>T, c.2601C>A, c.2600C>T, c.2599C>A, c.2598C>T, c.2597C>A, c.2596C>T, c.2595C>A, c.2594C>T, 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DNM1	NM_004408.4	c.679dup, c.767del, c.1025del, c.1153C>T, c.1347C>A, c.1626del, c.2296C>T, c.2299dup, c.128G>A, c.431C>T, c.442C>A, c.529G>C, c.545C>A, c.618G>C, c.632A>T, c.854T>A, c.865A>T, c.1036G>A, c.1037G>A, c.1076G>A, c.1190G>A, c.1603A>G, c.127G>A, c.134G>A, c.415G>A, c.443A>G, c.709C>T, c.1075G>A, c.766C>T, c.850C>T, c.112G>A, c.416G>A, c.457G>A, c.590>2A>G, c.1195A>T, c.261C>T
DYNC1H1	NM_001376.5	c.926G>A, c.1792C>T, c.1793G>C, c.1793G>T, c.1808A>T, c.1834G>A, c.2327C>T, c.3347T>C, c.4699C>T, c.4700G>T, c.4867C>T, c.6271C>T, c.6272G>A, c.6880G>A, c.6989G>A, c.6995G>A, c.7793G>T, c.10420C>T, c.10574G>A, c.3278T>C, c.4700G>A, c.4868G>A, c.5884C>T, c.5885G>A, c.6994C>T, c.10031G>A, c.10151G>A, c.10573C>T, c.751C>T, c.752G>A, c.791G>A, c.874C>T, c.1750A>C, c.4552G>A, c.9142G>A
DYRK1A	NM_001347721.2	c.143dup, c.151C>T, c.187C>T, c.197dup, c.210dup, c.250>251del, c.262del, c.267>268dup, c.285C>A, c.286A>T, c.293>294dup, c.317del, c.331C>T, c.341del, c.343G>T, c.367G>T, c.449dup, c.471del, c.474del, c.497dup, c.507dup, c.531>532dup, c.550>552delinsTTCT, c.606dup, c.658>659del, c.701dup, c.763A>T, c.803del, c.814dup, c.840dup, c.840del, c.931>932del, c.931C>T, c.940C>T, c.980>981insCCCA, c.986dup, c.987T>A, c.1000>1001del, c.1003dup, c.1025del, c.1098del, c.1112del, c.1132C>T, c.1135dup, c.1173del, c.1191>1192insTT, c.1214>1217dup, c.1218C>G, c.1224dup, c.1239del, c.1237A>T, c.1259>1260dup, c.1271dup, c.1270G>T, c.1271>1272insT, c.1275dup, c.1336del, c.1343>1346dup, c.1357dup, c.1378C>T, c.484G>A, c.593T>A, c.821T>G, c.833A>G, c.887T>G, c.895T>G, c.896T>C, c.905C>A, c.945T>A, c.947G>A, c.1009T>C, c.1010C>T, c.1015G>A, c.476G>A, c.833A>T, c.856C>T, c.905C>T, c.1373G>A, c.143>144del, c.201>204del, c.208dup, c.220dup, c.263>264del, c.284dup, c.285C>G, c.322C>T, c.334C>T, c.425dup, c.434del, c.450del, c.450C>G, c.542>545del, c.545>548del, c.574C>T, c.586C>T, c.594>597delinsGAA, c.630C>A, c.664C>T, c.675>676del, c.678>680delinsAC, c.687del, c.736C>T, c.748>749del, c.755del, c.760C>T, c.772C>T, c.817dup, c.822dup, c.848dup, c.909T>A, c.910C>T, c.918dup, c.930C>G, c.930C>A, c.977del, c.1006del, c.1008G>A, c.1069G>A, c.1074>1077del, c.1158dup, c.1190>1193insTAA, c.1205dup, c.1213>1213insTAA, c.1221del, c.1243del, c.1282C>T, c.1289del, c.1306dup, c.1373dup, c.1372C>T, c.707T>G, c.301>2A>G, c.489+2T>C, c.489+2T>A, c.490>1G>A, c.637>2T>C, c.638>2A>G, c.638>1G>A, c.638-1G>T, c.924+4>924+7del, c.924+1G>A, c.924+1G>C, c.924+2T>A, c.925-1G>C, c.1071+1G>A, c.1072>2A>T, c.1213>2A>G, c.1351>1354del, c.1464del, c.1519+1G>A, c.1626C>A, c.1951del, c.205C>T, c.1526C>G
EBP	NM_006579.3	c.182G>A, c.225dup, c.261C>G, c.304A>T, c.312T>A, c.329>332dup, c.387G>A, c.204G>T, c.238G>A, c.329G>A, c.439C>T, c.440G>A, c.149C>A, c.187C>T, c.203G>A, c.328C>T, c.386G>A, c.390del, c.404G>A, c.302-1G>A, c.338+1G>C, c.338+1G>T, c.523C>T, c.587G>A
EFNB1	NM_004429.5	c.111G>A, c.142A>T, c.149T>A, c.155>159delinsCAAG, c.219T>G, c.237C>G, c.403dup, c.457>458dup, c.523C>T, c.161C>T, c.451G>A, c.332C>T, c.452G>T, c.472A>G, c.473T>G, c.110G>A, c.196del, c.196C>T, c.253C>T, c.324dup, c.496C>T, c.128+1G>C, c.128+2>T>A, c.129-1G>T, c.499+1G>A, c.614>615del, c.635>636del, c.640C>T
EFTUD2	NM_004247.4	c.2595>2596insC, c.2551del, c.2544del, c.2505>2506insAG, c.2352>2353insT, c.2166C>A, c.2026>2027insTTTG, c.1947>1948insA, c.1946C>G, c.1923>1924insGATG, c.1901>1902insAA, c.1888del, c.1742del, c.1669C>T, c.1641>1642insC, c.1567del, c.1554del, c.1542C>A, c.1513C>T, c.1465>1466del, c.1402>1403insT, c.1332del, c.1331del, c.1060C>T, c.882del, c.828C>T, c.793del, c.742C>T, c.593>594insA, c.579del, c.423del, c.378del, c.342T>G, c.334>335insA, c.291>292insT, c.158del, c.103G>T, c.1910T>G, c.857A>G, c.623A>G, c.422G>A, c.784C>T, c.2496G>C, c.2493C>T, c.2419del, c.2199G>A, c.2198G>A, c.1759>1760del, c.1732C>T, c.1705C>T, c.1297>1298del, c.1012G>T, c.764>765insT, c.2770C>T, c.2698>2701del, c.2562>2-2562del, c.2562-1G>A, c.2562>2del, c.2466+1G>A, c.2347+1G>T, c.2133>1G>T, c.2045+2T>G, c.1963-1G>T, c.1719+1G>A, c.1607+1G>T, c.1414>2A>G, c.1286-2del, c.1286-2A>G, c.1150-1G>T, c.1150-2A>G, c.1149+1G>C, c.1149+1G>A, c.1096C>T, c.1058+1G>A, c.870-1G>T, c.703>2A>G, c.702+2T>C, c.702+1G>T, c.702G>T, c.523C>T, c.426+1>26+2insG, c.351-2A>G, c.3416>1C, c.427del, c.45del
EHMT1	NM_024757.5	c.162del, c.294>297del, c.302C>G, c.355del, c.391del, c.417dup, c.508del, c.555del, c.682del, c.719up, c.733G>T, c.749>750delinsG, c.756del, c.756dup, c.756del, c.784C>T, c.1042>1043dup, c.1051del, c.1058C>G, c.1052>1052del, c.1122up, c.1180del, c.1262del, c.1292del, c.1311G>A, c.1349+1352del, c.1408C>T, c.1420G>T, c.1438up, c.1543C>T, c.1597del, c.1602dup, c.380T>G, c.673C>T, c.870up, c.871C>T, c.1434>1435del, c.1468C>T, c.1495del, c.1533>1536del, c.1588C>T, c.2018+1G>A, c.2712+1G>C, c.546dup, c.642+1G>C, c.642+2+1G>T, c.643-2A>G, c.736C>T, c.824-2del, c.1249>2A>G, c.1501+1del, c.1502>2A>G, c.1647+2T>C, c.1858C>T, c.2018+1G>C, c.2079dup, c.2193-1G>C, c.2382+1G>A, c.2505+1G>A, c.2506-2A>G, c.2566-2C>T, c.2608-1G>A, c.2608-1G>G, c.2626G>A, c.2625del, c.2704C>T, c.2712+1G>A, c.2863>2864del, c.2877>2880del, c.2950C>T, c.3000del, c.3072>3073del, c.3126>3127del, c.3127C>T, c.3258+1del, c.3259+1G>A, c.3413G>A, c.3462-1G>A, c.3502C>T, c.3520del, c.404up, c.508C>T, c.1800up, c.2161del, c.2188up, c.2198del, c.2222del, c.2224del, c.2408C>T, c.2431>2435delinsTG, c.2498del, c.2610>2613up, c.2649C>A, c.2799del, c.2825C>T, c.2959G>A, c.2976del, c.3046C>T, c.3252C>G, c.3271up, c.3310G>T, c.3321>3324del, c.3342delinsAAG, c.3346C>T, c.3393C>A, c.3443del, c.3455-3458del, c.3482C>G, c.3488C>G, c.3649C>T
EP300	NM_001429.4	c.108>111del, c.272dup, c.272del, c.319C>T, c.478C>T, c.584C>A, c.832del, c.1066C>T, c.1164C>A, c.1423dup, c.1508up, c.1517del, c.1709>1710del, c.1738C>T, c.2164up, c.2225del, c.2305C>T, c.2333up, c.2540dup, c.2734C>T, c.2774up, c.2855C>G, c.2893C>T, c.2983G>T, c.3014>3015del, c.3070>3071insT, c.3139>3140del, c.3412dup, c.3451C>T, c.3472C>T, c.3485dup, c.3573T>A, c.3672>3673up, c.3684del, c.3685G>T, c.3707up, c.3733del, c.3826>3827up, c.3858>3859up, c.3889A>T, c.3905dup, c.4065C>A, c.4065C>T, c.4074>4074del, c.4103del, c.4190>4191up, c.4213del, c.4216>4219up, c.4307G>A, c.4444>4445up, c.4484T>A, c.4575up, c.4595del, c.4692up, c.4785del, c.4836up, c.4843del, c.4873C>T, c.4915T>A, c.4973T>G, c.5015T>A, c.5030del, c.3000del, c.3072>3073del, c.3126>3127del, c.3127C>T, c.3258+1del, c.3259+1G>A, c.3413G>A, c.3462-1G>A, c.3502C>T, c.3520del, c.7244A>T, c.7272C>T, c.7272del, c.7280up, c.7298del, c.7305C>T, c.7306del, c.7307C>T, c.7308del, c.7309C>T, c.7310G>T, c.7310C>T, 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KANSL1	NM_015443.4	c.2692_2693del, c.2591del, c.2519C>A, c.2132_2133insT, c.2130del, c.2109_2110insA, c.2105del, c.2058del, c.1958_1959insC, c.1944_1945inst, c.1802_1803insA, c.1726C>T, c.1725_1726insA, c.1579_1582del, c.1532del, c.1485_1488del, c.1450del, c.1412_1413del, c.1410C>A, c.1384C>T, c.1353G>A, c.1347G>A, c.1328_1329insC, c.1327_1328insATTAC, c.3031C>T, c.2788_2789del, c.2470C>T, c.1867_1870del, c.1816C>T, c.1651_1652insA, c.1463_1464insC, c.2542_1G>A, c.1186C>T, c.2837_2T>C, c.2837_1G>A, c.2667_1G>A, c.2542_2A>G, c.2020+1G>A, c.1652+1G>A, c.1268_1269del, c.1042C>T, c.930del, c.916C>T, c.878del, c.876del, c.699_700insT, c.611_612insG, c.611_612insGGGG, c.572del, c.540del, c.537_538insGA, c.449del, c.2956_2957del, c.2939_2940del, c.2908_2909insC, c.2902C>T, c.2835_2836insG, c.2824del, c.2284C>T, c.1288_1289del, c.1269del, c.1220C>G, c.1185_1186insA, c.1101del, c.1006G>T, c.993_994insA, c.808del, c.788del, c.779del, c.774_775inst, c.611del, c.295C>T
KAT6B	NM_012330.4	c.2616del, c.2658_2659del, c.2742_2743del, c.2848_2849del, c.3041_3042del, c.3118C>T, c.3169dup, c.3213del, c.3217G>T, c.3223G>T, c.3246del, c.3253del, c.3286G>T, c.3295_3298del, c.3373_3381del, c.3428del, c.3477_3480dup, c.3499_3500del, c.3147G>A, c.708_709del, c.3004C>T, c.3018del, c.3040C>T, c.3216del, c.3172C>T, c.3256G>T, c.3264dup, c.3349_3350del, c.3349C>T, c.3366_3369del, c.3401del, c.3399_3402del, c.847_2A>G, c.1030C>T, c.1061_11G>A, c.1993_11G>A, c.2347C>T, c.2629_2T>C, c.3021_1G>C, c.3022_2A>C, c.3581del, c.3606_3609del, c.3664_1G>A, c.3665_1G>C, c.3892G>T, c.3906del, c.3962_3963del, c.3995del, c.3998dup, c.4069G>T, c.4089_4092del, c.4205_4206del, c.4231G>T, c.4379del, c.4405dup, c.4461_4464del, c.4538dup, c.4572_4573dup, c.4592del, c.4602del, c.5058C>A, c.5146_5147del, c.5254C>T, c.5302C>T, c.5389C>T, c.5646del, c.130A>T, c.257del, c.282del, c.457C>T, c.466del, c.1009C>T, c.1770_1773del, c.1864C>T, c.2569_272del, c.3788_3789del, c.3802G>T, c.4096G>T, c.4102dup, c.4162C>T
KCNB1	NM_004975.4	c.307C>T, c.117_118insG, c.117del, c.1265A>C, c.1240A>G, c.1226T>C, c.1222C>C, c.1190G>T, c.1184G>A, c.1153C>A, c.1145A>T, c.1144G>C, c.1144G>A, c.1136G>T, c.1135G>A, c.1133T>G, c.1133T>C, c.1130C>A, c.1110T>C, c.1057G>A, c.1040G>T, c.990G>C, c.988G>A, c.956C>T, c.938A>G, c.935G>C, c.814C>T, c.629C>G, c.629G>A, c.623T>G, c.595A>T, c.587T>G, c.128A>G, c.1248C>A, c.1246T>C, c.1201G>A, c.1136G>A, c.1132G>C, c.1106G>T, c.1045G>T, c.1041C>G, c.1041C>A, c.1001T>C, c.995G>T, c.935G>A, c.934C>T, c.916C>T, c.908G>A, c.629C>T, c.1190G>A, c.1183G>A, c.1142G>A, c.1139A>G, c.1105T>C, c.937C>T, c.907C>T, c.1747C>T, c.1503_1504inst, c.1489G>T, c.1109G>A, c.1107G>A, c.1088del, c.984C>G, c.857del, c.820_821inst, c.661delinsAT
KCNJ2	NM_000891.3	c.199C>T, c.200G>A, c.211G>A, c.212A>T, c.224C>G, c.232G>T, c.233A>G, c.244C>T, c.245G>A, c.430G>A, c.431G>A, c.431G>C, c.434A>G, c.557C>A, c.557C>T, c.565A>G, c.566G>T, c.574A>G, c.644G>A, c.646A>T, c.646A>T, c.653G>A, c.653G>T, c.653G>T, c.779G>A, c.779G>C, c.896A>G, c.899G>A, c.902G>T, c.904G>A, c.913A>G, c.919A>G, c.926C>T, c.934C>T, c.935G>A, c.211G>C
KCNQ2	NM_172107.4	c.1742G>T, c.1734G>C, c.1734G>A, c.1733T>C, c.1732A>G, c.1732A>C, c.1721G>A, c.1720G>A, c.1700T>A, c.1688A>G, c.1687G>T, c.1684T>A, c.1681C>T, c.1679G>C, c.1666A>C, c.1665C>A, c.1663G>A, c.1662G>T, c.1658G>C, c.1657C>G, c.1640G>A, c.1639C>G, c.1636A>G, c.1619T>G, c.1618A>G, c.1617C>T, c.1076C>A, c.1067T>G, c.1067T>A, c.1065C>A, c.1064A>T, c.1057C>G, c.1056_1057delinsCG, c.1052T>G, c.1051C>T, c.1051C>G, c.1050C>A, c.1049A>T, c.1030T>C, c.1023G>C, c.1020C>G, c.1010C>G, c.1009G>A, c.1003C>T, c.1003C>G, c.970C>T, c.955A>G, c.943G>C, c.937G>A, c.923C>T, c.922C>T, c.920T>C, c.917C>A, c.916G>C, c.915C>G, c.911T>G, c.886A>C, c.880G>T, c.887T>G, c.873G>A, c.869G>A, c.854C>T, c.850T>G, c.845A>T, c.842G>A, c.839A>G, c.836G>T, c.835G>A, c.834T>G, c.833T>A, c.830C>T, c.832A>G, c.830C>G, c.829A>G, c.829A>C, c.827C>T, c.827C>A, c.826A>C, c.812G>A, c.811G>T, c.807G>T, c.806G>T, c.803T>C, c.797A>T, c.788C>T, c.787A>G, c.766G>T, c.766G>A, c.749T>C, c.747C>T, c.740C>T, c.740C>G, c.739T>C, c.727C>G, c.710A>G, c.700A>G, c.693G>T, c.682C>T, c.684C>A, c.668C>T, c.667T>C, c.649A>G, c.649A>C, c.641G>C, c.641G>A, c.638G>T, c.636C>T, c.635A>G, c.634G>A, c.592C>T, c.578C>T, c.523G>T, c.419A>C, c.388G>A, c.316T>G, c.242T>C, c.1749G>C, c.1742G>C, c.1741G>A, c.1687G>A, c.1682C>T, c.1678C>T, c.1663C>T, c.1658G>A, c.1657C>T, c.1639C>T, c.1630C>T, c.1058G>A, c.1004C>T, c.998G>A, c.997C>T, c.994A>G, c.949G>A, c.926C>T, c.917C>A, c.916G>A, c.915del, c.915C>A, c.911T>C, c.902G>A, c.902G>C, c.901G>A, c.881C>T, c.875T>C, c.853C>T, c.868G>T, c.868G>A, c.844G>C, c.841G>T, c.844G>A, c.841G>C, c.838T>C, c.833T>A, c.829A>T, c.821C>T, c.811G>C, c.802C>T, c.794C>T, c.797A>G, c.793G>A, c.782T>G, c.778C>T, c.748G>T, c.704C>T, c.693G>C, c.650C>T, c.640C>A, c.638G>A, c.629G>C, c.628G>T, c.620G>A, c.619C>T, c.602G>A, c.593G>A, c.587C>T, c.584C>T, c.583T>C, c.431G>A, c.430C>G, c.365C>T, c.1742G>C, c.1637T>C, c.1632_1G>T, c.1627G>A, c.1302_1G>C, c.1067T>C, c.1057C>A, c.1022A>G, c.928G>A, c.783G>C, c.841G>T, c.835G>T, c.799G>A, c.716G>T, c.715G>A, c.610C>T, c.593G>T, c.553G>C, c.545T>C, c.544G>A, c.430C>T, c.2597delinsTC, c.2331_2332insC, c.2318_2319insG, c.2245G>T, c.2127del, c.2126_2127del, c.2126_2127insC, c.1955del, c.1816A>T, c.1709del, c.1708C>T, c.1686C>A, c.1680_1681insG, c.1632_1G>A, c.1631_2T>C, c.1631_1G>A, c.1525_1G>A, c.1508C>G, c.1420G>T, c.1388_1389del, c.1229_1230insC, c.1220_1223del, c.1204G>T, c.1192_1193del, c.1160_1161insC, c.1160del, c.1154del, c.1123C>T, c.1118_2T>C, c.1117_1118insA, c.1114_1115insA, c.1093C>T, c.1086G>A, c.1079G>A, c.1041C>G, c.1024_2A>C, c.927+1T>C, c.927+1G>A, c.927+1G>C, c.848del, c.845del, c.817_1_81delinsAT, c.816_2T>A, c.816_1G>A, c.809G>A, c.807G>A, c.781_782insCT, c.691_1G>A, c.690_1G>A, c.687del, c.624del, c.514_1G>A, c.471G>A, c.439del, c.388_1G>C, c.388_1G>A, c.388_2A>G, c.387_2T>C, c.387+1G>A, c.367del, c.333_334del, c.297_1G>C, c.294C>G, c.286_287insC, c.2322C>T, c.222C>G, c.204_205insC, c.144_145insC, c.106del, c.2T>C, c.1686G>C, c.1583_1584delinsG, c.1540G>T, c.1539del, c.1534del, c.1528del, c.1525G>T, c.1480C>T, c.1452_1453insG, c.1452G>A, c.1444A>T, c.1411C>T, c.1375C>T, c.1368del, c.1356_1357insC, c.1342del, c.1273_1274del, c.1197del, c.1178del, c.1168C>T, c.1154_1155insT, c.1141_1142insG, c.1140_1141del, c.1137C>A, c.1133del, c.1104del, c.1089del, c.1089C>A, c.1089G>A, c.1080_1083del, c.1080G>A, c.1049_1050insA, c.1021C>T, c.1009_1010insGCGAC, c.992del, c.967del, c.883del, c.880_881insG, c.875_876delinsCT, c.856_857insC, c.856del, c.794_795insATCC, c.792C>G, c.792C>A, c.769G>T, c.753G>C, c.721_724del, c.668_669del, c.654G>A, c.653G>A, c.649_650del, c.644del, c.634_635insG, c.610del, c.605_606insG, c.562del, c.550del, c.518_519insT, c.493_494insC, c.477del, c.459del, c.437G>A, c.432del, c.418_421del, c.401del, c.354T>G, c.329_332del, c.330del, c.315del, c.297del, c.285C>A, c.274_275insG, c.273G>A, c.272G>A, c.236del, c.223del, c.216_217insC, c.204del, c.195_198del, c.197del, c.195_196del, c.144del, c.135del, c.119_120del, c.102del
KCNT1	NM_020822.3	c.862G>A, c.1309C>T, c.1420C>T, c.2677G>A, c.2678A>T, c.2687T>A, c.2688G>A, c.2688G>C, c.2782C>T, c.1193G>A, c.1283G>A, c.2386T>C, c.2686A>G, c.2800G>A, c.108del, c.785G>A, c.1421G>A, c.2849G>A, c.2882G>A, c.2896G>A
KIF1A	NM_001244008.2	c.920G>A, c.760C>T, c.757G>A, c.647G>A, c.646C>T, c.643A>C, c.296C>T, c.173C>T, c.38G>A, c.37C>T, c.32G>A, c.31C>T, c.967T>C, c.920G>C, c.919C>G, c.764C>A, c.641G>A, c.604G>C, c.556G>C, c.506G>C, c.467A>T, c.444G>T, c.442G>A, c.430G>T, c.275G>A, c.265T>G, c.254C>A, c.232G>C, c.799A>T, c.38G>T, c.37C>A, c.32G>T, c.2478C>G, c.3465_1G>A, c.3374_2T>G, c.2528_21G>A, c.2116_1G>C, c.1894_1G>C, c.1578_2A>G, c.1421_22T>A, c.1037_1G>C, c.958_1G>C, c.798_2T>C, c.798_1G>A, c.609_1G>A, c.1139G>C, c.1138C>T, c.821C>T, c.647G>C, c.452G>A, c.1048C>T, c.1031C>T, c.946C>T, c.773C>T, c.761G>A, c.746T>C, c.595G>A, c.500G>A, c.499G>C, c.304G>A, c.206C>T, c.1048C>G, c.746T>A, c.518T>C, c.512A>C, c.317C>A, c.308A>C, c.305G>A, c.233G>A, c.947G>A
KMT2A	NM_001197104.2	c.126del, c.166_167insA, c.293del, c.308del, c.416dup, c.572C>A, c.731T>G, c.766dup, c.766del, c.865A>T, c.883_886del, c.934_935insC, c.1065del, c.1147del, c.1274del, c.1274del, c.1395_1396insG, c.1470_1471del, c.15122G>T, c.1652dup, c.1660C>T, c.1700del, c.1704del, c.1808del, c.1822C>T, c.1837C>T, c.1975G>T, c.2068del, c.2193del, c.2322_2323del, c.2348_2349del, c.2388dup, c.2429_2430insG, c.2490dup, c.2517dup, c.2522del, c.2569G>T, c.2608A>T, c.2619_2620del, c.2635G>T, c.2865del, c.2958_2959insTTG, c.2968_2969insAGAG, c.3044del, c.3070dup, c.3190C>T, c.3193G>T, c.3337A>T, c.3340del, c.3373_3374del, c.3462del, c.3515del, c.3558_3559delinsT, c.3559C>T, c.3615C>A, c.3724C>T, c.3794del, c.3853C>T, c.3889G>T, c.3890del, c.3920dup, c.3926del, c.3933dup, c.3935_3936del, c.4069C>T, c.4246G>T, c.4331_4332del, c.4532del, c.4988T>A, c.5116del, c.5116G>A, c.5116del, c.5116G>T, c.5116del, c.5455C>T, c.5656del, c.5657del, c.5708_5709del, c.5822del, c.5887C>T, c.5901del, c.6033_6036del, c.6034A>T, c.6141_6142del, c.6150dup, c.5332del, c.6530del, c.6535del, c.6564del, c.6598del, c.6798del, c.7020del, c.7124del, c.7155del, c.7248del, c.7302del, c.7349del, c.7579del, c.7753del, c.7773T>A, c.7841del, c.7914T>A, c.7926del, c.7941_7942del, c.7990del, c.8055C>G, c.8185_8186del, c.8349_8352del, c.8454del, c.8874_8875del, c.9035del, c.9103C>T, c.9112_9115del, c.9139C>T, c.9195del, c.9275_9276del, c.9383del, c.9436C>T, c.9447_9450del, c.9449del, c.9519_9520del, c.9684del, c.9694C>T, c.9768_9769del, c.9839C>A, c.9847C>T, c.9910_9911del, c.10023del, c.10051del, c.10139C>G, c.10153_10154del, c.10171C>T, c.10322del, c.10550dup, c.10608del, c.10786del, c.10816G>T, c.10853del, c.10934del, c.10934dup, c.11073del, c.11110C>T, c.1122del, c.11230C>T, c.11280del, c.1138del, c.3215G>T, c.3463T>C, c.3464G>T, c.3472T>C, c.3473G>A, c.3473G>C, c.3539G>A, c.35395A>C, c.3557A>C, c.3560A>T, c.3566G>A, c.4342C>T, c.4343G>A, c.3460C>T, c.3464G>A, c.3565T>C, c.4367A>G, c.4696G>A, c.173del, c.173dup, c.458C>G, c.269C>A, c.478C>T, c.517C>T, c.835_838del, c.889del, c.1038del, c.1142del, c.1868del, c.2115del, c.2233C>T, c.2262del, c.2318del, c.2319del, c.2452A>T, c.2455C>T, c.2483C>G, c.2514del, c.2552_2553del, c.2629_2630del, c.2629_2630del, c.2673_2674del, c.2690C>G, c.3034C>T, c.3241C>T, c.3247C>T, c.3301C>T, c.3452C>T, c.3592C>T, c.3651_3652del, c.3651dup, c.3740_3741del, c.3790C>T, c.3991C>T, c.4029del, c.4032del, c.4177dup, c.4422G>A, c.4433del, c.4599dup, c.4504C>T, c.4906C>T, c.5256dup, c.5251A>T, c.5419C>T, c.5431C>T, c.5572C>T, c.5621del, c.578del, c.5935C>T, c.6002_6005del, c.6329C>G, c.6379C>T, c.6445C>T, c.6487C>T, c.6811del, c.6571C>T, c.6913del, c.7007del, c.7071del, c.7144C>T, c.7150C>T, c.7438C>T, c.7479del, c.7567_7570del, c.7831G>T, c.7975C>T, c.8095C>T, c.8183_8186del, c.8267del, c.8270dup, c.8407C>T, c.8767_8768del, c.8806_8809del, c.9068del, c.9133C>T, c.9921del, c.10064dup, c.10217C>A, c.10217C>G, c.10334del, c.11084C>G, c.11530del, c.3019G>T, c.3

		c.16445_16446del, c.16391_16392insAG, c.16361del, c.16336_16337insC, c.16184G>A, c.16170_16171insG, c.16154C>G, c.16125del, c.16109del, c.16084_16085del, c.15970del, c.15947del, c.15943C>T, c.15902_15903del, c.15858_15859del, c.15834del, c.15791G>A, c.15473del, c.15286C>T, c.15172del, c.15171G>A, c.15050_15051insdel, c.15013del, c.14989C>T, c.14976del, c.14971_14972insT, c.14949del, c.14925_14926ins, c.14923del, c.14885_14886insA, c.14868del, c.14867del, c.14843C>G, c.14833delinsA, c.14826_14827insT, c.14823_14825delinsTT, c.14726del, c.14722del, c.14713C>T, c.14700del, c.14688C>A, c.14669_14672del, c.14580_14581insT, c.14575C>T, c.11746C>T, c.11739del, c.11675_11676insA, c.11635del, c.11578C>T, c.11519_11520insT, c.11473del, c.11461C>T, c.11422del, c.11395C>T, c.11386del, c.11321_11340insG, c.11214del, c.11201_11202del, c.10999C>T, c.10957G>T, c.10858_10859insA, c.10834C>T, c.10825C>T, c.10777del, c.10707_10708insAG, c.10688_10689insA, c.10688_10689insAGA, c.10653_10654insT, c.10653_10654insA, c.10638del, c.10624_10625del, c.10552C>T, c.10538G>A, c.10496_10497del, c.10378C>T, c.10295_10296insT, c.10198C>T, c.10109del, c.10021C>T, c.10018C>T, c.9964C>T, c.9947del, c.9937_9939delinsA, c.9843_9844delinsCT, c.9835C>T, c.9824_9825insA, c.9790C>T, c.9773del, c.9730G>T, c.9730_9731del, c.9595del, c.9417del, c.9367del, c.9322G>T, c.93214del, c.93215del, c.93216del, c.93217del, c.93218del, c.93219del, c.93220del, c.93221del, c.93222del, c.93223del, c.93224del, c.93225del, c.93226del, c.93227del, c.93228del, c.93229del, c.93230del, c.93231del, c.93232del, c.93233del, c.93234del, c.93235del, c.93236del, c.93237del, c.93238del, c.93239del, c.932310del, c.932311del, c.932312del, c.932313del, c.932314del, c.932315del, c.932316del, c.932317del, c.932318del, c.932319del, c.932320del, c.932321del, c.932322del, c.932323del, c.932324del, c.932325del, c.932326del, c.932327del, c.932328del, c.932329del, c.932330del, c.932331del, c.932332del, c.932333del, c.932334del, c.932335del, c.932336del, c.932337del, c.932338del, c.932339del, c.932340del, c.932341del, c.932342del, c.932343del, c.932344del, c.932345del, c.932346del, c.932347del, c.932348del, c.932349del, c.932350del, c.932351del, c.932352del, c.932353del, c.932354del, c.932355del, c.932356del, c.932357del, c.932358del, c.932359del, c.932360del, c.932361del, c.932362del, c.932363del, c.932364del, c.932365del, c.932366del, c.932367del, c.932368del, c.932369del, c.932370del, c.932371del, c.932372del, c.932373del, c.932374del, c.932375del, c.932376del, c.932377del, c.932378del, c.932379del, c.932380del, c.932381del, c.932382del, c.932383del, c.932384del, c.932385del, c.932386del, c.932387del, c.932388del, c.932389del, c.932390del, c.932391del, c.932392del, c.932393del, c.932394del, c.932395del, c.932396del, c.932397del, c.932398del, c.932399del, c.932310del, c.932311del, c.932312del, c.932313del, c.932314del, c.932315del, c.932316del, c.932317del, c.932318del, c.932319del, c.932320del, c.932321del, c.932322del, c.932323del, c.932324del, c.932325del, c.932326del, c.932327del, c.932328del, c.932329del, c.932330del, c.932331del, c.932332del, c.932333del, c.932334del, c.932335del, c.932336del, c.932337del, c.932338del, c.932339del, c.932340del, c.932341del, c.932342del, c.932343del, c.932344del, c.932345del, c.932346del, c.932347del, c.932348del, c.932349del, c.932350del, c.932351del, c.932352del, c.932353del, c.932354del, c.932355del, c.932356del, c.932357del, c.932358del, c.932359del, c.932360del, c.932361del, c.932362del, c.932363del, c.932364del, c.932365del, c.932366del, c.932367del, c.932368del, c.932369del, c.932370del, c.932371del, c.932372del, c.932373del, c.932374del, c.932375del, c.932376del, c.932377del, c.932378del, c.932379del, c.932380del, c.932381del, c.932382del, c.932383del, c.932384del, c.932385del, c.932386del, c.932387del, c.932388del, c.932389del, c.932390del, c.932391del, c.932392del, c.932393del, c.932394del, c.932395del, c.932396del, c.932397del, c.932398del, c.932399del, c.932310del, c.932311del
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RAD21	NM_006265.3	c.1306C>T, c.1222G>T, c.1142G>A, c.1045_1046insA, c.593_594insG, c.579_580insA, c.578T>A, c.548T>C, c.466C>T, c.233_234del, c.208A>T, c.193C>T, c.182_183insCT, c.1808T>C, c.1127C>G, c.1550_1551insC, c.1548delinsTC, c.943_946del, c.704del, c.593_594insAG, c.589C>T, c.1753T>C, c.1635del, c.1432C>T, c.1161+1G>A, c.85delinsCCT, c.628G>T
RAF1	NM_002880.4	c.1837C>G, c.1472C>T, c.1082G>C, c.788T>G, c.781C>T, c.776C>A, c.775T>G, c.775T>A, c.770C>T, c.769T>C, c.768G>T, c.1472C>G, c.1457A>G, c.788T>A, c.786T>A, c.785A>T, c.785A>G, c.782C>T, c.782C>A, c.781C>G, c.778A>C, c.776C>T, c.776C>G, c.775T>C, c.769T>A, c.766A>G
RERE	NM_001042681.2	c.3785>G, c.2278C>T, c.659del, c.248_249insA, c.4456G>T, c.4246C>T, c.4023_4024insT, c.3732del, c.3586C>T, c.3249_3250insG, c.2721_2722delinsAT, c.2271_2272insGG, c.1920del, c.1681_1684del, c.452_453insC, c.348del, c.73_74insGA
RIT1	NM_006912.6	c.284G>C, c.270G>T, c.270G>A, c.268A>G, c.265T>C, c.246G>C, c.246T>A, c.244T>C, c.229G>T, c.229G>C, c.229G>A, c.104G>C, c.69A>C, c.270G>C, c.247A>C, c.245T>G, c.245T>C, c.244T>G, c.242A>G, c.241G>C, c.230C>G, c.170C>G, c.67A>C
RPS6KA3	NM_004586.3	c.1996C>T, c.1970C>G, c.1964del, c.1859del, c.1845del, c.1740del, c.1612del, c.1606del, c.1587T>G, c.1539_1542del, c.1308C>A, c.1155_1156del, c.1022del, c.958G>T, c.814del, c.770_773del, c.740_741insA, c.718_721del, c.646A>T, c.442del, c.432T>G, c.298A>T, c.251del, c.212_213insT, c.212T>G, c.185del, c.2186G>A, c.2185C>T, c.1762G>C, c.679T>G, c.5667A>T, c.224G>T, c.1606G>T, c.7097C>T, c.340C>T, c.1934G>A, c.1699C>T, c.1672C>T, c.1540C>T, c.1522C>T, c.1463_1464del, c.1304_1307del, c.1152del, c.932T>G, c.913C>T, c.898C>T, c.889_890del, c.817C>T, c.727C>T, c.451_452del, c.334C>T, c.327del, c.262_263insA, c.205G>T, c.2144del, c.2101_2A>G, c.2065C>T, c.2005_2008del, c.1959_2A>del, c.1894C>T, c.1765_2A>C, c.1000_2A>G, c.846_2A>G, c.845_5G>A, c.774_1G>A, c.774_1G>A, c.632_1G>A, c.632_2A>C, c.593_2_593+5del, c.486_3A>G, c.407_1G>T, c.328C>T, c.326_1G>C, c.326_11A>G, c.243_1G>A
RUNX2	NM_001024630.4	c.436G>A, c.476G>A, c.506G>A, c.506G>A, c.539C>A, c.524T>G, c.572G>A, c.571A>G, c.597G>T, c.578G>A, c.652A>G, c.659C>A, c.1085C>T, c.568C>T, c.569G>A, c.673C>T, c.674G>A, c.295dup, c.90dup, c.203delinsG, c.274del, c.423_2del, c.423_2T>G, c.424_2A>G, c.424_1G>C, c.577C>T, c.581_581+5del, c.594_595delinsG, c.625C>T, c.739del, c.859_1G>C, c.860_1G>T, c.891G>A, c.901C>T, c.909C>G, c.1171C>T, c.1205dup, c.1228dup, c.1281del, c.1379dup, c.1385dup, c.90del, c.163C>T, c.184C>T, c.187C>T, c.211C>T, c.217del, c.274dup, c.371C>A, c.411del, c.422dup, c.516dup, c.617del, c.637_640dup, c.645_646del, c.765del, c.770_771del, c.823del, c.850C>T, c.912del, c.407T>C, c.190C>T, c.278T>A, c.329del, c.604del, c.636dup, c.868C>T, c.874_875del, c.884del, c.1081C>T, c.1182T>A, c.1191del
SATB2	NM_001172509.2	c.1543G>A, c.1286G>A, c.1204G>A, c.1195C>T, c.1175G>A, c.1166G>T, c.392T>A, c.545T>C, c.346G>C, c.287T>G, c.185T>A, c.1196G>A, c.1174G>A, c.1165C>T, c.1946C>T, c.1166G>A, c.1942_1943del, c.1826_1827insGA, c.1826del, c.1756C>T, c.1741_161G>A, c.1728del, c.1627del, c.1624_1625insC, c.1610del, c.1592_1593insA, c.1515_1516insA, c.1515del, c.1504del, c.1495A>T, c.1477C>T, c.1474C>T, c.1393_1394insA, c.1375C>T, c.1314_1315insGGAG, c.1285C>T, c.1196del, c.1174_1G>A, c.1174_2A>G, c.1148del, c.1135C>T, c.1131_1132del, c.1105_1106insT, c.1016del, c.997C>T, c.955C>T, c.868>C, c.847C>T, c.808_809del, c.805G>T, c.715C>T, c.700_701G>A, c.652_654del, c.643_644insGT, c.622_623insA, c.598_2A>G, c.597_1G>A, c.581_584del, c.562C>T, c.553G>T, c.482del, c.474_2A>G, c.473_1G>A, c.3461_3464insAAGG, c.343C>T, c.337C>T, c.318T>G, c.163del, c.138del, c.124G>T, c.19_20insA, c.1654_1655del, c.1610_1611del, c.1606G>T, c.1569G>A, c.1330_1331insCCA, c.1249del, c.1225C>T, c.1218_1221del, c.1153del, c.1103_1106del, c.925C>T, c.913C>T, c.895del, c.853C>T, c.823C>T, c.728C>G, c.722_723insA, c.721_722del, c.674G>A, c.630T>G, c.568_569insA, c.554del, c.553_554insA, c.550A>T, c.509G>A, c.430C>T, c.382del, c.305del, c.225T>A, c.150del, c.115_116insA
SATB2	NM_001172509.2	c.4794T>G, c.4783_4784del, c.4777_4778del, c.4764T>A, c.4693C>T, c.4681G>T, c.4656T>A, c.4571del, c.4562del, c.4554del, c.4549A>T, c.4548del, c.4541T>G, c.4539_4540insA, c.4521C>G, c.4515_4516del, c.4462del, c.4450_4453del, c.4380T>A, c.4374C>A, c.4372del, c.4359T>G, c.4348C>T, c.4333A>T, c.4318del, c.4302G>A, c.4301G>A, c.4279C>T, c.4269_4272del, c.4266T>A, c.42565T>A, c.4244_4245del, c.4236del, c.4223C>A, c.4205_4208del, c.4216_4217insG, c.4188C>A, c.4166del, c.4177_4180delinsTG, c.4073G>A, c.3997del, c.4024G>T, c.3971_3972insT, c.3971_3972insinsA, c.3897_3890insG, c.3869T>A, c.3858G>A, c.3857G>A, c.3853_3854insT, c.3852del, c.3852G>A, c.3851G>A, c.3836_3837del, c.3829C>T, c.3828T>A, c.3783C>A, c.3812G>A, c.3813G>A, c.3822T>G, c.3730C>T, c.3736_3777insT, c.3757G>T, c.3726del, c.3631_3632ins, c.3635_3636delinsA, c.3635G>A, c.3612G>A, c.3704del, c.3690_3691insT, c.3614G>A, c.3611G>A, c.3609del, c.3607C>T, c.3553del, c.3487del, c.3477del, c.3455C>A, c.3457G>T, c.3439G>T, c.3431_3432del, c.3409del, c.3306C>A, c.3343_3344del, c.3361G>T, c.3327_3328insC, c.3295G>T, c.3241G>T, c.3195_3196del, c.3282del, c.3268_3269insA, c.3182del, c.3173_3176del, c.3139G>T, c.3124C>T, c.3082del, c.3017del, c.2980_2981del, c.2975_2976insT, c.2961del, c.2958_2959del, c.2904C>A, c.2893C>T, c.2877T>A, c.2863_2864insGA, c.2856G>A, c.2855G>A, c.2851del, c.2856_2859insATCT, c.2523_2524del, c.2495G>A, c.2460_2461insT, c.2486T>C, c.2043G>A, c.5582G>A, c.5563C>T, c.5383G>A, c.3765G>A, c.3726_3727insT, c.3635_3636delinsA, c.3635G>A, c.3612G>A, c.3704del, c.3690_3691insT, c.3614G>A, c.3611G>A, c.3609del, c.3607C>T, c.3553del, c.3496del, c.3497_3498insGTC, c.3534del, c.3529_3530insA, c.3514C>A, c.3502del, c.3503del, c.4996_4997insATGT, c.4954G>T, c.4905C>T, c.4884T>A, c.4811G>A, c.2402_2403insT, c.2350_2351del, c.2318_2319insG, c.2302_2303insGACC, c.2286del, c.2266A>T, c.2264T>G, c.2261G>A, c.2244G>A, c.2243G>A, c.2214G>A, c.2212G>A, c.2208del, c.2194C>T, c.2167del, 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c.3705_55G>A, c.3705_51G>A, c.3705_51G>C, c.3705_51G>T, c.3705_51G>A, c.3551_3551delinsT, c.3551_3551delinsC, c.3551_3551delinsA, c.3553del, c.5549del, c.5539del, c.5539_5540insTCCA, c.5539del, c.5506del, c.5489_5490insA, c.5486del, c.5479del, c.5479_5480insGTC, c.5452del, c.5392del, c.5383G>T, c.5373del, c.531_5316insC, c.5249del, c.5233A>T, c.5217del, c.5212_5213del, c.5209_5210del, c.5201_5202insTCT, c.5115del, c.5087del, c.5045del, c.5016del, c.4999del, c.4980_4981del, c.4972_4973delinsT, c.4969_4970insATTC, c.4969del, c.4965del, c.4964del, c.4965del, c.4906del, c.4985_4985insAT, c.4854_4855insT, c.2440G>T, c.2420_2421insT, c.2374del, c.2314del, c.2314del, c.2295del, c.2287del, c.2253del, c.2232_2233del, c.2226del, c.2217T>G, c.2208_2209del, c.2179_2180insA, c.2173G>T, c.2172del, c.2147_2150del, c.2111_2112insT, c.2081C>T, c.2061_2062del, c.2044G>T, c.2011del, c.1972del, c.1922_1923delinsATC, c.1900del, c.1895insC, c.1883_1883insC, c.1853_1854del, c.1843del, c.1839_1840del, 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c.2287del, c.2253del, c.2232_2233del, c.2226del, c.2217T>G, c.2208_2209del, c.2179_2180insA, c.2173G>T, c.2172del, c.2147_2150del, c.2111_2112insT, c.2081C>T, c.2061_2062del, c.2044G>T, c.2011del, c.1972del, c.1922_1923delinsATC, c.1900del, c.1895insC, c.1883_1883insC, c.1853_1854del, c.1843del, c.1839_1840del, c.1835_1835insC, c.1810_1811insC, c.1807_1808del, c.1752_1753insT, c.1745_1746insA, c.1709del, c.1685_1686del, c.1660C>T, c.1659_1660insT, c.1634_1635del, c.1635T>G, c.1598del, c.1561C>T, c.1543A>T, c.1517del, c.1486del, c.1466T>A, c.1463del, c.1439C>A, c.1439del, c.1397del, c.1363C>T, c.1354A>T, c.1349del, c.1344_1345insT, c.1337del, c.1324del, c.1303del, c.1301_1302insT, c.1278_1279delinsA, c.1273del, c.1259del, c.1239C>

SMARCE1	NM_003079.5	c.218A>G, c.227A>G, c.218A>C, c.541+1G>A, c.959del, c.816+1del, c.816+1G>T, c.714+1G>A, c.369+1G>T, c.369+1G>A, c.238-2A>G, c.237+1G>T, c.871C>T, c.808del, c.757C>T, c.751C>T, c.688C>T, c.683_685delinsT, c.673C>T, c.587del, c.585_586delinsA, c.554del, c.493G>T, c.472C>T, c.454_455delinsT, c.417C>G, c.411del, c.340G>T, c.328G>T, c.313C>G, c.292del, c.92del
SMC1A	NM_006306.4	c.3568A>G, c.3497A>C, c.3367C>T, c.3344G>T, c.3070A>T, c.2683C>G, c.2493_2495del, c.2341T>C, c.2078G>A, c.2046_2048del, c.1903C>T, c.1714C>T, c.1504G>A, c.1478A>C, c.802_804del, c.587G>A, c.586C>G, c.586C>T, c.421G>A, c.397T>G, c.128A>T, c.110G>T, c.3254A>G, c.3146G>A, c.2420G>A, c.2369G>A, c.2351T>C, c.2368C>T, c.2132G>A, c.2131C>T, c.1756C>T, c.1487G>A, c.1486C>T, c.1193G>A, c.3497A>G, c.3178G>A, c.2948A>G, c.2095C>T, c.3552_3553insGGCC, c.3508-2A>G, c.3437+1G>A, c.3249_3250insG, c.3181C>T, c.3151C>T, c.3145C>T, c.3123del, c.3103C>T, c.2973+1G>A, c.2923C>T, c.2873del, c.2853_2856del, c.2814_2815insT, c.2769_2770insC, c.2611C>T, c.2562+1G>A, c.2547del, c.2477del, c.2394_2395insA, c.2394del, c.2314-2A>G, c.2272C>T, c.2197G>T, c.2161C>T, c.1911+1G>T, c.1609del, c.1495C>T, c.1299del, c.1171C>T, c.1114-2A>G, c.855-2A>G, c.655del, c.616-2A>G, c.411+1G>C, c.157_158insA, c.116C>G, c.3283C>T, c.3255T>A, c.3119_3120insA, c.3040C>T, c.3022C>T, c.2899C>T, c.2872_2873insC, c.2845_2846insGATG, c.2437C>T, c.2398C>T, c.2173C>T, c.2146C>T, c.2122del, c.2076del, c.2069del, c.2039_2040del, c.2035G>T, c.1990del, c.1977del, c.1910del, c.1813_1814del, c.1785_1788del, c.1692_1693insT, c.1388_1389insC, c.1192C>T, c.1072_1073del, c.761del, c.733_734del, c.628C>T, c.583G>T, c.547-548del, c.547C>T, c.481del, c.392del, c.374del, c.371T>A, c.302del, c.52_53del, c.3362G>A, c.3115C>T, c.2364del, c.511C>T
SMC3	NM_005445.4	c.139T>C, c.562A>G, c.587T>C, c.703_705del, c.707G>C, c.720_722del, c.859_861dup, c.1453_1455del, c.1464_1466del, c.1964G>A, c.1997G>C, c.2750A>C, c.3298G>T, c.3439C>G, c.3362C>T, c.778C>T, c.1812+1G>A
SOS1	NM_005633.4	c.508A>G, c.322G>A, c.1867T>A, c.1656G>C, c.1656G>T, c.1655G>T, c.1655G>C, c.1655G>A, c.1654A>T, c.1654A>G, c.1642A>C, c.806T>C, c.2536G>A, c.1294T>C, c.1294T>A, c.3134C>G, c.2197A>T, c.1649T>C, c.1644T>A, c.1322G>A, c.1310T>G, c.1310T>C, c.1300_1301delinsAA, c.1300G>C, c.1300G>A, c.1297G>A, c.925G>T, c.806T>G, c.797C>A, c.305G>G
SOS2	NM_006939.4	c.800T>G, c.800T>C, c.800T>A, c.791C>G, c.791C>A, c.1127C>G, c.1126A>T
SOX9	NM_000346.4	c.326T>C, c.327G>T, c.337A>T, c.339G>A, c.358C>G, c.491A>C, c.493C>T, c.494A>G, c.508C>A, c.509C>T, c.526C>T, c.227C>A, c.455G>C, c.337A>G, c.462C>G, c.472G>A, c.473C>A, c.508C>T, c.517A>G, c.527C>T, c.196G>T, c.252C>G, c.296del, c.344G>A, c.359G>T, c.349C>T, c.432-2A>C, c.442G>T, c.557_560del, c.583C>T, c.685+2T>C, c.686-2A>G, c.736dup, c.788dup, c.788del, c.855_858dup, c.1103dup, c.1249C>T, c.1320C>A, c.68del, c.436_437dup, c.517A>T, c.522C>A, c.522C>G
SPECC1L	NM_015330.6	c.1189A>C, c.3293G>A, c.3247G>A
SPTAN1	NM_001130438.3	c.6605_6607del, c.4828C>T, c.6619_6621del, c.6781C>T, c.415C>T, c.1006C>T, c.1086-2A>C, c.1879C>T, c.4615C>T, c.4936C>T, c.5832+1G>C, c.6367del, c.58dup, c.133C>T, c.315_316insA, c.466C>T, c.840dup, c.1347_1348del, c.1367G>A, c.1642C>T, c.1773_1774del, c.1834C>T, c.2401C>T, c.2860del, c.2961del, c.2988dup, c.3251_3252del, c.3279del, c.3292del, c.3388C>T, c.5468_5471del, c.5567_5568del, c.6019_6020del, c.6100C>T, c.6937dup, c.6993dup, c.7225_7228dup
SRCAP	NM_006662.3	c.6985C>T, c.7303C>T, c.7330C>T, c.6208C>T, c.4557_4560del, c.55-1G>A, c.148del, c.435del, c.833del, c.1608_1611delinsCA, c.1793del, c.2518C>T, c.3833C>A, c.4648del, c.4925T>A, c.5102_5103del, c.5387del, c.5527del, c.5977_5980del, c.5984del, c.6889del, c.6901del, c.7000C>T, c.7189G>T, c.7215_7216del, c.7218dup, c.7219C>T, c.7220_7221del, c.7262dup, c.7275_7276del, c.7287C>A, c.7300G>T, c.7376del, c.7394del, c.7466C>G, c.7549del, c.7863dup, c.7937_7938del, c.8242C>T, c.577C>T, c.2471_2474dup, c.3269del, c.3628C>T, c.3738dup, c.4286_4287del, c.6709C>T
STAT3	NM_139276.3	c.2144C>T, c.2114A>G, c.1988C>T, c.1974G>T, c.1938C>G, c.1934T>A, c.1915C>A, c.1915C>G, c.1910T>C, c.1909G>T, c.1907C>A, c.1863C>A, c.1859C>G, c.1311C>A, c.1243G>A, c.1182G>A, c.1175A>G, c.1145G>T, c.1145G>C, c.1032G>C, c.1022C>T, c.988C>T, c.833G>A, c.454C>T, c.2147C>T, c.2134T>C, c.2117T>C, c.2116C>A, c.1979T>C, c.1976T>A, c.1974G>C, c.1970A>G, c.1909G>A, c.1865C>T, c.1858A>G, c.1699A>G, c.1397A>C, c.1387_1389del, c.1268G>A, c.1261G>A, c.1166C>T, c.1003C>T, c.995A>T, c.994C>T, c.1234A>T, c.832C>T
STXBP1	NM_001032221.6	c.107T>G, c.145_148del, c.148_149del, c.225T>G, c.241G>T, c.260_261del, c.294del, c.298del, c.360del, c.374_375del, c.400_401del, c.420T>A, c.451_454del, c.463del, c.512_513del, c.520del, c.586A>T, c.636T>A, c.636T>G, c.639del, c.661del, c.660_661del, c.662dup, c.685T>A, c.692dup, c.704_705insGG, c.785del, c.792C>A, c.912_915del, c.915del, c.920_921del, c.956del, c.971del, c.980dup, c.989dup, c.101C>G, c.1012C>T, c.1017del, c.1028del, c.1030del, c.1067dup, c.1072del, c.1074C>A, c.1074C>G, c.1079del, c.1092dup, c.1092del, c.1153dup, c.1176del, c.1236del, c.1258G>T, c.1304del, c.1354dup, c.1360-1_1360insC, c.1392del, c.1419C>A, c.1427C>A, c.1470del, c.1492dup, c.1495_1496del, c.1503T>A, c.1510del, c.1516_1517del, c.1570A>T, c.1597del, c.1606del, c.1607_1608del, c.1641_1642insC, c.122T>A, c.122T>G, c.125C>A, c.136A>G, c.164T>C, c.224A>G, c.236C>T, c.251T>A, c.416C>A, c.536T>A, c.539G>A, c.620A>G, c.701A>G, c.733C>A, c.733C>G, c.748C>G, c.767T>C, c.785A>T, c.874C>G, c.875G>C, c.875G>T, c.1060T>C, c.1060T>G, c.1216C>A, c.1216C>G, c.1227_1229del, c.1277T>G, c.1315A>T, c.1328T>G, c.1438C>T, c.1454T>A, c.1457T>G, c.1627G>C, c.1630G>A, c.1631G>C, c.1652G>C, c.1657G>C, c.1706G>C, c.1759del, c.1247T>C, c.175G>A, c.328T>C, c.416C>T, c.560C>T, c.568C>T, c.704G>A, c.734A>G, c.842T>A, c.847C>A, c.847G>A, c.847T>A, c.1003C>T, c.1061G>A, c.1216C>T, c.1217G>A, c.1217G>T, c.1277T>C, c.1439C>T, c.1630G>A, c.1631G>T, c.1651C>A, c.1651C>G, c.1652G>A, c.1652G>C, c.1077C>A, c.636C>T, c.637del, c.1246del, c.1247del, c.1570C>A, c.1934del, c.2061_2062del, c.2062_2063del, c.2063_2064del, c.2064_2065del, c.2065_2066del, c.2066_2067del, c.2067_2068del, c.2068_2069del, c.2069_2070del, c.2070_2071del, c.2071_2072del, c.2072_2073del, c.2073_2074del, c.2074_2075del, c.2075_2076del, c.2076_2077del, c.2077_2078del, c.2078_2079del, c.2079_2080del, c.2080_2081del, c.2081_2082del, c.2082_2083del, c.2083_2084del, c.2084_2085del, c.2085_2086del, c.2086_2087del, c.2087_2088del, 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TUBA1A	NM_006009.4	c.1285G>C, c.1265G>A, c.1264C>T, c.1264C>A, c.1256C>T, c.1246G>A, c.1226T>C, c.1225G>A, c.1216C>T, c.1216C>G, c.1205G>T, c.1204C>T, c.1204C>A, c.1190T>C, c.1186G>T, c.1177C>T, c.1169G>C, c.1169G>A, c.1168C>T, c.1168C>G, c.1160C>T, c.1148C>T, c.1148C>A, c.1112T>A, c.1105G>A, c.1096G>A, c.998C>T, c.995T>C, c.978A>C, c.959G>A, c.958C>T, c.922C>T, c.920C>T, c.919C>T, c.908T>G, c.875C>T, c.856C>T, c.848A>G, c.818C>A, c.790C>T, c.787C>A, c.746A>G, c.703G>T, c.703G>C, c.657T>G, c.656T>C, c.655A>G, c.652G>T, c.641G>A, c.640C>T, c.629A>G, c.626T>C, c.615C>A, c.545T>C, c.481T>G, c.481T>C, c.473C>T, c.467G>A, c.455T>A, c.431G>T, c.368G>A, c.367C>T, c.352G>C, c.352G>A, c.337G>A, c.321C>G, c.302A>G, c.283G>T, c.236G>A, c.215C>G, c.214C>T, c.209T>C, c.191G>A, c.190C>T, c.167C>T, c.163G>A, c.79G>C, c.74G>T, c.74G>A, c.4C>A, c.791G>A, c.790C>G, c.521C>T, c.518C>T, c.449C>T, c.235C>T, c.82C>T, c.53A>G
TUBB	NM_178014.4	c.361C>T, c.895A>G, c.897G>A, c.1057G>A, c.1201G>A, c.860C>T
TUBB2A	NM_001069.3	c.1228G>A, c.1073C>T, c.1072C>A, c.1070C>T, c.1033A>T, c.785G>A, c.741C>G, c.689G>T, c.580G>A, c.293G>T, c.292G>C, c.872A>C, c.728C>T, c.533C>T, c.5G>A
TUBB4A	NM_006087.4	c.1228G>A, c.1172G>A, c.1164G>C, c.1164G>A, c.1162A>G, c.1062C>G, c.811G>A, c.785G>A, c.763G>A, c.745G>A, c.731G>T, c.730G>C, c.730G>A, c.539T>C, c.535G>C, c.533C>T, c.533C>G, c.467G>T, c.286G>A, c.4C>T, c.4C>G, c.1172G>T, c.1163T>C, c.538G>A
WDR45	NM_001029896.2	c.891_892insT, c.872_873insCCGT, c.797_798del, c.790_791insACT, c.771_772insA, c.770_771insA, c.672del, c.667_668del, c.657_658del, c.618_619insA, c.618del, c.616del, c.604del, c.599_600insT, c.561_562insCACG, c.510_511insA, c.501del, c.498_499del, c.488del, c.487G>T, c.457del, c.307A>T, c.244del, c.227del, c.213_214insTAGT, c.209del, c.180del, c.139_140insC, c.139C>T, c.236_21A>G, c.749_751del, c.884G>A, c.870C>G, c.879_880del, c.865C>T, c.777del, c.697C>T, c.659_660del, c.634C>T, c.598_599del, c.604C>T, c.584_585del, c.434_435insA, c.408_409insT, c.397C>T, c.371_372del, c.246G>A, c.225_226insA, c.436+5G>C, c.1004_1005del, c.974_1G>A, c.973+1G>A, c.966_967insT, c.828_2A>G, c.827+5G>C, c.827+2_8273del, c.827+1G>T, c.827+1G>C, c.827+1G>A, c.725+1G>T, c.517_2A>G, c.516+1_516+3del, c.516+2T>C, c.516+1G>T, c.516+1G>A, c.516G>C, c.508C>T, c.437_1G>A, c.437_2A>T, c.437_2A>C, c.437_4_437_2delinsT, c.436+2T>G, c.436+2T>C, c.342_2A>C, c.340_341+2del, c.236_1G>T, c.235+1G>A, c.226G>T, c.131_2A>G, c.64del, c.55+2_55+3del, c.19C>T, c.574C>T, c.292G>T, c.81_82del, c.78_79del
ZBTB20	NM_001348800.3	c.1955A>T, c.1955A>G, c.1931C>T, c.1916G>T, c.1916G>A, c.1906T>C, c.1862T>G, c.1862T>C, c.1861C>T, c.1861C>G, c.1786C>T, c.1788C>G, c.1766C>A, c.1739G>A, c.1787A>G, c.1024del, c.918del, c.1734T>A, c.1727_1728insA, c.1431_1432insA, c.1412_1413insC, c.1350del, c.1256_1257insG, c.744C>G
ZC4H2	NM_018684.4	c.320_321del, c.637C>T, c.593G>A, c.592C>T, c.650A>G, c.575G>A, c.197T>A, c.187G>C, c.627G>C, c.617G>T, c.601C>T, c.275_276del, c.243_246del, c.199C>T, c.172G>T, c.148C>T, c.618C>A, c.561+1G>A, c.450_451insC, c.450del, c.426G>A, c.225G>T, c.672C>T, c.672C>A, c.225+2_225+3del, c.19C>T, c.574C>T, c.292G>T, c.81_82del, c.78_79del
ZEB2	NM_014795.4	c.2986_2987del, c.2908C>T, c.2894T>A, c.2890G>T, c.2885C>G, c.2803_2804insA, c.2757del, c.2745del, c.2609C>A, c.2592del, c.2579_2580insCT, c.2577_2578insT, c.2467C>T, c.2404_2407del, c.2398_2399insT, c.2384del, c.2374_2375insT, c.2365del, c.2357_2358insT, c.2337del, c.2303del, c.2273_2274del, c.2242_2243insC, c.2214_2215insTCC, c.2177_2178del, c.2177_2178insACTC, c.2142_2143del, c.2122del, c.2102C>A, c.2100C>G, c.2095C>T, c.2072G>A, c.2064del, c.2061del, c.2037del, c.2032del, c.2024del, c.1910C>A, c.1870A>T, c.1866_1867insC, c.1848del, c.1821_1824del, c.1806C>T, c.1792C>T, c.1770del, c.1747del, c.1703_1704insCTGA, c.1700_1703del, c.1687del, c.1653del, c.1648C>T, c.1630_1631del, c.1630_1631insA, c.1586del, c.1570_1571insA, c.1556del, c.1541_1542insC, c.1541_1542insA, c.1537del, c.1474A>T, c.1451C>A, c.1434C>A, c.1420C>T, c.1408_1409insA, c.1384_1385del, c.1364del, c.1349_1350del, c.1277T>A, c.1218del, c.1209del, c.1200_1201del, c.1138_1139insCTTA, c.1115del, c.1106T>A, c.1092_1093insCA, c.1038_1039del, c.1038del, c.997A>T, c.909_910insTTTC, c.904del, c.891_892delinsAT, c.876_877insA, c.874A>T, c.858del, c.855_856insA, c.846del, c.821_822insAAC, c.789_790del, c.782_783del, c.776_777insA, c.769G>T, c.760_761del, c.703del, c.703G>T, c.696C>A, c.674C>A, c.639C>A, c.621_622insT, c.502C>T, c.498C>A, c.493G>T, c.441C>G, c.329del, c.313del, c.304_305insA, c.280G>T, c.247_248insG, c.234_235del, c.232del, c.194_195del, c.3218A>G, c.3211T>C, c.3178T>C, c.3170G>A, c.3134A>G, c.3133C>T, c.3023T>G, c.899A>G, c.2866C>T, c.2806del, c.2769C>G, c.2761C>T, c.2718del, c.2717del, c.2701C>T, c.2688_2689insA, c.2688_2689insCA, c.2555C>G, c.2501del, c.2453_2454insT, c.2425G>T, c.2228_2229del, c.2227del, c.2180del, c.2179_2180del, c.2083C>T, c.2056G>T, c.1944del, c.1956C>A, c.1921del, c.1892del, c.1884del, c.1876G>T, c.1862del, c.1843C>T, c.1825G>T, c.1754del, c.1749C>A, c.1645A>T, c.1640C>G, c.1489C>T, c.1426_1427insA, c.1387del, c.1381C>T, c.1309C>T, c.1277T>G, c.1257del, c.1173_1176del, c.1170del, c.1168del, c.1102C>T, c.1052del, c.1027C>T, c.1017del, c.999del, c.980C>A, c.904C>T, c.857_858del, c.823C>T, c.805C>T, c.770_771del, c.761_762insAC, c.660C>G, c.656del, c.640_641insTGCC, c.553_554insTG, c.585del, c.481del, c.460del, c.432_433insT, c.425C>G, c.310C>T, c.236_239del, c.289del, c.3214C>T, c.3098del, c.3095del, c.3067+2T>G, c.3067+1G>A, c.3031del, c.3025C>T, c.3002del, c.2886+1G>A, c.1966_1967del, c.1467T>A, c.917-1G>A, c.917-2A>G, c.916+1G>A, c.808-1G>T, c.808-2A>G, c.712G>T, c.625C>T, c.379del, c.350_351del, c.73+2T>G, c.73+1del, c.73+1G>A, c.930C>A, c.363_364insGG, c.351_352insT, c.351T>A, c.175G>T