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|--------|-------------|---|
|        |             | c.133C>T, c.2389C>T, c.5167C>T, c.1885C>T, c.1660C>T  |
| NOTCH2 | NM_024408.3 | c.6424_6427delTCTG, c.6853C>T, c.6787C>T  |
| NR2F1  | NM_005654.4 | c.335G>A, c.317G>A, c.2T>C, c.382T>C, c.425G>A, c.425G>T  |
| NRAS   | NM_002524.4 | c.71T>A, c.179G>A, c.149C>T, c.182A>G, c.38G>A, c.35G>T, c.34G>C, c.34G>A   |
| NSD1   | NM_022455.4 | 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 ) |
| NSDHL  | NM_015922.2 | c.1038_1041dupCATG, c.208C>T  |
| PACS1  | NM_018026.3 | c.607C>T  |
| PIK3CA | NM_006218.2 | 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  |
| PIK3R2  | NM_005027.2 | c.1117G>A, c.1694C>G   |
| PPP2R1A | NM_014225.5 | c.773G>A, c.548G>A, c.536C>T, c.544C>T, c.547C>T, c.656C>T   |
| PPP2R5D | NM_006245.3 | 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  |
| PRKAR1A | NM_002734.3 | 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  |
| PTPN11  | NM_002834.3 | 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 |
| PURA    | NM_005859.4 | 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 )  |
| RAD21   | NM_006265.2    | c.1306C>T   |
| RAF1    | NM_002880.3    | 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  |
| RERE    | NM_012102.3    | c.4304A>T, c.4304A>G, c.4293C>A, c.3146C>T  |
| RIT1    | NM_006912.5    | 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  |
| RPS6KA3 | NM_004586.2    | 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   |
| RUNX2   | NM_001024630.3 | c.674G>A, c.673C>T, c.577C>T, c.569G>A, c.568C>T, c.1171C>T, c.278T>A, c.1085C>T  |
| SATB2   | NM_015265.3    | 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   |
| SCN1A   | NM_001165963.1 | 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, |

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|--|--|---|
|  |  | <p>c.2946+5G&gt;A, c.4219C&gt;T, c.1113T[6&gt;5] (std: c.1113T[5] alt: c.1118delT ), c.4223G&gt;A, c.4002+1G&gt;T, c.3985C&gt;T, c.602+1G&gt;T, c.602+1G&gt;C, c.602+1G&gt;A, c.3977C&gt;A, c.2946G[2&gt;1] (std: c.2946G[1] alt: c.2947delG ), c.4384T&gt;C, c.1112C&gt;T, c.4757G&gt;A, c.434T&gt;C, c.505T&gt;C, c.4762T&gt;C, c.2946+1G&gt;T, c.3968C&gt;A, c.580G&gt;A, c.3106C&gt;T, c.1662+2T&gt;C, c.4477-2A&gt;G, c.425GT[3&gt;2] (std: c.425GT[2] alt: c.429_430delGT ), c.4168G&gt;A, c.1662+3A&gt;G, c.3860T&gt;C, c.575G&gt;A, c.1662+1G&gt;T, c.1662+1G&gt;A, c.3879+1G&gt;T, c.2615C&gt;A, c.3818C&gt;T, c.3829C&gt;T, c.749T&gt;A, c.1096G&gt;C, c.563A&gt;T, c.1087A&gt;C, c.2213G&gt;A, c.264+4_264+7delAGTG, c.2624C&gt;T, c.748G&gt;A, c.4133A&gt;C, c.568T&gt;C, c.264+5G&gt;C, c.264+5G&gt;A, c.3864CTT[2&gt;1] (std: c.3864CTT[1] alt: c.3867_3869delCTT ), c.769T&gt;C, c.3295G&gt;T, c.3858G&gt;A, c.811_815dupGGCAA, c.3850T&gt;C, c.763GT[2&gt;1] (std: c.763GT[1] alt: c.765_766delGT ), c.2299_2302dupGACC, c.3812G&gt;A, c.3370T[3&gt;1] (std: c.3370T[1] alt: c.3371_3372delTT ), c.677C&gt;T, c.539T&gt;A, c.2935G&gt;A, c.680T&gt;G, c.3496C&gt;T, c.777C&gt;A, c.4853-1G&gt;C, c.3778A&gt;C, c.675G&gt;C, c.3852G&gt;A, c.5744_5748delAGGAA, c.530G&gt;T, c.530G&gt;A, c.2956C&gt;T, c.4862T&gt;C, c.251A&gt;G, c.1378C&gt;T, c.3880-2A&gt;G, c.1076A&gt;T, c.1076A&gt;G, c.4495T&gt;C, c.4879_4883dupAAGTA, c.661C&gt;G, c.2348T[4&gt;2] (std: c.2348T[2] alt: c.2350_2351delTT ), c.2354T&gt;C, c.2928G&gt;A, c.3880-1G&gt;T, c.2353A&gt;G, c.3940TC[3&gt;2] (std: c.3940TC[2] alt: c.3944_3945delTC ), c.4352C&gt;T, c.4359T&gt;G, c.243C&gt;A, c.1624C&gt;T, c.3705+1G&gt;A, c.5719ACT[3&gt;2] (std: c.5719ACT[2] alt: c.5725_5727delACT ), c.5726C&gt;T, c.5741_5742delAA, c.1348C&gt;T, c.664C&gt;T, c.235G&gt;A, c.5765T&gt;C, c.5734C&gt;T, c.1277A&gt;G, c.5710C&gt;T, c.5708_5711dupATCA, c.1049TG[4&gt;3] (std: c.1049TG[3] alt: c.1055_1056delTG ), c.4913T&gt;A, c.4633A&gt;G, c.4521C&gt;G, c.1665T[3&gt;2] (std: c.1665T[2] alt: c.1667delT ), c.4906C&gt;T, c.4907G&gt;A, c.2131C&gt;T, c.2665G&gt;A, c.1261G&gt;A, c.1264G&gt;A, c.2669T&gt;C, c.4581+1G&gt;A, c.2134C&gt;T, c.1259C&gt;T, c.2678T&gt;A, c.3629C&gt;A, c.4529C&gt;T, c.3615G&gt;A, c.603-2A&gt;G, c.652T&gt;C, c.2904C&gt;A, c.603-1G&gt;A, c.1177C&gt;T, c.4573C&gt;T, c.3656G&gt;A, c.1178G&gt;A, c.3610T&gt;C, c.1702C&gt;T, c.3611G&gt;A, c.3612G&gt;A, c.4934G&gt;A, c.2893C&gt;T, c.4933C&gt;T, c.4942C&gt;T, c.4943G&gt;A, c.3637C&gt;T, c.4549A&gt;T, c.4549A[6&gt;7] (std: c.4554dupA</p> |
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|-------|-------------|---|
|       |             | <p>alt: c.4554_4555insA ), c.4549A[6&gt;5] (std: c.4549A[5] alt: c.4554delA ), c.2791C&gt;T, c.4555C&gt;T, c.2792G&gt;T, c.2792G&gt;C, c.2792G&gt;A, c.2796G&gt;A, c.2802G&gt;A, c.2867T&gt;C, c.4547C&gt;A, c.2875T&gt;C, c.2876G&gt;A, c.2860G&gt;A, c.2819C&gt;T, c.2855G&gt;A, c.2831T&gt;C, c.2495G&gt;A, c.2836C&gt;T, c.2815C&gt;T, c.4543G&gt;A, c.2854T&gt;G, c.1506GAAAA[2&gt;1] (std: c.1506GAAAA[1] alt: c.1511_1515delGAAAA ), c.2849G&gt;A, c.1516C&gt;T, c.121A[6&gt;5] (std: c.121A[5] alt: c.126delA ), c.1738C&gt;T, c.2837G&gt;A, c.4968C&gt;G, c.5532AAAC[2&gt;1] (std: c.5532AAAC[1] alt: c.5536_5539delAAAC ), c.5536A&gt;T, c.2569delG, c.5555T&gt;C, c.5674C&gt;T, c.4970G&gt;A, c.4997C&gt;T, c.2589+3A&gt;T, c.5000T&gt;C, c.2575C&gt;T, c.2589+1G&gt;T, c.2576G&gt;A, c.4985C&gt;T, c.5261G&gt;A, c.5164A&gt;G, c.5506G&gt;T, c.5162C&gt;A, c.2585G&gt;T, c.2585G&gt;A, c.2584C&gt;T, c.5468T&gt;C, c.5014A&gt;C, c.5306A&gt;G, c.4973C&gt;T, c.4979T&gt;C, c.5422T&gt;C, c.5656C&gt;T, c.5048T&gt;C, c.5168C&gt;T, c.5006C&gt;A, c.5010_5013delGTTT, c.5334delG, c.5434T&gt;G, c.5435G&gt;A, c.5436G&gt;A, c.5218G&gt;A, c.5222G&gt;C, c.5222G&gt;A, c.5346C&gt;G, c.5347G&gt;A, c.5348C&gt;T, c.1804G&gt;T, c.1876A&gt;G, c.1834C&gt;T, c.1837C&gt;T</p> |
| SCN2A | NM_021007.2 | <p>c.4303C&gt;T, c.4264A&gt;G, c.304C&gt;T, c.605+1G&gt;A, c.605C&gt;T, c.386+2T&gt;C, c.4223T&gt;C, c.4446+9A&gt;G, c.408G&gt;A, c.3967A&gt;G, c.3964G&gt;A, c.1147C&gt;G, c.3956G&gt;A, c.3956G&gt;T, c.823C&gt;T, c.3955C&gt;T, c.3947C&gt;T, c.4879G&gt;A, c.4877G&gt;A, c.4886G&gt;A, c.3977T&gt;A, c.4123T&gt;G, c.4766A&gt;G, c.4468A&gt;G, c.2715G&gt;C, c.2877C&gt;A, c.2995G&gt;A, c.4036A&gt;G, c.4787T&gt;G, c.2722A&gt;G, c.4025T&gt;C, c.781G&gt;A, c.3631G&gt;A, c.2558G&gt;A, c.4904G&gt;A, c.668G&gt;A, c.4687C&gt;G, c.2674G&gt;A, c.751G&gt;A, c.5645G&gt;A, c.2960G&gt;T, c.5644C&gt;G, c.647T&gt;G, c.2548C&gt;T, c.2687C&gt;T, c.1289A&gt;G, c.1312G&gt;A, c.718G&gt;A, c.2809C&gt;T, c.2774T&gt;C, c.2566C&gt;T, c.4633A&gt;G, c.719C&gt;T, c.2567G&gt;A, c.2810G&gt;A, c.620T&gt;C, c.2627A&gt;G, c.1819C&gt;T, c.1827CT[3&gt;2] (std: c.1827CT[2] alt: c.1831_1832delCT), c.5308A&gt;T, c.4949T&gt;C, c.5317G&gt;A, c.5318C&gt;T, c.4976C&gt;T</p>   |
| SCN8A | NM_014191.3 | <p>c.2300C&gt;T, c.2287A&gt;G, c.2537T&gt;C, c.4408C&gt;A, c.4819G&gt;A, c.632T&gt;C, c.641G&gt;A, c.4850G&gt;A, c.4859G&gt;T, c.2879T&gt;A, c.614C&gt;A, c.4426G&gt;A, c.4423G&gt;A, c.2549G&gt;A, c.5279T&gt;C, c.4447G&gt;A, c.5302A&gt;G, c.5630A&gt;G, c.2603T&gt;C, c.2620G&gt;A,</p>   |



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|----------|----------------|--|
|          |                | 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   |
| SETBP1   | NM_015559.2    | c.1765C>T, c.1873C>T, c.1876C>T, c.1630C>T, c.2612T>C, c.2608G>A, c.2602G>A  |
| SETD2    | NM_014159.6    | c.4449AGAA[3>2] (std:c.4449AGAA[2] alt: c.4457_4460delAGAA )   |
| SETD5    | NM_001080517.1 | 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  |
| SHANK3   | NM_033517.1    | 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   |
| SHOC2    | NM_007373.3    | c.517A>G, c.4A>G   |
| SKI      | NM_003036.3    | c.103C>T, c.100G>A, c.100G>T, c.349G>C, c.347G>A, c.94C>G  |
| SLC25A24 | NM_213651.2    | c.593G>A, c.592C>T   |
| SMAD3    | NM_005902.3    | 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 |

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| SMAD4   | NM_005359.5    | 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 )  |
| SMARCA2 | NM_003070.3    | 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  |
| SMARCA4 | NM_001128849.1 | 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  |
| SMARCB1 | NM_003073.3    | 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   |
| SMARCE1 | NM_003079.4    | c.751C>T, c.715C>T, c.218A>G, c.314G>A, c.624_627delTGAG   |
| SMC1A   | NM_006306.2    | 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   |
| SMC3    | NM_005445.3    | c.3373C>G, c.1942A>G   |

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| SOS1    | NM_005633.3    | 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   |
| SOS2    | NM_006939.2    | c.1127C>G, c.800T>G, c.800T>C, c.800T>A, c.791C>A   |
| SOX9    | NM_000346.3    | 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 |
| SPECC1L | NM_015330.3    | c.3293G>A   |
| SPTAN1  | NM_001130438.2 | c.4828C>T, c.6616GAG[2>1] (std: c.6616GAG[1] alt: c.6619_6621delGAG )   |
| SRCAP   | NM_006662.2    | c.7727C>A, c.8242C>T, c.7303C>T, c.7330C>T  |

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| STAT3   | NM_139276.2 | <p>c.1988C&gt;T, c.2147C&gt;T, c.1979T&gt;C, c.1970A&gt;G, c.2117T&gt;C, c.2144C&gt;T, c.1909G&gt;A, c.1414A&gt;G, c.1139+1G&gt;T, c.1268G&gt;A, c.1859C&gt;G, c.1384GTG[2&gt;1] (std: c.1384GTG[1] alt: c.1387_1389delGTG ), c.1397A&gt;G, c.1397A&gt;C, c.1261G&gt;A, c.1924A&gt;G, c.1243G&gt;A, c.1032G&gt;C, c.1144C&gt;T, c.1145G&gt;A, c.1003C&gt;T, c.454C&gt;T, c.1699A&gt;G, c.2134T&gt;C, c.1593A&gt;T, c.1276T&gt;C, c.995A&gt;C, c.2137G&gt;C, c.2132T&gt;C, c.2129T&gt;G, c.2125A&gt;G, c.2116C&gt;A, c.1994T&gt;A, c.1974G&gt;C, c.1970A&gt;C, c.1913A&gt;G, c.1865C&gt;T, c.1863C&gt;G, c.1858A&gt;G, c.1850G&gt;A, c.1591A&gt;G, c.1407G&gt;T, c.1406A&gt;G, c.1310A&gt;C, c.1309C&gt;T, c.1294G&gt;C, c.1166C&gt;T, c.1152T&gt;A, c.1151T&gt;G, c.1151T&gt;C, c.1144C&gt;G, c.1140-2A&gt;G, c.1110-1G&gt;T, c.1025G&gt;A, c.995A&gt;T, c.994C&gt;T</p> |
| STXBP1  | NM_003165.3 | <p>c.37+1_37+2insT, c.794+5G&gt;A, c.1029+1G&gt;A, c.1029+1G&gt;T, c.794+1G&gt;A, c.386CT[2&gt;1] (std: c.386CT[1] alt: c.388_389delCT ), c.416C&gt;T, c.1359+1G&gt;A, c.57_59delAAA, c.1277T&gt;C, c.734A&gt;G, c.87+1G&gt;A, c.256TC[3&gt;4] (std: c.260_261dupTC alt: c.261_262insTC ), c.170-2A&gt;G, c.1216C&gt;T, c.1217G&gt;A, c.1217G&gt;T, c.364C&gt;T, c.1461+1G&gt;T, c.326-1G&gt;T, c.1162C&gt;T, c.1099C&gt;T, c.847G&gt;A, c.685C&gt;T, c.703C&gt;T, c.874C&gt;T, c.704G&gt;A, c.862T&gt;C, c.875G&gt;A, c.875G&gt;T, c.539G&gt;A, c.579-2A&gt;G, c.578+1G&gt;A, c.578+1G&gt;T, c.1060T&gt;C, c.157G&gt;T, c.148dupA, c.1439C&gt;T, c.1434G&gt;A, c.1702+1G&gt;A, c.569G&gt;A, c.1631G&gt;T, c.1630G&gt;T, c.568C&gt;T, c.1651C&gt;A, c.1651C&gt;T, c.1427C&gt;G, c.1611CAT[2&gt;1] (std: c.1611CAT[1] alt: c.1614_1616delCAT ), c.607C&gt;T</p>           |
| SYNGAP1 | NM_006772.2 | <p>c.1685C&gt;T, c.1717C&gt;T, c.1144G&gt;T, c.1735C&gt;T, c.823C[6&gt;7] (std: c.828dupC alt: c.828_829insC ), c.3416dupA, c.3277C&gt;T, c.654_655delCT, c.3583-6G&gt;A, c.2438delT, c.3408+1G&gt;A, c.1822T[3&gt;1] (std: c.1822T[1] alt: c.1823_1824delTT ), c.333delA, c.1782C[2&gt;1] (std: c.1782C[1] alt: c.1783delC ), c.2755C&gt;T, c.2383C[5&gt;4] (std: c.2383C[4] alt: c.2387delC ), c.2764C&gt;T, c.3706C&gt;T, c.3073C&gt;T, c.1452C[2&gt;1] (std: c.1452C[1] alt: c.1453delC ), c.980T&gt;C, c.2294+1G&gt;A, c.2782C&gt;T, c.3233_3236delTCAG, c.3718C&gt;T, c.739C&gt;T, c.2916delT, c.1861C&gt;T, c.2899C&gt;T, c.412A&gt;T, c.403C&gt;T, c.1252A[3&gt;1] (std: c.1252A[1] alt: c.1253_1254delAA ), c.427C&gt;T, c.1630C&gt;T, c.583G&gt;C, c.490C&gt;T, c.2104C&gt;T, c.509G&gt;A, c.2059C&gt;T, c.1167_1168delAG, c.1030G&gt;A</p>                    |

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| TBL1XR1 | NM_024665.4    | c.1043A>G, c.1108G>A, c.1337A>G  |
| TBX5    | NM_000192.3    | 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 )  |
| TCF4    | NM_001083962.1 | 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 |
| TGFB2   | NM_003238.3    | 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  |
| TGFBR1  | NM_004612.2    | 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  |
| TGFBR2  | NM_003242.5    | 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  |
| TRAF7   | NM_032271.2    | c.1964G>A  |
| TRPS1   | NM_014112.2    | 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   |

















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| TUBA1A | NM_006009.3    | 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 |
| TUBB   | NM_178014.2    | c.1201G>A   |
| TUBB2A | NM_001069.2    | c.292G>A, c.1033A>T, c.394G>A, c.728C>T   |
| TUBB4A | NM_006087.2    | 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  |
| TWIST1 | NM_000474.3    | c.301C>T, c.309C>G, c.346C>G, c.376G>T, c.433A>G  |
| WDR45  | NM_007075.3    | 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  |
| ZBTB20 | NM_001164342.1 | c.1811A>C, c.1817A>C, c.1939A>C, c.1873A>G, c.1760T>G   |
| ZC4H2  | NM_018684.3    | 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  |
| ZEB2   | NM_014795.3    | 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   |