Gene：

ACTB

NM ID：

NM_001101.3

Variant：

c.19G>A, c.220G>A, c.1043C>T, c.547C>T, c.625G>A, c.629G>A, c.616C>T, c.617G>A, c.586C>T, c.587G>A

Gene：

ACTG1

NM ID：

NM_001614.3

Variant：

c.791C>T, c.94C>T, c.464C>T, c.766C>T, c.773C>T, c.760C>T, c.353A>T, c.1003C>T, c.611C>G, c.721G>A

Gene：

ACTG2

NM ID：

NM_001615.3

Variant：

c.613G>A, c.187C>G, c.593G>A, c.769C>T, c.770G>A, c.118C>T, c.119G>A, c.532C>T, c.533G>A, c.533G>T

Gene：

ACVR1

NM ID：

NM_001105.4

Variant：

c.983G>A, c.617G>A

Gene：

ADNP

NM ID：

NM_015339.2

Variant：

c.2492TAAA[2>1] (std: c.2492TAAA[1] alt: c.2496_2499delTAAA), c.190dupA, c.517C>T, c.2491_2494delTTAA, c.3067AG[3>1] (std: c.3067AG[1] alt: c.3069_3072delAGAG), c.58A[7>8] (std: c.64dupA alt: c.64_65insA), c.539_542delTTAG, c.54TG[2>1] (std: c.54TG[1] alt: c.56_57delTG), c.790C>T, c.2156dupA, c.2157C>G, c.2157C>A, c.2213C>G, c.2213C>A, c.817C[3>2] (std: c.817C[2] alt: c.819delC), c.2188C>T, c.1235delT, c.2286T[2>1] (std: c.2286T[1] alt: c.2287delT)

Gene：

AKT3

NM ID：

NM_005465.4

Variant：

c.1330A>G, c.49G>A, c.548T>A, c.1393C>T, c.803T>C

Gene：

ANKRD11

NM ID：

NM_013275.5

Variant：

c.2398_2401delGAAA, c.2327T>G, c.2293AGAA[2>1] (std: c.2293AGAA[1] alt: c.2297_2300delAGAA), c.2407A[6>2] (std: c.2407A[2] alt:

c.2409_2412delAAAA), c.2407A[6>1] (std: c.2407A[1] alt: c.2408_2412delAAAAA), c.3198_3199delTA, c.1801C>T, c.2171CAAA[2>1] (std: c.2171CAAA[1] alt: c.2175_2178delCAAA), c.1977C>G, c.1977C>A, c.3084C>A, c.3220AAAG[2>1] (std: c.3220AAAG[1] alt: c.3224_3227delAAAG), c.1898AAACA[2>1] (std: c.1898AAACA[1] alt: c.1903_1907delAAACA), c.2197C>T, c.2518delC, c.4384dupA, c.2512C>T, c.1318C>T, c.2692C>T, c.4383GA[4>3] (std: c.4383GA[3] alt: c.4389_4390delGA), c.2704G>T, c.160C>T, c.3302A[8>9] (std: c.3309dupA alt: c.3309_3310insA), c.2647G>T, c.3019C>T, c.4396_4397delAG, c.1372C>T, c.867C>A, c.1458AG[3>2] (std: c.1458AG[2] alt: c.1462_1463delAG), c.1373GAAA[3>2] (std: c.1373GAAA[2] alt: c.1381_1384delGAAA), c.976G[2>1] (std: c.976G[1] alt: c.977delG), c.6786C[7>6] (std: c.6786C[6] alt: c.6792delC), c.3702_3705delTAAA, c.2824AG[3>1] (std: c.2824AG[1] alt: c.2826_2829delAGAG), c.3704_3707delAACA, c.3769A[3>1] (std: c.3769A[1] alt: c.3770_3771delAA), c.4087C>T, c.7569+1G>A, c.7814T>G, c.7535G>A, c.7825C>T, c.5772C[6>7] (std: c.5777dupC alt: c.5777_5778insC), c.6834TG[2>1] (std: c.6834TG[1] alt: c.6836_6837delTG), c.6847C>T, c.6977C[6>7] (std: c.6982dupC alt: c.6982_6983insC)

Gene：

ARID1A

NM ID：

NM_006015.4

Variant：

c.3230C>A, c.3067T>C

Gene：

ARID1B

NM ID：

NM_020732.3

Variant：

c.1239G[5>4] (std: c.1239G[4] alt: c.1243delG), c.1258A[2>3] (std: c.1259dupA alt: c.1259_1260insA), c.3581C[6>7] (std: c.3586dupC alt: c.3586_3587insC), c.4144C[5>4] (std: c.4144C[4] alt: c.4148delC), c.3689+1G>C, c.3535C>T, c.1960C>T, c.4870C>T, c.4273dupT, c.2393_2396delGAAA, c.2371G>A, c.3136-2A>G, c.4014-1G>A, c.4009C>T, c.2248C>T, c.3322A[3>1] (std: c.3322A[1] alt: c.3323_3324delAA), c.1762G>T, c.2692C>T, c.6382C>T, c.3223C>T, c.3304C>T, c.3898C>T, c.5338C>T, c.6322C>T, c.5404C>T, c.4110G>A, c.3096_3100delCAAAG, c.5390TGTT[2>1] (std: c.5390TGTT[1] alt: c.5394_5397delTGTT), c.1914C>A, c.5267_5270delAAAG, c.4741C>T, c.5566AAGA[2>1] (std: c.5566AAGA[1] alt: c.5570_5573delAAGA), c.2077G>T, c.1618C>T, c.5542C[6>7] (std: c.5547dupC alt: c.5547_5548insC), c.5830C>T, c.1621C>T, c.6041G>A, c.5703dupT, c.5776C>T, c.2196G[6>7] (std: c.2201dupG alt: c.2201_2202insG), c.5968C>T, c.2149C>T

Gene：

ASXL1

NM ID：

NM_015338.5

Variant：

c.1720-1G>A, c.3633CTCC[2>1] (std: c.3633CTCC[1] alt: c.3637_3640delCTCC), c.2893C>T, c.1927G[8>9] (std: c.1934dupG alt: c.1934_1935insG), c.4243C>T, c.1542TG[2>1] (std: c.1542TG[1] alt: c.1544_1545delTG), c.1158GT[3>2] (std: c.1158GT[2] alt: c.1162_1163delGT), c.1210C>T

Gene：

ASXL3

NM ID：

NM_030632.1

Variant：

c.1978_1981delGACA, c.1471C>T, c.4215CT[3>2] (std: c.4215CT[2] alt: c.4219_4220delCT), c.3349C>T, c.3106C>T, c.4330C>T, c.1188ACAG[2>1] (std: c.1188ACAG[1] alt: c.1192_1195delACAG), c.4399C>T

Gene：

ATP1A2

NM ID：

NM_000702.3

Variant：

c.1127C>G, c.1127C>T, c.2723G>A, c.1091C>T, c.1148G>A, c.2563G>A, c.2501G>A, c.3027T>A, c.1843G>A, c.2066G>A, c.2936C>T, c.1643G>A, c.1642C>T, c.3005G>A, c.1777C>T, c.788C>T, c.1882G>A, c.1816G>A, c.2143G>A, c.1261C>T

Gene：

ATP1A3

NM ID：

NM_152296.4

Variant：

c.2443G>A, c.2452G>A, c.967C>T, c.2323C>A, c.1838C>T, c.2315G>A, c.2266C>T, c.2839G>T, c.2839G>A, c.2267G>A, c.2767G>T, c.2767G>A, c.829G>A, c.1072G>C, c.1072G>A, c.2224G>T, c.2116G>A, c.2401G>A, c.410C>T, c.410C>A, c.2263G>T

Gene：

BCL11A

NM ID：

NM_022893.3

Variant：

c.787C[7>8] (std: c.793dupC alt: c.793_794insC), c.385+2T>C

Gene：

BICD2

NM ID：

NM_001003800.1

Variant：

c.2320G>A, c.2321A>G, c.320C>T, c.1667A>C, c.1636_1638delAAT, c.2080C>T, c.2108C>T, c.2239C>T

Gene：

BRAF

NM ID：

NM_004333.4

Variant：

c.1914T>G, c.1914T>A, c.2135C>A, c.1409C>G, c.1406G>A, c.1405G>C, c.1403T>C, c.1399T>G, c.1391G>T, c.1391G>A, c.1743T>A, c.1390G>A, c.1447A>C, c.1460T>G, c.1781A>T, c.1789C>G, c.1787G>T, c.1785T>G, c.1785T>A, c.1783T>C, c.1801A>C, c.1794_1796dupTAC, c.1796C>T, c.1796C>G, c.1799T>G, c.1799T>A, c.1454T>C, c.1455G>T, c.1455G>C, c.1741A>G, c.1574T>C, c.1574T>A, c.1502A>T, c.1502A>G, c.1502A>C, c.1495A>G, c.1501G>A, c.1497A>C, c.1695T>G, c.1593G>C, c.1595G>A, c.1600G>C, c.721A>C, c.722C>T, c.722C>G, c.722C>A, c.730A>C, c.1722C>G,

c.735A>T, c.735A>C, c.736G>C, c.739T>G, c.739T>C, c.740T>C, c.741T>G, c.785A>G, c.785A>C, c.793G>C, c.755G>C, c.784C>A, c.769C>A, c.770A>G, c.2126A>G, c.823G>A, c.1442C>A

Gene:

CACNA1A

NM ID:

NM_001127221.1

Variant:

c.3993-2A>G, c.4037G>A, c.4055G>T, c.4046G>A, c.4054C>T, c.4036C>T, c.4982G>A, c.4991G>A, c.4990C>T, c.4252C>T, c.439G>A, c.4999C>T, c.1597G>A, c.5569C>T, c.5559_5560delCA, c.4075C>T, c.5251C>T, c.6100C>T, c.4497CTT[3>2] (std: c.4497CTT[2] alt: c.4503_4505delCTT), c.4517T>C, c.3695+1G>A, c.2042_2043delAG, c.3825+1G>A, c.2042delA, c.5263G>A, c.5428A>C, c.1363C>T, c.2009G>A, c.5059C>T, c.3106dupT, c.6205C>T, c.1060C>T, c.653C>T, c.2002G>A, c.6193-2A>C, c.4177G>A, c.1997C>T, c.1442delG, c.4636C>T, c.1747C>T, c.1748G>A, c.757C>T, c.877G>A, c.5126T>C, c.5396C>T, c.2960C[3>5] (std: c.2960C[5] alt: c.2962_2963insCC), c.904G>A, c.592C>T, c.835C>T, c.2554C>T, c.2542C>T, c.3460C>T, c.3408C[7>6] (std: c.3408C[6] alt: c.3414delC), c.2134A>G, c.2137G>A

Gene:

CAMTA1

NM ID:

NM_015215.2

Variant:

c.838delA

Gene：

CASK

NM ID：

NM_003688.3

Variant：

c.2041C>T, c.1641_1644delAACA, c.1609C>T, c.2074C>T, c.2129A>G, c.79C>T, c.2168A>G, c.67T[2>1] (std: c.67T[1] alt: c.68delT), c.2506-2A>G, c.845dupA, c.846C>G, c.2302+1G>A, c.316C>T, c.1915C>T, c.626T>C, c.1466G>A, c.1837C>T, c.1465C>T, c.763C>T, c.764G>A

Gene：

CBL

NM ID：

NM_005188.3

Variant：

c.1096-4_1096-1delAAAG, c.1096-2A>T, c.1096-1G>C, c.1096-1G>T, c.1100A>C, c.1111T>A, c.1111T>C, c.1144A>G, c.1150T>C, c.1166A>C, c.1186T>C, c.1228-2A>G, c.1259G>A

Gene：

CDKL5

NM ID：

NM_003159.2

Variant：

c.404-2A>G, c.404-1G>T, c.413C>T, c.400C>T, c.352C>T, c.380A>G, c.119C>T, c.453delG, c.578A>G, c.100-2A>G, c.587C>T, c.146-1G>A, c.602T>C, c.2046+1G>A, c.163_166delGAAA, c.2372A[3>4] (std: c.2374dupA alt: c.2374_2375insA), c.99+5G>A, c.65G>C, c.622C>T, c.175C>T, c.2047-1G>A,

c.2345C>A, c.2360_2363delAGAA, c.71A>G, c.744+1G>A, c.198TC[2>1] (std: c.198TC[1] alt: c.200_201delTC), c.73G>A, c.99+1G>T, c.554+1G>A, c.532C>T, c.533G>A, c.533G>C, c.514G>A, c.513C>A, c.745-1_745insT, c.211A>G, c.1006C>T, c.710T[4>3] (std: c.710T[3] alt: c.713delT), c.215T>C, c.766C>T, c.2152G>A, c.854G>A, c.2413C>T, c.59G>T, c.868C>T, c.2810G>A, c.786C>A, c.2596C>T, c.2593C>T, c.1152C>G, c.1246G>T, c.1245AG[2>1] (std: c.1245AG[1] alt: c.1247_1248delAG), c.1209AC[2>3] (std: c.1211_1212dupAC alt: c.1212_1213insAC), c.1675C>T, c.1110C[2>1] (std: c.1110C[1] alt: c.1111delC), c.1813C>T, c.1449_1452dupGACC, c.2635_2636delCT, c.1671dupA, c.1648C>T

Gene：

CHD2

NM ID：

NM_001271.3

Variant：

c.1809+1G>A, c.1719G>A, c.3454C>T, c.340C>T, c.3214C>T, c.947dupA, c.3937C>T, c.982C>T, c.1396C>T, c.3782G[6>7] (std: c.3787dupG alt: c.3787_3788insG), c.3782G>A, c.3782G>T, c.4165A[9>10] (std: c.4173dupA alt: c.4173_4174insA), c.4921C>T, c.4909C>T, c.4767CT[3>2] (std: c.4767CT[2] alt: c.4771_4772delCT), c.515A[8>7] (std: c.515A[7] alt: c.522delA), c.4948G[2>3] (std: c.4949dupG alt: c.4949_4950insG), c.2425C>T, c.3725A[11>10] (std: c.3725A[10] alt: c.3735delA), c.4971G>A, c.3521G>A, c.5035C>T, c.2699G>A, c.2636C>T

Gene：

CHD7

NM ID：

NM_017780.3

Variant：

c.2520G>A, c.2504_2508delATCTT, c.2572C>T, c.5706C>G, c.2238+1G>A, c.6937-2A>G, c.2839C>T, c.2836-2A>T, c.2613G>T, c.2919GGAG[2>1] (std: c.2919GGAG[1] alt: c.2923_2926delGGAG), c.2905_2906delAG, c.4353+3A>G, c.5534+1G>A, c.4353+1G>T, c.4379_4380delTA, c.5210+3A>G, c.2442+5G>C, c.6955C>T, c.5607+1G>A, c.8023G>T, c.3205C>T, c.4533+1G>A, c.8055G>A, c.2959C>T, c.5833C>T, c.3241A>T, c.3301T>C, c.3302G>A, c.4393C>T, c.7891C>T, c.4787A>G, c.4795C>T, c.3059T>C, c.5944G>T, c.2642dupA, c.7879C>T, c.3379-2A>C, c.3226A>G, c.5968C>T, c.2440C>T, c.6079C>T, c.7957C>T, c.6070C>T, c.2257C>T, c.934C>T, c.6995G>A, c.4318C>T, c.3106C>T, c.3082A>G, c.7803C>G, c.7132G>T, c.4593G>A, c.3571A[3>1] (std: c.3571A[1] alt: c.3572_3573delAA), c.5211-1G>C, c.4507G>T, c.4480C>T, c.6165_6166delGT, c.1808_1811delACAA, c.1803_1806delGAAA, c.6157C>T, c.7763A>G, c.6175G>T, c.3623T>A, c.1714C>T, c.4247C>G, c.6850C>T, c.8956G[7>8] (std: c.8962dupG alt: c.8962_8963insG), c.6757G>T, c.3655C>T, c.6148C>T, c.3768C>G, c.7252C>T, c.3420TG[2>1] (std: c.3420TG[1] alt: c.3422_3423delTG), c.7282C>T, c.7296G[2>1] (std: c.7296G[1] alt: c.7297delG), c.6199C>T, c.5405-17G>A, c.5458C>T, c.7285G>T, c.3881T>C, c.5405-7G>A, c.5050G>A, c.5029C>T, c.4015C>T, c.6287A>G, c.6272G>A, c.604C>T, c.3905T>C, c.6292C>T, c.5428C>T, c.5436C>A, c.282delT, c.538C>T, c.1480C>T, c.511C>T, c.1312C>T, c.1465C>T, c.469C>T, c.496C>T, c.1170T>G, c.2096G>A, c.2219A>G, c.2957+5G>A, c.3202-3T>G, c.3378+5G>C, c.3952T>C, c.4406A>G, c.4644+5G>A, c.5050+3A>T, c.5390G>T, c.5534G>A, c.6103+5G>C, c.6271T>C, c.6857G>C, c.7165-4A>G, c.8077-10T>A, c.232C>T, c.715C[4>3] (std: c.715C[3] alt: c.718delC), c.1919A[7>6] (std: c.1919A[6] alt:

c.1925delA), c.2988T[3>2] (std: c.2988T[2] alt: c.2990delT), c.3209delT, c.3490C>T, c.3526C>T, c.3934T[4>3] (std: c.3934T[3] alt: c.3937delT), c.4633T[2>1] (std: c.4633T[1] alt: c.4634delT), c.5210+2T>C, c.5405-2A>G, c.6243C>A, c.6892C>T, c.7522C[3>2] (std: c.7522C[2] alt: c.7524delC), c.77_78delAA, c.158C[3>2] (std: c.158C[2] alt: c.160delC), c.219dupT, c.222delG, c.235A>T, c.256C[2>1] (std: c.256C[1] alt: c.257delC), c.283G[3>2] (std: c.283G[2] alt: c.285delG), c.283G[3>1] (std: c.283G[1] alt: c.284_285delGG), c.317delA, c.360delC, c.378C>A, c.591T[5>4] (std: c.591T[4] alt: c.595delT), c.626G[2>1] (std: c.626G[1] alt: c.627delG), c.635delA, c.669_670insG, c.673C[2>1] (std: c.673C[1] alt: c.674delC), c.727C[3>2] (std: c.727C[2] alt: c.729delC), c.777C[4>3] (std: c.777C[3] alt: c.780delC), c.781delT, c.785delC, c.789TC[2>1] (std: c.789TC[1] alt: c.791_792delTC), c.799G>T, c.808delG, c.863A[3>2] (std: c.863A[2] alt: c.865delA), c.921_922delAG, c.959_960delAG, c.964_965delTT, c.1041C[4>3] (std: c.1041C[3] alt: c.1044delC), c.1141_1142delAT, c.1152delA, c.1167T[2>4] (std: c.1167T[4] alt: c.1168_1169insTT), c.1295delA, c.1315C[5>4] (std: c.1315C[4] alt: c.1319delC), c.1372TC[2>1] (std: c.1372TC[1] alt: c.1374_1375delTC), c.1387G[2>1] (std: c.1387G[1] alt: c.1388delG), c.1528delC, c.1543C[2>1] (std: c.1543C[1] alt: c.1544delC), c.1665+1G>C, c.1677G[2>1] (std: c.1677G[1] alt: c.1678delG), c.1681C[3>2] (std: c.1681C[2] alt: c.1683delC), c.1683_1684delCT, c.1684T[3>2] (std: c.1684T[2] alt: c.1686delT), c.1740delA, c.1792A[2>1] (std: c.1792A[1] alt: c.1793delA), c.1918delG, c.2032A[3>2] (std: c.2032A[2] alt: c.2034delA), c.2049delG, c.2117A[2>1] (std: c.2117A[1] alt: c.2118delA), c.2145delC, c.2157delA, c.2179T[2>1] (std: c.2179T[1] alt: c.2180delT), c.2236delC, c.2238delG, c.2244_2245delAC, c.2362C>T, c.2443-1delG, c.2464G>T, c.2490C>A, c.2499-2A>C, c.2517delG, c.2561A[6>5] (std: c.2561A[5] alt:

c.2566delA), c.2583A[3>2] (std: c.2583A[2] alt: c.2585delA), c.2619G[2>1] (std: c.2619G[1] alt: c.2620delG), c.2668T[3>1] (std: c.2668T[1] alt: c.2669_2670delTT), c.2706_2707delTC, c.2734C[2>1] (std: c.2734C[1] alt: c.2735delC), c.2739delT, c.2829delG, c.2836-2A>G, c.2858G[2>1] (std: c.2858G[1] alt: c.2859delG), c.2886T>G, c.2906G[3>2] (std: c.2906G[2] alt: c.2908delG), c.2916_2917delGT, c.2966delG, c.3023_3024delAT, c.3138delT, c.3164C[2>1] (std: c.3164C[1] alt: c.3165delC), c.3177T>G, c.3317A[2>1] (std: c.3317A[1] alt: c.3318delA), c.3320C[3>2] (std: c.3320C[2] alt: c.3322delC), c.3336delC, c.3338G[2>1] (std: c.3338G[1] alt: c.3339delG), c.3357G[3>2] (std: c.3357G[2] alt: c.3359delG), c.3376T[2>3] (std: c.3377dupT alt: c.3377_3378insT), c.3514_3515delGA, c.3546A[3>2] (std: c.3546A[2] alt: c.3548delA), c.3640C>T, c.3691A[3>2] (std: c.3691A[2] alt: c.3693delA), c.3734delT, c.3750delG, c.3805T[3>2] (std: c.3805T[2] alt: c.3807delT), c.4012_4013delGG, c.4086C[2>1] (std: c.4086C[1] alt: c.4087delC), c.4168C[4>3] (std: c.4168C[3] alt: c.4171delC), c.4182C[2>1] (std: c.4182C[1] alt: c.4183delC), c.4203_4204delTA, c.4224TG[2>1] (std: c.4224TG[1] alt: c.4226_4227delTG), c.4251A[3>2] (std: c.4251A[2] alt: c.4253delA), c.4256C[2>1] (std: c.4256C[1] alt: c.4257delC), c.4268A[3>2] (std: c.4268A[2] alt: c.4270delA), c.4295_4296delTG, c.4354-1G>A, c.4361_4362delAG, c.4393delC, c.4399G[4>3] (std: c.4399G[3] alt: c.4402delG), c.4424delA, c.4516G[2>3] (std: c.4517dupG alt: c.4517_4518insG), c.4525T[3>2] (std: c.4525T[2] alt: c.4527delT), c.4645-1G>C, c.4655T[2>3] (std: c.4656dupT alt: c.4656_4657insT), c.4665delT, c.4685C[2>1] (std: c.4685C[1] alt: c.4686delC), c.4731delA, c.4862G>A, c.5051-1G>A, c.5052T[3>2] (std: c.5052T[2] alt: c.5054delT), c.5074G>T, c.5095A[3>4] (std: c.5097dupA alt: c.5097_5098insA), c.5249A[2>1] (std: c.5249A[1] alt: c.5250delA), c.5498A[3>1] (std: c.5498A[1]

alt: c.5499_5500delAA), c.5572A[3>2] (std: c.5572A[2] alt: c.5574delA), c.5587C[2>1] (std: c.5587C[1] alt: c.5588delC), c.5592delC, c.5627C>G, c.5656delC, c.5678AG[2>1] (std: c.5678AG[1] alt: c.5680_5681delAG), c.5687delC, c.5768dupG, c.5768_5769delGC, c.5773A[4>3] (std: c.5773A[3] alt: c.5776delA), c.5798C[3>2] (std: c.5798C[2] alt: c.5800delC), c.5931G[2>1] (std: c.5931G[1] alt: c.5932delG), c.5989T[3>2] (std: c.5989T[2] alt: c.5991delT), c.6013A[6>5] (std: c.6013A[5] alt: c.6018delA), c.6043T[2>1] (std: c.6043T[1] alt: c.6044delT), c.6070delC, c.6179delT, c.6209delA, c.6217C>T, c.6222G[3>2] (std: c.6222G[2] alt: c.6224delG), c.6228delG, c.6268dupT, c.6303G[2>1] (std: c.6303G[1] alt: c.6304delG), c.6318AC[2>1] (std: c.6318AC[1] alt: c.6320_6321delAC), c.6326delT, c.6391T[3>2] (std: c.6391T[2] alt: c.6393delT), c.6403AG[2>1] (std: c.6403AG[1] alt: c.6405_6406delAG), c.6460C[2>1] (std: c.6460C[1] alt: c.6461delC), c.6502delC, c.6571delG, c.6666G[2>1] (std: c.6666G[1] alt: c.6667delG), c.6703A[3>2] (std: c.6703A[2] alt: c.6705delA), c.6712G>T, c.6716delA, c.6746delA, c.6835delG, c.6904delG, c.6997C[2>1] (std: c.6997C[1] alt: c.6998delC), c.7026C[2>1] (std: c.7026C[1] alt: c.7027delC), c.7047C>A, c.7106delT, c.7179C[2>1] (std: c.7179C[1] alt: c.7180delC), c.7180CT[3>2] (std: c.7180CT[2] alt: c.7184_7185delCT), c.7215A[5>4] (std: c.7215A[4] alt: c.7219delA), c.7230G[2>1] (std: c.7230G[1] alt: c.7231delG), c.7234G>T, c.7249delA, c.7316A[5>4] (std: c.7316A[4] alt: c.7320delA), c.7328delA, c.7344_7345delAA, c.7382T[3>2] (std: c.7382T[2] alt: c.7384delT), c.7399T[2>1] (std: c.7399T[1] alt: c.7400delT), c.7574T[4>3] (std: c.7574T[3] alt: c.7577delT), c.7648GA[2>1] (std: c.7648GA[1] alt: c.7650_7651delGA), c.7769delA, c.7781G[2>1] (std: c.7781G[1] alt: c.7782delG), c.7803delC, c.7875_7876delGA, c.7884_7885delTA, c.7893A[3>2] (std: c.7893A[2] alt: c.7895delA), c.7920T[3>1] (std: c.7920T[1] alt: c.7921_7922delTT), c.151C>T,

c.253C>T, c.325C[3>4] (std: c.327dupC alt: c.327_328insC), c.388C>T, c.406C>T, c.490C>T, c.502C>T, c.550C>T, c.562C>T, c.601C>T, c.608dupA, c.619C>T, c.664C>T, c.689C>G, c.718C>T, c.763C>T, c.804_805insAT, c.844C>T, c.889C>T, c.897C[4>5] (std: c.900dupC alt: c.900_901insC), c.925C>T, c.939T>G, c.995T>G, c.1024C>T, c.1036A>T, c.1078G>T, c.1093C>T, c.1095_1096insTC, c.1116dupA, c.1135C>T, c.1153C>T, c.1159C>T, c.1190C>G, c.1234C>T, c.1245G[3>4] (std: c.1247dupG alt: c.1247_1248insG), c.1310dupA, c.1366C>T, c.1488dupA, c.1510C>T, c.1576C>T, c.1610G>A, c.1610_1611insA, c.1615C>T, c.1645C>T, c.1689dupA, c.1729A[2>3] (std: c.1730dupA alt: c.1730_1731insA), c.1735C>T, c.1774C>T, c.1786C>T, c.1818_1819insAT, c.1864G[2>3] (std: c.1865dupG alt: c.1865_1866insG), c.1919A[7>8] (std: c.1925dupA alt: c.1925_1926insA), c.1933A[4>6] (std: c.1933A[6] alt: c.1936_1937insAA), c.1940C>G, c.1947A[7>8] (std: c.1953dupA alt: c.1953_1954insA), c.1972G>T, c.1973_1974insT, c.1983dupG, c.1984A[6>8] (std: c.1984A[8] alt: c.1989_1990insAA), c.1984A[6>7] (std: c.1989dupA alt: c.1989_1990insA), c.1990G>T, c.2096+2T>A, c.2096+2T>C, c.2097-1G>A, c.2114C>A, c.2181dupA, c.2196dupA, c.2254A>T, c.2311G>T, c.2374C>T, c.2433A[6>7] (std: c.2438dupA alt: c.2438_2439insA), c.2443-2A>T, c.2443-2A>G, c.2498+1G>T, c.2498+1_2498+2insT, c.2498+2T>C, c.2501C>T, c.2505T>A, c.2505T>G, c.2520G>C, c.2569A[3>4] (std: c.2571dupA alt: c.2571_2572insA), c.2575T[3>4] (std: c.2577dupT alt: c.2577_2578insT), c.2584A>T, c.2688C[2>3] (std: c.2689dupC alt: c.2689_2690insC), c.2724G>A, c.2737_2738insTC, c.2753G>A, c.2757dupG, c.2764C>T, c.2815G>T, c.2858G>A, c.2957+2T>C, c.2957+2T>G, c.2958-2A>T, c.2958-1G>A, c.3024T>G, c.3053_3054insA, c.3089A>G, c.3091T>G, c.3091T>C, c.3117dupA, c.3121T[2>3] (std: c.3122dupT alt: c.3122_3123insT), c.3169C>T, c.3173T>A,

c.3202-2A>G, c.3202-1G>A, c.3245C>A, c.3297G>A, c.3379-1G>A, c.3522+2T>C, c.3522+2T>G, c.3523-1G>C, c.3535C>T, c.3571A[3>4] (std: c.3573dupA alt: c.3573_3574insA), c.3616dupA, c.3641A>G, c.3646A>T, c.3654C>G, c.3726A[3>4] (std: c.3728dupA alt: c.3728_3729insA), c.3770T>G, c.3778+1G>A, c.3802G>T, c.3811G>T, c.3847C>T, c.3856dupA, c.3875T>C, c.3896T>C, c.3907A>T, c.3989+1G>A, c.3990-2A>G, c.3990-1G>C, c.3993C>G, c.4034G>A, c.4036C>T, c.4075dupA, c.4084T>C, c.4102_4103dupGC, c.4110T[3>4] (std: c.4112dupT alt: c.4112_4113insT), c.4113_4114insCA, c.4138dupA, c.4157C>G, c.4164G>A, c.4185G>C, c.4186-1G>A, c.4213C>T, c.4324_4325dupAT, c.4353+1G>A, c.4353+2T>C, c.4357_4358dupCA, c.4357C>T, c.4375G>T, c.4441A>T, c.4477dupC, c.4534-1G>A, c.4601G>A, c.4602G>A, c.4644+1G>A, c.4666C[2>3] (std: c.4667dupC alt: c.4667_4668insC), c.4722G[2>3] (std: c.4723dupG alt: c.4723_4724insG), c.4730C>G, c.4753G>T, c.4783C>T, c.4849G[2>3] (std: c.4850dupG alt: c.4850_4851insG), c.4850+1G>A, c.4850+2T>A, c.4851-2A>T, c.4851-2A>G, c.4853G>A, c.4854G>A, c.5050+1G>A, c.5068C[2>3] (std: c.5069dupC alt: c.5069_5070insC), c.5094dupG, c.5101C>T, c.5122C>T, c.5136G>A, c.5164_5165insC, c.5178_5179dupCT, c.5205dupT, c.5216T>G, c.5225T>A, c.5234T>C, c.5245A>T, c.5297C>G, c.5300+1G>T, c.5331C[2>3] (std: c.5332dupC alt: c.5332_5333insC), c.5355G>A, c.5402A>C, c.5418C>G, c.5434G>C, c.5435A>G, c.5444T>C, c.5453dupT, c.5534+1G>T, c.5539G>T, c.5548dupA, c.5562C[3>4] (std: c.5564dupC alt: c.5564_5565insC), c.5635A[2>3] (std: c.5636dupA alt: c.5636_5637insA), c.5665+1G>T, c.5666-2A>C, c.5668A>T, c.5676dupT, c.5706C>A, c.5709G>A, c.5752dupA, c.5782C>T, c.5791C>T, c.5895-2A>G, c.5898G>A, c.5908G>T, c.5909A[2>3] (std: c.5910dupA alt: c.5910_5911insA), c.5981G>A, c.5982G>A, c.6013A[6>7]

(std: c.6018dupA alt: c.6018_6019insA), c.6018_6019insG, c.6041dupA, c.6051T>A, c.6103+1G>A, c.6168C[2>3] (std: c.6169dupC alt: c.6169_6170insC), c.6196G>T, c.6316A>T, c.6322G>A, c.6322G>T, c.6397C>T, c.6473C>G, c.6492_6493dupTG, c.6520A>T, c.6526G>T, c.6620dupG, c.6745dupG, c.6766C>T, c.6775_6775+1insGT, c.6856G[2>3] (std: c.6857dupG alt: c.6857_6858insG), c.6884C>G, c.6888_6889insT, c.6937-1G>C, c.6991A>T, c.7075C>T, c.7141_7142insGT, c.7160C>A, c.7164+1G>A, c.7195C>T, c.7226T[2>3] (std: c.7227dupT alt: c.7227_7228insT), c.7276C>T, c.7302dupA, c.7367C>G, c.7400T>A, c.7422dupG, c.7425dupT, c.7441C>T, c.7447G>T, c.7451dupT, c.7454dupT, c.7456C>T, c.7485_7486dupGC, c.7592G[2>3] (std: c.7593dupG alt: c.7593_7594insG), c.7636G>T, c.7652C[4>5] (std: c.7655dupC alt: c.7655_7656insC), c.7824T>A, c.7884_7885dupTA, c.7933G>T, c.8016G>A, c.8077-1G>A, c.8093C>A, c.8956_8957insA

Gene：

CHD8

NM ID：

NM_001170629.1

Variant：

c.7105A[8>9] (std: c.7112dupA alt: c.7112_7113insA), c.4871G>A, c.5389C>T, c.4009C>T, c.1690C>T, c.5500C>T, c.3519-2A>G, c.3724C>T, c.3725G>A, c.634C>T, c.3562C>T

Gene：

COL11A1

NM ID：

NM_001854.3

Variant：

c.3978+1G>A, c.3168+1G>T, c.3816+5G>A, c.1630-2delA, c.3816+2_3816+3insT, c.3816+1G>C, c.3816+1G>A, c.2754+5G>A, c.652-1G>C, c.4547G>T, c.4554+1G>C, c.4538G>A, c.3168+5G>A, c.2808+1G>C, c.2043+1G>T, c.1845+1G>A, c.1191delT, c.4554+1G>A, c.4519-2A>G, c.2043+1G>A, c.1681C>T, c.1245+1G>A

Gene：

COL1A1

NM ID：

NM_000088.3

Variant：

c.454C[5>6] (std: c.458dupC alt: c.458_459insC), c.436C[6>5] (std: c.436C[5] alt: c.441delC), c.427C[6>7] (std: c.432dupC alt: c.432_433insC), c.671G>T, c.391C>T, c.658C>T, c.661G>T, c.643-2A>G, c.381C[6>5] (std: c.381C[5] alt: c.386delC), c.358C>T, c.2445C[6>5] (std: c.2445C[5] alt: c.2450delC), c.563_564delGTinsA, c.334-9A>G, c.262G>T, c.3421C>T, c.572G>C, c.3207+1G>A, c.573C[6>7] (std: c.578dupC alt: c.578_579insC), c.579delT, c.1200+1G>A, c.3118G>A, c.1718C[3>2] (std: c.1718C[2] alt: c.1720delC), c.1084G>A, c.1192G>T, c.1696G>A, c.2362G>A, c.1081C>T, c.3567delT, c.1678G>T, c.1678G>A, c.599G>T, c.1821+1G>T, c.1821+1G>C, c.1821+1G>A, c.2028+2T>G, c.1768-1G>A, c.595C>T, c.2829+1G>A, c.3790A>G, c.3806G>A, c.1792C>T, c.1614+1G>C, c.1614+1G>A, c.2128-1G>C, c.2128-1G>A, c.1777G>A, c.3531+1G>A, c.2010delT, c.3360delT, c.3135delT, c.608G>T, c.3817G>T, c.3065G>T, c.3065G>C, c.3061G>T, c.3301G>T, c.3815G>T, c.1354-12G>A, c.3824G>A, c.3162delT, c.2596G>A, c.959G>A, c.3226G>A, c.4321G>C, c.3505G>A, c.4316T>C, c.958-1G>A, c.3652G>A, c.697-1G>C, c.1012G>A, c.3647A>G, c.1299+5G>A, c.985G>C, c.1299+1G>A, c.976G>C,

c.4248+1G>A, c.1273G>A, c.3235G>A, c.2062C>T, c.2667+1G>A, c.2644C>T, c.2127+2T>A, c.3495delT, c.3076C>T, c.3490C[5>4] (std: c.3490C[4] alt: c.3494delC), c.2155G>A, c.3910C>T, c.1369C>T, c.2161C>T, c.3470G>A, c.3479G>A, c.769G>A, c.2784delT, c.2110G>A, c.1875+1G>C, c.3261+1G>A, c.3040C>T, c.2452-1G>C, c.2452-1G>A, c.1103G>C, c.2569G>T, c.3451G>A, c.2299G>A, c.994G>A, c.4237G>A, c.1247G>A, c.1099C>T, c.2461G>A, c.1002+2T>C, c.1588G>A, c.3003C[6>5] (std: c.3003C[5] alt: c.3008delC), c.2073delT, c.3099+1G>A, c.3936G>A, c.2680C[5>4] (std: c.2680C[4] alt: c.2684delC), c.1138G>A, c.2685delT, c.1243C>T, c.1201-1G>A, c.2089C>T, c.1155+3_1155+6delAAGT, c.1462G>A, c.2523delT, c.1122C[6>5] (std: c.1122C[5] alt: c.1127delC), c.757C>T, c.1414C>T, c.751G>T, c.751-2A>G, c.751-1G>A, c.2533G>A, c.1405C>T, c.934C>T, c.1121G>C, c.4375G>C, c.4364G>T, c.4166T>C, c.3853G>C, c.3380G>A, c.3290G>T, c.904-9G>A, c.671G>A, c.281T>A, c.4391T>C, c.4321G>T, c.4163T>G, c.3515G>A, c.3496G>T, c.3263G>A, c.3208G>A, c.3073G>A, c.2937+4A>C, c.2605G>T, c.2515G>A, c.2317G>T, c.1976G>C, c.1966G>A, c.1777G>T, c.1579G>T, c.1102G>A, c.1066G>T, c.957+5G>A, c.788G>T, c.760G>A, c.696+3A>C, c.588+5G>A, c.588+4A>T, c.543+5G>A, c.64G>C, c.3825G>A, c.3535C[6>7] (std: c.3540dupC alt: c.3540_3541insC), c.3207+1_3207+2delGT, c.3022C[5>4] (std: c.3022C[4] alt: c.3026delC), c.2991delT, c.2775delT, c.2419C[6>5] (std: c.2419C[5] alt: c.2424delC), c.1812delT, c.1452delT, c.1375C[5>4] (std: c.1375C[4] alt: c.1379delC), c.1290G[3>2] (std: c.1290G[2] alt: c.1292delG), c.1266C[4>3] (std: c.1266C[3] alt: c.1269delC), c.1258C[4>3] (std: c.1258C[3] alt: c.1261delC), c.1128delT, c.1057-1G>A, c.517G>T, c.148C>T, c.1A>G, c.4308C[2>1] (std: c.4308C[1] alt: c.4309delC), c.4156A>T, c.4054delC, c.4051C>T, c.3987delC, c.3766delG, c.3762C[4>3] (std: c.3762C[3] alt:

c.3765delC), c.3747G[3>2] (std: c.3747G[2] alt: c.3749delG), c.3718C>T, c.3637G[2>1] (std: c.3637G[1] alt: c.3638delG), c.3589_3590delGA, c.3583G[2>1] (std: c.3583G[1] alt: c.3584delG), c.3578GC[2>1] (std: c.3578GC[1] alt: c.3580_3581delGC), c.3559G[2>1] (std: c.3559G[1] alt: c.3560delG), c.3535C[6>5] (std: c.3535C[5] alt: c.3540delC), c.3477delT, c.3450dupT, c.3401C[2>1] (std: c.3401C[1] alt: c.3402delC), c.3370G[2>1] (std: c.3370G[1] alt: c.3371delG), c.3328delC, c.3313delA, c.3309delC, c.3273C[5>4] (std: c.3273C[4] alt: c.3277delC), c.3262G>T, c.3255C[4>3] (std: c.3255C[3] alt: c.3258delC), c.3241delG, c.3208-1G>A, c.3201delT, c.3114delG, c.3079delG, c.3064G[2>1] (std: c.3064G[1] alt: c.3065delG), c.3027delT, c.2785G[2>1] (std: c.2785G[1] alt: c.2786delG), c.2770C[5>4] (std: c.2770C[4] alt: c.2774delC), c.2731G[2>1] (std: c.2731G[1] alt: c.2732delG), c.2646_2647delAG, c.2614-1G>A, c.2608C[5>4] (std: c.2608C[4] alt: c.2612delC), c.2518C[5>4] (std: c.2518C[4] alt: c.2522delC), c.2451delT, c.2443G[2>1] (std: c.2443G[1] alt: c.2444delG), c.2425G[2>3] (std: c.2426dupG alt: c.2426_2427insG), c.2410G>T, c.2389G[2>1] (std: c.2389G[1] alt: c.2390delG), c.2364C[3>2] (std: c.2364C[2] alt: c.2366delC), c.2329delG, c.2319C[3>2] (std: c.2319C[2] alt: c.2321delC), c.2266TC[2>1] (std: c.2266TC[1] alt: c.2268_2269delTC), c.2248C[3>2] (std: c.2248C[2] alt: c.2250delC), c.2236-1G>A, c.2208delT, c.2096C[2>1] (std: c.2096C[1] alt: c.2097delC), c.2085_2086delTC, c.2083G[2>1] (std: c.2083G[1] alt: c.2084delG), c.2068C[5>4] (std: c.2068C[4] alt: c.2072delC), c.2033AG[3>2] (std: c.2033AG[2] alt: c.2037_2038delAG), c.1993G[2>1] (std: c.1993G[1] alt: c.1994delG), c.1983+2T>G, c.1944delT, c.1930-2A>C, c.1900C>T, c.1897G>T, c.1889AG[3>2] (std: c.1889AG[2] alt: c.1893_1894delAG), c.1885G[2>1] (std: c.1885G[1] alt: c.1886delG), c.1876-2delA, c.1866delT, c.1861C[5>4] (std: c.1861C[4] alt: c.1865delC), c.1858G>T, c.1813G[2>1] (std: c.1813G[1] alt:

c.1814delG), c.1786G[2>1] (std: c.1786G[1] alt: c.1787delG), c.1668delT, c.1662C[6>5] (std: c.1662C[5] alt: c.1667delC), c.1660G[2>1] (std: c.1660G[1] alt: c.1661delG), c.1629C[3>2] (std: c.1629C[2] alt: c.1631delC), c.1608delT, c.1485C[3>1] (std: c.1485C[1] alt: c.1486_1487delCC), c.1263G[3>2] (std: c.1263G[2] alt: c.1265delG), c.1248C[4>3] (std: c.1248C[3] alt: c.1251delC), c.1207C[2>1] (std: c.1207C[1] alt: c.1208delC), c.1201_1202delGG, c.1200+1_1200+2delGT, c.1200+1G>C, c.1155+2T>G, c.1128_1129insC, c.1069C[4>3] (std: c.1069C[3] alt: c.1072delC), c.1065delT, c.982_983dupAC, c.898C>T, c.807T[3>2] (std: c.807T[2] alt: c.809delT), c.804+2_804+3delTG, c.775delG, c.750+2T>A, c.697G[4>3] (std: c.697G[3] alt: c.700delG), c.695delA, c.670G[2>1] (std: c.670G[1] alt: c.671delG), c.669delA, c.630delG, c.610C[5>4] (std: c.610C[4] alt: c.614delC), c.567C[3>2] (std: c.567C[2] alt: c.569delC), c.532C[2>1] (std: c.532C[1] alt: c.533delC), c.481C[4>3] (std: c.481C[3] alt: c.484delC), c.484C>T, c.268G>T, c.253G>T, c.243C[4>3] (std: c.243C[3] alt: c.246delC), c.189C>A, c.155TG[2>1] (std: c.155TG[1] alt: c.157_158delTG), c.144delT, c.143dupA, c.103+2T>C, c.67C>T, c.38T>G, c.4021C>T, c.4009G>T, c.3975G>A, c.3974G>A, c.3969dupT, c.3925C>T, c.3897C>A, c.3838C>T, c.3815-2A>G, c.3814+2T>C, c.3814+2T>G, c.3814+1G>C, c.3807G>A, c.3698C[2>3] (std: c.3699dupC alt: c.3699_3700insC), c.3607C>T, c.3532-1G>A, c.3424-1G>C, c.3385C>T, c.3370G>T, c.3349C>T, c.3285dupC, c.3262-1G>A, c.3208-1G>C, c.3207+2T>C, c.3207+1G>C, c.3165C[4>5] (std: c.3168dupC alt: c.3168_3169insC), c.3100-1G>A, c.3045+1G>T, c.3045+1G>A, c.3003C[6>7] (std: c.3008dupC alt: c.3008_3009insC), c.2938-1G>A, c.2869C>T, c.2668-1G>A, c.2594dupG, c.2560-1G>A, c.2559+2T>A, c.2559+1G>A, c.2550dupT, c.2464C>T, c.2451+1G>A, c.2451+1G>T, c.2451_2451+1insT, c.2445C[6>7] (std: c.2450dupC alt: c.2450_2451insC),

c.2398-1G>A, c.2397_2397+1insC, c.2344-1G>A, c.2235+1G>A, c.2228G>T, c.2227G>A, c.2075G>C, c.2065G>A, c.2032G>T, c.2028+1G>A, c.2013C[4>5] (std: c.2016dupC alt: c.2016_2017insC), c.1984-1G>A, c.1983+1G>A, c.1930-1G>A, c.1930-1G>C, c.1930-2A>G, c.1929+1G>C, c.1929+1G>A, c.1916C[5>6] (std: c.1920dupC alt: c.1920_1921insC), c.1876-1G>A, c.1876-1G>C, c.1876-2A>G, c.1875+1G>A, c.1767+2T>C, c.1718_1719insAC, c.1705G>C, c.1669-1G>C, c.1669-2A>G, c.1662C[6>7] (std: c.1667dupC alt: c.1667_1668insC), c.1615-1G>C, c.1615-1G>T, c.1516-1G>C, c.1516-1G>T, c.1516-2A>G, c.1461+1G>C, c.1461+1G>A, c.1375C[5>6] (std: c.1379dupC alt: c.1379_1380insC), c.1354-1G>A, c.1354-1G>T, c.1354-2A>G, c.1300-1G>A, c.1299+1G>C, c.1200+1G>T, c.1156-1G>A, c.1155+1G>A, c.1155+1G>C, c.1057-2A>T, c.1003-1G>C, c.1003-2A>G, c.983_984dupCT, c.967G>T, c.957+2T>A, c.957+1G>T, c.859-1G>A, c.859-2A>C, c.807T[3>4] (std: c.809dupT alt: c.809_810insT), c.766C[3>4] (std: c.768dupC alt: c.768_769insC), c.750+1G>A, c.750+1G>C, c.750+1G>T, c.697-1G>A, c.697-2A>T, c.642+5G>A, c.642+1G>A, c.642+1G>T, c.589-1G>A, c.589-2A>G, c.537C[4>5] (std: c.540dupC alt: c.540_541insC), c.370-1G>A, c.334-2A>G, c.333+2T>C, c.333+1G>A, c.333+1G>T, c.299-1G>A, c.299-1G>C, c.299-2A>G, c.141C>A, c.103+1G>A, c.103+1G>C, c.2T>G, c.2T>C

Gene：

COL1A2

NM ID：

NM_000089.3

Variant：

c.595-2A>G, c.226-2A>G, c.1557+3A>G, c.279+2T>C, c.432+1G>A, c.432+1G>T, c.432+1_432+4delGTAA, c.1774G>A, c.1981G>T, c.1801G>A,

c.1991G>A, c.2314G>A, c.2332G>A, c.1378G>A, c.739G>T, c.2673+1G>A, c.407G>A, c.2387G>C, c.2215G>C, c.767G>A, c.767G>T, c.389G>A, c.2242G>A, c.1045G>T, c.577G>A, c.2260G>T, c.2674-3T>G, c.3305G>C, c.2989G>A, c.1127G>T, c.2565+1G>A, c.298G>A, c.1658G>A, c.910G>A, c.821G>A, c.2045G>A, c.371G>A, c.875G>A, c.1503+1G>C, c.874G>A, c.920G>A, c.1576G>A, c.2835+1G>A, c.2133+6T>A, c.838G>A, c.353G>T, c.1072G>A, c.3034G>A, c.1270G>A, c.1197+5G>A, c.1171G>A, c.1342G>C, c.2845G>A, c.1252G>A, c.2746G>A, c.1009G>A, c.2512G>A, c.946G>A, c.2405G>T, c.2531G>C, c.956G>A, c.2503G>A, c.964G>A, c.982G>A, c.1208G>A, c.1863G>A, c.432+5G>A, c.1298G>A, c.3527G>T, c.12T>G, c.3773G>A, c.694-2A>C, c.486+1G>T, c.486+1G>C, c.693+1G>A, c.792+1G>A, c.792+1G>T, c.792+2T>A, c.792+2T>C, c.793G>C, c.1099G>T, c.1162G>C, c.1316G>A, c.1361G>T, c.1406G>C, c.1433G>A, c.1532G>A, c.1937G>T, c.2027G>T, c.2188G>T, c.2297G>T, c.2386G>A, c.3008G>A, c.3089G>C, c.3105+2T>C, c.3304G>T, c.3350A>G, c.3487T>C, c.3944A>T, c.3952dupT

Gene：

COL2A1

NM ID：

NM_001844.4

Variant：

c.2355+5G>A, c.2355+1G>A, c.2353C>T, c.1887+1G>A, c.762+1G>A, c.2428G>T, c.1861G>A, c.719G>A, c.519delT, c.2485C[6>5] (std: c.2485C[5] alt: c.2490delC), c.2491G>A, c.1996-9G>A, c.258C>A, c.2024G>A, c.115C>T, c.156C>A, c.486C[6>5] (std: c.486C[5] alt: c.491delC), c.1357G>A, c.1957C>T, c.3311G>A, c.1941+1G>A, c.1034G>A, c.3121G>A, c.647G>A, c.1833+1G>A, c.2710C>T, c.619G>A, c.2673C[6>7] (std: c.2678dupC alt: c.2678_2679insC),

c.625C>T, c.1331G>T, c.3106C>T, c.2376C[6>5] (std: c.2376C[5] alt: c.2381delC), c.1510G>A, c.2382delT, c.3138delT, c.3624delT, c.3597+1G>A, c.3139G>A, c.1123-1G>A, c.1924G>A, c.1221+1G>A, c.3589G>A, c.2659C>T, c.1177G>A, c.3149G>A, c.905C>T, c.908G>A, c.823C>T, c.85C>T, c.2159G>A, c.1537G>A, c.882C[2>1] (std: c.882C[1] alt: c.883delC), c.3508G>A, c.2059G>A, c.3574C>T, c.1546G>A, c.2536G>A, c.2214C[6>7] (std: c.2219dupC alt: c.2219_2220insC), c.655G>A, c.655-1G>C, c.3714C>A, c.1597C>T, c.2862C>T, c.4172A>G, c.1693C>T, c.2101C>T, c.2833G>A, c.1636G>A, c.2965C>T, c.2818C>T, c.2808C[6>5] (std: c.2808C[5] alt: c.2813delC), c.2794C>T, c.3355G[2>1] (std: c.3355G[1] alt: c.3356delG), c.2814delT, c.4074+2_4074+3delTG, c.3903C[4>3] (std: c.3903C[3] alt: c.3906delC), c.3872_3873delCT, c.3862CT[2>1] (std: c.3862CT[1] alt: c.3864_3865delCT), c.3778_3779delAA, c.3619C[5>4] (std: c.3619C[4] alt: c.3623delC), c.3495delT, c.3488delG, c.3419G>C, c.3400G[2>1] (std: c.3400G[1] alt: c.3401delG), c.3324C[2>1] (std: c.3324C[1] alt: c.3325delC), c.3228delT, c.3010C[3>2] (std: c.3010C[2] alt: c.3012delC), c.2997C[3>2] (std: c.2997C[2] alt: c.2999delC), c.2972AG[3>2] (std: c.2972AG[2] alt: c.2976_2977delAG), c.2854C[5>4] (std: c.2854C[4] alt: c.2858delC), c.2788G[2>1] (std: c.2788G[1] alt: c.2789delG), c.2785A[3>2] (std: c.2785A[2] alt: c.2787delA), c.2751delT, c.2673C[6>5] (std: c.2673C[5] alt: c.2678delC), c.2536G[4>3] (std: c.2536G[3] alt: c.2539delG), c.2474GA[3>2] (std: c.2474GA[2] alt: c.2478_2479delGA), c.2471delG, c.2302-1G>A, c.2261AG[2>1] (std: c.2261AG[1] alt: c.2263_2264delAG), c.2077_2078delGG, c.1968G[3>2] (std: c.1968G[2] alt: c.1970delG), c.1963G>T, c.1927C[5>4] (std: c.1927C[4] alt: c.1931delC), c.1908delT, c.1828delG, c.1818C[4>3] (std: c.1818C[3] alt: c.1821delC), c.1731T[2>1] (std: c.1731T[1] alt: c.1732delT), c.1674delA, c.1522G>T, c.1415A[2>1] (std: c.1415A[1] alt:

c.1416delA), c.1310G[4>3] (std: c.1310G[3] alt: c.1313delG), c.1171C[2>1] (std: c.1171C[1] alt: c.1172delC), c.1095delT, c.1032delA, c.994AG[2>1] (std: c.994AG[1] alt: c.996_997delAG), c.938C[4>3] (std: c.938C[3] alt: c.941delC), c.933delT, c.793delG, c.744delT, c.721C[4>3] (std: c.721C[3] alt: c.724delC), c.572delC, c.510delT, c.309G[3>2] (std: c.309G[2] alt: c.311delG), c.292G[3>2] (std: c.292G[2] alt: c.294delG), c.237delG, c.164GT[2>1] (std: c.164GT[1] alt: c.166_167delGT), c.145C[2>1] (std: c.145C[1] alt: c.146delC), c.4074+1G>T, c.3886+2T>C, c.3886+1G>A, c.3886+1G>T, c.3878G>A, c.3709C>T, c.3637C[5>6] (std: c.3641dupC alt: c.3641_3642insC), c.3596dupC, c.3463G[2>3] (std: c.3464dupG alt: c.3464_3465insG), c.3419G>A, c.3280C>T, c.3132C[6>7] (std: c.3137dupC alt: c.3137_3138insC), c.3112-1G>A, c.3111+1G>T, c.3003+1G>A, c.3003+1G>T, c.2896-1G>A, c.2869G>T, c.2839C>T, c.2719C[5>6] (std: c.2723dupC alt: c.2723_2724insC), c.2714T[2>3] (std: c.2715dupT alt: c.2715_2716insT), c.2653G>T, c.2625+1G>T, c.2518-1G>A, c.2493dupA, c.2473G>T, c.2467G>T, c.2436dupA, c.2194-1G>A, c.2193+2T>C, c.2095-1G>A, c.2094+1G>A, c.2050-1G>C, c.2049+1G>A, c.1996-1G>A, c.1942G[2>3] (std: c.1943dupG alt: c.1943_1944insG), c.1942-2A>G, c.1779G[5>6] (std: c.1783dupG alt: c.1783_1784insG), c.1777C>T, c.1735-1G>A, c.1714C>T, c.1600G[3>4] (std: c.1602dupG alt: c.1602_1603insG), c.1528-2A>G, c.1527+1G>C, c.1399_1400dupCA, c.1366-2A>T, c.1030C>T, c.870+1G>A, c.756dupT, c.654+1G>A, c.581C[3>4] (std: c.583dupC alt: c.583_584insC), c.556G>T, c.504C[6>7] (std: c.509dupC alt: c.509_510insC), c.430-1G>C, c.403G[4>5] (std: c.406dupG alt: c.406_407insG), c.293-1G>A, c.192C>A, c.123T>A, c.3G>T, c.1962C>T

Gene：

COL4A1

NM ID：

NM_001845.4

Variant：

c.3556G>A, c.2458+1G>A, c.2494G>A, c.2086G>A, c.3715G>A, c.2662G>A, c.1493G>A, c.2228G>T, c.2317G>A, c.3307G>A, c.2263G>A

Gene：

COL9A2

NM ID：

NM_001852.3

Variant：

c.186G>A

Gene：

COL9A3

NM ID：

NM_001853.3

Variant：

c.148-1G>A

Gene：

COMP

NM ID：

NM_000095.2

Variant：

c.874T>C, c.1153G>A, c.1201G>A, c.1405GAC[5>6] (std: c.1417_1419dupGAC alt: c.1419_1420insGAC), c.1405GAC[5>4] (std: c.1405GAC[4] alt: c.1417_1419delGAC), c.1754C>T, c.1760A>G, c.1318G>A, c.1368GGA[2>1] (std: c.1368GGA[1] alt: c.1371_1373delGGA), c.2155G>A, c.1444G>A,

c.811G>C, c.2152C>T, c.1502G>A, c.1675G>A

Gene：

CREBBP

NM ID：

NM_004380.2

Variant：

c.3836+1G>A, c.3832G>A, c.1237C>T, c.1270C>T, c.4398T>A, c.3779+1G>A, c.3982+1G>A, c.3369+1G>T, c.1941+1G>A, c.4439A>G, c.4133+1G>A, c.2302C>T, c.6324C>A, c.5831C[7>8] (std: c.5837dupC alt: c.5837_5838insC), c.4672C>T, c.5036CCT[2>1] (std: c.5036CCT[1] alt: c.5039_5041delCCT), c.1108C>T, c.4492C>T, c.4078C>T, c.5356C>T, c.1447C>T, c.5639AG[2>1] (std: c.5639AG[1] alt: c.5641_5642delAG), c.5614A>G, c.5599C>T, c.5600G>A, c.5602C>T, c.5603G>A

Gene：

CTCF

NM ID：

NM_006565.3

Variant：

c.942CA[5>6] (std: c.950_951dupCA alt: c.951_952insCA), c.1699C>T, c.615_618delGAAA, c.1670_1674delGTTCT

Gene：

CTNNB1

NM ID：

NM_001904.3

Variant：

c.268C>T, c.274C>T, c.283C>T, c.1759C>T, c.337C>T, c.1923AG[2>1] (std:

c.1923AG[1] alt: c.1925_1926delAG), c.1981C>T, c.1543C>T, c.1041_1044delATCT, c.998dupA, c.1494dupA, c.999C>A, c.999C>G, c.1420C>T, c.1603C>T

Gene：

DNM1

NM ID：

NM_004408.2

Variant：

c.127G>A, c.134G>A, c.139G>A, c.1190G>A, c.709C>T, c.1075G>A

Gene：

DYNC1H1

NM ID：

NM_001376.4

Variant：

c.917A>G, c.4868G>A, c.4700G>A, c.6122T>C, c.751C>T, c.9754AAG[3>2] (std: c.9754AAG[2] alt: c.9760_9762delAAG), c.10151G>A, c.3278T>C, c.1792C>T, c.3370C>T, c.10973G>A, c.6994C>T, c.2327C>T, c.10031G>A, c.10573C>T, c.5885G>A, c.5884C>T

Gene：

DYRK1A

NM ID：

NM_001396.3

Variant：

c.951+1_951+4delGTAA, c.932C>T, c.923T>C, c.358C>T, c.349C>T, c.447A[6>7] (std: c.452dupA alt: c.452_453insA), c.476dupA, c.665-11_665-7delTTCTC, c.1309C>T, c.1159C>T, c.572_575delAGAA, c.569_572delTAAA, c.685_686delAT,

c.883C>T, c.1405C>T, c.1488C[4>3] (std: c.1488C[3] alt: c.1491delC), c.691C>T, c.1400G>A, c.860A>T, c.1399C>T, c.1639C>T, c.613C>T, c.787C>T, c.763C>T

Gene：

EBP

NM ID：

NM_006579.2

Variant：

c.329_332dupGATA, c.329G>A, c.328C>T, c.439C>T, c.238G>A, c.141G>T, c.204G>T, c.203G>A, c.187C>T

Gene：

EFNB1

NM ID：

NM_004429.4

Variant：

c.409A>G, c.453TG[3>4] (std: c.457_458dupTG alt: c.458_459insTG), c.451G>A, c.161C>T, c.561delC, c.196C>T, c.612CT[2>1] (std: c.612CT[1] alt: c.614_615delCT)

Gene：

EFTUD2

NM ID：

NM_004247.3

Variant：

c.1058+1G>A, c.1732C>T, c.702+1G>T, c.1060C>T, c.784C>T, c.2694GTCT[2>1] (std: c.2694GTCT[1] alt: c.2698_2701delGTCT), c.2770C>T

Gene：

EHMT1

NM ID：

NM_024757.4

Variant：

c.2873TTCT[2>1] (std: c.2873TTCT[1] alt: c.2877_2880delTTCT), c.2426C>T, c.2712+1G>A, c.35G[6>7] (std: c.40dupG alt: c.40_41insG), c.508C>T, c.2516G>T, c.673C>T, c.3589C>T, c.1059AG[2>1] (std: c.1059AG[1] alt: c.1061_1062delAG), c.736C>T, c.1647+2T>C, c.3122TC[3>2] (std: c.3122TC[2] alt: c.3126_3127delTC)

Gene：

EP300

NM ID：

NM_001429.3

Variant：

c.102CT[3>1] (std: c.102CT[1] alt: c.104_107delCTCT), c.3857A>G, c.3163C>T, c.3684_3687delAGAA, c.2660C>T, c.4783T>G, c.4763T>C, c.4954_4957dupATGT, c.4933C>T

Gene：

ERF

NM ID：

NM_006494.2

Variant：

c.266A>G, c.1201_1202delAA, c.547C>T, c.566_567delGT, c.619C>T, c.891_892delAG, c.256C>T

Gene：

FBN1

NM ID：

NM_000138.4

Variant：

c.7205-2A>G, c.7205-1G>A, c.2806C>T, c.1714+1G>T, c.3964+1G>A, c.2168-2A>G, c.2168-1G>T, c.6487G>T, c.1693C>T, c.2180G>A, c.759C[4>3] (std: c.759C[3] alt: c.762delC), c.7253G>A, c.1679G>T, c.772C>T, c.5296+5G>A, c.1670G>A, c.7240C>T, c.6453C>T, c.347-2A>G, c.4748-1G>A, c.1606C>T, c.1601G>A, c.4096G>A, c.1665C>A, c.3886T>C, c.5671G>C, c.5666G>A, c.6628T>C, c.1643A>T, c.5560C>T, c.3839-1G>T, c.6388G>A, c.2201G>A, c.3902G>T, c.4043G>A, c.3596A>G, c.3851G>A, c.4337-2A>G, c.4988G>T, c.6393C>G, c.4955G>A, c.4747+5G>T, c.364C>T, c.3855C[2>1] (std: c.3855C[1] alt: c.3856delC), c.5015G>A, c.5014T>G, c.442+1G>A, c.1633C>T, c.440A[2>1] (std: c.440A[1] alt: c.441delA), c.6694T>C, c.434G>A, c.433T>C, c.439C>T, c.5626T>C, c.5021G>A, c.2216G>A, c.6689G>A, c.4348T>C, c.401G>A, c.6610T>C, c.6609_6610delAT, c.1468+5G>A, c.3623G>T, c.5905delA, c.6425G>A, c.1837+5G>A, c.5284G>A, c.2581C>T, c.1837+1G>T, c.5368C>T, c.5372G>A, c.2237A>G, c.5371T>C, c.2243G>A, c.6418G>A, c.2242T>C, c.1849T>C, c.6446A>G, c.3977G>A, c.5801G>A, c.1468+2T>C, c.7003C>T, c.6740-1G>A, c.4781G>A, c.6650G>A, c.5422+1G>A, c.2539+1G>A, c.3973G>T, c.5065+1G>T, c.5065+1G>C, c.5065+1G>A, c.1462T>G, c.1462T>C, c.1909T>C, c.5593_5594dupTG, c.811T>G, c.1426T>G, c.1426T>C, c.4786C>T, c.915A[3>2] (std: c.915A[2] alt: c.917delA), c.2495G>A, c.6431A>G, c.6997+1G>A, c.5918-2A>G, c.6379+1G>A, c.1787G>A, c.1427G>A, c.5918-1G>C, c.5919dupT, c.1786T>G, c.2559C>A, c.2627G>A, c.6739+1G>A, c.6616+1G>A, c.1879C>T, c.8038C>T, c.5874C>A, c.1904A>G, c.2227C>T, c.3725G>A, c.4388A>G, c.6751T>C, c.6751T>A, c.6164-2A>G, c.6752G>A,

c.1868G>T, c.3838G>A, c.6325C>T, c.6164-1G>A, c.2563C>T, c.4367G>A, c.247+1G>A, c.5788+5G>A, c.2669G>A, c.2673A>G, c.5683T>C, c.2562G>A, c.5839T>C, c.5840G>A, c.6661T>C, c.6037+2T>C, c.5345G>A, c.6871+1G>A, c.6508T>C, c.6339T>G, c.7342T>C, c.4406G>C, c.5431G>T, c.5431G>A, c.5788G>C, c.5434T>C, c.6037+1G>A, c.7339G>A, c.6331T>C, c.7532G>A, c.6583G>T, c.1481G>A, c.5863C>T, c.6332G>A, c.4505G>A, c.526C>T, c.5437C>T, c.5788+1G>T, c.6658C>T, c.1838-2A>G, c.1949GT[5>4] (std: c.1949GT[4] alt: c.1957_1958delGT), c.2438C>G, c.2489G>C, c.2433C>A, c.529T>C, c.5783G>A, c.7531T>C, c.1817C>A, c.7015T>C, c.530G>A, c.7402T>C, c.1948C>T, c.2639G>A, c.8020T>C, c.2248T>C, c.6169C>T, c.7498T>C, c.2638G>A, c.7399C>T, c.4531T>C, c.2419+1G>A, c.5309G>A, c.6806T>C, c.1766A>G, c.239G>A, c.7606G>A, c.4532G>A, c.5699G>A, c.7453+1G>A, c.6354C>T, c.6354C>G, c.1148-2A>G, c.5330G>A, c.4538G>A, c.6049T>C, c.7376G>A, c.3794G>C, c.5512G>T, c.2306G>A, c.5513G>A, c.5504G>A, c.3761G>A, c.7409G>A, c.478T>C, c.1961-1G>A, c.200G>T, c.7330+1G>A, c.3778G>T, c.7410C>G, c.2647T>C, c.7364G>A, c.1759T>G, c.7605C>G, c.7605C>A, c.4205G>A, c.4429G>T, c.6953G>A, c.203G>T, c.203G>A, c.2977T>C, c.2261A>G, c.2645C>T, c.2114-2A>G, c.2114-2A>C, c.3337+1G>A, c.2369G>A, c.2415T>G, c.3656A>G, c.6071G>A, c.188A>G, c.7582T>C, c.4460-8G>A, c.6784C>T, c.4567C>T, c.5726T>C, c.4560C[3>2] (std: c.4560C[2] alt: c.4562delC), c.679C>T, c.510C>G, c.3513C>A, c.4467T>A, c.8005G>T, c.3083-2A>C, c.8014T>G, c.8006G>T, c.266G>T, c.266G>C, c.266G>A, c.5250T>G, c.493C>T, c.7204+1G>A, c.1285C>T, c.3012C>G, c.3012C>A, c.184C>T, c.6940_6943dupTACA, c.6087C>A, c.1726T>G, c.496T>A, c.503G>T, c.503G>A, c.3037G>A, c.502T>C, c.6113G>A, c.2375G>A, c.7432_7435delGAGG, c.6884G>A, c.1715-2A>G, c.3712G>A, c.4888C>T,

c.1010dupA, c.5066-1G>A, c.2860C>T, c.6904T>A, c.4816+2T>C, c.7654T>C, c.5959G>C, c.2858delT, c.661delT, c.718C>T, c.1011C>A, c.1585C>T, c.1583G>A, c.4852C>T, c.701G>A, c.3302A>G, c.5099A>G, c.3095G>A, c.4942+2T>C, c.5993G>A, c.7454A>G, c.5097C>G, c.7039_7040delAT, c.2268G[2>1] (std: c.2268G[1] alt: c.2269delG), c.3475T>C, c.4942+1G>A, c.649T>G, c.4937G>A, c.2945G>C, c.1098G>T, c.7656C>A, c.4588C>T, c.1051C>T, c.5073AAG[2>1] (std: c.5073AAG[1] alt: c.5076_5078delAAG), c.6159C>A, c.2896G>T, c.2920C>T, c.1207C[5>4] (std: c.1207C[4] alt: c.1211delC), c.2728+1G>C, c.1090C>T, c.1538G>C, c.1129T>C, c.640G>A, c.1570dupA, c.7663G>A, c.2953G>A, c.2293+1G>A, c.1147G>A, c.4691G>A, c.4930C>T, c.7982A>G, c.643C>T, c.8326C>T, c.1522C>T, c.1132C[3>2] (std: c.1132C[2] alt: c.1134delC), c.7820-1G>C, c.7800C>G, c.1546C>T, c.164+1G>A, c.7801C>T, c.7865G>C, c.1035C>A, c.1042C>T, c.2723G>C, c.2723G>A, c.7832G>A, c.7806G>A, c.8525_8529delTTAAC, c.8521G>T, c.7828G>A, c.5203dupC, c.7977C>A, c.2722T>C, c.5162G>A, c.4243dupT, c.4615C>T, c.539-1G>A, c.4817-1G>A, c.1995C>G, c.7819+1G>A, c.8488C>T, c.7879G>C, c.7879G>A, c.4259G>A, c.5156G>A, c.7168T>C, c.7165CT[2>1] (std: c.7165CT[1] alt: c.7167_7168delCT), c.127A>T, c.4222T>C, c.8226+5G>A, c.4224CT[2>1] (std: c.4224CT[1] alt: c.4226_4227delCT), c.7726C>T, c.7180C>T, c.5183C>T, c.3458G>A, c.7916A>G, c.3164G>A, c.5187G>A, c.3413G>C, c.3217G>T, c.3217G>A, c.6296G>A, c.4331G>A, c.3193delG, c.3463G>A, c.4292G>A, c.7955G>A, c.4293C>A, c.3373C>T, c.8226+1G>T, c.8226+1G>A, c.3143T>C, c.7966C>T, c.7769G>A, c.8149G>T, c.7141C>T, c.3338A>G, c.5197T>G, c.2113+2T>G, c.6244G>T, c.7712G>A, c.4621C>T, c.7711T>C, c.3344A>G, c.2019TTTG[2>1] (std: c.2019TTTG[1] alt: c.2023_2026delTTTG), c.7125T>A, c.7708G>A, c.7754T>C, c.2080G>T, c.1A>T, c.1A>C, c.3G>T,

c.3G>A, c.2051G>T, c.2051G>A, c.8080C>T, c.2055C>G, c.7C>T

Gene：

FGFR1

NM ID：

NM_023110.2

Variant：

c.1921G>A, c.2038C>T, c.1869C>G, c.1864C>T, c.1864C>G, c.880G>A, c.961A[3>1] (std: c.961A[1] alt: c.962_963delAA), c.1638C>A, c.1601TGA[2>1] (std: c.1601TGA[1] alt: c.1604_1606delTGA), c.1097C>T, c.1460G>A, c.821A>G, c.296A>G, c.1042G>A, c.232C>T, c.755C>G, c.817G>A, c.1977+1G>A, c.214C>T, c.2059G>A, c.1825C>T

Gene：

FGFR2

NM ID：

NM_000141.4

Variant：

c.1882G>A, c.1942G>A, c.1084+3A>G, c.1922A>G, c.940-2A>G, c.940-1G>A, c.1075G>T, c.940G>T, c.1645A>C, c.1694A>G, c.1694A>C, c.943G>T, c.983A>G, c.1021A>C, c.1024T>G, c.1024T>C, c.1024T>A, c.1019A>G, c.1007A>G, c.2032A>G, c.1025G>T, c.1025G>C, c.1025G>A, c.1052C>G, c.1061C>T, c.1061C>G, c.1026C>G, c.1032G>A, c.1012G>C, c.1018T>C, c.1013G>A, c.1030G>C, c.962A>C, c.1040C>G, c.314A>G, c.842A>G, c.826T>G, c.870G>T, c.870G>C, c.833G>T, c.1172T>G, c.868T>G, c.868T>C, c.755C>G, c.758C>T, c.758C>G, c.866A>C, c.1150G>A, c.812G>T, c.1144T>C, c.1141T>G, c.799T>C, c.1115C>G, c.1124A>G, c.1988G>A, c.1198A>G, c.184T>C, c.1851G>C, c.1084+1G>T, c.958_959delAC, c.812GAG[2>3] (std:

c.815_817dupGAG alt: c.817_818insGAG), c.1646A>C, c.1084+2T>C, c.1084+1G>A, c.940-2A>T, c.923A>G, c.863T>A

Gene：

FGFR3

NM ID：

NM_000142.4

Variant：

c.1612A>G, c.1862G>A, c.1619A>G, c.1620C>A, c.1620C>G, c.833A>G, c.835A>T, c.1118A>G, c.1111A>T, c.1172C>A, c.2419T>G, c.2421A>G, c.2420G>C, c.2420G>T, c.1948A>G, c.1108G>T, c.1949A>C, c.1949A>T, c.1950G>C, c.1138G>A, c.1138G>C, c.1052C>G, c.749C>G, c.746C>G, c.742C>T, c.1031C>G, c.1180A>T, c.1130T>G

Gene：

FLNA

NM ID：

NM_001456.3

Variant：

c.148dupA, c.257AGA[3>2] (std: c.257AGA[2] alt: c.263_265delAGA), c.760G>A, c.3596C>T, c.728C[5>6] (std: c.732dupC alt: c.732_733insC), c.2761C>T, c.3562G>A, c.5193G>A, c.3557C>T, c.676C>T, c.544C>T, c.4071_4074dupGGTG, c.4543C>T, c.1924G>T, c.4599-1G>A, c.4446_4447dupAT, c.620C>T, c.586C>T, c.4660G>A, c.6611_6614delTCAG, c.4726G>A, c.7917_7918delCT, c.6700C>T, c.7231C>T

Gene：

FLNB

NM ID：

NM_001457.3

Variant：

c.1945C>T, c.5071G>A, c.679G>A, c.502G>A

Gene：

FOXG1

NM ID：

NM_005249.4

Variant：

c.250C[7>8] (std: c.256dupC alt: c.256_257insC), c.250C[7>6] (std: c.250C[6] alt: c.256delC), c.294C[5>4] (std: c.294C[4] alt: c.298delC), c.256C>T, c.214C>T, c.301C>T, c.1081G[2>3] (std: c.1082dupG alt: c.1082_1083insG), c.944C[3>2] (std: c.944C[2] alt: c.946delC), c.577G>A, c.799G>A, c.755G>T, c.730C>T, c.624C>A, c.624C>G, c.1023C>G, c.1011C[4>5] (std: c.1014dupC alt: c.1014_1015insC), c.812G>A, c.561C>A, c.561C>G, c.553A>T, c.703C>T, c.543G>T, c.651C>G, c.653A>G, c.645C>G, c.454G[7>8] (std: c.460dupG alt: c.460_461insG), c.454G[7>6] (std: c.454G[6] alt: c.460delG), c.537C>G, c.501G[6>5] (std: c.501G[5] alt: c.506delG), c.406G>T, c.407_458del

Gene：

FOXP1

NM ID：

NM_032682.5

Variant：

c.1393A>G, c.1540C>T, c.1541G>A, c.1574G>A, c.1573C>T, c.1489C>T, c.1507C>T, c.1146+1G>A, c.1235C[6>7] (std: c.1240dupC alt: c.1240_1241insC), c.1240_1241delCT

Gene：

FREM1

NM ID：

NM_144966.5

Variant：

c.3971T>G

Gene：

GABRA1

NM ID：

NM_000806.5

Variant：

c.335G>A, c.857-2A>G, c.1313A>G, c.881C>T, c.884C>T, c.640C>T, c.641G>A

Gene：

GABRB2

NM ID：

NM_021911.2

Variant：

c.911C>T, c.902A>G, c.909G>T, c.904G>A, c.946G>A

Gene：

GATAD2B

NM ID：

NM_020699.2

Variant：

c.346C>T, c.597+1G>A, c.658C>T, c.1196AG[2>1] (std: c.1196AG[1] alt:

c.1198_1199delAG), c.1432C>T, c.535C>T, c.1241G>A, c.1408C>T, c.917T[2>1]

(std: c.917T[1] alt: c.918delT), c.973C>T

Gene：

GFAP

NM ID：

NM_002055.4

Variant：

c.218T>C, c.1157A>T, c.235C>T, c.1154C>G, c.236G>A, c.208C>T, c.715C>T, c.262C>T, c.716G>A, c.758C>G, c.1246C>T

Gene：

GNAO1

NM ID：

NM_020988.2

Variant：

c.724-8G>A, c.680C>T, c.709G>A, c.692A>G, c.723+1G>A, c.736G>A, c.662C>A, c.133G>C, c.649G>A, c.626G>A, c.626G>T, c.625C>T, c.607G>A, c.118G>A, c.118G>C, c.118G>T

Gene：

GRIN1

NM ID：

NM_007327.3

Variant：

c.2479G>A, c.679G>C, c.2530C>T, c.2443G>A, c.1975C>T, c.1910C>T, c.1670C>G, c.1852G>C, c.1858G>C, c.1643G>A

Gene：

GRIN2B

NM ID：

NM_000834.3

Variant：

c.1088delT, c.448A>G, c.2172-2A>G, c.2360-2A>G, c.2131C>T, c.1658C>T, c.1675T>C, c.1672G>A, c.1367G>A, c.1382G>T, c.1677G>A, c.2116A>G, c.2002G>T, c.2044C>T, c.649C>T, c.1495G>A, c.2201C>T, c.1985A>C, c.2087G>A, c.2084T>C, c.1844A>T, c.1821G>T, c.1845C>G, c.2065G>A, c.1858G>A, c.1853T>G, c.2060C>T, c.2060C>G, c.1547A>G, c.2539C>T, c.1555C>T, c.1963A>T, c.737C>A, c.1238A>G, c.1966C>T, c.2429G>A, c.2430C>A, c.1970A>G, c.801CA[2>1] (std: c.801CA[1] alt: c.803_804delCA), c.411+1G>A, c.1883C>T, c.2252T>C, c.1619G>A, c.2515G>A, c.93C[7>8] (std: c.99dupC alt: c.99_100insC), c.2452A>C, c.2453T>C, c.3332G>A, c.1916C>T, c.2458G>A, c.2459G>T, c.2459G>C, c.2459G>A, c.1907C>T, c.1906G>C, c.2473T>G, c.2477G>A

Gene：

HDAC8

NM ID：

NM_018486.2

Variant：

c.130TTGA[2>1] (std: c.130TTGA[1] alt: c.134_137delTTGA), c.738-1G>A, c.932C>T, c.958G>A, c.562G>A, c.466A>G, c.490C>T, c.496C>T, c.787C>T, c.356C>T

Gene：

HNRNPK

NM ID：

NM_002140.3

Variant：

c.214-35A>G, c.257G>A, c.673T>C, c.248G>A, c.778G[2>3] (std: c.779dupG alt: c.779_780insG), c.1008+1G>A, c.859C>T, c.998dupA

Gene：

HNRNPU

NM ID：

NM_031844.2

Variant：

c.1676C[6>7] (std: c.1681dupC alt: c.1681_1682insC), c.2317AG[2>1] (std: c.2317AG[1] alt: c.2319_2320delAG), c.1852C>T, c.2270_2271delCT

Gene：

HRAS

NM ID：

NM_005343.2

Variant：

c.64C>A, c.38G>T, c.38G>A, c.37G>T, c.35_36delGCinsAA, c.173C>T, c.35G>T, c.35G>C, c.35G>A, c.175G>A, c.34G>T, c.34G>A, c.179G>T, c.179G>A, c.187G>A, c.181C>A, c.437C>T, c.350A>G, c.436G>A

Gene：

IFITM5

NM ID：

NM_001025295.2

Variant：

c.119C>T

Gene：

JAG1

NM ID：

NM_000214.2

Variant：

c.1395+3A>G, c.3048+1G>T, c.3048+1G>A, c.3006C>A, c.1156G>A, c.1325G>A, c.1326G>A, c.2963T[4>5] (std: c.2966dupT alt: c.2966_2967insT), c.1897TG[2>1] (std: c.1897TG[1] alt: c.1899_1900delTG), c.1528C>T, c.2230C>T, c.1561CT[2>1] (std: c.1561CT[1] alt: c.1563_1564delCT), c.1200C[6>7] (std: c.1205dupC alt: c.1205_1206insC), c.1207C>T, c.1485_1486delCT, c.2473C>T, c.2122C>T, c.2118CAGT[2>1] (std: c.2118CAGT[1] alt: c.2122_2125delCAGT), c.701G>A, c.1057G>T, c.2698C>T, c.2706C>A, c.703C>T, c.3G>A, c.910C>T, c.2688G>A, c.3164_3167delTAAG, c.265G[6>7] (std: c.270dupG alt: c.270_271insG), c.2566C>T, c.389G[2>3] (std: c.390dupG alt: c.390_391insG), c.2091GAAAG[2>1] (std: c.2091GAAAG[1] alt: c.2096_2100delGAAAG), c.439+1G>A, c.439C>T, c.2418C>A, c.588C>A, c.551G>A, c.1657G>T, c.886+3A>G, c.841C>T

Gene：

KANSL1

NM ID：

NM_001193466.1

Variant：

c.1652+1G>A, c.868C>T, c.537A[4>3] (std: c.537A[3] alt: c.540delA), c.1042C>T, c.808_809delCT, c.1816C>T

Gene：

KAT6B

NM ID：

NM_012330.3

Variant：

c.3347CA[2>1] (std: c.3347CA[1] alt: c.3349_3350delCA), c.3349C>T, c.3664+1G>A, c.3765TCTA[2>1] (std: c.3765TCTA[1] alt: c.3769_3772delTCTA), c.3962_3963delAA, c.3172C>T, c.3787A[3>1] (std: c.3787A[1] alt: c.3788_3789delAA), c.3147G>A, c.4203CT[2>1] (std: c.4203CT[1] alt: c.4205_4206delCT), c.5389C>T, c.3606_3609delAACA, c.4101G[2>3] (std: c.4102dupG alt: c.4102_4103insG)

Gene：

KCNB1

NM ID：

NM_004975.2

Variant：

c.1217C>T, c.629C>T, c.1133T>C, c.934C>T, c.935G>A, c.908G>A, c.1132G>C, c.1747C>T, c.916C>T, c.1297C>T, c.643C>T, c.995G>T, c.1001T>C, c.1109G>A, c.1121C>T, c.1045G>T, c.1041C>G, c.1041C>A

Gene：

KCNJ2

NM ID：

NM_000891.2

Variant：

c.779G>C, c.896A>T, c.899G>A, c.899G>T, c.913A>G, c.934C>T, c.653G>A, c.652C>T, c.430G>A, c.200G>A, c.199C>T, c.574A>G, c.431G>A, c.434A>G, c.211G>A, c.644G>A, c.236T>C, c.514G>A, c.224C>T, c.232G>T, c.244C>T, c.245G>A

Gene：

KCNQ2

NM ID：

NM_172107.2

Variant：

c.1632-1G>A, c.1A>G, c.2T>A, c.875T>C, c.693G>T, c.1639C>T, c.710A>G, c.637C>T, c.704C>T, c.881C>T, c.638G>A, c.640C>T, c.868G>A, c.869G>A, c.829A>T, c.506G>T, c.622A>G, c.629G>C, c.629G>A, c.628C>T, c.821C>T, c.926C>T, c.833T>C, c.1224C[6>7] (std: c.1229dupC alt: c.1229_1230insC), c.1224C[6>5] (std: c.1224C[5] alt: c.1229delC), c.620G>A, c.2595G[3>2] (std: c.2595G[2] alt: c.2597delG), c.684C>A, c.841G>T, c.841G>A, c.593G>A, c.844G>A, c.583T>C, c.380A>G, c.602G>A, c.917C>T, c.619C>T, c.587C>T, c.902G>A, c.601C>T, c.916G>A, c.901G>A, c.915C>A, c.911T>C, c.748G>T, c.740C>T, c.365C>T, c.285C>A, c.782T>G, c.325CTGT[2>1] (std: c.325CTGT[1] alt: c.329_332delCTGT), c.1658G>T, c.1658G>A, c.2245G>T, c.1118+2T>G, c.1118+2T>C, c.1051C>G, c.1679G>A, c.788C>T, c.1682C>T, c.1093C>T, c.1678C>T, c.297-2A>G, c.794C>T, c.807G>A, c.1058G>A, c.793G>A, c.1342C>T, c.1687G>A, c.1149-1G>T, c.1525+1G>A, c.431G>A, c.1155C[6>5] (std: c.1155C[5] alt: c.1160delC), c.430C>T, c.430C>G, c.437G[3>2] (std: c.437G[2] alt: c.439delG), c.1749G>C, c.1004C>T, c.2127delT, c.2126_2127delCT, c.2122C[5>6] (std: c.2126dupC alt: c.2126_2127insC), c.1741C>T, c.1741C>G, c.1742G>A, c.998G>A, c.994A>G, c.997C>T, c.140C[5>6] (std: c.144dupC alt: c.144_145insC), c.140C[5>4] (std: c.140C[4] alt: c.144delC)

Gene：

KCNT1

NM ID：

NM_020822.2

Variant：

c.1283G>A, c.1546A>G, c.2849G>A, c.2686A>G, c.1885A>G, c.2280C>G, c.2896G>A, c.785G>A, c.862G>A, c.1421G>A, c.1420C>T, c.811G>T, c.808C>G, c.1193G>A, c.2800G>A, c.2386T>C, c.2782C>T

Gene：

KIF1A

NM ID：

NM_004321.6

Variant：

c.430G>T, c.914C>T, c.773C>T, c.946C>T, c.920G>A, c.757G>A, c.760C>T, c.761G>A, c.643A>C, c.646C>T, c.647G>A, c.821C>T, c.206C>T, c.173C>T, c.38G>A, c.2323C>T, c.32G>A, c.595G>A, c.31C>T, c.232G>A, c.499C>T, c.500G>A, c.1040A>G, c.296C>T, c.304G>A, c.1048C>G

Gene：

KMT2A

NM ID：

NM_001197104.1

Variant：

c.4343G>A, c.4367A>G, c.5621dupG, c.8095C>T, c.4504C>T, c.6571C>T, c.5431C>T, c.3301C>T, c.1137A[6>7] (std: c.1142dupA alt: c.1142_1143insA), c.3461G>A, c.5251A>T, c.3464G>A, c.3460C>T, c.4906C>T, c.7438C>T, c.3190C>T, c.6487C>T, c.502+1G>A, c.3565T>C, c.3451C>T, c.2513G[2>1] (std: c.2513G[1] alt: c.2514delG), c.3034C>T, c.2312C[7>8] (std: c.2318dupC alt: c.2318_2319insC), c.478C>T, c.7069C[3>2] (std: c.7069C[2] alt: c.7071delC),

c.9694C>T, c.3790C>T, c.11230C>T

Gene：

KMT2D

NM ID：

NM_003482.3

Variant：

c.14075+1G>A, c.5166delT, c.4135_4136delAT, c.4148G>A, c.4163G[6>7] (std: c.4168dupG alt: c.4168_4169insG), c.13818C>G, c.16342C>T, c.13996_13997delAG, c.16052G>A, c.15629A>G, c.16360C>T, c.8053C>T, c.15640C>T, c.1967delT, c.15641G>A, c.15536G>A, c.5627_5630delACAG, c.8059C>T, c.15535C>T, c.11047C>T, c.16019G>A, c.5120AC[3>2] (std: c.5120AC[2] alt: c.5124_5125delAC), c.11089G[5>6] (std: c.11093dupG alt: c.11093_11094insG), c.8401C>T, c.2994delT, c.8171_8175delCCAGC, c.3754C>T, c.11149C>T, c.1934C[7>6] (std: c.1934C[6] alt: c.1940delC), c.10999C>T, c.6295C>T, c.7611T[3>4] (std: c.7613dupT alt: c.7613_7614insT), c.15673C>T, c.5269C>T, c.3585dupA, c.6172delG, c.15185G>A, c.16438_16441delAACT, c.16442G>A, c.7411C>T, c.15949TATT[2>1] (std: c.15949TATT[1] alt: c.15953_15956delTATT), c.15061C>T, c.11692C>T, c.8200C>T, c.1139C[5>4] (std: c.1139C[4] alt: c.1143delC), c.16501C>T, c.16294C>T, c.4843C>T, c.3121C>T, c.5104C>T, c.15256C>T, c.15143G>A, c.401-3A>G, c.15142C>T, c.4525_4528delATCT, c.7228C>T, c.8311C>T, c.7480T[2>3] (std: c.7481dupT alt: c.7481_7482insT), c.15461G>A, c.12469C>T, c.7933C>T, c.13606C>T, c.5677C>T, c.15079C>T, c.15088C>T, c.858dupT, c.3699G[6>5] (std: c.3699G[5] alt: c.3704delG), c.15104G>C, c.14189G>A, c.12667C>T, c.6010C>T, c.12844C>T, c.7903C>T, c.11290C>T, c.12268C>T, c.4392C[4>5] (std: c.4395dupC alt: c.4395_4396insC), c.8743C>T, c.5707C>T,

c.11743C>T, c.8488C>T, c.1329_1332delACCT, c.12592C>T, c.6992delT, c.12895G[2>1] (std: c.12895G[1] alt: c.12896delG), c.13028C[5>4] (std: c.13028C[4] alt: c.13032delC), c.11421G[2>1] (std: c.11421G[1] alt: c.11422delG), c.6595delT, c.13450C>T, c.11944C>T, c.14710C>T, c.14713C>T, c.11383C[4>3] (std: c.11383C[3] alt: c.11386delC)

Gene：

KRAS

NM ID：

NM_004985.3

Variant：

c.15A>T, c.468C>G, c.13A>G, c.458A>T, c.458A>G, c.466T>G, c.466T>A, c.34G>A, c.35G>T, c.35G>A, c.38G>A, c.40G>A, c.37G>T, c.64C>G, c.65A>G, c.439A>G, c.173C>T, c.175G>T, c.178G>C, c.178G>A, c.211T>G, c.194G>T, c.108A>G, c.101C>T, c.101C>G, c.355G>A, c.455T>G

Gene：

LMNA

NM ID：

NM_170707.3

Variant：

c.481G>A, c.427T>C, c.448A>C, c.475G>T, c.412G>A, c.497G>C, c.513+1G>A, c.513+1G>C, c.513G>A, c.1609-3C>G, c.11C>G, c.1609-1G>A, c.1380+1G>A, c.194A>G, c.3G>A, c.3G>T, c.16C>T, c.356+1G>A, c.356+1G>C, c.348dupG, c.29C>T, c.832G>C, c.1619T>C, c.917T>C, c.1368C>A, c.1368C>G, c.244G>A, c.1366A>G, c.1622G>A, c.1622G>C, c.1621C>A, c.1621C>T, c.184C>G, c.1357C>T, c.904CT[3>2] (std: c.904CT[2] alt: c.908_909delCT), c.398G>T, c.1394G>A, c.799T>C, c.949G>A, c.1399T>C, c.1346G>A, c.892C>T, c.274C>T,

c.1608+1G>A, c.886_887insA, c.357-1G>T, c.266G>T, c.1401G>A, c.855G[5>4] (std: c.855G[4] alt: c.859delG), c.958delC, c.162_163delGG, c.1444C>T, c.1445G>A, c.639+1G>A, c.1608+5G>A, c.94_96delAAG, c.777T>A, c.158A>G, c.778AAG[2>1] (std: c.778AAG[1] alt: c.781_783delAAG), c.863C>G, c.305T>C, c.364AAG[2>1] (std: c.364AAG[1] alt: c.367_369delAAG), c.122G>A, c.94A>G, c.154C>G, c.73C>G, c.871G>A, c.116A>G, c.762C[2>1] (std: c.762C[1] alt: c.763delC), c.1116G>C, c.1412G>A, c.127G>A, c.608A>G, c.294GC[3>2] (std: c.294GC[2] alt: c.298_299delGC), c.607G>A, c.1296GCAC[3>4] (std: c.1304_1307dupGCAC alt: c.1307_1308insGCAC), c.80C>T, c.91_93delGAG, c.1458G>T, c.746G>A, c.82C>T, c.745C>T, c.1294C>T, c.618C>G, c.961C>T, c.83G>A, c.1112_1115dupTGGA, c.640-10A>G, c.1072G>A, c.1961dupG, c.736C>T, c.1824C>T, c.1057C>T, c.978_979delAC, c.1130G>A, c.1129C>T, c.1477C>T, c.1436delT, c.1142delA, c.1579C>T, c.1580G>A, c.1580G>C, c.1146C>T, c.1745G>A, c.1583C>A, c.1583C>G, c.1494G>A, c.1494G>T, c.644T>C, c.568C>T, c.695G>A, c.1157+1G>T, c.1157G>A, c.700C>T, c.646C>T, c.585C>G, c.569G>A, c.992G>A, c.991_992delCG, c.1045C>T, c.1540T>C, c.1558T>C, c.1521C[6>7] (std: c.1526dupC alt: c.1526_1527insC), c.1158-2A>G, c.1003C>T, c.673C>T

Gene：

LZTR1

NM ID：

NM_006767.3

Variant：

c.401-2_401-1delAG, c.264-13G>A, c.628C>T, c.791+1G>A, c.365C>T, c.486G>A, c.848G>A, c.772T[3>2] (std: c.772T[2] alt: c.774delT), c.2227T[3>2] (std: c.2227T[2] alt: c.2229delT), c.509G>A, c.850C>T, c.1687G>C, c.742G>A,

c.2178C>A, c.21G[7>6] (std: c.21G[6] alt: c.27delG), c.993+1G>A, c.1149+1G>A, c.2407-2A>G, c.1029T[2>1] (std: c.1029T[1] alt: c.1030delT), c.1084C>T, c.1018C>T

Gene：

MAP2K1

NM ID：

NM_002755.3

Variant：

c.323G>T, c.608A>G, c.275T>G, c.199G>A, c.355C>T, c.364A>G, c.158T>C, c.169A>C, c.370C>T, c.371C>T, c.383G>T, c.388T>A, c.388T>C, c.389A>G, c.607G>C

Gene：

MAP2K2

NM ID：

NM_030662.3

Variant：

c.383C>A, c.395G>A, c.400T>C, c.401A>G, c.171T>A, c.170T>G, c.169T>G, c.181A>G, c.619G>A

Gene：

MECP2

NM ID：

NM_004992.3

Variant：

c.298C>G, c.272G[4>6] (std: c.272G[6] alt: c.275_276insGG), c.272G[4>3] (std: c.272G[3] alt: c.275delG), c.316C>T, c.317G>A, c.301C>T, c.302C>G, c.377+1G>T, c.364G>A, c.209_210insA, c.1357C>T, c.212C[4>3] (std: c.212C[3]

alt: c.215delC), c.1308_1309delTC, c.452A>G, c.1159C[5>6] (std: c.1163dupC alt: c.1163_1164insC), c.1162_1163delCCinsTA, c.451G>T, c.1024_1025insAG, c.27-6C>G, c.454C>G, c.410A>G, c.455C>G, c.403A>G, c.1A>T, c.467A>G, c.423C>G, c.468C>G, c.468C>A, c.380C>T, c.378-3C>G, c.839C[2>1] (std: c.839C[1] alt: c.840delC), c.1287C[4>5] (std: c.1290dupC alt: c.1290_1291insC), c.419C>T, c.401C>T, c.401C>G, c.397C>T, c.397C>A, c.398G>T, c.398G>A, c.674C>G, c.471C>G, c.472A>G, c.382C>T, c.691G[5>4] (std: c.691G[4] alt: c.695delG), c.965C>T, c.964C>T, c.473C>T, c.904C>T, c.1087_1088insC, c.763C>T, c.1140dupG, c.-99+2_-99+3delTG, c.-99+2T>G, c.-99+2T>A, c.-99+1G>A, c.705G[6>7] (std: c.710dupG alt: c.710_711insG), c.705G[6>5] (std: c.705G[5] alt: c.710delG), c.905C>T, c.905C>A, c.910A>G, c.1081C[6>7] (std: c.1086dupC alt: c.1086_1087insC), c.1081C[6>5] (std: c.1081C[5] alt: c.1086delC), c.852AAAG[2>1] (std: c.852AAAG[1] alt: c.856_859delAAAG), c.916C>T, c.1203C[6>7] (std: c.1208dupC alt: c.1208_1209insC), c.1203C[6>5] (std: c.1203C[5] alt: c.1208delC), c.807C[2>1] (std: c.807C[1] alt: c.808delC), c.808C>T, c.502C>T, c.803G[4>3] (std: c.803G[3] alt: c.806delG), c.612dupA, c.880C>T, c.730C>T, c.611C>G, c.917G>C, c.917G>A, c.499C>T, c.749_750insT, c.749dupG, c.750C[4>5] (std: c.753dupC alt: c.753_754insC), c.750C[4>6] (std: c.750C[6] alt: c.753_754insCC), c.750C[4>3] (std: c.750C[3] alt: c.753delC), c.754G[2>1] (std: c.754G[1] alt: c.755delG), c.925C>T, c.569G>A, c.1216C>T, c.1132_1133insT, c.1118C>G, c.1133C[5>6] (std: c.1137dupC alt: c.1137_1138insC), c.1133C[5>4] (std: c.1133C[4] alt: c.1137delC)

Gene：

MED13L

NM ID：

NM_015335.4

Variant：

c.6418C>T, c.2579A>G, c.5173C>T, c.2524C>T, c.2605C>T, c.6488C>T, c.6485C>T, c.5588+1G>A, c.263G>A, c.5278C>T, c.1704AG[3>2] (std: c.1704AG[2] alt: c.1708_1709delAG), c.1690C>T, c.4076G>A

Gene：

MEF2C

NM ID：

NM_002397.4

Variant：

c.47ACAG[2>1] (std: c.47ACAG[1] alt: c.51_54delACAG), c.2T>C, c.45dupT, c.43C>T, c.44G>A, c.830T[4>3] (std: c.830T[3] alt: c.833delT), c.766C>T, c.403-1G>T, c.71G>A, c.565C>T, c.113T>C

Gene：

MSX2

NM ID：

NM_002449.4

Variant：

c.443C>A, c.443C>T

Gene：

NALCN

NM ID：

NM_052867.2

Variant：

c.4197+1G>A, c.2203C>T, c.2563C>T, c.1733A>G, c.3542G>A, c.3050T[7>8] (std: c.3056dupT alt: c.3056_3057insT), c.985A>G, c.965T>C, c.4755+1G>T

Gene：

NF1

NM ID：

NM_000267.3

Variant：

c.6641+1G>A, c.6641+1G>T, c.6641delG, c.6622T>C, c.1260+5G>A, c.1260+5G>C, c.6619C>T, c.6624G>A, c.1260+2T>C, c.1260+1G>A, c.1260+1G>C, c.6611G>A, c.6612G>A, c.5749+2T>C, c.5749+2T>G, c.6642-1G>T, c.6606C>A, c.5749+1G>A, c.5944-5A>G, c.6642-3C>A, c.6642-3C>G, c.1393-2A>G, c.1393-2_1393-1delAG, c.1393-1G>T, c.1399dupA, c.586+5G>A, c.574C>T, c.5944-2A>G, c.541C>T, c.540dupA, c.586+1G>A, c.5944-1G>C, c.7267dupA, c.7267A[1>3] (std: c.7267A[3] alt: c.7267_7268insAA), c.569T>G, c.6670C>T, c.1408G>T, c.5725T>C, c.205-2A>C, c.205-2A>G, c.205-2_205-1delAG, c.5724GT[2>1] (std: c.5724GT[1] alt: c.5726_5727delGT), c.1246C>T, c.5722G>T, c.205-1G>A, c.205-1G>C, c.1186-1G>T, c.5546+5G>A, c.5546+5G>T, c.1062+3A>G, c.1541delA, c.1541_1542delAG, c.1540C>T, c.6364+2T>G, c.1436A[5>4] (std: c.1436A[4] alt: c.1440delA), c.6858G>C, c.1062+2T>C, c.6858+1G>A, c.6858+1G>C, c.6858+1G>T, c.7296C>A, c.1237T>C, c.487G>T, c.1721+3A>G, c.480-1G>A, c.7285C>T, c.1238C>G, c.1013A>G, c.1062+1G>A, c.1062G>A, c.1062G>C, c.1017CT[2>1] (std: c.1017CT[1] alt: c.1019_1020delCT), c.2991-2A>G, c.2991-2A>T, c.2990+5G>A, c.5543T>G, c.496_497delGT, c.495TGTT[2>1] (std: c.495TGTT[1] alt: c.499_502delTGTT), c.730+1G>A, c.730+1G>T, c.6364G>A, c.1466A>G, c.1216T[3>2] (std: c.1216T[2] alt: c.1218delT), c.1721+2T>G, c.5546+1G>A, c.484C>T, c.2990G>A, c.6709C>T, c.7000-4A>G, c.6756+2T>A, c.6756+2T>C, c.6756+2T>G, c.5546G>A, c.1020dupT, c.7000-1G>A, c.7000-1G>T, c.2991-1G>A, c.2991-1G>C, c.1720A>C, c.7000-2A>G, c.2991G>T, c.4367+1G>A,

c.4367+1G>C, c.7994AG[2>1] (std: c.7994AG[1] alt: c.7996_7997delAG), c.1009G>T, c.479+1G>A, c.479+1G>T, c.7329delT, c.5719G>T, c.1721+1G>A, c.1721+1G>T, c.1224T>G, c.1007G>A, c.1721G>A, c.1198C>T, c.503C>G, c.6362delG, c.1571A[2>1] (std: c.1571A[1] alt: c.1572delA), c.6756+1G>A, c.4773-2A>C, c.4773-2A>T, c.1453G>T, c.6085-1G>C, c.1570G>T, c.999C>A, c.998dupA, c.6365-11T>G, c.479+5G>A, c.7054C>T, c.6789_6792delTTAC, c.6839T>G, c.1527+5G>A, c.953A[3>4] (std: c.955dupA alt: c.955_956insA), c.6841C>T, c.943C>T, c.1527+1G>A, c.1527+1G>C, c.1527+1G>T, c.1527+1_1527+4delGTAA, c.4514+1G>A, c.4666C>T, c.6999+1G>A, c.889-2A>G, c.724dupA, c.889-1G>A, c.889-1G>C, c.1527+2T>C, c.4773-1G>A, c.1496T>G, c.4662-1G>A, c.989C>T, c.4662-2A>G, c.3198-2A>G, c.3198-2A>T, c.4681_4682delGA, c.888+1G>A, c.888+1G>T, c.6792C>A, c.6792C>G, c.4684G>T, c.6791dupA, c.6770dupG, c.2978T>G, c.6349C>T, c.981GT[2>1] (std: c.981GT[1] alt: c.983_984delGT), c.2953C>T, c.6365-3C>G, c.1641+2T>C, c.7131C>G, c.7552+1G>T, c.6084+2T>C, c.1641+1G>A, c.6365-2A>G, c.227A[7>6] (std: c.227A[6] alt: c.233delA), c.587-3C>A, c.1121_1122delAT, c.2970_2971delAA, c.2970_2972delAAT, c.1039C>T, c.6365-1G>A, c.587-2A>G, c.715C>T, c.6084+1G>A, c.3977T>G, c.1392+1G>A, c.1392+1G>C, c.6974_6977delATAG, c.7633C[6>7] (std: c.7638dupC alt: c.7638_7639insC), c.7633C[6>5] (std: c.7633C[5] alt: c.7638delC), c.240TC[4>3] (std: c.240TC[3] alt: c.246_247delTC), c.240TC[4>2] (std: c.240TC[2] alt: c.244_247delTCTC), c.1139T>C, c.236T>G, c.4495C>T, c.667T>C, c.1063-2A>G, c.7539dupT, c.4276C>T, c.1094C>G, c.7682AG[2>1] (std: c.7682AG[1] alt: c.7684_7685delAG), c.669G>A, c.4319T>A, c.1063-13G>A, c.7806+1G>A, c.4277A>C, c.3113+5G>A, c.4278G>C, c.4309GAA[2>1] (std: c.4309GAA[1] alt: c.4312_4314delGAA), c.3113+2T>C, c.3113+2T>G, c.655-2A>C, c.4493G>A,

c.4312G>T, c.245C>T, c.5943+1G>A, c.5943+1G>T, c.6955C>T, c.6339T>A, c.5205+5G>A, c.5205+5G>C, c.4270-2A>G, c.1070T>C, c.4306A>G, c.1073_1074insA, c.7702C>T, c.247C>T, c.8097+1G>A, c.6579+1G>A, c.6579+1G>T, c.2888A[3>2] (std: c.2888A[2] alt: c.2890delA), c.1713G>A, c.1627C>T, c.7699C>T, c.663G>A, c.4661+1G>A, c.5667dupT, c.6336GT[2>1] (std: c.6336GT[1] alt: c.6338_6339delGT), c.7697C>G, c.662G>A, c.2887C>T, c.6330CT[3>2] (std: c.6330CT[2] alt: c.6334_6335delCT), c.1712G[3>2] (std: c.1712G[2] alt: c.1714delG), c.1712G>A, c.5205+1G>T, c.288+5G>A, c.288+5G>C, c.1381C>T, c.5941C>T, c.3975-2A>G, c.3709-2A>G, c.5547-2A>T, c.4006C>T, c.3033A[5>4] (std: c.3033A[4] alt: c.3037delA), c.6874A[3>2] (std: c.6874A[2] alt: c.6876delA), c.4480C>T, c.3113+1G>A, c.3113+1G>C, c.3113+1G>T, c.3044T>C, c.3045GT[2>1] (std: c.3045GT[1] alt: c.3047_3048delGT), c.7774C[2>1] (std: c.7774C[1] alt: c.7775delC), c.6907C>T, c.5547-1G>C, c.3114-2A>G, c.5498T>G, c.3208C>T, c.910C>T, c.7528C>T, c.3114-1G>A, c.2851-12_2851-9delTTCT, c.7739C>G, c.2410-16A>G, c.277T>C, c.288+1G>A, c.288+1G>C, c.288+1G>T, c.5851dupA, c.269T>C, c.3046T>C, c.286G[3>2] (std: c.286G[2] alt: c.288delG), c.4021C>T, c.5843A[3>1] (std: c.5843A[1] alt: c.5844_5845delAA), c.2869A[2>1] (std: c.2869A[1] alt: c.2870delA), c.3047G>A, c.1660C>T, c.3318C>A, c.5798C>G, c.1646T>C, c.1642-7A>G, c.1642-10A>G, c.3104T>A, c.3104T>G, c.5768T[3>2] (std: c.5768T[2] alt: c.5770delT), c.1658A>G, c.5839C>T, c.7190CT[2>1] (std: c.7190CT[1] alt: c.7192_7193delCT), c.1607C>A, c.3049C>T, c.5750-1G>A, c.4469T>C, c.4473G>A, c.4071C[6>7] (std: c.4076dupC alt: c.4076_4077insC), c.4071C[6>5] (std: c.4071C[5] alt: c.4076delC), c.1642-3C>G, c.1642-2A>G, c.654+1G>A, c.654+1G>T, c.4110+1G>A, c.4110+1G>C, c.4110+1G>T, c.1642-1G>A, c.3315-3C>G, c.3315-2A>G, c.3197+1G>A, c.5626G[2>1] (std:

c.5626G[1] alt: c.5627delG), c.1845+1G>A, c.5624C>G, c.1845G>A, c.1845G>T, c.5791T>C, c.5793G>A, c.7209A[3>2] (std: c.7209A[2] alt: c.7211delA), c.2339C>A, c.2339C>G, c.1722-2A>G, c.1862C[2>1] (std: c.1862C[1] alt: c.1863delC), c.5792G>A, c.4078C>T, c.1603C>T, c.5772T[3>2] (std: c.5772T[2] alt: c.5774delT), c.5766C[2>1] (std: c.5766C[1] alt: c.5767delC), c.2002-1G>A, c.1180T[3>4] (std: c.1182dupT alt: c.1182_1183insT), c.844C>T, c.4108C>T, c.1185G>A, c.705C>G, c.1185+1G>A, c.1185+1G>T, c.1182_1185+1delTAAGG, c.3712G>T, c.7251C>G, c.1185+2T>G, c.5776G>T, c.5904C>G, c.1A>G, c.5928G>A, c.2410-1G>A, c.7204CA[2>1] (std: c.7204CA[1] alt: c.7206_7207delCA), c.2T>A, c.2T>C, c.2T>G, c.7783C>T, c.4368-1G>A, c.4368-1G>T, c.688G>T, c.7238AG[2>1] (std: c.7238AG[1] alt: c.7240_7241delAG), c.1733T[6>5] (std: c.1733T[5] alt: c.1738delT), c.1733T>G, c.4369T[5>4] (std: c.4369T[4] alt: c.4373delT), c.7237C>T, c.6410T>A, c.4084C>T, c.2326-2A>G, c.2326-6T>G, c.4111-2A>G, c.1748A>G, c.60G>C, c.4751T[6>7] (std: c.4756dupT alt: c.4756_4757insT), c.3097C>T, c.2326-3T>G, c.6399dupA, c.625C>T, c.1726C>T, c.2326-1G>C, c.7254AGC[2>1] (std: c.7254AGC[1] alt: c.7257_7259delAGC), c.3089C>G, c.2329T>A, c.2329T>C, c.60+2delT, c.1955delG, c.79C>T, c.1754T>G, c.1756_1759delACTA, c.7515G[4>3] (std: c.7515G[3] alt: c.7518delG), c.4761T>A, c.58C>T, c.6238C[3>2] (std: c.6238C[2] alt: c.6240delC), c.3871-2A>G, c.1884C>A, c.2330G>C, c.5896C>T, c.82C>T, c.3086TC[2>3] (std: c.3088_3089dupTC alt: c.3089_3090insTC), c.1877T[6>7] (std: c.1882dupT alt: c.1882_1883insT), c.1877T[6>5] (std: c.1877T[5] alt: c.1882delT), c.4841_4842insAAT, c.60+1G>C, c.7411C>T, c.7843C>T, c.5329C>T, c.2850+1G>A, c.2850+1G>T, c.4402A>G, c.1885G[4>3] (std: c.1885G[3] alt: c.1888delG), c.1885G>A, c.3974+1G>A, c.3974+1G>C, c.3974+1G>T, c.4173A>T, c.3892C>T, c.2693T>C, c.2356C>T, c.4172G>C,

c.3974G>C, c.2850G>A, c.2251+1G>C, c.3076A>T, c.1261-19G>A, c.731-2A>C, c.731-2A>G, c.1811T>A, c.2163T>A, c.128T>C, c.4982_4983delGT, c.3909T[3>2] (std: c.3909T[2] alt: c.3911delT), c.2509T>A, c.4120C>T, c.731-1G>A, c.3721C>T, c.7900C[2>1] (std: c.7900C[1] alt: c.7901delC), c.1912G>T, c.7419G>A, c.3144G>A, c.4171A>G, c.7418G>A, c.1318C>T, c.2350T>C, c.7846C>T, c.2848C>T, c.5380C>T, c.5286T>G, c.4992G>A, c.2510G>A, c.4168C>G, c.4168C>T, c.2506G>T, c.55G>T, c.1783_1784delGA, c.1261-2A>G, c.3916C>T, c.3161_3165delACCAA, c.311T>G, c.3790G>A, c.3790G>T, c.2446C>T, c.339delG, c.7907+1G>A, c.6514GA[5>4] (std: c.6514GA[4] alt: c.6522_6523delGA), c.6514GA[5>3] (std: c.6514GA[3] alt: c.6520_6523delGAGA), c.7882GT[2>1] (std: c.7882GT[1] alt: c.7884_7885delGT), c.3737dupT, c.3739_3742delTTTG, c.5206-2A>G, c.1797G>A, c.6513T>A, c.4132C>T, c.765T[4>3] (std: c.765T[3] alt: c.768delT), c.5453delT, c.1796G>A, c.808C>T, c.61-1G>A, c.2503C>T, c.2827A>T, c.2125T>C, c.5294C>A, c.1924C>T, c.2027C[7>8] (std: c.2033dupC alt: c.2033_2034insC), c.2027C[7>6] (std: c.2027C[6] alt: c.2033delC), c.337T[2>3] (std: c.338dupT alt: c.338_339insT), c.337T[2>1] (std: c.337T[1] alt: c.338delT), c.7907+5G>A, c.7907+5G>T, c.2709G>A, c.2734C>T, c.6483C>G, c.5206-1delG, c.5206-1G>A, c.2481T[3>2] (std: c.2481T[2] alt: c.2483delT), c.6494C>G, c.4637C>A, c.4637C>G, c.4950C>A, c.139T>C, c.4772+1G>A, c.4772+1G>T, c.4931T[5>4] (std: c.4931T[4] alt: c.4935delT), c.5458C>T, c.801G>A, c.5406dupT, c.2536G>C, c.2072T>G, c.2464G>T, c.61-2A>G, c.4922G>A, c.2537C>A, c.5429G>A, c.4923G>A, c.2560C>T, c.2540T>C, c.2540T>G, c.2543G>A, c.3773G>A, c.3367G>T, c.2084T>C, c.6476C[2>1] (std: c.6476C[1] alt: c.6477delC), c.2542G[5>4] (std: c.2542G[4] alt: c.2546delG), c.2542G>A, c.2542G>C, c.2842C>T, c.1299T>A, c.1299T>G, c.6425T>A,

c.2531T>C, c.2531T>G, c.2530C>T, c.2534G>A, c.3763C>T, c.7486C>T, c.3814C>T, c.2533T>C, c.2041C>T, c.2050C>T, c.7432G[2>1] (std: c.7432G[1] alt: c.7433delG), c.5401C>T, c.317A[3>2] (std: c.317A[2] alt: c.319delA), c.3376C>T, c.2044C>T, c.2830T[6>5] (std: c.2830T[5] alt: c.2835delT), c.6440delG, c.1294GT[2>3] (std: c.1296_1297dupGT alt: c.1297_1298insGT), c.5440C>T, c.2453C[3>2] (std: c.2453C[2] alt: c.2455delC), c.2674delA, c.4912CT[3>1] (std: c.4912CT[1] alt: c.4914_4917delCTCT), c.1275G>A, c.3834C>G, c.334C>T, c.3825C[2>1] (std: c.3825C[1] alt: c.3826delC), c.31C>T, c.3821dupT, c.3818CT[3>2] (std: c.3818CT[2] alt: c.3822_3823delCT), c.1278G>A, c.5438C>G, c.5425C>A, c.5425C>G, c.5425C>T, c.4620C[2>1] (std: c.4620C[1] alt: c.4621delC), c.2409+2T>G, c.4606dupA, c.3230T[3>4] (std: c.3232dupT alt: c.3232_3233insT), c.2764G>A, c.5426G>T, c.3826C>G, c.3826C>T, c.147C>G, c.5234C>G, c.3707G>A, c.5264C>G, c.2409+1G>A, c.2409+1G>C, c.2409+1G>T, c.3685A[2>1] (std: c.3685A[1] alt: c.3686delA), c.3827G>A, c.3827G>C, c.3827G>T, c.3703C>T, c.5224C>T, c.4614G>A, c.2407C>T, c.4613G>A, c.2622_2623insA, c.2665dupA, c.3942G>A, c.3663C[3>2] (std: c.3663C[2] alt: c.3665delC), c.3497-1G>A, c.204+2T>G, c.3427C>T, c.5248A>G, c.204+1G>A, c.204+1G>T, c.5242C>T, c.2619dupT, c.3445A>G, c.3233C>G, c.2325+3A>G, c.2325+2T>C, c.2252-3T>G, c.4269+2T>C, c.2252-2A>C, c.2252-2A>G, c.2325+1G>A, c.2325+1G>T, c.3639_3641delAAT, c.4269+1G>A, c.4269+1G>T, c.3628G>T, c.2252-1G>C, c.4269G>A, c.4269G>C, c.4265C>T, c.4267A>C, c.4267A>G, c.3525_3526delAA, c.4268A>G, c.3870+1G>A, c.3870+1G>C, c.3870+1G>T, c.3461A>T, c.3447G>A, c.3447G>C, c.3447G>T, c.4243G>T, c.3520C>T, c.3616G>T, c.3456dupA, c.3457_3460delCTCA, c.3449C>G, c.3544G>T, c.4253T>A, c.2266C>T, c.4515-2A>G, c.3587T>G, c.2269A[4>2] (std: c.2269A[2] alt:

c.2271_2272delAA), c.4515-1G>A, c.3565C>T, c.4537C>T, c.3578T>G, c.3610C>G, c.3610C>T, c.3603G[2>1] (std: c.3603G[1] alt: c.3604delG), c.3596C>T, c.2288T>C, c.2288T>G, c.3256C>T, c.4572C>G, c.4557A[2>1] (std: c.4557A[1] alt: c.4558delA), c.3277G>A, c.3299CA[2>1] (std: c.3299CA[1] alt: c.3301_3302delCA)

Gene：

NF2

NM ID：

NM_000268.3

Variant：

c.58A>T, c.241-2A>G, c.354CTT[2>1] (std: c.354CTT[1] alt: c.357_359delCTT), c.810+2T>C, c.810+1G>A, c.1737+1G>T, c.516+1G>A, c.240+2T>C, c.240+1G>C, c.784C>T, c.1550T>C, c.600-2A>G, c.122G>A, c.193C>T, c.517-2A>G, c.517-1G>A, c.493C>T, c.1021C>T, c.592C>T, c.586C>T, c.169C>T, c.531T>A, c.658A>T, c.432C>G, c.1228C>T, c.1396C>T, c.1387G>T, c.1198C>T, c.1345A[3>1] (std: c.1345A[1] alt: c.1346_1347delAA), c.1575-2A>G, c.1340+2T>G, c.999+1G>A, c.1606C>T

Gene：

NFIX

NM ID：

NM_001271043.1

Variant：

c.592C>T, c.544G>T, c.385C>T, c.371G>A

Gene：

NIPBL

NM ID：

NM_133433.3

Variant：

c.6647ATA[3>2] (std: c.6647ATA[2] alt: c.6653_6655delATA), c.4321G>T, c.6589+5G>A, c.4782TCAGT[2>1] (std: c.4782TCAGT[1] alt: c.4787_4791delTCAGT), c.771+1G>A, c.7435GA[3>2] (std: c.7435GA[2] alt: c.7439_7440delGA), c.5329-15A>G, c.4291C>T, c.4593T>G, c.5509C>T, c.737A>G, c.4606C>T, c.3058AG[3>1] (std: c.3058AG[1] alt: c.3060_3063delAGAG), c.7410+4A>G, c.5808+1G>A, c.65-5A>G, c.5440C>T, c.5455C>T, c.5483G>A, c.1435C>T, c.7012G>C, c.64+1G>A, c.5262C>A, c.7219C>T, c.6108+2T>C, c.8377C>T, c.459-2A>G, c.2500C>T, c.2479_2480delAG, c.2494C>T, c.7849C>T, c.85C[2>1] (std: c.85C[1] alt: c.86delC), c.2464_2467delCAAA, c.3445C>T, c.7788C[2>1] (std: c.7788C[1] alt: c.7789delC), c.3439C>T, c.7301A>G, c.7175G>A, c.7327C>T, c.7168G>A, c.7150C>T, c.6892C>T, c.6893G>A, c.2422C>T, c.2602C>T, c.133C>T, c.2389C>T, c.5167C>T, c.1885C>T, c.1660C>T

Gene：

NOTCH2

NM ID：

NM_024408.3

Variant：

c.6424_6427delTCTG, c.6853C>T, c.6787C>T

Gene：

NR2F1

NM ID：

NM_005654.4

Variant：

c.335G>A, c.317G>A, c.2T>C, c.382T>C, c.425G>A, c.425G>T

Gene：

NRAS

NM ID：

NM_002524.4

Variant：

c.71T>A, c.179G>A, c.149C>T, c.182A>G, c.38G>A, c.35G>T, c.34G>C, c.34G>A

Gene：

NSD1

NM ID：

NM_022455.4

Variant：

c.4297A[5>4] (std: c.4297A[4] alt: c.4301delA), c.1262G>A, c.5903T>C, c.4378+1G>A, c.4378+1_4378+4delGTGA, c.4379-2A>G, c.4753G>T, c.5146+1G>A, c.4417C>T, c.4217_4220delGAAA, c.4966+1G>A, c.1318C>T, c.2049_2053delGATAA, c.5927T>C, c.6086C>T, c.4411C>T, c.5892+1G>T, c.3379CT[3>2] (std: c.3379CT[2] alt: c.3383_3384delCT), c.5885T>C, c.6013C>T, c.6070C>T, c.5616TA[2>1] (std: c.5616TA[1] alt: c.5618_5619delTA), c.3958C>T, c.3290T>A, c.6490T>C, c.6014G>A, c.5965C>T, c.3964C>T, c.2760_2763delTAAG, c.6020T>C, c.6019A>T, c.6258+1G>A, c.5989T>C, c.5609A>G, c.5990A>G, c.2672_2676delCTTCT, c.3839G>A, c.1492C>T, c.6059A>G, c.5950C>T, c.5758T>A, c.5304-1G>C, c.6412T>C, c.5951G>A, c.5309G>A, c.3546CT[2>1] (std: c.3546CT[1] alt: c.3548_3549delCT), c.6049C>T, c.3549dupT, c.5581C>T, c.5303+1G>C, c.6050G>A, c.5740C>T, c.3214C>T, c.1894C>T, c.5566C>T, c.5296C>T, c.4817G>A, c.1831C>T,

c.6421delG, c.1810C>T, c.6437dupG, c.6521_6523delTCT, c.5854C>T, c.6426C>G, c.1828C>T, c.5332C>T, c.6455G>A, c.2859dupT, c.4192+2T>G, c.6288A[3>4] (std: c.6290dupA alt: c.6290_6291insA), c.6454C>T, c.5177C>G, c.4057G>T, c.5279_5282delTCTG, c.3172C>T, c.4108C>T, c.6366T>G, c.5431C>T, c.2362C>T, c.5017T[4>3] (std: c.5017T[3] alt: c.5020delT), c.5098C>T, c.2386_2389delGAAA, c.6356A>G, c.3657AG[2>1] (std: c.3657AG[1] alt: c.3659_3660delAG), c.6349C>T, c.5229G>A, c.6559C>T, c.3091C>T, c.6605G>A, c.3067C>T, c.3004_3005delAA, c.2952CT[2>1] (std: c.2952CT[1] alt: c.2954_2955delCT)

Gene:

NSDHL

NM ID:

NM_015922.2

Variant:

c.1038_1041dupCATG, c.208C>T

Gene:

PACS1

NM ID:

NM_018026.3

Variant:

c.607C>T

Gene:

PIK3CA

NM ID:

NM_006218.2

Variant:

c.1048G>A, c.1030G>A, c.1133G>A, c.1356AGA[2>1] (std: c.1356AGA[1] alt: c.1359_1361delAGA), c.1357G>A, c.1093G>A, c.1258T>C, c.1633G>A, c.1624G>A, c.263G>A, c.323G>A, c.325GAA[2>1] (std: c.325GAA[1] alt: c.328_330delGAA), c.2740G>A, c.3139C>T, c.2816A>G, c.3140A>G, c.3140A>T, c.3129G>A, c.3129G>T, c.2176G>A

Gene：

PIK3R2

NM ID：

NM_005027.2

Variant：

c.1117G>A, c.1694C>G

Gene：

PPP2R1A

NM ID：

NM_014225.5

Variant：

c.773G>A, c.548G>A, c.536C>T, c.544C>T, c.547C>T, c.656C>T

Gene：

PPP2R5D

NM ID：

NM_006245.3

Variant：

c.1258G>A, c.619T>C, c.748G>A, c.751G>T, c.598G>A, c.752A>C, c.752A>T, c.602C>G, c.592G>A, c.632A>C

Gene：

PRKAR1A

NM ID：

NM_002734.3

Variant：

c.891+3A>G, c.535C>T, c.682C>T, c.738T>G, c.177+3A>G, c.177+1G>A, c.892-2A>G, c.-6-2A>G, c.672G>A, c.1A>G, c.671G>A, c.502+1G>A, c.46C>T, c.124C>T, c.489TG[2>1] (std: c.489TG[1] alt: c.491_492delTG), c.622G[2>1] (std: c.622G[1] alt: c.623delG), c.286C>T, c.289C>T, c.1003C>T, c.1102C>T

Gene：

PTPN11

NM ID：

NM_002834.3

Variant：

c.5C>T, c.767A>G, c.766C>A, c.774G>T, c.782T>A, c.781C>T, c.785T>G, c.794G>A, c.598A>T, c.329A>C, c.836A>C, c.836A>G, c.184T>G, c.328G>A, c.182A>C, c.182A>G, c.181G>A, c.181G>C, c.181G>T, c.155C>T, c.317A>C, c.853T>A, c.853T>C, c.209A>G, c.211T>C, c.846C>G, c.188A>G, c.218C>T, c.174C>A, c.174C>G, c.206A>T, c.179G>C, c.179G>T, c.166A>G, c.205G>A, c.205G>C, c.172A>C, c.172A>G, c.172A>T, c.213T>G, c.844A>G, c.167T>C, c.228G>C, c.228G>T, c.178G>A, c.178G>C, c.178G>T, c.215C>G, c.227A>C, c.227A>G, c.227A>T, c.214G>A, c.214G>C, c.214G>T, c.802G>A, c.802G>T, c.236A>G, c.855T>G, c.854T>A, c.854T>C, c.854T>G, c.923A>C, c.923A>G, c.922A>G, c.1381G>A, c.1381G>T, c.1391G>C, c.124A>G, c.1403C>T, c.1402A>C, c.1529A>C, c.1529A>G, c.1530G>C, c.1530G>T, c.1528C>G, c.412C>T, c.417G>C, c.417G>T, c.1517A>C, c.1510A>G, c.1504T>A, c.1504T>G, c.1520C>A, c.1505C>T, c.1502G>A, c.1507G>A, c.1507G>C, c.1508G>A,

c.1493G>T, c.1492C>T, c.1471C>A, c.1471C>G, c.1471C>T, c.1472C>A, c.1472C>T, c.1282G>T

Gene：

PURA

NM ID：

NM_005859.4

Variant：

c.691TTC[3>2] (std: c.691TTC[2] alt: c.697_699delTTC), c.710C[2>3] (std: c.711dupC alt: c.711_712insC), c.675TG[2>1] (std: c.675TG[1] alt: c.677_678delTG), c.205C>T, c.487C>T, c.640G>T, c.563T>C, c.572C>T, c.163C>T, c.596G>C, c.363C>G, c.812_814delTCT, c.430A>G, c.154G[6>7] (std: c.159dupG alt: c.159_160insG), c.303TC[3>2] (std: c.303TC[2] alt: c.307_308delTC)

Gene：

RAD21

NM ID：

NM_006265.2

Variant：

c.1306C>T

Gene：

RAF1

NM ID：

NM_002880.3

Variant：

c.1423T>C, c.1082G>C, c.1457A>G, c.1472C>T, c.505G>C, c.785A>T, c.786T>A, c.788T>G, c.788T>C, c.788T>A, c.781C>T, c.781C>G, c.781C>A, c.782C>T,

c.782C>G, c.779C>A, c.775T>G, c.775T>C, c.775T>A, c.769T>C, c.770C>T, c.776C>T, c.776C>G, c.776C>A, c.768G>T, c.766A>G, c.1279A>G, c.1837C>G

Gene：

RERE

NM ID：

NM_012102.3

Variant：

c.4304A>T, c.4304A>G, c.4293C>A, c.3146C>T

Gene：

RIT1

NM ID：

NM_006912.5

Variant：

c.170C>G, c.230C>G, c.229G>C, c.229G>A, c.104G>C, c.69A>C, c.67A>C, c.242A>G, c.244T>G, c.244T>C, c.365G>T, c.265T>C, c.246T>G, c.246T>A, c.247A>C, c.268A>G, c.270G>T, c.270G>C, c.270G>A, c.251C>T, c.284G>C

Gene：

RPS6KA3

NM ID：

NM_004586.2

Variant：

c.1894C>T, c.1959+2delT, c.1934G>A, c.356G>C, c.632-1G>C, c.328C>T, c.334C>T, c.727C>T, c.817C>T, c.205G>T, c.212T>G, c.1699C>T, c.898C>T, c.913C>T, c.2185C>T, c.2186G>A

Gene：

RUNX2

NM ID：

NM_001024630.3

Variant：

c.674G>A, c.673C>T, c.577C>T, c.569G>A, c.568C>T, c.1171C>T, c.278T>A, c.1085C>T

Gene：

SATB2

NM ID：

NM_015265.3

Variant：

c.1169C>T, c.715C>T, c.1165C>T, c.1756C>T, c.474-2A>G, c.1495A>T, c.1127GT[3>2] (std: c.1127GT[2] alt: c.1131_1132delGT), c.480A[3>2] (std: c.480A[2] alt: c.482delA), c.1196G>A, c.346G>C, c.1375C>T, c.1286G>A, c.1285C>T, c.597+1G>A, c.847C>T, c.1728delT, c.868C>T, c.1946C>T, c.1964C>T, c.124G>T

Gene：

SCN1A

NM ID：

NM_001165963.1

Variant：

c.265-1G>A, c.269T>C, c.1170+1G>T, c.3706G>C, c.1162T>C, c.278_279dupTG, c.4284+1G>T, c.4284+2T>C, c.962C>G, c.3716AT[5>6] (std: c.3724_3725dupAT alt: c.3725_3726insAT), c.1028+2T>C, c.4300T>C, c.4298G>A, c.4301G>A, c.1028+1G>T, c.1007G>A, c.2590-2A>G, c.965-1G>A, c.4266T>A, c.1150T>A, c.1025C>T, c.4321G>C, c.3730C>T, c.301C>T, c.302G>A, c.3429+1G>T,

c.3424A[3>1] (std: c.3424A[1] alt: c.3425_3426delAA), c.974A>G, c.474-1G>A, c.971ATT[2>1] (std: c.971ATT[1] alt: c.974_976delATT), c.3734G>A, c.3733C>T, c.2593C>T, c.4003G>A, c.2T>C, c.1133delT, c.4016C>T, c.4016C>A, c.4033C>T, c.985G>T, c.715G>A, c.314C>T, c.1130G>A, c.1129C>T, c.4013A>C, c.4822G>T, c.4441G>A, c.838T>C, c.4412C>T, c.337C>A, c.719T>C, c.4002+2T>C, c.4787G>A, c.4786C>T, c.4428C>A, c.354G>T, c.842C>T, c.350T>C, c.4224G>A, c.2946+5G>A, c.4219C>T, c.1113T[6>5] (std: c.1113T[5] alt: c.1118delT), c.4223G>A, c.4002+1G>T, c.3985C>T, c.602+1G>T, c.602+1G>C, c.602+1G>A, c.3977C>A, c.2946G[2>1] (std: c.2946G[1] alt: c.2947delG), c.4384T>C, c.1112C>T, c.4757G>A, c.434T>C, c.505T>C, c.4762T>C, c.2946+1G>T, c.3968C>A, c.580G>A, c.3106C>T, c.1662+2T>C, c.4477-2A>G, c.425GT[3>2] (std: c.425GT[2] alt: c.429_430delGT), c.4168G>A, c.1662+3A>G, c.3860T>C, c.575G>A, c.1662+1G>T, c.1662+1G>A, c.3879+1G>T, c.2615C>A, c.3818C>T, c.3829C>T, c.749T>A, c.1096G>C, c.563A>T, c.1087A>C, c.2213G>A, c.264+4_264+7delAGTG, c.2624C>T, c.748G>A, c.4133A>C, c.568T>C, c.264+5G>C, c.264+5G>A, c.3864CTT[2>1] (std: c.3864CTT[1] alt: c.3867_3869delCTT), c.769T>C, c.3295G>T, c.3858G>A, c.811_815dupGGCAA, c.3850T>C, c.763GT[2>1] (std: c.763GT[1] alt: c.765_766delGT), c.2299_2302dupGACC, c.3812G>A, c.3370T[3>1] (std: c.3370T[1] alt: c.3371_3372delTT), c.677C>T, c.539T>A, c.2935G>A, c.680T>G, c.3496C>T, c.777C>A, c.4853-1G>C, c.3778A>C, c.675G>C, c.3852G>A, c.5744_5748delAGGAA, c.530G>T, c.530G>A, c.2956C>T, c.4862T>C, c.251A>G, c.1378C>T, c.3880-2A>G, c.1076A>T, c.1076A>G, c.4495T>C, c.4879_4883dupAAGTA, c.661C>G, c.2348T[4>2] (std: c.2348T[2] alt: c.2350_2351delTT), c.2354T>C, c.2928G>A, c.3880-1G>T, c.2353A>G, c.3940TC[3>2] (std: c.3940TC[2] alt: c.3944_3945delTC), c.4352C>T, c.4359T>G,

c.243C>A, c.1624C>T, c.3705+1G>A, c.5719ACT[3>2] (std: c.5719ACT[2] alt: c.5725_5727delACT), c.5726C>T, c.5741_5742delAA, c.1348C>T, c.664C>T, c.235G>A, c.5765T>C, c.5734C>T, c.1277A>G, c.5710C>T, c.5708_5711dupATCA, c.1049TG[4>3] (std: c.1049TG[3] alt: c.1055_1056delTG), c.4913T>A, c.4633A>G, c.4521C>G, c.1665T[3>2] (std: c.1665T[2] alt: c.1667delT), c.4906C>T, c.4907G>A, c.2131C>T, c.2665G>A, c.1261G>A, c.1264G>A, c.2669T>C, c.4581+1G>A, c.2134C>T, c.1259C>T, c.2678T>A, c.3629C>A, c.4529C>T, c.3615G>A, c.603-2A>G, c.652T>C, c.2904C>A, c.603-1G>A, c.1177C>T, c.4573C>T, c.3656G>A, c.1178G>A, c.3610T>C, c.1702C>T, c.3611G>A, c.3612G>A, c.4934G>A, c.2893C>T, c.4933C>T, c.4942C>T, c.4943G>A, c.3637C>T, c.4549A>T, c.4549A[6>7] (std: c.4554dupA alt: c.4554_4555insA), c.4549A[6>5] (std: c.4549A[5] alt: c.4554delA), c.2791C>T, c.4555C>T, c.2792G>T, c.2792G>C, c.2792G>A, c.2796G>A, c.2802G>A, c.2867T>C, c.4547C>A, c.2875T>C, c.2876G>A, c.2860G>A, c.2819C>T, c.2855G>A, c.2831T>C, c.2495G>A, c.2836C>T, c.2815C>T, c.4543G>A, c.2854T>G, c.1506GAAAA[2>1] (std: c.1506GAAAA[1] alt: c.1511_1515delGAAAA), c.2849G>A, c.1516C>T, c.121A[6>5] (std: c.121A[5] alt: c.126delA), c.1738C>T, c.2837G>A, c.4968C>G, c.5532AAAC[2>1] (std: c.5532AAAC[1] alt: c.5536_5539delAAAC), c.5536A>T, c.2569delG, c.5555T>C, c.5674C>T, c.4970G>A, c.4997C>T, c.2589+3A>T, c.5000T>C, c.2575C>T, c.2589+1G>T, c.2576G>A, c.4985C>T, c.5261G>A, c.5164A>G, c.5506G>T, c.5162C>A, c.2585G>T, c.2585G>A, c.2584C>T, c.5468T>C, c.5014A>C, c.5306A>G, c.4973C>T, c.4979T>C, c.5422T>C, c.5656C>T, c.5048T>C, c.5168C>T, c.5006C>A, c.5010_5013delGTTT, c.5334delG, c.5434T>G, c.5435G>A, c.5436G>A, c.5218G>A, c.5222G>C, c.5222G>A, c.5346C>G, c.5347G>A, c.5348C>T, c.1804G>T, c.1876A>G, c.1834C>T, c.1837C>T

Gene：

SCN2A

NM ID：

NM_021007.2

Variant：

c.4303C>T, c.4264A>G, c.304C>T, c.605+1G>A, c.605C>T, c.386+2T>C, c.4223T>C, c.4446+9A>G, c.408G>A, c.3967A>G, c.3964G>A, c.1147C>G, c.3956G>A, c.3956G>T, c.823C>T, c.3955C>T, c.3947C>T, c.4879G>A, c.4877G>A, c.4886G>A, c.3977T>A, c.4123T>G, c.4766A>G, c.4468A>G, c.2715G>C, c.2877C>A, c.2995G>A, c.4036A>G, c.4787T>G, c.2722A>G, c.4025T>C, c.781G>A, c.3631G>A, c.2558G>A, c.4904G>A, c.668G>A, c.4687C>G, c.2674G>A, c.751G>A, c.5645G>A, c.2960G>T, c.5644C>G, c.647T>G, c.2548C>T, c.2687C>T, c.1289A>G, c.1312G>A, c.718G>A, c.2809C>T, c.2774T>C, c.2566C>T, c.4633A>G, c.719C>T, c.2567G>A, c.2810G>A, c.620T>C, c.2627A>G, c.1819C>T, c.1827CT[3>2] (std: c.1827CT[2] alt: c.1831_1832delCT), c.5308A>T, c.4949T>C, c.5317G>A, c.5318C>T, c.4976C>T

Gene：

SCN8A

NM ID：

NM_014191.3

Variant：

c.2300C>T, c.2287A>G, c.2537T>C, c.4408C>A, c.4819G>A, c.632T>C, c.641G>A, c.4850G>A, c.4859G>T, c.2879T>A, c.614C>A, c.4426G>A, c.4423G>A, c.2549G>A, c.5279T>C, c.4447G>A, c.5302A>G, c.5630A>G, c.2603T>C, c.2620G>A, c.3953A>G, c.3979A>G, c.5615G>A, c.3985A>G,

c.3943G>A, c.5614C>T, c.718A>G, c.800T>C, c.3967G>A, c.4594A>T, c.2932A>G, c.779T>C, c.4948G>A

Gene：

SETBP1

NM ID：

NM_015559.2

Variant：

c.1765C>T, c.1873C>T, c.1876C>T, c.1630C>T, c.2612T>C, c.2608G>A, c.2602G>A

Gene：

SETD2

NM ID：

NM_014159.6

Variant：

c.4449AGAA[3>2] (std: c.4449AGAA[2] alt: c.4457_4460delAGAA)

Gene：

SETD5

NM ID：

NM_001080517.1

Variant：

c.2347-7A>G, c.3214C>T, c.988A>G, c.3001C>T, c.1783-2A>T, c.2302C>T, c.922C>T, c.1333C>T, c.3849C[7>8] (std: c.3855dupC alt: c.3855_3856insC), c.3856delT

Gene：

SHANK3

NM ID：

NM_033517.1

Variant：

c.271C>T, c.4644GCAGC[2>3] (std: c.4649_4653dupGCAGC alt: c.4653_4654insGCAGC), c.4972C>T, c.4402C>T, c.3637dupC, c.3672G[8>9] (std: c.3679dupG alt: c.3679_3680insG), c.3672G[8>7] (std: c.3672G[7] alt: c.3679delG), c.2265+1G>A, c.4065_4066delTG, c.3424_3425delCT

Gene：

SHOC2

NM ID：

NM_007373.3

Variant：

c.517A>G, c.4A>G

Gene：

SKI

NM ID：

NM_003036.3

Variant：

c.103C>T, c.100G>A, c.100G>T, c.349G>C, c.347G>A, c.94C>G

Gene：

SLC25A24

NM ID：

NM_213651.2

Variant：

c.593G>A, c.592C>T

Gene：

SMAD3

NM ID：

NM_005902.3

Variant：

c.1175C[5>6] (std: c.1179dupC alt: c.1179_1180insC), c.401-6G>A, c.1A>T, c.5C>A, c.1247C>T, c.53G[2>1] (std: c.53G[1] alt: c.54delG), c.667C[2>1] (std: c.667C[1] alt: c.668delC), c.652A[2>1] (std: c.652A[1] alt: c.653delA), c.154G>T, c.82G>T, c.450C[6>5] (std: c.450C[5] alt: c.455delC), c.803G>A, c.989C[2>3] (std: c.990dupC alt: c.990_991insC), c.802C>T, c.754C>T, c.797C>A, c.715G>A, c.733G>A, c.859C>T, c.860G>A, c.277C>T, c.1118G>A, c.1091A>G, c.1102C>T

Gene：

SMAD4

NM ID：

NM_005359.5

Variant：

c.461C>G, c.505C>T, c.687G[6>7] (std: c.692dupG alt: c.692_693insG), c.1333C>T, c.426TC[3>2] (std: c.426TC[2] alt: c.430_431delTC), c.533C>G, c.1308+1G>A, c.1345C>T, c.1130AG[3>2] (std: c.1130AG[2] alt: c.1134_1135delAG), c.1096C>T, c.403C>T, c.1081C>T, c.1245_1248delCAGA, c.1271dupA, c.1198delA, c.1082G>A, c.787+2T>C, c.1258_1259dupCG, c.1194G>A, c.1229AG[2>1] (std: c.1229AG[1] alt: c.1231_1232delAG), c.250-1G>C, c.263_267delAAGGA, c.1228_1229delCA, c.1022T[2>1] (std: c.1022T[1] alt: c.1023delT), c.906G>A, c.1035C[3>2] (std: c.1035C[2] alt: c.1037delC), c.989A>G, c.148A[6>7] (std: c.153dupA alt: c.153_154insA), c.1547dupA, c.1547AG[2>1] (std: c.1547AG[1] alt: c.1549_1550delAG), c.1499T>C,

c.1498A>G, c.1486C>T, c.937C[3>4] (std: c.939dupC alt: c.939_940insC)

Gene：

SMARCA2

NM ID：

NM_003070.3

Variant：

c.3476G>A, c.2348C>G, c.3485G>A, c.2554G>A, c.3602C>T, c.2564G>A, c.3314G>A, c.1573C>T, c.1574G>A, c.2648C>A, c.2648C>T

Gene：

SMARCA4

NM ID：

NM_001128849.1

Variant：

c.2032C>T, c.2653C>T, c.2438C>T, c.3277C>T, c.1757_1760delAGAA, c.4567C>T, c.3608G>A, c.3565C>T, c.3559C>T, c.2936G>A, c.2935C>T, c.1141C>T, c.2859+1G>C, c.3922C>T

Gene：

SMARCB1

NM ID：

NM_003073.3

Variant：

c.141C>A, c.118C>T, c.1070C>T, c.1087A>G, c.1085AGA[3>2] (std: c.1085AGA[2] alt: c.1091_1093delAGA), c.1118+1G>A, c.1121G>A, c.362+1G>A, c.472C>T, c.601C>T, c.364G>T

Gene：

SMARCE1

NM ID：

NM_003079.4

Variant：

c.751C>T, c.715C>T, c.218A>G, c.314G>A, c.624_627delTGAG

Gene：

SMC1A

NM ID：

NM_006306.2

Variant：

c.2447G>A, c.2853_2856delTCAG, c.1193G>A, c.1192C>T,

c.3549_3552dupGGCC, c.2420G>A, c.547C>T, c.1495C>T, c.2351T>C,

c.2870C[3>4] (std: c.2872dupC alt: c.2872_2873insC), c.2131C>T, c.1486C>T,

c.1487G>A, c.796AAG[3>2] (std: c.796AAG[2] alt: c.802_804delAAG),

c.2161C>T, c.586C>T, c.2369G>A, c.2368C>T, c.3103C>T, c.3197G>A,

c.3254A>G, c.3145C>T, c.3146G>A, c.3362G>A

Gene：

SMC3

NM ID：

NM_005445.3

Variant：

c.3373C>G, c.1942A>G

Gene：

SOS1

NM ID：

NM_005633.3

Variant：

c.512T>C, c.2183A>T, c.2197A>T, c.1867T>A, c.508A>G, c.2536G>A, c.2104T>C, c.1132A>G, c.335C>G, c.1430A>G, c.1433C>G, c.1654A>T, c.1654A>G, c.1642A>C, c.1644T>A, c.1655G>T, c.1655G>C, c.1655G>A, c.1656G>T, c.1656G>C, c.1649T>C, c.322G>A, c.305C>G, c.1294T>C, c.844T>C, c.1297G>A, c.925G>T, c.1322G>A, c.1300G>C, c.1300G>A, c.806T>G, c.806T>C, c.1310T>G, c.1310T>C, c.797C>A

Gene：

SOS2

NM ID：

NM_006939.2

Variant：

c.1127C>G, c.800T>G, c.800T>C, c.800T>A, c.791C>A

Gene：

SOX9

NM ID：

NM_000346.3

Variant：

c.507C>G, c.508C>T, c.527C>G, c.1320C>G, c.1180C>T, c.236A>C, c.316A>G, c.323C>T, c.338T>C, c.358C>T, c.427T>C, c.462C>G, c.473C>T, c.506A>C, c.517A>G, c.738delG, c.296delG, c.441delC, c.596C[5>4] (std: c.596C[4] alt: c.600delC), c.610T[2>1] (std: c.610T[1] alt: c.611delT), c.1371C[2>1] (std: c.1371C[1] alt: c.1372delC), c.122C>A, c.227C>A, c.257G[2>3] (std: c.258dupG alt: c.258_259insG), c.257G>A, c.337A>G, c.349C>T, c.432-2A>G, c.432-2A>C, c.442G>T, c.451A>T, c.455G>C, c.472G>A, c.495C>G, c.509C>G, c.527C>T, c.583C>T, c.685+1G>A, c.694C>T, c.715dupA, c.736dupC, c.783G[6>7] (std:

c.788dupG alt: c.788_789insG), c.885C[2>3] (std: c.886dupC alt: c.886_887insC), c.984G[2>3] (std: c.985dupG alt: c.985_986insG), c.1123C>T, c.1198G>T, c.1242_1243insT, c.1493G[3>4] (std: c.1495dupG alt: c.1495_1496insG), c.1514_1515insC

Gene：

SPECC1L

NM ID：

NM_015330.3

Variant：

c.3293G>A

Gene：

SPTAN1

NM ID：

NM_001130438.2

Variant：

c.4828C>T, c.6616GAG[2>1] (std: c.6616GAG[1] alt: c.6619_6621delGAG)

Gene：

SRCAP

NM ID：

NM_006662.2

Variant：

c.7727C>A, c.8242C>T, c.7303C>T, c.7330C>T

Gene：

STAT3

NM ID：

NM_139276.2

Variant：

c.1988C>T, c.2147C>T, c.1979T>C, c.1970A>G, c.2117T>C, c.2144C>T, c.1909G>A, c.1414A>G, c.1139+1G>T, c.1268G>A, c.1859C>G, c.1384GTG[2>1] (std: c.1384GTG[1] alt: c.1387_1389delGTG), c.1397A>G, c.1397A>C, c.1261G>A, c.1924A>G, c.1243G>A, c.1032G>C, c.1144C>T, c.1145G>A, c.1003C>T, c.454C>T, c.1699A>G, c.2134T>C, c.1593A>T, c.1276T>C, c.995A>C, c.2137G>C, c.2132T>C, c.2129T>G, c.2125A>G, c.2116C>A, c.1994T>A, c.1974G>C, c.1970A>C, c.1913A>G, c.1865C>T, c.1863C>G, c.1858A>G, c.1850G>A, c.1591A>G, c.1407G>T, c.1406A>G, c.1310A>C, c.1309C>T, c.1294G>C, c.1166C>T, c.1152T>A, c.1151T>G, c.1151T>C, c.1144C>G, c.1140-2A>G, c.1110-1G>T, c.1025G>A, c.995A>T, c.994C>T

Gene：

STXBP1

NM ID：

NM_003165.3

Variant：

c.37+1_37+2insT, c.794+5G>A, c.1029+1G>A, c.1029+1G>T, c.794+1G>A, c.386CT[2>1] (std: c.386CT[1] alt: c.388_389delCT), c.416C>T, c.1359+1G>A, c.57_59delAAA, c.1277T>C, c.734A>G, c.87+1G>A, c.256TC[3>4] (std: c.260_261dupTC alt: c.261_262insTC), c.170-2A>G, c.1216C>T, c.1217G>A, c.1217G>T, c.364C>T, c.1461+1G>T, c.326-1G>T, c.1162C>T, c.1099C>T, c.847G>A, c.685C>T, c.703C>T, c.874C>T, c.704G>A, c.862T>C, c.875G>A, c.875G>T, c.539G>A, c.579-2A>G, c.578+1G>A, c.578+1G>T, c.1060T>C, c.157G>T, c.148dupA, c.1439C>T, c.1434G>A, c.1702+1G>A, c.569G>A,

c.1631G>T, c.1630G>T, c.568C>T, c.1651C>A, c.1651C>T, c.1427C>G, c.1611CAT[2>1] (std: c.1611CAT[1] alt: c.1614_1616delCAT), c.607C>T

Gene:

SYNGAP1

NM ID:

NM_006772.2

Variant:

c.1685C>T, c.1717C>T, c.1144G>T, c.1735C>T, c.823C[6>7] (std: c.828dupC alt: c.828_829insC), c.3416dupA, c.3277C>T, c.654_655delCT, c.3583-6G>A, c.2438delT, c.3408+1G>A, c.1822T[3>1] (std: c.1822T[1] alt: c.1823_1824delTT), c.333delA, c.1782C[2>1] (std: c.1782C[1] alt: c.1783delC), c.2755C>T, c.2383C[5>4] (std: c.2383C[4] alt: c.2387delC), c.2764C>T, c.3706C>T, c.3073C>T, c.1452C[2>1] (std: c.1452C[1] alt: c.1453delC), c.980T>C, c.2294+1G>A, c.2782C>T, c.3233_3236delTCAG, c.3718C>T, c.739C>T, c.2916delT, c.1861C>T, c.2899C>T, c.412A>T, c.403C>T, c.1252A[3>1] (std: c.1252A[1] alt: c.1253_1254delAA), c.427C>T, c.1630C>T, c.583G>C, c.490C>T, c.2104C>T, c.509G>A, c.2059C>T, c.1167_1168delAG, c.1030G>A

Gene:

TBL1XR1

NM ID:

NM_024665.4

Variant:

c.1043A>G, c.1108G>A, c.1337A>G

Gene:

TBX5

NM ID：

NM_000192.3

Variant：

c.242+1G>A, c.243-2A>G, c.668C>T, c.756-2A>G, c.205G>T, c.835C>T, c.588A[6>7] (std: c.593dupA alt: c.593_594insA), c.587C>A, c.709C>T, c.710G>A, c.192G>A, c.456delC, c.142C>T, c.101C[5>4] (std: c.101C[4] alt: c.105delC)

Gene：

TCF4

NM ID：

NM_001083962.1

Variant：

c.1146+3A>G, c.1146+1G>A, c.2010_2011delGA, c.1153C>T, c.1086G>A, c.759C>G, c.1167T[3>2] (std: c.1167T[2] alt: c.1169delT), c.1413G[2>1] (std: c.1413G[1] alt: c.1414delG), c.1486G>T, c.1486+1G>T, c.510AAAG[2>1] (std: c.510AAAG[1] alt: c.514_517delAAAG), c.1498G>T, c.923-2A>G, c.1916_1917delGA, c.1964C[2>3] (std: c.1965dupC alt: c.1965_1966insC), c.520C>T, c.1953TC[3>2] (std: c.1953TC[2] alt: c.1957_1958delTC), c.990G>A, c.1570C>T, c.655+1G>T, c.466C[4>3] (std: c.466C[3] alt: c.469delC), c.469C>T, c.1826T>C, c.1705C>T, c.656-1G>C, c.1876C>T, c.1720A>G, c.1719_1722dupCAAT, c.1841C>T, c.1739G>A, c.1738C>T, c.1727G>A, c.1726C>T, c.1733G>C, c.1733G>A

Gene：

TGFB2

NM ID：

NM_003238.3

Variant：

c.814A[8>9] (std: c.821dupA alt: c.821_822insA), c.576_577delAA, c.544C>T, c.391C>T, c.583G>T, c.905G>A, c.958C>T, c.904C>T, c.896G>A, c.895C>T

Gene：

TGFBR1

NM ID：

NM_004612.2

Variant：

c.1240C>T, c.700T>C, c.680AAG[2>1] (std: c.680AAG[1] alt: c.683_685delAAG), c.757A>G, c.934G>A, c.722C>T, c.1061T>C, c.1457T>C, c.1460G>A, c.1459C>T

Gene：

TGFBR2

NM ID：

NM_003242.5

Variant：

c.1378C>T, c.1379G>A, c.1336G>A, c.95-2A>G, c.1489C>T, c.1408T>G, c.1483C>T, c.1397-1G>A, c.1052G>A, c.1067G>C, c.1136A>T, c.1151A>G, c.1609C>T, c.1580C>T, c.1579G>A, c.1540T>C, c.1582C>T, c.1570G>A, c.1583G>A

Gene：

TRAF7

NM ID：

NM_032271.2

Variant：

c.1964G>A

Gene：

TRPS1

NM ID：

NM_014112.2

Variant：

c.2086C>T, c.2557C>T, c.1870C>T, c.2194C>T, c.2720T[6>7] (std: c.2725dupT alt: c.2725_2726insT), c.2794G>A, c.2762G>A, c.2795C>T, c.1630C>T, c.2893C>T, c.2761C>T

Gene：

TSC1

NM ID：

NM_000368.4

Variant：

c.664-15A>G, c.2215C>T, c.1431_1434delAGAA, c.737+3A>G, c.664-1G>A, c.2227C>T, c.737+1G>A, c.737G>A, c.671T>G, c.733C>T, c.107-1G>A, c.682C>T, c.645T[4>3] (std: c.645T[3] alt: c.648delT), c.645T[4>2] (std: c.645T[2] alt: c.647_648delTT), c.663+1G>A, c.738-2A>T, c.2389C>T, c.182T>G, c.182T>C, c.749T>A, c.2249G>A, c.1233C[5>4] (std: c.1233C[4] alt: c.1237delC), c.1959dupA, c.1997+1G>A, c.1960C>T, c.1029+1G>A, c.973C>T, c.1905AG[2>1] (std: c.1905AG[1] alt: c.1907_1908delAG), c.897CA[3>2] (std: c.897CA[2] alt: c.901_902delCA), c.901C>T, c.772G>T, c.1987G>T, c.1963C>T, c.989_990delTG, c.1252C[6>5] (std: c.1252C[5] alt: c.1257delC), c.1439-2A>G, c.989dupT, c.982C>T, c.211-2A>C, c.1902CA[2>1] (std: c.1902CA[1] alt: c.1904_1905delCA), c.211-1G>A, c.1884AAAG[2>1] (std: c.1884AAAG[1] alt: c.1888_1891delAAAG), c.2272C>T, c.2362G>T, c.2356C>T, c.2208+2T>A, c.2524C>T, c.1530_1531delCA, c.569G>C, c.2515_2518delGAGT, c.2283C>A,

c.1525C>T, c.2503-2A>G, c.2503-1G>C, c.325C>T, c.309G>A, c.2509_2512delAACA, c.2041+1G>A, c.2507C>G, c.2341C>T, c.2818C>T, c.1498C>T, c.492G>A, c.2626G>T, c.2074C>T, c.2293C>T, c.2023delG, c.1695C[3>2] (std: c.1695C[2] alt: c.1697delC), c.261dupA, c.2027G>A, c.1580_1581delAG, c.269TC[2>1] (std: c.269TC[1] alt: c.271_272delTC), c.1579C>T, c.2806C>T, c.2111_2112delAT, c.2144G[2>1] (std: c.2144G[1] alt: c.2145delG), c.445C>T, c.2716C>T, c.2692C>T, c.2666A[7>8] (std: c.2672dupA alt: c.2672_2673insA), c.2689C>T, c.1030-3C>G, c.611G>C, c.473T>C, c.350T>C, c.2666A[7>6] (std: c.2666A[6] alt: c.2672delA), c.2566G[4>3] (std: c.2566G[3] alt: c.2569delG), c.2530C>T, c.1781delT, c.1708_1709delAG, c.1453G>T, c.986CT[2>1] (std: c.986CT[1] alt: c.988_989delCT), c.657G[2>1] (std: c.657G[1] alt: c.658delG), c.2699AG[2>1] (std: c.2699AG[1] alt: c.2701_2702delAG), c.2698_2699delCA, c.2668A>T, c.2582delT, c.2503-2delA, c.2468AC[2>1] (std: c.2468AC[1] alt: c.2470_2471delAC), c.2363A[2>1] (std: c.2363A[1] alt: c.2364delA), c.2318C[2>1] (std: c.2318C[1] alt: c.2319delC), c.2287C>T, c.2263C>T, c.2155delC, c.2107CT[2>1] (std: c.2107CT[1] alt: c.2109_2110delCT), c.2022delC, c.1997+1G>C, c.1966G>T, c.1964delA, c.1958dupT, c.1955_1956delTG, c.1846delG, c.1839A[3>2] (std: c.1839A[2] alt: c.1841delA), c.1820T[5>3] (std: c.1820T[3] alt: c.1823_1824delTT), c.1799delA, c.1786T[3>2] (std: c.1786T[2] alt: c.1788delT), c.1615delT, c.1560delC, c.1550G[2>1] (std: c.1550G[1] alt: c.1551delG), c.1533delT, c.1488TG[2>1] (std: c.1488TG[1] alt: c.1490_1491delTG), c.1433A[2>1] (std: c.1433A[1] alt: c.1434delA), c.1421A[5>4] (std: c.1421A[4] alt: c.1425delA), c.1267GA[3>2] (std: c.1267GA[2] alt: c.1271_1272delGA), c.1250delC, c.1203delT, c.1119C>G, c.1027C>T, c.1019A[2>1] (std: c.1019A[1] alt: c.1020delA), c.989delT, c.988delC, c.913+1G>A, c.893dupC, c.832TC[2>1] (std: c.832TC[1] alt:

c.834_835delTC), c.829GT[2>1] (std: c.829GT[1] alt: c.831_832delGT), c.801dupA, c.748T[2>1] (std: c.748T[1] alt: c.749delT), c.745delA, c.591dupC, c.585C>A, c.562T[3>1] (std: c.562T[1] alt: c.563_564delTT), c.562T>G, c.533dupT, c.527_528delAT, c.508+1G>A, c.431CA[2>1] (std: c.431CA[1] alt: c.433_434delCA), c.433C>T, c.338T>A, c.203_204delAT, c.194A[2>1] (std: c.194A[1] alt: c.195delA), c.147C[2>1] (std: c.147C[1] alt: c.148delC), c.88A[3>2] (std: c.88A[2] alt: c.90delA), c.70delG, c.60TG[2>3] (std: c.62_63dupTG alt: c.63_64insTG), c.2698C>T, c.2650dupT, c.2566G[4>5] (std: c.2569dupG alt: c.2569_2570insG), c.2508A[3>4] (std: c.2510dupA alt: c.2510_2511insA), c.2503-1G>T, c.2498A[4>5] (std: c.2501dupA alt: c.2501_2502insA), c.2497C>T, c.2342A[1>3] (std: c.2342A[3] alt: c.2342_2343insAA), c.2332C>T, c.2299C>T, c.2283C>G, c.2250G>A, c.2173A[3>4] (std: c.2175dupA alt: c.2175_2176insA), c.2041+2T>C, c.2028G>A, c.1883T>A, c.1791A[2>4] (std: c.1791A[4] alt: c.1792_1793insAA), c.1773_1774dupGA, c.1759A>T, c.1729G>T, c.1717C>T, c.1676dupG, c.1654T[3>4] (std: c.1656dupT alt: c.1656_1657insT), c.1513TC[2>3] (std: c.1515_1516dupTC alt: c.1516_1517insTC), c.1432_1433dupGA, c.1411_1412insT, c.1331C>G, c.1118dupA, c.1030-1G>A, c.1014A[2>3] (std: c.1015dupA alt: c.1015_1016insA), c.1001C>A, c.988dupC, c.936C>A, c.891T>G, c.814G>T, c.748T[2>3] (std: c.749dupT alt: c.749_750insT), c.749T>G, c.745A>T, c.722A[2>3] (std: c.723dupA alt: c.723_724insA), c.709dupT, c.653A[2>3] (std: c.654dupA alt: c.654_655insA), c.647T>C, c.572T>G, c.572T>A, c.539T>C, c.495C>A, c.397G>T, c.395G>A, c.372dupT, c.308G>A, c.278T>G, c.272C>A, c.251C[2>3] (std: c.252dupC alt: c.252_253insC), c.215T>C, c.210+1G>A, c.196G>T, c.149T>C, c.107-2A>G

Gene：

TSC2

NM ID：

NM_000548.3

Variant：

c.225+1G>A, c.1258G>T, c.569dupA, c.138+2T>C, c.598C>T, c.133_136delCTGA, c.138+5G>A, c.135GA[3>2] (std: c.135GA[2] alt: c.139_140delGA), c.600-1G>A, c.730T>C, c.1283_1285delCCT, c.774+1G>A, c.3611G>A, c.4989+1G>A, c.487T[3>1] (std: c.487T[1] alt: c.488_489delTT), c.2743-9C>G, c.501G>A, c.500G>A, c.496C>T, c.1714C>T, c.2824G>T, c.2098-1G>A, c.2743-1G>C, c.4952A>G, c.649-1G>A, c.139-1G>A, c.975+1G>T, c.3696dupT, c.3693_3696delGTCT, c.4936G>A, c.2113G>A, c.3747_3748delGT, c.3787C[5>4] (std: c.3787C[4] alt: c.3791delC), c.658C>T, c.648+1G>A, c.2089T[2>3] (std: c.2090dupT alt: c.2090_2091insT), c.336+5G>C, c.4933T[3>1] (std: c.4933T[1] alt: c.4934_4935delTT), c.2769T[4>3] (std: c.2769T[3] alt: c.2772delT), c.2108G>A, c.3797dupT, c.4839CAT[2>1] (std: c.4839CAT[1] alt: c.4842_4844delCAT), c.1599+2T>C, c.1443+5G>C, c.3750C>G, c.3685C>T, c.1599+1G>A, c.2221-1G>A, c.2087G>A, c.3662C>A, c.4993C>T, c.2070C[2>1] (std: c.2070C[1] alt: c.2071delC), c.4685T>C, c.1195G>T, c.4846C>T, c.226-2A>G, c.4830G>A, c.4927A>C, c.2221-2A>G, c.4569+1G>A, c.2328C>A, c.1322G>A, c.336+1G>A, c.4823_4825delACT, c.2251C>T, c.848+1G>A, c.4005+1G>T, c.1953AG[4>3] (std: c.1953AG[3] alt: c.1959_1960delAG), c.2546-2A>G, c.2639+1G>A, c.2639+1G>C, c.5024C>A, c.5024C>T, c.337-1G>A, c.4813C>T, c.4762C>T, c.4918C>T, c.4471A[3>2] (std: c.4471A[2] alt: c.4473delA), c.1432C>T, c.976-15G>A, c.1444-2A>G, c.1257+1G>A, c.2355+2T>C, c.1111C>T, c.3610+1G>A, c.3610+1G>T, c.1783C>T, c.358A>T, c.2354_2355+2delAGGT, c.2545+5G>C, c.3398-1delG,

c.1255C>T, c.1864C>T, c.1792T>C, c.5034C>G, c.2353C>T, c.4909AAG[2>1] (std: c.4909AAG[1] alt: c.4912_4914delAAG), c.849-1G>A, c.1249C>T, c.826_827delAT, c.4573C>T, c.1108C>T, c.3598C>T, c.4493+1G>A, c.5056C>T, c.2356-2A>C, c.2194C>T, c.1929C>G, c.2531T>C, c.2590C>T, c.1839+1G>T, c.1348G>T, c.2356-1G>A, c.4620C>G, c.4662G>T, c.5138G>A, c.1085T>C, c.5140C>T, c.268C>T, c.2377G>T, c.3412C>T, c.4318C>T, c.1096G>T, c.5150T>C, c.3355C>T, c.1831C>T, c.3532C>T, c.2410T>C, c.4655_4657delAAG, c.2453TCA[3>2] (std: c.2453TCA[2] alt: c.2459_2461delTCA), c.3442C>T, c.5126C>G, c.5126C>T, c.5069-18A>G, c.3884-1G>C, c.4508A>C, c.2477T>C, c.1447G>T, c.4594C>T, c.4507C>T, c.4006-2A>G, c.4878_4881delCACC, c.4544_4547delACAA, c.5069-2A>G, c.5068+2T>C, c.1832G>A, c.5160+1G>A, c.4646A>G, c.1372C>T, c.4298C>A, c.1060C>T, c.4255C>T, c.4258_4261delTCAG, c.3094C>T, c.3095G>C, c.976-1G>A, c.3099C>A, c.2690T>C, c.2688G>A, c.1362-2A>G, c.5161-1G>A, c.5161-1G>C, c.4096G>T, c.2713C>T, c.4129C>T, c.2714G>A, c.4375C>T, c.5220G>A, c.3236C>A, c.5212delT, c.1507C>T, c.3180G>A, c.5228G>A, c.5258G[2>1] (std: c.5258G[1] alt: c.5259delG), c.1513C>T, c.5227C>T, c.3203C>T, c.5254C>T, c.3204TG[2>1] (std: c.3204TG[1] alt: c.3206_3207delTG), c.5286C[4>3] (std: c.5286C[3] alt: c.5289delC), c.3131+1G>A, c.4174C>T, c.3284+1G>A, c.4183C>T, c.478C>G, c.1599G>C, c.2197C>G, c.4807G>T, c.4942A>T, c.5160+4A>T, c.849-3T>G, c.976-3C>G, c.1001T>C, c.1001T>G, c.1347G>C, c.1789C>T, c.1796A>T, c.2114T>A, c.2150T>G, c.2198T>C, c.2713C>G, c.2726T>A, c.2747T>G, c.3082G>A, c.3106T>C, c.3178T>C, c.3182T>C, c.3596T>G, c.3608C>A, c.3884A>T, c.4489C>A, c.4490C>G, c.4601T>A, c.4604A>C, c.4643T>C, c.4700G>A, c.4708A>T, c.4711T>A, c.4735G>A, c.4751T>G, c.4780C>A, c.4787G>T, c.4790T>C, c.4828T>G, c.4859A>G, c.4868C>T, c.4928A>G,

c.4928A>T, c.4929C>G, c.4937T>G, c.4949A>G, c.4957T>C, c.5017G>T, c.5018T>A, c.5057A>C, c.5160+5G>T, c.1474C>T, c.1559AC[4>3] (std: c.1559AC[3] alt: c.1565_1566delAC), c.1760_1761delAT, c.1946+1G>A, c.3999C>A, c.4113TG[2>1] (std: c.4113TG[1] alt: c.4115_4116delTG), c.4178CT[2>1] (std: c.4178CT[1] alt: c.4180_4181delCT), c.4204G[4>3] (std: c.4204G[3] alt: c.4207delG), c.4418AG[3>2] (std: c.4418AG[2] alt: c.4422_4423delAG), c.4542A[3>2] (std: c.4542A[2] alt: c.4544delA), c.4641C[2>1] (std: c.4641C[1] alt: c.4642delC), c.5160+1G>T, c.43T[3>2] (std: c.43T[2] alt: c.45delT), c.100A>T, c.115delA, c.169delC, c.187delC, c.217T[3>2] (std: c.217T[2] alt: c.219delT), c.266T>A, c.334C>T, c.337-2A>G, c.357delC, c.413A[3>2] (std: c.413A[2] alt: c.415delA), c.438CA[2>1] (std: c.438CA[1] alt: c.440_441delCA), c.461C[2>1] (std: c.461C[1] alt: c.462delC), c.509_510insAT, c.516_517delGT, c.528C[2>1] (std: c.528C[1] alt: c.529delC), c.602delT, c.606delC, c.632C[2>1] (std: c.632C[1] alt: c.633delC), c.644_645delTA, c.648+1G>T, c.701AG[2>1] (std: c.701AG[1] alt: c.703_704delAG), c.727CT[2>1] (std: c.727CT[1] alt: c.729_730delCT), c.797C[3>2] (std: c.797C[2] alt: c.799delC), c.826delA, c.855C>A, c.899_900insT, c.905CT[3>2] (std: c.905CT[2] alt: c.909_910delCT), c.908T>C, c.949_950insTA, c.975G[2>1] (std: c.975G[1] alt: c.976delG), c.1000GT[2>1] (std: c.1000GT[1] alt: c.1002_1003delGT), c.1008T>G, c.1094_1095delTC, c.1142G[2>1] (std: c.1142G[1] alt: c.1143delG), c.1181TG[3>2] (std: c.1181TG[2] alt: c.1185_1186delTG), c.1228_1229insG, c.1323G>A, c.1330A[2>1] (std: c.1330A[1] alt: c.1331delA), c.1361+1G>A, c.1390A[2>1] (std: c.1390A[1] alt: c.1391delA), c.1407delG, c.1409C[2>1] (std: c.1409C[1] alt: c.1410delC), c.1488C[4>3] (std: c.1488C[3] alt: c.1491delC), c.1528C>T, c.1599+2T>A, c.1616delT, c.1640A[3>4] (std: c.1642dupA alt: c.1642_1643insA), c.1692C[2>1]

(std: c.1692C[1] alt: c.1693delC), c.1795A[2>1] (std: c.1795A[1] alt: c.1796delA), c.1839+1G>A, c.1960_1961delGG, c.1976_1977insA, c.2074delG, c.2157T>A, c.2163delG, c.2181T[2>1] (std: c.2181T[1] alt: c.2182delT), c.2235_2236delGA, c.2240_2241delTG, c.2394C[2>1] (std: c.2394C[1] alt: c.2395delC), c.2417_2418dupTG, c.2433delC, c.2452dupA, c.2530delC, c.2538C[2>1] (std: c.2538C[1] alt: c.2539delC), c.2638A[2>1] (std: c.2638A[1] alt: c.2639delA), c.2639+1G>T, c.2641T[3>2] (std: c.2641T[2] alt: c.2643delT), c.2649_2650dupGT, c.2670delT, c.2689T[2>1] (std: c.2689T[1] alt: c.2690delT), c.2737_2738delAC, c.2766delG, c.2780C[5>4] (std: c.2780C[4] alt: c.2784delC), c.2785delG, c.2814_2815delTA, c.2816_2817dupGT, c.3097_3098dupTA, c.3099delC, c.3127A[2>1] (std: c.3127A[1] alt: c.3128delA), c.3131G>C, c.3131+2T>C, c.3213A[2>1] (std: c.3213A[1] alt: c.3214delA), c.3240dupA, c.3291C[4>5] (std: c.3294dupC alt: c.3294_3295insC), c.3312_3313delGA, c.3316A>T, c.3318G[2>3] (std: c.3319dupG alt: c.3319_3320insG), c.3336G[2>3] (std: c.3337dupG alt: c.3337_3338insG), c.3349G[3>2] (std: c.3349G[2] alt: c.3351delG), c.3383delT, c.3389C[2>1] (std: c.3389C[1] alt: c.3390delC), c.3396G[6>5] (std: c.3396G[5] alt: c.3401delG), c.3397+1G>A, c.3518C[3>2] (std: c.3518C[2] alt: c.3520delC), c.3572C[3>2] (std: c.3572C[2] alt: c.3574delC), c.3581G>A, c.3599G>C, c.3611-1delG, c.3695delC, c.3755C>A, c.3884-1G>A, c.3982dupA, c.3983G[2>1] (std: c.3983G[1] alt: c.3984delG), c.4013_4014delCA, c.4014dupA, c.4027G>T, c.4030G>T, c.4051G>T, c.4176_4177delGA, c.4235_4236delCT, c.4236dupT, c.4255delC, c.4264delA, c.4277A[3>2] (std: c.4277A[2] alt: c.4279delA), c.4344dupC, c.4471A[3>1] (std: c.4471A[1] alt: c.4472_4473delAA), c.4503C[2>1] (std: c.4503C[1] alt: c.4504delC), c.4537G>T, c.4569+2T>C, c.4577C[2>1] (std: c.4577C[1] alt: c.4578delC), c.4590G[2>1] (std: c.4590G[1] alt: c.4591delG),

c.4630A>T, c.4662+1G>T, c.4663-2_4663-1delAG, c.4703C[2>1] (std: c.4703C[1] alt: c.4704delC), c.4709G[2>1] (std: c.4709G[1] alt: c.4710delG), c.4716G[2>3] (std: c.4717dupG alt: c.4717_4718insG), c.4743delC, c.4761C[2>3] (std: c.4762dupC alt: c.4762_4763insC), c.4854delC, c.4870_4871delCT, c.4909_4910delAA, c.4923G[3>2] (std: c.4923G[2] alt: c.4925delG), c.4936GT[2>1] (std: c.4936GT[1] alt: c.4938_4939delGT), c.5073G[2>3] (std: c.5074dupG alt: c.5074_5075insG), c.5135C[3>1] (std: c.5135C[1] alt: c.5136_5137delCC), c.5160+2T>G, c.5168CA[2>1] (std: c.5168CA[1] alt: c.5170_5171delCA), c.5172G[2>3] (std: c.5173dupG alt: c.5173_5174insG), c.5402_5403delAG, c.5418TG[3>2] (std: c.5418TG[2] alt: c.5422_5423delTG), c.3G>A, c.34A>T, c.43T[3>4] (std: c.45dupT alt: c.45_46insT), c.108G[2>3] (std: c.109dupG alt: c.109_110insG), c.138+1G>A, c.225+2T>A, c.246G>A, c.337-1G>T, c.403dupG, c.451A>T, c.465C>G, c.469G>T, c.600-2A>G, c.618C>A, c.684C>A, c.707dupT, c.731G>A, c.759C>A, c.760G>T, c.767dupG, c.768C>A, c.770G>A, c.775-1G>A, c.831C[2>3] (std: c.832dupC alt: c.832_833insC), c.849-2A>T, c.868C[4>5] (std: c.871dupC alt: c.871_872insC), c.880G>A, c.905CT[3>4] (std: c.909_910dupCT alt: c.910_911insCT), c.911G>A, c.972C>G, c.979_980dupAT, c.1042A>T, c.1117C>T, c.1257+2T>C, c.1258-2A>C, c.1258-1G>A, c.1327C>T, c.1336C>T, c.1472C>A, c.1563dupA, c.1624C[5>6] (std: c.1628dupC alt: c.1628_1629insC), c.1661C>A, c.1790A>G, c.1794C>G, c.1820C>A, c.1840-2A>T, c.1841C>A, c.1844T>C, c.1901_1902insAT, c.1942dupT, c.1965dupT, c.1969A>T, c.2083C>T, c.2087_2088insAA, c.2098-2A>G, c.2098G>A, c.2103dupT, c.2106dupC, c.2109G>A, c.2158A>T, c.2221-2A>C, c.2225C>G, c.2294C[2>4] (std: c.2294C[4] alt: c.2295_2296insCC), c.2355+1G>T, c.2370C>G, c.2375T>G, c.2404dupA, c.2430dupC, c.2461A>T, c.2489T>G, c.2491_2492insTA,

c.2531T>G, c.2540T>C, c.2546-2A>T, c.2661T>A, c.2665G>C, c.2666C>T, c.2687G>A, c.2707C[3>4] (std: c.2709dupC alt: c.2709_2710insC), c.2716A[2>3] (std: c.2717dupA alt: c.2717_2718insA), c.2743-2A>C, c.2743-2A>G, c.2768dupC, c.2780C[5>6] (std: c.2784dupC alt: c.2784_2785insC), c.2785G>T, c.2974C>T, c.3028C>T, c.3076dupT, c.3098dupA, c.3099C>G, c.3212C>G, c.3265C>T, c.3397+2T>G, c.3425dupT, c.3574C>T, c.3575dupA, c.3755C>G, c.4037C>A, c.4289G>A, c.4397C>A, c.4438_4439insA, c.4439T>A, c.4440A[3>4] (std: c.4442dupA alt: c.4442_4443insA), c.4493G>C, c.4493G>A, c.4494-1_4494insC, c.4515C>A, c.4606C>T, c.4620C>A, c.4663-1G>A, c.4672G>A, c.4700G>T, c.4713C>G, c.4744dupA, c.4779C>G, c.4850-2A>C, c.4850-1G>A, c.4858C>T, c.4871T>C, c.4925G>A, c.4989G[3>4] (std: c.4991dupG alt: c.4991_4992insG), c.4989+1_4989+2insA, c.5032dupT, c.5043C>G, c.5112dupT, c.5160+1_5160+2insT, c.5170C>T, c.5208C>A, c.5246G[2>3] (std: c.5247dupG alt: c.5247_5248insG), c.5388dupC, c.5407T[2>3] (std: c.5408dupT alt: c.5408_5409insT)

Gene：

TUBA1A

NM ID：

NM_006009.3

Variant：

c.367C>T, c.368G>A, c.5G>A, c.344T>C, c.352G>A, c.17C>G, c.698A>G, c.712A>G, c.715A>C, c.629A>G, c.641G>T, c.641G>A, c.283G>T, c.652G>A, c.269A>G, c.379G>A, c.1274T>A, c.1246G>A, c.1304T>C, c.481T>G, c.1025A>C, c.787C>A, c.1226T>C, c.1225G>A, c.152C>T, c.1264C>T, c.1265G>A, c.1256C>T, c.139G>C, c.995T>C, c.449C>T, c.455T>A, c.424G>T, c.424G>A, c.167C>T, c.427G>A, c.1204C>T, c.1204C>A, c.1205G>T,

c.1205G>A, c.518C>T, c.162T>A, c.190C>T, c.986A>G, c.562A>C, c.791G>A, c.790C>T, c.790C>G, c.970G>C, c.53A>G, c.1190T>C, c.1055A>T, c.1096G>A, c.1076C>T, c.959G>A, c.1091C>G, c.808G>T, c.815A>G, c.1168C>T, c.1168C>G, c.1144A>G, c.79G>C, c.1160C>T, c.1169G>C, c.1169G>A, c.1148C>T, c.1148C>A, c.1129A>G, c.1105G>A, c.920C>T, c.916G>T, c.919C>T, c.908T>G, c.856C>T, c.887T>C, c.878A>G

Gene:

TUBB

NM ID:

NM_178014.2

Variant:

c.1201G>A

Gene:

TUBB2A

NM ID:

NM_001069.2

Variant:

c.292G>A, c.1033A>T, c.394G>A, c.728C>T

Gene:

TUBB4A

NM ID:

NM_006087.2

Variant:

c.5G>A, c.941C>T, c.467G>T, c.286G>A, c.535G>C, c.533C>T, c.1228G>A, c.785G>A, c.763G>A, c.1172G>A, c.745G>A

Gene：

TWIST1

NM ID：

NM_000474.3

Variant：

c.301C>T, c.309C>G, c.346C>G, c.376G>T, c.433A>G

Gene：

WDR45

NM ID：

NM_007075.3

Variant：

c.1005AT[2>1] (std: c.1005AT[1] alt: c.1007_1008delAT), c.1030delT, c.235+1G>A, c.1032dupC, c.749CCT[2>1] (std: c.749CCT[1] alt: c.752_754delCCT), c.374_375delAT, c.409T[3>4] (std: c.411dupT alt: c.411_412insT), c.183C>A, c.400C>T, c.2T>A, c.873C>G, c.830+1G>C, c.830+1G>A, c.698G>A, c.700C>T, c.19C>T, c.666CA[3>2] (std: c.666CA[2] alt: c.670_671delCA), c.587_588delTA, c.614G>A, c.46C>T, c.52C>T, c.519+1_519+3delGTG

Gene：

ZBTB20

NM ID：

NM_001164342.1

Variant：

c.1811A>C, c.1817A>C, c.1939A>C, c.1873A>G, c.1760T>G

Gene：

ZC4H2

NM ID：

NM_018684.3

Variant：

c.243_246delACAA, c.631C>T, c.199C>T, c.637C>T, c.225+5G>A, c.592C>T, c.412C>T, c.427C>T

Gene：

ZEB2

NM ID：

NM_014795.3

Variant：

c.823C>T, c.1640C>G, c.855AG[2>1] (std: c.855AG[1] alt: c.857_858delAG), c.1489C>T, c.73+2T>C, c.3046C>T, c.904C>T, c.1421A[6>7] (std: c.1426dupA alt: c.1426_1427insA), c.2894T>A, c.1381C>T, c.3218A>G, c.980C>A, c.2499A[3>2] (std: c.2499A[2] alt: c.2501delA), c.2701C>T, c.2083C>T, c.1027C>T, c.1956C>A, c.2761C>T, c.1102C>T, c.674C>A, c.1876G>T, c.289delT