

| ID | Rf.Nr | M/F | SMCHD1 mutationcDNA (NM_015295.2) | status | 450K array | RRBS | qPCR | ChIP |
|-------|------------|-----|--------------------------------------|------------------|------------|------|------|------|
| w412 | Rf7.204 | F | none | Control | x | | | |
| w1848 | Rf28.102 | F | none | Control | x | | | |
| r450 | Rf204.306 | F | none | Control | x | | | |
| r74 | Rf208.318 | F | none | Control | x | | | |
| r285 | Rf209.210 | F | none | Control | x | | | |
| r352 | Rf210.104 | F | none | Control | x | | | |
| r63 | Rf268.1 | F | none | Control | x | | | |
| w1825 | Rf300.5 | F | none | Control | x | | | |
| r264 | Rf385.302 | F | none | Control | x | | | |
| r322 | Rf400.2 | F | none | Control | x | | | |
| r650 | Rf649.4 | F | none | Control | x | | | |
| r860 | Rf739.3 | F | none | Control | x | | | |
| b59 | Rf696.2 | F | none | Control | x | | | |
| r795 | Rf742.3 | F | none | Control | x | | | |
| r977 | Rf844.4 | F | none | Control | x | | | |
| b46 | Rf909.2 | F | none | Control | x | | | |
| r997 | Rf910.3 | F | none | Control | x | | | |
| b61 | Rf952.2 | F | none | Control | x | | | |
| b93 | Rf969.1 | F | none | Control | x | | | |
| b323 | Rf1021.5 | F | none | Control | x | | | |
| b288 | Rf1110.2 | F | none | Control | x | | | |
| b480 | Rf1126.202 | F | none | Control | x | | | |
| b567 | Rf1232.4 | F | none | Control | x | | | |
| b609 | Rf629.2 | F | c.5602C>T | Mutation carrier | x | | | |
| b227 | Rf393.302 | F | c.1608del | Mutation carrier | x | | | |
| r272 | Rf385.102 | F | c.5596C>G | Mutation carrier | x | | | |
| r77 | Rf399.3 | F | c.2068C>T | Mutation carrier | x | | | |
| r837 | Rf683.1 | F | c.4661T>C | Mutation carrier | x | x | x | x |
| r861 | Rf743.2 | F | c.182_183dup | Mutation carrier | x | | | |
| r864 | Rf744.4 | F | unknown | Mutation carrier | x | | | |
| r893 | Rf562.1 | F | c.2191C>T | Mutation carrier | x | | | |
| r944 | Rf874.3 | F | c.3274_3276+1del | Mutation carrier | x | | | |
| r950 | Rf878.2 | F | c.3276+4_3276+7del | Mutation carrier | x | | | |
| r973 | Rf649.3 | F | c.3801+1G>A | Mutation carrier | x | | | |
| b13 | Rf898.2 | F | 1,2 Mb deletion SMCHD1 | Mutation carrier | x | | | |
| b17 | Rf922.2 | F | c.412C>T | Mutation carrier | x | | | |
| b37 | Rf936.1 | F | c.3276_3276+4del | Mutation carrier | x | | | |
| b109 | Rf975.201 | F | c.24del | Mutation carrier | x | | | |
| b206 | Rf1033.1 | F | c.1302_1306delTGATA | Mutation carrier | x | | x | x |
| b260 | Rf1021.2 | F | c.1580C>T | Mutation carrier | x | | | |
| b286 | Rf1110.3 | F | c.3048+2T>C | Mutation carrier | x | | | |
| b376 | Rf392.102 | F | c.4566G>A | Mutation carrier | x | | | |
| b398 | Rf1014.4 | F | c.4566G>A | Mutation carrier | x | | | |
| b454 | Rf1101.2 | F | c.582dupT | Mutation carrier | x | | | |
| b483 | Rf1126.204 | F | c.4404G>A | Mutation carrier | x | | | |
| b493 | Rf1196.1 | F | c.1273G>A | Mutation carrier | x | | | |
| b565 | Rf909.206 | F | c.2665dupA | Mutation carrier | x | | | |
| r730 | Rf745 | F | none | Control | | x | x | x |
| b118 | Rf854 | M | none | Control | | x | | |
| r870 | Rf844 | M | c.3274_3276+1del | Mutation carrier | | x | x | x |
| b194 | Rf1024 | M | none | Control | | | x | x |
| v96 | Rf899 | M | none | Control | | | x | x |
| b887 | Rf943 | F | none | Control | | | x | x |
| b735 | Rf1033 | F | c.1302_1306delTGATA | Mutation carrier | | | x | x |
| b206 | Rf1033 | F | c.1302_1306delTGATA | Mutation carrier | | | x | x |
| r648 | Rf696 | M | c.1647+3A>G | Mutation carrier | | | x | x |
| r887P | Rf854 | M | c.3444T>A | Mutation carrier | | | x | |
| r562 | Rf645 | F | none | Contracted D4Z4 | | | x | x |
| r707 | Rf731 | F | none | Contracted D4Z4 | | | x | x |
| r473 | Rf584 | F | none | Contracted D4Z4 | | | x | |
| r603 | Rf675 | F | none | Contracted D4Z4 | | | x | |
| r738 | Rf752 | F | none | Contracted D4Z4 | | | x | |
| r719 | Rf737 | M | none | Contracted D4Z4 | | | x | |
| b22 | Rf925 | F | none | Contracted D4Z4 | | | x | |
| r966 | Rf887 | F | none | Contracted D4Z4 | | | x | |
| r937 | Rf870 | M | none | Contracted D4Z4 | | | x | |
| r218 | Rf353 | F | none | Contracted D4Z4 | | | x | |

Cells which contain two 4QB alleles used for SMCHD1 KD

| ID | Rf.Nr | M/F | SMCHD1 mutationcDNA (NM_015295.2) | 4q allele 1 | 4q allele 2 |
|------|--------|-----|--------------------------------------|-------------|-------------|
| r879 | Rf852 | M | none | B | B |
| b194 | Rf1024 | M | none | B | B |
| b94 | Rf964 | F | none | B | B |