# APPENDIX

Table 1 - Gene lists from Round 1 of exome sequencing

| **GENES WITH VARIANTS SHARED BETWEEN 7 PATIENTS** | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| PCLO - protein piccolo | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 52,27 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX1662 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 54,32 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX1560 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 42,24 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX4581 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 44,23 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX4481 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 41,20 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX797 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 45,36 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| OX2636 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 59,15 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| NDUFS1 - NADH-ubiquinone oxidoreductase 75 kDa subunit, | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 27,11 \| 11,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| OX1662 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | possibly damaging | 15,14 \| 11,0 |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| OX1560 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 21,7 \| 13,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |

| | | | | | |
|---|---|---|---|---|---|
| OX4581 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | possibly damaging | 21,15 \| 18,0 |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| OX4481 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 10,10 \| 12,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| OX797 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | possibly damaging | 17,4 \| 15,0 |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| OX2636 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 22,12 \| 8,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |

| | | | | | |
|---|---|---|---|---|---|
| **GENES WITH VARIANTS SHARED BETWEEN 6 PATIENTS** | | | | | |
| | | | | | |
| LTBP4 - latent-transforming growth factor beta-binding | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,3 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| OX1662 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,4 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| OX1560 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,2 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| OX4581 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,4 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| OX4481 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 0,7 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |

| | | | | | |
|---|---|---|---|---|---|
| OX2636 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 2,3 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| **GENES WITH VARIANTS SHARED BETWEEN 5 PATIENTS** | | | | | |
| | | | | | |
| C20orf194 - hypothetical protein LOC25943 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 10,7 \| 0,7 |
| OX1560 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 9,5 \| 0,9 |
| OX4581 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 16,11 \| 0,13 |
| OX4481 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 12,8 \| 0,12 |
| OX2636 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 12,7 \| 0,9 |
| | | | | | |
| | | | | | |
| RSBN1L - round spermatid basic protein 1-like protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 4,20 \| 0,17 |

| | | | | | |
|---|---|---|---|---|---|
| OX1560 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 6,31 \| 0,14 |
| OX4581 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 5,33 \| 0,15 |
| OX4481 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 2,27 \| 0,15 |
| OX2636 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 3,24 \| 0,11 |
| | | | | | |
| | | | | | |
| MUC5B - mucin-5B precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr11:1271485 | G > A | nonsynonymous; MUC5B:NM_002458:exon31:c.G13375A:p.V4459M | unknown | 21,16 \| 12,14 |
| OX1662 | chr11:1270647 | G > C | nonsynonymous; MUC5B:NM_002458:exon31:c.G12537C:p.Q4179H | unknown | 22,5 \| 16,1 |
| OX1662 | chr11:1274084 | G > A | nonsynonymous; MUC5B:NM_002458:exon33:c.G15091A:p.V5031M | unknown | 1,0 \| 3,1 |
| OX1560 | chr11:1263932 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C5822G:p.T1941S | benign | 40,20 \| 25,14 |
| OX1560 | chr11:1265384 | T > C | nonsynonymous; MUC5B:NM_002458:exon31:c.T7274C:p.M2425T | unknown | 21,17 \| 19,2 |
| OX1560 | chr11:1265671 | G > A | nonsynonymous; MUC5B:NM_002458:exon31:c.G7561A:p.A2521T | unknown | 67,19 \| 13,14 |
| OX1560 | chr11:1265687 | G > T | nonsynonymous; MUC5B:NM_002458:exon31:c.G7577T:p.R2526I | unknown | 65,21 \| 21,12 |
| OX1560 | chr11:1266892 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C8782G:p.R2928G | unknown | 17,6 \| 23,0 |
| OX1560 | chr11:1267010 | A > G | nonsynonymous; MUC5B:NM_002458:exon31:c.A8900G:p.N2967S | unknown | 45,24 \| 29,12 |
| OX1560 | chr11:1267289 | A > C | nonsynonymous; MUC5B:NM_002458:exon31:c.A9179C:p.K3060T | unknown | 31,22 \| 20,36 |

| | | | | | |
|---|---|---|---|---|---|
| OX1560 | chr11:1267291 | T > C | nonsynonymous; MUC5B:NM_002458:exon31:c.T9181C:p.S3061P | unknown | 32,23 \| 20,36 |
| OX1560 | chr11:1267852 | G > A | nonsynonymous; MUC5B:NM_002458:exon31:c.G9742A:p.A3248T | unknown | 16,16 \| 2,7 |
| OX1560 | chr11:1269770 | C > T | nonsynonymous; MUC5B:NM_002458:exon31:c.C11660T:p.P3887L | unknown | 3,12 \| 2,5 |
| OX1560 | chr11:1270361 | C > T | nonsynonymous; MUC5B:NM_002458:exon31:c.C12251T:p.T4084M | unknown | 15,38 \| 6,13 |
| OX1560 | chr11:1270781 | A > G | nonsynonymous; MUC5B:NM_002458:exon31:c.A12671G:p.N4224S | unknown | 26,21 \| 12,13 |
| OX1560 | chr11:1271894 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C13784G:p.T4595S | unknown | 7,16 \| 7,3 |
| OX4581 | chr11:1270647 | G > C | nonsynonymous; MUC5B:NM_002458:exon31:c.G12537C:p.Q4179H | unknown | 19,1 \| 11,3 |
| OX4581 | chr11:1271180 | C > T | nonsynonymous; MUC5B:NM_002458:exon31:c.C13070T:p.P4357L | unknown | 17,40 \| 11,24 |
| OX797 | chr11:1270647 | G > C | nonsynonymous; MUC5B:NM_002458:exon31:c.G12537C:p.Q4179H | unknown | 21,3 \| 10,1 |
| | | | | | |
| **GENES WITH VARIANTS SHARED BETWEEN 4 PATIENTS** | | | | | |
| | | | | | |
| SIGLEC12 - sialic acid-binding Ig-like lectin 12 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 5,9 \| 3,11 |
| OX1560 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 0,0 \| 10,27 |
| OX4581 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 0,0 \| 6,34 |
| OX797 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 0,0 \| 13,34 |
| | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| OR4N3P - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 92,29 \| 30,9 |
| OX4581 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 77,14 \| 59,27 |
| OX4481 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 119,29 \| 30,9 |
| OX797 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 87,23 \| 54,19 |
| | | | | | |
| | | | | | |
| PIK3R6 - phosphoinositide 3-kinase regulatory subunit 6 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr17:8725216 | - > G | frameshift insertion; PIK3R6:NM_001010855:exon17:c.1826_1827insC:p.S609fs | | 0,0 \| 0,3 |
| OX1560 | chr17:8725216 | - > G | frameshift insertion; PIK3R6:NM_001010855:exon17:c.1826_1827insC:p.S609fs | | 0,0 \| 1,2 |
| OX1560 | chr17:8731443 | C > G | nonsynonymous; PIK3R6:NM_001010855:exon12:c.G1378C:p.V460L | benign | 9,1 \| 8,1 |
| OX4581 | chr17:8725216 | - > G | frameshift insertion; PIK3R6:NM_001010855:exon17:c.1826_1827insC:p.S609fs | | 0,0 \| 0,3 |
| OX2636 | chr17:8725216 | - > G | frameshift insertion; PIK3R6:NM_001010855:exon17:c.1826_1827insC:p.S609fs | | 0,0 \| 0,4 |
| | | | | | |
| | | | | | |

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|---|---|---|---|---|---|
| TMC1 - transmembrane channel-like protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 17,5 \| 9,4 |
| OX1662 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 14,1 \| 10,2 |
| OX4481 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 9,3 \| 3,5 |
| OX2636 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 18,3 \| 11,5 |
| | | | | | |
| | | | | | |
| PRAMEF4 - PRAME family member 4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr1:12939552 | - > G | frameshift insertion; PRAMEF4:NM_001009611:exon4:c.1250_1251insC:p.A417fs | | 37,47 \| 29,23 |
| OX1560 | chr1:12941966 | C > T | nonsynonymous; PRAMEF4:NM_001009611:exon3:c.G584A:p.R195H | benign | 67,85 \| 25,27 |
| OX797 | chr1:12942179 | T > C | nonsynonymous; PRAMEF4:NM_001009611:exon3:c.A371G:p.H124R | benign | 34,22 \| 14,2 |
| OX2636 | chr1:12939552 | - > G | frameshift insertion; PRAMEF4:NM_001009611:exon4:c.1250_1251insC:p.A417fs | | 33,49 \| 18,23 |
| | | | | | |
| | | | | | |
| FAM22F - hypothetical protein LOC54754 | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 2,5 \| 0,5 |
| OX1560 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 3,3 \| 1,3 |
| OX4481 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 1,2 \| 0,3 |
| OX797 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 4,6 \| 2,4 |
| | | | | | |
| **GENES WITH VARIANTS SHARED BETWEEN 3 PATIENTS** | | | | | |
| | | | | | |
| FAM21C - WASH complex subunit FAM21C | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr10:46245557 | T > C | nonsynonymous; FAM21C:NM_001169106:exon9:c.T746C:p.M249T | benign | 24,8 \| 12,1 |
| | | | FAM21C:NM_015262:exon9:c.T746C:p.M249T | | |
| | | | FAM21C:NM_001169107:exon9:c.T746C:p.M249T | | |
| OX4481 | chr10:46245557 | T > C | nonsynonymous; FAM21C:NM_001169106:exon9:c.T746C:p.M249T | benign | 16,4 \| 9,4 |
| | | | FAM21C:NM_015262:exon9:c.T746C:p.M249T | | |
| | | | FAM21C:NM_001169107:exon9:c.T746C:p.M249T | | |
| OX2636 | chr10:46245557 | T > C | nonsynonymous; FAM21C:NM_001169106:exon9:c.T746C:p.M249T | benign | 27,2 \| 17,3 |
| | | | FAM21C:NM_015262:exon9:c.T746C:p.M249T | | |

| | | | | | |
|---|---|---|---|---|---|
| | | | FAM21C:NM_001169107:exon9:c.T746C:p.M249T | | |
| | | | | | |
| | | | | | |
| MST1P2 - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr1:16975285 | - > C | frameshift insertion; MST1P2:NR_027504:exon8:c.1654_1655insC:p.Q552fs | | 17,12 \| 11,8 |
| OX1560 | chr1:16974312 | G > C | nonsynonymous; MST1P2:NR_027504:exon7:c.G772C:p.V258L | | 13,3 \| 4,0 |
| OX797 | chr1:16975285 | - > CC | frameshift insertion; MST1P2:NR_027504:exon8:c.1654_1655insCC:p.Q552fs | | 26,17 \| 2,3 |
| | | | | | |
| | | | | | |
| ALDH3B1 - aldehyde dehydrogenase family 3 member B1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr11:67795380 | - > C | frameshift insertion; ALDH3B1:NM_001161473:exon10:c.1378_1379insC:p.Q460fs | | 0,0 \| 3,0 |
| | | | ALDH3B1:NM_000694:exon10:c.1378_1379insC:p.Q460fs | | |
| | | | ALDH3B1:NM_001030010:exon9:c.1268_1269insC:p.P423fs | | |
| OX1662 | chr11:67795380 | - > C | frameshift insertion; ALDH3B1:NM_000694:exon10:c.1378_1379insC:p.Q460fs | | 0,0 \| 2,1 |
| | | | ALDH3B1:NM_001161473:exon10:c.1378_1379insC:p.Q460fs | | |
| | | | ALDH3B1:NM_001030010:exon9:c.1268_1269insC:p.P423fs | | |

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|---|---|---|---|---|---|
| OX4581 | chr11:67795380 | - > C | frameshift insertion; ALDH3B1:NM_000694:exon10:c.1378_1379insC:p.Q460fs | | 0,0 \| 1,2 |
| | | | ALDH3B1:NM_001161473:exon10:c.1378_1379insC:p.Q460fs | | |
| | | | ALDH3B1:NM_001030010:exon9:c.1268_1269insC:p.P423fs | | |
| | | | | | |
| | | | | | |
| TTN - titin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr2:179434293 | - > CTT | nonframeshift insertion; TTN:NM_133432:exon155:c.49746_49747insAAG:p.I16582delinsIR | | 3,6 \| 1,4 |
| | | | TTN:NM_003319:exon154:c.49371_49372insAAG:p.I16457delinsIR | | |
| | | | TTN:NM_133437:exon155:c.49947_49948insAAG:p.I16649delinsIR | | |
| | | | TTN:NM_133378:exon275:c.68862_68863insAAG:p.I22954delinsIR | | |
| OX4481 | chr2:179433388 | C > T | nonsynonymous; TTN:NM_133432:exon155:c.G50651A:p.R16884H | | 3,22 \| 5,17 |
| | | | TTN:NM_003319:exon154:c.G50276A:p.R16759H | | |
| | | | TTN:NM_133437:exon155:c.G50852A:p.R16951H | | |
| | | | TTN:NM_133378:exon275:c.G69767A:p.R23256H | | |
| OX797 | chr2:179483108 | C > T | nonsynonymous; TTN:NM_133432:exon81:c.G20257A:p.V6753I | unknown | 6,25 \| 14,25 |
| | | | TTN:NM_133437:exon81:c.G20458A:p.V6820I | | |
| | | | TTN:NM_133378:exon201:c.G39373A:p.V13125I | | |
| | | | TTN:NM_003319:exon80:c.G19882A:p.V6628I | | |
| OX797 | chr2:179544700 | - > TCT | nonframeshift insertion; TTN:NM_133378:exon137:c.29769_29770insAGA:p.V9923delinsVE | | 5,14 \| 6,4 |

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| MARK1 - serine/threonine-protein kinase MARK1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr1:220835506 | T > G | stoploss; MARK1:NM_018650:exon18:c.T2386G:p.X796E | | 0,13 \| 0,7 |
| OX1662 | chr1:220835506 | T > G | stoploss; MARK1:NM_018650:exon18:c.T2386G:p.X796E | | 0,18 \| 0,10 |
| OX4481 | chr1:220835506 | T > G | stoploss; MARK1:NM_018650:exon18:c.T2386G:p.X796E | | 0,6 \| 0,4 |
| | | | | | |
| | | | | | |
| RARRES1;RARRES1 - retinoic acid receptor responder (tazarotene) | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr3:158431648 | T > G | nonsynonymous; RARRES1:NM_002888:exon2:c.A277C:p.I93L | benign | 13,16 \| 0,15 |
| | | | RARRES1:NM_206963:exon2:c.A277C:p.I93L | | |
| OX1662 | chr3:158431648 | T > G | nonsynonymous; RARRES1:NM_206963:exon2:c.A277C:p.I93L | benign | 13,23 \| 0,15 |
| | | | RARRES1:NM_002888:exon2:c.A277C:p.I93L | | |
| OX2636 | chr3:158431648 | T > G | nonsynonymous; RARRES1:NM_002888:exon2:c.A277C:p.I93L | benign | 4,22 \| 0,15 |
| | | | RARRES1:NM_206963:exon2:c.A277C:p.I93L | | |

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| HLA-DRB1 - major histocompatibility complex, class II, DR beta 1 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr6:32552043 | G > A | nonsynonymous; HLA-DRB1:NM_002124:exon2:c.G213T:p.E71D | | 3,19 \| 0,6 |
| OX1560 | chr6:32549362 | G > A | nonsynonymous; HLA-DRB1:NM_002124:exon3:c.G624T:p.Q208H | | 30,16 \| 11,12 |
| OX4581 | chr6:32552043 | G > A | nonsynonymous; HLA-DRB1:NM_002124:exon2:c.G213T:p.E71D | | 3,9 \| 0,4 |
| | | | | | |
| | | | | | |
| PRSS48 - serine protease 48 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr4:152201026 | - > GTCAG | frameshift insertion; PRSS48:NM_183375:exon2:c.131_132insGTCAG:p.S44fs | | 17,5 \| 16,4 |
| OX4581 | chr4:152201026 | - > GTCAG | frameshift insertion; PRSS48:NM_183375:exon2:c.131_132insGTCAG:p.S44fs | | 16,3 \| 7,4 |
| OX797 | chr4:152201026 | - > GTCAG | frameshift insertion; PRSS48:NM_183375:exon2:c.131_132insGTCAG:p.S44fs | | 10,3 \| 14,2 |
| | | | | | |
| | | | | | |
| LGALS9C - galectin-9C | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX484 | chr17:18394056 | G > A | nonsynonymous; LGALS9C:NM_001040078:exon6:c.G550A:p.V184M | benign | 15,8 \| 14,11 |
| OX484 | chr17:18397546 | G > C | nonsynonymous; LGALS9C:NM_001040078:exon11:c.G936C:p.L312F | possibly damaging | 12,6 \| 4,5 |
| OX1662 | chr17:18397546 | G > C | nonsynonymous; LGALS9C:NM_001040078:exon11:c.G936C:p.L312F | possibly damaging | 21,16 \| 11,6 |
| OX797 | chr17:18397546 | G > C | nonsynonymous; LGALS9C:NM_001040078:exon11:c.G936C:p.L312F | possibly damaging | 12,3 \| 9,6 |
| | | | | | |
| | | | | | |
| SLC38A3;SLC38A3 - sodium-coupled neutral amino acid transporter 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4481 | chr3:50251835 | - > G | frameshift insertion; SLC38A3:NM_006841:exon3:c.103_104insG:p.V35fs | | 0,0 \| 1,3 |
| OX797 | chr3:50251835 | - > G | frameshift insertion; SLC38A3:NM_006841:exon3:c.103_104insG:p.V35fs | | 0,0 \| 1,2 |
| OX2636 | chr3:50251835 | - > G | frameshift insertion; SLC38A3:NM_006841:exon3:c.103_104insG:p.V35fs | | 0,0 \| 3,2 |
| | | | | | |
| | | | | | |
| PABPC3 - polyadenylate-binding protein 3 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX1662 | chr13:25671456 | C > T | nonsynonymous; PABPC3:NM_030979:exon1:c.C1120T:p.R374C | benign | 30,54 \| 7,14 |
| OX1560 | chr13:25671456 | C > T | nonsynonymous; PABPC3:NM_030979:exon1:c.C1120T:p.R374C | benign | 32,32 \| 2,10 |
| OX4581 | chr13:25671477 | G > C | nonsynonymous; PABPC3:NM_030979:exon1:c.G1141C:p.E381Q | benign | 48,39 \| 12,19 |
| | | | | | |
| | | | | | |
| AHNAK2 - protein AHNAK2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr14:105413152 | A > C | nonsynonymous; AHNAK2:NM_138420:exon7:c.T8636G:p.V2879G | benign | 23,20 \| 19,17 |
| OX484 | chr14:105417848 | G > C | nonsynonymous; AHNAK2:NM_138420:exon7:c.C3940G:p.L1314V | unknown | 10,40 \| 11,48 |
| OX4581 | chr14:105413207 | T > G | nonsynonymous; AHNAK2:NM_138420:exon7:c.A8581C:p.I2861L | unknown | 37,26 \| 20,17 |
| OX4581 | chr14:105416174 | A > T | nonsynonymous; AHNAK2:NM_138420:exon7:c.T5614A:p.C1872S | benign | 37,19 \| 24,23 |
| OX4581 | chr14:105419232 | C > G | nonsynonymous; AHNAK2:NM_138420:exon7:c.G2556C:p.M852I | benign | 46,28 \| 38,16 |
| OX2636 | chr14:105418445 | C > T | nonsynonymous; AHNAK2:NM_138420:exon7:c.G3343A:p.A1115T | benign | 7,25 \| 17,13 |
| | | | | | |
| | | | | | |
| OR2T2 - olfactory receptor 2T2 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX484 | chr1:248616710 | CGTGCTG > - | frameshift deletion; OR2T2:NM_001004136:exon1:c.612_618del:p.204_206del | | 12,36 \| 2,14 |
| OX4581 | chr1:248616710 | CGTGCTG > - | frameshift deletion; OR2T2:NM_001004136:exon1:c.612_618del:p.204_206del | | 12,29 \| 10,13 |
| OX4481 | chr1:248616710 | CGTGCTG > - | frameshift deletion; OR2T2:NM_001004136:exon1:c.612_618del:p.204_206del | | 11,22 \| 3,10 |
| | | | | | |
| | | | | | |
| FCGBP - IgGFc-binding protein precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:40382736 | A > T | nonsynonymous; FCGBP:NM_003890:exon22:c.T10150A:p.S3384T | unknown | 0,1 \| 4,9 |
| OX4481 | chr19:40392544 | TCT > - | nonframeshift deletion; FCGBP:NM_003890:exon16:c.7958_7960del:p.2653_2654del | | 5,31 \| 2,3 |
| OX2636 | chr19:40396277 | C > T | nonsynonymous; FCGBP:NM_003890:exon15:c.G7120A:p.D2374N | unknown | 7,7 \| 7,7 |
| | | | | | |
| | | | | | |
| SSPO - SCO-spondin precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 2,2 |

| | | | | | |
|---|---|---|---|---|---|
| OX1560 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 0,3 |
| OX4481 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 1,3 |
| | | | | | |
| **GENES WITH VARIANTS SHARED BETWEEN 2 PATIENTS** | | | | | |
| | | | | | |
| KIAA1467 - hypothetical protein LOC57613 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr12:13208856 | A > G | nonsynonymous; KIAA1467:NM_020853:exon2:c.A409G:p.S137G | benign | 2,7 \| 0,3 |
| OX2636 | chr12:13208856 | A > G | nonsynonymous; KIAA1467:NM_020853:exon2:c.A409G:p.S137G | benign | 3,4 \| 0,4 |
| | | | | | |
| | | | | | |
| MYOM1 - myomesin-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr18:3141984 | G > A | nonsynonymous; MYOM1:NM_019856:exon14:c.C1978T:p.P660S | probably damaging | 14,10 \| 8,6 |
| | | | MYOM1:NM_003803:exon14:c.C1978T:p.P660S | | |
| OX1560 | chr18:3135692 | T > A | nonsynonymous; MYOM1:NM_019856:exon15:c.A2062T:p.T688S | benign | 3,6 \| 3,6 |
| | | | MYOM1:NM_003803:exon15:c.A2062T:p.T688S | | |

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|---|---|---|---|---|---|
| | | | | | |
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| TCF12 - transcription factor 12 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr15:57545548 | C > - | frameshift deletion; TCF12:NM_207036:exon16:c.1349delC:p.P450fs | | 7,19 \| 4,13 |
| | | | TCF12:NM_207040:exon8:c.767delC:p.P256fs | | |
| | | | TCF12:NM_207037:exon16:c.1349delC:p.P450fs | | |
| | | | TCF12:NM_207038:exon15:c.1277delC:p.P426fs | | |
| | | | TCF12:NM_003205:exon15:c.1277delC:p.P426fs | | |
| OX4581 | chr15:57555369 | GAAA > - | frameshift deletion; TCF12:NM_207040:exon10:c.1060_1063del:p.354_355del | | 9,5 \| 6,4 |
| | | | TCF12:NM_207037:exon18:c.1642_1645del:p.548_549del | | |
| | | | TCF12:NM_207038:exon17:c.1570_1573del:p.524_525del | | |
| | | | TCF12:NM_003205:exon17:c.1570_1573del:p.524_525del | | |
| | | | TCF12:NM_207036:exon18:c.1642_1645del:p.548_549del | | |
| | | | | | |
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| ADAM29 - disintegrin and metalloproteinase | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| OX1560 | chr4:175898748 | TAT > - | nonframeshift deletion; ADAM29:NM_001130704:exon4:c.2072_2074del:p.691_692del | | 19,5 \| 5,1 |
| | | | ADAM29:NM_014269:exon5:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130705:exon3:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130703:exon4:c.2072_2074del:p.691_692del | | |
| OX4481 | chr4:175898748 | TAT > - | nonframeshift deletion; ADAM29:NM_001130704:exon4:c.2072_2074del:p.691_692del | | 12,5 \| 11,4 |
| | | | ADAM29:NM_014269:exon5:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130705:exon3:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130703:exon4:c.2072_2074del:p.691_692del | | |
| | | | | | |
| | | | | | |
| PRPF19 - pre-mRNA-processing factor 19 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr11:60666746 | T > G | nonsynonymous; PRPF19:NM_014502:exon11:c.A859C:p.T287P | probably damaging | 2,4 \| 0,4 |
| OX4481 | chr11:60666746 | T > G | nonsynonymous; PRPF19:NM_014502:exon11:c.A859C:p.T287P | probably damaging | 5,2 \| 0,3 |
| | | | | | |
| | | | | | |
| LOC399753 - unknown | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX1560 | chr10:49219100 | T > C | nonsynonymous; LOC399753:NR_029396:exon8:c.A1198G:p.S400G | | 14,17 \| 23,8 |
| OX4581 | chr10:49218451 | G > A | nonsynonymous; LOC399753:NR_029396:exon8:c.C1847T:p.T616I | | 11,29 \| 12,6 |
| | | | | | |
| | | | | | |
| ABCA2 - ABC transporter A family member 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4481 | chr9:139906404 | G > T | nonsynonymous; ABCA2:NM_212533:exon34:c.C5522A:p.A1841D | probably damaging | 3,1 \| 3,0 |
| | | | ABCA2:NM_001606:exon34:c.C5432A:p.A1811D | | |
| OX797 | chr9:139905705 | G > A | nonsynonymous; ABCA2:NM_212533:exon37:c.C5951T:p.T1984I | | 2,2 \| 0,6 |
| | | | ABCA2:NM_001606:exon37:c.C5861T:p.T1954I | | |
| | | | | | |
| | | | | | |
| NT5C3 - 5'-nucleotidase, cytosolic III | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr7:33075588 | G > - | frameshift deletion; NT5C3:NR_029372:exon2:c.243delC:p.N81fs | | 5,27 \| 1,28 |
| OX2636 | chr7:33075588 | G > - | frameshift deletion; NT5C3:NR_029372:exon2:c.243delC:p.N81fs | | 9,16 \| 1,14 |

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| TRIOBP - TRIO and F-actin-binding protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr22:38109227 | C > G | nonsynonymous; TRIOBP:NM_001039141:exon5:c.C265G:p.P89A | benign | 10,0 \| 6,0 |
| OX4481 | chr22:38119197 | G > A | nonsynonymous; TRIOBP:NM_001039141:exon7:c.G634A:p.G212S | possibly damaging | 1,2 \| 2,1 |
| | | | | | |
| | | | | | |
| OR2T27 - olfactory receptor 2T27 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr1:248813656 | AAG > - | nonframeshift deletion; OR2T27:NM_001001824:exon1:c.528_530del:p.176_177del | | 10,5 \| 3,6 |
| OX4481 | chr1:248813656 | AAG > - | nonframeshift deletion; OR2T27:NM_001001824:exon1:c.528_530del:p.176_177del | | 16,5 \| 6,7 |
| | | | | | |
| | | | | | |
| VPS13D - vacuolar protein sorting-associated protein 13D | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX484 | chr1:12336408 | G > C | nonsynonymous; VPS13D:NM_018156:exon19:c.G2763C:p.E921D | benign | 12,7 \| 10,6 |
| | | | VPS13D:NM_015378:exon19:c.G2763C:p.E921D | | |
| OX1662 | chr1:12387842 | A > T | nonsynonymous; VPS13D:NM_018156:exon36:c.A8128T:p.S2710C | benign | 4,32 \| 3,28 |
| | | | VPS13D:NM_015378:exon36:c.A8128T:p.S2710C | | |
| | | | | | |
| | | | | | |
| LFNG - beta-1,3-N-acetylglucosaminyltransferase lunatic | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX797 | chr7:2552886 | - > GTGG | frameshift insertion; LFNG:NM_001166355:exon2:c.143_144insGTGG:p.G48fs | | 5,21 \| 1,4 |
| OX797 | chr7:2552909 | - > GATG | stopgain; LFNG:NM_001166355:exon2:c.166_167insGATG:p.E56_W57delinsGX | | 1,3 \| 1,4 |
| OX2636 | chr7:2552909 | - > GATG | stopgain; LFNG:NM_001166355:exon2:c.166_167insGATG:p.E56_W57delinsGX | | 0,9 \| 1,2 |
| | | | | | |
| | | | | | |
| HEMGN - hemogen | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr9:100692325 | A > G | nonsynonymous; HEMGN:NM_197978:exon3:c.T1352C:p.F451S | possibly damaging | 49,1 \| 30,1 |

| | | | | | |
|---|---|---|---|---|---|
| | | | HEMGN:NM_018437:exon4:c.T1352C:p.F451S | | |
| OX1560 | chr9:100693398 | T > G | nonsynonymous; HEMGN:NM_197978:exon3:c.A279C:p.E93D | benign | 28,13 \| 17,7 |
| | | | HEMGN:NM_018437:exon4:c.A279C:p.E93D | | |
| | | | | | |
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| ZDBF2 - DBF4-type zinc finger-containing protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr2:207175370 | C > T | nonsynonymous; ZDBF2:NM_020923:exon5:c.C6118T:p.R2040W | benign | 4,2 \| 1,4 |
| OX4481 | chr2:207170388 | A > G | nonsynonymous; ZDBF2:NM_020923:exon5:c.A1136G:p.Q379R | benign | 6,5 \| 2,7 |
| | | | | | |
| | | | | | |
| OBSCN - obscurin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX797 | chr1:228481122 | C > T | nonsynonymous; OBSCN:NM_052843:exon41:c.C10936T:p.R3646W | probably damaging | 5,6 \| 7,2 |
| | | | OBSCN:NM_001098623:exon41:c.C10936T:p.R3646W | | |
| OX2636 | chr1:228456382 | A > C | nonsynonymous; OBSCN:NM_001098623:exon17:c.A5013C:p.K1671N | probably damaging | 1,6 \| 4,7 |
| | | | OBSCN:NM_052843:exon17:c.A5013C:p.K1671N | | |

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| GPRIN1 - G protein-regulated inducer of neurite outgrowth | | | | | |
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| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr5:176026138 | T > A | nonsynonymous; GPRIN1:NM_052899:exon2:c.A698T:p.E233V | possibly damaging | 15,33 \| 4,10 |
| OX2636 | chr5:176026138 | T > A | nonsynonymous; GPRIN1:NM_052899:exon2:c.A698T:p.E233V | possibly damaging | 8,27 \| 5,3 |
| | | | | | |
| | | | | | |
| ZNF585B - zinc finger protein 585B | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr19:37677027 | C > T | nonsynonymous; ZNF585B:NM_152279:exon5:c.G1412A:p.C471Y | probably damaging | 22,18 \| 18,11 |
| OX797 | chr19:37677723 | T > C | nonsynonymous; ZNF585B:NM_152279:exon5:c.A716G:p.E239G | probably damaging | 23,43 \| 12,22 |
| | | | | | |
| | | | | | |
| MOGAT1 - 2-acylglycerol O-acyltransferase 1 | | | | | |

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|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr2:223559988 | G > - | frameshift deletion; MOGAT1:NM_058165:exon5:c.834delG:p.R278fs | | 1,4 \| 1,4 |
| OX2636 | chr2:223553200 | T > C | nonsynonymous; MOGAT1:NM_058165:exon2:c.T232C:p.W78R | benign | 4,7 \| 1,6 |
| | | | | | |
| | | | | | |
| TANC1 - protein TANC1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr2:160087099 | C > T | nonsynonymous; TANC1:NM_033394:exon27:c.C5162T:p.T1721M | possibly damaging | 10,23 \| 15,8 |
| OX4481 | chr2:160053219 | C > T | nonsynonymous; TANC1:NM_033394:exon18:c.C3080T:p.P1027L | benign | 1,4 \| 1,7 |
| | | | TANC1:NM_001145909:exon18:c.C3056T:p.P1019L | | |
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| TPSB2 - tryptase beta-2 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr16:1279732 | C > A | nonsynonymous; TPSB2:NM_024164:exon3:c.G68T:p.G23V | benign | 2,5 \| 1,4 |
| OX4481 | chr16:1279945 | T > C | nonsynonymous; TPSB2:NM_024164:exon2:c.A8G:p.N3S | unknown | 4,3 \| 4,0 |

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| MUC20 - mucin-20 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr3:195453019 | C > - | frameshift deletion; MUC20:NM_001098516:exon2:c.927delC:p.A309fs | | 16,13 \| 4,2 |
| | | | MUC20:NM_152673:exon3:c.1032delC:p.A344fs | | |
| OX1662 | chr3:195453019 | C > - | frameshift deletion; MUC20:NM_001098516:exon2:c.927delC:p.A309fs | | 12,7 \| 7,3 |
| | | | MUC20:NM_152673:exon3:c.1032delC:p.A344fs | | |
| | | | | | |
| | | | | | |
| H6PD - GDH/6PGL endoplasmic bifunctional protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr1:9304987 | CCCAGGCA > - | frameshift deletion; H6PD:NM_004285:wholegene | | 14,2 \| 5,3 |
| OX2636 | chr1:9304987 | CCCAGGCA > - | frameshift deletion; H6PD:NM_004285:wholegene | | 0,0 \| 4,0 |
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|---|---|---|---|---|---|
| BAZ2A - bromodomain adjacent to zinc finger domain | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr12:56994855 | C > T | nonsynonymous; BAZ2A:NM_013449:exon22:c.G4328A:p.R1443Q | benign | 3,4 \| 5,2 |
| OX4481 | chr12:56994855 | C > T | nonsynonymous; BAZ2A:NM_013449:exon22:c.G4328A:p.R1443Q | benign | 3,2 \| 7,4 |
| | | | | | |
| | | | | | |
| CNTNAP5 - contactin-associated protein-like 5 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr2:125504872 | C > G | nonsynonymous; CNTNAP5:NM_130773:exon14:c.C2141G:p.P714R | benign | 11,1 \| 11,3 |
| OX4481 | chr2:125405388 | C > T | nonsynonymous; CNTNAP5:NM_130773:exon13:c.C1927T:p.R643W | possibly damaging | 1,1 \| 2,3 |
| | | | | | |
| | | | | | |
| LRIG1 - leucine-rich repeats and immunoglobulin-like | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

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|---|---|---|---|---|---|
| OX1560 | chr3:66431273 | C > T | nonsynonymous; LRIG1:NM_015541:exon18:c.G2783A:p.R928Q | benign | 3,11 \| 2,9 |
| OX4581 | chr3:66431953 | G > A | nonsynonymous; LRIG1:NM_015541:exon17:c.C2720T:p.P907L | benign | 32,3 \| 16,10 |
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| DIDO1 - death-inducer obliterator 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX797 | chr20:61526477 | G > A | nonsynonymous; DIDO1:NM_001193370:exon9:c.C2255T:p.A752V | benign | 21,44 \| 20,40 |
| | | | DIDO1:NM_001193369:exon9:c.C2255T:p.A752V | | |
| | | | DIDO1:NM_033081:exon9:c.C2255T:p.A752V | | |
| | | | DIDO1:NM_080797:exon9:c.C2255T:p.A752V | | |
| OX2636 | chr20:61511139 | G > C | nonsynonymous; DIDO1:NM_001193369:exon16:c.C6169G:p.Q2057E | benign | 0,11 \| 3,14 |
| | | | DIDO1:NM_033081:exon16:c.C6169G:p.Q2057E | | |
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| | | | | | |
| HSH2D - hematopoietic SH2 domain containing | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr19:16268326 | G > A | nonsynonymous; HSH2D:NM_032855:exon8:c.G781A:p.G261R | | 5,8 \| 4,14 |
| OX2636 | chr19:16268539 | G > T | nonsynonymous; HSH2D:NM_032855:exon8:c.G994T:p.A332S | benign | 2,1 \| 3,0 |

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| SDCCAG3 - serologically defined colon cancer antigen 3 | | | | | |
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| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr9:139301782 | G > T | nonsynonymous; SDCCAG3:NM_001039707:exon5:c.C634A:p.L212I | benign | 0,4 \| 4,3 |
| | | | SDCCAG3:NM_006643:exon4:c.C565A:p.L189I | | |
| | | | SDCCAG3:NM_001039708:exon3:c.C415A:p.L139I | | |
| OX1662 | chr9:139301806 | C > T | nonsynonymous; SDCCAG3:NM_006643:exon4:c.G541A:p.G181S | benign | 1,3 \| 2,1 |
| | | | SDCCAG3:NM_001039708:exon3:c.G391A:p.G131S | | |
| | | | SDCCAG3:NM_001039707:exon5:c.G610A:p.G204S | | |
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| | | | | | |
| OR11H2 - olfactory receptor 11H2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr14:20181502 | C > T | nonsynonymous; OR11H2:NM_001197287:exon2:c.G574A:p.V192I | | 40,36 \| 17,19 |
| OX2636 | chr14:20181502 | C > T | nonsynonymous; OR11H2:NM_001197287:exon2:c.G574A:p.V192I | | 41,35 \| 58,61 |
| | | | | | |

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|---|---|---|---|---|---|
| ACTN1 - alpha-actinin-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr14:69349272 | G > A | nonsynonymous; ACTN1:NM_001130004:exon16:c.C1856T:p.T619M | benign | 2,1 \| 3,1 |
| | | | ACTN1:NM_001130005:exon16:c.C1856T:p.T619M | | |
| | | | ACTN1:NM_001102:exon16:c.C1856T:p.T619M | | |
| OX1560 | chr14:69376064 | C > T | nonsynonymous; ACTN1:NM_001130004:exon6:c.G565A:p.E189K | benign | 6,1 \| 3,1 |
| | | | ACTN1:NM_001130005:exon6:c.G565A:p.E189K | | |
| | | | ACTN1:NM_001102:exon6:c.G565A:p.E189K | | |
| | | | | | |
| | | | | | |
| ZSCAN29 - zinc finger and SCAN domain-containing protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr15:43658669 | C > A | nonsynonymous; ZSCAN29:NM_152455:exon3:c.G861T:p.R287S | possibly damaging | 12,12 \| 16,29 |
| OX2636 | chr15:43653374 | C > T | nonsynonymous; ZSCAN29:NM_152455:exon5:c.G2456A:p.G819E | benign | 20,28 \| 16,23 |
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|---|---|---|---|---|---|
| ALG3 - Alg3p | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr3:183963114 | G > T | stopgain; ALG3:NM_005787:exon4:c.C477A:p.C159X | | 9,10 \| 3,0 |
| | | | ALG3:NR_024534:exon4:c.C471A:p.C157X | | |
| | | | ALG3:NR_024533:exon3:c.C408A:p.C136X | | |
| | | | ALG3:NM_001006941:exon4:c.C333A:p.C111X | | |
| OX4581 | chr3:183963134 | C > T | nonsynonymous; ALG3:NM_005787:exon4:c.G457A:p.V153I | probably damaging | 4,2 \| 1,6 |
| | | | ALG3:NR_024534:exon4:c.G451A:p.V151I | | |
| | | | ALG3:NR_024533:exon3:c.G388A:p.V130I | | |
| | | | ALG3:NM_001006941:exon4:c.G313A:p.V105I | | |
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| | | | | | |
| MYO9B - myosin-IXb | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:17298787 | G > A | nonsynonymous; MYO9B:NM_004145:exon19:c.G2621A:p.R874H | probably damaging | 3,1 \| 2,1 |
| | | | MYO9B:NM_001130065:exon19:c.G2621A:p.R874H | | |
| OX4481 | chr19:17212814 | A > G | nonsynonymous; MYO9B:NM_004145:exon2:c.A287G:p.Q96R | benign | 1,1 \| 3,0 |
| | | | MYO9B:NM_001130065:exon2:c.A287G:p.Q96R | | |

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| CCDC155 - coiled-coil domain-containing protein 155 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:49898404 | G > A | nonsynonymous; CCDC155:NM_144688:exon4:c.G190A:p.V64M | probably damaging | 6,0 \| 8,2 |
| OX797 | chr19:49898375 | T > G | nonsynonymous; CCDC155:NM_144688:exon4:c.T161G:p.V54G | probably damaging | 5,1 \| 4,0 |
| | | | | | |
| | | | | | |
| APLF - aprataxin and PNK-like factor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr2:68805146 | A > - | frameshift deletion; APLF:NM_173545:exon10:c.1528delA:p.R510fs | | 0,25 \| 1,16 |
| OX1560 | chr2:68805127 | A > T | nonsynonymous; APLF:NM_173545:exon10:c.A1509T:p.E503D | probably damaging | 0,24 \| 1,13 |
| | | | | | |
| | | | | | |
| SIRT7 - NAD-dependent deacetylase sirtuin-7 | | | | | |

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|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr17:79872316 | T > G | nonsynonymous; SIRT7:NM_016538:exon7:c.A670C:p.T224P | benign | 1,7 \| 6,0 |
| OX797 | chr17:79872316 | T > G | nonsynonymous; SIRT7:NM_016538:exon7:c.A670C:p.T224P | benign | 0,8 \| 5,0 |
| | | | | | |
| | | | | | |
| ERBB2IP - protein LAP2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr5:65350003 | G > A | nonsynonymous; ERBB2IP:NM_018695:exon21:c.G2857A:p.E953K | possibly damaging | 14,23 \| 4,11 |
| | | | ERBB2IP:NM_001006600:exon21:c.G2857A:p.E953K | | |
| OX2636 | chr5:65321357 | G > A | nonsynonymous; ERBB2IP:NM_018695:exon11:c.G867A:p.M289I | benign | 11,22 \| 13,9 |
| | | | ERBB2IP:NM_001006600:exon11:c.G867A:p.M289I | | |
| | | | | | |
| | | | | | |
| CTAG2 - cancer/testis antigen 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chrX:153880699 | G > - | frameshift deletion; CTAG2:NM_020994:exon2:c.476delC:p.S159fs | | 0,0 \| 17,5 |

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|---|---|---|---|---|---|
| OX4581 | chrX:153881543 | G > T | nonsynonymous; CTAG2:NM_172377:exon1:c.C247A:p.P83T | benign | 1,5 \| 0,3 |
| | | | CTAG2:NM_020994:exon1:c.C247A:p.P83T | | |
| | | | | | |
| | | | | | |
| MUC2 - mucin-2 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr11:1096411 | G > A | nonsynonymous; MUC2:NM_002457:exon35:c.G6424A:p.V2142M | | 3,1 \| 6,2 |
| OX4481 | chr11:1095708 | C > A | stopgain; MUC2:NM_002457:exon34:c.C6207A:p.C2069X | | 2,4 \| 3,0 |
| | | | | | |
| | | | | | |
| GPX1 - Gpx1p | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr3:49394834 | G > A | nonsynonymous; GPX1:NM_000581:exon2:c.C599T:p.P200L | | 0,1 \| 2,3 |
| OX2636 | chr3:49394834 | G > A | nonsynonymous; GPX1:NM_000581:exon2:c.C599T:p.P200L | | 6,5 \| 6,1 |
| | | | | | |
| | | | | | |
| DNAH11 - dynein heavy chain 11, axonemal | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX4581 | chr7:21630992 | G > A | nonsynonymous; DNAH11:NM_003777:exon14:c.G2464A:p.E822K | benign | 10,4 \| 5,6 |
| OX4481 | chr7:21818707 | T > C | nonsynonymous; DNAH11:NM_003777:exon57:c.T9490C:p.C3164R | | 1,3 \| 1,3 |
| | | | | | |
| | | | | | |
| ITGB4 - integrin beta-4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr17:73736913 | C > A | nonsynonymous; ITGB4:NM_001005619:exon21:c.C2590A:p.L864I | benign | 6,11 \| 3,0 |
| | | | ITGB4:NM_000213:exon22:c.C2590A:p.L864I | | |
| | | | ITGB4:NM_001005731:exon22:c.C2590A:p.L864I | | |
| OX1560 | chr17:73735778 | G > A | nonsynonymous; ITGB4:NM_000213:exon19:c.G2246A:p.C749Y | probably damaging | 9,6 \| 0,3 |
| | | | ITGB4:NM_001005619:exon18:c.G2246A:p.C749Y | | |
| | | | ITGB4:NM_001005731:exon19:c.G2246A:p.C749Y | | |
| | | | | | |
| | | | | | |
| CPAMD8 - C3 and PZP-like alpha-2-macroglobulin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

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|---|---|---|---|---|---|
| OX4481 | chr19:17014633 | T > C | nonsynonymous; CPAMD8:NM_015692:exon33:c.A4427G:p.Y1476C | | 4,27 \| 1,27 |
| OX2636 | chr19:17086023 | C > T | nonsynonymous; CPAMD8:NM_015692:exon17:c.G2095A:p.V699M | | 1,5 \| 0,3 |
| | | | | | |
| | | | | | |
| OR2T29 - olfactory receptor 2T29 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr1:248722624 | C > T | nonsynonymous; OR2T29:NM_001004694:exon1:c.G169A:p.A57T | benign | 32,57 \| 35,20 |
| OX2636 | chr1:248722621 | G > A | nonsynonymous; OR2T29:NM_001004694:exon1:c.C172T:p.H58Y | benign | 14,19 \| 39,24 |
| OX2636 | chr1:248722624 | C > T | nonsynonymous; OR2T29:NM_001004694:exon1:c.G169A:p.A57T | benign | 12,19 \| 40,26 |
| | | | | | |
| | | | | | |
| MDP1,NEDD8-MDP1 - NEDD8-MDP1 protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr14:24683269 | A > - | frameshift deletion; MDP1:NM_001199821:exon5:c.352delT:p.F118fs | | 29,7 \| 16,1 |
| | | | MDP1:NM_138476:exon6:c.492delT:p.P164fs | | |
| | | | NEDD8-MDP1:NM_001199823:exon7:c.543delT:p.P181fs | | |
| OX1560 | chr14:24683269 | A > - | frameshift deletion; MDP1:NM_001199821:exon5:c.352delT:p.F118fs | | 7,7 \| 13,6 |
| | | | MDP1:NM_138476:exon6:c.492delT:p.P164fs | | |

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|---|---|---|---|---|---|
| | | | NEDD8-MDP1:NM_001199823:exon7:c.543delT:p.P181fs | | |
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| | | | | | |
| ZBTB38 - zinc finger and BTB domain-containing protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr3:141163079 | A > G | nonsynonymous; ZBTB38:NM_001080412:exon8:c.A1849G:p.N617D | benign | 19,18 \| 17,22 |
| OX2636 | chr3:141163079 | A > G | nonsynonymous; ZBTB38:NM_001080412:exon8:c.A1849G:p.N617D | benign | 17,13 \| 16,8 |
| | | | | | |
| | | | | | |
| SNORD50A - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr6:86387034 | AA > - | frameshift deletion; SNORD50A:NR_002743:exon1:c.52_0del:p.18_0del | | 17,4 \| 12,3 |
| OX797 | chr6:86387034 | AA > - | frameshift deletion; SNORD50A:NR_002743:exon1:c.52_0del:p.18_0del | | 12,4 \| 9,3 |
| | | | | | |
| | | | | | |
| SSH2 - protein phosphatase Slingshot homolog 2 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| OX1662 | chr17:27963651 | C > G | nonsynonymous; SSH2:NM_033389:exon14:c.G1516C:p.V506L | benign | 15,15 \| 18,11 |
| OX4581 | chr17:27963431 | G > C | nonsynonymous; SSH2:NM_033389:exon14:c.C1736G:p.P579R | possibly damaging | 17,9 \| 18,9 |
| | | | | | |
| | | | | | |
| SYCE2 - synaptonemal complex central element protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr19:13015347 | G > A | nonsynonymous; SYCE2:NM_001105578:exon3:c.C265T:p.H89Y | possibly damaging | 28,11 \| 28,4 |
| OX797 | chr19:13015347 | G > A | nonsynonymous; SYCE2:NM_001105578:exon3:c.C265T:p.H89Y | possibly damaging | 21,2 \| 16,12 |
| | | | | | |
| | | | | | |
| GPR111 - probable G-protein coupled receptor 111 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr6:47649849 | T > G | nonsynonymous; GPR111:NM_153839:exon7:c.T1350G:p.C450W | probably damaging | 6,11 \| 8,18 |

| | | | | | |
|---|---|---|---|---|---|
| OX2636 | chr6:47650349 | T > C | nonsynonymous; GPR111:NM_153839:exon7:c.T1850C:p.L617P | probably damaging | 7,7 \| 2,7 |
| | | | | | |
| | | | | | |
| PCNXL2 - pecanex-like protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr1:233190016 | C > T | nonsynonymous; PCNXL2:NM_014801:exon25:c.G4349A:p.R1450Q | probably damaging | 7,0 \| 5,0 |
| OX2636 | chr1:233275593 | G > A | nonsynonymous; PCNXL2:NM_014801:exon20:c.C3526T:p.H1176Y | probably damaging | 2,2 \| 4,2 |
| | | | | | |
| | | | | | |
| ASMTL - acetylserotonin O-methyltransferase-like | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chrX:1558028 | G > A | nonsynonymous; ASMTL:NM_001173473:exon3:c.C61T:p.R21W | probably damaging | 1,6 \| 4,8 |
| | | | ASMTL:NM_004192:exon3:c.C235T:p.R79W | | |
| OX2636 | chrX:1522164 | A > - | stoploss; ASMTL:NM_001173474:exon12:c.1816delT:p.X606E | | 6,1 \| 4,0 |
| | | | ASMTL:NM_001173473:exon13:c.1690delT:p.X564E | | |
| | | | ASMTL:NM_004192:exon13:c.1864delT:p.X622E | | |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| | | | | | |
| ZNF474 - zinc finger protein 474 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr5:121488638 | T > - | stopgain; ZNF474:NM_207317:exon2:c.953delT:p.L318X | | 8,10 \| 6,6 |
| OX4581 | chr5:121488638 | T > - | stopgain; ZNF474:NM_207317:exon2:c.953delT:p.L318X | | 14,6 \| 11,2 |
| | | | | | |
| | | | | | |
| DNM1P46 - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr15:100340097 | C > T | nonsynonymous; DNM1P46:NR_003260:exon4:c.G830A:p.R277H | | 4,3 \| 5,3 |
| OX2636 | chr15:100340126 | AGA > - | frameshift deletion; DNM1P46:NR_003260:exon4:c.799_0del:p.267_0del | | 5,10 \| 1,3 |
| | | | | | |
| | | | | | |
| PRKDC - DNA-dependent protein kinase catalytic subunit | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| OX4581 | chr8:48846622 | C > T | nonsynonymous; PRKDC:NM_006904:exon15:c.G1526A:p.R509H | benign | 12,17 \| 8,8 |
| | | | PRKDC:NM_001081640:exon15:c.G1526A:p.R509H | | |
| OX2636 | chr8:48771413 | T > C | nonsynonymous; PRKDC:NM_006904:exon47:c.A6341G:p.K2114R | | 4,2 \| 9,6 |
| | | | PRKDC:NM_001081640:exon47:c.A6341G:p.K2114R | | |
| | | | | | |
| | | | | | |
| FAM82A2;FAM82A2 - regulator of microtubule dynamics protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr15:41029923 | A > C | nonsynonymous; FAM82A2:NM_018145:exon10:c.T1127G:p.V376G | probably damaging | 3,6 \| 0,5 |
| OX797 | chr15:41029923 | A > C | nonsynonymous; FAM82A2:NM_018145:exon10:c.T1127G:p.V376G | probably damaging | 14,2 \| 0,6 |
| | | | | | |
| | | | | | |
| CALHM2 - calcium homeostasis modulator protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr10:105209357 | AG > - | frameshift deletion; CALHM2:NR_024552:exon3:c.849_0del:p.283_0del | | 1,6 \| 2,4 |
| | | | CALHM2:NM_015916:exon3:c.341_342del:p.114_114del | | |

| | | | | | |
|---|---|---|---|---|---|
| OX4481 | chr10:105209357 | AG > - | frameshift deletion; CALHM2:NR_024552:exon3:c.849_0del:p.283_0del | | 5,6 \| 2,7 |
| | | | CALHM2:NM_015916:exon3:c.341_342del:p.114_114del | | |
| | | | | | |
| | | | | | |
| AQP12B - aquaporin-12B | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX797 | chr2:241622034 | G > C | nonsynonymous; AQP12B:NM_001102467:exon1:c.C221G:p.T74S | probably damaging | 5,3 \| 4,1 |
| OX2636 | chr2:241621908 | G > A | nonsynonymous; AQP12B:NM_001102467:exon1:c.C347T:p.A116V | benign | 4,2 \| 1,4 |
| | | | | | |
| | | | | | |
| PTCHD3;PTCHD3 - patched domain-containing protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr10:27700857 | A > C | nonsynonymous; PTCHD3:NM_001034842:exon2:c.T1091G:p.V364G | probably damaging | 2,13 \| 0,9 |
| OX4481 | chr10:27700857 | A > C | nonsynonymous; PTCHD3:NM_001034842:exon2:c.T1091G:p.V364G | probably damaging | 2,17 \| 0,8 |
| | | | | | |
| | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| SH3RF3 - SH3 domain-containing RING finger protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX484 | chr2:110065896 | C > T | nonsynonymous; SH3RF3:NM_001099289:exon8:c.C2099T:p.S700F | benign | 2,1 \| 2,1 |
| OX4581 | chr2:110065649 | C > G | nonsynonymous; SH3RF3:NM_001099289:exon8:c.C1852G:p.Q618E | benign | 1,1 \| 0,3 |
| | | | | | |
| | | | | | |
| MUC16 - mucin-16 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX4581 | chr19:9005567 | T > A | nonsynonymous; MUC16:NM_024690:exon46:c.A39839T:p.Y13280F | | 22,3 \| 17,6 |
| OX2636 | chr19:9047685 | G > A | nonsynonymous; MUC16:NM_024690:exon5:c.C33946T:p.P11316S | | 3,11 \| 6,11 |
| | | | | | |
| | | | | | |
| CCDC41 - coiled-coil domain-containing protein 41 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr12:94806191 | C > T | nonsynonymous; CCDC41:NM_001042399:exon2:c.G76A:p.G26S | | 15,21 \| 10,24 |
| | | | CCDC41:NM_016122:exon3:c.G76A:p.G26S | | |

| | | | | | |
|---|---|---|---|---|---|
| OX2636 | chr12:94763737 | G > T | nonsynonymous; CCDC41:NM_001042399:exon8:c.C1009A:p.L337I | | 36,10 \| 18,11 |
| | | | CCDC41:NM_016122:exon9:c.C1009A:p.L337I | | |
| | | | | | |
| | | | | | |
| PLA2G4F - cytosolic phospholipase A2 zeta | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr15:42434285 | A > C | nonsynonymous; PLA2G4F:NM_213600:exon20:c.T2447G:p.V816G | probably damaging | 9,4 \| 0,7 |
| OX797 | chr15:42442662 | T > C | nonsynonymous; PLA2G4F:NM_213600:exon9:c.A794G:p.E265G | benign | 2,0 \| 2,1 |
| | | | | | |
| | | | | | |
| TBC1D3G - TBC1 domain family member 3G | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1560 | chr17:34797608 | T > C | nonsynonymous; TBC1D3G:NM_001040282:exon14:c.A1528G:p.S510G | | 33,43 \| 6,11 |
| OX2636 | chr17:34797608 | T > C | nonsynonymous; TBC1D3G:NM_001040282:exon14:c.A1528G:p.S510G | | 36,29 \| 10,15 |
| | | | | | |
| | | | | | |
| STK33 - serine/threonine-protein kinase 33 | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| OX1662 | chr11:8414092 | - > ACCAGA | nonframeshift insertion; STK33:NM_030906:exon14:c.1510_1511insTCTGGT:p.S504delinsSGS | | 17,15 \| 5,2 |
| OX4481 | chr11:8414092 | - > ACCAGA | nonframeshift insertion; STK33:NM_030906:exon14:c.1510_1511insTCTGGT:p.S504delinsSGS | | 8,12 \| 4,3 |
| | | | | | |

| | | | | |
|---|---|---|---|---|
| **Screening** | | | | |
| | Primer sequence 5′→3′ | | Product size (bp) | Amplification conditions[a] |
| Amplicon | Forward | Reverse | | |
| Ex2 F/R | GCG TAA TCT TCC CCA GTA CCT CTC TCT GTT C | CTG CCT AGT CTC CAC GTT GCT TCC CCT CAC | 293 | 65 |
| Ex3 F/R2 | GAT GGA TGA CTT TTG GTG GAC TTT TGT TTG C | CTA TTT TTC TAC TAA AAT GTA TCA CAC CAC | 223 | 65 |
| Ex4 F/R | CTT GCT AAA TTG TAC TCT GCT GTC CCG TAC | CAA AAA GGT ACT AGG TAT TCT ACT GCA TTT C | 300 | 65 |
| Ex5 F/R | TAA GTC TGT ATA TTA ATT AGG TGA GTG TC | GTC CCT CCT GGA CAA AGG GAT AAA AAA TG | 279 | 65 |

| | | | | |
|---|---|---|---|---|
| Ex6 F/R | GCA ATA TAA TTA CCA TGA ATA GTC TAG C | CTT AGT TAC AAA TTA AAC AAA CAG TAG TG | 278 | 65 |
| Ex7 F/R | GGG TGC GTT CTA AGT TAA GAC ATT GCT G | CCA GAA GTA CAG ATT ATG GGA GG | 329 | 65 |
| Ex8 F/R | CTG TTA ATT TGA AGT CTT GAT TTT TTT C | GTA TGC CCA CTC GCA TCA ATC GAA TGT AC | 226 | 65 |
| Ex9 F/R | GCC CCT TAC AGA ATA TAG AAC TCA TTT TAC | CTG GCT GAT ATA CAG ATG ACT GCA GTA G | 247 | 65 |
| Ex 9A F/R | GTT GGT TCA GTC CTA TTT TGC ATA AGT TC | GCG AGT AAT GTA TAC TGC TTT CCT TTA AAC | 288 | 65 |
| Ex10 F/R | GTA CCC CTT GAA TTT TTA TAA GCA ACA TC | CTA ATC CAA AAA GAA ACA CAA GTG AGG AC | 292 | 65 |
| Ex11 F/R | GAC CTG TTA TCA TAG TTG TCC ATT TTA TG | GGT CAC TGA AAG TCC ACA AAC TTT TAG G | 279 | 65 |
| Ex12 F/R | GAA TAA TGG CAG AGT GCT AAA ATA TAA TAC | GAT ACT TAT CAC CTA CAA AAT CTA ATC ACT G | 220 | 65 |
| Ex 13 F/R | CTC CAT GTG AAC GGA TTT GTG TAT GTG CTG | CAC ACT TTC TTA TAG TCA AAA TCT AAA TG | 281 | 65 |
| Ex14 F/R | GTT ATT GAG TAT CTT TAC CCT TTC CTT CAC AAC | CTT TAT TGT TAT GCC AGA TTG ACC AAA AAC | 241 | 65 |
| Ex15F/R | GCA TAT CAG CTA TTT CCT CAT CTC TTT TGG AG | GTT TGA TTT TTA AAG ATG GGA GTC TGT ACA GC | 260 | 65 |
| Ex16 F/R | CAC ATA TCA CAT AGT AGG AAA CGT AGC AG | GAC TAA TAC TTA GCC AAT ACT TT A CTG C | 460 | 65 |
| Ex 17 F/R | CAG AAT AAT GGC ATC CAG AAA AAG TTG CTG | CCA GCT GTG AGA AAG GTG ACA GTG TTC CAA ATG | 335 | 65 |
| Ex 18 F/R2 | GAT GTG GAT ATA TTC TTT TCA GCT CTA AAT AG | GAG ACA ACA TAC ACA AAA ATC CTA ACA C | 475 | 65 |
| Ex 19 F/R | GTG CAC AAT CAG CAT ATC TTA CTT TAT AG | GGA AAA ACG ACC CTG ATG ATA CAG TAG AAG | 440 | 65 |
| Ex 20 F/R | GTA TGG GAA TGA AGT TAC ACA AAA CAA CAG | CTG TTC AAA GAT TCT AAC CCA GTT TAA G | 334 | 65 |
| | | | | |

Table 2 *TCF12* Primers and amplification conditions

Table 3 Mutation confirmation

| | | Primer sequence 5′→3′ | | Product size (bp) | Amplification conditions[a] | Digest/dideoxy seq[b] |
|---|---|---|---|---|---|---|
| Family # | Mutation | Forward | Reverse | | | |
| **1** | **526+1G>A** | Ex7F | Ex7R | 329 | 65 | HphI |
| **2** | **722C>G** | Ex10F | Ex10R | 292 | 65 | dideoxy seq |
| **3** | **778_779delAT** | Ex10F | Ex10R | 292 | 65 | dideoxy seq |
| **4** | **812C>A** | Ex10F | Ex10R | 292 | 65 | MseI |
| **5** | **822C>G** | Ex10F | Ex10R | 292 | 65 | MspI |
| **6** | **825C>G** | Ex10F | Ex10R | 292 | 65 | dideoxy seq |
| **7** | **842_843delCA** | Ex11F | Ex11R | 279 | 65 | dideoxy seq |
| **8** | **842C>G** | Ex11F | Ex11R | 279 | 65 | dideoxy seq |
| **9** | **842C>G** | Ex11F | Ex11R | 279 | 65 | dideoxy seq |

| | | | | | | |
|---|---|---|---|---|---|---|
| **10** | **970G>T** | Ex11F | Ex11R | 279 | 65 | dideoxy seq |
| **11** | **1035+3G>C** | Ex12F | Ex12R | 220 | 65 | dideoxy seq |
| **12** | **1073T>G** | Ex13F | Ex13R | 281 | 65 | dideoxy seq |
| **13** | **1115-2A>G** | Ex14F | Ex14R | 241 | 65 | dideoxy seq |
| **14** | **1123C>T** | Ex14F | Ex14R | 241 | 65 | dideoxy seq |
| **15** | **1127C>A** | Ex14F | Ex14R | 241 | 65 | HaeIII |
| **16** | **1283T>G** | Ex16F | Ex16R | 460 | 65 | TseI |
| **17** | **1312delC** | Ex16F | Ex16R | 460 | 65 | TseI |
| **18** | **1328dupT** | Ex16F | Ex16R | 460 | 65 | dideoxy seq |
| **19** | **1349delC** | Ex16F | Ex16R | 460 | 65 | TseI |
| **20** | **1376T>G** | Ex16F | Ex16R | 460 | 65 | BsaBI |
| **21** | **1453C>T** | Ex16F | Ex16R | 460 | 65 | BclI |
| **22** | **1468-20T>A** | Ex17F | Ex17R | 335 | 65 | dideoxy seq |
| **23** | **1491dupT** | Ex17F | Ex17R | 335 | 65 | PflFI |
| **24** | **1491dupT** | Ex17F | Ex17R | 335 | 65 | PflFI |
| **25** | **1514C>G** | Ex17F | Ex17R | 335 | 65 | dideoxy seq |
| **26** | **1530dupA** | Ex17F | Ex17R | 335 | 65 | HpyCH4III |
| **27** | **1540_1543dupTCAA** | Ex17F | Ex17R | 335 | 65 | dideoxy seq |
| **28** | **1582+2T>G** | Ex17F | Ex17R | 335 | 65 | MaeIII |
| **29** | **1628delA** | Ex18F | Ex18R2 | 475 | 65 | Hpy188III |
| **30** | **1642_1645delGAAA** | Ex18F | Ex18R2 | 475 | 65 | dideoxy seq |
| **31** | **1646delA** | Ex18F | Ex18R2 | 475 | 65 | dideoxy seq |
| **32** | **1691C>G** | Ex18F | Ex18R2 | 475 | 65 | dideoxy seq |
| **33** | **1691C>G** | Ex18F | Ex18R2 | 475 | 65 | dideoxy seq |
| **34** | **1713delA** | Ex18F | Ex18R2 | 475 | 65 | dideoxy seq |
| **35** | **1796_1799dupAGAA** | Ex19F | Ex19R | 440 | 65 | dideoxy seq |
| **36** | **1806delG** | Ex19F | Ex19R | 440 | 65 | BsrBI |
| **37** | **1871T>C** | Ex19F | Ex19R | 440 | 65 | BstNI |
| **38** | **1912C>G** | Ex19F | Ex19R | 440 | 65 | dideoxy seq |
| **39** | **1963G>T** | Ex19F | Ex19R | 440 | 65 | dideoxy seq |

Table 4 Sample relationships for *de novo* TCF12 mutations

| | OX3956 | | | OX4618 | | | LAH4951 | | | GOS4437 | | | OX838 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Microsatellite | P | M | F | P | M | F | P | M | F | P | M | F | P | M | F |
| D1S2868 | 2,2 (208,208) | 1,2(214,208) | 2,3(208,212) | 2,3 (214,212) | 3,3 (212,212) | 1,2(216,214) | 1,1(162,162) | 1,2(162,154) | 1,1(162,162) | 1,2(214,208) | 1,1(214,214) | 2,3(216,208) | 1,1(154,154) | 1,1(154,154) | 1,1(154,154) |
| D3S1311 | 1,3(150,136) | 1,4(150,148) | 2,3(144,136) | 1,2(148,136) | 1,2(148,136) | 1,2(148,136) | 1,3(156,152) | 1,4(156,142) | 2,3(154,152) | 1,2(150,146) | 2,2(146,146) | 1,2(150,146) | 3,3(154,154) | 2,3(156,154) | 1,3(158,154) |
| D4S403 | 1,1(184,184) | 1,2(184,174) | 1,2(184,174) | 1,3(184,174) | 1,2(184,182) | 1,3(184,174) | 1,2(236,232) | 2,3(234,232) | 1,4(236,226) | 2,2 (174,174) | 1,2(182,174) | F | 1,2(236,230) | 2,3(230,226) | 1,1(236,236) |
| D5S2027 | 1,3(200,198) | 1,2(200,196) | 3,3(198,198) | 1,3(198,192) | 2,3(196,192) | 1,4(198,194) | 1,2(166,164) | 1,2(166,164) | 2,3(166,170) | 1,2(198,194) | 1,2(198,194) | 1,2(198,194) | 3,3(166,166) | 2,3(168,166) | 1,3(172,166) |
| D6S1610 | 1,2(208,204) | 2,3(204,200) | 1,1(204,204) | 2,2(210,210) | 1,2(212,210) | 2,3(210,202) | 1,3(136,146) | 1,1(146,146) | 2,3(140,138) | 3,4(206,202) | 1,4(212,202) | 2,4(210,206) | 2,3(146,142) | 1,2(150,146) | F |
| D7S519 | 1,2(266,260) | 2,3(260,264) | 1,1(260,260) | 2,2(342,342) | 2,3(342,338) | 1,2(348,338) | 1,2(268,266) | 1,2(268,266) | 1,2(268,266) | 2,3(266,262) | 2,3(266,262) | 1,2(268,266) | 1,3(272,264) | 1,3(272,264) | 2,3(268,264) |
| D9S158 | 1,2(340,338) | 1,2(340,338) | 2,2(338,338) | 2,3(266,262) | 1,3(274,262) | 2,4(266,270) | 1,3(238,232) | F | 1,2 (238,230) | 2,3(340,334) | 3,3(334,334) | 1,2(340,338) | 1,2(234,230) | 2,3(230,228) | 1,1(234,234) |
| D10S548 | 2,3(190,188) | 1,2(194,190) | 3,3(188,188) | 1,2 (188,186) | 1,2(188,186) | 1,2(188,186) | 1,3(156,150) | 2,3(154,150) | 1,3(156,150) | 1,2(190,188) | 1,2(190,188) | 1,2(190,188) | 2,3(154,150) | 3,3(150,150) | 1,2(158,154) |
| D11S898 | 1,2(158,156) | 2,2 (156,156) | 1,2 (158,156) | 1,2(158,154) | 2,3(154,160) | 1,2(158,154) | 1,2(164,162) | 1,2(164,162) | 1,3(164,154) | 1,2(158,148) | 2,2(150,150) | 1,2(158,150) | 1,2(166,164) | 1,3(166,156) | 2,4(166,140) |
| D13S1265 | 1,1 (298,298) | 1,2 (298,290) | 1,3 (298,296) | 1,2(298,294) | 1,2(298,294) | 1,2(298,294) | 1,2(158,152) | 1,1(158,158) | 1,2(158,152) | 2,3(300,296) | 1,2(302,296) | 2,3(300,296) | 1,3(158,154) | 1,2(158,154) | 3,4(154,142) |
| D14S280 | 1,2(248,254) | 1,2(248,254) | 1,2 (248,254) | 1,3(250,248) | 1,2(250,246) | 2,3(250,246) | 2,3(242,240) | 2,3(242,240) | 1,3(244,240) | 2,3(246,244) | 1,3(248,244) | 2,4(246,242) | 2,2(240,240) | 1,2(242,240) | 1,2(242,240) |
| D16S415 | 1,2 (230, 228) | 1,3(230, 222) | 2,4(228, 214) | 1,2(232,228) | 1,2(232,228) | 2,3(228,214) | 1,3(234,218) | 1,2(234,232) | 1,3(234,218) | 2,3(228,224) | 2,3(228,224) | 1,2(232,228) | 3,4(228,218) | 1,3(234,228) | 2,4(232,218) |
| D18S474 | 1,2 (136,124) | 2,3(124,128) | 1,4(136,138) | 1,2 (134,130) | 2,3(130,128) | 1,3(134,128) | 1,4(146,130) | 1,3(146,134) | 2,4(144,130) | 1,3(138,126) | 2,3(132,126) | 1,3(138,126) | 2,3(142,134) | 1,3(144,134) | 1,2(144,140) |
| | 10/13 correct | | | 13/13 SRs correct | | | 12/12 correct | | | 12/13 correct | | | 12/13 correct | | |

| | OX2065 | | | OX4855 | | | OX5189 | | | OX4379 | | | 98D4142 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Microsatellite | P | M | F | P | M | F | P | M | F | P | M | F | P | M | F |
| D1S2868 | 2,3(214,210) | 1,2(220,214) | 3,4(210,212) | 2,3(212,210) | 1,3(214,210) | 1,3(214,210) | F | F | F | 2,2(154,154) | 2,2(154,154) | 2,2(154,154) | 1,3(158,154) | 1,3(158,154) | 2,3(156,154) |
| D3S1311 | 1,2(154,148) | 2,3(148,150) | 1,4(154,136) | 1,3(148,136) | 1,2(148,146) | 3,4(136,138) | 1,2(152,142) | 1,2(152,142) | 1,1(152,152) | 2,4(154,152) | 2,4(154,142) | 2,2(142,142) | 1,2(158,152) | 1,2(158,152) | 2,3(152,148) |
| D4S403 | 2,3(182,174) | 1,3(184,174) | 2,3(182,174) | 1,2(184,182) | 1,2(184,182) | 1,3(184,180) | 2,3(232,226) | 1,3(236,226) | 1,2(236,232) | 2,2 (234,234) | 2,2(234,234) | 2,2 (234,234) | 1,2(236,232) | 1,2(236,232) | 1,3(236,228) |
| D5S2027 | 1,2(192,190) | 1,2(192,190) | 1,2(192,190) | 2,3(196,194) | 1,3(198,194) | 2,2(196,196) | 1,2(172,166) | 1,2(172,166) | 1,2(172,166) | 1,2 (172,170) | 2,3 (170,168) | 1,1(172,172) | 1,1(172,172) | 1,1(172,172) | 1,1(172,172) |
| D6S1610 | 1,2(206,204) | 1,3(206,208) | 1,2(206,204) | 1,1(210,210) | 1,2(210,208) | 1,3(210,198) | 1,3(148,138) | 1,4(148,146) | 2,3(148,138) | 1,2(148,146) | 2,4(146,140) | 1,3(148,144) | 1,2(148,138) | 1,2(148,132) | F |
| D7S519 | 1,2(270,262) | 2,2(262,262) | 1,3(270,266) | 1,2(266,260) | 1,2(266,260) | 1,2(266,260) | 1,3(274,268) | 2,3(270,268) | 1,2(274,270) | 1,2(274,264) | 1,1(274,274) | 2,2(264,264) | 2,3(270,264) | 1,2(274,270) | 1,3(274,264) |
| D9S158 | 2,3(338,336) | 2,3(338,336) | 1,2(342,338) | 1,2 (338,336) | 1,2 (338,336) | 1,2 (338,336) | 2,3(228,226) | 1,2(232,228) | 3,3(226,226) | 1,1(232,232) | 1,1(232,232) | 1,2(232,230) | 1,2(238,234) | 2,3(234,226) | 1,4(238,224) |
| D10S548 | 1,3(194,188) | 1,2(194,192) | 2,3(192,188) | 1,2(190,188) | 1,2(190,188) | 1,2(190,188) | 1,2(154,150) | 1,2(154,152) | 2,2(150,150) | 2,2(150,150) | 1,2(156,150) | 1,2(156,150) | 1,3(156,148) | 1,2(156,150) | 3,3(148,148) |
| D11S898 | 1,2(158,156) | 1,2(158,156) | 1,2(158,156) | 2,3(150,148) | 1,2(158,150) | 1,3(158,148) | 1,2(164,158) | 1,3(164,154) | 1,4(164,160) | 1,3(164,158) | 1,2(164,162) | 2,3(162,158) | 1,2(164,156) | 1,2(164,156) | 1,2(164,156) |
| D13S1265 | 2,3(294,284) | 1,3(300,284) | 2,4(294,280) | 1,2(306,294) | 2,4(306,278) | 1,3(306,292) | 2,3(154,142) | 1,2(162,154) | 1,3(162,142) | 1,1(156,156) | 1,2(156,142) | 1,1(156,156) | 2,3(154,152) | 1,2(158,154) | 3,3(152,152) |
| D14S280 | 1,3(248,244) | 1,1(248,248) | 2,3(246,244) | 2,3(246,242) | 2,3(246,242) | 1,2(248,246) | 1,2(242,240) | 1,2(242,240) | 1,2(242,240) | 1,2(246,244) | 1,3(246,240) | 2,4(244,238) | 1,2(244,240) | 1,2(244,240) | 2,3(240,236) |
| D16S415 | 2,2(226,226) | 2,2(226,226) | 1,2(230,226) | 1,2(228,226) | 1,2(228,226) | 1,1(228,228) | 1,3(246,228) | 2,3(232,228) | 1,3(246,228) | 3,4(230,218) | 1,4(234,218) | 2,3(232,230) | 1,3(234,230) | 2,3(232,230) | 1,3(234,230) |
| D18S474 | 2,2(126,126) | 2,2(126,126) | 1,2(140,126) | 1,2(140,124) | 1,2(140,124) | 1,2(140,124) | 1,3(140,132) | 1,2(140,136) | f | 2,3(132,130) | 1,3(144,130) | 1,2(144,132) | 3,3(130,130) | 1,3(144,130) | 2,3(142,130) |
| | 13/13 correct | | | 13/13 correct | | | 12/13 correct | | | 13/13 correct | | | 12/13 correct | | |

| | 01D4012 | | | 08D7786 | | |
|---|---|---|---|---|---|---|
| Microsatellite | P | M | F | P | M | F |
| D1S2868 | 1,3 (158,154) | 2,3 (156, 154) | 1,2 (158,156) | 1,3(160,158) | 1,2(160,158) | 3,3(158, 158) |
| D3S1311 | 2,3 (156,154) | 3,3 (154, 154) | 1,2 (160, 156) | 2,3 (156,152) | 2,4 (156, 146) | 1,3 (162, 152) |
| D4S403 | 2,4 (234, 228) | 1,4 (236, 228) | 2,3 (234, 232) | 2,3 (236, 228) | 2,2 (236, 236) | 1,3 (240, 228) |
| D5S2027 | 2,2 (166,166) | 1,2 (170, 166) | 2,2 (166,166) | 2,2 (170, 170 | 1,2 (172, 170) | 2,3 (170, 166) |
| D6S1610 | 1,3(148,144) | 2,3(146,144) | 1,3(148,144) | 2,3 (142, 140) | 3,3 (140, 140) | 1,2 (144, 142) |
| D7S519 | 1,2 (276,274) | 1,2 (274,266) | 1,2 (276,266) | 2,3 (268, 264) | 1,2 (276, 268) | 2,3 (268, 264) |
| D9S158 | 1,2 (226,224) | 1,2 (226,224) | 1,2 (226,224) | 1,2 (234, 226) | 1,2 (234, 226) | 1,2 (234, 226 |
| D10S548 | 1,3 (154,148) | 1,2 (154,152) | 3,4 (148,140) | 1,1 (156, 154) | 1,3 (156, 148) | 1,2 (156, 154) |
| D11S898 | 1,3(164, 154) | 2,3(162,154) | 1,2 (164,162) | 1,2 (164, 160) | 1,2 (164, 160) | 1,2 (164, 154) |
| D13S1265 | 2,3 (156,154) | 1,3 (162,154) | 2,3 (165, 142) | 1,2 (160, 142) | 1,2 (160, 142) | 1,2 (160, 142) |
| D14S280 | 1,2 (244,238) | 2,2 (238,238) | 1,2 (244, 238) | 2,2 (240, 240) | 2,2 (240, 240) | 1,2 (242, 240) |
| D16S415 | 2,3 (232,218) | 3,3 (218, 218) | 1,2 (234, 232) | 3,3 (218, 218) | 1,3 (234, 218) | 2,3 (228, 218) |
| D18S474 | 1,3 (146, 142) | 1,4 (146, 132) | 2,3 (144, 142) | 1,3 (146, 130) | 1,2 (146, 134) | 3,3 (130, 130) |
| | 13/13 Correct | | | 13/13 CORRECT | | |

Table 5 Summary of clinical features of *TCF12* mutation positive patients and their family members

| Family | ID | Proband (P), Mother (M), Father (F) | Sex | Mutation | Age at assessment | Diagnosis (SCS, O, FHx, NS) | Craniosynostosis (Y/N) | Pattern of suture fusion (BC, RC, LC, UC, S, L) | Brain anatomy on radiological imaging (CT or MRI) | Normal ICP at invasive monitoring? (if performed) | Number of major craniofacial | Age at first craniofacial procedure | Neurocognitive development | Speech and language developmental quotient | Other characteristics |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | |
| 1 | III-1 | P | F | c.526+1G>A | 16 | NS | Y | RC | normal | Y | 2 | 1 | Normal | Griffiths score = 108 (at 1yo) | Class II malocclusion with dental crowding' bimaxillary osteotomy at 17yo, good outcome/no complications |
| 1 | II-2 | M | F | Negative | 36 | - | - | - | - | - | - | - | Normal | - | - |
| 1 | I-1 | F | M | c.526+1G>A | 63 | - | - | - | - | - | - | - | Normal | - | - |
| | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 2 | II-1 | P | F | c.722C>G; p.S241* | 22 | FHx | Y | RC, S | N/A | n/a | 2 | 0.66 | Mild delay | - | Blepharoptosis requiring repair, strabismus, mild midfacial hypoplasia, class I malocclusion, camptodactyly digits 3+5, brachydactyly; further correction of supraorbital rim and forehead |
| 2 | I-2 | | F | c.722C>G; p.S241* | 48 | FH | - | | | | | | Normal | - | |
| 2 | I-3 | | M | c.722C>G; p.S241* | 53 | FH | - | | | | | | Normal | - | |
| 2 | II-2 | | M | c.722C>G; p.S241* | 27 | FHx | Y | S | normal | n/a | 1 | 1.2 | Mild LD | - | partial sclerosis of both coronal sutures on plain radiography, class II-1 malocclusion |
| | | | | | | | | | | | | | | | |
| 3 | II-2 | P | F | c.778_779delAT; p.M260Vfs*5 | 12 | SCS, FHx | Y | BC | Hypoplastic corpus callosum | n/a | 1 | 0.5 | - | - | Conchal hypertrophy of Right ear |
| 3 | II-1 | | M | c.778_779delAT; p.M260Vfs*5 | 15 | FHx | Y | RC | normal | n/a | 1 | 1 | - | - | L cleft lip, cleft palate |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 3 | I-1 | | M | c.778_779delAT; p.M260Vfs*5 | 47 | FHx | y | LC | n/a | n/a | 0 | - | - | - | bilateral transverse palmar creases |
| | | | | | | | | | | | | | | | |
| 4 | III-1 | P | F | c.812C>A;p.S271* | 7 | FHx | Y | RC | normal | n/a | 1 | 0.6 | normal | - | - |
| | | | | | | | | | | | | | | | |
| 5 | II-1 | P | F | p.822C>G (sp) | 13 | NS | Y | RC | normal | n/a | 1 | 1.3 | normal | - | bilateral parietal foramina |
| | | | | | | | | | | | | | | | |
| 6 | II-1 | P | F | p.825G>C; p.L275F (sp) | 13 | SCS | Y | BC | normal | n/a | 1 | 0.9 | normal | - | bilateral auricular prominence with conchal hypertrophy, dental crowding |
| | | | | | | | | | | | | | | | |
| 7 | II-1 | P | F | p.842_843delCA;p.S281Cfs*56 | 19 | NS | Y | BC | Chiari I malformation, enlarged third and lateral ventricles | Y | 1 | 11 | Normal | WISC-III UK = 111 (at 11.5yr) | |
| | | | | | | | | | | | | | | | |
| 8 | II-1 | P | M | c.842C>G ;p.S281* | 7 | SCS, FHx | Y | RC | Small mass close | n/a | 1 | 1 | normal | - | |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | to pineal gland | | | | | | |
| 8 | I-1 | | M | c.842C>G ;p.S281* | 37 | FHx | Y | LC | n/a | n/a | 0 | - | normal | - | Corneal abnormalities |
| | | | | | | | | | | | | | | |
| 9 | II-1 | P | M | c.842C>G ;p.S281* | 27 | SCS | Y | BC | Mild ventriculomegaly | n/a | 2 | 0.4 | Mild delay | - | low frontal hairline, incomplete descent of testes; additional procedure to correct supraorbital retrusion |
| 9 | I-11 | M | M | Negative | | - | - | - | - | - | - | - | - | - | - |
| 9 | I-1 | F | F | c.842C>G ;p.S281* | | NS | N | - | - | - | - | - | Normal | - | - |
| | | | | | | | | | | | | | | |
| 10 | II-1 | Y | F | c.970G>T;p. G324* | 11 | Fhx | Y | BC | n/a | n/a | 1 | 1 | normal | - | - |
| | | | | | | | | | | | | | | |
| 11 | III-1 | Y | F | c.1035+3G>C (sp) | 3.6 | SCS, FHx | Y | BC | Normal | n/a | 2 | 1.8 | Mild language delay | - | FOAR age 2.0 years following initial posterior release |
| 11 | I-2 | | F | c.1035+3G>C (sp) | 56 | - | - | - | - | - | - | - | Normal | - | Rheumatoid arthritis |
| 11 | II-3 | | F | c.1035+3G>C (sp) | 38 | FHx | - | - | - | - | - | - | normal | - | |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | II-1 | | F | c.1035+3G>C (sp) | 36 | SCS, FHx | Y | BC | n/a | n/a | 0 | - | Need S&LT | - | mild brachydactyly of toes, severe early onset rheumatoid arthritis, Crohn's disease |
| | | | | | | | | | | | | | | | |
| 12 | II-1 | Y | F | c.1073C>G; p.S358* | 6.8 | NS | Y | LC | normal | n/a | 1 | 0.9 | normal | - | low frontal hairline, strabismus, prominent ears with conchal hypertrophy |
| 12 | I-2 | | F | c.1073C>G; p.S358* | 29 | - | - | - | - | | | | | | Facial nerve paralysis |
| | | | | | | | | | | | | | | | |
| 13 | II-1 | Y | M | c. 1115-2A>G (sp) | 17 | SCS, FHx | Y | BC | normal | n/a | 1 | 0.9 | normal | | low frontal hairline, mild mandibular retrusion |
| 13 | I-2 | | F | c. 1115-2A>G (sp) | 49 | SCS, FHx | Y | BC | n/a | | | | normal | | |
| | | | | | | | | | | | | | | | |
| 14 | II-2 | Y | F | c.1123C>T;p.Q375* | 3.5 | FHx | Y | BC | Left paraventricular cyst | n/a | 1 | 0.7 | Mild developmental delay | - | Mild webbed neck, syndactylyl of $2^{nd}/3^{rd}$ toes |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | I-1 | | M | c.1123C>T;p.Q375* | 36 | - | - | - | - | - | - | - | normal | - | |
| 14 | II-1 | | F | c.1123C>T;p.Q375* | 5.3 | FHx | | | Thinning of optic chiasm | n/a | 1 | 0.8 | normal | - | Syndactyly of 2nd/3rd toes |
| | | | | | | | | | | | | | | | |
| 15 | II-1 | Y | F | c.1127G>A;p. W376* | 7.3 | NS | Y | RC | normal | n/a | 1 | 1.1 | normal | Griffiths score = 104 (at 1.1yo) | Left strabismus |
| 15 | I-1 | | M | c.1127G>A;p. W376* | 46 | - | - | - | - | - | - | - | normal | - | - |
| | | | | | | | | | | | | | | | |
| 16 | II-1 | Y | F | c.1283T>G; p. L428* | 12.4 | SCS | Y | BC | normal | n/a | 2 | 1 | normal | - | myopia, small ears, low frontal hairline. Second procedure = FOAR @ 11.9 yr |
| | | | | | | | | | | | | | | | |
| 17 | Ii-1 | Y | M | c.1312delC;p. L438Cfs*81 | 7.1 | SCS | Y | BC | Midline cyst at septum pellucidium, partial agenesis of | n/a | 1 | 0.9 | Normal | CELF-UK socre = 103 at 6.9yo | low frontal hairline, L single palmar crease |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | corpus callosum | | | | | | |
| | | | | | | | | | | | | | | |
| 18 | II-1 | Y | F | c.1328dupT;p.445Tfs*11 | 4.1 | NS | Y | RC | Prominent lateral ventricles | n/a | 1 | 0.9 | Normal | - | Strabismus |
| | | | | | | | | | | | | | | |
| 19 | III-1 | Y | M | c.1349delC;p.P450Lfs*69 | 20 | NS | Y | BC | Normla | Y | 1 | 0.5 | Low average | WISC-III score = 6.3yr | low hairline, small cupped ears with prominent crura, hallux valgus, recurrent absence seizures until mid-teens, abnormal EEG with focal spike and slow wave discharges |
| 19 | II-2 | | F | c.1349delC;p.P450Lfs*69 | 45 | - | - | | Normal | | | | | | small ears, prominent crura helicis, prominent nose with deviated septum, unilateral transverse palmar crease |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 20 | II-1 | Y | F | c.1376T>G; p. L459* | 18 | FHx | Y | BC | Normal | n/a | 1 | 0.7 | Severe LD due to MPSIIIA | Confounded by MPSIIIA | coincidental diagnosis of mucopolysaccharidosis type IIIA (MPSIIIA) |
| 20 | II-2 | | M | c.1376T>G; p. L459* | 18 | FHx | Y | BC | Normal | n/a | 1 | 0.6 | Diagnosis of infantile autism at 2.1yo | exostosis on R tibia; unaffected by MPSIIIA | |
| 20 | I-2 | | F | c.1376T>G; p. L459* | 52 | - | - | | | | | | Normal | | |
| | | | | | | | | | | | | | | | |
| 21 | II-1 | Y | F | c.1453C>T; p.R485* | 0.9 | SCS | Y | BC | Normal | n/a | 1 | 0.7 | Normal | - | low frontal hairline, prominent crura, post-axial polydactyly L hand, broad thumbs, bilateral lamellar lens opacities (also present in unaffected father) |
| | | | | | | | | | | | | | | | |
| | | | | | | | | | | | | | | | |
| | | | | | | | | | | | | | | | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 22 | I-2 | Y | M | c.1468-20T>A; V490Gfs*4 | 54 | FHx | Y | BC | Normal | Y | 0 | | Mild LD | - | myopia, coronary heart disease, type 2 diabetes |
| 22 | II-3 | | F | c.1468-20T>A; V490Gfs*4 | 25 | - | - | | | | | | Normal | - | |
| | | | | | | | | | | | | | | | |
| 23 | III-1 | Y | M | c.1491dupT;p. V498Cfs*12 | 23 | NS | Y | RC | normal | Y | 2 | 0.4 | Normal | Griffiths score=113 (at 5yo) | tall (165 cm), normal pituitary testing, R transverse palmar crease, flat thumbs, hallux valgus, vertical strabismus; additional procedure = onlay cranioplasty of forehead @18 yr |
| 23 | II-1 | | M | c.1491dupT;p. V498Cfs*12 | 37 | - | - | | | | | | Normal | - | |
| | | | | | | | | | | | | | | | |
| 24 | II-1 | Y | M | c.1491dupT;p. V498Cfs*12 | 7.1 | N | Y | BC | Normal | n/a | 1 | 1.5 | Normal | REEL-2 score = 86 (at1.8yo) | dental crowding, bilateral transverse palmar creases, hallux valgus |
| 24 | I-2 | | F | c.1491dupT;p. V498Cfs*12 | 48 | - | - | | | | | | Normal | - | - |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 25 | II-1 | Y | F | c.1514C>G;p.S505* | 10 | SCS | Y | BC | Normal | n/a | 1 | 0.8 | normal | - | prominent Left ear with conchal hypertrophy, Left transverse palmar crease |
| | | | | | | | | | | | | | | | |
| 26 | II-1 | Y | F | c. 1530dupA;p.V511Sfs*11 | 1.4 | NS | Y | RC | Widened CSF spaces | n/a | 1 | 1 | Normal | REEL-3 score =102 (at 1yo) | Left divergent strabismus |
| | | | | | | | | | | | | | | | |
| 27 | III-1 | Y | F | c. 1540_1543dupTCAA; p. S515Ifs*8 | 3.2 | SCS, FHx | Y | RC | Normal | n/a | 1 | 1.1 | Delay in fine and gross motor skills, visual, hearing and language compre-hension@ 3.0 yr. | - | low frontal hairline, beaked nose, R divergent strabismus, hallux valgus, 2nd +4th toes overlap 3rd bilaterally |
| 27 | II-2 | | F | c.1540_1543dupTCAA ;p. S515Ifs*8 | 25 | SCS, FHx | Y | RC | n/a | n/a | 1 | 0.4 | Minor dyslexia/dyspraxia | - | prominent ear crura with folded pinnae, bilateral 5th finger clinodactyly, hallux valgus, |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | incomplete 2/3 toe syndactyly |
| 27 | I-2 | | F | c.1540_1543dupTCAA ;p. S515Ifs*8 | 53 | FHx | Y | RC | - | n/a | 0 | | - | - | facial asymmetry, hallux valgus, severe strabismus as child - operated |
| | | | | | | | | | | | | | | |
| 28 | II-1 | Y | F | 1582+2T>G (sp) | 7.6 | SCS | Y | BC | Normal with prominent extra-axial fluid spaces | n/a | 2 | 0.7 | Normal | Bayley Cognitive scale = 105 (at 2.3yo) | prominent crura helicis, additional procedure = FOAR @ 2.5 yr following initial posterior calvarial release |
| 28 | I-2 | | F | 1582+2T>G (sp) | 43 | - | - | | | | | | Normal | - | |
| | | | | | | | | | | | | | | | |
| 29 | II-1 | Y | M | c.1628delA; p. K543Rfs*20 | 3.9 | NS | Y | RC | Normal | n/a | 1 | 1.3 | normal motor development immature listening, attention, eye contact, social | 72 @ 3.9 yr (verbal compre-hension and expressive language) | (father with orbital hypertelorism and midface hypoplasia presumed affected, TCF12 mutation negative) |

| | | | | | | | | | | | | | commun-ication | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | | |
| 30 | II-1 | Y | F | c. 1642_1645delGAAA;p. E548Rfs*14 | 2.8 | NS | Y | BC | Normal | n/a | 2 | 1.1 | mild language delay | 100 @ 1.1yr (Bayley cognitive scale); 85 @1.1 yr (REEL 3) | additional procedure = FOAR aged 2.8 yr following initial posterior calvarial release/distraction |
| 30 | I-2 | | F | c. 1642_1645delGAAA;p. E548Rfs*14 | 26 | - | - | | | | | | Normal | - | Flat parietal region, orthodontic brace, myopia |
| | | | | | | | | | | | | | | | |
| 31 | I-1 | Y | M | c. 1646delA;p. K549Rfs*14 | 48 | FH | Y | RC | n/a | n/a | 0 | | Normal | - | bilateral transverse palmar creases, brachydactyly of hands |
| 31 | II-1 | | M | c. 1646delA;p. K549Rfs*14 | 13 | FH | Y | S | Chiari I malformation, slightly enlarged | n/a | 1 | 1 | Asperger syndrome | - | bilateral transverse palmar creases, brachydactyly of hands |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | lateral ventricles | | | | | | |
| 31 | II-2 | | M | c. 1646delA;p. K549Rfs*14 | 9 | FH | Y | RC | slightly enlarged lateral ventricles | n/a | 1 | 0.7 | Autism | - | deafness, relapsing respiratory tract infections |
| | | | | | | | | | | | | | | |
| 32 | II-1 | Y | F | c. 1691C>G; p.S564* | 1.6 | NS | Y | BC | Normal | n/a | 1 | 1 | Normal | - | - |
| | | | | | | | | | | | | | | |
| 33 | II-1 | | M | c. 1713delA;p. D572Ifs*19 | 4.8 | NS | Y | RC | Wormian bones | n/a | 1 | 1 | Normal | - | - |
| | | | | | | | | | | | | | | |
| 34 | III-2 | Y | F | c. 1796_1799dupAGAA; p. R602Gfs*6 | 4.2 | FH | Y | BC | normal | n/a | 1 | 1.6 | normal | - | |
| 34 | III-3 | | M | c. 1796_1799dupAGAA; p. R602Gfs*6 | 1.3 | FH | Y | BC | Normal | n/a | 1 | 1.1 | Normal | - | Broad thumbs |
| 34 | II-1 | F | M | c. 1796_1799dupAGAA; p. R602Gfs*6 | 43 | FH | Y | RC | n/a | n/a | 0 | - | - | - | - |
| | | | | | | | | | | | | | | |
| 35 | III-1 | Y | M | c. 1806delG;p. R602Sfs*31 | 2.7 | SCS, FHx | Y | RC | n/a | n/a | 0 | - | Normal | 95 @ 1.2 yr | blepharoptosis, L divergent |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | (REEL 3) | strabismus, L torticollis |
| 35 | II-2 | | F | c. 1806delG;p. R602Sfs*31 | 33 | SCS, FHx | Y | RC | normal | n/a | 1 | 1.3 | normal | - | R sided strabismus (operated as child), myopia, asthma, hypertension |
| | | | | | | | | | | | | | | |
| 36 | | Y | M | c. 1871T>C;p.L624P | 12.9 | NS | Y | LC | Normal | n/a | 1 | 1.3 | Normal | - | R divergent strabismus (R eye amblyopic), asthma |
| 36 | | F | M | c. 1871T>C;p.L624P | 56 | - | - | - | - | - | - | - | Normal | - | - |
| | | | | | | | | | | | | | | |
| 37 | II-1 | Y | F | c. 1912C>G; p.Q638E | 14.2 | SCS, FHx | Y | BC | Normal | n/a | 1 | 0.8 | Normal | - | |
| 37 | I-2 | | F | c. 1912C>G; p.Q638E | 44 | SCS | - | | | | | | Normal | | bilateral blepharoptosis |
| | | | | | | | | | | | | | | |
| 38 | II-1 | Y | M | c. 1963G>T;p. E655* | 3.3 | NS | Y | BC+S | Normal | n/a | 1 | 1.1 | Normal | 107 @ 3.3 yr (REEL 3) | De novo. (mother - bony exostoses around jaw, TCF12 mutation negative) |
| | | | | | | | | | | | | | | |
| 39 | II-1 | P | F | c.268C>T; p.R90* | 2.5 | SCS | Y | RC | Non-spec | N/A | 1 | 0.75 | N/A | N/A | Anal atresia |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | periventricular white matter lesion | | | | | | |
| 39 | | M | F | Negative | | | | | | | | | | | |
| 39 | I-1 | F | M | c.268C>T; p.R90* | | | | | | | | | | | |
| | | | | | | | | | | | | | | | |
| 40 | II-1 | Y | F | c.1036-6G>A (sp) | 4 | SCS | Y | BC | n/a | n/a | ? | ? | - | - | De novo |
| | | | | | | | | | | | | | | | |
| 41 | II-1 | Y | M | c.1453C>T; p.R485* | 3 | NS | Y | BC | Normal | n/a | 1 | 1 | Normal | - | De novo |
| | | | | | | | | | | | | | | | |
| 42 | II-1 | Y | F | *1583-2delA (sp)* | 9 | NS | Y | RC | Normal | n/a | 1 | 2 | LDs ? due to deprivation | Significant LD and speech problems pre-op, now mild expressive language delay | Minor ocular torticollis |
| 42 | I-2 | M | F | *1583-2delA (sp)* | 33 | - | No | - | | | | | | | Mother has isolated cleft lip |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 43 | II-1 | Y | M | c. *1637delA;p. N546Tfs*17* | 5 | NS | Y | RC +S | Normal | n/a | 1 | 4 | Complex neurodevelopmental delay, Significant neglect and understmulation, ~ 2years behind peers, taken into foster care | | Hepatosplenomegaly noted, but no other medical problems; Has non-identical twin brother, no CRS, 3 other sibs, no CF abnormalities |
| 43 | I-2 | M | F | c.*1637delA;p. N546Tfs*17* | ? | NS | N | - | | | - | - | IQ of 69 | | Mother has thin eyebrows, large ear lobes, bilateral single palmar creases; second son died of pineoblastoma |
| 43 | II-6 | | M | c.*1637delA;p. N546Tfs*17* | 15 | NS | N | | | | - | - | No learning difficulties | | Similar appearance to mother |
| | | | | | | | | | | | | | | | |
| 44 | II-1 | Y | F | c.1838G>A;p.R613H | 2 | NS | Y | BC | Normal | n/a | 1 | ? | Normal | ? | ? |
| | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 45 | II-1 | Y | F | c. *1876C>T;p.R626** | 5 | NS | Y | LC | Right cerebellar hypoplasia | n/a | 1 | 3 | Normal | - | Mother has porphyria, proband negative |
| 45 | | M | F | Negative | 44 | - | - | - | - | - | - | - | - | - | - |
| | | | | | | | | | | | | | | | |
| 46 | II-1 | Y | M | c. *1928T>A;p.I643N* | 5 | NS | Y | BC | Normal | n/a | 1 | ? | Significant behavioural problems | - | Still quite asymmetric despite surgical correction |
| 46 | I-1 | F | M | c. *1928T>A;p.I643N* | 43 | NS | N | - | - | - | - | - | - | - | Paternal grandfather apparently mutation positive |
| | | | | | | | | | | | | | | | |
| 47 | II-1 | Y | F | c. *2026_2027ins11;p.E676fs** | 2 | NS | Y | RC | Normal | n/a | 1 | ? | Normal | ? | |
| 47 | I-1 | F | M | c. 2026_2027ins11;p.E676fs* | 37 | NS | N | - | - | - | - | - | - | - | - |
| | | | | | | | | | | | | | | | |

Table 6 *TCF12* Cephalometric Measurements - familial cases only

| Individual | Status in family | Head Circumference(cm) (M=54.73-60.27mm, mean of 57.50mm;F=5 | Inter Pupillary Distance (mm) Male = 57.7mm-69.4mm, mean= 63.5; | Inner Inter-canthal distance (mm) | Outer Inter-Canthal Distance (mm) Male = 88.3-103.0mm;mea | Bicoronal Circumference (cm) | Antero-Posterior Length (cm) | CI/% | Height | *TCF12* mutation +ve |
|---|---|---|---|---|---|---|---|---|---|---|
| Family # 19 III-1 | Proband | 57.4 | 65 | 30 | 110 | | | | 5'6" | Y |
| Family # 19 II-2 | Mother | 57.5 | 60 | 30 | 100 | | | | 5'2" | Y |
| Family # 19 II-1 | Father | 59 | 65 | 35 | 105 | | | | | N |
| Family #30 II-1 | Proband | | | | | | | | | Y |
| Family #30 I-2 | Mother | | 65 | 30 | 105 | | | | 5'1" | Y |
| Family #30 I-1 | Father | | 60 | 35 | 100 | | | | 5'7" | N |
| Family #11 III-1 | Proband | | | | | | | | | Y |
| Family #11 II-3 | Mother | 58 | 60 | 35 | 100 | | | | | Y |
| Family #11 II-2 | Father | 58 | 60 | 35 | 100 | | | | | N |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| Family #11 II-1 | Maternal Aunt | 55 | 65 | 35 | 110 | | | | | Y |
| Family #11 I-1 | Maternal Grandfather | 58.5 | 65 | 30 | 100 | | | | | N |
| Family #11 I-2 | Maternal Grandmother | 53.5 | 60 | 30 | 100 | | | | | Y |
| Family #35 III-1 | Proband | | | | | | | | | Y |
| Family #35 II-2 | Mother | 55 | 65 | 35 | 100 | | | | | Y |
| Family #35 II-1 | Father | 58.5 | 60 | 35 | 105 | | | | | N |
| Family #35 I-2 | Maternal Grandmother | 54.5 | 60 | 30 | 95 | | | | | N |
| Family #35 I-1 | Maternal Grandfather | 59.5 | 65 | 40 | 100 | | | | | N |
| Family #23 III-1 | Proband | 52.6 | | | | | | | | Y |
| Family #23 II-2 | Mother | 56.2 | 55 | 30 | 85 | 14.5 | 17.8 | 81.5 | 159.1 | N |
| Family #23 II-1 | Father | 56.1 | 60 | 30 | 90 | 14.3 | 18 | 79.4 | 180.2 | Y |
| Family #23 I-2 | Paternal grandmother | 56 | 55 | 30 | 90 | 14.6 | 17.9 | 81.6 | 5'6" | ? |
| Family #36 II-1 | Proband | 58.5 | | | | | | | | Y |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Family #36 I-2 | Mother | | | | | | | | N |
| Family #36 I-1 | Father | 55.4 | 65 | 35 | 105 | 15 | 19.2 | 78.1 | Y |
| Family #15 II-1 | Proband | | | | | | | | Y |
| Family #15 I-2 | Mother | 57 | 65 | 35 | 100 | 15 | 19.5 | 76.9 | N |
| Family#15 I-1 | Father | 55 | 60 | 30 | 95 | 15.2 | 18.2 | 83.5 | Y |
| Family #24 II-1 | Proband | 58.4 | 65 | 35 | 115 | 15.2 | 19.5 | 77.8 | Y |
| Family #24 II-3 | Sister | 55.6 | 55 | 30 | 95 | | | | N |
| Family #24 II-2 | Brother | 57 | 60 | 30 | 100 | | | | N |
| Family #24 I-2 | Mother | 54 | 55 | 30 | 95 | | | | Y |
| Family #34 III-3 | Proband | 45 | | | | | | | Y |
| Family #34 III-2 | Brother | 44.5 | | | | | | | Y |
| Family #34 III-1 | Brother | | | | | | | | N |
| Family #34 II-2 | Mother | | | | | | | | N |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Family #34 II-1 | Father | 52 | | | | | | | Y |
| Family #34 I-2 | Paternal grandmother | | | | | | | | N |
| Family #22 I-2 | Proband | 58.4 | 70 | 40 | 125 | | | | Y |
| Family #22 II-3 | Daughter | 56.8 | 70 | 40 | 113 | | | | Y |
| Family #1 III-1 | Proband | | | | | | | | Y |
| Family #1 II-1 | Mother | 55.2 | 60 | 30 | 100 | 14.5 | 18.7 | 77.5 | Y |
| Family #1 I-1 | Maternal Grandmother | | | | | | | | N |
| Family #1 I-1 | Maternal Grandfather | 56.2 | 60 | 35 | 100 | | | | Y |
| Family #20 II-2 | Brother | | | | | | | | Y |
| Family # 20 II-1 | Proband | | | | | | | | Y |
| Family #20 I-1 | Father | | | | | | | | N |
| Family #20 I-2 | Mother | 55.4 | 55 | 25 | 85 | 15.4 | 16.5 | | Y |

Source = ADULTDATA – the Handbook of Adult Anthropometric and Strength Measurements – Data for Design Safety (Values given for mean value and 5$^{th}$-95$^{th}$ centiles). Red = Mutation-positive+ craniosynostosis in proband; Orange = Mutation-positive+ craniosynostosis in adult; Yellow= Mutation-positive+ no craniofacial features in adult; White = mutation-negative.

Table 7 Primers for Parental Origin investigation

| | | | 500bp screening primers – 5' of mutation | | | 500bp screening primers- 3' of mutation | | | | | | | GENOTYPES | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Proband ID | Exon with mutation | *De novo* mutation | Forward screening primer | Reverse screening primer | Amplification conditions | Forward screening primer | Reverse screening primer | Amplification conditions | Informative SNP on trio analysis? | Extended screen? | Informative SNP on extended trio analysis? | SNP ID | Proband | Mother | Father |
| OX2065 | 10 | 822C>G | CAAACAATTGCCAA ATACTACTGCAGTCA T | TGATGTTGCTTATAA AAATTCAAGGGGTA C | 65 | GCTATAACACGT GACTAGGGTACA GCAACA | ACT GAG TGT GGA GGA TAA CTC TAG ATG GA | 65 | Y | N/A | | rs12439017 [T/C] | TC | TT | TC |
| 98D4142 | 10 | 825G>C | CAAACAATTGCCAA ATACTACTGCAGTCA T | TGATGTTGCTTATAA AAATTCAAGGGGTA C | 65 | GCTATAACACGT GACTAGGGTACA GCAACA | ACT GAG TGT GGA GGA TAA CTC TAG ATG GA | 65 | N | N/A | | | | | |
| OX838 | 11 | 842_843 delCA | GCTATAACACGTGA CTAGGGTACAGCAA CA | ACT GAG TGT GGA GGA TAA CTC TAG ATG GA | 65 | CAGCATTCTAGG TGAGCTTTTTGA GTTGGC | GAT AAT AAA AAA GCT TCT TGT CTT CCT GTC | 65 | Y | N/A | | rs2290938 [T/C] | TC | TC | CC |
| 01D3258 | 11 | 842C>G | GCTATAACACGTGA CTAGGGTACAGCAA CA | ACT GAG TGT GGA GGA TAA CTC TAG ATG GA | 65 | CAGCATTCTAGG TGAGCTTTTTGA GTTGGC | GAT AAT AAA AAA GCT TCT TGT CTT CCT GTC | 65 | Y | N/A | | rs2290938 [T/C] | TC | TC | CC |
| GOS5463 | 13 | c.1036-6G>A | CTTCAAGGATGACA GAGTATCTTCTGAGA | CTGTGAGAGGTGAA GGTGATCCAACTGG T | 65 | ACCAGTTGGATC ACCTTCACCTCT CACAG | AGGAGGAAGTT ACATGTACTTTT AAATAG | 65 | Y | N/A | | rs12904933 [T/C] | TC | TT | CC |
| GOS4437 | 16 | 1283T>G | CTATTTCTTTTAGAT GGTGCCTTTTTTGTG | TGT ATG GAT TAA AAA AAA AAC ATG GTG GTT | 65 | GTAGTAAACCCT AAAGATTCTTGT CCTAAA | TAC AGA ATA CAA GCC TTC AGC ATT TAG GGC C | 65 | N | Y | N | | | | |
| OX3956 | 16 | 1312delC | CTATTTCTTTTAGAT GGTGCCTTTTTTGTG | TGT ATG GAT TAA AAA AAA AAC ATG GTG GTT | 65 | GTAGTAAACCCT AAAGATTCTTGT CCTAAA | TAC AGA ATA CAA GCC TTC AGC ATT TAG GGC C | 65 | N | Y | N | | | | |
| OX5189 | 16 | 1453C>T | CTATTTCTTTTAGAT GGTGCCTTTTTTGTG | TGT ATG GAT TAA AAA AAA AAC ATG GTG GTT | 65 | GTAGTAAACCCT AAAGATTCTTGT CCTAAA | TAC AGA ATA CAA GCC TTC AGC ATT TAG GGC C | 65 | Y | N/A | N/A | rs2615227 [C>G] | CG | CC | GG |
| BCH5547 | 16 | 1453C>T | CTATTTCTTTTAGAT GGTGCCTTTTTTGTG | TGT ATG GAT TAA AAA AAA AAC ATG GTG GTT | 65 | GTAGTAAACCCT AAAGATTCTTGT CCTAAA | GTAGTAAACCCT AAAGATTCTTGT CCTAAA | 65 | N | Y | N | | | | |

| | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 01D4012 | 17 | 1514C>G | CAAGGGAGTAGTGA CACAAATAATGCTC AG<br><br>*CTTGGTGTGATAGTTC AAAATGTTATGTAA* | AGG AAA ACC CAA AAA TAA AAA AAG ATT AAG<br><br>*CTG AGC ATT ATT TGT GTC ACT ACT CCC TTG* | 65 | GATATCATTTGG AACACTGTCACC TTTCTC<br><br>*GTC TTA GCT GTA ATT CAT ATA TGA GTC TCG* | CGA GAC TCA TAT ATG AAT TAC AGC TAA GAC<br><br>*GGT ATA CAG GTA TAC AGC CTA GGA ACA AC* | 65 | N | Y | N | | | | |
| OX4855IS | 17 | 1530dupA | CAAGGGAGTAGTGA CACAAATAATGCTC AG<br><br>*CTTGGTGTGATAGTTC AAAATGTTATGTAA* | AGG AAA ACC CAA AAA TAA AAA AAG ATT AAG<br><br>*CTG AGC ATT ATT TGT GTC ACT ACT CCC TTG* | 65 | GATATCATTTGG AACACTGTCACC TTTCTC<br><br>*GTC TTA GCT GTA ATT CAT ATA TGA GTC TCG* | CGA GAC TCA TAT ATG AAT TAC AGC TAA GAC<br><br>*GGT ATA CAG GTA TAC AGC CTA GGA ACA AC* | 65 | N | Y | N | | | | |
| OX4618 | 18 | 1628delA | GATATCATTTGGAAC ACTGTCACCTTTCTC<br><br>*GTC TTA GCT GTA ATT CAT ATA TGA GTC TCG* | CGA GAC TCA TAT ATG AAT TAC AGC TAA GAC<br><br>*GGT ATA CAG GTA TAC AGC CTA GGA ACA AC* | 65 | GTTAGCATCCAG GTTTTAAATTTT ATTCAT<br><br>*CTTACTGTATGTC ATTTACTTTATAAA C* | TTT TAA GCT ATT ATT ACA AGA GTC AGA AAG<br><br>*GCA TGT TAC TGC CTG TGA AGA CAT TCA G* | 65 | N | Y | Y | rs11631084 [G/A]<br><br>rs11636340 [T/A] | GA<br><br>TA | GA<br><br>TA | GG<br><br>TT |
| LAH4951 | 18 | 1691C>G | GATATCATTTGGAAC ACTGTCACCTTTCTC | CGA GAC TCA TAT ATG AAT TAC AGC TAA GAC | 65 | GTTAGCATCCAG GTTTTAAATTTT ATTCAT | TTT TAA GCT ATT ATT ACA AGA GTC AGA AAG | 65 | Y | N/A | N/A | rs56163695 [A/G] | AG | AA | AG |
| 08D7786 | 18 | 1713delA | GATATCATTTGGAAC ACTGTCACCTTTCTC | CGA GAC TCA TAT ATG AAT TAC AGC TAA GAC | 65 | GTTAGCATCCAG GTTTTAAATTTT ATTCAT | TTT TAA GCT ATT ATT ACA AGA GTC AGA AAG | 65 | N | Y | N | | | | |
| OX4594 | 19 | 1806delG | GTACTATGATTTATT GAACCCATTTGCTAT | GGT TTC CTA TAA AGT AAG ATA TGC TGA TTG | 65 | GTTTACAGTTGC TCTTCTGCTTGG AGAAG | GCT CCA TGG CTG TAG TTT TCA AGC ATC TGC | 65 | N | Y | N | | | | |
| OX4379 | 19 | 1963G>T | GTACTATGATTTATT GAACCCATTTGCTAT | GGT TTC CTA TAA AGT AAG ATA TGC TGA TTG | 65 | GTTTACAGTTGC TCTTCTGCTTGG AGAAG | GCT CCA TGG CTG TAG TTT TCA AGC ATC TGC | 65 | N | Y | N | | | | |

N.B. Italicised primers for patient OX4618 indicated a further 500 bp either side of the mutation were screened (i.e. 1kb 5' and 3' of the mutation) to identify informative SNPs. Screening beyond this distance from the mutation was not performed as LD is likely to have broken down.

Table 8 Primers for allele-specific PCR to identify parental origin of mutation. Note (cmn) denotes common primer for trio. Underlined and emboldened nucleotide indicates mismatched base, italicised nucleotide indicates specific base.

| | | | Primer sequence 5'→3' | | | | |
|---|---|---|---|---|---|---|---|
| Family # | *TCF12* mutation | Informative SNP | Forward primer(s) | Reverse primers (s) | Product size (bp) | Annealing temperature (ºC) | Restriction digest/dideoxy seq |
| 5 | 822C>G | rs12439017 [T/C] | GTGTTATGCATTAATAACT**G***T*<br><br>GTGTTATGCATTAATAAC*T***G***C* | GGCTATAACACGTGACTAGGG (cmn) | 525 | 56 (T allele)<br><br>58 (C allele) | HpaII |
| 7 | 842_843delCA | rs2290938 [T>C] | GACCTCCACTTCCCACATGTCTCAATC (cmn) | TCG TGT TTA TGT CTG TTG GTG AAA CT (wt)<br><br>TCG TGT TTA TGT CTG TTG GTG AAA CA (mt) | 388 | 65 | Dideoxy seq |
| 21 | 1453C>T | rs2615227 [C>G] | TCAAACTATGGAGGATCAAG (cmn) | GAATAAAGTTTAAATT**C***G*<br><br>GAATAAAGTTTAAATT**C***C* | 468 | 51 (G allele)<br><br>51 (C allele) | BclI |
| 29 | 1628delA | rs11631084 [G>A]<br><br>rs11636340 [A>T] | TGTCTTAGCTGTAATTCATATATGAGT CTC (cmn) | CTT ACA AAG GCC GGG TGC GGT GGC TCA T**G***C*<br><br>CTT ACA AAG GCC GGG TGC GGT GGC TCT TG*T* | 1654 | Unable to amplify (G allele)<br><br>66 (A allele) | Dideoxy seq |
| 32 | 1691C>G | rs56163695 [A>G] | TTTACCTCTTCCTGTTCTGT**A***A*<br><br>TTTACCTCTTCCTGTTCTGT**A***G* | TTTAATGCTCACTTTCAGGC (cmn) | 816 | 52 (A allele)<br><br>52 (G allele) | AhdI |
| 41 | *c.1036-6G>A* | rs12904933 [C/T] | ACATGTGATCTATTTTATTCCTC**G***C*<br><br>ACATGTGATCTATTTTATTCCTC**G***T* | AAAACAGAAGCCTACCTGTG AGAG (cmn) | 323 | 56 (C allele)<br><br>58 (T allele) | BsaI |

Table 9 Primers for *TCF12* Haplotyping

| Primer sequence 5′→3′ | | | | | |
|---|---|---|---|---|---|
| SNP | Forward | Reverse | Product size (bp) | Amplification conditions | Digest |
| 1 rs4774246 | GAA ATT CTG AGG TTT TAC TTA CCC TAA CA | CAC ATC TCC AAC CCT AAA TCA AAT AGC CAG | 204 | 65 | RsaI |
| 2 rs2585082 | CAA ACA TTC CTT TCA ACC TTC ATC ATA AGA C | AAT GAA GAG GAA GGT GAA ACT AGC TGC TGA | 614 | 59 | BfaI |
| 3 rs1628067 | GTC ACT TGA TGT CCG ACT ACC AGA TAC ACG | GCC ACC GTG CCC AGC CAA CTT TTT TAT TCC | 254 | 65 | MseI |
| 4 rs12901270 | CAG GGT GTC ATT ATT AGA AAA ACA TTA GTT GAT ACT | GAC AGT TGA AGG AAG GCT AAG TCA GCA TTT C | 351 | 60 | BfaI |
| 5 rs2440979 | CAA TGT GGT GGG AGT TTT AGA GCA GGG AC | CTG TCT TGG TGA GAG CCT CCC AAT GGG TTG | 112 | 65 | BstXI |

Table 10 Genotyping results for association study of rs1291270 with craniosynostosis

| | Numbers | | | Proportions | Allele 1 | Allele 2 |
|---|---|---|---|---|---|---|
| | A | C | | | A | C |
| Master Plates | 597 | 223 | | Mixed CRS cohort | 0.825 | 0.175 |
| Mutant | 33 | 7 | 40 | Mutant *TCF12* allele | 0.913 | 0.087 |
| Partner + CRS | 33 | 16 | 49 | Partner TCF12 allele (CRS) | 0.673 | 0.327 |
| Partner + NP | 18 | 7 | 25 | Partner TCF12 allele (non-pen) | 0.72 | 0.28 |
| Bystander | 38 | 3 | 41 | Bystander allele | 0.927 | 0.073 |
| HapMap | 163 | 71 | 234 | HapMap CEU | 0.697 | 0.303 |
| Control Plates | 274 | 96 | 370 | UK Control population | 0.718 | 0.282 |

Table 11 HapMap haplotypes for CEU population

| **CEU ONLY** | SNP 1 | SNP 2 | SNP 3 | SNP 4 | SNP 5 | | |
|---|---|---|---|---|---|---|---|
| | rs447426 | rs2585082 | rs1628067 | rs12901270 | rs2440979 | AUTO | MANUAL |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | T | C GCAT | CGCAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | A | C | C GCAC | CGCAC |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | T | T | A | C | C TTAC | CTTAC |

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|---|---|---|---|---|---|---|---|
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | A | T | C GCAT | CGCAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | C | A | T | T TCAT | TTCAT |
| | T | T | T | A | T | T TTAT | TTTAT |

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|---|---|---|---|---|---|---|---|
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | C | T TTAC | TTTAC |
| | T | T | C | A | C | T TCAC | TTCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |

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|---|---|---|---|---|---|---|---|
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | T | C GCAT | CGCAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |

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|---|---|---|---|---|---|---|---|
| | T | T | C | A | T | T TCAT | TTCAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | T | T | C | A | T | T TCAT | TTCAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | C | A | T | T TCAT | TTCAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | T | C GCAT | CGCAT |
| | N | N | N | N | N | N NNNN | NNNNN |

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|---|---|---|---|---|---|---|---|
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | T | T | A | T | C TTAT | CTTAT |
| | N | N | N | N | N | N NNNN | NNNNN |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | N | N | N | N | N | N NNNN | NNNNN |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | T | T | C | A | C | T TCAC | TTCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | C | T TTAC | TTTAC |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | C | T | T | A | T | C TTAT | CTTAT |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | C | C TTAC | CTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | T | C GCCT | CGCCT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | A | C | C GCAC | CGCAC |
| | T | T | T | A | T | T TTAT | TTTAT |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | T | T | A | T | C TTAT | CTTAT |
| | C | G | C | C | T | C GCCT | CGCCT |
| | T | T | T | A | C | T TTAC | TTTAC |
| | C | G | C | A | C | C GCAC | CGCAC |
| | C | G | C | C | C | C GCCC | CGCCC |
| | C | G | C | A | C | C GCAC | CGCAC |

Table 12

| CEU Population only | |
|---|---|
| NNNNN | 8 |
| **CGCAC** | **64** |
| **CGCAT** | **4** |
| **CGCCC** | **47** |
| **CGCCT** | **24** |
| **CTTAC** | **8** |
| **CTTAT** | **29** |
| **TTCAC** | **2** |
| **TTCAT** | **4** |
| **TTTAC** | **8** |
| **TTTAT** | **44** |
| **Total** | 242 |
| **Total known** | **234** |

Table 13 HapMap Legitimate *TCF12* haplotypes for all ethnic populations

| | SNP 1 | SNP 2 | SNP 3 | SNP 4 | SNP 5 | |
|---|---|---|---|---|---|---|
| phased_haplotypes | rs447426 | rs2585082 | rs1628067 | rs12901270 | rs2440979 | |
| - NA19909_c1 | C | T | T | A | T | CT T A T |
| - NA19909_c2 | C | T | T | A | T | CT T A T |
| - NA19908_c1 | C | T | T | A | T | CT T A T |
| - NA19908_c2 | C | T | T | A | T | CT T A T |
| - NA19901_c1 | C | T | T | A | T | CT T A T |
| - NA19901_c2 | T | G | T | A | T | TG T A T |
| - NA19900_c1 | C | G | C | A | C | CG C A C |
| - NA19900_c2 | T | T | T | A | T | TT T A T |
| - NA20127_c1 | T | T | C | A | T | TT C A T |
| - NA20127_c2 | C | T | T | A | T | CT T A T |
| - NA20126_c1 | T | T | C | A | C | TT C A C |
| - NA20126_c2 | T | G | T | A | T | TG T A T |
| - NA20357_c1 | C | T | T | A | T | CT T A T |
| - NA20357_c2 | C | T | T | A | T | CT T A T |
| - NA20356_c1 | C | T | T | A | T | CT T A T |
| - NA20356_c2 | T | T | C | A | C | TT C A C |
| - NA19704_c1 | C | G | C | A | C | CG C A C |
| - NA19704_c2 | T | T | C | A | C | TT C A C |
| - NA19703_c1 | T | T | C | A | T | TT C A T |
| - NA19703_c2 | T | T | C | A | C | TT C A C |
| - NA19835_c1 | C | T | T | A | T | CT T A T |
| - NA19835_c2 | C | T | T | A | T | CT T A T |
| - NA19834_c1 | C | T | T | A | T | CT T A T |
| - NA19834_c2 | C | T | T | A | T | CT T A T |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA19701_c1 | C | T | T | A | T | CT T A T |
| - NA19701_c2 | T | G | T | A | T | TG T A T |
| - NA19700_c1 | T | T | C | A | C | TT C A C |
| - NA19700_c2 | C | T | T | A | T | CT T A T |
| - NA19917_c1 | C | G | C | A | C | CG C A C |
| - NA19917_c2 | C | T | T | A | T | CT T A T |
| - NA19916_c1 | C | T | T | A | T | CT T A T |
| - NA19916_c2 | C | T | T | A | T | CT T A T |
| - NA19819_c1 | C | G | C | C | C | CG C C C |
| - NA19819_c2 | C | T | T | A | T | CT T A T |
| - NA19818_c1 | T | G | T | A | T | TG T A T |
| - NA19818_c2 | C | T | T | A | C | CT T A C |
| - NA19713_c1 | C | T | T | A | T | CT T A T |
| - NA19713_c2 | C | T | T | A | T | CT T A T |
| - NA19982_c1 | T | T | T | A | T | TT T A T |
| - NA19982_c2 | T | T | T | A | T | TT T A T |
| - NA20289_c1 | C | T | T | A | T | CT T A T |
| - NA20289_c2 | T | T | C | A | T | TT C A T |
| - NA20289_0_c1 | C | T | T | A | T | CT T A T |
| - NA20289_0_c2 | N | N | N | N | N | NN N N N |
| - NA20334_c1 | C | G | C | A | C | CG C A C |
| - NA20334_c2 | C | T | T | A | T | CT T A T |
| - NA20334_0_c1 | C | T | T | A | T | CT T A T |
| - NA20334_0_c2 | N | N | N | N | N | NN N N N |
| - NA20336_c1 | T | T | T | A | T | TT T A T |
| - NA20336_c2 | C | T | T | A | T | CT T A T |
| - NA20336_0_c1 | C | T | T | A | C | CT T A C |
| - NA20336_0_c2 | N | N | N | N | N | NN N N N |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA20294_c1 | C | T | T | A | T | CT T A T |
| - NA20294_c2 | C | G | C | C | C | CG C C C |
| - NA20294_0_c1 | T | G | T | A | T | TG T A T |
| - NA20294_0_c2 | N | N | N | N | N | NN N N N |
| - NA20276_c1 | T | T | C | A | C | TT C A C |
| - NA20276_c2 | C | T | T | A | T | CT T A T |
| - NA20276_0_c1 | T | G | T | A | T | TG T A T |
| - NA20276_0_c2 | N | N | N | N | N | NN N N N |
| - NA20332_c1 | C | G | C | C | C | CG C C C |
| - NA20332_c2 | C | T | T | A | T | CT T A T |
| - NA20332_0_c1 | T | T | T | A | T | TT T A T |
| - NA20332_0_c2 | N | N | N | N | N | NN N N N |
| - NA20291_c1 | C | T | T | A | C | CT T A C |
| - NA20291_c2 | C | T | T | A | T | CT T A T |
| - NA20291_0_c1 | T | T | T | A | C | TT T A C |
| - NA20291_0_c2 | N | N | N | N | N | NN N N N |
| - NA20342_c1 | C | T | T | A | T | CT T A T |
| - NA20342_c2 | T | G | T | A | T | TG T A T |
| - NA20342_0_c1 | C | G | C | A | C | CG C A C |
| - NA20342_0_c2 | N | N | N | N | N | NN N N N |
| - NA20287_c1 | C | T | T | A | T | CT T A T |
| - NA20287_c2 | C | T | T | A | T | CT T A T |
| - NA20287_0_c1 | C | T | T | A | T | CT T A T |
| - NA20287_0_c2 | N | N | N | N | N | NN N N N |
| - NA20349_c1 | C | T | T | A | T | CT T A T |
| - NA20349_c2 | T | T | T | A | T | TT T A T |
| - NA20349_0_c1 | C | T | T | A | T | CT T A T |
| - NA20349_0_c2 | N | N | N | N | N | NN N N N |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA19985_c1 | C | T | T | A | T | CT T A T |
| - NA19985_c2 | C | T | T | A | T | CT T A T |
| - NA19985_0_c1 | C | T | T | A | T | CT T A T |
| - NA19985_0_c2 | N | N | N | N | N | NN N N N |
| - NA20282_c1 | T | T | T | A | T | TT T A T |
| - NA20282_c2 | C | T | C | A | C | CT C A C |
| - NA20282_0_c1 | C | T | C | A | T | CT C A T |
| - NA20282_0_c2 | N | N | N | N | N | NN N N N |
| - NA19921_c1 | T | T | T | A | T | TT T A T |
| - NA19921_c2 | C | G | C | C | C | CG C C C |
| - NA19921_NA19920_c1 | C | T | T | A | T | CT T A T |
| - NA19921_NA19920_c2 | N | N | N | N | N | NN N N N |
| - NA20359_c1 | C | T | T | A | T | CT T A T |
| - NA20359_c2 | C | T | T | A | T | CT T A T |
| - NA20359_0_c1 | C | G | C | A | C | CG C A C |
| - NA20359_0_c2 | N | N | N | N | N | NN N N N |
| - NA20344_c1 | C | T | T | A | T | CT T A T |
| - NA20344_c2 | C | T | T | A | T | CT T A T |
| - NA20344_0_c1 | T | T | C | A | C | TT C A C |
| - NA20344_0_c2 | N | N | N | N | N | NN N N N |
| - NA20317_c1 | T | T | C | A | C | TT C A C |
| - NA20317_c2 | C | T | T | A | T | CT T A T |
| - NA20317_0_c1 | T | T | C | A | C | TT C A C |
| - NA20317_0_c2 | N | N | N | N | N | NN N N N |
| - NA20346_c1 | T | T | T | A | T | TT T A T |
| - NA20346_c2 | T | G | T | A | T | TG T A T |
| - NA20346_0_c1 | C | T | T | A | T | CT T A T |
| - NA20346_0_c2 | N | N | N | N | N | NN N N N |

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|---|---|---|---|---|---|---|
| - NA20301_c1 | T | G | T | A | T | TG T A T |
| - NA20301_c2 | C | T | T | A | T | CT T A T |
| - NA20301_0_c1 | C | T | T | A | T | CT T A T |
| - NA20301_0_c2 | N | N | N | N | N | NN N N N |
| - NA19914_c1 | C | T | T | A | T | CT T A T |
| - NA19914_c2 | C | T | T | A | T | CT T A T |
| - NA19914_0_c1 | C | T | T | A | T | CT T A T |
| - NA19914_0_c2 | N | N | N | N | N | NN N N N |
| - NA20363_c1 | C | T | T | A | T | CT T A T |
| - NA20363_c2 | C | T | T | A | T | CT T A T |
| - NA20363_0_c1 | C | T | T | A | T | CT T A T |
| - NA20363_0_c2 | N | N | N | N | N | NN N N N |
| - NA19904_c1 | C | T | T | A | C | CT T A C |
| - NA19904_c2 | C | G | C | C | C | CG C C C |
| - NA20340_c1 | T | T | C | A | C | TT C A C |
| - NA20340_c2 | T | T | T | A | T | TT T A T |
| - NA20297_c1 | T | T | C | A | C | TT C A C |
| - NA20297_c2 | C | T | T | A | T | CT T A T |
| - NA20281_c1 | C | T | T | A | T | CT T A T |
| - NA20281_c2 | T | T | C | A | C | TT C A C |
| - NA20348_c1 | C | G | C | C | C | CG C C C |
| - NA20348_c2 | C | T | T | A | T | CT T A T |
| - NA20300_c1 | T | T | C | A | C | TT C A C |
| - NA20300_c2 | C | T | T | A | T | CT T A T |
| - NA19625_c1 | T | T | T | A | C | TT T A C |
| - NA19625_c2 | C | G | C | A | C | CG C A C |
| - NA20279_c1 | C | T | T | A | T | CT T A T |
| - NA20279_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19712_c1 | C | T | T | A | T | CT T A T |
| - NA19712_c2 | T | T | T | A | T | TT T A T |
| - NA20322_c1 | T | T | T | A | T | TT T A T |
| - NA20322_c2 | C | T | T | A | T | CT T A T |
| - NA19711_c1 | C | T | T | A | T | CT T A T |
| - NA19711_c2 | T | T | T | A | T | TT T A T |
| - NA19708_c1 | C | T | T | A | T | CT T A T |
| - NA19708_c2 | T | G | T | A | T | TG T A T |
| - NA20341_c1 | T | T | C | A | C | TT C A C |
| - NA20341_c2 | C | G | C | C | C | CG C C C |
| - NA12749_c1 | C | G | C | C | C | CG C C C |
| - NA12749_c2 | C | G | C | C | C | CG C C C |
| - NA12748_c1 | C | T | T | A | C | CT T A C |
| - NA12748_c2 | C | G | C | C | T | CG C C T |
| - NA12890_c1 | C | T | T | A | T | CT T A T |
| - NA12890_c2 | T | T | T | A | T | TT T A T |
| - NA12889_c1 | C | G | C | C | C | CG C C C |
| - NA12889_c2 | T | T | T | A | T | TT T A T |
| - NA11931_c1 | T | T | T | A | T | TT T A T |
| - NA11931_c2 | C | T | T | A | T | CT T A T |
| - NA11930_c1 | C | T | T | A | T | CT T A T |
| - NA11930_c2 | T | T | T | A | T | TT T A T |
| - NA12348_c1 | C | G | C | A | C | CG C A C |
| - NA12348_c2 | C | G | C | A | C | CG C A C |
| - NA12347_c1 | C | G | C | C | T | CG C C T |
| - NA12347_c2 | C | T | T | A | T | CT T A T |
| - NA12273_c1 | C | G | C | C | C | CG C C C |
| - NA12273_c2 | C | G | C | C | C | CG C C C |

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|---|---|---|---|---|---|---|
| - NA12272_c1 | C | G | C | A | C | CG C A C |
| - NA12272_c2 | C | G | C | C | C | CG C C C |
| - NA12776_c1 | C | G | C | C | C | CG C C C |
| - NA12776_c2 | C | G | C | A | C | CG C A C |
| - NA12775_c1 | C | G | C | A | C | CG C A C |
| - NA12775_c2 | C | G | C | C | T | CG C C T |
| - NA12828_c1 | C | G | C | A | C | CG C A C |
| - NA12828_c2 | C | G | C | A | C | CG C A C |
| - NA12827_c1 | T | T | T | A | T | TT T A T |
| - NA12827_c2 | C | G | C | C | C | CG C C C |
| - NA12287_c1 | C | G | C | A | C | CG C A C |
| - NA12287_c2 | C | G | C | A | C | CG C A C |
| - NA12286_c1 | C | G | C | C | T | CG C C T |
| - NA12286_c2 | C | T | T | A | T | CT T A T |
| - NA07045_c1 | C | G | C | C | C | CG C C C |
| - NA07045_c2 | C | T | T | A | C | CT T A C |
| - NA06986_c1 | C | T | T | A | T | CT T A T |
| - NA06986_c2 | C | G | C | C | T | CG C C T |
| - NA12343_c1 | C | G | C | C | T | CG C C T |
| - NA12343_c2 | C | G | C | A | C | CG C A C |
| - NA12342_c1 | C | G | C | C | T | CG C C T |
| - NA12342_c2 | C | G | C | C | T | CG C C T |
| - NA07346_c1 | C | G | C | C | T | CG C C T |
| - NA07346_c2 | C | T | T | A | T | CT T A T |
| - NA07347_c1 | T | T | T | A | T | TT T A T |
| - NA07347_c2 | T | T | T | A | T | TT T A T |
| - NA12341_c1 | C | G | C | C | C | CG C C C |
| - NA12341_c2 | C | G | C | A | T | CG C A T |

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|---|---|---|---|---|---|---|
| - NA12340_c1 | C | G | C | C | C | CG C C C |
| - NA12340_c2 | C | T | T | A | T | CT T A T |
| - NA12843_c1 | T | T | T | A | T | TT T A T |
| - NA12843_c2 | C | G | C | C | C | CG C C C |
| - NA12842_c1 | C | G | C | C | T | CG C C T |
| - NA12842_c2 | C | G | C | A | C | CG C A C |
| - NA11892_c1 | C | G | C | C | C | CG C C C |
| - NA11892_c2 | C | G | C | A | C | CG C A C |
| - NA11891_c1 | C | G | C | A | C | CG C A C |
| - NA11891_c2 | T | T | T | A | T | TT T A T |
| - NA11894_c1 | C | G | C | A | C | CG C A C |
| - NA11894_c2 | C | G | C | A | C | CG C A C |
| - NA11893_c1 | C | G | C | A | C | CG C A C |
| - NA11893_c2 | C | G | C | A | C | CG C A C |
| - NA12830_c1 | C | T | T | A | T | CT T A T |
| - NA12830_c2 | C | G | C | A | C | CG C A C |
| - NA12829_c1 | T | T | T | A | T | TT T A T |
| - NA12829_c2 | C | G | C | A | C | CG C A C |
| - NA11920_c1 | C | T | T | A | C | CT T A C |
| - NA11920_c2 | C | G | C | A | C | CG C A C |
| - NA11919_c1 | T | T | T | A | T | TT T A T |
| - NA11919_c2 | T | T | T | A | T | TT T A T |
| - NA07031_c1 | C | G | C | A | C | CG C A C |
| - NA07031_c2 | C | G | C | A | C | CG C A C |
| - NA07051_c1 | C | G | C | A | C | CG C A C |
| - NA07051_c2 | C | T | T | A | T | CT T A T |
| - NA12778_c1 | T | T | T | A | T | TT T A T |
| - NA12778_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA12777_c1 | T | T | T | A | T | TT T A T |
| - NA12777_c2 | T | T | T | A | T | TT T A T |
| - NA12400_c1 | T | T | T | A | T | TT T A T |
| - NA12400_c2 | C | G | C | C | C | CG C C C |
| - NA12399_c1 | C | G | C | C | T | CG C C T |
| - NA12399_c2 | C | T | T | A | C | CT T A C |
| - NA07037_c1 | C | G | C | C | T | CG C C T |
| - NA07037_c2 | C | G | C | C | C | CG C C C |
| - NA07435_c1 | T | T | T | A | T | TT T A T |
| - NA07435_c2 | C | G | C | A | C | CG C A C |
| - NA12489_c1 | C | G | C | C | C | CG C C C |
| - NA12489_c2 | C | G | C | C | C | CG C C C |
| - NA12546_c1 | C | G | C | A | C | CG C A C |
| - NA12546_c2 | T | T | T | A | C | TT T A C |
| - NA12763_c1 | C | G | C | C | C | CG C C C |
| - NA12763_c2 | C | G | C | C | T | CG C C T |
| - NA12762_c1 | T | T | T | A | T | TT T A T |
| - NA12762_c2 | C | G | C | A | C | CG C A C |
| - NA11882_c1 | C | G | C | A | C | CG C A C |
| - NA11882_c2 | T | T | T | A | T | TT T A T |
| - NA11881_c1 | T | T | T | A | T | TT T A T |
| - NA11881_c2 | C | G | C | C | C | CG C C C |
| - NA07345_c1 | C | G | C | A | C | CG C A C |
| - NA07345_c2 | C | T | T | A | T | CT T A T |
| - NA07357_c1 | C | T | T | A | T | CT T A T |
| - NA07357_c2 | C | G | C | A | C | CG C A C |
| - NA12892_c1 | C | G | C | C | T | CG C C T |
| - NA12892_c2 | C | G | C | C | C | CG C C C |

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|---|---|---|---|---|---|---|
| - NA12891_c1 | T | T | T | A | T | TT T A T |
| - NA12891_c2 | C | T | T | A | T | CT T A T |
| - NA11830_c1 | C | G | C | A | T | CG C A T |
| - NA11830_c2 | C | G | C | C | C | CG C C C |
| - NA11829_c1 | T | T | C | A | T | TT C A T |
| - NA11829_c2 | T | T | T | A | T | TT T A T |
| - NA12249_c1 | C | T | T | A | T | CT T A T |
| - NA12249_c2 | C | T | T | A | C | CT T A C |
| - NA12248_c1 | C | T | T | A | T | CT T A T |
| - NA12248_c2 | T | T | T | A | C | TT T A C |
| - NA12006_c1 | C | G | C | C | T | CG C C T |
| - NA12006_c2 | T | T | T | A | T | TT T A T |
| - NA12005_c1 | C | T | T | A | T | CT T A T |
| - NA12005_c2 | C | T | T | A | T | CT T A T |
| - NA12234_c1 | T | T | T | A | T | TT T A T |
| - NA12234_c2 | C | G | C | C | C | CG C C C |
| - NA12264_c1 | C | G | C | C | C | CG C C C |
| - NA12264_c2 | T | T | T | A | C | TT T A C |
| - NA12239_c1 | C | G | C | A | C | CG C A C |
| - NA12239_c2 | T | T | T | A | C | TT T A C |
| - NA12146_c1 | C | G | C | C | T | CG C C T |
| - NA12146_c2 | C | G | C | A | C | CG C A C |
| - NA12156_c1 | T | T | T | A | T | TT T A T |
| - NA12156_c2 | T | T | T | A | T | TT T A T |
| - NA12155_c1 | T | T | T | A | C | TT T A C |
| - NA12155_c2 | T | T | C | A | C | TT C A C |
| - NA06985_c1 | T | T | T | A | T | TT T A T |
| - NA06985_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA06993_c1 | C | T | T | A | T | CT T A T |
| - NA06993_c2 | C | T | T | A | T | CT T A T |
| - NA12815_c1 | C | G | C | C | C | CG C C C |
| - NA12815_c2 | C | G | C | C | T | CG C C T |
| - NA12814_c1 | T | T | T | A | T | TT T A T |
| - NA12814_c2 | C | G | C | C | C | CG C C C |
| - NA11832_c1 | C | G | C | A | C | CG C A C |
| - NA11832_c2 | C | G | C | A | T | CG C A T |
| - NA11831_c1 | C | T | T | A | T | CT T A T |
| - NA11831_c2 | C | T | T | A | C | CT T A C |
| - NA12873_c1 | C | G | C | C | C | CG C C C |
| - NA12873_c2 | C | G | C | A | C | CG C A C |
| - NA12872_c1 | C | G | C | C | C | CG C C C |
| - NA12872_c2 | C | G | C | A | C | CG C A C |
| - NA11995_c1 | C | G | C | C | T | CG C C T |
| - NA11995_c2 | T | T | T | A | T | TT T A T |
| - NA11994_c1 | C | G | C | C | T | CG C C T |
| - NA11994_c2 | C | G | C | C | T | CG C C T |
| - NA07000_c1 | C | G | C | C | C | CG C C C |
| - NA07000_c2 | C | G | C | C | C | CG C C C |
| - NA06994_c1 | C | G | C | A | C | CG C A C |
| - NA06994_c2 | C | G | C | A | C | CG C A C |
| - NA12145_c1 | T | T | T | A | T | TT T A T |
| - NA12145_c2 | C | G | C | C | T | CG C C T |
| - NA12144_c1 | T | T | T | A | T | TT T A T |
| - NA12144_c2 | C | G | C | A | C | CG C A C |
| - NA12813_c1 | C | G | C | C | C | CG C C C |
| - NA12813_c2 | C | T | T | A | C | CT T A C |

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|---|---|---|---|---|---|---|
| - NA12812_c1 | C | G | C | A | C | CG C A C |
| - NA12812_c2 | C | G | C | A | C | CG C A C |
| - NA12751_c1 | C | G | C | A | C | CG C A C |
| - NA12751_c2 | C | G | C | C | C | CG C C C |
| - NA12750_c1 | C | G | C | C | C | CG C C C |
| - NA12750_c2 | C | G | C | A | C | CG C A C |
| - NA12761_c1 | T | T | C | A | T | TT C A T |
| - NA12761_c2 | C | G | C | A | C | CG C A C |
| - NA12760_c1 | T | T | T | A | T | TT T A T |
| - NA12760_c2 | C | G | C | A | C | CG C A C |
| - NA11840_c1 | T | T | T | A | T | TT T A T |
| - NA11840_c2 | T | T | C | A | T | TT C A T |
| - NA11839_c1 | C | G | C | A | C | CG C A C |
| - NA11839_c2 | C | G | C | A | C | CG C A C |
| - NA12875_c1 | C | G | C | C | C | CG C C C |
| - NA12875_c2 | T | T | C | A | T | TT C A T |
| - NA12874_c1 | C | G | C | A | C | CG C A C |
| - NA12874_c2 | C | T | T | A | T | CT T A T |
| - NA12718_c1 | T | T | T | A | T | TT T A T |
| - NA12718_c2 | C | G | C | C | C | CG C C C |
| - NA12718_0_c1 | C | T | T | A | T | CT T A T |
| - NA12718_0_c2 | N | N | N | N | N | NN N N N |
| - NA12045_c1 | C | G | C | C | C | CG C C C |
| - NA12045_c2 | T | T | T | A | T | TT T A T |
| - NA12045_0_c1 | C | G | C | A | C | CG C A C |
| - NA12045_0_c2 | N | N | N | N | N | NN N N N |
| - NA11843_c1 | C | G | C | C | C | CG C C C |
| - NA11843_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA11843_0_c1 | C | G | C | A | C | CG C A C |
| - NA11843_0_c2 | N | N | N | N | N | NN N N N |
| - NA12383_c1 | C | G | C | A | C | CG C A C |
| - NA12383_c2 | C | G | C | C | C | CG C C C |
| - NA12383_0_c1 | C | G | C | A | T | CG C A T |
| - NA12383_0_c2 | N | N | N | N | N | NN N N N |
| - NA12716_c1 | C | G | C | A | C | CG C A C |
| - NA12716_c2 | C | G | C | A | C | CG C A C |
| - NA12716_NA12717_c1 | C | T | T | A | T | CT T A T |
| - NA12716_NA12717_c2 | N | N | N | N | N | NN N N N |
| - NA12275_c1 | C | G | C | C | C | CG C C C |
| - NA12275_c2 | C | G | C | C | C | CG C C C |
| - NA12275_NA12274_c1 | C | G | C | A | C | CG C A C |
| - NA12275_NA12274_c2 | N | N | N | N | N | NN N N N |
| - NA12003_c1 | C | G | C | C | C | CG C C C |
| - NA12003_c2 | T | T | T | A | T | TT T A T |
| - NA12003_NA12004_c1 | C | T | T | A | T | CT T A T |
| - NA12003_NA12004_c2 | N | N | N | N | N | NN N N N |
| - NA12154_c1 | T | T | T | A | T | TT T A T |
| - NA12154_c2 | C | G | C | C | C | CG C C C |
| - NA12154_NA12236_c1 | C | G | C | A | C | CG C A C |
| - NA12154_NA12236_c2 | N | N | N | N | N | NN N N N |
| - NA06989_c1 | C | G | C | A | C | CG C A C |
| - NA06989_c2 | C | G | C | C | T | CG C C T |
| - NA10850_c1 | T | T | T | A | T | TT T A T |
| - NA10850_c2 | C | G | C | A | C | CG C A C |
| - NA06984_c1 | C | G | C | C | C | CG C C C |
| - NA06984_c2 | T | T | C | A | C | TT C A C |

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|---|---|---|---|---|---|---|
| - NA11917_c1 | C | G | C | C | C | CG C C C |
| - NA11917_c2 | C | T | T | A | T | CT T A T |
| - NA12282_c1 | T | T | T | A | T | TT T A T |
| - NA12282_c2 | C | G | C | A | C | CG C A C |
| - NA12283_c1 | C | T | T | A | T | CT T A T |
| - NA12283_c2 | T | T | T | A | C | TT T A C |
| - NA11918_c1 | C | T | T | A | T | CT T A T |
| - NA11918_c2 | T | T | T | A | C | TT T A C |
| - NA12413_c1 | C | G | C | A | C | CG C A C |
| - NA12413_c2 | T | T | T | A | T | TT T A T |
| - NA07056_c1 | C | G | C | A | C | CG C A C |
| - NA07056_c2 | C | T | T | A | C | CT T A C |
| - NA12044_c1 | C | G | C | A | C | CG C A C |
| - NA12044_c2 | C | G | C | C | T | CG C C T |
| - NA11992_c1 | C | G | C | C | C | CG C C C |
| - NA11992_c2 | C | G | C | A | C | CG C A C |
| - NA07022_c1 | T | T | T | A | T | TT T A T |
| - NA07022_c2 | C | G | C | A | C | CG C A C |
| - NA07055_c1 | T | T | T | A | T | TT T A T |
| - NA07055_c2 | C | G | C | C | C | CG C C C |
| - NA12056_c1 | C | G | C | C | C | CG C C C |
| - NA12056_c2 | C | G | C | A | C | CG C A C |
| - NA11993_c1 | C | T | T | A | T | CT T A T |
| - NA11993_c2 | C | G | C | C | T | CG C C T |
| - NA12057_c1 | T | T | T | A | C | TT T A C |
| - NA12057_c2 | C | G | C | A | C | CG C A C |
| - NA12043_c1 | C | G | C | C | C | CG C C C |
| - NA12043_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA18597_c1 | C | G | C | A | C | CG C A C |
| - NA18597_c2 | C | T | T | A | T | CT T A T |
| - NA18615_c1 | C | T | T | A | T | CT T A T |
| - NA18615_c2 | C | G | C | A | C | CG C A C |
| - NA18557_c1 | C | G | C | A | C | CG C A C |
| - NA18557_c2 | C | T | T | A | T | CT T A T |
| - NA18628_c1 | C | T | T | A | T | CT T A T |
| - NA18628_c2 | C | G | C | A | C | CG C A C |
| - NA18745_c1 | C | T | T | A | T | CT T A T |
| - NA18745_c2 | C | G | C | A | C | CG C A C |
| - NA18640_c1 | C | G | C | C | C | CG C C C |
| - NA18640_c2 | C | G | C | C | C | CG C C C |
| - NA18747_c1 | C | T | T | A | T | CT T A T |
| - NA18747_c2 | T | T | T | A | T | TT T A T |
| - NA18596_c1 | C | T | T | A | T | CT T A T |
| - NA18596_c2 | C | G | C | A | C | CG C A C |
| - NA18536_c1 | T | T | C | A | C | TT C A C |
| - NA18536_c2 | C | T | T | A | T | CT T A T |
| - NA18599_c1 | C | T | T | A | T | CT T A T |
| - NA18599_c2 | C | T | T | A | T | CT T A T |
| - NA18544_c1 | C | T | T | A | T | CT T A T |
| - NA18544_c2 | T | T | T | A | T | TT T A T |
| - NA18602_c1 | C | G | C | C | C | CG C C C |
| - NA18602_c2 | C | G | C | C | T | CG C C T |
| - NA18614_c1 | T | T | T | A | C | TT T A C |
| - NA18614_c2 | C | T | T | A | C | CT T A C |
| - NA18548_c1 | T | T | T | A | T | TT T A T |
| - NA18548_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA18616_c1 | T | T | T | A | C | TT T A C |
| - NA18616_c2 | C | G | C | C | T | CG C C T |
| - NA18559_c1 | T | T | T | A | T | TT T A T |
| - NA18559_c2 | T | T | T | A | C | TT T A C |
| - NA18619_c1 | C | T | T | A | T | CT T A T |
| - NA18619_c2 | C | G | C | A | C | CG C A C |
| - NA18638_c1 | C | G | C | C | T | CG C C T |
| - NA18638_c2 | C | T | T | A | T | CT T A T |
| - NA18639_c1 | C | T | T | A | T | CT T A T |
| - NA18639_c2 | C | T | T | A | T | CT T A T |
| - NA18627_c1 | C | G | C | A | C | CG C A C |
| - NA18627_c2 | C | T | T | A | T | CT T A T |
| - NA18631_c1 | C | T | T | A | T | CT T A T |
| - NA18631_c2 | C | G | C | A | C | CG C A C |
| - NA18634_c1 | C | G | C | C | C | CG C C C |
| - NA18634_c2 | C | T | T | A | T | CT T A T |
| - NA18642_c1 | C | T | T | A | T | CT T A T |
| - NA18642_c2 | T | T | C | A | C | TT C A C |
| - NA18626_c1 | T | T | C | A | C | TT C A C |
| - NA18626_c2 | C | T | T | A | T | CT T A T |
| - NA18543_c1 | T | T | T | A | T | TT T A T |
| - NA18543_c2 | C | G | C | A | C | CG C A C |
| - NA18610_c1 | C | T | T | A | T | CT T A T |
| - NA18610_c2 | C | G | C | C | C | CG C C C |
| - NA18617_c1 | C | G | C | A | C | CG C A C |
| - NA18617_c2 | T | T | T | A | C | TT T A C |
| - NA18613_c1 | T | T | T | A | C | TT T A C |
| - NA18613_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18647_c1 | C | T | T | A | T | CT T A T |
| - NA18647_c2 | C | T | T | A | T | CT T A T |
| - NA18630_c1 | T | T | T | A | T | TT T A T |
| - NA18630_c2 | C | T | T | A | T | CT T A T |
| - NA18641_c1 | C | G | C | A | C | CG C A C |
| - NA18641_c2 | C | G | C | A | C | CG C A C |
| - NA18748_c1 | C | G | C | A | C | CG C A C |
| - NA18748_c2 | C | T | T | A | T | CT T A T |
| - NA18749_c1 | C | T | T | A | C | CT T A C |
| - NA18749_c2 | C | T | T | A | T | CT T A T |
| - NA18757_c1 | C | G | C | C | C | CG C C C |
| - NA18757_c2 | C | G | C | A | C | CG C A C |
| - NA18546_c1 | T | T | T | A | T | TT T A T |
| - NA18546_c2 | C | T | T | A | C | CT T A C |
| - NA18643_c1 | C | T | T | A | T | CT T A T |
| - NA18643_c2 | C | G | C | A | C | CG C A C |
| - NA18645_c1 | C | T | T | A | C | CT T A C |
| - NA18645_c2 | C | T | T | A | T | CT T A T |
| - NA18534_c1 | C | G | C | C | T | CG C C T |
| - NA18534_c2 | C | G | C | A | C | CG C A C |
| - NA18595_c1 | C | G | C | A | T | CG C A T |
| - NA18595_c2 | C | T | T | A | T | CT T A T |
| - NA18618_c1 | C | G | C | A | C | CG C A C |
| - NA18618_c2 | C | G | C | A | C | CG C A C |
| - NA18740_c1 | C | G | C | A | C | CG C A C |
| - NA18740_c2 | T | T | T | A | T | TT T A T |
| - NA18524_c1 | T | T | T | A | C | TT T A C |
| - NA18524_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA18635_c1 | C | T | T | A | T | CT T A T |
| - NA18635_c2 | C | T | T | A | T | CT T A T |
| - NA18537_c1 | C | G | C | C | T | CG C C T |
| - NA18537_c2 | C | G | T | A | T | CG T A T |
| - NA18572_c1 | C | G | C | A | C | CG C A C |
| - NA18572_c2 | C | T | T | A | T | CT T A T |
| - NA18592_c1 | C | T | T | A | T | CT T A T |
| - NA18592_c2 | C | G | T | A | T | CG T A T |
| - NA18526_c1 | T | T | T | A | C | TT T A C |
| - NA18526_c2 | C | T | T | A | T | CT T A T |
| - NA18529_c1 | C | T | T | A | T | CT T A T |
| - NA18529_c2 | C | T | T | A | T | CT T A T |
| - NA18558_c1 | C | T | T | A | T | CT T A T |
| - NA18558_c2 | C | T | T | A | T | CT T A T |
| - NA18562_c1 | C | T | T | A | C | CT T A C |
| - NA18562_c2 | C | G | C | A | C | CG C A C |
| - NA18545_c1 | C | T | T | A | T | CT T A T |
| - NA18545_c2 | C | G | C | A | C | CG C A C |
| - NA18609_c1 | C | T | T | A | T | CT T A T |
| - NA18609_c2 | C | G | C | A | C | CG C A C |
| - NA18552_c1 | C | T | T | A | T | CT T A T |
| - NA18552_c2 | C | G | C | A | C | CG C A C |
| - NA18611_c1 | C | T | T | A | T | CT T A T |
| - NA18611_c2 | C | G | C | A | C | CG C A C |
| - NA18555_c1 | T | T | T | A | T | TT T A T |
| - NA18555_c2 | C | T | T | A | T | CT T A T |
| - NA18566_c1 | C | G | C | C | T | CG C C T |
| - NA18566_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18563_c1 | C | G | C | C | C | CG C C C |
| - NA18563_c2 | T | T | C | A | T | TT C A T |
| - NA18570_c1 | C | T | T | A | T | CT T A T |
| - NA18570_c2 | C | G | C | A | C | CG C A C |
| - NA18612_c1 | C | T | T | A | T | CT T A T |
| - NA18612_c2 | T | T | C | A | C | TT C A C |
| - NA18621_c1 | C | T | T | A | T | CT T A T |
| - NA18621_c2 | C | G | C | A | C | CG C A C |
| - NA18622_c1 | C | T | T | A | T | CT T A T |
| - NA18622_c2 | T | T | T | A | C | TT T A C |
| - NA18573_c1 | C | G | C | A | C | CG C A C |
| - NA18573_c2 | C | T | T | A | T | CT T A T |
| - NA18577_c1 | C | T | T | A | T | CT T A T |
| - NA18577_c2 | C | T | T | A | T | CT T A T |
| - NA18579_c1 | T | T | T | A | T | TT T A T |
| - NA18579_c2 | C | T | T | A | T | CT T A T |
| - NA18632_c1 | C | T | T | A | T | CT T A T |
| - NA18632_c2 | C | T | T | A | T | CT T A T |
| - NA18636_c1 | C | T | T | A | T | CT T A T |
| - NA18636_c2 | C | G | C | A | C | CG C A C |
| - NA18593_c1 | C | G | C | A | C | CG C A C |
| - NA18593_c2 | C | T | T | A | T | CT T A T |
| - NA18603_c1 | C | T | T | A | C | CT T A C |
| - NA18603_c2 | C | T | T | A | T | CT T A T |
| - NA18624_c1 | C | G | C | A | C | CG C A C |
| - NA18624_c2 | T | T | T | A | T | TT T A T |
| - NA18550_c1 | C | T | T | A | T | CT T A T |
| - NA18550_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18605_c1 | C | T | T | A | C | CT T A C |
| - NA18605_c2 | C | T | T | A | T | CT T A T |
| - NA18542_c1 | C | G | C | C | T | CG C C T |
| - NA18542_c2 | C | T | T | A | C | CT T A C |
| - NA18532_c1 | C | T | T | A | T | CT T A T |
| - NA18532_c2 | C | G | C | A | C | CG C A C |
| - NA18561_c1 | C | T | T | A | T | CT T A T |
| - NA18561_c2 | C | T | T | A | C | CT T A C |
| - NA18608_c1 | T | T | T | A | C | TT T A C |
| - NA18608_c2 | C | T | T | A | T | CT T A T |
| - NA18564_c1 | C | G | C | A | C | CG C A C |
| - NA18564_c2 | C | T | T | A | T | CT T A T |
| - NA18571_c1 | C | G | C | A | C | CG C A C |
| - NA18571_c2 | C | T | T | A | T | CT T A T |
| - NA18620_c1 | C | T | T | A | C | CT T A C |
| - NA18620_c2 | T | T | C | A | T | TT C A T |
| - NA18623_c1 | C | T | T | A | C | CT T A C |
| - NA18623_c2 | C | G | C | C | T | CG C C T |
| - NA18576_c1 | C | T | T | A | C | CT T A C |
| - NA18576_c2 | T | T | T | A | C | TT T A C |
| - NA18582_c1 | C | G | C | A | C | CG C A C |
| - NA18582_c2 | C | T | T | A | T | CT T A T |
| - NA18633_c1 | T | T | T | A | C | TT T A C |
| - NA18633_c2 | T | T | T | A | C | TT T A C |
| - NA18637_c1 | C | T | T | A | T | CT T A T |
| - NA18637_c2 | C | G | C | A | C | CG C A C |
| - NA18594_c1 | C | T | T | A | T | CT T A T |
| - NA18594_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA17970_c1 | T | T | C | A | C | TT C A C |
| - NA17970_c2 | C | T | T | A | T | CT T A T |
| - NA17977_c1 | C | G | C | C | C | CG C C C |
| - NA17977_c2 | C | G | C | A | C | CG C A C |
| - NA17981_c1 | C | G | C | C | C | CG C C C |
| - NA17981_c2 | T | T | T | A | C | TT T A C |
| - NA17993_c1 | C | G | C | A | T | CG C A T |
| - NA17993_c2 | C | T | T | A | C | CT T A C |
| - NA18101_c1 | T | T | T | A | T | TT T A T |
| - NA18101_c2 | C | G | C | A | C | CG C A C |
| - NA18105_c1 | T | T | T | A | C | TT T A C |
| - NA18105_c2 | C | T | T | A | T | CT T A T |
| - NA18109_c1 | C | G | C | A | C | CG C A C |
| - NA18109_c2 | T | T | T | A | T | TT T A T |
| - NA18129_c1 | T | T | T | A | T | TT T A T |
| - NA18129_c2 | C | G | C | A | C | CG C A C |
| - NA18135_c1 | C | T | T | A | T | CT T A T |
| - NA18135_c2 | C | T | T | A | T | CT T A T |
| - NA18139_c1 | C | T | T | A | T | CT T A T |
| - NA18139_c2 | T | T | T | A | C | TT T A C |
| - NA18144_c1 | C | T | T | A | T | CT T A T |
| - NA18144_c2 | C | G | C | A | C | CG C A C |
| - NA18150_c1 | C | G | C | A | C | CG C A C |
| - NA18150_c2 | C | T | T | A | T | CT T A T |
| - NA18154_c1 | T | T | T | A | C | TT T A C |
| - NA18154_c2 | C | G | C | A | T | CG C A T |
| - NA18162_c1 | C | T | T | A | T | CT T A T |
| - NA18162_c2 | C | G | C | A | T | CG C A T |

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|---|---|---|---|---|---|---|
| - NA17974_c1 | C | G | C | A | C | CG C A C |
| - NA17974_c2 | C | G | C | A | C | CG C A C |
| - NA17975_c1 | C | T | T | A | T | CT T A T |
| - NA17975_c2 | C | T | T | A | T | CT T A T |
| - NA17980_c1 | T | T | T | A | T | TT T A T |
| - NA17980_c2 | C | G | C | A | C | CG C A C |
| - NA17986_c1 | C | T | T | A | T | CT T A T |
| - NA17986_c2 | T | T | T | A | C | TT T A C |
| - NA17989_c1 | C | G | C | A | C | CG C A C |
| - NA17989_c2 | C | T | T | A | T | CT T A T |
| - NA17997_c1 | C | T | T | A | T | CT T A T |
| - NA17997_c2 | C | T | T | A | T | CT T A T |
| - NA18106_c1 | C | G | C | A | C | CG C A C |
| - NA18106_c2 | C | G | C | A | C | CG C A C |
| - NA18114_c1 | C | G | C | C | C | CG C C C |
| - NA18114_c2 | C | T | T | A | T | CT T A T |
| - NA18122_c1 | C | T | T | A | C | CT T A C |
| - NA18122_c2 | T | T | T | A | T | TT T A T |
| - NA18138_c1 | T | T | T | A | T | TT T A T |
| - NA18138_c2 | C | G | C | A | C | CG C A C |
| - NA18149_c1 | C | G | C | A | C | CG C A C |
| - NA18149_c2 | C | T | T | A | T | CT T A T |
| - NA17987_c1 | C | G | C | A | C | CG C A C |
| - NA17987_c2 | C | T | T | A | T | CT T A T |
| - NA17988_c1 | C | G | C | A | T | CG C A T |
| - NA17988_c2 | C | G | C | A | C | CG C A C |
| - NA17998_c1 | C | G | C | A | T | CG C A T |
| - NA17998_c2 | C | T | C | A | C | CT C A C |

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|---|---|---|---|---|---|---|
| - NA18107_c1 | T | T | C | A | C | TT C A C |
| - NA18107_c2 | C | G | C | A | C | CG C A C |
| - NA18108_c1 | C | G | C | A | C | CG C A C |
| - NA18108_c2 | C | T | T | A | T | CT T A T |
| - NA18128_c1 | T | T | C | A | C | TT C A C |
| - NA18128_c2 | T | T | T | A | C | TT T A C |
| - NA18146_c1 | C | G | C | A | C | CG C A C |
| - NA18146_c2 | T | T | T | A | T | TT T A T |
| - NA18148_c1 | C | T | T | A | T | CT T A T |
| - NA18148_c2 | C | G | C | A | C | CG C A C |
| - NA18161_c1 | T | T | T | A | C | TT T A C |
| - NA18161_c2 | C | T | T | A | T | CT T A T |
| - NA18670_c1 | C | T | T | A | T | CT T A T |
| - NA18670_c2 | C | T | T | A | C | CT T A C |
| - NA18694_c1 | T | T | T | A | T | TT T A T |
| - NA18694_c2 | C | T | T | A | T | CT T A T |
| - NA18696_c1 | C | T | T | A | T | CT T A T |
| - NA18696_c2 | C | T | T | A | T | CT T A T |
| - NA17965_c1 | T | T | T | A | C | TT T A C |
| - NA17965_c2 | C | T | T | A | T | CT T A T |
| - NA17967_c1 | C | T | T | A | T | CT T A T |
| - NA17967_c2 | C | G | C | A | C | CG C A C |
| - NA17969_c1 | C | T | T | A | T | CT T A T |
| - NA17969_c2 | C | T | T | A | C | CT T A C |
| - NA17983_c1 | C | G | C | C | C | CG C C C |
| - NA17983_c2 | T | T | T | A | C | TT T A C |
| - NA18117_c1 | C | G | C | A | C | CG C A C |
| - NA18117_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18120_c1 | C | T | T | A | T | CT T A T |
| - NA18120_c2 | C | G | C | C | T | CG C C T |
| - NA18124_c1 | C | T | T | A | T | CT T A T |
| - NA18124_c2 | C | G | C | A | T | CG C A T |
| - NA18125_c1 | C | T | T | A | T | CT T A T |
| - NA18125_c2 | T | T | T | A | C | TT T A C |
| - NA18127_c1 | C | T | T | A | T | CT T A T |
| - NA18127_c2 | T | T | T | A | T | TT T A T |
| - NA18132_c1 | C | T | T | A | T | CT T A T |
| - NA18132_c2 | C | T | T | A | T | CT T A T |
| - NA18141_c1 | C | T | T | A | T | CT T A T |
| - NA18141_c2 | C | G | C | A | C | CG C A C |
| - NA18143_c1 | C | T | T | A | T | CT T A T |
| - NA18143_c2 | C | T | T | A | T | CT T A T |
| - NA18147_c1 | T | T | C | A | T | TT C A T |
| - NA18147_c2 | T | T | T | A | C | TT T A C |
| - NA18155_c1 | T | T | T | A | T | TT T A T |
| - NA18155_c2 | C | T | T | A | T | CT T A T |
| - NA18158_c1 | T | T | C | A | C | TT C A C |
| - NA18158_c2 | C | T | T | A | T | CT T A T |
| - NA18674_c1 | C | T | T | A | T | CT T A T |
| - NA18674_c2 | C | T | T | A | T | CT T A T |
| - NA17982_c1 | C | T | T | A | C | CT T A C |
| - NA17982_c2 | C | T | T | A | T | CT T A T |
| - NA17990_c1 | C | G | C | A | C | CG C A C |
| - NA17990_c2 | C | T | T | A | T | CT T A T |
| - NA18112_c1 | T | T | T | A | T | TT T A T |
| - NA18112_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA18118_c1 | T | T | C | A | C | TT C A C |
| - NA18118_c2 | C | G | C | A | C | CG C A C |
| - NA18151_c1 | C | T | T | A | T | CT T A T |
| - NA18151_c2 | C | G | C | C | C | CG C C C |
| - NA18153_c1 | C | T | T | A | T | CT T A T |
| - NA18153_c2 | C | G | C | A | C | CG C A C |
| - NA18702_c1 | C | T | T | A | T | CT T A T |
| - NA18702_c2 | T | T | T | A | T | TT T A T |
| - NA18704_c1 | T | T | C | A | C | TT C A C |
| - NA18704_c2 | C | T | T | A | T | CT T A T |
| - NA17976_c1 | T | T | T | A | C | TT T A C |
| - NA17976_c2 | T | T | T | A | T | TT T A T |
| - NA17979_c1 | C | G | C | A | C | CG C A C |
| - NA17979_c2 | C | G | C | A | C | CG C A C |
| - NA18156_c1 | C | T | T | A | C | CT T A C |
| - NA18156_c2 | T | T | T | A | T | TT T A T |
| - NA18152_c1 | C | G | C | A | C | CG C A C |
| - NA18152_c2 | C | T | T | A | C | CT T A C |
| - NA18160_c1 | C | G | C | A | C | CG C A C |
| - NA18160_c2 | C | T | T | A | T | CT T A T |
| - NA18682_c1 | C | G | C | C | C | CG C C C |
| - NA18682_c2 | C | T | T | A | T | CT T A T |
| - NA18689_c1 | T | T | T | A | T | TT T A T |
| - NA18689_c2 | C | G | C | C | C | CG C C C |
| - NA18685_c1 | C | G | C | A | C | CG C A C |
| - NA18685_c2 | C | G | C | A | C | CG C A C |
| - NA17962_c1 | C | G | C | C | T | CG C C T |
| - NA17962_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA17966_c1 | C | T | T | A | T | CT T A T |
| - NA17966_c2 | C | T | T | A | T | CT T A T |
| - NA17999_c1 | C | G | C | A | T | CG C A T |
| - NA17999_c2 | C | G | C | A | C | CG C A C |
| - NA17968_c1 | C | T | T | A | T | CT T A T |
| - NA17968_c2 | C | G | C | A | C | CG C A C |
| - NA17995_c1 | C | G | C | A | C | CG C A C |
| - NA17995_c2 | C | G | C | A | C | CG C A C |
| - NA17996_c1 | C | T | T | A | T | CT T A T |
| - NA17996_c2 | C | G | C | A | C | CG C A C |
| - NA18131_c1 | C | T | T | A | T | CT T A T |
| - NA18131_c2 | C | T | T | A | T | CT T A T |
| - NA18134_c1 | C | T | T | A | T | CT T A T |
| - NA18134_c2 | C | G | C | C | T | CG C C T |
| - NA18157_c1 | T | T | T | A | T | TT T A T |
| - NA18157_c2 | C | T | T | A | C | CT T A C |
| - NA18159_c1 | C | T | T | A | T | CT T A T |
| - NA18159_c2 | C | T | T | A | T | CT T A T |
| - NA17972_c1 | C | T | T | A | T | CT T A T |
| - NA17972_c2 | C | T | T | A | T | CT T A T |
| - NA18133_c1 | C | T | T | C | C | CT T C C |
| - NA18133_c2 | T | T | T | A | T | TT T A T |
| - NA18166_c1 | C | G | C | A | C | CG C A C |
| - NA18166_c2 | C | G | C | A | C | CG C A C |
| - NA18102_c1 | C | G | C | A | C | CG C A C |
| - NA18102_c2 | T | T | T | A | C | TT T A C |
| - NA18136_c1 | C | T | T | A | T | CT T A T |
| - NA18136_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18140_c1 | C | G | C | A | C | CG C A C |
| - NA18140_c2 | C | G | C | C | C | CG C C C |
| - NA20847_c1 | C | G | C | C | C | CG C C C |
| - NA20847_c2 | C | T | T | A | T | CT T A T |
| - NA20849_c1 | C | T | T | A | T | CT T A T |
| - NA20849_c2 | C | G | C | C | C | CG C C C |
| - NA20851_c1 | C | T | T | A | T | CT T A T |
| - NA20851_c2 | C | G | C | A | C | CG C A C |
| - NA20853_c1 | C | G | C | C | T | CG C C T |
| - NA20853_c2 | T | T | T | A | T | TT T A T |
| - NA20906_c1 | C | G | C | A | C | CG C A C |
| - NA20906_c2 | T | T | T | A | T | TT T A T |
| - NA20866_c1 | C | G | C | A | C | CG C A C |
| - NA20866_c2 | C | T | T | A | T | CT T A T |
| - NA21104_c1 | C | T | T | A | T | CT T A T |
| - NA21104_c2 | T | T | T | A | T | TT T A T |
| - NA20907_c1 | C | G | C | A | T | CG C A T |
| - NA20907_c2 | C | G | C | A | C | CG C A C |
| - NA20908_c1 | C | G | C | C | C | CG C C C |
| - NA20908_c2 | C | T | T | A | T | CT T A T |
| - NA21086_c1 | T | T | T | A | T | TT T A T |
| - NA21086_c2 | C | T | T | A | T | CT T A T |
| - NA21101_c1 | C | T | T | A | T | CT T A T |
| - NA21101_c2 | C | G | C | A | C | CG C A C |
| - NA21106_c1 | C | T | T | A | T | CT T A T |
| - NA21106_c2 | C | T | T | A | T | CT T A T |
| - NA21125_c1 | C | T | T | A | T | CT T A T |
| - NA21125_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21137_c1 | C | G | C | C | C | CG C C C |
| - NA21137_c2 | C | T | T | A | T | CT T A T |
| - NA21141_c1 | C | G | C | A | C | CG C A C |
| - NA21141_c2 | C | T | T | A | T | CT T A T |
| - NA21142_c1 | C | T | T | A | T | CT T A T |
| - NA21142_c2 | C | G | C | A | C | CG C A C |
| - NA20883_c1 | C | G | C | C | C | CG C C C |
| - NA20883_c2 | T | T | T | A | T | TT T A T |
| - NA20891_c1 | C | G | C | A | T | CG C A T |
| - NA20891_c2 | C | G | C | A | T | CG C A T |
| - NA20901_c1 | T | T | T | A | T | TT T A T |
| - NA20901_c2 | C | G | C | A | C | CG C A C |
| - NA21090_c1 | C | T | T | A | T | CT T A T |
| - NA21090_c2 | C | T | T | A | T | CT T A T |
| - NA21092_c1 | T | T | T | A | T | TT T A T |
| - NA21092_c2 | C | T | T | A | T | CT T A T |
| - NA21094_c1 | C | T | T | A | T | CT T A T |
| - NA21094_c2 | C | T | T | A | T | CT T A T |
| - NA21098_c1 | T | T | T | A | T | TT T A T |
| - NA21098_c2 | C | G | C | C | C | CG C C C |
| - NA21100_c1 | C | G | C | A | C | CG C A C |
| - NA21100_c2 | C | G | C | A | C | CG C A C |
| - NA21105_c1 | C | G | C | C | C | CG C C C |
| - NA21105_c2 | C | G | C | A | C | CG C A C |
| - NA21107_c1 | T | T | T | A | T | TT T A T |
| - NA21107_c2 | C | T | T | A | T | CT T A T |
| - NA21109_c1 | C | G | C | A | C | CG C A C |
| - NA21109_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA20888_c1 | C | T | T | A | T | CT T A T |
| - NA20888_c2 | C | T | T | A | T | CT T A T |
| - NA20892_c1 | C | G | C | A | C | CG C A C |
| - NA20892_c2 | T | T | T | A | T | TT T A T |
| - NA20900_c1 | C | G | C | A | C | CG C A C |
| - NA20900_c2 | C | G | C | A | T | CG C A T |
| - NA20910_c1 | C | G | C | A | C | CG C A C |
| - NA20910_c2 | C | G | C | A | C | CG C A C |
| - NA21088_c1 | C | T | T | A | T | CT T A T |
| - NA21088_c2 | C | G | C | C | C | CG C C C |
| - NA21089_c1 | C | G | C | A | C | CG C A C |
| - NA21089_c2 | C | G | C | A | C | CG C A C |
| - NA21102_c1 | C | G | C | C | C | CG C C C |
| - NA21102_c2 | T | T | T | A | T | TT T A T |
| - NA20845_c1 | C | T | T | A | T | CT T A T |
| - NA20845_c2 | C | G | C | A | C | CG C A C |
| - NA20850_c1 | C | T | T | A | T | CT T A T |
| - NA20850_c2 | C | G | C | A | T | CG C A T |
| - NA20852_c1 | C | G | C | C | C | CG C C C |
| - NA20852_c2 | C | G | C | A | C | CG C A C |
| - NA20858_c1 | C | G | C | C | C | CG C C C |
| - NA20858_c2 | C | T | T | A | T | CT T A T |
| - NA20870_c1 | C | T | T | A | C | CT T A C |
| - NA20870_c2 | C | G | C | A | C | CG C A C |
| - NA20889_c1 | C | G | C | A | C | CG C A C |
| - NA20889_c2 | C | T | T | A | C | CT T A C |
| - NA20897_c1 | C | T | T | A | T | CT T A T |
| - NA20897_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA20904_c1 | C | T | T | A | T | CT T A T |
| - NA20904_c2 | T | T | T | A | T | TT T A T |
| - NA20911_c1 | C | G | C | A | C | CG C A C |
| - NA20911_c2 | T | T | T | A | T | TT T A T |
| - NA21099_c1 | C | G | C | A | C | CG C A C |
| - NA21099_c2 | C | G | C | C | C | CG C C C |
| - NA21112_c1 | C | G | C | A | C | CG C A C |
| - NA21112_c2 | C | T | T | A | T | CT T A T |
| - NA21116_c1 | C | G | C | A | T | CG C A T |
| - NA21116_c2 | C | T | T | A | C | CT T A C |
| - NA20862_c1 | C | G | C | C | C | CG C C C |
| - NA20862_c2 | C | G | C | C | C | CG C C C |
| - NA21143_c1 | C | T | T | A | T | CT T A T |
| - NA21143_c2 | C | G | C | A | C | CG C A C |
| - NA21144_c1 | C | G | C | C | C | CG C C C |
| - NA21144_c2 | T | T | T | A | T | TT T A T |
| - NA20871_c1 | C | T | T | A | T | CT T A T |
| - NA20871_c2 | C | G | C | A | C | CG C A C |
| - NA20887_c1 | T | T | C | A | T | TT C A T |
| - NA20887_c2 | C | G | C | A | C | CG C A C |
| - NA20890_c1 | C | G | C | A | C | CG C A C |
| - NA20890_c2 | T | T | T | A | T | TT T A T |
| - NA20898_c1 | C | T | T | A | T | CT T A T |
| - NA20898_c2 | C | G | C | A | C | CG C A C |
| - NA20903_c1 | C | T | T | A | T | CT T A T |
| - NA20903_c2 | C | G | C | A | T | CG C A T |
| - NA20909_c1 | C | G | C | A | C | CG C A C |
| - NA20909_c2 | C | G | C | C | C | CG C C C |

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|---|---|---|---|---|---|---|
| - NA21113_c1 | C | T | T | A | T | CT T A T |
| - NA21113_c2 | C | G | C | A | C | CG C A C |
| - NA21115_c1 | C | T | T | A | T | CT T A T |
| - NA21115_c2 | C | T | T | A | T | CT T A T |
| - NA21118_c1 | T | T | T | A | T | TT T A T |
| - NA21118_c2 | C | G | C | A | C | CG C A C |
| - NA21119_c1 | C | T | T | A | T | CT T A T |
| - NA21119_c2 | C | T | T | A | T | CT T A T |
| - NA21123_c1 | T | T | T | A | T | TT T A T |
| - NA21123_c2 | T | T | T | A | T | TT T A T |
| - NA21097_c1 | C | G | C | C | C | CG C C C |
| - NA21097_c2 | T | T | T | A | T | TT T A T |
| - NA20882_c1 | T | T | T | A | T | TT T A T |
| - NA20882_c2 | C | G | C | A | C | CG C A C |
| - NA20894_c1 | C | T | T | A | T | CT T A T |
| - NA20894_c2 | T | T | T | A | T | TT T A T |
| - NA20896_c1 | C | T | T | A | T | CT T A T |
| - NA20896_c2 | T | T | T | A | T | TT T A T |
| - NA20902_c1 | C | T | T | A | T | CT T A T |
| - NA20902_c2 | C | T | T | A | T | CT T A T |
| - NA20895_c1 | C | G | C | A | T | CG C A T |
| - NA20895_c2 | C | G | C | C | T | CG C C T |
| - NA20856_c1 | C | T | T | A | T | CT T A T |
| - NA20856_c2 | T | T | T | A | T | TT T A T |
| - NA20869_c1 | C | G | C | C | C | CG C C C |
| - NA20869_c2 | C | G | C | A | C | CG C A C |
| - NA20872_c1 | C | G | C | A | C | CG C A C |
| - NA20872_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA20874_c1 | C | T | T | A | T | CT T A T |
| - NA20874_c2 | C | T | T | A | T | CT T A T |
| - NA20854_c1 | C | G | C | A | C | CG C A C |
| - NA20854_c2 | C | G | C | C | C | CG C C C |
| - NA20879_c1 | C | T | T | A | T | CT T A T |
| - NA20879_c2 | T | T | T | A | T | TT T A T |
| - NA21111_c1 | T | T | T | A | T | TT T A T |
| - NA21111_c2 | C | G | C | A | C | CG C A C |
| - NA20846_c1 | C | T | T | A | T | CT T A T |
| - NA20846_c2 | C | G | C | C | T | CG C C T |
| - NA20899_c1 | T | T | T | A | T | TT T A T |
| - NA20899_c2 | C | T | T | A | T | CT T A T |
| - NA21108_c1 | C | T | T | A | T | CT T A T |
| - NA21108_c2 | C | G | C | C | C | CG C C C |
| - NA20861_c1 | C | T | T | A | C | CT T A C |
| - NA20861_c2 | C | G | C | A | C | CG C A C |
| - NA20873_c1 | T | T | C | A | C | TT C A C |
| - NA20873_c2 | C | G | C | A | C | CG C A C |
| - NA20884_c1 | T | T | T | A | T | TT T A T |
| - NA20884_c2 | C | T | T | A | T | CT T A T |
| - NA21091_c1 | C | G | C | A | C | CG C A C |
| - NA21091_c2 | T | T | T | A | T | TT T A T |
| - NA20875_c1 | T | T | T | A | T | TT T A T |
| - NA20875_c2 | T | T | T | A | T | TT T A T |
| - NA20876_c1 | C | G | C | C | C | CG C C C |
| - NA20876_c2 | C | T | T | A | T | CT T A T |
| - NA20877_c1 | C | G | C | A | T | CG C A T |
| - NA20877_c2 | C | T | T | A | C | CT T A C |

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|---|---|---|---|---|---|---|
| - NA21103_c1 | C | T | T | A | T | CT T A T |
| - NA21103_c2 | C | G | C | A | C | CG C A C |
| - NA20885_c1 | T | T | T | A | T | TT T A T |
| - NA20885_c2 | C | T | T | A | T | CT T A T |
| - NA21117_c1 | C | G | C | C | T | CG C C T |
| - NA21117_c2 | C | G | C | A | C | CG C A C |
| - NA20859_c1 | T | T | T | A | T | TT T A T |
| - NA20859_c2 | C | T | T | A | T | CT T A T |
| - NA20881_c1 | C | T | T | A | T | CT T A T |
| - NA20881_c2 | T | T | T | A | T | TT T A T |
| - NA18946_c1 | C | G | C | A | C | CG C A C |
| - NA18946_c2 | C | T | T | A | T | CT T A T |
| - NA18979_c1 | C | G | C | A | C | CG C A C |
| - NA18979_c2 | C | T | T | A | T | CT T A T |
| - NA19058_c1 | T | T | T | A | T | TT T A T |
| - NA19058_c2 | C | G | C | A | C | CG C A C |
| - NA18993_c1 | C | T | T | A | T | CT T A T |
| - NA18993_c2 | C | T | T | A | T | CT T A T |
| - NA19060_c1 | C | T | T | A | T | CT T A T |
| - NA19060_c2 | C | G | C | C | C | CG C C C |
| - NA19062_c1 | C | G | C | C | T | CG C C T |
| - NA19062_c2 | C | T | T | A | T | CT T A T |
| - NA18955_c1 | C | G | C | A | C | CG C A C |
| - NA18955_c2 | C | T | T | A | T | CT T A T |
| - NA18962_c1 | C | G | C | A | C | CG C A C |
| - NA18962_c2 | T | T | T | A | T | TT T A T |
| - NA18977_c1 | C | T | T | A | T | CT T A T |
| - NA18977_c2 | T | T | T | A | C | TT T A C |

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|---|---|---|---|---|---|---|
| - NA18954_c1 | C | T | T | A | T | CT T A T |
| - NA18954_c2 | C | T | T | A | T | CT T A T |
| - NA19066_c1 | T | T | T | A | C | TT T A C |
| - NA19066_c2 | T | T | T | A | T | TT T A T |
| - NA19010_c1 | C | T | T | A | T | CT T A T |
| - NA19010_c2 | C | G | C | A | C | CG C A C |
| - NA19054_c1 | C | T | T | A | C | CT T A C |
| - NA19054_c2 | C | G | C | A | C | CG C A C |
| - NA19064_c1 | T | T | T | A | T | TT T A T |
| - NA19064_c2 | C | G | C | A | C | CG C A C |
| - NA19072_c1 | C | T | T | A | T | CT T A T |
| - NA19072_c2 | T | T | T | A | T | TT T A T |
| - NA19075_c1 | C | T | T | A | T | CT T A T |
| - NA19075_c2 | C | G | C | A | C | CG C A C |
| - NA19065_c1 | C | G | C | A | C | CG C A C |
| - NA19065_c2 | T | T | T | A | T | TT T A T |
| - NA19074_c1 | C | G | C | A | C | CG C A C |
| - NA19074_c2 | C | T | T | A | T | CT T A T |
| - NA19076_c1 | T | T | T | A | T | TT T A T |
| - NA19076_c2 | C | G | C | A | C | CG C A C |
| - NA19063_c1 | C | G | C | C | C | CG C C C |
| - NA19063_c2 | T | T | T | A | C | TT T A C |
| - NA19002_c1 | C | T | T | A | T | CT T A T |
| - NA19002_c2 | C | G | C | C | T | CG C C T |
| - NA19067_c1 | C | T | T | A | T | CT T A T |
| - NA19067_c2 | C | G | C | C | T | CG C C T |
| - NA19057_c1 | C | G | C | A | C | CG C A C |
| - NA19057_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19068_c1 | C | T | T | A | T | CT T A T |
| - NA19068_c2 | C | G | C | A | C | CG C A C |
| - NA19059_c1 | C | T | T | A | T | CT T A T |
| - NA19059_c2 | C | T | T | A | T | CT T A T |
| - NA19070_c1 | T | T | C | A | C | TT C A C |
| - NA19070_c2 | C | G | C | A | C | CG C A C |
| - NA19077_c1 | C | T | T | A | T | CT T A T |
| - NA19077_c2 | C | T | T | A | C | CT T A C |
| - NA19078_c1 | C | T | C | A | C | CT C A C |
| - NA19078_c2 | C | T | T | A | T | CT T A T |
| - NA19079_c1 | C | G | C | A | C | CG C A C |
| - NA19079_c2 | C | T | T | A | T | CT T A T |
| - NA19080_c1 | C | G | C | A | C | CG C A C |
| - NA19080_c2 | C | T | T | A | T | CT T A T |
| - NA19081_c1 | T | T | T | A | T | TT T A T |
| - NA19081_c2 | T | T | T | A | T | TT T A T |
| - NA19083_c1 | C | G | C | A | C | CG C A C |
| - NA19083_c2 | C | T | T | A | T | CT T A T |
| - NA19085_c1 | T | T | T | A | T | TT T A T |
| - NA19085_c2 | C | G | C | C | C | CG C C C |
| - NA19084_c1 | T | T | T | A | T | TT T A T |
| - NA19084_c2 | T | T | T | A | C | TT T A C |
| - NA19086_c1 | C | T | T | A | T | CT T A T |
| - NA19086_c2 | T | T | T | A | T | TT T A T |
| - NA18939_c1 | C | G | C | A | C | CG C A C |
| - NA18939_c2 | C | T | T | A | T | CT T A T |
| - NA19088_c1 | C | G | C | A | C | CG C A C |
| - NA19088_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA19087_c1 | T | T | T | A | T | TT T A T |
| - NA19087_c2 | C | G | C | C | C | CG C C C |
| - NA19055_c1 | C | T | T | A | T | CT T A T |
| - NA19055_c2 | C | G | C | A | C | CG C A C |
| - NA18957_c1 | C | T | T | A | T | CT T A T |
| - NA18957_c2 | C | G | C | C | T | CG C C T |
| - NA18963_c1 | C | G | C | C | T | CG C C T |
| - NA18963_c2 | C | G | C | A | C | CG C A C |
| - NA19001_c1 | C | G | C | C | T | CG C C T |
| - NA19001_c2 | C | G | C | C | C | CG C C C |
| - NA19009_c1 | T | T | T | A | T | TT T A T |
| - NA19009_c2 | C | T | T | A | T | CT T A T |
| - NA19056_c1 | C | T | T | A | T | CT T A T |
| - NA19056_c2 | C | T | T | A | T | CT T A T |
| - NA18942_c1 | C | T | T | A | T | CT T A T |
| - NA18942_c2 | C | G | C | A | C | CG C A C |
| - NA18949_c1 | C | T | T | A | T | CT T A T |
| - NA18949_c2 | C | G | C | A | C | CG C A C |
| - NA18970_c1 | C | G | C | A | C | CG C A C |
| - NA18970_c2 | C | G | C | C | C | CG C C C |
| - NA18945_c1 | T | T | T | A | C | TT T A C |
| - NA18945_c2 | T | T | T | A | T | TT T A T |
| - NA18940_c1 | C | G | C | A | C | CG C A C |
| - NA18940_c2 | T | T | C | A | C | TT C A C |
| - NA18964_c1 | C | G | C | A | C | CG C A C |
| - NA18964_c2 | C | G | C | A | C | CG C A C |
| - NA18953_c1 | C | T | T | A | T | CT T A T |
| - NA18953_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA18961_c1 | C | T | T | A | C | CT T A C |
| - NA18961_c2 | C | T | T | A | T | CT T A T |
| - NA18972_c1 | T | T | T | A | T | TT T A T |
| - NA18972_c2 | C | T | T | A | T | CT T A T |
| - NA18967_c1 | C | T | T | A | T | CT T A T |
| - NA18967_c2 | T | T | T | A | T | TT T A T |
| - NA18976_c1 | T | T | T | A | T | TT T A T |
| - NA18976_c2 | C | G | C | C | C | CG C C C |
| - NA18981_c1 | C | G | C | A | C | CG C A C |
| - NA18981_c2 | C | T | T | A | T | CT T A T |
| - NA18971_c1 | T | T | T | A | T | TT T A T |
| - NA18971_c2 | C | T | T | A | T | CT T A T |
| - NA18994_c1 | C | T | C | A | T | CT C A T |
| - NA18994_c2 | C | T | T | A | T | CT T A T |
| - NA18998_c1 | C | G | C | A | C | CG C A C |
| - NA18998_c2 | C | T | T | A | T | CT T A T |
| - NA19000_c1 | T | T | T | A | T | TT T A T |
| - NA19000_c2 | C | T | T | A | T | CT T A T |
| - NA18943_c1 | C | G | C | A | C | CG C A C |
| - NA18943_c2 | C | T | T | A | T | CT T A T |
| - NA18947_c1 | C | G | C | A | C | CG C A C |
| - NA18947_c2 | C | T | T | A | T | CT T A T |
| - NA18944_c1 | C | G | C | A | C | CG C A C |
| - NA18944_c2 | C | G | C | A | C | CG C A C |
| - NA18948_c1 | C | T | T | A | T | CT T A T |
| - NA18948_c2 | C | T | T | A | T | CT T A T |
| - NA18951_c1 | C | T | T | A | T | CT T A T |
| - NA18951_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA18952_c1 | C | T | T | A | T | CT T A T |
| - NA18952_c2 | T | T | T | A | C | TT T A C |
| - NA18956_c1 | C | T | T | A | T | CT T A T |
| - NA18956_c2 | C | T | T | A | T | CT T A T |
| - NA18968_c1 | C | G | C | C | T | CG C C T |
| - NA18968_c2 | T | T | T | A | T | TT T A T |
| - NA18959_c1 | C | T | T | A | T | CT T A T |
| - NA18959_c2 | C | T | T | A | T | CT T A T |
| - NA18969_c1 | C | T | T | A | T | CT T A T |
| - NA18969_c2 | C | T | T | A | T | CT T A T |
| - NA18960_c1 | C | G | C | C | T | CG C C T |
| - NA18960_c2 | C | T | T | A | T | CT T A T |
| - NA18965_c1 | C | G | C | A | C | CG C A C |
| - NA18965_c2 | C | T | T | A | T | CT T A T |
| - NA18973_c1 | T | T | T | A | C | TT T A C |
| - NA18973_c2 | C | T | T | A | T | CT T A T |
| - NA18966_c1 | C | G | C | A | C | CG C A C |
| - NA18966_c2 | T | T | T | A | T | TT T A T |
| - NA18975_c1 | C | T | T | A | T | CT T A T |
| - NA18975_c2 | C | G | C | C | T | CG C C T |
| - NA18978_c1 | T | T | T | A | C | TT T A C |
| - NA18978_c2 | C | G | C | A | C | CG C A C |
| - NA18980_c1 | C | T | T | A | T | CT T A T |
| - NA18980_c2 | C | G | C | A | T | CG C A T |
| - NA18974_c1 | C | G | C | A | C | CG C A C |
| - NA18974_c2 | C | T | T | A | T | CT T A T |
| - NA18987_c1 | C | G | C | C | C | CG C C C |
| - NA18987_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA18990_c1 | T | T | C | A | C | TT C A C |
| - NA18990_c2 | C | T | T | A | C | CT T A C |
| - NA18991_c1 | C | T | C | A | C | CT C A C |
| - NA18991_c2 | T | T | T | A | T | TT T A T |
| - NA18995_c1 | T | T | T | A | T | TT T A T |
| - NA18995_c2 | C | G | C | C | T | CG C C T |
| - NA18997_c1 | C | G | C | A | C | CG C A C |
| - NA18997_c2 | C | G | C | A | C | CG C A C |
| - NA19005_c1 | C | G | C | A | C | CG C A C |
| - NA19005_c2 | T | T | T | A | C | TT T A C |
| - NA18999_c1 | C | G | C | A | C | CG C A C |
| - NA18999_c2 | C | G | C | A | C | CG C A C |
| - NA19007_c1 | C | T | T | A | T | CT T A T |
| - NA19007_c2 | C | G | C | A | C | CG C A C |
| - NA19028_c1 | C | T | T | A | T | CT T A T |
| - NA19028_c2 | C | T | T | A | T | CT T A T |
| - NA19031_c1 | C | T | T | A | T | CT T A T |
| - NA19031_c2 | C | T | T | A | C | CT T A C |
| - NA19035_c1 | C | T | T | A | T | CT T A T |
| - NA19035_c2 | C | T | T | A | T | CT T A T |
| - NA19027_c1 | C | T | T | A | T | CT T A T |
| - NA19027_c2 | C | T | T | A | T | CT T A T |
| - NA19041_c1 | T | T | T | A | T | TT T A T |
| - NA19041_c2 | T | T | C | A | T | TT C A T |
| - NA19046_c1 | C | T | T | A | T | CT T A T |
| - NA19046_c2 | C | T | T | A | T | CT T A T |
| - NA19308_c1 | C | T | T | A | T | CT T A T |
| - NA19308_c2 | T | T | C | A | C | TT C A C |

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|---|---|---|---|---|---|---|
| - NA19311_c1 | C | T | T | A | T | CT T A T |
| - NA19311_c2 | C | T | T | A | C | CT T A C |
| - NA19317_c1 | C | T | T | A | T | CT T A T |
| - NA19317_c2 | T | T | C | A | T | TT C A T |
| - NA19376_c1 | C | T | T | A | T | CT T A T |
| - NA19376_c2 | C | T | T | A | T | CT T A T |
| - NA19380_c1 | T | G | T | A | T | TG T A T |
| - NA19380_c2 | T | T | T | A | T | TT T A T |
| - NA19383_c1 | T | T | C | A | C | TT C A C |
| - NA19383_c2 | T | T | C | A | C | TT C A C |
| - NA19393_c1 | C | T | T | A | C | CT T A C |
| - NA19393_c2 | C | G | C | A | C | CG C A C |
| - NA19397_c1 | C | T | T | A | T | CT T A T |
| - NA19397_c2 | C | T | T | A | T | CT T A T |
| - NA19430_c1 | C | T | T | A | T | CT T A T |
| - NA19430_c2 | C | G | C | A | C | CG C A C |
| - NA19466_c1 | C | T | T | A | T | CT T A T |
| - NA19466_c2 | C | T | C | A | T | CT C A T |
| - NA19038_c1 | C | T | T | A | T | CT T A T |
| - NA19038_c2 | C | T | T | A | C | CT T A C |
| - NA19314_c1 | T | T | C | A | T | TT C A T |
| - NA19314_c2 | T | G | T | A | T | TG T A T |
| - NA19315_c1 | C | T | T | A | T | CT T A T |
| - NA19315_c2 | T | G | T | A | T | TG T A T |
| - NA19324_c1 | C | G | C | A | C | CG C A C |
| - NA19324_c2 | C | T | T | A | T | CT T A T |
| - NA19328_c1 | C | T | T | A | T | CT T A T |
| - NA19328_c2 | C | T | C | A | T | CT C A T |

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|---|---|---|---|---|---|---|
| - NA19377_c1 | T | T | C | A | C | TT C A C |
| - NA19377_c2 | C | T | T | A | C | CT T A C |
| - NA19381_c1 | C | T | T | A | T | CT T A T |
| - NA19381_c2 | T | T | T | A | T | TT T A T |
| - NA19398_c1 | C | T | T | A | T | CT T A T |
| - NA19398_c2 | C | T | T | A | T | CT T A T |
| - NA19403_c1 | T | T | C | A | C | TT C A C |
| - NA19403_c2 | C | T | T | A | T | CT T A T |
| - NA19437_c1 | C | T | T | A | T | CT T A T |
| - NA19437_c2 | C | T | T | A | T | CT T A T |
| - NA19439_c1 | C | T | T | A | T | CT T A T |
| - NA19439_c2 | C | T | T | A | T | CT T A T |
| - NA19440_c1 | C | T | T | A | T | CT T A T |
| - NA19440_c2 | T | T | C | A | C | TT C A C |
| - NA19463_c1 | C | T | T | A | T | CT T A T |
| - NA19463_c2 | C | T | T | A | T | CT T A T |
| - NA19467_c1 | T | T | C | A | T | TT C A T |
| - NA19467_c2 | T | T | C | A | T | TT C A T |
| - NA19470_c1 | T | T | T | A | T | TT T A T |
| - NA19470_c2 | T | T | C | A | C | TT C A C |
| - NA19471_c1 | C | T | T | A | T | CT T A T |
| - NA19471_c2 | C | T | T | A | T | CT T A T |
| - NA19473_c1 | C | T | T | A | T | CT T A T |
| - NA19473_c2 | T | T | T | A | C | TT T A C |
| - NA19307_c1 | C | T | T | A | C | CT T A C |
| - NA19307_c2 | C | T | T | A | T | CT T A T |
| - NA19318_c1 | T | T | C | A | C | TT C A C |
| - NA19318_c2 | T | T | T | A | C | TT T A C |

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|---|---|---|---|---|---|---|
| - NA19319_c1 | T | T | C | A | C | TT C A C |
| - NA19319_c2 | C | T | T | A | T | CT T A T |
| - NA19334_c1 | C | T | T | A | T | CT T A T |
| - NA19334_c2 | C | T | T | A | C | CT T A C |
| - NA19350_c1 | T | T | C | A | C | TT C A C |
| - NA19350_c2 | C | T | T | A | T | CT T A T |
| - NA19352_c1 | C | T | T | A | T | CT T A T |
| - NA19352_c2 | C | T | T | A | T | CT T A T |
| - NA19359_c1 | T | T | C | A | C | TT C A C |
| - NA19359_c2 | C | T | T | A | T | CT T A T |
| - NA19375_c1 | C | T | T | A | T | CT T A T |
| - NA19375_c2 | C | T | T | A | T | CT T A T |
| - NA19428_c1 | C | T | T | A | T | CT T A T |
| - NA19428_c2 | C | T | T | A | T | CT T A T |
| - NA19429_c1 | C | T | T | A | T | CT T A T |
| - NA19429_c2 | T | T | C | A | C | TT C A C |
| - NA19443_c1 | T | T | C | A | T | TT C A T |
| - NA19443_c2 | T | T | T | A | C | TT T A C |
| - NA19455_c1 | T | T | T | A | T | TT T A T |
| - NA19455_c2 | C | T | T | A | T | CT T A T |
| - NA19313_c1 | C | T | T | A | T | CT T A T |
| - NA19313_c2 | T | T | C | A | T | TT C A T |
| - NA19316_c1 | C | T | T | A | C | CT T A C |
| - NA19316_c2 | C | T | T | A | C | CT T A C |
| - NA19321_c1 | T | T | T | A | T | TT T A T |
| - NA19321_c2 | C | T | T | A | T | CT T A T |
| - NA19332_c1 | C | T | T | A | T | CT T A T |
| - NA19332_c2 | T | T | C | A | C | TT C A C |

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|---|---|---|---|---|---|---|
| - NA19379_c1 | T | T | T | A | T | TT T A T |
| - NA19379_c2 | C | T | T | A | T | CT T A T |
| - NA19391_c1 | C | T | T | A | T | CT T A T |
| - NA19391_c2 | T | T | C | A | T | TT C A T |
| - NA19396_c1 | C | T | T | A | T | CT T A T |
| - NA19396_c2 | C | T | T | A | T | CT T A T |
| - NA19434_c1 | C | T | T | A | T | CT T A T |
| - NA19434_c2 | T | T | C | A | C | TT C A C |
| - NA19435_c1 | C | T | T | A | T | CT T A T |
| - NA19435_c2 | C | T | T | A | T | CT T A T |
| - NA19436_c1 | C | T | T | A | T | CT T A T |
| - NA19436_c2 | T | T | C | A | C | TT C A C |
| - NA19438_c1 | T | T | C | A | C | TT C A C |
| - NA19438_c2 | C | T | T | A | T | CT T A T |
| - NA19445_c1 | T | T | C | A | C | TT C A C |
| - NA19445_c2 | C | T | T | A | T | CT T A T |
| - NA19446_c1 | C | T | T | A | T | CT T A T |
| - NA19446_c2 | T | T | C | A | C | TT C A C |
| - NA19462_c1 | T | G | T | A | T | TG T A T |
| - NA19462_c2 | C | T | T | A | T | CT T A T |
| - NA19468_c1 | T | T | T | A | T | TT T A T |
| - NA19468_c2 | C | T | T | A | T | CT T A T |
| - NA19469_c1 | C | T | T | A | T | CT T A T |
| - NA19469_c2 | T | T | T | A | T | TT T A T |
| - NA19472_c1 | C | T | T | A | T | CT T A T |
| - NA19472_c2 | T | G | T | A | T | TG T A T |
| - NA19346_c1 | C | T | T | A | T | CT T A T |
| - NA19346_c2 | T | T | T | A | C | TT T A C |

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|---|---|---|---|---|---|---|
| - NA19347_c1 | C | T | T | A | C | CT T A C |
| - NA19347_c2 | C | T | T | A | T | CT T A T |
| - NA19371_c1 | T | T | C | A | C | TT C A C |
| - NA19371_c2 | C | T | T | A | T | CT T A T |
| - NA19374_c1 | T | T | C | A | C | TT C A C |
| - NA19374_c2 | T | T | C | A | T | TT C A T |
| - NA19382_c1 | T | T | T | A | T | TT T A T |
| - NA19382_c2 | T | T | T | A | T | TT T A T |
| - NA19384_c1 | T | T | C | A | T | TT C A T |
| - NA19384_c2 | C | T | T | A | T | CT T A T |
| - NA19385_c1 | T | G | T | A | T | TG T A T |
| - NA19385_c2 | C | T | T | A | T | CT T A T |
| - NA19444_c1 | T | T | C | A | C | TT C A C |
| - NA19444_c2 | C | T | T | A | T | CT T A T |
| - NA19448_c1 | C | T | T | A | T | CT T A T |
| - NA19448_c2 | C | T | T | A | T | CT T A T |
| - NA19452_c1 | C | T | T | A | T | CT T A T |
| - NA19452_c2 | T | T | T | A | T | TT T A T |
| - NA19327_c1 | T | T | C | A | C | TT C A C |
| - NA19327_c2 | C | T | T | A | T | CT T A T |
| - NA19390_c1 | C | T | T | A | T | CT T A T |
| - NA19390_c2 | C | T | T | A | T | CT T A T |
| - NA19399_c1 | T | T | C | A | T | TT C A T |
| - NA19399_c2 | C | T | T | A | T | CT T A T |
| - NA19404_c1 | C | T | T | A | T | CT T A T |
| - NA19404_c2 | C | T | T | A | T | CT T A T |
| - NA19431_c1 | T | T | T | A | T | TT T A T |
| - NA19431_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19449_c1 | C | T | T | A | T | CT T A T |
| - NA19449_c2 | C | T | T | A | T | CT T A T |
| - NA19456_c1 | T | T | T | A | T | TT T A T |
| - NA19456_c2 | C | T | T | A | T | CT T A T |
| - NA19474_c1 | C | T | T | A | T | CT T A T |
| - NA19474_c2 | T | T | T | A | T | TT T A T |
| - NA19373_c1 | C | G | C | A | T | CG C A T |
| - NA19373_c2 | T | T | C | A | T | TT C A T |
| - NA19036_c1 | C | T | T | A | T | CT T A T |
| - NA19036_c2 | T | T | C | A | C | TT C A C |
| - NA19309_c1 | C | T | T | A | T | CT T A T |
| - NA19309_c2 | T | T | C | A | C | TT C A C |
| - NA19394_c1 | T | T | T | A | C | TT T A C |
| - NA19394_c2 | C | T | T | A | T | CT T A T |
| - NA19451_c1 | C | G | C | A | C | CG C A C |
| - NA19451_c2 | C | T | T | A | T | CT T A T |
| - NA19044_c1 | T | T | T | A | T | TT T A T |
| - NA19044_c2 | T | T | T | A | C | TT T A C |
| - NA19360_c1 | C | T | T | A | T | CT T A T |
| - NA19360_c2 | C | T | T | A | T | CT T A T |
| - NA19310_c1 | T | T | T | A | C | TT T A C |
| - NA19310_c2 | C | T | T | A | T | CT T A T |
| - NA19457_c1 | C | T | T | A | T | CT T A T |
| - NA19457_c2 | T | T | T | A | T | TT T A T |
| - NA19372_c1 | T | T | C | A | C | TT C A C |
| - NA19372_c2 | C | T | T | A | T | CT T A T |
| - NA19663_c1 | C | T | T | A | T | CT T A T |
| - NA19663_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19664_c1 | C | T | T | A | T | CT T A T |
| - NA19664_c2 | C | T | T | A | T | CT T A T |
| - NA19684_c1 | C | T | T | A | T | CT T A T |
| - NA19684_c2 | C | G | C | C | C | CG C C C |
| - NA19685_c1 | C | G | C | A | C | CG C A C |
| - NA19685_c2 | C | T | T | A | T | CT T A T |
| - NA19722_c1 | C | G | C | C | C | CG C C C |
| - NA19722_c2 | C | T | T | A | T | CT T A T |
| - NA19723_c1 | C | G | C | A | C | CG C A C |
| - NA19723_c2 | C | T | T | A | C | CT T A C |
| - NA19675_c1 | C | T | T | A | T | CT T A T |
| - NA19675_c2 | C | G | C | A | C | CG C A C |
| - NA19676_c1 | C | G | C | A | C | CG C A C |
| - NA19676_c2 | C | G | C | C | C | CG C C C |
| - NA19651_c1 | C | T | T | A | T | CT T A T |
| - NA19651_c2 | C | T | T | A | T | CT T A T |
| - NA19652_c1 | C | T | T | A | T | CT T A T |
| - NA19652_c2 | T | T | T | A | T | TT T A T |
| - NA19681_c1 | T | T | T | A | T | TT T A T |
| - NA19681_c2 | C | G | C | A | C | CG C A C |
| - NA19682_c1 | C | G | C | C | C | CG C C C |
| - NA19682_c2 | C | G | C | A | C | CG C A C |
| - NA19725_c1 | C | G | C | A | C | CG C A C |
| - NA19725_c2 | C | G | C | A | C | CG C A C |
| - NA19726_c1 | C | G | C | A | C | CG C A C |
| - NA19726_c2 | C | T | T | A | T | CT T A T |
| - NA19755_c1 | C | T | T | A | C | CT T A C |
| - NA19755_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA19756_c1 | C | G | C | A | C | CG C A C |
| - NA19756_c2 | C | G | C | A | C | CG C A C |
| - NA19770_c1 | C | T | T | A | T | CT T A T |
| - NA19770_c2 | C | G | C | C | C | CG C C C |
| - NA19771_c1 | C | T | T | A | T | CT T A T |
| - NA19771_c2 | C | T | T | A | T | CT T A T |
| - NA19773_c1 | C | G | C | A | C | CG C A C |
| - NA19773_c2 | C | T | T | A | T | CT T A T |
| - NA19774_c1 | C | T | T | A | C | CT T A C |
| - NA19774_c2 | C | T | T | A | T | CT T A T |
| - NA19776_c1 | C | G | C | C | C | CG C C C |
| - NA19776_c2 | C | T | T | A | C | CT T A C |
| - NA19777_c1 | C | G | C | A | C | CG C A C |
| - NA19777_c2 | C | T | T | A | C | CT T A C |
| - NA19782_c1 | C | G | C | C | C | CG C C C |
| - NA19782_c2 | C | T | T | A | C | CT T A C |
| - NA19783_c1 | C | G | C | A | C | CG C A C |
| - NA19783_c2 | C | T | T | A | T | CT T A T |
| - NA19794_c1 | C | T | T | A | T | CT T A T |
| - NA19794_c2 | T | T | C | A | T | TT C A T |
| - NA19795_c1 | C | T | T | A | T | CT T A T |
| - NA19795_c2 | C | G | C | A | C | CG C A C |
| - NA19669_c1 | C | T | T | A | C | CT T A C |
| - NA19669_c2 | C | T | T | A | T | CT T A T |
| - NA19670_c1 | C | G | C | A | C | CG C A C |
| - NA19670_c2 | C | T | T | A | T | CT T A T |
| - NA19779_c1 | C | T | T | A | T | CT T A T |
| - NA19779_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA19780_c1 | C | G | C | C | T | CG C C T |
| - NA19780_c2 | C | T | T | A | T | CT T A T |
| - NA19657_c1 | C | G | C | C | C | CG C C C |
| - NA19657_c2 | C | G | C | A | C | CG C A C |
| - NA19658_c1 | C | G | C | A | C | CG C A C |
| - NA19658_c2 | C | T | T | A | T | CT T A T |
| - NA19660_c1 | C | T | T | A | T | CT T A T |
| - NA19660_c2 | C | T | T | A | T | CT T A T |
| - NA19661_c1 | C | G | C | A | C | CG C A C |
| - NA19661_c2 | T | T | T | A | T | TT T A T |
| - NA19678_c1 | C | G | C | A | C | CG C A C |
| - NA19678_c2 | C | G | C | A | C | CG C A C |
| - NA19679_c1 | C | T | T | A | T | CT T A T |
| - NA19679_c2 | C | T | T | A | T | CT T A T |
| - NA19719_c1 | T | T | C | A | T | TT C A T |
| - NA19719_c2 | C | T | T | A | T | CT T A T |
| - NA19720_c1 | C | G | C | C | C | CG C C C |
| - NA19720_c2 | C | T | T | A | T | CT T A T |
| - NA19746_c1 | C | T | T | A | T | CT T A T |
| - NA19746_c2 | C | G | C | C | C | CG C C C |
| - NA19747_c1 | T | T | T | A | T | TT T A T |
| - NA19747_c2 | C | T | T | A | C | CT T A C |
| - NA19788_c1 | T | T | T | A | T | TT T A T |
| - NA19788_c2 | C | T | T | A | T | CT T A T |
| - NA19789_c1 | T | T | T | A | T | TT T A T |
| - NA19789_c2 | C | T | T | A | T | CT T A T |
| - NA19749_c1 | C | T | T | A | T | CT T A T |
| - NA19749_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19750_c1 | C | T | T | A | T | CT T A T |
| - NA19750_c2 | C | G | C | A | C | CG C A C |
| - NA19761_c1 | C | G | C | A | C | CG C A C |
| - NA19761_c2 | C | G | C | A | C | CG C A C |
| - NA19762_c1 | C | G | C | A | C | CG C A C |
| - NA19762_c2 | T | T | T | A | T | TT T A T |
| - NA19649_c1 | C | T | T | A | T | CT T A T |
| - NA19649_c2 | C | T | T | A | C | CT T A C |
| - NA19649_NA19648_c1 | C | T | T | A | T | CT T A T |
| - NA19649_NA19648_c2 | N | N | N | N | N | NN N N N |
| - NA19654_c1 | C | T | T | A | T | CT T A T |
| - NA19654_c2 | C | T | T | A | T | CT T A T |
| - NA19654_NA19655_c1 | C | T | T | A | C | CT T A C |
| - NA19654_NA19655_c2 | N | N | N | N | N | NN N N N |
| - NA19716_c1 | C | T | T | A | C | CT T A C |
| - NA19716_c2 | T | T | T | A | C | TT T A C |
| - NA19716_NA19717_c1 | C | T | T | A | T | CT T A T |
| - NA19716_NA19717_c2 | N | N | N | N | N | NN N N N |
| - NA19759_c1 | C | G | C | A | C | CG C A C |
| - NA19759_c2 | T | T | T | A | T | TT T A T |
| - NA19759_NA19758_c1 | C | G | C | C | C | CG C C C |
| - NA19759_NA19758_c2 | N | N | N | N | N | NN N N N |
| - NA21303_c1 | T | T | C | A | C | TT C A C |
| - NA21303_c2 | T | T | C | A | C | TT C A C |
| - NA21301_c1 | C | T | T | A | C | CT T A C |
| - NA21301_c2 | C | T | T | A | T | CT T A T |
| - NA21479_c1 | C | T | T | A | T | CT T A T |
| - NA21479_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21478_c1 | T | T | T | A | C | TT T A C |
| - NA21478_c2 | C | G | C | A | C | CG C A C |
| - NA21615_c1 | C | T | T | A | T | CT T A T |
| - NA21615_c2 | C | T | T | A | T | CT T A T |
| - NA21614_c1 | C | T | T | A | T | CT T A T |
| - NA21614_c2 | C | T | T | A | T | CT T A T |
| - NA21476_c1 | C | T | T | A | T | CT T A T |
| - NA21476_c2 | C | T | T | A | T | CT T A T |
| - NA21475_c1 | C | T | T | A | T | CT T A T |
| - NA21475_c2 | C | G | C | A | C | CG C A C |
| - NA21362_c1 | C | T | T | A | T | CT T A T |
| - NA21362_c2 | C | T | T | A | T | CT T A T |
| - NA21312_c1 | C | T | T | A | T | CT T A T |
| - NA21312_c2 | T | T | C | A | C | TT C A C |
| - NA21385_c1 | C | T | T | A | T | CT T A T |
| - NA21385_c2 | T | T | C | A | C | TT C A C |
| - NA21384_c1 | C | G | C | A | C | CG C A C |
| - NA21384_c2 | C | T | T | A | T | CT T A T |
| - NA21493_c1 | C | T | T | A | T | CT T A T |
| - NA21493_c2 | C | G | C | A | T | CG C A T |
| - NA21522_c1 | C | T | T | A | T | CT T A T |
| - NA21522_c2 | T | G | T | A | T | TG T A T |
| - NA21400_c1 | C | T | T | A | T | CT T A T |
| - NA21400_c2 | T | T | C | A | T | TT C A T |
| - NA21399_c1 | C | T | T | A | T | CT T A T |
| - NA21399_c2 | T | T | C | A | C | TT C A C |
| - NA21388_c1 | C | G | C | A | C | CG C A C |
| - NA21388_c2 | T | T | T | A | C | TT T A C |

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|---|---|---|---|---|---|---|
| - NA21387_c1 | C | T | T | A | T | CT T A T |
| - NA21387_c2 | C | T | T | A | T | CT T A T |
| - NA21489_c1 | C | T | T | A | T | CT T A T |
| - NA21489_c2 | C | T | T | A | T | CT T A T |
| - NA21488_c1 | C | G | C | A | C | CG C A C |
| - NA21488_c2 | T | T | C | A | C | TT C A C |
| - NA21403_c1 | C | T | T | A | C | CT T A C |
| - NA21403_c2 | C | G | C | A | C | CG C A C |
| - NA21402_c1 | C | T | T | A | C | CT T A C |
| - NA21402_c2 | C | T | T | A | T | CT T A T |
| - NA21382_c1 | T | T | C | A | T | TT C A T |
| - NA21382_c2 | C | T | T | A | T | CT T A T |
| - NA21381_c1 | C | T | T | A | T | CT T A T |
| - NA21381_c2 | C | T | T | A | T | CT T A T |
| - NA21454_c1 | T | T | C | A | T | TT C A T |
| - NA21454_c2 | C | T | T | A | T | CT T A T |
| - NA21453_c1 | C | T | T | A | T | CT T A T |
| - NA21453_c2 | C | T | T | A | T | CT T A T |
| - NA21524_c1 | T | T | T | A | C | TT T A C |
| - NA21524_c2 | C | T | T | A | T | CT T A T |
| - NA21523_c1 | C | T | T | A | T | CT T A T |
| - NA21523_c2 | T | T | C | A | C | TT C A C |
| - NA21635_c1 | C | T | T | A | T | CT T A T |
| - NA21635_c2 | C | T | T | A | T | CT T A T |
| - NA21634_c1 | C | T | T | A | T | CT T A T |
| - NA21634_c2 | C | G | C | A | C | CG C A C |
| - NA21686_c1 | C | T | T | A | T | CT T A T |
| - NA21686_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21647_c1 | C | G | C | A | C | CG C A C |
| - NA21647_c2 | C | G | C | A | C | CG C A C |
| - NA21360_c1 | C | T | T | A | T | CT T A T |
| - NA21360_c2 | C | T | T | A | T | CT T A T |
| - NA21359_c1 | T | T | C | A | C | TT C A C |
| - NA21359_c2 | C | T | T | A | T | CT T A T |
| - NA21513_c1 | T | T | C | A | C | TT C A C |
| - NA21513_c2 | C | T | T | A | T | CT T A T |
| - NA21512_c1 | T | T | C | A | C | TT C A C |
| - NA21512_c2 | C | T | T | A | T | CT T A T |
| - NA21365_c1 | C | T | T | A | T | CT T A T |
| - NA21365_c2 | T | T | C | A | C | TT C A C |
| - NA21344_c1 | C | T | T | A | C | CT T A C |
| - NA21344_c2 | C | T | T | A | T | CT T A T |
| - NA21580_c1 | C | T | T | A | T | CT T A T |
| - NA21580_c2 | C | T | T | A | T | CT T A T |
| - NA21316_c1 | T | T | C | A | C | TT C A C |
| - NA21316_c2 | C | T | T | A | T | CT T A T |
| - NA21526_c1 | T | T | C | A | C | TT C A C |
| - NA21526_c2 | C | G | C | A | T | CG C A T |
| - NA21583_c1 | T | T | T | A | T | TT T A T |
| - NA21583_c2 | T | T | T | A | T | TT T A T |
| - NA21717_c1 | C | G | C | A | C | CG C A C |
| - NA21717_c2 | C | T | T | A | T | CT T A T |
| - NA21716_c1 | T | T | C | A | T | TT C A T |
| - NA21716_c2 | T | T | C | A | C | TT C A C |
| - NA21438_c1 | C | T | T | A | T | CT T A T |
| - NA21438_c2 | T | T | C | A | C | TT C A C |

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|---|---|---|---|---|---|---|
| - NA21447_c1 | T | T | C | A | C | TT C A C |
| - NA21447_c2 | T | T | C | A | C | TT C A C |
| - NA21600_c1 | C | T | T | A | T | CT T A T |
| - NA21600_c2 | C | T | T | A | T | CT T A T |
| - NA21599_c1 | C | T | T | A | T | CT T A T |
| - NA21599_c2 | T | T | C | A | C | TT C A C |
| - NA21308_c1 | C | T | T | A | T | CT T A T |
| - NA21308_c2 | C | G | C | A | C | CG C A C |
| - NA21307_c1 | T | T | C | A | C | TT C A C |
| - NA21307_c2 | C | T | T | A | T | CT T A T |
| - NA21424_c1 | C | T | T | A | T | CT T A T |
| - NA21424_c2 | C | G | C | A | C | CG C A C |
| - NA21423_c1 | C | G | C | A | C | CG C A C |
| - NA21423_c2 | T | T | C | A | C | TT C A C |
| - NA21486_c1 | C | T | T | A | T | CT T A T |
| - NA21486_c2 | C | G | C | A | C | CG C A C |
| - NA21485_c1 | T | T | C | A | C | TT C A C |
| - NA21485_c2 | C | G | C | C | C | CG C C C |
| - NA21441_c1 | C | T | T | A | T | CT T A T |
| - NA21441_c2 | C | T | T | A | T | CT T A T |
| - NA21440_c1 | C | T | T | A | T | CT T A T |
| - NA21440_c2 | C | T | T | A | T | CT T A T |
| - NA21297_c1 | C | T | T | A | T | CT T A T |
| - NA21297_c2 | C | T | T | A | T | CT T A T |
| - NA21379_c1 | C | T | T | A | T | CT T A T |
| - NA21379_c2 | C | G | C | A | C | CG C A C |
| - NA21528_c1 | C | T | T | A | T | CT T A T |
| - NA21528_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21391_c1 | C | T | T | A | T | CT T A T |
| - NA21391_c2 | C | T | T | A | T | CT T A T |
| - NA21336_c1 | C | T | T | A | T | CT T A T |
| - NA21336_c2 | C | T | T | A | T | CT T A T |
| - NA21741_c1 | C | T | T | A | T | CT T A T |
| - NA21741_c2 | T | G | T | A | T | TG T A T |
| - NA21576_c1 | T | T | C | A | C | TT C A C |
| - NA21576_c2 | T | T | C | A | C | TT C A C |
| - NA21740_c1 | C | T | T | A | T | CT T A T |
| - NA21740_c2 | C | T | T | A | T | CT T A T |
| - NA21451_c1 | T | T | C | A | C | TT C A C |
| - NA21451_c2 | C | T | T | A | T | CT T A T |
| - NA21733_c1 | C | G | C | C | C | CG C C C |
| - NA21733_c2 | T | T | C | A | C | TT C A C |
| - NA21616_c1 | C | T | T | A | T | CT T A T |
| - NA21616_c2 | C | T | T | A | T | CT T A T |
| - NA21434_c1 | C | G | C | A | C | CG C A C |
| - NA21434_c2 | C | T | T | A | T | CT T A T |
| - NA21784_c1 | C | T | T | A | T | CT T A T |
| - NA21784_c2 | C | G | C | A | C | CG C A C |
| - NA21300_c1 | C | T | T | A | T | CT T A T |
| - NA21300_c2 | C | T | T | A | T | CT T A T |
| - NA21435_c1 | C | T | T | A | T | CT T A T |
| - NA21435_c2 | C | T | T | A | T | CT T A T |
| - NA21357_c1 | C | T | T | A | C | CT T A C |
| - NA21357_c2 | T | T | C | A | T | TT C A T |
| - NA21355_c1 | T | T | T | A | T | TT T A T |
| - NA21355_c2 | C | G | C | A | C | CG C A C |

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|---|---|---|---|---|---|---|
| - NA21578_c1 | T | T | C | A | C | TT C A C |
| - NA21578_c2 | C | T | T | A | T | CT T A T |
| - NA21414_c1 | C | T | T | A | T | CT T A T |
| - NA21414_c2 | T | T | T | A | C | TT T A C |
| - NA21491_c1 | C | G | C | C | C | CG C C C |
| - NA21491_c2 | C | T | T | A | T | CT T A T |
| - NA21405_c1 | T | T | C | A | C | TT C A C |
| - NA21405_c2 | C | T | T | A | T | CT T A T |
| - NA21390_c1 | C | T | T | A | T | CT T A T |
| - NA21390_c2 | T | T | T | A | T | TT T A T |
| - NA21650_c1 | T | T | C | A | C | TT C A C |
| - NA21650_c2 | C | T | T | A | T | CT T A T |
| - NA21768_c1 | T | G | T | A | T | TG T A T |
| - NA21768_c2 | C | T | T | A | T | CT T A T |
| - NA21582_c1 | C | G | C | C | C | CG C C C |
| - NA21582_c2 | C | T | T | A | T | CT T A T |
| - NA21776_c1 | C | T | T | A | T | CT T A T |
| - NA21776_c2 | C | T | T | A | T | CT T A T |
| - NA21575_c1 | C | T | T | A | T | CT T A T |
| - NA21575_c2 | C | T | T | A | T | CT T A T |
| - NA21825_c1 | C | G | C | A | T | CG C A T |
| - NA21825_c2 | C | T | T | A | T | CT T A T |
| - NA21719_c1 | C | T | T | A | T | CT T A T |
| - NA21719_c2 | C | T | T | A | T | CT T A T |
| - NA21378_c1 | C | T | T | A | C | CT T A C |
| - NA21378_c2 | C | T | T | A | T | CT T A T |
| - NA21368_c1 | C | T | T | A | T | CT T A T |
| - NA21368_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21573_c1 | T | T | C | A | C | TT C A C |
| - NA21573_c2 | T | T | T | A | C | TT T A C |
| - NA21320_c1 | C | G | C | A | C | CG C A C |
| - NA21320_c2 | C | T | T | A | T | CT T A T |
| - NA21678_c1 | C | T | T | A | T | CT T A T |
| - NA21678_c2 | C | T | T | A | T | CT T A T |
| - NA21682_c1 | C | T | T | A | T | CT T A T |
| - NA21682_c2 | T | T | T | A | C | TT T A C |
| - NA21723_c1 | T | T | C | A | C | TT C A C |
| - NA21723_c2 | C | T | T | A | T | CT T A T |
| - NA21356_c1 | T | T | C | A | C | TT C A C |
| - NA21356_c2 | T | T | C | A | C | TT C A C |
| - NA21685_c1 | C | T | T | A | T | CT T A T |
| - NA21685_c2 | C | T | T | A | T | CT T A T |
| - NA21339_c1 | C | T | T | A | C | CT T A C |
| - NA21339_c2 | T | T | T | A | T | TT T A T |
| - NA21574_c1 | C | T | T | A | T | CT T A T |
| - NA21574_c2 | C | G | C | A | C | CG C A C |
| - NA21826_c1 | C | T | T | A | C | CT T A C |
| - NA21826_c2 | T | T | C | A | C | TT C A C |
| - NA21519_c1 | C | T | T | A | T | CT T A T |
| - NA21519_c2 | T | G | T | A | T | TG T A T |
| - NA21611_c1 | C | T | T | A | T | CT T A T |
| - NA21611_c2 | T | T | T | A | T | TT T A T |
| - NA21632_c1 | C | T | T | A | T | CT T A T |
| - NA21632_c2 | C | T | T | A | T | CT T A T |
| - NA21509_c1 | C | T | T | A | T | CT T A T |
| - NA21509_c2 | T | T | C | A | C | TT C A C |

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|---|---|---|---|---|---|---|
| - NA21473_c1 | C | G | C | A | C | CG C A C |
| - NA21473_c2 | T | T | C | A | C | TT C A C |
| - NA21408_c1 | T | T | T | A | C | TT T A C |
| - NA21408_c2 | C | T | T | A | T | CT T A T |
| - NA21510_c1 | C | T | T | A | T | CT T A T |
| - NA21510_c2 | C | T | T | A | T | CT T A T |
| - NA21352_c1 | C | T | T | A | T | CT T A T |
| - NA21352_c2 | C | T | T | A | T | CT T A T |
| - NA21617_c1 | C | T | T | A | T | CT T A T |
| - NA21617_c2 | C | T | T | A | T | CT T A T |
| - NA21520_c1 | C | T | T | A | T | CT T A T |
| - NA21520_c2 | C | T | T | A | T | CT T A T |
| - NA21418_c1 | T | T | C | A | C | TT C A C |
| - NA21418_c2 | T | T | C | A | C | TT C A C |
| - NA21367_c1 | C | T | T | A | T | CT T A T |
| - NA21367_c2 | T | T | C | A | C | TT C A C |
| - NA21448_c1 | C | T | T | A | T | CT T A T |
| - NA21448_c2 | C | G | C | A | C | CG C A C |
| - NA21683_c1 | C | T | T | A | T | CT T A T |
| - NA21683_c2 | C | T | T | A | T | CT T A T |
| - NA21738_c1 | C | T | T | A | T | CT T A T |
| - NA21738_c2 | C | T | T | A | T | CT T A T |
| - NA21739_c1 | C | T | T | A | T | CT T A T |
| - NA21739_c2 | T | T | T | A | T | TT T A T |
| - NA21417_c1 | C | T | T | A | T | CT T A T |
| - NA21417_c2 | T | T | C | A | C | TT C A C |
| - NA21415_c1 | T | T | T | A | C | TT T A C |
| - NA21415_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21521_c1 | C | T | T | A | T | CT T A T |
| - NA21521_c2 | C | T | T | A | T | CT T A T |
| - NA21311_c1 | T | T | T | A | T | TT T A T |
| - NA21311_c2 | T | T | C | A | C | TT C A C |
| - NA21363_c1 | C | G | C | A | C | CG C A C |
| - NA21363_c2 | C | T | T | A | T | CT T A T |
| - NA21596_c1 | T | T | C | A | T | TT C A T |
| - NA21596_c2 | C | G | C | A | C | CG C A C |
| - NA21693_c1 | C | T | T | A | T | CT T A T |
| - NA21693_c2 | C | T | T | A | T | CT T A T |
| - NA21587_c1 | C | T | T | A | T | CT T A T |
| - NA21587_c2 | C | T | T | A | T | CT T A T |
| - NA21529_c1 | C | G | C | A | C | CG C A C |
| - NA21529_c2 | C | G | C | A | C | CG C A C |
| - NA21436_c1 | C | G | C | A | C | CG C A C |
| - NA21436_c2 | C | T | T | A | T | CT T A T |
| - NA21420_c1 | C | T | T | A | T | CT T A T |
| - NA21420_c2 | C | T | T | A | T | CT T A T |
| - NA21364_c1 | C | T | T | A | T | CT T A T |
| - NA21364_c2 | C | T | T | A | T | CT T A T |
| - NA21689_c1 | C | T | T | A | T | CT T A T |
| - NA21689_c2 | T | T | T | A | T | TT T A T |
| - NA21597_c1 | T | T | C | A | C | TT C A C |
| - NA21597_c2 | T | T | T | A | C | TT T A C |
| - NA21353_c1 | T | T | C | A | C | TT C A C |
| - NA21353_c2 | T | G | T | A | T | TG T A T |
| - NA21613_c1 | C | G | C | A | C | CG C A C |
| - NA21613_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA21421_c1 | C | T | T | A | T | CT T A T |
| - NA21421_c2 | C | T | T | A | T | CT T A T |
| - NA21631_c1 | C | T | T | A | T | CT T A T |
| - NA21631_c2 | C | T | T | A | T | CT T A T |
| - NA21620_c1 | C | T | T | A | T | CT T A T |
| - NA21620_c2 | C | T | T | A | T | CT T A T |
| - NA21619_c1 | C | T | T | A | T | CT T A T |
| - NA21619_c2 | C | T | T | A | T | CT T A T |
| - NA21295_c1 | T | T | T | A | T | TT T A T |
| - NA21295_c2 | T | T | T | A | T | TT T A T |
| - NA21517_c1 | C | T | T | A | T | CT T A T |
| - NA21517_c2 | C | T | T | A | T | CT T A T |
| - NA21314_c1 | T | T | C | A | C | TT C A C |
| - NA21314_c2 | C | G | C | A | C | CG C A C |
| - NA21333_c1 | C | G | C | A | C | CG C A C |
| - NA21333_c2 | C | G | C | A | C | CG C A C |
| - NA21457_c1 | C | T | T | A | T | CT T A T |
| - NA21457_c2 | C | T | T | A | T | CT T A T |
| - NA21722_c1 | C | T | T | A | T | CT T A T |
| - NA21722_c2 | C | T | T | A | T | CT T A T |
| - NA21318_c1 | C | T | T | A | T | CT T A T |
| - NA21318_c2 | T | T | C | A | C | TT C A C |
| - NA21370_c1 | T | G | T | A | T | TG T A T |
| - NA21370_c2 | C | T | T | A | T | CT T A T |
| - NA21577_c1 | C | T | T | A | T | CT T A T |
| - NA21577_c2 | T | T | C | A | C | TT C A C |
| - NA21371_c1 | C | T | T | A | T | CT T A T |
| - NA21371_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA20505_c1 | C | G | C | C | T | CG C C T |
| - NA20505_c2 | T | T | T | A | C | TT T A C |
| - NA20504_c1 | C | G | C | C | T | CG C C T |
| - NA20504_c2 | C | G | C | A | T | CG C A T |
| - NA20506_c1 | T | T | T | A | T | TT T A T |
| - NA20506_c2 | T | T | T | A | T | TT T A T |
| - NA20502_c1 | C | T | T | A | T | CT T A T |
| - NA20502_c2 | C | G | C | C | C | CG C C C |
| - NA20528_c1 | C | G | C | A | C | CG C A C |
| - NA20528_c2 | C | G | C | A | C | CG C A C |
| - NA20531_c1 | T | T | T | A | T | TT T A T |
| - NA20531_c2 | C | T | T | A | T | CT T A T |
| - NA20534_c1 | C | T | T | A | T | CT T A T |
| - NA20534_c2 | C | G | C | C | C | CG C C C |
| - NA20535_c1 | T | T | T | A | T | TT T A T |
| - NA20535_c2 | C | G | C | C | C | CG C C C |
| - NA20586_c1 | C | G | C | A | C | CG C A C |
| - NA20586_c2 | T | T | T | A | T | TT T A T |
| - NA20756_c1 | C | G | C | A | C | CG C A C |
| - NA20756_c2 | C | G | C | C | C | CG C C C |
| - NA20760_c1 | C | G | C | A | C | CG C A C |
| - NA20760_c2 | C | T | T | A | T | CT T A T |
| - NA20765_c1 | T | T | T | A | T | TT T A T |
| - NA20765_c2 | T | T | T | A | T | TT T A T |
| - NA20766_c1 | C | G | C | A | C | CG C A C |
| - NA20766_c2 | C | G | C | A | C | CG C A C |
| - NA20769_c1 | C | T | T | A | T | CT T A T |
| - NA20769_c2 | C | G | C | C | C | CG C C C |

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|---|---|---|---|---|---|---|
| - NA20771_c1 | C | G | C | A | C | CG C A C |
| - NA20771_c2 | C | G | C | C | C | CG C C C |
| - NA20512_c1 | C | T | T | A | C | CT T A C |
| - NA20512_c2 | T | T | T | A | T | TT T A T |
| - NA20515_c1 | C | G | C | A | T | CG C A T |
| - NA20515_c2 | T | T | T | A | T | TT T A T |
| - NA20516_c1 | C | G | C | A | C | CG C A C |
| - NA20516_c2 | C | G | C | C | C | CG C C C |
| - NA20517_c1 | C | G | C | A | C | CG C A C |
| - NA20517_c2 | T | T | T | A | T | TT T A T |
| - NA20518_c1 | C | G | C | C | C | CG C C C |
| - NA20518_c2 | T | T | T | A | T | TT T A T |
| - NA20530_c1 | C | T | T | A | T | CT T A T |
| - NA20530_c2 | C | T | T | A | T | CT T A T |
| - NA20538_c1 | C | T | T | A | T | CT T A T |
| - NA20538_c2 | T | T | T | A | T | TT T A T |
| - NA20539_c1 | C | G | C | C | C | CG C C C |
| - NA20539_c2 | C | G | C | C | C | CG C C C |
| - NA20542_c1 | C | T | T | A | T | CT T A T |
| - NA20542_c2 | T | T | T | A | T | TT T A T |
| - NA20544_c1 | C | T | T | A | T | CT T A T |
| - NA20544_c2 | C | G | C | A | C | CG C A C |
| - NA20588_c1 | C | G | C | C | C | CG C C C |
| - NA20588_c2 | C | T | T | A | T | CT T A T |
| - NA20752_c1 | C | G | C | A | C | CG C A C |
| - NA20752_c2 | T | T | T | A | T | TT T A T |
| - NA20753_c1 | C | T | T | A | C | CT T A C |
| - NA20753_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA20755_c1 | T | T | T | A | T | TT T A T |
| - NA20755_c2 | C | G | C | A | C | CG C A C |
| - NA20759_c1 | C | G | C | A | C | CG C A C |
| - NA20759_c2 | T | T | T | A | T | TT T A T |
| - NA20770_c1 | C | G | C | A | C | CG C A C |
| - NA20770_c2 | T | T | T | A | T | TT T A T |
| - NA20775_c1 | C | G | C | C | C | CG C C C |
| - NA20775_c2 | T | T | T | A | T | TT T A T |
| - NA20785_c1 | T | T | T | A | T | TT T A T |
| - NA20785_c2 | C | T | T | A | T | CT T A T |
| - NA20796_c1 | T | T | T | A | T | TT T A T |
| - NA20796_c2 | C | G | C | C | C | CG C C C |
| - NA20799_c1 | T | T | T | A | C | TT T A C |
| - NA20799_c2 | T | T | T | A | T | TT T A T |
| - NA20808_c1 | T | T | T | A | T | TT T A T |
| - NA20808_c2 | C | T | T | A | T | CT T A T |
| - NA20810_c1 | T | T | T | A | T | TT T A T |
| - NA20810_c2 | C | G | C | C | C | CG C C C |
| - NA20812_c1 | C | G | C | A | C | CG C A C |
| - NA20812_c2 | C | G | C | A | C | CG C A C |
| - NA20813_c1 | C | G | C | C | C | CG C C C |
| - NA20813_c2 | C | G | C | A | C | CG C A C |
| - NA20815_c1 | C | G | C | C | C | CG C C C |
| - NA20815_c2 | C | G | C | A | C | CG C A C |
| - NA20816_c1 | C | G | C | C | C | CG C C C |
| - NA20816_c2 | C | G | C | A | C | CG C A C |
| - NA20819_c1 | C | G | C | C | C | CG C C C |
| - NA20819_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA20826_c1 | C | G | C | C | C | CG C C C |
| - NA20826_c2 | T | T | T | A | T | TT T A T |
| - NA20509_c1 | C | T | T | A | T | CT T A T |
| - NA20509_c2 | C | T | T | A | T | CT T A T |
| - NA20521_c1 | T | T | T | A | T | TT T A T |
| - NA20521_c2 | C | G | C | C | C | CG C C C |
| - NA20529_c1 | T | T | T | A | T | TT T A T |
| - NA20529_c2 | C | G | C | C | C | CG C C C |
| - NA20540_c1 | C | G | C | A | C | CG C A C |
| - NA20540_c2 | T | T | T | A | T | TT T A T |
| - NA20541_c1 | C | G | C | C | C | CG C C C |
| - NA20541_c2 | C | T | T | A | T | CT T A T |
| - NA20581_c1 | T | T | T | A | T | TT T A T |
| - NA20581_c2 | C | T | T | A | T | CT T A T |
| - NA20582_c1 | C | T | T | A | C | CT T A C |
| - NA20582_c2 | C | G | C | C | C | CG C C C |
| - NA20589_c1 | C | T | T | A | T | CT T A T |
| - NA20589_c2 | C | T | T | A | C | CT T A C |
| - NA20754_c1 | C | G | C | C | T | CG C C T |
| - NA20754_c2 | T | T | T | A | T | TT T A T |
| - NA20772_c1 | C | G | C | C | C | CG C C C |
| - NA20772_c2 | C | G | C | C | C | CG C C C |
| - NA20773_c1 | C | G | C | A | C | CG C A C |
| - NA20773_c2 | C | G | C | C | C | CG C C C |
| - NA20778_c1 | C | G | C | C | C | CG C C C |
| - NA20778_c2 | C | G | C | A | C | CG C A C |
| - NA20787_c1 | C | G | C | A | C | CG C A C |
| - NA20787_c2 | C | G | C | C | T | CG C C T |

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|---|---|---|---|---|---|---|
| - NA20790_c1 | C | G | C | A | C | CG C A C |
| - NA20790_c2 | T | T | T | A | T | TT T A T |
| - NA20792_c1 | C | T | T | A | T | CT T A T |
| - NA20792_c2 | T | T | T | A | T | TT T A T |
| - NA20795_c1 | T | T | T | A | T | TT T A T |
| - NA20795_c2 | C | T | T | A | T | CT T A T |
| - NA20797_c1 | C | G | C | C | C | CG C C C |
| - NA20797_c2 | C | T | T | A | T | CT T A T |
| - NA20800_c1 | C | T | T | A | T | CT T A T |
| - NA20800_c2 | C | T | T | A | T | CT T A T |
| - NA20801_c1 | C | G | C | A | C | CG C A C |
| - NA20801_c2 | C | G | C | C | C | CG C C C |
| - NA20804_c1 | C | T | T | A | T | CT T A T |
| - NA20804_c2 | C | G | C | A | C | CG C A C |
| - NA20806_c1 | C | T | T | A | T | CT T A T |
| - NA20806_c2 | T | T | T | A | T | TT T A T |
| - NA20807_c1 | C | T | T | A | T | CT T A T |
| - NA20807_c2 | T | T | T | A | T | TT T A T |
| - NA20809_c1 | C | T | T | A | T | CT T A T |
| - NA20809_c2 | C | T | T | A | T | CT T A T |
| - NA20510_c1 | C | G | C | C | T | CG C C T |
| - NA20510_c2 | C | T | T | A | T | CT T A T |
| - NA20519_c1 | C | T | T | A | T | CT T A T |
| - NA20519_c2 | T | T | C | A | T | TT C A T |
| - NA20543_c1 | C | G | C | C | C | CG C C C |
| - NA20543_c2 | T | T | T | A | T | TT T A T |
| - NA20758_c1 | C | G | C | C | T | CG C C T |
| - NA20758_c2 | T | T | T | A | T | TT T A T |

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|---|---|---|---|---|---|---|
| - NA20761_c1 | T | T | T | A | T | TT T A T |
| - NA20761_c2 | C | G | C | A | C | CG C A C |
| - NA20768_c1 | C | G | C | A | C | CG C A C |
| - NA20768_c2 | C | T | T | A | T | CT T A T |
| - NA20774_c1 | C | T | T | A | T | CT T A T |
| - NA20774_c2 | T | T | T | A | T | TT T A T |
| - NA20786_c1 | C | T | T | A | T | CT T A T |
| - NA20786_c2 | T | T | C | A | C | TT C A C |
| - NA20802_c1 | T | T | T | A | T | TT T A T |
| - NA20802_c2 | C | G | C | C | C | CG C C C |
| - NA20805_c1 | C | T | T | A | T | CT T A T |
| - NA20805_c2 | C | G | C | C | C | CG C C C |
| - NA20818_c1 | C | T | T | A | T | CT T A T |
| - NA20818_c2 | C | T | T | A | C | CT T A C |
| - NA20508_c1 | T | T | T | A | T | TT T A T |
| - NA20508_c2 | T | T | T | A | T | TT T A T |
| - NA20520_c1 | C | T | T | A | T | CT T A T |
| - NA20520_c2 | C | T | T | A | C | CT T A C |
| - NA20524_c1 | C | T | T | A | T | CT T A T |
| - NA20524_c2 | T | T | T | A | C | TT T A C |
| - NA20525_c1 | C | T | T | A | T | CT T A T |
| - NA20525_c2 | C | G | C | A | C | CG C A C |
| - NA20527_c1 | T | T | T | A | T | TT T A T |
| - NA20527_c2 | C | G | C | A | C | CG C A C |
| - NA20811_c1 | C | G | C | A | C | CG C A C |
| - NA20811_c2 | C | T | T | A | T | CT T A T |
| - NA20522_c1 | C | G | C | A | C | CG C A C |
| - NA20522_c2 | C | G | C | C | C | CG C C C |

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|---|---|---|---|---|---|---|
| - NA20783_c1 | C | T | T | A | T | CT T A T |
| - NA20783_c2 | C | T | T | A | T | CT T A T |
| - NA20803_c1 | C | T | T | A | T | CT T A T |
| - NA20803_c2 | C | T | T | A | T | CT T A T |
| - NA20828_c1 | C | T | T | A | T | CT T A T |
| - NA20828_c2 | T | T | T | A | T | TT T A T |
| - NA20757_c1 | C | G | C | A | T | CG C A T |
| - NA20757_c2 | C | G | C | C | C | CG C C C |
| - NA19095_c1 | T | T | T | A | T | TT T A T |
| - NA19095_c2 | T | T | T | A | T | TT T A T |
| - NA19096_c1 | C | T | T | A | T | CT T A T |
| - NA19096_c2 | C | T | T | A | T | CT T A T |
| - NA18867_c1 | T | T | T | A | T | TT T A T |
| - NA18867_c2 | C | T | T | A | T | CT T A T |
| - NA18868_c1 | T | T | T | A | T | TT T A T |
| - NA18868_c2 | T | G | T | A | T | TG T A T |
| - NA18924_c1 | C | T | T | A | T | CT T A T |
| - NA18924_c2 | T | T | C | A | T | TT C A T |
| - NA18923_c1 | C | T | T | A | T | CT T A T |
| - NA18923_c2 | C | T | T | A | T | CT T A T |
| - NA18488_c1 | C | T | T | A | T | CT T A T |
| - NA18488_c2 | C | T | T | A | T | CT T A T |
| - NA18486_c1 | T | G | T | A | T | TG T A T |
| - NA18486_c2 | T | T | C | A | T | TT C A T |
| - NA19122_c1 | C | T | T | A | T | CT T A T |
| - NA19122_c2 | C | T | T | A | T | CT T A T |
| - NA19121_c1 | C | T | T | A | T | CT T A T |
| - NA19121_c2 | T | T | C | A | T | TT C A T |

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|---|---|---|---|---|---|---|
| - NA19247_c1 | C | T | T | A | T | CT T A T |
| - NA19247_c2 | T | T | C | A | C | TT C A C |
| - NA19248_c1 | C | T | T | A | T | CT T A T |
| - NA19248_c2 | C | T | T | A | T | CT T A T |
| - NA18933_c1 | T | T | C | A | C | TT C A C |
| - NA18933_c2 | C | T | T | A | T | CT T A T |
| - NA18934_c1 | T | T | C | A | C | TT C A C |
| - NA18934_c2 | C | T | T | A | T | CT T A T |
| - NA18499_c1 | C | T | T | A | T | CT T A T |
| - NA18499_c2 | C | T | T | A | T | CT T A T |
| - NA18498_c1 | C | T | T | A | T | CT T A T |
| - NA18498_c2 | T | G | T | A | T | TG T A T |
| - NA19118_c1 | C | T | T | A | C | CT T A C |
| - NA19118_c2 | C | T | T | A | T | CT T A T |
| - NA19117_c1 | C | T | T | A | T | CT T A T |
| - NA19117_c2 | C | T | T | A | T | CT T A T |
| - NA19149_c1 | T | T | C | A | T | TT C A T |
| - NA19149_c2 | C | T | T | A | T | CT T A T |
| - NA19150_c1 | C | T | T | A | T | CT T A T |
| - NA19150_c2 | T | T | C | A | T | TT C A T |
| - NA18489_c1 | C | T | T | A | C | CT T A C |
| - NA18489_c2 | C | T | T | A | T | CT T A T |
| - NA18487_c1 | C | T | T | A | T | CT T A T |
| - NA18487_c2 | C | T | T | A | T | CT T A T |
| - NA19235_c1 | C | T | T | A | T | CT T A T |
| - NA19235_c2 | C | T | T | A | T | CT T A T |
| - NA19236_c1 | C | T | T | A | T | CT T A T |
| - NA19236_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19225_c1 | C | T | T | A | T | CT T A T |
| - NA19225_c2 | T | G | T | A | T | TG T A T |
| - NA19226_c1 | C | T | T | A | T | CT T A T |
| - NA19226_c2 | C | T | T | A | T | CT T A T |
| - NA19190_c1 | T | T | C | A | C | TT C A C |
| - NA19190_c2 | C | T | T | A | T | CT T A T |
| - NA19189_c1 | T | T | T | A | T | TT T A T |
| - NA19189_c2 | C | T | T | A | T | CT T A T |
| - NA18909_c1 | T | T | C | A | C | TT C A C |
| - NA18909_c2 | T | T | C | A | C | TT C A C |
| - NA18910_c1 | C | T | T | A | T | CT T A T |
| - NA18910_c2 | C | T | T | A | T | CT T A T |
| - NA18520_c1 | T | T | T | A | T | TT T A T |
| - NA18520_c2 | T | G | C | A | T | TG C A T |
| - NA18519_c1 | C | T | T | A | T | CT T A T |
| - NA18519_c2 | T | T | C | A | C | TT C A C |
| - NA18916_c1 | C | T | T | A | T | CT T A T |
| - NA18916_c2 | T | G | T | A | T | TG T A T |
| - NA18917_c1 | T | T | C | A | T | TT C A T |
| - NA18917_c2 | C | T | T | A | T | CT T A T |
| - NA19114_c1 | C | T | T | A | C | CT T A C |
| - NA19114_c2 | T | T | T | A | T | TT T A T |
| - NA19113_c1 | T | T | C | A | T | TT C A T |
| - NA19113_c2 | T | T | C | A | C | TT C A C |
| - NA19214_c1 | C | T | T | A | T | CT T A T |
| - NA19214_c2 | T | T | C | A | C | TT C A C |
| - NA19213_c1 | C | T | T | A | T | CT T A T |
| - NA19213_c2 | C | T | T | A | T | CT T A T |

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|---|---|---|---|---|---|---|
| - NA19182_c1 | C | T | T | A | T | CT T A T |
| - NA19182_c2 | T | T | C | A | T | TT C A T |
| - NA19181_c1 | C | T | T | A | T | CT T A T |
| - NA19181_c2 | C | T | T | A | T | CT T A T |
| - NA19179_c1 | C | T | T | A | T | CT T A T |
| - NA19179_c2 | C | T | T | A | T | CT T A T |
| - NA19178_c1 | C | T | T | A | T | CT T A T |
| - NA19178_c2 | C | T | T | A | T | CT T A T |
| - NA19147_c1 | C | T | T | A | T | CT T A T |
| - NA19147_c2 | T | T | C | A | T | TT C A T |
| - NA19146_c1 | C | T | T | A | T | CT T A T |
| - NA19146_c2 | C | T | T | A | C | CT T A C |
| - NA19108_c1 | T | T | C | A | C | TT C A C |
| - NA19108_c2 | C | T | T | A | T | CT T A T |
| - NA19107_c1 | T | T | T | A | T | TT T A T |
| - NA19107_c2 | C | T | T | A | T | CT T A T |
| - NA19185_c1 | C | T | T | A | T | CT T A T |
| - NA19185_c2 | C | T | T | A | T | CT T A T |
| - NA19184_c1 | C | T | T | A | T | CT T A T |
| - NA19184_c2 | C | T | T | A | T | CT T A T |
| - NA19257_c1 | C | T | T | A | T | CT T A T |
| - NA19257_c2 | T | T | C | A | T | TT C A T |
| - NA19256_c1 | T | G | T | A | T | TG T A T |
| - NA19256_c2 | T | G | T | A | T | TG T A T |
| - NA19197_c1 | C | T | T | A | C | CT T A C |
| - NA19197_c2 | C | T | T | A | T | CT T A T |
| - NA19198_c1 | T | T | C | A | C | TT C A C |
| - NA19198_c2 | C | T | T | A | T | CT T A T |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA18873_c1 | T | G | T | A | T | TG T A T |
| - NA18873_c2 | T | T | C | A | C | TT C A C |
| - NA18874_c1 | C | T | T | A | C | CT T A C |
| - NA18874_c2 | C | T | T | A | T | CT T A T |
| - NA19201_c1 | C | T | T | A | T | CT T A T |
| - NA19201_c2 | C | T | T | A | T | CT T A T |
| - NA19200_c1 | C | T | T | A | T | CT T A T |
| - NA19200_c2 | C | T | T | A | T | CT T A T |
| - NA19238_c1 | T | T | C | A | C | TT C A C |
| - NA19238_c2 | T | G | T | A | T | TG T A T |
| - NA19239_c1 | C | T | T | A | T | CT T A T |
| - NA19239_c2 | T | T | C | A | C | TT C A C |
| - NA18505_c1 | C | T | T | A | T | CT T A T |
| - NA18505_c2 | C | T | T | A | T | CT T A T |
| - NA18504_c1 | C | T | T | A | T | CT T A T |
| - NA18504_c2 | C | T | T | A | T | CT T A T |
| - NA19222_c1 | T | G | T | A | T | TG T A T |
| - NA19222_c2 | C | T | T | A | T | CT T A T |
| - NA19223_c1 | C | T | T | A | T | CT T A T |
| - NA19223_c2 | C | T | T | A | T | CT T A T |
| - NA18861_c1 | C | T | T | A | T | CT T A T |
| - NA18861_c2 | T | T | C | A | C | TT C A C |
| - NA18862_c1 | T | T | C | A | T | TT C A T |
| - NA18862_c2 | C | T | T | A | T | CT T A T |
| - NA18852_c1 | T | T | T | A | T | TT T A T |
| - NA18852_c2 | T | T | C | A | C | TT C A C |
| - NA18853_c1 | T | T | C | A | T | TT C A T |
| - NA18853_c2 | T | T | C | A | C | TT C A C |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA18517_c1 | T | T | T | A | T | TT T A T |
| - NA18517_c2 | T | T | C | A | T | TT C A T |
| - NA18516_c1 | C | T | T | A | T | CT T A T |
| - NA18516_c2 | C | T | T | A | T | CT T A T |
| - NA19140_c1 | C | T | T | A | T | CT T A T |
| - NA19140_c2 | T | T | T | A | T | TT T A T |
| - NA19141_c1 | C | T | T | A | T | CT T A T |
| - NA19141_c2 | C | T | T | A | T | CT T A T |
| - NA19137_c1 | T | T | T | A | T | TT T A T |
| - NA19137_c2 | C | T | T | A | T | CT T A T |
| - NA19138_c1 | C | T | T | A | C | CT T A C |
| - NA19138_c2 | C | T | T | A | T | CT T A T |
| - NA19127_c1 | C | T | T | A | T | CT T A T |
| - NA19127_c2 | C | T | T | A | T | CT T A T |
| - NA19128_c1 | C | T | T | A | T | CT T A T |
| - NA19128_c2 | T | T | T | A | T | TT T A T |
| - NA19193_c1 | T | G | T | A | T | TG T A T |
| - NA19193_c2 | T | G | T | A | T | TG T A T |
| - NA19192_c1 | T | T | C | A | C | TT C A C |
| - NA19192_c2 | C | T | T | A | T | CT T A T |
| - NA19152_c1 | C | T | T | A | T | CT T A T |
| - NA19152_c2 | T | T | C | A | C | TT C A C |
| - NA19153_c1 | C | T | T | A | T | CT T A T |
| - NA19153_c2 | C | T | T | A | T | CT T A T |
| - NA19116_c1 | C | T | T | A | T | CT T A T |
| - NA19116_c2 | C | T | T | A | T | CT T A T |
| - NA19119_c1 | T | G | T | A | T | TG T A T |
| - NA19119_c2 | T | T | T | A | T | TT T A T |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA19209_c1 | C | T | T | A | T | CT T A T |
| - NA19209_c2 | T | T | C | A | C | TT C A C |
| - NA19210_c1 | C | T | T | A | T | CT T A T |
| - NA19210_c2 | T | T | T | A | T | TT T A T |
| - NA18870_c1 | C | T | T | A | T | CT T A T |
| - NA18870_c2 | C | T | T | A | C | CT T A C |
| - NA18871_c1 | C | T | T | A | T | CT T A T |
| - NA18871_c2 | C | T | T | A | T | CT T A T |
| - NA19131_c1 | C | T | T | A | T | CT T A T |
| - NA19131_c2 | C | T | T | A | C | CT T A C |
| - NA19130_c1 | C | T | T | A | T | CT T A T |
| - NA19130_c2 | T | T | C | A | T | TT C A T |
| - NA18508_c1 | C | T | T | A | T | CT T A T |
| - NA18508_c2 | C | T | T | A | T | CT T A T |
| - NA18507_c1 | T | T | C | A | C | TT C A C |
| - NA18507_c2 | C | T | T | A | T | CT T A T |
| - NA19206_c1 | C | T | T | A | T | CT T A T |
| - NA19206_c2 | T | T | C | A | C | TT C A C |
| - NA19207_c1 | T | T | C | A | T | TT C A T |
| - NA19207_c2 | T | G | T | A | T | TG T A T |
| - NA18912_c1 | C | T | T | A | T | CT T A T |
| - NA18912_c2 | C | T | T | A | T | CT T A T |
| - NA18913_c1 | C | T | T | A | T | CT T A T |
| - NA18913_c2 | T | G | T | A | T | TG T A T |
| - NA19159_c1 | T | T | C | A | T | TT C A T |
| - NA19159_c2 | T | G | T | A | T | TG T A T |
| - NA19160_c1 | C | T | T | A | T | CT T A T |
| - NA19160_c2 | C | T | T | A | T | CT T A T |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA19172_c1 | T | T | C | A | C | TT C A C |
| - NA19172_c2 | C | T | T | A | T | CT T A T |
| - NA19171_c1 | T | T | C | A | C | TT C A C |
| - NA19171_c2 | C | T | T | A | C | CT T A C |
| - NA18858_c1 | T | T | C | A | T | TT C A T |
| - NA18858_c2 | T | T | C | A | C | TT C A C |
| - NA18859_c1 | C | T | T | A | T | CT T A T |
| - NA18859_c2 | C | T | T | A | T | CT T A T |
| - NA19102_c1 | C | T | T | A | T | CT T A T |
| - NA19102_c2 | T | T | C | A | C | TT C A C |
| - NA19101_c1 | T | G | T | A | T | TG T A T |
| - NA19101_c2 | C | T | T | A | T | CT T A T |
| - NA18855_c1 | T | G | T | A | T | TG T A T |
| - NA18855_c2 | C | T | T | A | T | CT T A T |
| - NA18855_NA18856_c1 | C | T | T | A | T | CT T A T |
| - NA18855_NA18856_c2 | N | N | N | N | N | NN N N N |
| - NA18511_c1 | C | T | T | A | T | CT T A T |
| - NA18511_c2 | C | T | T | A | T | CT T A T |
| - NA18511_0_c1 | T | T | C | A | T | TT C A T |
| - NA18511_0_c2 | N | N | N | N | N | NN N N N |
| - NA19093_c1 | C | T | T | A | T | CT T A T |
| - NA19093_c2 | C | T | T | A | T | CT T A T |
| - NA19093_NA19092_c1 | T | T | C | A | T | TT C A T |
| - NA19093_NA19092_c2 | N | N | N | N | N | NN N N N |
| - NA18501_c1 | C | T | T | A | T | CT T A T |
| - NA18501_c2 | C | T | T | A | T | CT T A T |
| - NA18501_NA18502_c1 | T | T | T | A | T | TT T A T |
| - NA18501_NA18502_c2 | N | N | N | N | N | NN N N N |

| | | | | | | |
|---|---|---|---|---|---|---|
| - NA19175_c1 | T | T | C | A | C | TT C A C |
| - NA19175_c2 | T | T | T | A | T | TT T A T |
| - NA19176_c1 | C | T | T | A | T | CT T A T |
| - NA19176_c2 | C | T | T | A | T | CT T A T |
| - NA18510_c1 | C | T | T | A | T | CT T A T |
| - NA18510_c2 | T | T | T | A | T | TT T A T |
| - NA19144_c1 | T | T | C | A | T | TT C A T |
| - NA19144_c2 | C | T | T | A | T | CT T A T |
| - NA19203_c1 | C | T | T | A | T | CT T A T |
| - NA19203_c2 | C | T | T | A | T | CT T A T |
| - NA19143_c1 | T | T | C | A | C | TT C A C |
| - NA19143_c2 | C | T | T | A | T | CT T A T |
| - NA19204_c1 | T | T | T | A | T | TT T A T |
| - NA19204_c2 | C | T | T | A | T | CT T A T |
| - NA19098_c1 | C | T | T | A | T | CT T A T |
| - NA19098_c2 | T | T | C | A | C | TT C A C |
| - NA19099_c1 | T | T | C | A | T | TT C A T |
| - NA19099_c2 | C | T | T | A | T | CT T A T |

Table 14 Summary of all *TCF12* haplotypes across all ethnic populations available on the HapMap Project website

| ALL HAPMAP populations | | |
|---|---|---|
| | 1st count | 2nd count |
| NNNNN | 36 | 36 |
| **CGCAC** | 339 | 339 |
| **CGCAT** | 29 | 29 |
| **CGCCC** | 147 | 147 |
| **CGCCT** | 56 | 56 |
| **CTTAC** | 86 | 86 |
| **CTTAT** | 822 | 822 |
| **TTCAC** | 130 | 130 |
| **TTCAT** | 53 | 53 |
| **TTTAC** | 64 | 64 |
| **TTTAT** | 244 | 244 |
| **TGTAT** | 40 | 40 |
| **CGTAT** | 2 | 2 |
| **CTCAC** | 4 | 4 |
| **CTCAT** | 4 | 4 |
| **CTTCC** | 1 | 1 |
| **TGCAT** | 1 | 1 |
| | | |
| | 2058 | 2058 |

Table 15 *TCF12* haplotypes for control individuals from the UK (Control Plate 3)

| CP3 | SNP1 | | SNP 2 | | SNP 3 | | SNP 4 | | SNP 5 | |
|---|---|---|---|---|---|---|---|---|---|---|
| Plate position | rs447426 | | rs2585082 | | rs1628067 | | rs12901270 | | rs2440979 | |
| 1 | F | F | G | G | C | C | A | A | T | T |
| 2 | F | F | G | G | C | C | C | C | C | T |
| 3 | F | F | G | G | C | C | F | | C | T |
| 4 | F | F | G | T | C | T | A | C | C | T |
| 5 | F | F | G | T | T | T | F | | C | T |
| 6 | F | F | G | T | C | T | A | A | C | T |
| 7 | C | T | G | T | C | T | A | A | C | T |
| 8 | C | C | G | G | C | C | A | C | T | T |
| 9 | C | C | G | T | C | T | A | C | C | T |
| 10 | T | T | T | T | T | T | A | A | C | C |
| 11 | F | F | G | G | C | C | C | C | T | T |
| 12 | C | C | G | G | F | | C | C | C | C |
| 13 | C | T | G | T | C | T | A | C | C | T |
| 14 | C | C | G | G | C | C | A | A | T | T |
| 15 | C | T | G | T | C | T | A | A | C | C |
| 16 | C | T | G | T | C | T | A | C | C | T |
| 17 | C | T | G | T | C | T | A | A | C | T |
| 18 | C | C | G | T | C | T | A | C | C | C |
| 19 | C | T | T | T | C | T | A | A | C | C |
| 20 | C | C | G | T | C | T | A | A | C | T |
| 21 | C | T | G | T | C | T | A | A | C | T |
| 22 | C | T | T | T | C | T | A | A | C | C |
| 23 | C | C | G | G | C | C | C | C | C | T |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 24 | C | T | G | T | C | T | A | A | C | T |
| 25 | C | C | G | T | C | T | A | A | C | T |
| 26 | C | C | G | T | C | T | A | A | C | T |
| 27 | C | C | T | T | T | T | A | A | C | T |
| 28 | C | C | G | T | C | T | A | C | C | C |
| 29 | F | F | G | T | C | T | A | A | C | C |
| 30 | C | C | G | T | C | T | A | A | C | T |
| 31 | C | T | G | T | C | T | A | C | C | T |
| 32 | C | T | G | T | C | T | A | A | C | T |
| 33 | C | C | G | G | C | C | A | C | T | T |
| 34 | C | T | T | T | T | T | A | A | C | C |
| 35 | C | T | G | T | C | T | A | C | C | T |
| 36 | C | C | G | T | C | T | A | C | C | T |
| 37 | C | C | G | G | C | C | A | C | T | T |
| 38 | C | C | G | T | C | T | A | C | C | T |
| 39 | C | C | G | G | C | C | A | C | C | T |
| 40 | T | T | T | T | T | T | A | A | T | T |
| 41 | C | C | G | G | C | C | A | A | C | C |
| 42 | C | T | T | T | T | T | A | A | C | C |
| 43 | C | T | G | T | C | T | A | A | C | T |
| 44 | C | C | G | T | C | T | A | C | C | T |
| 45 | C | C | G | G | C | C | A | C | T | T |
| 46 | C | T | T | T | T | T | A | A | C | C |
| 47 | C | C | G | G | C | C | C | C | C | T |
| 48 | C | T | G | T | C | T | A | C | C | T |
| 49 | C | T | G | T | C | T | A | A | C | C |
| 50 | C | T | T | T | C | C | A | A | C | C |
| 51 | C | T | G | T | C | T | A | C | C | T |
| 52 | C | C | G | G | C | C | A | A | T | T |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 53 | C | C | G | T | C | T | A | A | T | T |
| 54 | C | T | G | T | C | T | A | C | T | T |
| 55 | C | C | G | G | C | C | A | C | C | T |
| 56 | C | C | G | T | C | T | A | C | C | T |
| 57 | C | C | G | T | C | T | A | A | C | T |
| 58 | C | C | T | T | T | T | A | A | C | T |
| 59 | C | C | G | G | C | C | C | C | C | T |
| 60 | C | C | G | G | C | C | A | A | C | T |
| 61 | C | T | G | T | C | T | A | C | C | T |
| 62 | C | C | G | G | C | C | A | C | T | T |
| 63 | C | T | T | T | T | T | A | A | C | C |
| 64 | C | C | G | G | C | C | A | A | C | T |
| 65 | C | C | G | G | C | C | A | C | T | T |
| 66 | T | T | T | T | T | T | A | A | C | C |
| 67 | T | T | T | T | T | T | A | A | C | T |
| 68 | C | C | G | G | C | C | A | C | T | T |
| 69 | C | C | G | G | C | C | C | C | C | T |
| 70 | T | T | G | T | C | T | A | C | C | T |
| 71 | C | T | G | T | C | T | A | A | C | T |
| 72 | C | T | T | T | T | T | A | A | C | C |
| 73 | C | T | G | T | C | T | A | C | C | C |
| 74 | C | T | G | T | C | T | A | A | C | T |
| 75 | T | T | T | T | C | T | A | A | C | T |
| 76 | C | C | G | G | C | C | A | A | T | T |
| 77 | C | C | G | G | C | C | A | A | T | T |
| 78 | C | T | G | T | C | T | A | C | C | C |
| 79 | C | T | G | T | C | T | A | A | C | T |
| 80 | C | T | G | T | C | T | A | C | T | T |
| 81 | C | T | G | T | C | T | A | A | C | C |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 82 | C | C | G | T | C | T | A | A | C | T |
| 83 | C | T | T | T | T | T | A | A | C | C |
| 84 | C | C | G | G | C | C | A | A | T | T |
| 85 | C | T | T | T | C | T | A | A | C | C |
| 86 | C | C | G | T | C | T | A | A | C | T |
| 87 | C | C | G | G | C | C | C | C | T | T |
| 88 | C | C | G | T | C | T | A | A | C | T |
| 89 | C | C | G | G | C | C | A | C | T | T |
| 90 | C | T | G | T | C | T | A | A | C | T |
| 91 | C | T | G | T | C | T | A | C | C | T |
| 92 | C | C | G | G | C | C | C | C | T | T |
| 93 | C | T | G | T | C | T | A | C | C | T |
| 94 | C | T | G | T | C | T | A | C | T | T |
| 95 | C | C | G | G | C | C | C | C | T | T |
| 96 | C | T | G | T | C | T | A | A | C | C |

Table 16 Proportion of each genotype for a control UK population (Control plate 3)

| CP3 | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | | | | | |
| 1 | | | 2 | | | 3 | | | 4 | | | 5 | | |
| CC | 44 | | GG | 30 | | CC | 30 | | AA | 51 | | TT | 23 | |
| CT | 38 | | GT | 49 | | CT | 52 | | AC | 33 | | CT | 50 | |
| TT | 6 | | TT | 17 | | TT | 13 | | CC | 10 | | CC | 23 | |
| C | 126 | 0.715 | G | 109 | 0.545 | C | 112 | 0.589 | A | 135 | 0.718 | C | 96 | 0.5 |
| T | 50 | 0.284 | T | 83 | 0.455 | T | 78 | 0.411 | C | 53 | 0.282 | T | 96 | 0.5 |
| | 176 | | | 192 | | | 190 | | | 188 | | | 192 | |

Table 17 *TCF12* haplotypes for control individuals from the UK (Control Plate 4)

| Plate position | rs447426 | | rs2585082 | | rs1628067 | | rs12901270 | | rs2440979 | |
|---|---|---|---|---|---|---|---|---|---|---|
| 1 | F | F | G | G | C | C | A | A | T | T |
| 2 | F | F | G | G | C | C | C | C | C | T |
| 3 | F | F | G | G | C | C | F | F | C | T |
| 4 | F | F | G | T | C | T | A | C | C | T |
| 5 | F | F | G | T | T | T | F | F | C | T |
| 6 | F | F | G | T | C | T | A | A | C | T |
| 7 | C | T | G | T | C | T | A | A | C | T |
| 8 | C | C | G | G | C | C | A | C | T | T |
| 9 | C | C | G | T | C | T | A | C | C | T |
| 10 | T | T | T | T | T | T | A | A | C | C |
| 11 | F | F | G | G | C | C | C | C | T | T |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 12 | C | C | G | G | F | F | C | C | C | C |
| 13 | C | T | G | T | C | T | A | C | C | T |
| 14 | C | C | G | G | C | C | A | A | T | T |
| 15 | C | T | G | T | C | T | A | A | C | C |
| 16 | C | T | G | T | C | T | A | C | C | T |
| 17 | C | T | G | T | C | T | A | A | C | T |
| 18 | C | C | G | T | C | T | A | C | C | C |
| 19 | C | T | T | T | C | T | A | A | C | C |
| 20 | C | C | G | T | C | T | A | A | C | T |
| 21 | C | T | G | T | C | T | A | A | C | T |
| 22 | C | T | T | T | C | T | A | A | C | C |
| 23 | C | C | G | G | C | C | C | C | C | T |
| 24 | C | T | G | T | C | T | A | A | C | T |
| 25 | C | C | G | T | C | T | A | A | C | T |
| 26 | C | C | G | T | C | T | A | A | C | T |
| 27 | C | C | T | T | T | T | A | A | C | T |
| 28 | C | C | G | T | C | T | A | C | C | C |
| 29 | F | F | G | T | C | T | A | A | C | C |
| 30 | C | C | G | T | C | T | A | A | C | T |
| 31 | C | T | G | T | C | T | A | C | C | T |
| 32 | C | T | G | T | C | T | A | A | C | T |
| 33 | C | C | G | G | C | C | A | C | T | T |
| 34 | C | T | T | T | T | T | A | A | C | C |
| 35 | C | T | G | T | C | T | A | C | C | T |
| 36 | C | C | G | T | C | T | A | C | C | T |
| 37 | C | C | G | G | C | C | A | C | T | T |
| 38 | C | C | G | T | C | T | A | C | C | T |
| 39 | C | C | G | G | C | C | A | C | C | T |
| 40 | T | T | T | T | T | T | A | A | T | T |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 41 | C | C | G | G | C | C | A | A | C | C |
| 42 | C | T | T | T | T | T | A | A | C | C |
| 43 | C | T | G | T | C | T | A | A | C | T |
| 44 | C | C | G | T | C | T | A | C | C | T |
| 45 | C | C | G | G | C | C | A | C | T | T |
| 46 | C | T | T | T | T | T | A | A | C | C |
| 47 | C | C | G | G | C | C | C | C | C | T |
| 48 | C | T | G | T | C | T | A | C | C | T |
| 49 | C | T | G | T | C | T | A | A | C | C |
| 50 | C | T | T | T | C | C | A | A | C | C |
| 51 | C | T | G | T | C | T | A | C | C | T |
| 52 | C | C | G | G | C | C | A | A | T | T |
| 53 | C | C | G | T | C | T | A | A | T | T |
| 54 | C | T | G | T | C | T | A | C | T | T |
| 55 | C | C | G | G | C | C | A | C | C | T |
| 56 | C | C | G | T | C | T | A | C | C | T |
| 57 | C | C | G | T | C | T | A | A | C | T |
| 58 | C | C | T | T | T | T | A | A | C | T |
| 59 | C | C | G | G | C | C | C | C | C | T |
| 60 | C | C | G | G | C | C | A | A | C | T |
| 61 | C | T | G | T | C | T | A | C | C | T |
| 62 | C | C | G | G | C | C | A | C | T | T |
| 63 | C | T | T | T | T | T | A | A | C | C |
| 64 | C | C | G | G | C | C | A | A | C | T |
| 65 | C | C | G | G | C | C | A | C | T | T |
| 66 | T | T | T | T | T | T | A | A | C | C |
| 67 | T | T | T | T | T | T | A | A | C | T |
| 68 | C | C | G | G | C | C | A | C | T | T |
| 69 | C | C | G | G | C | C | C | C | C | T |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 70 | T | T | G | T | C | T | A | C | C | T |
| 71 | C | T | G | T | C | T | A | A | C | T |
| 72 | C | T | T | T | T | T | A | A | C | C |
| 73 | C | T | G | T | C | T | A | C | C | C |
| 74 | C | T | G | T | C | T | A | A | C | T |
| 75 | T | T | T | T | C | T | A | A | C | T |
| 76 | C | C | G | G | C | C | A | A | T | T |
| 77 | C | C | G | G | C | C | A | A | T | T |
| 78 | C | T | G | T | C | T | A | C | C | C |
| 79 | C | T | G | T | C | T | A | A | C | T |
| 80 | C | T | G | T | C | T | A | C | T | T |
| 81 | C | T | G | T | C | T | A | A | C | C |
| 82 | C | C | G | T | C | T | A | A | C | T |
| 83 | C | T | T | T | T | T | A | A | C | C |
| 84 | C | C | G | G | C | C | A | A | T | T |
| 85 | C | T | T | T | C | T | A | A | C | C |
| 86 | C | C | G | T | C | T | A | A | C | T |
| 87 | C | C | G | G | C | C | C | C | T | T |
| 88 | C | C | G | T | C | T | A | A | C | T |
| 89 | C | C | G | G | C | C | A | C | T | T |
| 90 | C | T | G | T | C | T | A | A | C | T |
| 91 | C | T | G | T | C | T | A | C | C | T |
| 92 | C | C | G | G | C | C | C | C | T | T |
| 93 | C | T | G | T | C | T | A | C | C | T |
| 94 | C | T | G | T | C | T | A | C | T | T |
| 95 | C | C | G | G | C | C | C | C | T | T |
| 96 | C | T | G | T | C | T | A | A | C | C |

Table 18 Proportion of each genotype for a control UK population (Control plate 4)

| 1 | | | 2 | | | 3 | | | 4 | | | 5 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| CC | 49 | | GG | 27 | | CC | 31 | | AA | 54 | | CC | 24 | |
| CT | 38 | | GT | 43 | | CT | 59 | | AC | 31 | | CT | 47 | |
| TT | 9 | | TT | 25 | | TT | 25 | | CC | 6 | | TT | 25 | |
| C | 136 | 0.708 | G | 97 | 0.511 | C | 102 | 0.534 | A | 139 | 0.764 | C | 95 | 0.495 |
| T | 56 | 0.292 | T | 93 | 0.489 | T | 89 | 0.466 | C | 43 | 0.236 | T | 97 | 0.505 |
| | 192 | | | 190 | | | 191 | | | 182 | | | 192 | |

Table 19 Summary of haplotype results for all 48 families

| Family # | Individual | | | | | | Family # | Individual | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | III-1 | II-1 | II-2 | I-1 | I-2 | | 25 | II-1 | I-1 | I-2 | | | |
| | CTTAC | N/A | CTTAC | CGCAC | CTTAT | | | CGCCC | CGCAC | CGCCC | | | |
| | TTTAC | N/A | TTTAT | CTTAC | TTTAT | | | CGCCC | CGCCC | CTTAT | | | |
| 2 | II-1 | I-1 | I-2 | II-2 | I-3 | I-4 | 26 | II-1 | I-1 | I-2 | | | |
| | CGCAC | CGCAC | CGCAC | CGCAC | CGCAC | CGCAC | | CGCCC | CGCCC | CGCAC | | | |
| | CGCAC | CGCAC | CGCAC | CGCAC | CGCAC | CGCAC | | CGCAC | CGCCC | TTTCT | | | |
| 3 | II-2 | II-1 | I-1 | I-2 | | | 27 | IV-1 | III-1 | III-2 | II-1 | II-2 | I-1 | I-2 |

| | | | | | |
|---|---|---|---|---|---|
| | CTTAT | CTTAT | TTTAC | CTTAT | |
| | TTTAC | TTTAC | CGCCC | CGCAC | |
| 4 | III-1 | II-1 | II-2 | I-1 | I-2 |
| | CGCAC | N/A | N/A | N/A | N/A |
| | CGCAC | N/A | N/A | N/A | N/A |
| 5 | II-1 | I-1 | I-2 | | |
| | CTTAC | CTTAC* | CTTAT | | |
| | TTTAT | CTTAC | TTTAT | | |
| 6 | II-1 | I-1 | I-2 | | |
| | CGCAC | CGCAC | CGCCT | | |
| | CTTAT | CGCCC | CTTAT | | |
| 7 | II-1 | I-1 | I-2 | | |
| | CGCCC | CGCCC* | CGCAC | | |
| | TTCAC | CGCCC | TTCAC | | |
| 8 | II-1 | I-1 | I-2 | | |
| | CGCAC | CGCAT | CGCAC | | |
| | CGCAT | CTTAT | CTTAT | | |
| 9 | II-1 | II-2 | I-1 | I-2 | |
| | CGCCC | CTTAT | CTTAT | CGCCC | |
| | CGCAT | CTTAT | CGCAT | CTTAT | |
| 10 | II-1 | I-2 | I-3 | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | CGCCT | N/A | CGCAC | N/A | TTTAT | N/A | CGCAC |
| | TTTAT | N/A | TTTAT | N/A | CGCAC | N/A | TTTAC |
| 28 | III-1 | II-1 | II-2 | I-1 | I-2 | | |
| | CGCCC | CGCAC | CGCAC | CGCAC | CGCCC* | | |
| | TTTAT | CGCCC | TTTAT | TTTAT | TTTAT | | |
| 29 | II-1 | I-1 | I-2 | | | | |
| | TTTAT | CTTAT | CGCCC | | | | |
| | CGCCC | TTTAT* | TTTAT | | | | |
| 30 | II-1 | I-1 | I-2 | | | | |
| | CGCAC | CGCAC | CGCAC | | | | |
| | TTTAT | CGCAC | TTTAT | | | | |
| 31 | II-1 | II-2 | I-1 | I-2 | | | |
| | CGCAC | N/A | CGCAC | N/A | | | |
| | CGCAC | N/A | TTTAT | N/A | | | |
| 32 | II-1 | I-1 | I-2 | | | | |
| | CGCAC | TTTAT | CGCAC | | | | |
| | TTTAT | TTTAT* | CGCAC | | | | |
| 33 | II-1 | I-1 | I-2 | | | | |
| | CGCCC | CGCCC | CGCAC | | | | |
| | CGCAC | TTCAT | CGCCT | | | | |
| 34 | III-1 | III-2 | III-3 | II-1 | II-2 | I-1 | I-2 |

| | | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | TTTAT | TTTAT | TTTAC | | | | | CGCCC | CGCAC | CGCCC | CGCCC | CGCAC | N/A | CGCCC |
| | TTTAC | TTTAT | CGCAC | | | | | CGCCC | TTTAT | TTTAT | TTTAT | CGCCC | N/A | CTTAC |
| 11 | III-1 | II-1 | II-2 | II-3 | I-1 | I-2 | | 35 | III-1 | III-2 | II-1 | II-2 | I-1 | I-2 | | |
| | CGCAC | CGCAC | CGCCT | CGCAC | CGCCC | CGCAC | | | TTTAC | TTTAC | CTTAT | CGCCT | CGCCT | CTTAT | | |
| | CGCCT | CGCCC | CGCAC | CGCCC | TTTAC | CGCCC | | | TTTAC | TTTAC | TTTAC | TTTAC | CTTAT | TTTAC* | | |
| 12 | II-1 | I-1 | I-2 | | | | | 36 | II-1 | I-1 | I-2 | | | | | |
| | CGCAC | N/A | CGCAC | | | | | | CGCAC | CGCAC | CTTAT | | | | | |
| | CTTAC | N/A | TGTAT | | | | | | TTTAT | CGCAC | TTTAT | | | | | |
| 13 | II-1 | I-1 | I-2 | | | | | 37 | II-1 | I-1 | I-2 | | | | | |
| | CTTAT | CTTAT | CTTAT | | | | | | CGTCT | CGTCT | TTCAC | | | | | |
| | CTTAT | CTTAT | CGCCC | | | | | | CTTAT | CTCAC | CTTAT | | | | | |
| 14 | II-1 | II-2 | I-1 | I-2 | | | | 38 | II-1 | I-1 | I-2 | | | | | |
| | CGCAT | CGCAT | CGCAT | CGCAC | | | | | CGCCC | CGCCC | CGCCC | | | | | |
| | CGCAC | CGCAC | CTTAT | TTTAT | | | | | CGCCT | CGCCT | CTTAT | | | | | |
| 15 | II-1 | I-1 | I-2 | | | | | 39 | II-1 | I-1 | I-2 | | | | | |
| | TTTAT | TTTAT | CGCAC | | | | | | CGCAC | CGCAC | CGCAC | | | | | |
| | CGCAC | CTTAT | CTTAT | | | | | | TTTAT | TTTAT | CGCCC | | | | | |
| 16 | II-1 | I-1 | I-2 | | | | | 40 | II-1 | I-1 | I-2 | | | | | |
| | CGCCC | CGCCT | GCCCC | | | | | | CGCAC | N/A | CGCAC | | | | | |
| | CGCCT | TTTAT | CGCAC | | | | | | TTTAT | N/A | TTTAT | | | | | |
| | | | | | | | | | | | | | | | | |

| 17 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCAC | CGCAC | CGCAC |
| | CGCAC | CGCAC | TTTAT |

| 18 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCCC | CGCCC | CGCCC |
| | CGCCC | CGCCC | CTTAT |

| 19 | III-1 | II-1 | II-2 | I-1 | I-2 |
|---|---|---|---|---|---|
| | TTTAT | CGCCT | TTTAT | N/A | CGCCC |
| | CGCCT | CTTAT | CGCCC | N/A | TTTAC |

| 20 | II-1 | II-2 | I-1 | I-2 |
|---|---|---|---|---|
| | CGCAC | CGCAC | TTTAT | CGCAC |
| | TTTAT | TTTAT | TTTAT | CTTAC |

| 21 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CTTAT | CTTAT* | CGCAT |
| | CGCAT | CTCAT | CGCAC |

| 22 | II-3 | II-2 | II-3 |
|---|---|---|---|
| | TTTAT | TTTAT | CGCCC |
| | CGCCC | CTTAT | CGCAC |

| 23 | III-1 | II-1 | II-2 | I-1 | I-2 |
|---|---|---|---|---|---|
| | CGCCT | CGCCC | CTTAT | N/A | CGCCC |
| | TTTAT | CGCCT | TTTAT | N/A | TTTAT |

| 41 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCCC | CTTAT | CGCAC |
| | TTTAT | TTTAT* | CGCCC |

| 42 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCAC | CGCAC | CTTAT |
| | TTTAT | TTTAT | TTTAT |

| 43 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CTTAT | N/A | N/A |
| | CTTAT | N/A | N/A |

| 44 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCCC | CTTAC | CGCCC |
| | CTTAT | CTTAT | CTTAT |

| 45 | II-1 | II-2 | II-3 | II-4 | II-5 | II-6 | I-1 | I-2 |
|---|---|---|---|---|---|---|---|---|
| | CGCAC | N/A | N/A | N/A | N/A | CGCAC | CGCAT | CGCAC |
| | CGCAT | N/A | N/A | N/A | N/A | CGCAT | CTTAC | TTCAT |

| 46 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCCC | N/A | CGCCC |
| | CGCCC | N/A | CTTAT |

| 47 | II-1 | I-1 | I-2 |
|---|---|---|---|
| | CGCCC | CGCCC | CGCAC |
| | CTTAT | CGCAC | CTTAT |

| 24 | II-1 | II-2 | II-3 | I-1 | I-2 | I-3 | 48 | II-1 | I-1 | I-2 |
|---|---|---|---|---|---|---|---|---|---|---|
| | TTTAT | CGCCC | CGCCC | N/A | TTTAT | N/A | | CGCAC | CGCAT | CGCAC |
| | CGCAC | TTTAT | CGCCC | N/A | CGCCC | N/A | | CGCAT | CTTAT | CGCCC |

Table 20.1 Genotyping results of mutant *TCF12* alleles in 46 families – excluding uninformative cases

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | C | T | T | A | C |
| 2 | C | G | C | A | C |
| 3 | T | T | T | A | C |
| 5 | C | T | T | A | C |
| 6 | | | | | |
| 7 | C | G | C | C | C |
| 8 | C | G | C | A | T |
| 9 | C | G | C | A | T |
| 10 | T | T | T | A | T |
| 11 | C | G | C | A | C |
| 12 | C | G | C | A | C |
| 13 | C | T | T | A | T |
| 14 | C | G | C | A | T |
| 15 | T | T | T | A | T |
| 16 | | | | | |
| 17 | C | G | C | A | C |
| 18 | C | G | C | C | C |
| 19 | T | T | T | A | T |

| | | | | | |
|---|---|---|---|---|---|
| 20 | C | G | C | A | C |
| 21 | C | T | T | A | T |
| 22 | T | T | T | A | T |
| 23 | C | G | C | C | T |
| 24 | T | T | T | A | T |
| 25 | C | G | C | C | C |
| 26 | | | | | |
| 27 | T | T | T | A | T |
| 28 | T | T | T | A | T |
| 29 | T | T | T | A | T |
| 30 | T | T | T | A | T |
| 31 | C | G | C | A | C |
| 32 | T | T | T | A | T |
| 33 | | | | | |
| 34 | T | T | T | A | T |
| 35 | T | T | T | A | C |
| 36 | C | G | C | A | C |
| 37 | C | T | T | A | T |
| 38 | | | | | |
| 39 | T | T | T | A | T |
| 41 | T | T | T | A | T |
| 42 | | | | | |
| 43 | C | T | T | A | T |
| 44 | C | G | C | C | C |
| 45 | C | G | C | A | C |
| 46 | C | G | C | C | C |
| 47 | C | G | C | C | C |
| 48 | C | G | C | A | T |

| | | | | | |
|---|---|---|---|---|---|
| ALLELE 1 | 25 | 21 | 19 | 33 | 22 |
| ALLELE 2 | 15 | 19 | 21 | 7 | 18 |
| n= 40 | 40 | 40 | 40 | 40 | 40 |

Table 20.2 Proportions of mutant *TCF12* alleles in 46 families – excluding uninformative cases

| MUTANT ALLELE | | |
|---|---|---|
| SNP 1 rs 4774246(C>T) | Numbers | Proportions |
| C | 25 | 0.625 |
| T | 15 | 0.375 |
| | | |
| SNP 2 rs2585082 (T>G) | | |
| T | 21 | 0.525 |
| G | 19 | 0.475 |
| | | |
| SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| C | 19 | 0.475 |
| T | 21 | 0.525 |
| | | |
| SNP 4 rs12901270(A>C) | Numbers | Proportions |
| A | 33 | 0.825 |

| | | |
|---|---|---|
| C | 7 | 0.175 |
| | | |
| SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| T | 22 | 0.55 |
| C | 18 | 0.45 |

Table 20.3 Fisher's exact test of mutant *TCF12* alleles in 46 families – excluding uninformative cases

| | | | |
|---|---|---|---|
| | rs4774246 | | |
| | T | C | |
| Gp1 | 15 | 25 | 40 |
| Gp2 | 106 | 262 | 368 |
| | 121 | 287 | 408 |
| 2-tailed p value | 0.2755 | **Not sig** | |
| | rs2585082 (T/G) | | |
| | T | G | |
| Gp1 | 21 | 19 | 40 |
| Gp2 | 176 | 206 | 382 |
| | 197 | 225 | 422 |
| | | | |

| | | | |
|---|---|---|---|
| 2 tailed p value | 0.5062 | **Not sig** | |
| | SNP 3 rs1628067 (C>T) | | |
| | T | C | |
| Gp1 | 21 | 19 | 40 |
| Gp2 | 167 | 214 | 381 |
| | 188 | 233 | 421 |
| | | | |
| | | | |
| 2 tailed p value | 0.3188 | **Not sig** | |
| | rs12901270 (A/C) | | |
| | A | C | |
| Gp1 | 33 | 7 | 40 |
| Gp2 | 274 | 96 | 370 |
| | 307 | 103 | 410 |
| 2 tailed p value | 0.3367 | **Not sig** | |
| | SNP 5 rs2440979 (T>C) | | |

| | T | C | |
|---|---|---|---|
| Gp1 | 22 | 18 | 40 |
| Gp2 | 193 | 191 | 384 |
| | 215 | 209 | 424 |
| 2 tailed p value | 0.6202 | **Not sig** | |

Table 21.1 Numbers of mutant *TCF12* alleles in 46 families –including uninformative cases where mutant and partner alleles apportioned 50:50

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | C | T | T | A | C |
| 2 | C | G | C | A | C |
| 3 | T | T | T | A | C |
| 5 | C | T | T | A | C |
| 6 | *C* | *G* | *C* | *A* | *C* |
| 7 | C | G | C | C | C |
| 8 | C | G | C | A | T |

| | | | | | |
|---|---|---|---|---|---|
| 9 | C | G | C | A | T |
| 10 | T | T | T | A | T |
| 11 | C | G | C | A | C |
| 12 | C | G | C | A | C |
| 13 | C | T | T | A | T |
| 14 | C | G | C | A | T |
| 15 | T | T | T | A | T |
| 16 | *C* | *G* | *C* | *C* | *T* |
| 17 | C | G | C | A | C |
| 18 | C | G | C | C | C |
| 19 | T | T | T | A | T |
| 20 | C | G | C | A | C |
| 21 | C | T | T | A | T |
| 22 | T | T | T | A | T |
| 23 | C | G | C | C | T |
| 24 | T | T | T | A | T |
| 25 | C | G | C | C | C |
| 26 | *C* | *G* | *C* | *C* | *C* |
| 27 | T | T | T | A | T |

| | | | | | |
|---|---|---|---|---|---|
| 28 | T | T | T | A | T |
| 29 | T | T | T | A | T |
| 30 | T | T | T | A | T |
| 31 | C | G | C | A | C |
| 32 | T | T | T | A | T |
| 33 | *C* | *G* | *C* | *C* | *C* |
| 34 | T | T | T | A | T |
| 35 | T | T | T | A | C |
| 36 | C | C | C | A | C |
| 37 | C | T | T | A | T |
| 38 | *C* | *G* | *C* | *C* | *C* |
| 39 | T | T | T | A | T |
| 41 | T | T | T | A | T |
| 42 | *C* | *G* | *C* | *A* | *C* |
| 43 | C | T | T | A | T |
| 44 | C | G | C | C | C |
| 45 | C | G | C | A | C |
| 46 | C | G | C | C | C |
| 47 | C | G | C | C | C |

| | | | | | |
|---|---|---|---|---|---|
| 48 | C | G | C | A | T |
| ALLELE 1 | 28 | 21 | 22 | 34 | 23 |
| ALLELE 2 | 15 | 22 | 21 | 9 | 20 |
| n= 46 | 43 | 43 | 43 | 43 | 43 |

Table 21.2 Proportions of mutant *TCF12* alleles in 46 families – including uninformative cases (italicised) where mutant and partner alleles apportioned 50:50

| SNP 1 rs 4774246(C>T) | Numbers | Proportions |
|---|---|---|
| C | 28 | 0.651162791 |
| T | 15 | 0.348837209 |
| | | |
| SNP 2 rs2585082 (T>G) | | |
| T | 21 | 0.488372093 |
| G | 22 | 0.511627907 |
| | | |
| SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| C | 22 | 0.511627907 |
| T | 21 | 0.488372093 |
| | | |

| SNP 4 rs12901270(A>C) | Numbers | Proportions |
|---|---|---|
| A | 34 | 0.790697674 |
| C | 9 | 0.209302326 |
| | | |
| SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| T | 23 | 0.534883721 |
| C | 20 | 0.465116279 |

Table 21.3 Fisher's exact test for mutant *TCF12* alleles in 46 families – including uninformative cases where mutant and partner alleles apportioned 50:50

| | rs4774246 | | |
|---|---|---|---|
| | T | C | |
| Gp1 | 15 | 28 | 43 |
| Gp2 | 106 | 262 | 368 |
| | 121 | 290 | 411 |
| 2-tailed p value | 0.4793 | **Not sig** | |
| | rs2585082 (T/G) | | |
| | T | G | |
| Gp1 | 21 | 22 | 43 |
| Gp2 | 176 | 206 | 382 |
| | 197 | 228 | 425 |
| | | | |
| 2 tailed p value | 0.7494 | **Not sig** | |
| | | | |
| | T | C | |

| | | | |
|---|---|---|---|
| Gp1 | 21 | 22 | 43 |
| Gp2 | 167 | 214 | 381 |
| | 188 | 236 | 424 |
| | | | |
| | | | |
| 2 tailed p value | 0.6276 | **Not sig** | |
| | rs12901270 (A/C) | | |
| | A | C | |
| Gp1 | 34 | 9 | 43 |
| Gp2 | 274 | 96 | 370 |
| | 308 | 105 | 413 |
| 2 tailed p value | 0.5803 | **Not sig** | |
| | | | |
| | T | C | |
| Gp1 | 23 | 20 | 43 |
| Gp2 | 193 | 191 | 384 |
| | 216 | 211 | 427 |
| 2 tailed p value | 0.7489 | **Not sig** | |
| | | | |
| Decimal places rounded up to nearest whole number for statistical analysis | | | |

Table 22.1 Genotyping results of partner *TCF12* alleles in the presence of craniosynostosis in 46 families – excluding uninformative cases

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | T | T | T | A | C |
| 2 | C | G | C | A | C |
| 3 | C | T | T | A | T |
| 3 | C | G | C | C | C |
| 5 | T | T | T | A | T |
| 6 | | | | | |
| 7 | T | T | C | A | C |
| 8 | C | T | T | A | T |
| 8 | C | G | C | A | C |
| 9 | C | G | C | C | C |
| 10 | T | T | T | A | C |
| 11 | C | G | C | C | C |
| 11 | C | G | C | C | T |
| 12 | C | T | T | A | C |

| | | | | | |
|---|---|---|---|---|---|
| 13 | C | T | T | A | T |
| 13 | C | G | C | C | C |
| 14 | C | G | C | A | C |
| 15 | C | G | C | A | C |
| 16 | | | | | |
| 17 | C | G | C | A | C |
| 18 | C | G | C | C | C |
| 19 | C | G | C | C | T |
| 20 | T | T | T | A | T |
| 21 | C | G | C | A | T |
| 22 | C | T | T | A | T |
| 23 | T | T | T | A | T |
| 24 | C | G | C | A | C |
| 25 | C | G | C | C | C |
| 26 | | | | | |
| 27 | C | G | C | A | C |
| 27 | C | G | C | C | T |
| 28 | C | G | C | C | C |
| 29 | C | G | C | C | C |

| | | | | | |
|---|---|---|---|---|---|
| 30 | C | G | C | A | C |
| 31 | C | G | C | A | C |
| 31 | T | T | T | A | T |
| 32 | C | G | C | A | C |
| 33 | | | | | |
| 34 | C | G | C | C | C |
| 34 | C | G | C | A | C |
| 35 | T | T | T | A | C |
| 35 | C | G | C | C | T |
| 36 | T | T | T | A | T |
| 37 | C | G | T | C | T |
| 37 | T | T | C | A | C |
| 38 | | | | | |
| 39 | C | G | C | A | C |
| 41 | C | G | C | C | C |
| 42 | | | | | |
| 43 | C | T | T | A | T |
| 44 | C | T | T | A | T |
| 45 | C | G | C | A | T |

| | | | | | |
|---|---|---|---|---|---|
| 46 | C | G | C | C | C |
| 47 | C | T | T | A | T |
| 48 | C | G | C | A | C |
| Allele 1 | 39 | 18 | 32 | 33 | 19 |
| Allele 2 | 10 | 31 | 17 | 16 | 30 |
| n = 48 | 49 | 49 | 49 | 49 | 49 |

Table 22.2 Proportions of partner *TCF12* alleles in 46 families

| SNP 1 rs 4774246(C>T) | Numbers | Proportions |
|---|---|---|
| C | 39 | 0.795918367 |
| T | 10 | 0.204081633 |
| | | |
| SNP 2 rs2585082 | | |
| T | 18 | 0.367346939 |
| G | 31 | 0.632653061 |
| | | |
| SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| C | 32 | 0.653061224 |
| T | 17 | 0.346938776 |
| | | |

| SNP 4 rs12901270(A>C) | Numbers | Proportions |
|---|---|---|
| A | 33 | 0.673469388 |
| C | 16 | 0.326530612 |
| | | |
| SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| T | 19 | 0.387755102 |
| C | 30 | 0.612244898 |

Table 22.3 Fisher's exact test for partner *TCF12* alleles in 46 families

| | rs4774246 | | |
|---|---|---|---|
| | T | C | |
| Gp1 | 10 | 39 | 49 |
| Gp2 | 106 | 262 | 368 |
| | 116 | 301 | 417 |
| 2-tailed p value | 0.2398 | **not sig** | |
| | rs2585082 (T/G) | | |
| | T | G | |
| Gp1 | 18 | 31 | 49 |
| Gp2 | 176 | 206 | 382 |
| | 194 | 237 | |
| 2 tailed p value | 0.2268 | **Not sig** | |
| | rs1628067 (T/C) | | |
| | T | C | |
| Gp1 | 17 | 32 | 49 |

| | | | |
|---|---|---|---|
| Gp2 | 167 | 214 | 381 |
| | 184 | 246 | |
| 2 tailed p value | 0.283 | **Not sig** | |
| | rs12901270 (A/C) | | |
| | A | C | |
| Gp1 | 33 | 16 | 49 |
| Gp2 | 274 | 96 | 370 |
| | 307 | 112 | |
| 2 tailed p value | 0.3086 | **Not sig** | |
| | rs2440979 (T/C) | | |
| | T | C | |
| Gp1 | 19 | 30 | 49 |
| Gp2 | 193 | 191 | 384 |
| | 212 | 221 | |
| 2 tailed p value | 0.1716 | **Not sig** | |

Table 23.1 Genotyping results of partner *TCF12* alleles in 46 families– including uninformative cases

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | T | T | T | A | C |
| 2 | C | G | C | A | C |
| 3 | C | T | T | A | T |
| 3 | C | G | C | C | C |
| 5 | T | T | T | A | T |
| 6 | *C* | *T* | *T* | *A* | *T* |
| 7 | T | T | C | A | C |
| 8 | C | G | C | A | C |
| 9 | C | G | C | C | C |
| 10 | T | T | T | A | C |
| 11 | C | G | C | C | C |
| 11 | C | G | C | C | T |
| 12 | C | T | T | A | C |
| 13 | C | T | T | A | T |
| 13 | C | G | C | C | C |
| 14 | C | G | C | A | C |

| | | | | | |
|---|---|---|---|---|---|
| 15 | C | G | C | A | C |
| 16 | *C* | *G* | *C* | *C* | *C* |
| 17 | C | G | C | A | C |
| 18 | C | G | C | C | C |
| 19 | C | G | C | C | T |
| 20 | T | T | T | A | T |
| 21 | C | G | C | A | T |
| 22 | C | T | T | A | T |
| 23 | T | T | T | A | T |
| 24 | C | G | C | A | C |
| 25 | C | G | C | C | C |
| 26 | *C* | *G* | *C* | *A* | *C* |
| 27 | C | G | C | A | C |
| 27 | C | G | C | C | T |
| 28 | C | G | C | C | C |
| 29 | C | G | C | C | C |
| 30 | C | G | C | A | C |
| 31 | C | G | C | A | C |
| 31 | T | T | T | A | T |

| | | | | | |
|---|---|---|---|---|---|
| 32 | C | G | C | A | C |
| 33 | *C* | *G* | *C* | *A* | *C* |
| 34 | C | G | C | C | C |
| 34 | C | G | C | A | C |
| 35 | T | T | T | A | C |
| 35 | C | G | C | C | T |
| 36 | T | T | T | A | T |
| 37 | C | G | T | C | T |
| 37 | T | T | C | A | C |
| 38 | *C* | *G* | *C* | *C* | *C* |
| 39 | C | G | C | A | C |
| 41 | C | G | C | C | C |
| 42 | *T* | *T* | *T* | *A* | *T* |
| 43 | C | T | T | A | T |
| 44 | C | T | T | A | T |
| 45 | C | G | C | A | T |
| 46 | C | G | C | C | C |
| 47 | C | T | T | A | T |
| 48 | C | G | C | A | C |

| | | | | | |
|---|---|---|---|---|---|
| Allele 1 | 40.5 | 18 | 35 | 34 | 18 |
| Allele 2 | 10.5 | 33 | 16 | 17 | 33 |
| n = 51 | 51 | 51 | 51 | 51 | 51 |

Table 23.2 Proportions of partner *TCF12* alleles in 46 families – including uninformative cases

| SNP 1 rs 4774246(C>T) | Numbers | Proportions |
|---|---|---|
| C | 40.5 | 0.794117647 |
| T | 10.5 | 0.205882353 |
| | | |
| SNP 2 rs2585082 | | |
| T | 18 | 0.352941176 |
| G | 33 | 0.647058824 |
| | | |
| SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| C | 35 | 0.68627451 |
| T | 16 | 0.31372549 |
| | | |
| SNP 4 rs12901270(A>C) | Numbers | Proportions |
| A | 34 | 0.666666667 |

| | | |
|---|---|---|
| C | 17 | 0.333333333 |
| | | |
| SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| T | 18 | 0.352941176 |
| C | 33 | 0.647058824 |

Table 23.3 Fisher's exact test for partner *TCF12* alleles in 46 families – including uninformative cases

| | | | |
|---|---|---|---|
| | rs4774246 | | |
| | T | C | |
| Gp1 | 10.5 | 40.5 | 51 |
| Gp2 | 106 | 262 | 368 |
| | 116.5 | 302.5 | 419 |
| 2-tailed p value | 0.3214 | **not sig** | |
| | rs2585082 (T/G) | | |
| | T | G | |
| Gp1 | 18 | 33 | 51 |
| Gp2 | 176 | 206 | 382 |
| | 194 | 239 | |
| | | | |
| 2 tailed p value | 0.1774 | **Not sig** | |
| | rs1628067 (T/C) | | |
| | T | C | |
| Gp1 | 16 | 35 | 51 |

| | | | |
|---|---|---|---|
| Gp2 | 167 | 214 | 381 |
| | 183 | 249 | |
| | | | |
| | | | |
| 2 tailed p value | 0.0986 | **Not sig** | |
| | | | |
| | rs12901270 (A/C) | | |
| | | | |
| | A | C | |
| Gp1 | 34 | 17 | 51 |
| Gp2 | 274 | 96 | 370 |
| | 308 | 113 | |
| 2 tailed p value | 0.3114 | **Not sig** | |
| | rs2440979 (T/C) | | |
| | T | C | |
| Gp1 | 18 | 33 | 51 |
| Gp2 | 193 | 191 | 384 |
| | 211 | 224 | |
| | | | |
| 2 tailed p value | 0.0525 | **Not sig** | |
| | | | |

Table 24.1 Genotyping results of partner *TCF12* alleles in non-penetrant individuals in 46 families

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | T | T | T | A | T |
| 1 | C | G | C | A | C |
| 2 | C | G | C | C | C |
| 9 | C | T | T | A | T |
| 10 | T | T | T | A | T |
| 11 | C | G | C | C | C |
| 12 | T | G | T | A | T |
| 14 | C | T | T | A | T |
| 15 | C | T | T | A | T |
| 18 | C | G | C | C | C |
| 19 | C | G | C | C | C |
| 20 | C | T | T | A | C |
| 22 | C | G | C | C | C |
| 23 | C | G | C | C | C |
| 24 | C | G | C | C | C |
| 28 | C | G | C | A | C |
| 30 | C | G | C | A | C |
| 35 | T | T | T | A | C |
| 36 | C | G | C | A | C |
| 39 | C | G | C | A | C |
| 44 | C | T | T | A | T |
| 45 | C | G | C | A | T |
| 45 | T | T | C | A | T |
| 47 | C | G | C | A | C |
| 48 | C | T | T | A | T |

| | | | | | |
|---|---|---|---|---|---|
| Allele 1 | 20 | 10 | 15 | 18 | 10 |
| Allele 2 | 5 | 15 | 10 | 7 | 15 |
| n=26 | 25 | 25 | 25 | 25 | 25 |

Table 24.2 Proportions of partner *TCF12* alleles in non-penetrant individuals in 46 families

| n = | 26 | 26 | |
|---|---|---|---|
| | **Partner allele (in non-penetrant person)** | | |
| | SNP 1 rs 4774246(C>T) | Numbers | Proportions |
| | C | 20 | 0.8 |
| | T | 5 | 0.2 |
| | | | |
| | SNP 2 rs2585082 | | |
| | T | 10 | 0.4 |
| | G | 15 | 0.6 |
| | | | |
| | SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| | C | 15 | 0.6 |
| | T | 10 | 0.4 |
| | | | |
| | SNP 4 rs12901270(A>C) | Numbers | Proportions |
| | A | 18 | 0.72 |

| | | | |
|---|---|---|---|
| | C | 7 | 0.28 |
| | | | |
| | SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| | T | 10 | 0.4 |
| | C | 15 | 0.6 |

Table 24.3 Fisher's exact test for partner *TCF12* alleles in non-penetrant individuals in 46 families

| | | | |
|---|---|---|---|
| | rs4774246 | | |
| | T | C | |
| Gp1 | 5 | 20 | 19 |
| Gp2 | 106 | 262 | 368 |
| | 108 | 278 | 386 |
| 2-tailed p value | 0.4912 | not sig | |
| | rs2585082 (T/G) | | |
| | T | G | |
| Gp1 | 10 | 15 | 25 |
| Gp2 | 176 | 206 | 382 |
| | 180 | 220 | 400 |
| | | | |
| 2 tailed p value | 0.6797 | not sig | |
| | rs1628067 (T/C) | | |
| | T | C | |

| | | | |
|---|---|---|---|
| Gp1 | 10 | 15 | 25 |
| Gp2 | 167 | 214 | 381 |
| | 172 | 227 | 399 |
| 2 tailed p value | 0.8359 | Not sig | |
| | rs12901270 (A/C) | | |
| | A | C | |
| Gp1 | 18 | 7 | 25 |
| Gp2 | 274 | 96 | 370 |
| | 292 | 103 | 395 |
| 2 tailed p value | 0.8158 | Not sig | |
| | | | |
| | rs2440979 (T/C) | | |
| | T | C | |
| Gp1 | 10 | 15 | 25 |
| Gp2 | 193 | 191 | 384 |
| | 197 | 205 | 402 |
| | | | |
| 2 tailed p value | 0.4099 | Not sig | |

Table 25.1 Genotyping results of bystander *TCF12* alleles in 46 families

| Family # | SNP 1 [C/T] | SNP 2 [T>G] | SNP 3 [C>T] | SNP 4 [A/C] | SNP 5 [T>C] |
|---|---|---|---|---|---|
| 1 | C | T | T | A | T |
| 2 | C | G | C | A | C |
| 2 | C | G | C | A | C |
| 3 | C | G | C | A | C |
| 5 | C | T | T | A | T |
| 6 | C | G | C | C | T |
| 7 | C | G | C | A | C |
| 8 | C | T | T | A | T |
| 9 | C | T | T | A | T |
| 10 | C | G | C | A | C |
| 11 | C | G | C | A | C |
| 11 | T | T | T | A | C |
| 13 | C | T | T | A | T |
| 14 | T | T | T | A | T |
| 15 | C | T | T | A | T |
| 18 | C | T | T | A | T |
| 19 | C | T | T | A | T |
| 19 | T | T | T | A | C |
| 20 | T | T | T | A | T |
| 21 | C | G | C | A | C |
| 22 | C | G | C | A | C |
| 23 | C | T | T | A | T |
| 23 | T | T | T | A | T |
| 27 | T | T | T | A | C |
| 28 | C | G | C | A | C |

| | | | | | |
|---|---|---|---|---|---|
| 28 | T | T | T | A | T |
| 29 | T | T | T | A | T |
| 30 | C | G | C | A | C |
| 32 | C | G | C | A | C |
| 34 | C | T | T | A | C |
| 35 | C | T | T | A | T |
| 35 | C | T | T | A | T |
| 36 | C | T | T | A | T |
| 37 | C | T | C | A | C |
| 39 | C | G | C | C | C |
| 41 | C | G | C | A | C |
| 44 | C | T | T | A | C |
| 45 | C | T | T | A | C |
| 46 | C | T | T | A | T |
| 47 | C | G | C | A | C |
| 48 | C | G | C | C | C |
| Allele 1 | 33 | 25 | 17 | 38 | 19 |
| Allele 2 | 8 | 16 | 24 | 3 | 22 |
| | 41 | 41 | 41 | 41 | 41 |

Table 25.2 Proportions of bystander *TCF12* alleles in 46 families

| Bystander allele (parent not carrying mutation) | | |
|---|---|---|
| SNP 1 rs 4774246(C>T) | Numbers | Proportions |
| C | 33 | 0.804878049 |
| T | 8 | 0.195121951 |
| | | |
| SNP 2 rs2585082 (T>G) | | |
| T | 25 | 0.609756098 |
| G | 16 | 0.390243902 |
| | | |
| SNP 3 rs1628067 (C>T) | Numbers | Proportions |
| C | 17 | 0.414634146 |
| T | 24 | 0.585365854 |
| | | |
| | | |
| SNP 4 rs12901270(A>C) | Numbers | Proportions |
| A | 38 | 0.926829268 |
| C | 3 | 0.073170732 |
| SNP 5 rs2440979 (T>C) | Numbers | Proportions |
| T | 19 | 0.463414634 |
| C | 22 | 0.536585366 |

Table 25.3 Fisher's exact test for bystander *TCF12* alleles in 46 families

| | T | C | |
|---|---|---|---|
| Gp1 | 8 | 33 | 41 |
| Gp2 | 106 | 262 | 368 |
| | 114 | 295 | 409 |
| 2-tailed p value | 0.2707 | Not sig | |
| | | | |
| rs2585082 (T/G) | | | |
| | T | G | |
| Gp1 | 25 | 16 | 41 |
| Gp2 | 176 | 206 | 382 |
| | 201 | 222 | 423 |
| | | | |
| 2 tailed p value | 0.0728 | Not sig | |
| rs1628067 (T/C) | | | |
| | T | C | |
| Gp1 | 24 | 17 | 41 |
| Gp2 | 167 | 214 | 381 |
| | 191 | 231 | 422 |
| | | | |
| 2 tailed p value | 0.0978 | Not sig | |
| rs12901270 (A/C) | | | |
| | A | C | |

| | | | |
|---|---|---|---|
| Gp1 | 38 | 3 | 41 |
| Gp2 | 274 | 96 | 370 |
| | 312 | 99 | 411 |
| 2 tailed p value | 0.0064 | **Stat sig** | |
| | | | |
| rs2440979 (T/C) | | | |
| | T | C | |
| Gp1 | 19 | 22 | 41 |
| Gp2 | 193 | 191 | 384 |
| | 212 | 213 | 425 |
| 2 tailed p value | 0.7429 | Not sig | |

Table 26.1 Total proportions of each *TCF12* allele as per each tag-SNP – excluding uninformative cases

| | 1 | 2 | 3 | 4 | 5 |
|---|---|---|---|---|---|
| | rs4774246 (T/C) | rs2585082 (T/G) | rs1628067 (T/C) | rs12901270 (A/C) | rs2440979 (T/C) |
| Mutant *TCF12* allele (40) | 0.375 | 0.525 | 0.525 | 0.825 | 0.55 |
| Partner TCF12 allele (in presence of craniosynostosis) (49) | 0.204081633 | 0.367346939 | 0.346938776 | 0.673469388 | 0.387755102 |
| Partner TCF12 allele (in non-penetrant person) (25) | 0.2 | 0.4 | 0.4 | 0.72 | 0.4 |
| Bystander TCF12 allele (in parent not carrying mutation) (41) | 0.195121951 | 0.609756098 | 0.585365854 | 0.926829268 | 0.463414634 |
| HapMap CEU (234) | 0.248 | 0.406 | 0.38 | 0.697 | 0.449 |
| Control plates (156-192) | 0.288 | 0.461 | 0.438 | 0.741 | 0.503 |

Table 26.2 Total proportions of each *TCF12* allele as per each tag-SNP – including uninformative cases

| | 1 | 2 | 3 | 4 | 5 |
|---|---|---|---|---|---|
| | rs4774246 (T/C) | rs2585082 (T/G) | rs1628067 (T/C) | rs12901270 (A/C) | rs2440979 (T/C) |
| Mutant *TCF12* allele (43) | 0.348837209 | 0.488372093 | 0.488372093 | 0.790697674 | 0.534883721 |
| Partner TCF12 allele (in presence of craniosynostosis) (51) | 0.205882353 | 0.352941176 | 0.31372549 | 0.666666667 | 0.352941176 |
| Partner TCF12 allele (in non-penetrant person) (26) | 0.192307692 | 0.384615385 | 0.384615385 | 0.692307692 | 0.384615385 |
| Bystander TCF12 allele (in parent not carrying mutation) (40) | 0.175 | 0.6 | 0.575 | 0.9 | 0.45 |
| HapMap CEU (234) | 0.248 | 0.406 | 0.38 | 0.697 | 0.449 |
| Control plates (156-192) | 0.288 | 0.461 | 0.438 | 0.741 | 0.503 |

Table 27.1 *TCF12* expression data derived from B-cells in 45 CEPH pedigrees (original dataset mined from (Cheung et al., 2010).

| **rs4774246** | | | | | | | |
|---|---|---|---|---|---|---|---|
| | C/C | T/T | T/C | | | | |
| | 8.70563 | 8.81506 | 9.24864 | | | | |
| | 8.75975 | 9.02181 | 9.268605 | rs477426 genotype | C/C | T/C | T/T |
| | 9.26608 | 9.32787 | 9.390115 | Expression level | 9.7967 | 9.8241 | 9.342 |
| | 9.274375 | 9.71716 | 9.518375 | | | | |
| | 9.28504 | 9.82988 | 9.53372 | | | | |
| | 9.31424 | **9.342356** | 9.76272 | | | | |
| | 9.369645 | 0.43564847 | 9.772705 | | | | |
| | 9.55349 | | 9.805625 | | | | |
| | 9.574905 | | 9.944595 | | | | |
| | 9.59861 | | 10.1744 | | | | |
| | 9.65015 | | 10.35745 | | | | |
| | 9.666305 | | 10.38355 | | | | |
| | 9.7286 | | 10.55295 | | | | |
| | 9.75789 | | **9.8241115** | | | | |
| | 9.83395 | | 0.4357473 | | | | |
| | 9.905895 | | | | | | |
| | 9.91643 | | | | | | |
| | 9.94627 | | | AVG | 9.796679 | 9.342356 | 9.82411154 |
| | 9.950025 | | | SD | 0.46949573 | 0.43564847 | 0.4357473 |
| | 10.04385 | | | SQRT MEAN | 5.47722558 | 2.23606798 | 3.60555128 |
| | 10.073065 | | | SEM | 0.0857178 | 0.19482792 | 0.12085456 |
| | 10.10076 | | | | | | |
| | 10.16035 | | | | | | |
| | 10.23555 | | | | | | |
| | 10.2372 | | | | | | |
| | 10.238815 | | | | | | |
| | 10.2702 | | | | | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | 10.47745 | | | | | | |
| | 10.49225 | | | | | | |
| | 10.5136 | | | | | | |
| | **9.796679** | | | | | | |
| | 0.46949573 | | | | | | |

Table 27.2

| **rs2585082** | | | | | | | |
|---|---|---|---|---|---|---|---|
| | T/T | G/T | G/G | rs2585082 genotype | T/T | G/T | G/G |
| | 8.81506 | 8.70563 | 8.75975 | Expression level | 9.459 | 9.748 | 9.9 |
| | 8.95245 | 9.24864 | 9.26608 | | | | |
| | 9.02181 | 9.268605 | 9.31424 | | | | |
| | 9.28504 | 9.274375 | 9.574905 | | | | |
| | 9.32787 | 9.390115 | 9.59861 | | | | |
| | 9.53372 | 9.518375 | 9.65015 | | | | |
| | 9.59507 | 9.76272 | 9.666305 | AVG | 9.45942364 | 9.74885333 | 9.90042286 |
| | 9.71716 | 9.772705 | 9.7286 | SD | 0.4177005 | 0.5157805 | 0.45145371 |
| | 9.82988 | 9.805625 | 9.75789 | SQRT MEAN | 3.31662479 | 3.87298335 | 4.58257569 |
| | 9.83395 | 9.944595 | 9.905895 | SEM | 0.12594144 | 0.13317395 | 0.09851528 |
| | 10.14165 | 10.073065 | 9.91643 | | | | |
| | **9.4594236** | 10.1744 | 10.04385 | | | | |
| | 0.4177005 | 10.35745 | 10.10076 | | | | |
| | 3.31662479 | 10.38355 | 10.16035 | | | | |
| | 104.05366 | 10.55295 | 10.23555 | | | | |
| | | **9.7488533** | 10.2372 | | | | |
| | | 0.5157805 | 10.238815 | | | | |
| | | 3.87298335 | 10.2702 | | | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | | | 10.47745 | | | | |
| | | | 10.49225 | | | | |
| | | | 10.5136 | | | | |
| | | | **9.9004229** | | | | |
| | | | 0.45145371 | | | | |
| | | | 4.58257569 | | | | |

Table 27.3

| **rs12901270** | | | | | | | |
|---|---|---|---|---|---|---|---|
| | A/A | A/C | C/C | rs12901270 | A/A | A/C | C/C |
| | 8.70563 | 8.75975 | 9.59861 | Expression level | 9.63069583 | 9.71155 | 10.200402 |
| | 8.81506 | 9.26608 | 10.16035 | | | | |
| | 8.95245 | 9.268605 | 10.2372 | | | | |
| | 9.02181 | 9.37829 | 10.49225 | | | | |
| | 9.24864 | 9.45121 | 10.5136 | | | | |
| | 9.274375 | 9.518375 | **10.200402** | | | | |
| | 9.28504 | 9.666305 | 5 | | | | |
| | 9.31424 | 9.68912 | | | | | |
| | 9.32787 | 9.772705 | | | | | |
| | 9.369645 | 9.805625 | | AVG | 9.63069583 | 9.71155 | 10.200402 |
| | 9.390115 | 9.905115 | | SD | 0.46678212 | 0.38362352 | 0.37026346 |
| | 9.431765 | 9.905895 | | SQRT MEAN | 5.47722558 | 4.24264069 | 2.23606798 |
| | 9.53372 | 9.91643 | | SEM | 0.08522237 | 0.09042093 | 0.16558685 |
| | 9.55349 | 9.944595 | | | | | |
| | 9.59507 | 9.950025 | | | | | |
| | 9.711215 | 10.10076 | | | | | |
| | 9.71716 | 10.238815 | | | | | |
| | 9.7286 | 10.2702 | | | | | |
| | 9.75789 | **9.71155** | | | | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | 9.76272 | 18 | | | | | |
| | 9.82988 | | | | | | |
| | 9.83395 | | | | | | |
| | 9.94627 | | | | | | |
| | 10.04385 | | | | | | |
| | 10.09927 | | | | | | |
| | 10.14165 | | | | | | |
| | 10.23555 | | | | | | |
| | 10.35745 | | | | | | |
| | 10.38355 | | | | | | |
| | 10.55295 | | | | | | |
| | **9.6306958** | | | | | | |
| | 30 | | | | | | |

Table 27.4

| **rs1628067** | T/T | C/T | C/C | | | | |
|---|---|---|---|---|---|---|---|
| | 8.95245 | 8.81506 | 8.75975 | | | | |
| | 9.02181 | 9.24864 | 9.26608 | rs1628067(C>T) | T/T | C/T | C/C |
| | 9.32787 | 9.268605 | 9.31424 | Expression level | 9.468575 | 9.717215 | 9.894709 |
| | 9.53372 | 9.274375 | 9.574905 | | | | |
| | 9.83395 | 9.37829 | 9.59861 | | | | |
| | 10.14165 | 9.390115 | 9.65015 | | | | |
| | **9.468575** | 9.431765 | 9.666305 | | | | |
| | 6 | 9.45121 | 9.7286 | | | | |
| | | 9.518375 | 9.75789 | | | | |
| | | 9.55349 | 9.905895 | | T/T | C/T | C/C |
| | | 9.68912 | 9.91643 | AVG | 9.468575 | 9.717215 | 9.894709 |
| | | 9.711215 | 10.04385 | SD | 0.46401608 | 0.38551165 | 0.44449054 |
| | | 9.71716 | 10.10076 | SQRT MEAN | 2.44948974 | 5.09901951 | 4.58257569 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | | 9.76272 | 10.16035 | SEM | 0.18943377 | 0.07560506 | 0.09699579 |
| | | 9.772705 | 10.23555 | | | | |
| | | 9.805625 | 10.2372 | | | | |
| | | 9.82988 | 10.238815 | | | | |
| | | 9.905115 | 10.2702 | | | | |
| | | 9.944595 | 10.35745 | | | | |
| | | 9.94627 | 10.49225 | | | | |
| | | 9.950025 | 10.5136 | | | | |
| | | 10.073065 | **9.8947086** | | | | |
| | | 10.09927 | 21 | | | | |
| | | 10.1744 | | | | | |
| | | 10.38355 | | | | | |
| | | 10.55295 | | | | | |
| | | **9.717215** | | | | | |
| | | 26 | | | | | |

Table 28 Combined gene list of 11 patients with mutation-negative coronal synostosis from Round 2 of exome sequencing and mutation-negative patients in Round 1. Note there were no genes with variants shared between all 11, 10 , 9, or 8 patients.

| GENES WITH VARIANTS SHARED BETWEEN 7 PATIENTS | | | | | |
|---|---|---|---|---|---|
| PPP1R9B - neurabin-2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr17:48227279 | G > A | nonsynonymous; PPP1R9B:NM_032595:exon1:c.C596T:p.T199M | | 26,30 \| 30,23 |
| VS10 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 9,14 |
| VS11 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 5,25 |
| VS12 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 5,17 |
| VS13 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 5,15 |
| VS14 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 10,20 |
| VS8 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 7,9 |
| VS9 | chr17:48218714 | C > T | stopgain; PPP1R9B:NM_032595:exon4:c.G1646A:p.W549X | | 14,14 \| 17,18 |
| VS9 | chr17:48227384 | - > GC | frameshift insertion; PPP1R9B:NM_032595:exon1:c.491_492insGC:p.A164fs | | 0,0 \| 5,22 |
| | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| MAPK8IP2 - C-Jun-amino-terminal kinase-interacting protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 13,11 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS11 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 8,8 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS12 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 13,8 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS13 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 11,7 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS14 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 14,7 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS8 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 9,8 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| VS9 | chr22:51042513 | - > G | frameshift insertion; MAPK8IP2:NM_012324:exon5:c.785_786insG:p.A262fs | | 0,0 \| 9,9 |
| | | | MAPK8IP2:NM_016431:exon3:c.704_705insG:p.A235fs | | |
| | | | | | |
| | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| KCNC3 - potassium voltage-gated channel subfamily C | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,9 |
| VS11 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,16 |
| VS12 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,4 |
| VS13 | chr19:50827057 | C > T | nonsynonymous; KCNC3:NM_004977:exon2:c.G1153A:p.D385N | probably damaging | 27,14 \| 20,17 |
| VS13 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,4 |
| VS14 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,9 |
| VS8 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,11 |
| VS9 | chr19:50832152 | T > C | nonsynonymous; KCNC3:NM_004977:exon1:c.A188G:p.D63G | benign | 0,0 \| 0,10 |
| | | | | | |
| GENES WITH VARIANTS SHARED BETWEEN 5 PATIENTS | | | | | |
| COPZ2 - coatomer subunit zeta-2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS10 | chr17:46115123 | - > G | frameshift insertion; COPZ2:NM_016429:exon1:c.17_18insC:p.A6fs | | 0,0 \| 0,5 |
| VS11 | chr17:46115123 | - > G | frameshift insertion; COPZ2:NM_016429:exon1:c.17_18insC:p.A6fs | | 0,0 \| 0,5 |
| VS13 | chr17:46115123 | - > G | frameshift insertion; COPZ2:NM_016429:exon1:c.17_18insC:p.A6fs | | 0,0 \| 1,7 |
| VS14 | chr17:46115123 | - > G | frameshift insertion; COPZ2:NM_016429:exon1:c.17_18insC:p.A6fs | | 0,0 \| 0,5 |
| VS8 | chr17:46115123 | - > G | frameshift insertion; COPZ2:NM_016429:exon1:c.17_18insC:p.A6fs | | 0,0 \| 0,9 |
| | | | | | |
| | | | | | |
| TTN - titin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chr2:179454930 | T > C | nonsynonymous; TTN:NM_133432:exon133:c.A34702G:p.I11568V | benign | 38,56 \| 43,47 |
| | | | TTN:NM_133437:exon133:c.A34903G:p.I11635V | | |
| | | | TTN:NM_133378:exon253:c.A53818G:p.I17940V | | |
| | | | TTN:NM_003319:exon132:c.A34327G:p.I11443V | | |
| VS3 | chr2:179434293 | - > CTT | nonframeshift insertion; TTN:NM_133432:exon155:c.49746_49747insAAG:p.I16582delinsIR | | 3,6 \| 1,4 |
| | | | TTN:NM_003319:exon154:c.49371_49372insAAG:p.I16457delinsIR | | |
| | | | TTN:NM_133437:exon155:c.49947_49948insAAG:p.I16649delinsIR | | |
| | | | TTN:NM_133378:exon275:c.68862_68863insAAG:p.I22954delinsIR | | |

| | | | | | |
|---|---|---|---|---|---|
| VS5 | chr2:179433388 | C > T | nonsynonymous; TTN:NM_133432:exon155:c.G50651A:p.R16884H | | 3,22 \| 5,17 |
| | | | TTN:NM_003319:exon154:c.G50276A:p.R16759H | | |
| | | | TTN:NM_133437:exon155:c.G50852A:p.R16951H | | |
| | | | TTN:NM_133378:exon275:c.G69767A:p.R23256H | | |
| VS6 | chr2:179483108 | C > T | nonsynonymous; TTN:NM_133432:exon81:c.G20257A:p.V6753I | unknown | 6,25 \| 14,25 |
| | | | TTN:NM_133437:exon81:c.G20458A:p.V6820I | | |
| | | | TTN:NM_133378:exon201:c.G39373A:p.V13125I | | |
| | | | TTN:NM_003319:exon80:c.G19882A:p.V6628I | | |
| VS6 | chr2:179544700 | - > TCT | nonframeshift insertion; TTN:NM_133378:exon137:c.29769_29770insAGA:p.V9923delinsVE | | 5,14 \| 6,4 |
| VS9 | chr2:179544700 | - > TCT | nonframeshift insertion; TTN:NM_133378:exon137:c.29769_29770insAGA:p.V9923delinsVE | | 10,8 \| 3,5 |
| | | | | | |
| | | | | | |
| DLX6 - homeobox protein DLX-6 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr7:96635449 | - > CGC | nonframeshift insertion; DLX6:NM_005222:exon1:c.160_161insCGC:p.Q54delinsPQ | | 0,0 \| 5,7 |
| VS11 | chr7:96635449 | - > CGC | nonframeshift insertion; DLX6:NM_005222:exon1:c.160_161insCGC:p.Q54delinsPQ | | 0,0 \| 8,14 |
| VS12 | chr7:96635449 | - > CGC | nonframeshift insertion; DLX6:NM_005222:exon1:c.160_161insCGC:p.Q54delinsPQ | | 6,4 \| 4,3 |
| VS13 | chr7:96635449 | - > CGC | nonframeshift insertion; DLX6:NM_005222:exon1:c.160_161insCGC:p.Q54delinsPQ | | 1,2 \| 0,3 |

| | | | | | |
|---|---|---|---|---|---|
| VS14 | chr7:96635449 | - > CGC | nonframeshift insertion; DLX6:NM_005222:exon1:c.160_161insCGC:p.Q54delinsPQ | | 5,1 \| 5,5 |
| | | | | | |
| GENES WITH VARIANTS SHARED BETWEEN 4 PATIENTS | | | | | |
| PCLO - protein piccolo | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 54,32 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| VS3 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 42,24 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| VS5 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 41,20 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| VS6 | chr7:82581493 | - > ATC | nonframeshift insertion; PCLO:NM_033026:exon5:c.8776_8777insGAT:p.D2926delinsDD | | 0,0 \| 45,36 |
| | | | PCLO:NM_014510:exon5:c.8776_8777insGAT:p.D2926delinsDD | | |
| | | | | | |
| | | | | | |
| LAMA5 - laminin subunit alpha-5 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS10 | chr20:60887285 | C > T | nonsynonymous; LAMA5:NM_005560:exon69:c.G9448A:p.G3150S | benign | 27,28 \| 10,11 |
| VS12 | chr20:60885296 | T > G | nonsynonymous; LAMA5:NM_005560:exon77:c.A10672C:p.I3558L | benign | 15,19 \| 10,15 |
| VS14 | chr20:60921603 | C > T | nonsynonymous; LAMA5:NM_005560:exon9:c.G1241A:p.R414H | benign | 6,8 \| 3,6 |
| VS2 | chr20:60892518 | C > T | nonsynonymous; LAMA5:NM_005560:exon55:c.G7394A:p.R2465Q | benign | 4,3 \| 3,4 |
| | | | | | |
| | | | | | |
| NDUFS1 - NADH-ubiquinone oxidoreductase 75 kDa subunit, | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | possibly damaging | 15,14 \| 11,0 |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| VS3 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 21,7 \| 13,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |

| | | | | | |
|---|---|---|---|---|---|
| VS5 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | possibly damaging | 10,10 \| 12,0 |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | | |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| VS6 | chr2:206992537 | A > C | nonsynonymous; NDUFS1:NM_001199983:exon15:c.T1697G:p.I566R | possibly damaging | 17,4 \| 15,0 |
| | | | NDUFS1:NM_001199984:exon16:c.T1910G:p.I637R | | |
| | | | NDUFS1:NM_001199981:exon15:c.T1760G:p.I587R | | |
| | | | NDUFS1:NM_005006:exon16:c.T1868G:p.I623R | | |
| | | | NDUFS1:NM_001199982:exon13:c.T1535G:p.I512R | | |
| | | | | | |
| | | | | | |
| SSPO - SCO-spondin precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr7:149484636 | C > T | nonsynonymous; SSPO:NM_198455:exon22:c.C3563T:p.P1188L | | 6,15 \| 6,9 |
| VS10 | chr7:149513596 | C > A | nonsynonymous; SSPO:NM_198455:exon74:c.C11227A:p.H3743N | | 44,9 \| 32,10 |
| VS2 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 2,2 |
| VS3 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 0,3 |
| VS5 | chr7:149476670 | - > C | frameshift insertion; SSPO:NM_198455:exon9:c.1122_1123insC:p.P374fs | | 0,0 \| 1,3 |

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|---|---|---|---|---|---|
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| MUC5B - mucin-5B precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr11:1250475 | G > T | nonsynonymous; MUC5B:NM_002458:exon9:c.G1052T:p.R351L | benign | 5,12 \| 0,5 |
| VS2 | chr11:1270647 | G > C | nonsynonymous; MUC5B:NM_002458:exon31:c.G12537C:p.Q4179H | unknown | 22,5 \| 16,1 |
| VS2 | chr11:1274084 | G > A | nonsynonymous; MUC5B:NM_002458:exon33:c.G15091A:p.V5031M | unknown | 1,0 \| 3,1 |
| VS3 | chr11:1263932 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C5822G:p.T1941S | benign | 40,20 \| 25,14 |
| VS3 | chr11:1265384 | T > C | nonsynonymous; MUC5B:NM_002458:exon31:c.T7274C:p.M2425T | unknown | 21,17 \| 19,2 |
| VS3 | chr11:1265671 | G > A | nonsynonymous; MUC5B:NM_002458:exon31:c.G7561A:p.A2521T | unknown | 67,19 \| 13,14 |
| VS3 | chr11:1265687 | G > T | nonsynonymous; MUC5B:NM_002458:exon31:c.G7577T:p.R2526I | unknown | 65,21 \| 21,12 |
| VS3 | chr11:1266892 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C8782G:p.R2928G | unknown | 17,6 \| 23,0 |
| VS3 | chr11:1267010 | A > G | nonsynonymous; MUC5B:NM_002458:exon31:c.A8900G:p.N2967S | unknown | 45,24 \| 29,12 |

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|---|---|---|---|---|---|
| VS3 | chr11:12672 89 | A > C | nonsynonymous; MUC5B:NM_002458:exon31:c.A9179C:p.K3060T | unknown | 31,22 \| 20,36 |
| VS3 | chr11:12672 91 | T > C | nonsynonymous; MUC5B:NM_002458:exon31:c.T9181C:p.S3061P | unknown | 32,23 \| 20,36 |
| VS3 | chr11:12678 52 | G > A | nonsynonymous; MUC5B:NM_002458:exon31:c.G9742A:p.A3248T | unknown | 16,16 \| 2,7 |
| VS3 | chr11:12697 70 | C > T | nonsynonymous; MUC5B:NM_002458:exon31:c.C11660T:p.P3887L | unknown | 3,12 \| 2,5 |
| VS3 | chr11:12703 61 | C > T | nonsynonymous; MUC5B:NM_002458:exon31:c.C12251T:p.T4084M | unknown | 15,38 \| 6,13 |
| VS3 | chr11:12707 81 | A > G | nonsynonymous; MUC5B:NM_002458:exon31:c.A12671G:p.N4224S | unknown | 26,21 \| 12,13 |
| VS3 | chr11:12718 94 | C > G | nonsynonymous; MUC5B:NM_002458:exon31:c.C13784G:p.T4595S | unknown | 7,16 \| 7,3 |
| VS6 | chr11:12706 47 | G > C | nonsynonymous; MUC5B:NM_002458:exon31:c.G12537C:p.Q4179H | unknown | 21,3 \| 10,1 |
| | | | | | |
| GENES WITH VARIANTS SHARED BETWEEN 3 PATIENTS | | | | | |
| PCDH17 - protocadherin-17 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chr13:58208 735 | C > G | nonsynonymous; PCDH17:NM_001040429:exon1:c.C2055G:p.S685R | benign | 15,14 \| 23,11 |

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|---|---|---|---|---|---|
| VS6 | chr13:58207504 | A > G | nonsynonymous; PCDH17:NM_001040429:exon1:c.A824G:p.N275S | probably damaging | 6,10 \| 5,10 |
| VS8 | chr13:58207351 | C > A | nonsynonymous; PCDH17:NM_001040429:exon1:c.C671A:p.S224Y | possibly damaging | 20,39 \| 18,15 |
| | | | | | |
| | | | | | |
| TCF7L1 - transcription factor 7-like 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr2:85360847 | GGC > - | nonframeshift deletion; TCF7L1:NM_031283:exon1:c.40_42del:p.14_14del | | 1,0 \| 5,0 |
| VS14 | chr2:85360847 | GGC > - | nonframeshift deletion; TCF7L1:NM_031283:exon1:c.40_42del:p.14_14del | | 0,0 \| 15,0 |
| VS8 | chr2:85360847 | GGC > - | nonframeshift deletion; TCF7L1:NM_031283:exon1:c.40_42del:p.14_14del | | 7,0 \| 8,2 |
| | | | | | |
| | | | | | |
| AMDHD2 - putative N-acetylglucosamine-6-phosphate | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr16:2578571 | T > - | frameshift deletion; AMDHD2:NM_015944:exon8:c.981delT:p.P327fs | | 29,10 \| 17,4 |
| | | | AMDHD2:NM_001145815:exon8:c.981delT:p.P327fs | | |

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|---|---|---|---|---|---|
| VS12 | chr16:2579545 | C > G | nonsynonymous; AMDHD2:NM_015944:exon10:c.C1301G:p.A434G | benign | 2,3 \| 5,0 |
| VS9 | chr16:2579545 | C > G | nonsynonymous; AMDHD2:NM_015944:exon10:c.C1301G:p.A434G | benign | 8,14 \| 11,0 |
| | | | | | |
| | | | | | |
| MST1P2 - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr1:16975285 | - > C | frameshift insertion; MST1P2:NR_027504:exon8:c.1654_1655insC:p.Q552fs | | 17,12 \| 11,8 |
| VS3 | chr1:16974312 | G > C | nonsynonymous; MST1P2:NR_027504:exon7:c.G772C:p.V258L | | 13,3 \| 4,0 |
| VS6 | chr1:16975285 | - > CC | frameshift insertion; MST1P2:NR_027504:exon8:c.1654_1655insCC:p.Q552fs | | 26,17 \| 2,3 |
| | | | | | |
| | | | | | |
| RSBN1L - round spermatid basic protein 1-like protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 4,20 \| 0,17 |
| VS3 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 6,31 \| 0,14 |

| | | | | | |
|---|---|---|---|---|---|
| VS5 | chr7:77379331 | T > G | nonsynonymous; RSBN1L:NM_198467:exon3:c.T1294G:p.L432V | possibly damaging | 2,27 \| 0,15 |
| | | | | | |
| | | | | | |
| TCHH - trichohyalin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr1:152084212 | - > GCTGCTCGCGCCTCTCCT | nonframeshift insertion; TCHH:NM_007113:exon2:c.1481_1482insAGGAGAGGCGCGAGCAGC:p.E494delinsEERREQQ | | 83,79 \| 45,42 |
| VS12 | chr1:152084212 | - > GCTGCTCGCGCCTCTCCT | nonframeshift insertion; TCHH:NM_007113:exon2:c.1481_1482insAGGAGAGGCGCGAGCAGC:p.E494delinsEERREQQ | | 0,0 \| 85,75 |
| VS13 | chr1:152084212 | - > GCTGCTCGCGCCTCTCCT | nonframeshift insertion; TCHH:NM_007113:exon2:c.1481_1482insAGGAGAGGCGCGAGCAGC:p.E494delinsEERREQQ | | 68,61 \| 28,21 |
| | | | | | |
| | | | | | |
| OR4N3P - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 92,29 \| 30,9 |

| | | | | | |
|---|---|---|---|---|---|
| VS5 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 119,29 \| 30,9 |
| VS6 | chr15:22413601 | - > T | frameshift insertion; OR4N3P:NR_028067:exon1:c.140_141insT:p.F47fs | | 87,23 \| 54,19 |
| | | | | | |
| | | | | | |
| FAM22F - hypothetical protein LOC54754 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 3,3 \| 1,3 |
| VS5 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 1,2 \| 0,3 |
| VS6 | chr9:97080947 | AAG > - | nonframeshift deletion; FAM22F:NM_017561:exon7:c.2069_2071del:p.690_691del | | 4,6 \| 2,4 |
| | | | | | |
| | | | | | |
| C9orf150 - hypothetical protein LOC286343 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr9:12775879 | - > GGGGGCGGC | nonframeshift insertion; C9orf150:NM_203403:exon1:c.165_166insGGGGGCGGC:p.G55delinsGGGGG | | 17,21 \| 47,18 |

| | | | | | |
|---|---|---|---|---|---|
| VS14 | chr9:12775879 | - > GGCAGC | nonframeshift insertion; C9orf150:NM_203403:exon1:c.165_166insGGCAGC:p.G55delinsGGS | | 24,24 \| 11,6 |
| VS8 | chr9:12775879 | - > GACGGC | nonframeshift insertion; C9orf150:NM_203403:exon1:c.165_166insGACGGC:p.G55delinsGDG | | 18,31 \| 10,8 |
| | | | | | |
| | | | | | |
| LTBP4 - latent-transforming growth factor beta-binding | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,4 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| VS3 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 1,2 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| VS5 | chr19:41123095 | - > G | frameshift insertion; LTBP4:NM_001042545:exon21:c.3034_3035insG:p.V1012fs | | 0,0 \| 0,7 |
| | | | LTBP4:NM_003573:exon24:c.3124_3125insG:p.V1042fs | | |
| | | | LTBP4:NM_001042544:exon24:c.3235_3236insG:p.V1079fs | | |
| | | | | | |
| | | | | | |
| ABCA2 - ABC transporter A family member 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS5 | chr9:139906404 | G > T | nonsynonymous; ABCA2:NM_212533:exon34:c.C5522A:p.A1841D | probably damaging | 3,1 \| 3,0 |
| | | | ABCA2:NM_001606:exon34:c.C5432A:p.A1811D | | |
| VS6 | chr9:139905705 | G > A | nonsynonymous; ABCA2:NM_212533:exon37:c.C5951T:p.T1984I | | 2,2 \| 0,6 |
| | | | ABCA2:NM_001606:exon37:c.C5861T:p.T1954I | | |
| VS9 | chr9:139910131 | C > T | nonsynonymous; ABCA2:NM_212533:exon22:c.G3605A:p.S1202N | | 11,12 \| 9,10 |
| | | | ABCA2:NM_001606:exon22:c.G3515A:p.S1172N | | |
| | | | | | |
| | | | | | |
| C20orf194 - hypothetical protein LOC25943 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr20:3270852 | G > T | nonsynonymous; C20orf194:NM_001009984:exon26:c.C2250A:p.S750R | possibly damaging | 11,7 \| 2,2 |
| VS3 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 9,5 \| 0,9 |
| VS5 | chr20:3362103 | T > G | nonsynonymous; C20orf194:NM_001009984:exon3:c.A206C:p.N69T | benign | 12,8 \| 0,12 |
| | | | | | |
| | | | | | |
| ANKRD43 - ankyrin repeat domain-containing protein 43 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS14 | chr5:132149339 | - > CGCCGCCGC | nonframeshift insertion; ANKRD43:NM_175873:exon1:c.26_27insCGCCGCCGC:p.A9delinsAAAA | | 9,1 \| 7,0 |
| VS8 | chr5:132149339 | - > CGCCGCCGC | nonframeshift insertion; ANKRD43:NM_175873:exon1:c.26_27insCGCCGCCGC:p.A9delinsAAAA | | 10,2 \| 6,1 |
| VS9 | chr5:132149339 | - > CGCCGCCGC | nonframeshift insertion; ANKRD43:NM_175873:exon1:c.26_27insCGCCGCCGC:p.A9delinsAAAA | | 4,3 \| 8,0 |
| | | | | | |
| | | | | | |
| AKD1 - adenylate kinase domain-containing protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr6:109867129 | G > A | nonsynonymous; AKD1:NM_001145128:exon26:c.C3166T:p.L1056F | possibly damaging | 7,2 \| 10,4 |
| VS12 | chr6:109835501 | C > T | nonsynonymous; AKD1:NM_001145128:exon32:c.G4205A:p.R1402H | possibly damaging | 2,8 \| 1,8 |
| VS14 | chr6:109962734 | T > C | nonsynonymous; AKD1:NM_145025:exon10:c.A920G:p.D307G | possibly damaging | 24,10 \| 26,16 |
| | | | AKD1:NM_001145128:exon10:c.A920G:p.D307G | | |
| | | | | | |
| | | | | | |
| PRAMEF4 - PRAME family member 4 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| VS2 | chr1:12939552 | - > G | frameshift insertion; PRAMEF4:NM_001009611:exon4:c.1250_1251insC:p.A417fs | | 37,47 \| 29,23 |
| VS3 | chr1:12941966 | C > T | nonsynonymous; PRAMEF4:NM_001009611:exon3:c.G584A:p.R195H | benign | 67,85 \| 25,27 |
| VS6 | chr1:12942179 | T > C | nonsynonymous; PRAMEF4:NM_001009611:exon3:c.A371G:p.H124R | benign | 34,22 \| 14,2 |
| | | | | | |
| | | | | | |
| NBEAL2 - neurobeachin-like protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr3:47037840 | C > T | nonsynonymous; NBEAL2:NM_015175:exon16:c.C2231T:p.P744L | benign | 60,87 \| 53,63 |
| VS14 | chr3:47045749 | C > A | nonsynonymous; NBEAL2:NM_015175:exon37:c.C6064A:p.P2022T | benign | 27,45 \| 40,42 |
| VS3 | chr3:47045998 | G > A | nonsynonymous; NBEAL2:NM_015175:exon38:c.G6212A:p.R2071H | probably damaging | 3,4 \| 4,2 |
| | | | | | |
| | | | | | |
| MAGEF1 - melanoma-associated antigen F1 | | | | | |

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|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr3:184429147 | - > ATC | nonframeshift insertion; MAGEF1:NM_022149:exon1:c.463_464insGAT:p.E155delinsDE | | 47,42 \| 107,71 |
| VS12 | chr3:184429142 | - > GCC | nonframeshift insertion; MAGEF1:NM_022149:exon1:c.468_469insGGC:p.E156delinsEA | | 46,33 \| 96,65 |
| VS14 | chr3:184429139 | - > ACC | nonframeshift insertion; MAGEF1:NM_022149:exon1:c.471_472insGGT:p.E157delinsEV | | 32,34 \| 94,62 |
| | | | | | |
| GENES WITH VARIANTS SHARED BETWEEN 2 PATIENTS | | | | | |
| QSOX2 - sulfhydryl oxidase 2 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr9:139137324 | C > A | nonsynonymous; QSOX2:NM_181701:exon1:c.G326T:p.R109L | benign | 14,3 \| 2,1 |
| VS8 | chr9:139137421 | T > C | nonsynonymous; QSOX2:NM_181701:exon1:c.A229G:p.N77D | probably damaging | 1,6 \| 10,7 |
| | | | | | |
| | | | | | |
| ERMN - ermin | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| VS10 | chr2:158182169 | T > A | nonsynonymous; ERMN:NM_001009959:exon2:c.A25T:p.I9F | benign | 42,21 \| 37,15 |
| VS12 | chr2:158178285 | A > G | nonsynonymous; ERMN:NM_020711:exon3:c.T353C:p.I118T | benign | 37,53 \| 25,41 |
| | | | ERMN:NM_001009959:exon4:c.T392C:p.I131T | | |
| | | | | | |
| | | | | | |
| MARK1 - serine/threonine-protein kinase MARK1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr1:220835506 | T > G | stoploss; MARK1:NM_018650:exon18:c.T2386G:p.X796E | | 0,18 \| 0,10 |
| VS5 | chr1:220835506 | T > G | stoploss; MARK1:NM_018650:exon18:c.T2386G:p.X796E | | 0,6 \| 0,4 |
| | | | | | |
| | | | | | |
| AIM1L - absent in melanoma 1-like protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS11 | chr1:26673081 | C > T | nonsynonymous; AIM1L:NM_001039775:exon2:c.G68A:p.R23Q | probably damaging | 49,34 \| 31,26 |
| VS13 | chr1:26670418 | C > T | nonsynonymous; AIM1L:NM_001039775:exon2:c.G2731A:p.G911S | | 20,20 \| 17,17 |
| | | | | | |
| | | | | | |
| PCDHGA10 - protocadherin gamma-A10 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr5:140793262 | T > C | nonsynonymous; PCDHGA10:NM_032090:exon1:c.T520C:p.Y174H | probably damaging | 31,18 \| 34,11 |
| | | | PCDHGA10:NM_018913:exon1:c.T520C:p.Y174H | | |
| VS2 | chr5:140795212 | A > T | stopgain; PCDHGA10:NM_032090:exon1:c.A2470T:p.K824X | | 0,12 \| 0,7 |
| | | | | | |
| | | | | | |
| ADAM29 - disintegrin and metalloproteinase | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr4:175898748 | TAT > - | nonframeshift deletion; ADAM29:NM_001130704:exon4:c.2072_2074del:p.691_692del | | 19,5 \| 5,1 |
| | | | ADAM29:NM_014269:exon5:c.2072_2074del:p.691_692del | | |

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|---|---|---|---|---|---|
| | | | ADAM29:NM_001130705:exon3:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130703:exon4:c.2072_2074del:p.691_692del | | |
| VS5 | chr4:175898748 | TAT > - | nonframeshift deletion; ADAM29:NM_001130704:exon4:c.2072_2074del:p.691_692del | | 12,5 \| 11,4 |
| | | | ADAM29:NM_014269:exon5:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130705:exon3:c.2072_2074del:p.691_692del | | |
| | | | ADAM29:NM_001130703:exon4:c.2072_2074del:p.691_692del | | |
| | | | | | |
| | | | | | |
| MAML3 - mastermind-like protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr4:140811110 | - > TTGCTGTTG | nonframeshift insertion; MAML3:NM_018717:exon2:c.1480_1481insCAACAGCAA:p.Q494delinsQQQQ | | 10,15 \| 0,9 |
| VS13 | chr4:140811110 | - > TTGCTGTTG | nonframeshift insertion; MAML3:NM_018717:exon2:c.1480_1481insCAACAGCAA:p.Q494delinsQQQQ | | 4,19 \| 2,8 |
| | | | | | |
| | | | | | |
| LGI4 - leucine-rich repeat LGI family member 4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS11 | chr19:35625544 | G > C | nonsynonymous; LGI4:NM_139284:exon1:c.C41G:p.A14G | benign | 5,14 \| 6,0 |
| VS8 | chr19:35625544 | G > C | nonsynonymous; LGI4:NM_139284:exon1:c.C41G:p.A14G | benign | 7,14 \| 10,0 |
| | | | | | |
| | | | | | |
| PRPF19 - pre-mRNA-processing factor 19 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr11:60666746 | T > G | nonsynonymous; PRPF19:NM_014502:exon11:c.A859C:p.T287P | probably damaging | 2,4 \| 0,4 |
| VS5 | chr11:60666746 | T > G | nonsynonymous; PRPF19:NM_014502:exon11:c.A859C:p.T287P | probably damaging | 5,2 \| 0,3 |
| | | | | | |
| | | | | | |
| JHDM1D - lysine-specific demethylase 7 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr7:139824526 | A > T | nonsynonymous; JHDM1D:NM_030647:exon7:c.T946A:p.S316T | benign | 13,18 \| 11,27 |
| VS11 | chr7:139801920 | A > G | nonsynonymous; JHDM1D:NM_030647:exon12:c.T1469C:p.I490T | benign | 1,11 \| 4,14 |
| | | | | | |
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|---|---|---|---|---|---|
| CBX4 - E3 SUMO-protein ligase CBX4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr17:77807926 | - > GCCGCC | nonframeshift insertion; CBX4:NM_003655:exon5:c.1515_1516insGGCGGC:p.V505delinsVAA | | 17,25 \| 17,20 |
| VS14 | chr17:77807926 | - > GCCGCC | nonframeshift insertion; CBX4:NM_003655:exon5:c.1515_1516insGGCGGC:p.V505delinsVAA | | 15,24 \| 14,38 |
| | | | | | |
| | | | | | |
| HEMGN - hemogen | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr9:100692325 | A > G | nonsynonymous; HEMGN:NM_197978:exon3:c.T1352C:p.F451S | possibly damaging | 49,1 \| 30,1 |
| | | | HEMGN:NM_018437:exon4:c.T1352C:p.F451S | | |
| VS3 | chr9:100693398 | T > G | nonsynonymous; HEMGN:NM_197978:exon3:c.A279C:p.E93D | benign | 28,13 \| 17,7 |
| | | | HEMGN:NM_018437:exon4:c.A279C:p.E93D | | |
| | | | | | |
| | | | | | |
| KIAA0748 - hypothetical protein LOC9840 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| VS10 | chr12:55356261 | C > A | nonsynonymous; KIAA0748:NM_001098815:exon9:c.G1421T:p.R474L | benign | 25,5 \| 18,3 |
| | | | KIAA0748:NM_001136030:exon9:c.G1421T:p.R474L | | |
| VS6 | chr12:55356526 | G > A | nonsynonymous; KIAA0748:NM_001098815:exon9:c.C1156T:p.P386S | benign | 6,17 \| 3,15 |
| | | | KIAA0748:NM_001136030:exon9:c.C1156T:p.P386S | | |
| | | | | | |
| | | | | | |
| IRS2 - insulin receptor substrate 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr13:110435744 | C > A | nonsynonymous; IRS2:NM_003749:exon1:c.G2657T:p.R886L | probably damaging | 8,4 \| 7,3 |
| VS8 | chr13:110434532 | A > G | nonsynonymous; IRS2:NM_003749:exon1:c.T3869C:p.L1290P | possibly damaging | 8,8 \| 6,7 |
| | | | | | |
| | | | | | |
| ZDBF2 - DBF4-type zinc finger-containing protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS3 | chr2:207175370 | C > T | nonsynonymous; ZDBF2:NM_020923:exon5:c.C6118T:p.R2040W | benign | 4,2 \| 1,4 |
| VS5 | chr2:207170388 | A > G | nonsynonymous; ZDBF2:NM_020923:exon5:c.A1136G:p.Q379R | benign | 6,5 \| 2,7 |
| | | | | | |
| | | | | | |
| ZSWIM6 - zinc finger SWIM domain-containing protein 6 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr5:60628199 | - > GCG | nonframeshift insertion; ZSWIM6:NM_020928:exon1:c.100_101insGCG:p.A34delinsGA | | 1,0 \| 0,3 |
| VS8 | chr5:60628173 | - > CGG | nonframeshift insertion; ZSWIM6:NM_020928:exon1:c.74_75insCGG:p.G25delinsGG | | 0,2 \| 0,3 |
| | | | | | |
| | | | | | |
| OBSCN - obscurin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr1:228547428 | C > A | nonsynonymous; OBSCN:NM_052843:exon81:c.C18835A:p.L6279M | possibly damaging | 10,3 \| 0,3 |
| VS6 | chr1:228481122 | C > T | nonsynonymous; OBSCN:NM_052843:exon41:c.C10936T:p.R3646W | probably damaging | 5,6 \| 7,2 |
| | | | OBSCN:NM_001098623:exon41:c.C10936T:p.R3646W | | |
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|---|---|---|---|---|---|
| POU2F2 - POU domain, class 2, transcription factor 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr19:42596244 | C > A | nonsynonymous; POU2F2:NM_001207025:exon13:c.G1377T:p.L459F | possibly damaging | 12,3 \| 0,8 |
| | | | POU2F2:NM_001207026:exon13:c.G1377T:p.L459F | | |
| | | | POU2F2:NM_002698:exon13:c.G1329T:p.L443F | | |
| VS8 | chr19:42596244 | C > A | nonsynonymous; POU2F2:NM_001207025:exon13:c.G1377T:p.L459F | possibly damaging | 12,1 \| 0,4 |
| | | | POU2F2:NM_001207026:exon13:c.G1377T:p.L459F | | |
| | | | POU2F2:NM_002698:exon13:c.G1329T:p.L443F | | |
| | | | | | |
| | | | | | |
| BIVM-ERCC5,ERCC5 - DNA repair protein complementing XP-G cells | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS5 | chr13:103519019 | C > A | nonsynonymous; ERCC5:NM_000123:exon11:c.C2357A:p.A786D | | 13,5 \| 1,2 |
| | | | BIVM-ERCC5:NM_001204425:exon19:c.C3719A:p.A1240D | | |
| VS8 | chr13:103527717 | C > T | nonsynonymous; ERCC5:NM_000123:exon15:c.C3025T:p.R1009C | benign | 12,2 \| 12,1 |
| | | | BIVM-ERCC5:NM_001204425:exon23:c.C4387T:p.R1463C | | |
| | | | | | |
| | | | | | |

| PHLDB3 - pleckstrin homology-like domain family B member | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr19:44008078 | T > C | nonsynonymous; PHLDB3:NM_198850:exon2:c.A193G:p.T65A | probably damaging | 101,154 \| 85,121 |
| VS5 | chr19:44008158 | G > C | nonsynonymous; PHLDB3:NM_198850:exon2:c.C113G:p.S38C | benign | 0,6 \| 1,6 |
| | | | | | |
| | | | | | |
| KIF1A - kinesin-like protein KIF1A | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr2:241680767 | G > A | nonsynonymous; KIF1A:NM_004321:exon33:c.C3365T:p.T1122M | possibly damaging | 2,3 \| 4,4 |
| VS14 | chr2:241700257 | G > C | nonsynonymous; KIF1A:NM_004321:exon24:c.C2242G:p.Q748E | benign | 7,8 \| 9,4 |
| | | | | | |
| | | | | | |
| ZNF772 - zinc finger protein 772 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS13 | chr19:57986395 | G > A | nonsynonymous; ZNF772:NM_001024596:exon4:c.C319T:p.P107S | probably damaging | 29,36 \| 21,42 |
| VS6 | chr19:57988675 | - > GCC | nonframeshift insertion; ZNF772:NM_001024596:exon1:c.3_4insGGC:p.M1delinsMA | | 0,20 \| 2,1 |
| | | | ZNF772:NM_001144068:exon1:c.3_4insGGC:p.M1delinsMA | | |
| | | | | | |
| | | | | | |
| FCGBP - IgGFc-binding protein precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr19:40408566 | C > T | nonsynonymous; FCGBP:NM_003890:exon8:c.G4273A:p.D1425N | benign | 0,2 \| 1,3 |
| VS5 | chr19:40392544 | TCT > - | nonframeshift deletion; FCGBP:NM_003890:exon16:c.7958_7960del:p.2653_2654del | | 5,31 \| 2,3 |
| | | | | | |
| | | | | | |
| TPSB2 - tryptase beta-2 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr16:1279732 | C > A | nonsynonymous; TPSB2:NM_024164:exon3:c.G68T:p.G23V | benign | 2,5 \| 1,4 |

| | | | | | |
|---|---|---|---|---|---|
| VS5 | chr16:12799 45 | T > C | nonsynonymous; TPSB2:NM_024164:exon2:c.A8G:p.N3S | unknown | 4,3 \| 4,0 |
| | | | | | |
| | | | | | |
| EML6 - echinoderm microtubule-associated protein-like | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr2:550565 86 | T > A | nonsynonymous; EML6:NM_001039753:exon6:c.T819A:p.D273E | probably damaging | 14,13 \| 14,14 |
| VS12 | chr2:551305 11 | C > T | nonsynonymous; EML6:NM_001039753:exon23:c.C3281T:p.T1094M | benign | 22,27 \| 21,13 |
| | | | | | |
| | | | | | |
| PALLD - palladin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr4:169846 390 | G > A | nonsynonymous; PALLD:NM_001166108:exon21:c.G3365A:p.R1122Q | benign | 22,13 \| 15,9 |
| | | | PALLD:NM_016081:exon20:c.G3314A:p.R1105Q | | |
| VS8 | chr4:169799 408 | G > C | nonsynonymous; PALLD:NM_001166110:exon2:c.G366C:p.Q122H | benign | 4,4 \| 7,7 |
| | | | | | |
| | | | | | |

| RRM2B - ribonucleoside-diphosphate reductase subunit M2 B | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr8:103250840 | - > G | frameshift insertion; RRM2B:NM_001172477:exon1:c.211_212insC:p.R71fs | | 13,7 \| 6,7 |
| VS8 | chr8:103250840 | - > G | frameshift insertion; RRM2B:NM_001172477:exon1:c.211_212insC:p.R71fs | | 10,3 \| 5,1 |
| | | | | | |
| | | | | | |
| VIL1 - villin-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr2:219296838 | G > A | nonsynonymous; VIL1:NM_007127:exon12:c.G1273A:p.D425N | probably damaging | 11,20 \| 10,12 |
| VS3 | chr2:219290479 | G > T | stopgain; VIL1:NM_007127:exon4:c.G292T:p.E98X | | 2,8 \| 3,0 |
| | | | | | |
| | | | | | |
| MYO7A - myosin-VIIa | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS10 | chr11:76905558 | G > A | nonsynonymous; MYO7A:NM_000260:exon32:c.G4312A:p.A1438T | benign | 9,9 \| 5,8 |
| | | | MYO7A:NM_001127180:exon32:c.G4312A:p.A1438T | | |
| VS10 | chr11:76919766 | A > G | nonsynonymous; MYO7A:NM_000260:exon44:c.A5969G:p.Q1990R | possibly damaging | 6,15 \| 8,14 |
| | | | MYO7A:NM_001127180:exon44:c.A5855G:p.Q1952R | | |
| VS13 | chr11:76871254 | A > G | nonsynonymous; MYO7A:NM_001127179:exon11:c.A1126G:p.I376V | benign | 8,8 \| 12,6 |
| | | | MYO7A:NM_000260:exon11:c.A1126G:p.I376V | | |
| | | | MYO7A:NM_001127180:exon11:c.A1126G:p.I376V | | |
| VS13 | chr11:76903159 | G > A | nonsynonymous; MYO7A:NM_000260:exon31:c.G3988A:p.A1330T | probably damaging | 9,7 \| 8,6 |
| | | | MYO7A:NM_001127180:exon31:c.G3988A:p.A1330T | | |
| | | | | | |
| | | | | | |
| ATN1 - atrophin-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr12:7045934 | - > CGC | nonframeshift insertion; ATN1:NM_001940:exon5:c.1504_1505insCGC:p.Q502delinsPQ | | 42,40 \| 9,26 |
| | | | ATN1:NM_001007026:exon5:c.1504_1505insCGC:p.Q502delinsPQ | | |
| VS3 | chr12:7045136 | A > C | nonsynonymous; ATN1:NM_001940:exon5:c.A706C:p.K236Q | benign | 3,3 \| 5,9 |
| | | | ATN1:NM_001007026:exon5:c.A706C:p.K236Q | | |
| | | | | | |
| | | | | | |
| CNTNAP5 - contactin-associated protein-like 5 precursor | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr2:125504872 | C > G | nonsynonymous; CNTNAP5:NM_130773:exon14:c.C2141G:p.P714R | benign | 11,1 \| 11,3 |
| VS5 | chr2:125405388 | C > T | nonsynonymous; CNTNAP5:NM_130773:exon13:c.C1927T:p.R643W | possibly damaging | 1,1 \| 2,3 |
| | | | | | |
| | | | | | |
| NAALAD2 - N-acetylated-alpha-linked acidic dipeptidase 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr11:89924875 | C > T | nonsynonymous; NAALAD2:NM_005467:exon19:c.C2183T:p.T728I | probably damaging | 10,5 \| 6,4 |
| VS5 | chr11:89885490 | G > T | nonsynonymous; NAALAD2:NM_005467:exon6:c.G634T:p.A212S | probably damaging | 16,5 \| 4,0 |
| | | | | | |
| | | | | | |
| LGALS9C - galectin-9C | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr17:18397546 | G > C | nonsynonymous; LGALS9C:NM_001040078:exon11:c.G936C:p.L312F | possibly damaging | 21,16 \| 11,6 |

| | | | | | |
|---|---|---|---|---|---|
| VS6 | chr17:18397546 | G > C | nonsynonymous; LGALS9C:NM_001040078:exon11:c.G936C:p.L312F | possibly damaging | 12,3 \| 9,6 |
| | | | | | |
| | | | | | |
| GRN - granulins | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr17:42428086 | C > T | nonsynonymous; GRN:NM_002087:exon7:c.C626T:p.P209L | probably damaging | 11,21 \| 5,13 |
| VS8 | chr17:42430069 | G > A | nonsynonymous; GRN:NM_002087:exon13:c.G1685A:p.G562D | probably damaging | 16,17 \| 13,27 |
| | | | | | |
| | | | | | |
| WDR96 - WD repeat-containing protein C10orf79 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr10:105923869 | C > T | nonsynonymous; WDR96:NM_025145:exon24:c.G3229A:p.V1077I | benign | 18,12 \| 21,8 |
| VS5 | chr10:105932284 | T > C | nonsynonymous; WDR96:NM_025145:exon20:c.A2470G:p.M824V | possibly damaging | 0,19 \| 3,16 |
| | | | | | |
| | | | | | |
| DDHD1 - phospholipase DDHD1 | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| VS10 | chr14:53619494 | - > CACCGC | nonframeshift insertion; DDHD1:NM_030637:exon1:c.323_324insGCGGTG:p.S108delinsSGG | | 4,5 \| 9,25 |
| | | | DDHD1:NM_001160147:exon1:c.323_324insGCGGTG:p.S108delinsSGG | | |
| | | | DDHD1:NM_001160148:exon1:c.323_324insGCGGTG:p.S108delinsSGG | | |
| VS9 | chr14:53619490 | - > GCGCCG | nonframeshift insertion; DDHD1:NM_001160148:exon1:c.327_328insCGGCGC:p.G109delinsGGA | | 4,15 \| 8,20 |
| | | | DDHD1:NM_030637:exon1:c.327_328insCGGCGC:p.G109delinsGGA | | |
| | | | DDHD1:NM_001160147:exon1:c.327_328insCGGCGC:p.G109delinsGGA | | |
| | | | | | |
| | | | | | |
| TXNDC5 - thioredoxin domain-containing protein 5 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr6:7910890 | - > GCC | nonframeshift insertion; TXNDC5:NM_030810:exon1:c.120_121insGGC:p.E40delinsEA | | 3,11 \| 5,15 |
| VS9 | chr6:7910888 | GCC > - | nonframeshift deletion; TXNDC5:NM_030810:exon1:c.120_122del:p.40_41del | | 1,10 \| 1,4 |
| | | | | | |
| | | | | | |
| OPLAH - 5-oxoprolinase | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS12 | chr8:145109572 | C > T | nonsynonymous; OPLAH:NM_017570:exon19:c.G2578A:p.G860R | probably damaging | 9,24 \| 4,25 |
| VS3 | chr8:145106943 | CC > - | frameshift deletion; OPLAH:NM_017570:exon25:c.3497_3498del:p.1166_1166del | | 0,0 \| 7,0 |
| | | | | | |
| | | | | | |
| GYLTL1B - glycosyltransferase-like protein LARGE2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS6 | chr11:45947819 | G > A | nonsynonymous; GYLTL1B:NM_152312:exon8:c.G929A:p.R310H | possibly damaging | 5,0 \| 17,0 |
| VS8 | chr11:45948991 | G > C | nonsynonymous; GYLTL1B:NM_152312:exon11:c.G1451C:p.R484P | probably damaging | 33,17 \| 30,14 |
| | | | | | |
| | | | | | |
| MAP2 - microtubule-associated protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr2:210557420 | G > A | nonsynonymous; MAP2:NM_002374:exon7:c.G526A:p.D176N | benign | 25,12 \| 26,7 |

| | | | | | |
|---|---|---|---|---|---|
| VS9 | chr2:210558830 | T > C | nonsynonymous; MAP2:NM_002374:exon7:c.T1936C:p.S646P | benign | 29,19 \| 21,16 |
| | | | | | |
| | | | | | |
| NEB - nebulin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr2:152529122 | T > C | nonsynonymous; NEB:NM_001164508:exon37:c.A4060G:p.K1354E | probably damaging | 25,25 \| 26,32 |
| | | | NEB:NM_001164507:exon37:c.A4060G:p.K1354E | | |
| | | | NEB:NM_004543:exon37:c.A4060G:p.K1354E | | |
| VS12 | chr2:152411477 | G > A | nonsynonymous; NEB:NM_001164508:exon125:c.C19396T:p.R6466W | probably damaging | 6,3 \| 7,0 |
| | | | NEB:NM_001164507:exon125:c.C19396T:p.R6466W | | |
| | | | NEB:NM_004543:exon97:c.C14257T:p.R4753W | | |
| | | | | | |
| | | | | | |
| MAP6D1 - MAP6 domain-containing protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr3:183543205 | C > T | nonsynonymous; MAP6D1:NM_024871:exon1:c.G131A:p.G44D | benign | 1,6 \| 3,3 |

| | | | | | |
|---|---|---|---|---|---|
| VS8 | chr3:183543070 | C > A | nonsynonymous; MAP6D1:NM_024871:exon1:c.G266T:p.R89L | benign | 5,12 \| 0,3 |
| | | | | | |
| | | | | | |
| TEX15 - testis-expressed sequence 15 protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr8:30694465 | A > G | nonsynonymous; TEX15:NM_031271:exon3:c.T8186C:p.V2729A | benign | 18,30 \| 21,20 |
| VS8 | chr8:30700839 | CT > - | frameshift deletion; TEX15:NM_031271:exon1:c.5694_5695del:p.1898_1899del | | 35,13 \| 37,14 |
| | | | | | |
| | | | | | |
| SUSD4 - sushi domain-containing protein 4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr1:223438092 | C > T | nonsynonymous; SUSD4:NM_001037175:exon5:c.G604A:p.V202M | possibly damaging | 18,18 \| 15,23 |
| | | | SUSD4:NM_017982:exon5:c.G604A:p.V202M | | |
| VS11 | chr1:223441918 | C > T | nonsynonymous; SUSD4:NM_017982:exon4:c.G461A:p.R154Q | probably damaging | 11,7 \| 9,15 |
| | | | SUSD4:NM_001037175:exon4:c.G461A:p.R154Q | | |

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|---|---|---|---|---|---|
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| PCDHA10 - protocadherin alpha-10 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr5:140238008 | A > G | nonsynonymous; PCDHA10:NM_018901:exon1:c.A2375G:p.D792G | benign | 14,26 \| 10,22 |
| | | | PCDHA10:NM_031859:exon1:c.A2375G:p.D792G | | |
| VS9 | chr5:140237413 | C > A | nonsynonymous; PCDHA10:NM_018901:exon1:c.C1780A:p.R594S | benign | 19,5 \| 2,2 |
| | | | PCDHA10:NM_031859:exon1:c.C1780A:p.R594S | | |
| | | | | | |
| | | | | | |
| ASXL3 - putative Polycomb group protein ASXL3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr18:31325384 | C > G | nonsynonymous; ASXL3:NM_030632:exon12:c.C5572G:p.P1858A | benign | 33,10 \| 39,5 |
| VS6 | chr18:31320333 | C > G | nonsynonymous; ASXL3:NM_030632:exon11:c.C2965G:p.R989G | possibly damaging | 3,2 \| 4,3 |
| | | | | | |
| | | | | | |
| TTC34 - tetratricopeptide repeat protein 34 | | | | | |

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|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr1:2572985 | C > T | unknown; UNKNOWN | | 6,6 \| 4,8 |
| VS12 | chr1:2573220 | G > C | unknown; UNKNOWN | | 4,4 \| 3,10 |
| | | | | | |
| | | | | | |
| ADRA2B - alpha-2B adrenergic receptor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr2:96781791 | A > G | nonsynonymous; ADRA2B:NM_000682:exon1:c.T98C:p.V33A | probably damaging | 4,22 \| 6,25 |
| VS8 | chr2:96780898 | G > A | nonsynonymous; ADRA2B:NM_000682:exon1:c.C991T:p.R331W | probably damaging | 26,18 \| 25,15 |
| | | | | | |
| | | | | | |
| SECISBP2 - selenocysteine insertion sequence-binding | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

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|---|---|---|---|---|---|
| VS10 | chr9:91964721 | C > G | nonsynonymous; SECISBP2:NM_024077:exon13:c.C1769G:p.T590S | benign | 28,15 \| 20,4 |
| VS6 | chr9:91943595 | G > C | nonsynonymous; SECISBP2:NM_024077:exon5:c.G595C:p.D199H | possibly damaging | 14,0 \| 18,3 |
| | | | | | |
| | | | | | |
| DMXL2 - dmX-like protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr15:51741245 | G > A | nonsynonymous; DMXL2:NM_015263:exon43:c.C9047T:p.T3016M | probably damaging | 6,4 \| 1,11 |
| | | | DMXL2:NM_001174117:exon41:c.C7139T:p.T2380M | | |
| | | | DMXL2:NM_001174116:exon43:c.C9050T:p.T3017M | | |
| VS8 | chr15:51741375 | C > A | nonsynonymous; DMXL2:NM_001174116:exon43:c.G8920T:p.G2974C | probably damaging | 2,10 \| 5,0 |
| | | | DMXL2:NM_015263:exon43:c.G8917T:p.G2973C | | |
| | | | DMXL2:NM_001174117:exon41:c.G7009T:p.G2337C | | |
| | | | | | |
| | | | | | |
| PLXNC1 - plexin-C1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr12:94620982 | A > C | nonsynonymous; PLXNC1:NM_005761:exon9:c.A1951C:p.K651Q | benign | 6,16 \| 5,14 |

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|---|---|---|---|---|---|
| VS8 | chr12:94653423 | C > A | nonsynonymous; PLXNC1:NM_005761:exon19:c.C3164A:p.A1055D | benign | 10,2 \| 3,0 |
| | | | | | |
| | | | | | |
| APLF - aprataxin and PNK-like factor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr2:68805146 | A > - | frameshift deletion; APLF:NM_173545:exon10:c.1528delA:p.R510fs | | 0,25 \| 1,16 |
| VS3 | chr2:68805127 | A > T | nonsynonymous; APLF:NM_173545:exon10:c.A1509T:p.E503D | probably damaging | 0,24 \| 1,13 |
| | | | | | |
| | | | | | |
| TBP - TATA-box-binding protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr6:170871028 | - > TAG | stopgain; TBP:NM_001172085:exon2:c.144_145insTAG:p.Q48delinsQX | | 5,7 \| 20,12 |
| | | | TBP:NM_003194:exon3:c.204_205insTAG:p.Q68delinsQX | | |
| VS14 | chr6:170871030 | - > ACA | nonframeshift insertion; TBP:NM_001172085:exon2:c.146_147insACA:p.Q49delinsQQ | | 8,7 \| 13,16 |
| | | | TBP:NM_003194:exon3:c.206_207insACA:p.Q69delinsQQ | | |
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|---|---|---|---|---|---|
| SLC2A5 - solute carrier family 2, facilitated glucose | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS5 | chr1:9100209 | G > A | nonsynonymous; SLC2A5:NM_003039:exon6:c.C611T:p.A204V | benign | 0,0 \| 0,3 |
| | | | SLC2A5:NR_024180:exon8:c.C779T:p.A260V | | |
| VS9 | chr1:9098957 | AGC > - | nonframeshift deletion; SLC2A5:NM_003039:exon9:c.1026_1028del:p.342_343del | | 16,13 \| 4,0 |
| | | | | | |
| | | | | | |
| SHISA7 - protein shisa-7 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr19:55953906 | T > G | nonsynonymous; SHISA7:NM_001145176:exon1:c.A325C:p.T109P | probably damaging | 0,7 \| 4,0 |
| VS13 | chr19:55954062 | C > A | nonsynonymous; SHISA7:NM_001145176:exon1:c.G169T:p.G57C | benign | 0,6 \| 0,3 |
| | | | | | |
| | | | | | |
| TUSC1 - tumor suppressor candidate gene 1 protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

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|---|---|---|---|---|---|
| VS11 | chr9:25678195 | - > CGC | nonframeshift insertion; TUSC1:NM_001004125:exon1:c.125_126insGCG:p.S42delinsSG | | 12,32 \| 16,16 |
| VS8 | chr9:25678195 | - > CGC | nonframeshift insertion; TUSC1:NM_001004125:exon1:c.125_126insGCG:p.S42delinsSG | | 4,15 \| 8,13 |
| | | | | | |
| | | | | | |
| DNAH11 - dynein heavy chain 11, axonemal | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr7:21906238 | C > A | nonsynonymous; DNAH11:NM_003777:exon71:c.C11669A:p.S3890Y | benign | 11,14 \| 2,2 |
| VS5 | chr7:21818707 | T > C | nonsynonymous; DNAH11:NM_003777:exon57:c.T9490C:p.C3164R | | 1,3 \| 1,3 |
| | | | | | |
| | | | | | |
| FRYL - protein furry homolog-like | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr4:48553541 | T > A | nonsynonymous; FRYL:NM_015030:exon37:c.A4490T:p.N1497I | benign | 11,5 \| 17,2 |
| VS8 | chr4:48555361 | T > C | nonsynonymous; FRYL:NM_015030:exon36:c.A4306G:p.M1436V | benign | 13,30 \| 7,28 |
| | | | | | |
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|---|---|---|---|---|---|
| SLC38A3;SLC38A3 - sodium-coupled neutral amino acid transporter 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS5 | chr3:50251835 | - > G | frameshift insertion; SLC38A3:NM_006841:exon3:c.103_104insG:p.V35fs | | 0,0 \| 1,3 |
| VS6 | chr3:50251835 | - > G | frameshift insertion; SLC38A3:NM_006841:exon3:c.103_104insG:p.V35fs | | 0,0 \| 1,2 |
| | | | | | |
| | | | | | |
| DOCK7 - dedicator of cytokinesis protein 7 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr1:63021589 | T > G | nonsynonymous; DOCK7:NM_033407:exon21:c.A2503C:p.N835H | benign | 14,18 \| 10,7 |
| VS11 | chr1:63021570 | C > A | nonsynonymous; DOCK7:NM_033407:exon21:c.G2522T:p.R841I | probably damaging | 19,27 \| 4,3 |
| | | | | | |
| | | | | | |
| DNHD1 - dynein heavy chain domain-containing protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS14 | chr11:6585158 | G > A | nonsynonymous; DNHD1:NM_144666:exon31:c.G10088A:p.R3363H | benign | 18,18 \| 22,15 |
| VS9 | chr11:6585585 | C > G | nonsynonymous; DNHD1:NM_144666:exon32:c.C10307G:p.T3436S | possibly damaging | 36,7 \| 27,8 |
| | | | | | |
| | | | | | |
| PABPC3 - polyadenylate-binding protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr13:25671456 | C > T | nonsynonymous; PABPC3:NM_030979:exon1:c.C1120T:p.R374C | benign | 30,54 \| 7,14 |
| VS3 | chr13:25671456 | C > T | nonsynonymous; PABPC3:NM_030979:exon1:c.C1120T:p.R374C | benign | 32,32 \| 2,10 |
| | | | | | |
| | | | | | |
| COL16A1 - collagen alpha-1(XVI) chain precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chr1:32138025 | CTC > - | nonframeshift deletion; COL16A1:NM_001856:exon47:c.3095_3097del:p.1032_1033del | | 72,37 \| 11,4 |
| VS3 | chr1:32158204 | C > T | nonsynonymous; COL16A1:NM_001856:exon15:c.G1153A:p.A385T | unknown | 5,4 \| 1,2 |
| | | | | | |
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|---|---|---|---|---|---|
| PPM1E - protein phosphatase 1E | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr17:56833479 | - > ACGAAC | nonframeshift insertion; PPM1E:NM_014906:exon1:c.121_122insACGAAC:p.P41delinsHEP | | 21,44 \| 19,53 |
| VS8 | chr17:56833471 | - > CCCCGA | nonframeshift insertion; PPM1E:NM_014906:exon1:c.113_114insCCCCGA:p.E38delinsDPE | | 16,27 \| 33,43 |
| | | | | | |
| | | | | | |
| NPHP4 - nephrocystin-4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr1:6008220 | G > A | nonsynonymous; NPHP4:NM_015102:exon8:c.C902T:p.P301L | possibly damaging | 6,20 \| 12,15 |
| VS3 | chr1:5935064 | T > C | nonsynonymous; NPHP4:NM_015102:exon21:c.A2914G:p.S972G | possibly damaging | 0,3 \| 0,4 |
| | | | | | |
| | | | | | |
| AKAP13 - A-kinase anchor protein 13 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS12 | chr15:86124333 | G > T | nonsynonymous; AKAP13:NM_006738:exon7:c.G3034T:p.G1012W | benign | 11,16 \| 10,13 |
| | | | AKAP13:NM_007200:exon7:c.G3034T:p.G1012W | | |
| VS3 | chr15:86228067 | A > G | nonsynonymous; AKAP13:NM_007200:exon16:c.A5252G:p.Y1751C | probably damaging | 16,14 \| 9,11 |
| | | | AKAP13:NM_006738:exon16:c.A5264G:p.Y1755C | | |
| | | | | | |
| | | | | | |
| PTCHD3 - patched domain-containing protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr10:27702262 | - > G | frameshift insertion; PTCHD3:NM_001034842:exon1:c.918_919insC:p.L306fs | | 21,15 \| 19,22 |
| VS6 | chr10:27702786 | A > C | nonsynonymous; PTCHD3:NM_001034842:exon1:c.T394G:p.W132G | benign | 3,3 \| 0,3 |
| | | | | | |
| | | | | | |
| SCARA3 - scavenger receptor class A member 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS3 | chr8:27528567 | T > C | nonsynonymous; SCARA3:NM_016240:exon6:c.T1520C:p.V507A | benign | 0,6 \| 0,4 |
| VS8 | chr8:27516048 | G > C | nonsynonymous; SCARA3:NM_182826:exon5:c.G361C:p.E121Q | benign | 12,9 \| 14,7 |
| | | | SCARA3:NM_016240:exon5:c.G361C:p.E121Q | | |
| | | | | | |
| | | | | | |
| TMC1 - transmembrane channel-like protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 14,1 \| 10,2 |
| VS5 | chr9:75315444 | GAA > - | nonframeshift deletion; TMC1:NM_138691:exon8:c.247_249del:p.83_83del | | 9,3 \| 3,5 |
| | | | | | |
| | | | | | |
| BRWD1 - bromodomain and WD repeat-containing protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr21:40685411 | C > T | nonsynonymous; BRWD1:NM_001007246:exon1:c.G7A:p.E3K | benign | 5,15 \| 8,9 |
| | | | BRWD1:NM_018963:exon1:c.G7A:p.E3K | | |
| | | | BRWD1:NM_033656:exon1:c.G7A:p.E3K | | |
| VS8 | chr21:40668277 | G > C | nonsynonymous; BRWD1:NM_018963:exon6:c.C362G:p.T121R | possibly damaging | 4,1 \| 5,2 |

| | | | | | |
|---|---|---|---|---|---|
| | | | BRWD1:NM_033656:exon6:c.C362G:p.T121R | | |
| | | | | | |
| | | | | | |
| KIAA1211 - hypothetical protein LOC57482 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr4:57182721 | C > T | nonsynonymous; KIAA1211:NM_020722:exon8:c.C3053T:p.P1018L | possibly damaging | 3,2 \| 3,3 |
| VS8 | chr4:57180639 | A > G | nonsynonymous; KIAA1211:NM_020722:exon8:c.A971G:p.Q324R | benign | 1,2 \| 3,0 |
| | | | | | |
| | | | | | |
| ITGB4 - integrin beta-4 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr17:73736913 | C > A | nonsynonymous; ITGB4:NM_001005619:exon21:c.C2590A:p.L864I | benign | 6,11 \| 3,0 |
| | | | ITGB4:NM_000213:exon22:c.C2590A:p.L864I | | |
| | | | ITGB4:NM_001005731:exon22:c.C2590A:p.L864I | | |
| VS3 | chr17:73735778 | G > A | nonsynonymous; ITGB4:NM_000213:exon19:c.G2246A:p.C749Y | probably damaging | 9,6 \| 0,3 |
| | | | ITGB4:NM_001005619:exon18:c.G2246A:p.C749Y | | |
| | | | ITGB4:NM_001005731:exon19:c.G2246A:p.C749Y | | |
| | | | | | |

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|---|---|---|---|---|---|
| C13orf23 - unknown | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr13:39602426 | T > C | nonsynonymous; C13orf23:NM_025138:exon5:c.A307G:p.I103V | benign | 17,50 \| 15,45 |
| | | | C13orf23:NM_170719:exon4:c.A241G:p.I81V | | |
| VS8 | chr13:39587285 | A > T | nonsynonymous; C13orf23:NM_025138:exon11:c.T2104A:p.S702T | probably damaging | 20,16 \| 14,22 |
| | | | C13orf23:NM_170719:exon10:c.T2038A:p.S680T | | |
| | | | | | |
| | | | | | |
| CWH43 - PGAP2-interacting protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS6 | chr4:49063895 | A > - | frameshift deletion; CWH43:NM_025087:exon16:c.2088delA:p.K696fs | | 3,6 \| 4,7 |
| VS8 | chr4:48994008 | A > G | nonsynonymous; CWH43:NM_025087:exon4:c.A412G:p.I138V | benign | 8,3 \| 3,0 |
| | | | | | |
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|---|---|---|---|---|---|
| MYH14 - myosin-14 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chr19:50726550 | C > A | nonsynonymous; MYH14:NM_024729:exon5:c.C637A:p.Q213K | probably damaging | 9,52 \| 13,39 |
| | | | MYH14:NM_001077186:exon5:c.C637A:p.Q213K | | |
| | | | MYH14:NM_001145809:exon5:c.C637A:p.Q213K | | |
| VS3 | chr19:50780078 | G > A | nonsynonymous; MYH14:NM_024729:exon27:c.G3622A:p.E1208K | possibly damaging | 1,10 \| 4,8 |
| | | | MYH14:NM_001077186:exon28:c.G3646A:p.E1216K | | |
| | | | MYH14:NM_001145809:exon29:c.G3745A:p.E1249K | | |
| | | | | | |
| | | | | | |
| HLA-DRB1 - major histocompatibility complex, class II, DR beta 1 precursor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr6:32552043 | G > A | nonsynonymous; HLA-DRB1:NM_002124:exon2:c.G213T:p.E71D | | 3,19 \| 0,6 |
| VS3 | chr6:32549362 | G > A | nonsynonymous; HLA-DRB1:NM_002124:exon3:c.G624T:p.Q208H | | 30,16 \| 11,12 |
| | | | | | |
| | | | | | |
| SIGLEC12 - sialic acid-binding Ig-like lectin 12 | | | | | |

| | | | | | |
|---|---|---|---|---|---|
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 0,0 \| 10,27 |
| VS6 | chr19:52004794 | - > C | frameshift insertion; SIGLEC12:NM_053003:exon1:c.194_195insG:p.R65fs | | 0,0 \| 13,34 |
| | | | | | |
| | | | | | |
| ABCB1 - multidrug resistance protein 1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr7:87183217 | T > C | nonsynonymous; ABCB1:NM_000927:exon10:c.A859G:p.I287V | benign | 3,17 \| 6,27 |
| VS9 | chr7:87165788 | C > T | nonsynonymous; ABCB1:NM_000927:exon21:c.G2467A:p.A823T | benign | 24,14 \| 33,11 |
| | | | | | |
| | | | | | |
| FAM21C - WASH complex subunit FAM21C | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS3 | chr10:46245557 | T > C | nonsynonymous; FAM21C:NM_001169106:exon9:c.T746C:p.M249T | benign | 24,8 \| 12,1 |
| | | | FAM21C:NM_015262:exon9:c.T746C:p.M249T | | |
| | | | FAM21C:NM_001169107:exon9:c.T746C:p.M249T | | |
| VS5 | chr10:46245557 | T > C | nonsynonymous; FAM21C:NM_001169106:exon9:c.T746C:p.M249T | benign | 16,4 \| 9,4 |
| | | | FAM21C:NM_015262:exon9:c.T746C:p.M249T | | |
| | | | FAM21C:NM_001169107:exon9:c.T746C:p.M249T | | |
| | | | | | |
| | | | | | |
| ZNF469 - zinc finger protein 469 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr16:88495913 | G > A | nonsynonymous; ZNF469:NM_001127464:exon1:c.G2035A:p.E679K | benign | 39,16 \| 35,5 |
| VS14 | chr16:88501430 | C > T | nonsynonymous; ZNF469:NM_001127464:exon2:c.C7468T:p.P2490S | possibly damaging | 9,30 \| 5,36 |
| | | | | | |
| | | | | | |
| MRO - protein maestro | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr18:48327781 | C > T | nonsynonymous; MRO:NM_001127176:exon5:c.G565A:p.V189I | probably damaging | 4,4 \| 4,2 |

| | | | | | |
|---|---|---|---|---|---|
| | | | MRO:NM_031939:exon6:c.G523A:p.V175I | | |
| VS12 | chr18:48331614 | AG > - | frameshift deletion;<br>MRO:NM_001127176:exon4:c.380_381del:p.127_127del | | 11,16 \| 20,8 |
| | | | MRO:NM_001127175:exon4:c.380_381del:p.127_127del | | |
| | | | MRO:NM_031939:exon5:c.338_339del:p.113_113del | | |
| | | | MRO:NM_001127174:exon4:c.338_339del:p.113_113del | | |
| | | | | | |
| | | | | | |
| ABCA13 - ATP-binding cassette sub-family A member 13 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr7:48315459 | A > T | nonsynonymous;<br>ABCA13:NM_152701:exon17:c.A6196T:p.M2066L | benign | 53,30 \| 35,24 |
| VS10 | chr7:48682937 | A > G | nonsynonymous;<br>ABCA13:NM_152701:exon60:c.A14891G:p.H4964R | benign | 14,12 \| 12,7 |
| VS3 | chr7:48452070 | C > A | nonsynonymous;<br>ABCA13:NM_152701:exon41:c.C12349A:p.P4117T | probably damaging | 7,3 \| 11,6 |
| | | | | | |
| | | | | | |
| C14orf21 - pumilio domain-containing protein C14orf21 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr14:24769865 | - > CGGAGG | nonframeshift insertion;<br>C14orf21:NM_174913:exon2:c.499_500insCGGAGG:p.E167delinsAEE | | 11,13 \| 9,3 |

| | | | | | |
|---|---|---|---|---|---|
| VS12 | chr14:24769873 | - > CAGGAGGAG | nonframeshift insertion; C14orf21:NM_174913:exon2:c.507_508insCAGGAGGAG:p.E169delinsEQEE | | 22,10 \| 8,3 |
| | | | | | |
| | | | | | |
| IL17RC - interleukin-17 receptor C | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr3:9970067 | C > T | nonsynonymous; IL17RC:NM_001203264:exon11:c.C956T:p.P319L | benign | 29,12 \| 26,19 |
| | | | IL17RC:NM_153460:exon11:c.C956T:p.P319L | | |
| | | | IL17RC:NM_001203263:exon11:c.C956T:p.P319L | | |
| | | | IL17RC:NM_153461:exon11:c.C1169T:p.P390L | | |
| | | | IL17RC:NM_032732:exon10:c.C911T:p.P304L | | |
| | | | IL17RC:NM_001203265:exon10:c.C911T:p.P304L | | |
| VS3 | chr3:995895 6 | - > G | frameshift insertion; IL17RC:NR_037807:exon1:c.199_200insG:p.A67fs | | 2,0 \| 3,0 |
| | | | | | |
| | | | | | |
| SOX1 - transcription factor SOX-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS12 | chr13:112722454 | TGGGCG > - | nonframeshift deletion; SOX1:NM_005986:exon1:c.482_487del:p.161_163del | | 6,13 \| 2,13 |
| VS9 | chr13:112722454 | TGGGCG > - | nonframeshift deletion; SOX1:NM_005986:exon1:c.482_487del:p.161_163del | | 0,9 \| 0,5 |
| | | | | | |
| | | | | | |
| BHLHB9 - protein BHLHb9 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chrX:102004021 | T > G | nonsynonymous; BHLHB9:NM_001142525:exon4:c.T98G:p.V33G | benign | 31,12 \| 31,20 |
| | | | BHLHB9:NM_001142530:exon2:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_001142524:exon4:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_001142528:exon4:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_001142527:exon4:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_001142529:exon3:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_001142526:exon4:c.T98G:p.V33G | | |
| | | | BHLHB9:NM_030639:exon3:c.T98G:p.V33G | | |
| VS9 | chrX:102004743 | T > C | nonsynonymous; BHLHB9:NM_001142525:exon4:c.T820C:p.C274R | benign | 29,45 \| 26,39 |
| | | | BHLHB9:NM_001142530:exon2:c.T820C:p.C274R | | |
| | | | BHLHB9:NM_001142524:exon4:c.T820C:p.C274R | | |
| | | | BHLHB9:NM_001142528:exon4:c.T820C:p.C274R | | |
| | | | BHLHB9:NM_001142527:exon4:c.T820C:p.C274R | | |
| | | | BHLHB9:NM_001142529:exon3:c.T820C:p.C274R | | |

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|---|---|---|---|---|---|
| | | | BHLHB9:NM_001142526:exon4:c.T820C:p.C274R | | |
| | | | BHLHB9:NM_030639:exon3:c.T820C:p.C274R | | |
| | | | | | |
| | | | | | |
| TNS3 - tensin-3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr7:47408378 | A > G | nonsynonymous; TNS3:NM_022748:exon17:c.T1865C:p.L622P | benign | 12,7 \| 3,12 |
| VS6 | chr7:47408712 | T > G | nonsynonymous; TNS3:NM_022748:exon17:c.A1531C:p.T511P | benign | 0,7 \| 3,0 |
| | | | | | |
| | | | | | |
| FAM120C - constitutive coactivator of PPAR-gamma-like protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chrX:54209317 | - > GGCGGC | nonframeshift insertion; FAM120C:NM_198456:exon1:c.315_316insGCCGCC:p.L105delinsLPP | | 7,14 \| 1,12 |
| | | | FAM120C:NM_017848:exon1:c.315_316insGCCGCC:p.L105delinsLPP | | |
| VS14 | chrX:54209317 | - > GGCGGC | nonframeshift insertion; FAM120C:NM_198456:exon1:c.315_316insGCCGCC:p.L105delinsLPP | | 3,18 \| 8,13 |
| | | | FAM120C:NM_017848:exon1:c.315_316insGCCGCC:p.L105delinsLPP | | |
| | | | | | |
| | | | | | |

| DNAH7 - dynein heavy chain 7, axonemal | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr2:196749503 | C > T | nonsynonymous; DNAH7:NM_018897:exon35:c.G5569A:p.V1857I | benign | 4,19 \| 8,18 |
| VS3 | chr2:196851841 | C > T | nonsynonymous; DNAH7:NM_018897:exon14:c.G1703A:p.C568Y | benign | 2,39 \| 4,27 |
| | | | | | |
| | | | | | |
| FAM83G - family with sequence similarity 83, member G | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr17:18881189 | G > A | nonsynonymous; FAM83G:NM_001039999:exon5:c.C1790T:p.S597L | possibly damaging | 42,32 \| 46,51 |
| VS3 | chr17:18874794 | C > G | nonsynonymous; FAM83G:NM_001039999:exon6:c.G2350C:p.G784R | probably damaging | 9,10 \| 16,6 |
| | | | | | |
| | | | | | |
| C3orf25 - EF-hand domain-containing protein C3orf25 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |

| | | | | | |
|---|---|---|---|---|---|
| VS13 | chr3:129140275 | GT > - | frameshift deletion; C3orf25:NM_207307:exon2:c.420_421del:p.140_141del | | 37,11 \| 30,11 |
| VS14 | chr3:129140263 | C > T | nonsynonymous; C3orf25:NM_207307:exon2:c.G433A:p.E145K | benign | 32,18 \| 25,11 |
| | | | | | |
| | | | | | |
| FAM82A2;FAM82A2 - regulator of microtubule dynamics protein 3 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr15:41029923 | A > C | nonsynonymous; FAM82A2:NM_018145:exon10:c.T1127G:p.V376G | probably damaging | 3,6 \| 0,5 |
| VS6 | chr15:41029923 | A > C | nonsynonymous; FAM82A2:NM_018145:exon10:c.T1127G:p.V376G | probably damaging | 14,2 \| 0,6 |
| | | | | | |
| | | | | | |
| CALHM2 - calcium homeostasis modulator protein 2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS2 | chr10:105209357 | AG > - | frameshift deletion; CALHM2:NR_024552:exon3:c.849_0del:p.283_0del | | 1,6 \| 2,4 |
| | | | CALHM2:NM_015916:exon3:c.341_342del:p.114_114del | | |
| VS5 | chr10:105209357 | AG > - | frameshift deletion; CALHM2:NR_024552:exon3:c.849_0del:p.283_0del | | 5,6 \| 2,7 |

| | | | | | |
|---|---|---|---|---|---|
| | | | CALHM2:NM_015916:exon3:c.341_342del:p.114_114del | | |
| | | | | | |
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| ATXN1 - ataxin-1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr6:16327850 | G > A | nonsynonymous; ATXN1:NM_000332:exon8:c.C692T:p.P231L | benign | 8,14 \| 2,11 |
| | | | ATXN1:NM_001128164:exon7:c.C692T:p.P231L | | |
| VS14 | chr6:16327323 | G > C | nonsynonymous; ATXN1:NM_000332:exon8:c.C1219G:p.P407A | probably damaging | 15,19 \| 12,21 |
| | | | ATXN1:NM_001128164:exon7:c.C1219G:p.P407A | | |
| | | | | | |
| | | | | | |
| NRG2 - Nrg2p | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr5:139422558 | - > GCTGCTGCT | nonframeshift insertion; NRG2:NM_013982:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | 4,6 \| 6,4 |
| | | | NRG2:NM_013981:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_004883:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_001184935:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_013983:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |

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|---|---|---|---|---|---|
| VS8 | chr5:139422558 | - > GCTGCTGCT | nonframeshift insertion; NRG2:NM_013982:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | 6,8 \| 10,8 |
| | | | NRG2:NM_013981:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_004883:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_001184935:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | NRG2:NM_013983:exon1:c.97_98insAGCAGCAGC:p.S33delinsSSSS | | |
| | | | | | |
| | | | | | |
| FOXD1 - forkhead box protein D1 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr5:72743299 | - > GC | frameshift insertion; FOXD1:NM_004472:exon1:c.889_890insGC:p.R297fs | | 0,0 \| 2,1 |
| VS8 | chr5:72743299 | - > GC | frameshift insertion; FOXD1:NM_004472:exon1:c.889_890insGC:p.R297fs | | 0,0 \| 3,3 |
| | | | | | |
| | | | | | |
| PLA2G4F - cytosolic phospholipase A2 zeta | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS3 | chr15:42434285 | A > C | nonsynonymous; PLA2G4F:NM_213600:exon20:c.T2447G:p.V816G | probably damaging | 9,4 \| 0,7 |
| VS6 | chr15:42442662 | T > C | nonsynonymous; PLA2G4F:NM_213600:exon9:c.A794G:p.E265G | benign | 2,0 \| 2,1 |

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|---|---|---|---|---|---|
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| ACVR2A - activin receptor type-2A | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS8 | chr2:148676144 | A > C | nonsynonymous; ACVR2A:NM_001616:exon7:c.A945C:p.K315N | probably damaging | 11,6 \| 0,9 |
| VS9 | chr2:148676144 | A > C | nonsynonymous; ACVR2A:NM_001616:exon7:c.A945C:p.K315N | probably damaging | 10,2 \| 0,9 |
| | | | | | |
| | | | | | |
| SYN2 - synapsin-2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr3:12046206 | - > GGC | nonframeshift insertion; SYN2:NM_003178:exon1:c.181_182insGGC:p.T61delinsRP | | 0,0 \| 0,3 |
| | | | SYN2:NM_133625:exon1:c.181_182insGGC:p.T61delinsRP | | |
| VS9 | chr3:12046206 | - > GGC | nonframeshift insertion; SYN2:NM_003178:exon1:c.181_182insGGC:p.T61delinsRP | | 0,0 \| 0,3 |
| | | | SYN2:NM_133625:exon1:c.181_182insGGC:p.T61delinsRP | | |
| | | | | | |
| | | | | | |
| FLJ36031 - FLJ36031Y | | | | | |
| | | | | | |

| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
|---|---|---|---|---|---|
| VS8 | chr7:106300898 | G > A | nonsynonymous; FLJ36031:NM_175884:exon1:c.C445T:p.P149S | benign | 3,2 \| 2,5 |
| VS9 | chr7:106300898 | G > A | nonsynonymous; FLJ36031:NM_175884:exon1:c.C445T:p.P149S | benign | 1,0 \| 2,4 |
| | | | | | |
| | | | | | |
| ZNF404 - zinc finger protein 404 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr19:44376894 | C > A | nonsynonymous; ZNF404:NM_001033719:exon2:c.G1463T:p.G488V | probably damaging | 26,19 \| 17,11 |
| VS3 | chr19:44377016 | - > TTA | nonframeshift insertion; ZNF404:NM_001033719:exon2:c.1341_1342insTAA:p.L447delinsLN | | 2,3 \| 0,7 |
| | | | | | |
| | | | | | |
| AR - androgen receptor | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS11 | chrX:66765228 | AGAGACTAGCCCCAG > - | nonframeshift deletion; AR:NM_000044:exon1:c.240_254del:p.80_85del | | 13,3 \| 1,2 |

| | | | | | |
|---|---|---|---|---|---|
| VS13 | chrX:66766385 | GCGGCGGCGGCGGCGGCGGCGGCG > - | nonframeshift deletion; AR:NM_000044:exon1:c.1397_1420del:p.466_474del | | 1,1 \| 4,2 |
| VS13 | chrX:66766408 | - > GCG | nonframeshift insertion; AR:NM_000044:exon1:c.1420_1421insGCG:p.E474delinsGE | | 1,1 \| 4,2 |
| | | | | | |
| | | | | | |
| HJURP - Holliday junction recognition protein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chr2:234746224 | T > C | stoploss; HJURP:NM_018410:exon9:c.A2246G:p.X749W | | 9,9 \| 14,11 |
| VS6 | chr2:234749748 | T > C | nonsynonymous; HJURP:NM_018410:exon8:c.A1678G:p.K560E | benign | 14,16 \| 12,6 |
| | | | | | |
| | | | | | |
| OR1J2 - olfactory receptor 1J2 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS14 | chr9:125273392 | - > T | frameshift insertion; OR1J2:NM_054107:exon1:c.312_313insT:p.F104fs | | 41,48 \| 41,41 |
| VS8 | chr9:125273392 | - > T | frameshift insertion; OR1J2:NM_054107:exon1:c.312_313insT:p.F104fs | | 45,48 \| 47,33 |
| | | | | | |

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|---|---|---|---|---|---|
| SCNN1D - amiloride-sensitive sodium channel subunit delta | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS13 | chr1:1223322 | G > A | nonsynonymous; SCNN1D:NM_001130413:exon12:c.G1567A:p.V523M | benign | 1,13 \| 3,15 |
| VS14 | chr1:1221412 | A > G | nonsynonymous; SCNN1D:NM_001130413:exon7:c.T665G:p.F222C | probably damaging | 26,8 \| 34,19 |
| | | | | | |
| | | | | | |
| RBM23 - probable RNA-binding protein 23 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr14:23371261 | - > TGC | nonframeshift insertion; RBM23:NM_018107:exon11:c.1126_1127insGCA:p.A376delinsAA | | 16,18 \| 25,95 |
| | | | RBM23:NM_001077352:exon10:c.1072_1073insGCA:p.A358delinsAA | | |
| | | | RBM23:NM_001077351:exon12:c.1174_1175insGCA:p.A392delinsAA | | |
| VS13 | chr14:23371262 | - > ACG | nonframeshift insertion; RBM23:NM_001077352:exon10:c.1071_1072insCGT:p.A357delinsAV | | 10,26 \| 30,94 |
| | | | RBM23:NM_001077351:exon12:c.1173_1174insCGT:p.A391delinsAV | | |
| | | | RBM23:NM_018107:exon11:c.1125_1126insCGT:p.A375delinsAV | | |
| | | | | | |

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|---|---|---|---|---|---|
| EPX - eosinophil peroxidase preproprotein | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr17:56271442 | C > T | nonsynonymous; EPX:NM_000502:exon5:c.C583T:p.L195F | benign | 1,15 \| 1,16 |
| VS14 | chr17:56274358 | C > T | nonsynonymous; EPX:NM_000502:exon7:c.C860T:p.S287L | probably damaging | 57,54 \| 59,57 |
| | | | | | |
| | | | | | |
| LOC100287177 - hypothetical protein LOC100287177 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS12 | chr19:46145700 | G > C | nonsynonymous; LOC100287177:NM_001242348:exon3:c.G241C:p.A81P | | 4,6 \| 0,5 |
| VS14 | chr19:46145700 | G > C | nonsynonymous; LOC100287177:NM_001242348:exon3:c.G241C:p.A81P | | 4,5 \| 0,5 |
| | | | | | |
| | | | | | |
| STK33 - serine/threonine-protein kinase 33 | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high qualit |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | y) reads |
| VS2 | chr11:84140 92 | - > ACCAGA | nonframeshift insertion; STK33:NM_030906:exon14:c.1510_1511insTCTGGT:p.S504delinsSGS | | 17,15 \| 5,2 |
| VS5 | chr11:84140 92 | - > ACCAGA | nonframeshift insertion; STK33:NM_030906:exon14:c.1510_1511insTCTGGT:p.S504delinsSGS | | 8,12 \| 4,3 |
| | | | | | |
| | | | | | |
| PLEC - plectin | | | | | |
| | | | | | |
| indiv | posn | snp | snp effect | polyphen prediction | (high quality) reads |
| VS10 | chr8:144995 098 | - > GCTCCT | nonframeshift insertion; PLEC:NM_201383:exon32:c.8903_8904insAGGAGC:p.E2968delinsEEQ | | 65,44 \| 34,34 |
| | | | PLEC:NM_201382:exon32:c.8891_8892insAGGAGC:p.E2964delinsEEQ | | |
| | | | PLEC:NM_201384:exon32:c.8891_8892insAGGAGC:p.E2964delinsEEQ | | |
| | | | PLEC:NM_201379:exon32:c.8825_8826insAGGAGC:p.E2942delinsEEQ | | |
| | | | PLEC:NM_201381:exon32:c.8795_8796insAGGAGC:p.E2932delinsEEQ | | |
| | | | PLEC:NM_201380:exon32:c.9302_9303insAGGAGC:p.E3101delinsEEQ | | |
| | | | PLEC:NM_201378:exon32:c.8849_8850insAGGAGC:p.E2950delinsEEQ | | |
| | | | PLEC:NM_000445:exon33:c.8972_8973insAGGAGC:p.E2991delinsEEQ | | |
| VS12 | chr8:145018 058 | T > C | nonsynonymous; PLEC:NM_201382:exon1:c.A1G:p.M1V | benign | 29,24 \| 22,20 |

Table 31.1 Gene lists (complied by Dr S Twigg) of genes associated with craniosynostosis, have no loss-of-function alleles in Exome Variant Server or are a known transcription factor

| Craniosynostosis Tier 1 genes | | Craniosynostosis Tier 2 genes | TWIST1 target genes |
|---|---|---|---|
| *ALPL* | *RAB23* | *ADAMTSL4* | *BRAP* |
| *ASXL1* | *RECQL4* | *AXIN2* | *CHD3* |
| *COLEC11* | *RUNX2* | *BMPR1B* | *CHD4* |
| *CTSK* | *SH3PXD2B* | *CDC45* | *HDAC2* |
| *CYP26B1* | *SKI* | *DEPTOR* | *HES6* |
| *EFNB1* | *TCF12* | *DUSP6* | *MTA2* |
| *ERF* | *TGFBR1* | *FGF3* | *PPP2CA* |
| *ESCO2* | *TGFBR2* | *FGF4* | *RBBP7* |
| *FAM20C* | *TWIST1* | *FLNA* | *RELA* |
| *FBN1* | *WDR35* | *GDF6* | *SETD8* |
| *FGFR1* | *ZIC1* | *HUWE1* | |
| *FGFR2* | *MSX2* | *IFT122* | |
| *FGFR3* | | *IGF1R* | |
| *FREM1* | | *IL6ST* | |
| *GLI3* | | *LMX1B* | |
| *HDAC4* | | *LRRN3* | |
| *IHH* | | *MNT* | |
| *IL11RA* | | *NELL1* | |
| *JAG1* | | *TCF20* | |
| *KRAS* | | *TECPR1* | |
| *LRP5* | | *WDR19* | |
| *MEGF8* | | *POR* | |
| *MLL2* | | | |

Table 31.2

| DECIPHER craniosynostosis Deleterious | | | | | Mouse knockout genes with craniosynostosis phenotype | | | Copenhagen genes | Genes expressed in fronto-nasal primordia | Genes expressed in scalp fibroblasts (Bouchekova et al., 2010) | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *ABI2* | *DLGAP1* | *KANSL1L* | *PIKFYVE* | *TMEM194B* | *ACAN* | *GLI3* | *PSIP1* | *ADO* | *ALDH1A3* | *ABHD5* | *DLG1* | *JAM3* | *PRDM1* | *UBE2E3* |
| *ABRACL* | *DLGAP2* | *KBTBD11* | *PLAC9* | *TMEM237* | *ACVR1* | *GNAS* | *PTCH1* | *AKAP5* | *ALX1* | *ABI1* | *DOCK2* | *KBTBD8* | *PRPF38B* | *UBE3C* |
| *ACAA1* | *DNAH7* | *KCTD18* | *PLAGL1* | *TMEM248* | *ACVR2A* | *GSC* | *PTH1R* | *ALPP* | *ALX3* | *ABI3BP* | *DPP4* | *KCNK2* | *PRPF4B* | *UBE4B* |
| *ACADL* | *DNAJC6* | *KCTD7* | *PLCD1* | *TMEM254* | *AKT1* | *GSK3B* | *PTHLH* | *ALPPL2* | *ANO1* | *ACIN1* | *DSP* | *KCNMB1* | *PRRT3* | *USP31* |
| *ACCS* | *DOCK4* | *KIAA1244* | *PLCL1* | *TNFAIP3* | *ALX1* | *GUSB* | *PTPN11* | *ALX4* | *ASCL1* | *ADAM10* | *DST* | *KCTD12* | *PRRX1* | *USP34* |
| *ACCSL* | *DOCK7* | *KIAA1919* | *PLEKHM3* | *TP53I11* | *ALX3* | *HAND2* | *PTPN9* | *ARID1B* | *CLDN3* | *ADAM12* | *DUSP4* | *KIAA1109* | *PRRX2* | *VAMP5* |
| *ACVR2B* | *DSE* | *KLF7* | *PLK4* | *TPST1* | *ALX4* | *HAPLN1* | *PYGO2* | *BARX2* | *CLDN9* | *ADAM9* | *DUSP5* | *KIF11* | *PSPH* | *VCAM1* |
| *ADAM23* | *DYDC2* | *KLHL22* | *PMS1* | *TRAF3IP2* | *ANKH* | *HDAC1* | *RAI1* | *BBS9* | *CPED1* | *ADAMTS6* | *DUSP6* | *KNTC1* | *PTAR1* | *VDR* |
| *ADAT2* | *DYTN* | *KPNA5* | *PNPLA7* | *TRAK2* | *APAF1* | *HDAC4* | *RB1* | *BGLAP* | *CPM* | *AKAP12* | *DYNC2H1* | *LHFP* | *PTBP2* | *VIM* |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *ADCYAP1* | *ECT2L* | *KYNU* | *POLR3C* | *TRMT2A* | *ARID3B* | *HECTD1* | *RBL1* | *BGN* | *CRABP1* | *AKAP9* | *E2F7* | *LIFR* | *PTGFR* | *VPS13C* |
| *ADGB* | *EEF1B2* | *L1TD1* | *PPIL3* | *TSPAN14* | *ARID5B* | *HEXA* | *RGMA* | *BMP2* | *CRABP2* | *ALDH1A3* | *ECT2* | *LITAF* | *PTGS2* | *VPS35* |
| *AHI1* | *EFCAB7* | *LAMA4* | *PPP1R3A* | *TSPAN18* | *ARSB* | *HIRA* | *ROR2* | *BMP3* | *CRTAC1* | *ANK3* | *EFHC1* | *LMNB1* | *PTPRF* | *WDHD1* |
| *AIFM3* | *EHMT1* | *LANCL1* | *PRDM11* | *TSPYL1* | *ATF4* | *HOXA1* | *RPGRIP1L* | *BMP4* | *CRYAB* | *ANKRD10* | *EFHD1* | *LMOD1* | *PVRL3* | *WFDC1* |
| *AIG1* | *ELF2* | *LARP1B* | *PRODH* | *TSPYL4* | *AXIN1* | *HOXA2* | *RPL24* | *BMP7* | *CRYM* | *ANKRD12* | *EFS* | *LNPEP* | *QPRT* | *WSB1* |
| *AK4* | *ELMOD2* | *LEPR* | *PTH2R* | *TSSK2* | *AXIN2* | *HOXA3* | *RPL29* | *C1QTNF3* | *CXCL13* | *ANKRD17* | *EGFR* | *LOX* | *RAB33A* | *ZBTB20* |
| *ALDH8A1* | *EMILIN2* | *LEPROT* | *RAB11FIP4* | *TUBE1* | *BGN* | *HOXD13* | *RPS6KA3* | *CASS4* | *CXCR4* | *ANKRD28* | *EGR1* | *LRRC8C* | *RAB8B* | *ZBTB38* |
| *ALG6* | *ENOSF1* | *LPCAT4* | *RAB32* | *TXLNB* | *BMP2* | *HSPG2* | *RUNX2* | *CDON* | *DCC* | *ANKRD32* | *EIF4A2* | *LRRFIP1* | *RALGPS2* | *ZC3H12C* |
| *ALS2* | *EPM2A* | *LPIN2* | *RAB33B* | *TXNRD2* | *BMP5* | *HTT* | *RYK* | *CER1* | *DMRT3* | *ANLN* | *ELL2* | *LSP1* | *RANBP2* | *ZCCHC2* |
| *ALS2CR11* | *EPM2AIP1* | *LRP1B* | *RABGEF1* | *TYMS* | *BMP6* | *HYDIN* | *SALL4* | *CKAP5* | *DMRTA2* | *APC* | *EMILIN2* | *LSS* | *RAPGEF2* | *ZHX1* |
| *ALS2CR12* | *ERBB4* | *LRRC37B* | *RANBP1* | *TYW1* | *BMP7* | *IDS* | *SATB2* | *CNTN4* | *ELAVL4* | *APCDD1* | *EML4* | *MALAT1* | *RASA4* | *ZNF207* |
| *ALX4* | *ERICH1* | *LRRC7* | *RAPH1* | *TYW5* | *BMPER* | *IDUA* | *SGPL1* | *COL2A1* | *EPS8* | *APLP2* | *EMP1* | *MAML2* | *RASEF* | *ZNF292* |
| *AMD1* | *EVI2A* | *LRRFIP2* | *RAVER2* | *UBE2U* | *BMPR2* | *IFT88* | *SGSH* | *CPS1* | *EYA2* | *ARFGEF1* | *EMX2OS* | *MAP1LC3A* | *RASSF8* | *ZNF367* |
| *ANGPTL3* | *EVI2B* | *LRRN3* | *RBFOX1* | *UCP1* | *BRD4* | *IGF1* | *SH3BP2* | *CYFIP2* | *FAM107A* | *ARHGAP18* | *ENO2* | *MASTL* | *RBMS1* | *ZNF395* |
| *ANKRD35* | *EXOG* | *LTV1* | *RBM8A* | *UFD1L* | *CACNA1S* | *IGF1R* | *SHH* | *CYP17A1* | *FAP* | *ARHGAP22* | *ENPP2* | *MBNL1* | *RBPMS* | *ZNF585A* |
| *ANKRD44* | *EXT2* | *LZTR1* | *REPS1* | *UNC80* | *CALCR* | *IGF2* | *SIX1* | *CYP19A1* | *FEZF1* | *ARHGAP5* | *EPB41L3* | *MBNL2* | *RCN1* | *ZNF708* |
| *ANKRD50* | *EYA4* | *MAML3* | *REV3L* | *USP1* | *CASP3* | *IGF2R* | *SIX3* | *CYP21A2* | *FOXG1* | *ARRDC3* | *EPHA5* | *MCM10* | *RICTOR* | |
| *ANXA11* | *FAM117B* | *MAP2* | *RFPL4B* | *USP14* | *CASR* | *IHH* | *SKI* | *CYP51A1* | *HAS2* | *ASPH* | *EPHB1* | *MEG3* | *RPL23A* | |
| *AOX1* | *FAM126B* | *MAP3K5* | *RFTN2* | *UTP6* | *CBFB* | *IRF6* | *SLC39A4* | *DLX4* | *HEY1* | *ASPM* | *ERO1L* | *MEGF6* | *RPL37A* | |
| *ARHGAP15* | *FAM162B* | *MAP7* | *RFX6* | *UTRN* | *CBX2* | *ITGAV* | *SLC4A4* | *DLX5* | *HHIP* | *ASXL2* | *ETNK1* | *MFAP2* | *RPS24* | |
| *ARHGEF10* | *FAM213A* | *MAPT* | *RHOT1* | *VGLL2* | *CCND1* | *ITGB1* | *SMO* | *EFNA2* | *HPGD* | *ATAD2* | *ETS1* | *MGC16121* | *RRAGD* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *ARRDC1* | *FAM26D* | *MARCKS* | *RIMBP3* | *VILL* | *CDKN1C* | *ITGB1BP1* | *SMPD3* | *EP300* | *HTR3B* | *ATM* | *EXT1* | *MGEA5* | *RTN4* | |
| *ARVCF* | *FAM26E* | *MARS2* | *RNF115* | *VKORC1L1* | *CDON* | *JMJD6* | *SNAI1* | *FGF1* | *IGF1* | *ATP2B4* | *F3* | *MKI67* | *S100A4* | |
| *ASL* | *FASTKD2* | *MAT1A* | *RNF150* | *VTA1* | *CDX1* | *JUNB* | *SOCS7* | *FGF10* | *INA* | *ATP6V1C1* | *FAM102B* | *MKLN1* | *SCFD2* | |
| *ASZ1* | *FAT4* | *MDFIC* | *ROR1* | *WBSCR17* | *CER1* | *KAT6B* | *SOSTDC1* | *FGF18* | *IRS4* | *ATRX* | *FAM91A2* | *MLF1IP* | *SCML1* | |
| *ATG4C* | *FBXO25* | *MDH1B* | *ROS1* | *WDR12* | *CHRD* | *KL* | *SOX11* | *FGF2* | *ITGA8* | *B4GALT6* | *FBLIM1* | *MME* | *SCYL2* | |
| *ATP13A2* | *FBXO30* | *MED15* | *RPE* | *WDR78* | *CHST11* | *KLF10* | *SOX9* | *FGF6* | *LAMP5* | *BAIAP2L1* | *FBXL7* | *MMP3* | *SDAD1* | |
| *AUTS2* | *FGGY* | *MET* | *RPE65* | *WISP3* | *CHUK* | *L1CAM* | *SPRY4* | *FGF7* | *LHX2* | *BARD1* | *FBXO31* | *MORF4L2* | *SDC2* | |
| *BCLAF1* | *FOXC1* | *METTL21A* | *RPF2* | *WLS* | *CITED2* | *LAMA5* | *SRC* | *FGF8* | *MAOB* | *BCL11B* | *FGF5* | *MRPS15* | *SDCCAG8* | |
| *BMPR2* | *FOXD3* | *METTL24* | *RSPH4A* | *WNT2* | *CLCN7* | *LECT1* | *SSBP3* | *FGF9* | *NEFL* | *BEX1* | *FGL2* | *MRPS6* | *SEMA3C* | |
| *BOLL* | *FOXF2* | *METTL4* | *RTN4R* | *XYLB* | *COL11A2* | *LHX1* | *STK36* | *FGFR4* | *NEUROD1* | *BMP2* | *FIGNL1* | *MRVI1* | *SENP6* | |
| *BZW1* | *FOXP2* | *MFAP2* | *RWDD1* | *YES1* | *COL1A1* | *LHX2* | *SUMF1* | *FGFRL1* | *NEUROG1* | *BMPER* | *FKBP5* | *MSRB3* | *SEPW1* | |
| *CACHD1* | *FOXQ1* | *MFSD6* | *SAMD5* | *ZC2HC1B* | *COL2A1* | *LMNB1* | *SUMO1* | *FRS2* | *NHLH2* | *BRCA2* | *FLJ43663* | *MSX2* | *SERAC1* | |
| *CALN1* | *FRK* | *MFSD8* | *SATB2* | *ZDBF2* | *CREBBP* | *LMO4* | *TAB1* | *FZR1* | *NR2E1* | *BRIP1* | *FMNL2* | *MT1H* | *SERPINB2* | |
| *CAPZA2* | *FUCA2* | *MGARP* | *SBDS* | *ZDHHC8* | *CRKL* | *LMX1A* | *TACC3* | *GDF7* | *OLIG2* | *BUB1B* | *FOS* | *MT1X* | *SERPINE2* | |
| *CASP10* | *FYN* | *MGST2* | *SCARF2* | *ZEB2* | *CRTAP* | *LMX1B* | *TBX1* | *GPC3* | *OTX1* | *C1QTNF5* | *FUBP1* | *MYBL1* | *SFPQ* | |
| *CASP8* | *FZD5* | *MIER1* | *SCLT1* | *ZMYND19* | *CSF1* | *LRP2* | *TBX15* | *GRB2* | *PAPSS2* | *CA12* | *GAS6* | *MYO10* | *SGCG* | |
| *CAV1* | *FZD7* | *MLH1* | *SCN5A* | *ZNF107* | *CSF1R* | *LRP5* | *TBX18* | *HMX2* | *PAX3* | *CALB2* | *GDAP2* | *MYOCD* | *SGOL2* | |
| *CAV2* | *GADD45A* | *MPP4* | *SCOC* | *ZNF117* | *CSNK2A1* | *LRP6* | *TCF15* | *HOPX* | *PAX6* | *CALD1* | *GK* | *NCKAP1* | *SH2D5* | |
| *CCDC150* | *GLS* | *MRPL41* | *SDHB* | *ZNF138* | *CTGF* | *LTBP3* | *TCOF1* | *IBSP* | *PAX7* | *CAMK2D* | *GM2A* | *NEDD9* | *SHCBP1* | |
| *CCDC28A* | *GMDS* | *MSTN* | *SDPR* | *ZNF273* | *CTNNB1* | *MAP3K3* | *TFAP2A* | *IGF2BP3* | *PCDH20* | *CASC5* | *GNA13* | *NEK1* | *SIPA1L2* | |
| *CCNYL1* | *GNB1L* | *MYB* | *SERBP1* | *ZNF277* | *CTNNBIP1* | *MAPK7* | *TGFB2* | *LAX1* | *PMAIP1* | *CBLB* | *GOLGA4* | *NETO2* | *SLC16A7* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *CCRN4L* | *GNG12* | *MYD88* | *SERPIND 1* | *ZNF596* | *CTSK* | *MGAT2* | *TGFBR2* | *LHX3* | *PRKCQ* | *CBRG support* | *GOLGA 8A* | *NFAT5* | *SLC19A 2* | |
| *CD28* | *GOLGA4* | *MYL1* | *SETD7* | *ZNF680* | *CYP24A1* | *MMP13* | *THRA* | *MMP23 B* | *PRR15* | *CCDC50* | *GPC4* | *NIPA1* | *SLC20A 1* | |
| *CD82* | *GOLGA8 A* | *MYL12A* | *SF3B1* | *ZNF727* | *CYP26A1* | *MMP14* | *THRB* | *MNX1* | *PTCHD1* | *CCDC58* | *GPC6* | *NIPBL* | *SLC24A 3* | |
| *CDC40* | *GOPC* | *MYL12B* | *SF3B5* | *ZNF735* | *CYP27B1* | *MMP16* | *TIPARP* | *MSX1* | *RGS4* | *CCND1* | *GPM6B* | *NKTR* | *SLC2A1 3* | |
| *CDC45* | *GP1BB* | *MYO1B* | *SGIP1* | *ZNF736* | *DDR1* | *MMP9* | *TP53* | *NCOA1* | *RPRM* | *CCNE2* | *GPNMB* | *NOTCH2* | *SLC2A3* | |
| *CDK15* | *GPR1* | *MYOM1* | *SGK1* | *ZNF74* | *DDR2* | *MN1* | *TP53BP2* | *NOG* | *SGPP2* | *CD109* | *GPX7* | *NPR3* | *SLC39A 4* | |
| *CDK19* | *GPR126* | *MYOM2* | *SGOL2* | *ZNF92* | *DIAPH1* | *MNT* | *TP73* | *PART1* | *SIX2* | *CD36* | *GRB14* | *NPTN* | *SLC4A4* | |
| *CETN1* | *GPR85* | *MYSM1* | *SH2D4B* | *ZUFSP* | *DISP1* | *MSX1* | *TSHZ1* | *PCYT1B* | *SIX6* | *CD44* | *GREM2* | *NPTX1* | *SLC6A1 5* | |
| *CFLAR* | *GPRC6A* | *NAA15* | *SHPRH* | | *DKK1* | *MSX2* | *TWIST1* | *PDE1C* | *SLC27A2* | *CD47* | *GSTA1* | *NR1D2* | *SLIT2* | |
| *CFTR* | *GRM1* | *NAB1* | *SLC16A1 0* | | *DLG1* | *MYC* | *TWSG1* | *PIK3R2* | *SNCA* | *CD59* | *GSTT2* | *NR3C1* | *SMCHD 1* | |
| *CHST1* | *GSC2* | *NABP1* | *SLC22A1 3* | | *DLG5* | *MYH10* | *VCL* | *PJA1* | *SP8* | *CDC42E P5* | *H19* | *NRIP1* | *SNRPN* | |
| *CITED2* | *GTDC1* | *NBEAL1* | *SLC22A1 4* | | *DLL3* | *NAPA* | *VDR* | *PLS3* | *ST18* | *CDC7* | *HAPLN3* | *NRP1* | *SORT1* | |
| *CLDN5* | *GTF3C3* | *NDC80* | *SLC22A1 6* | | *DLX1* | *NDST1* | *VEGFA* | *PRRX1* | *STMN3* | *CDH18* | *HELLS* | *NUPL1* | *SPRY2* | |
| *CLGN* | *GTF3C6* | *NDUFB3* | *SLC25A1* | | *DLX2* | *NELL1* | *WNT1* | *PTCH1* | *STRA6* | *CENPE* | *HES1* | *OCIAD2* | *STON2* | |
| *CLK1* | *GUSB* | *NDUFC1* | *SLC2A12* | | *DLX5* | *NFATC2* | *WNT3A* | *PTER* | *TFAP2B* | *CENPF* | *HES4* | *OPA1* | *STXBP6* | |
| *CLN8* | *HBS1L* | *NDUFS1* | *SLC35D1* | | *DLX6* | *NFIA* | *WNT5A* | *PTPRO* | *TUBB3* | *CEP152* | *HHIP* | *OSBPL8* | *SUPT16 H* | |
| *CLTCL1* | *HDAC2* | *NF1* | *SLC35D3* | | *DNAH5* | *NFIX* | *WNT9A* | *PYGO2* | *UNCX* | *CEP170* | *HINT3* | *OSBPL9* | *SVIL* | |
| *CLUL1* | *HEBP2* | *NFIA* | *SLC39A1 0* | | *DPH1* | *NKD1* | *WWTR1* | *RARRES 1* | *WSCD1* | *CFLAR* | *HIPK2* | *OSMR* | *SYNE2* | |
| *COL10A1* | *HECA* | *NHSL1* | *SLC7A11* | | *DUSP6* | *NKX3-2* | *XRCC2* | *RLIM* | | *CHAC1* | *HLA-DPA1* | *OSR1* | *SYNPO2* | |
| *COLEC1 2* | *HECW2* | *NIF3L1* | *SLC7A4* | | *E2F5* | *NOG* | *ZEB1* | *RPS6KB 2* | | *CHD1* | *HLA-DPB1* | *PABPC1* | *TAF15* | |
| *COMT* | *HIBCH* | *NMBR* | *SMCHD1* | | *EDNRB* | *NPHP3* | *ZFAND5* | *SATB2* | | *CHM* | *HMGA2* | *PAN3* | *TAF9B* | |
| *COPRS* | *HIRA* | *NOP58* | *SNAP29* | | *EFNB1* | *NPPC* | *ZIC3* | *SHH* | | *CLCN3* | *HMMR* | *PAPD4* | *TAOK1* | |
| *COQ10B* | *HIVEP2* | *NRG3* | *SPATA5* | | *EN1* | *NPR2* | *ZMPSTE 24* | *SOX6* | | *CLGN* | *HMOX1* | *PAPPA* | *TBX3* | |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *CPO* | *HOOK1* | *NRP2* | *SPATS2L* | | *EP300* | *NPR3* | | *SOX9* | | *CNN1* | *HNMT* | *PCBP2* | *TEAD1* | |
| *CPS1* | *HS3ST5* | *NT5DC1* | *SPPL2C* | | *ERBB2* | *OFD1* | | *SP7* | | *CNTN3* | *HPCAL1* | *PDE4DIP* | *TES* | |
| *CRCP* | *HSPA4L* | *NUDT17* | *SPRY1* | | *EVC* | *OTX1* | | *SPTA1* | | *COL13A1* | *HRK* | *PDGFC* | *TFPI2* | |
| *CREB1* | *HSPD1* | *NUS1* | *STAT1* | | *EXT2* | *PAPPA* | | *STRA6* | | *COL6A1* | *HS2ST1* | *PDLIM1* | *TFRC* | |
| *CRHR1* | *HSPE1* | *OLIG3* | *STAT4* | | *FANCA* | *PAPSS2* | | *TGFB1* | | *COL8A2* | *HSPA2* | *PDZRN3* | *TGFB1I1* | |
| *CRKL* | *ICA1L* | *OMA1* | *STK17B* | | *FBN1* | *PAX6* | | *TGFB2* | | *COPA* | *HSPA4* | *PGK1* | *TGFBR1* | |
| *CROCC* | *ICOS* | *ORC2* | *STRADB* | | *FGF18* | *PAX9* | | *TGFB3* | | *CPEB4* | *HSPB2* | *PHGDH* | *TGFBR2* | |
| *CRYGA* | *IDH1* | *OXSR1* | *STX11* | | *FGFR1* | *PBX1* | | *TINAGL1* | | *CSNK1A1* | *ID4* | *PHIP* | *TGFBR3* | |
| *CRYGB* | *IFNGR1* | *P2RX6* | *STXBP5* | | *FGFR2* | *PDGFA* | | *TSHR* | | *CTSC* | *IFI44* | *PHLDA1* | *THBD* | |
| *CRYGD* | *IFRD1* | *PABPC4L* | *SUMO1* | | *FGFR3* | *PDGFRA* | | *TULP4* | | *CTSK* | *IFI44L* | *PIK3C2A* | *THOC2* | |
| *CSMD1* | *IL12RB2* | *PARD3B* | *SUZ12* | | *FIGN* | *PDS5B* | | *VPREB1* | | *CXCL2* | *IGFBP2* | *PIK3R1* | *TM4SF1* | |
| *CTDSPL* | *IL20RA* | *PBOV1* | *SYT13* | | *FOS* | *PEX11B* | | *WIF1* | | *CXCL3* | *IGFBP5* | *PKIA* | *TMEM38B* | |
| *CTTNBP2* | *IL22RA2* | *PCDH10* | *TACSTD2* | | *FOSL2* | *PEX2* | | *WIPF1* | | *CXCL5* | *IL13RA2* | *PKN2* | *TNC* | |
| *CYP20A1* | *IL23R* | *PCDH18* | *TBC1D9* | | *FOXA2* | *PHEX* | | *WWTR1* | | *CYB5R2* | *IL15* | *PKP4* | *TNIK* | |
| *CYP2J2* | *IMMP2L* | *PDE4B* | *TBPL1* | | *FOXC1* | *PIGA* | | *ZFX* | | *CYLD* | *IL6ST* | *PLA2G4A* | *TOP2A* | |
| *DAB1* | *INADL* | *PDE7B* | *TBX1* | | *FOXC2* | *PITX2* | | | | *CYP1B1* | *IL7R* | *PLK4* | *TPD52L1* | |
| *DCBLD1* | *INO80D* | *PERP* | *TCF21* | | *FOXG1* | *PKD1* | | | | *DAB2* | *IMPA2* | *PLXNC1* | *TPM1* | |
| *DDO* | *INPP1* | *PEX11B* | *TCTEX1D1* | | *FOXH1* | *PLCB1* | | | | *DCBLD2* | *INSIG1* | *PNN* | *TPP1* | |
| *DEPDC1* | *INSL5* | *PEX3* | *TES* | | *FREM2* | *PLK2* | | | | *DCLRE1B* | *IQGAP1* | *PODXL* | *TPR* | |
| *DGCR14* | *ITGA10* | *PEX7* | *TFEC* | | *G6PD* | *POLL* | | | | *DDAH1* | *ITGA11* | *PPFIBP1* | *TPST2* | |
| *DGCR2* | *ITGA9* | *PGAP1* | *TGIF1* | | *GATA3* | *POSTN* | | | | *DDX17* | *ITGA2* | *PPIG* | *TRIM2* | |
| *DGCR6* | *ITGB3BP* | *PGM1* | *THAP7* | | *GCM2* | *PPT1* | | | | *DEPDC1* | *ITGA6* | *PPM1M* | *TRPC4* | |
| *DGCR6L* | *JAK1* | *PGRMC2* | *THOC1* | | *GDF6* | *PRKG2* | | | | *DES* | *ITGA8* | *PPP1R14A* | *TRPM7* | |
| *DGCR8* | *JUN* | *PHACTR2* | *TM2D1* | | *GFRA4* | *PRKRA* | | | | *DGKI* | *ITGAV* | *PPP3CA* | *TTK* | |
| *DIRAS3* | *KANK4* | *PI4KA* | *TMEM168* | | *GJA1* | *PRRX1* | | | | *DHX9* | *JAG1* | *PPP3R1* | *TUBB6* | |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *DLEC1* | *KANSL1* | *PIAS3* | *TMEM19 1B* | | *GLI2* | *PSEN1* | | | | *DKK3* | *JAK1* | *PRAC* | *UBE2D3* | |

Table 31.3

| *Coussen's suture list (Coussens et al., 2007)* | | | | | |
|---|---|---|---|---|---|
| *ABCG1* | *COL2A1* | *HCK* | *MMD* | *RAG1* | *TRIM58* |
| *ACSL1* | *COL3A1* | *HCLS1* | *MME* | *RAG2* | *TSPAN5* |
| *ADAM23* | *COL6A1* | *HERC5* | *MMP14* | *RARRES1* | *TTF2* |
| *ADCY2* | *COL6A2* | *HHEX* | *MMP2* | *RASGRP1* | *TTK* |
| *ADD2* | *COL6A3* | *HIST1H1E* | *MMP23B* | *RASGRP2* | *TXNIP* |
| *AEBP1* | *COL8A2* | *HIST1H2BD* | *MMP8* | *RASSF2* | *TYRO3* |
| *AIF1* | *CORO1A* | *HK3* | *MN1* | *RBP4* | *UCP2* |
| *AIM1* | *CREB5* | *HLA-DMA* | *MNDA* | *RETN* | *VNN1* |
| *ALAS2* | *CRHBP* | *HLA-DMB* | *MPO* | *RFC5* | *VPREB1* |
| *ALOX5* | *CRISP2* | *HLA-DPA1* | *MPP1* | *RGS2* | *VPREB3* |
| *ALOX5AP* | *CRISP3* | *HLA-DQA1* | *MS4A1* | *RHAG* | *VRK1* |
| *AMPD3* | *CRISPLD2* | *HLA-DQB1* | *MS4A3* | *RHCE* | *VWF* |
| *AMPH* | *CSF2RB* | *HLA-DRA* | *MS4A4A* | *RHD* | *WASF1* |
| *ANGPTL2* | *CSF3R* | *HLA-DRB1* | *MSH2* | *RHOH* | *WASL* |
| *ANK1* | *CST7* | *HLF* | *MT1X* | *RIF1* | *WHSC1* |
| *ANTXR1* | *CSTA* | *HMBS* | *MYB* | *RIMS3* | *WIF1* |
| *ANXA3* | *CTSG* | *HMGA1* | *MYL4* | *RNASE2* | *XK* |
| *APOC1* | *CTSS* | *HMGB2* | *MYO1F* | *RNASE3* | *XPNPEP2* |
| *ARG1* | *CXCR4* | *HMMR* | *NAP1L3* | *RNASE6* | *ZFHX4* |
| *ARG2* | *CYBB* | *HMOX1* | *NAV3* | *RNGTT* | *ZNF238* |
| *ARHGAP15* | *CYFIP2* | *HP* | *NCF1* | *ROR1* | |
| *ARHGAP19* | *CYP4F3* | *HS3ST3A1* | *NCF2* | *RPS6KA1* | |
| *ARHGAP25* | *DACT1* | *HSPA1B* | *NCF4* | *RRAGD* | |
| *ARHGDIB* | *DCHS1* | *ICAM3* | *NEDD4L* | *RRM2* | |

| | | | | | |
|---|---|---|---|---|---|
| *ARRB2* | *DCK* | *IGF1* | *NEIL1* | *RUNX1T1* | |
| *ASPM* | *DIRAS3* | *IGJ* | *NEK2* | *RYR3* | |
| *ASPN* | *DNM1* | *IGLL1* | *NELL2* | *S100A10* | |
| *ATAD2* | *DNMT3B* | *IL11RA* | *NFE2* | *S100A9* | |
| *ATF3* | *DNTT* | *IL18RAP* | *NPL* | *S100P* | |
| *ATF7IP2* | *DOCK2* | *IL1R2* | *NUP210* | *SAMSN1* | |
| *ATP2A3* | *DPYD* | *IL6ST* | *OAS2* | *SCRG1* | |
| *ATP8B4* | *DPYSL3* | *IL7R* | *OIP5* | *SELENBP1* | |
| *AURKB* | *DUSP10* | *IMPA2* | *OLFM1* | *SELL* | |
| *AZU1* | *E2F5* | *INHBA* | *OLFM4* | *SELPLG* | |
| *BANK1* | *EAF2* | *INPP5D* | *OLFML1* | *SEMA3C* | |
| *BCL11A* | *EDIL3* | *IQGAP2* | *OLR1* | *SERPINA1* | |
| *BCL11B* | *EFEMP1* | *IRF4* | *ORM1* | *SERPINB1* | |
| *BCL7A* | *EGFL6* | *IRF8* | *OSBPL3* | *SERPINB10* | |
| *BIN2* | *EGFR* | *ISG20* | *P2RX1* | *SETBP1* | |
| *BLM* | *EHD2* | *ISLR* | *P2RX5* | *SHOX2* | |
| *BLNK* | *ELOVL4* | *ITGA4* | *P2RY13* | *SIX2* | |
| *BPI* | *EMX2* | *ITGAM* | *P2RY14* | *SLC14A1* | |
| *BST1* | *ENPP4* | *ITGB2* | *PADI4* | *SLC15A2* | |
| *BTG2* | *EPB42* | *ITGB5* | *PAM* | *SLC22A4* | |
| *BTK* | *EPHA3* | *ITGBL1* | *PARP8* | *SLC2A5* | |
| *BUB1* | *EPHA4* | *ITK* | *PAX5* | *SLC38A1* | |
| *BUB1B* | *EPHB2* | *ITM2A* | *PCLO* | *SLC4A1* | |
| *C1QTNF3* | *EPX* | *JUN* | *PCSK5* | *SLC4A4* | |
| *CA1* | *EVI2B* | *KAL1* | *PDGFRL* | *SLCO4C1* | |
| *CALD1* | *EWSR1* | *KCNK1* | *PDK4* | *SLPI* | |
| *CAMP* | *EYA2* | *KIAA0125* | *PDZRN3* | *SNCA* | |
| *CAP2* | *EZH2* | *KIAA0922* | *PDZRN4* | *SNCAIP* | |
| *CAPN3* | *FABP4* | *KIF11* | *PELI2* | *SOCS2* | |

| | | | | | |
|---|---|---|---|---|---|
| *CAPN6* | *FAIM3* | *KIF14* | *PF4* | *SORL1* | |
| *CASP1* | *FAM60A* | *KIF15* | *PIK3CD* | *SPIB* | |
| *CAT* | *FAP* | *KIF23* | *PIP5K1B* | *SPON1* | |
| *CCNA2* | *FAT4* | *KIF2C* | *PITX2* | *SPON2* | |
| *CCNB2* | *FBLN1* | *KIF4A* | *PLAC8* | *SPTA1* | |
| *CCND1* | *FCAR* | *KLF1* | *PLAGL1* | *SSBP2* | |
| *CCND2* | *FCN1* | *LAIR1* | *PLCB1* | *SSPN* | |
| *CCND3* | *FECH* | *LAMA2* | *PLCG2* | *SSX2IP* | |
| *CCNE2* | *FEN1* | *LAMC1* | *PLEKHA1* | *ST6GAL1* | |
| *CCR2* | *FGFR2* | *LAPTM5* | *PLEKHF2* | *STXBP6* | |
| *CD163* | *FGR* | *LBR* | *PLK4* | *SYK* | |
| *CD19* | *FKBP14* | *LCN2* | *PLOD3* | *TACC2* | |
| *CD1D* | *FLI1* | *LCP1* | *PLSCR1* | *TACC3* | |
| *CD300A* | *FLRT3* | *LCP2* | *PNMA2* | *TACSTD2* | |
| *CD37* | *FMOD* | *LIG4* | *POLQ* | *TAGLN* | |
| *CD38* | *FOLR3* | *LILRA2* | *POSTN* | *TAL1* | |
| *CD48* | *FPR1* | *LILRB1* | *POU2AF1* | *TARP* | |
| *CD52* | *FUT4* | *LILRB2* | *PPP2R3A* | *TBC1D19* | |
| *CD53* | *FYB* | *LMNA* | *PRELP* | *TCEAL2* | |
| *CD5L* | *FZD1* | *LOXL1* | *PRG1* | *TCL1A* | |
| *CD69* | *GAP43* | *LOXL2* | *PRG2* | *TCN1* | |
| *CD72* | *GAS7* | *LRIG1* | *PRG3* | *TF* | |
| *CD74* | *GCA* | *LRMP* | *PRIM1* | *TGFB2* | |
| *CD79B* | *GCH1* | *LRRN3* | *PRKAR2B* | *TGFB3* | |
| *CDA* | *GDF10* | *LSS* | *PRKCD* | *THBS2* | |
| *CDC20* | *GEM* | *LTF* | *PRRX2* | *THBS3* | |
| *CDC42BPA* | *GIT2* | *LY75* | *PRSS23* | *THY1* | |
| *CDC7* | *GLUL* | *LY86* | *PRTN3* | *TIMP3* | |
| *CDKN2D* | *GMFG* | *LYN* | *PSMB9* | *TKT* | |

| | | | | | |
|---|---|---|---|---|---|
| *CDKN3* | *GNG4* | *MAB21L2* | *PSPH* | *TLE2* | |
| *CEACAM1* | *GNG7* | *MAPK14* | *PSTPIP2* | *TLE4* | |
| *CEACAM6* | *GPC1* | *MATN2* | *PTGER4* | *TLR2* | |
| *CECR1* | *GPC3* | *MBP* | *PTK2B* | *TMEFF1* | |
| *CENPA* | *GPLD1* | *MCC* | *PTN* | *TMEM30B* | |
| *CENPE* | *GPNMB* | *MCM10* | *PTPN22* | *TMPO* | |
| *CENPF* | *GPR126* | *MCTP2* | *PTPRC* | *TNFAIP8* | |
| *CHD7* | *GPR18* | *ME2* | *PTPRE* | *TNFRSF17* | |
| *CHI3L1* | *GRP* | *MEG3* | *PTPRF* | *TNFSF10* | |
| *CHL1* | *GULP1* | *MEIS2* | *PTTG1* | *TNN* | |
| *CLC* | *GYPA* | *MFAP2* | *PYGL* | *TOX* | |
| *CLEC1B* | *GYPB* | *MFAP3L* | *QRSL1* | *TPD52* | |
| *CLEC2D* | *GZMB* | *MFAP4* | *RAB11FIP1* | *TPSB2* | |
| *CLEC5A* | *HAPLN1* | *MFNG* | *RAB20* | *TPX2* | |
| *COL10A1* | *HBB* | *MGAM* | *RAB27A* | *TRIM10* | |
| *COL11A1* | *HBD* | *MICB* | *RAC2* | *TRIM14* | |
| *COL14A1* | *HBG1* | *MID1* | *RAD51* | *TRIM2* | |
| *COL16A1* | *HBG2* | *MKI67* | *RAD51AP1* | *TRIM22* | |

N.B. Full list of Non loss-of-function genes in Exome Variant Server totals > 4200 genes and is not given here, but is available online at https://gbrowse2.molbiol.ox.ac.uk/cgi-bin/cranio_snp_compare.cgi

Table 31.4

| *Wingender transcription factor database* | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *AEBP2* | *DLX2* | *FOXD4L3* | *HMGB3P1* | *KLF2* | *NFIC* | *PKNOX1* | *SIX1* | *TCF7L1* | *ZBTB39* | *ZNF182* | *ZNF358* | *ZNF551* | *ZNF700* | ZNF99 |
| *AFP* | *DLX3* | *FOXD4L5* | *HMGXB3* | *KLF3* | *NFIX* | *PKNOX2* | *SIX2* | *TCF7L2* | *ZBTB4* | *ZNF184* | *ZNF362* | *ZNF552* | *ZNF701* | ZNFX1 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *AHR* | *DLX4* | *FOXD4L6* | *HMGXB4* | *KLF4* | *NFKB1* | *PLAG1* | *SIX3* | *TCFL5* | *ZBTB40* | *ZNF189* | *ZNF366* | *ZNF554* | *ZNF702P* | ZSCAN1 |
| *AHRR* | *DLX5* | *FOXE1* | *HMX1* | *KLF5* | *NFKB2* | *PLAGL1* | *SIX4* | *TEAD1* | *ZBTB41* | *ZNF19* | *ZNF37A* | *ZNF555* | *ZNF705A* | ZSCAN10 |
| *AIRE* | *DLX6* | *FOXE3* | *HMX2* | *KLF6* | *NFKBIA* | *PLAGL2* | *SIX5* | *TEAD2* | *ZBTB42* | *ZNF195* | *ZNF382* | *ZNF556* | *ZNF705D* | ZSCAN12 |
| *ALX1* | *DMAP1* | *FOXF1* | *HMX3* | *KLF7* | *NFKBIB* | *POGZ* | *SIX6* | *TEAD3* | *ZBTB43* | *ZNF197* | *ZNF383* | *ZNF557* | *ZNF705G* | ZSCAN16 |
| *ALX3* | *DMRT1* | *FOXF2* | *HN1L* | *KLF8* | *NFKBID* | *POU2F1* | *SLC2A4RG* | *TEAD4* | *ZBTB44* | *ZNF2* | *ZNF384* | *ZNF558* | *ZNF707* | ZSCAN2 |
| *ALX4* | *DMRT2* | *FOXG1* | *HNF1A* | *KLF9* | *NFKBIE* | *POU2F2* | *SMAD1* | *TEF* | *ZBTB45* | *ZNF200* | *ZNF391* | *ZNF559* | *ZNF708* | ZSCAN20 |
| *APC* | *DMRT3* | *FOXH1* | *HNF1B* | *L3MBTL1* | *NFKBIL1* | *POU2F3* | *SMAD2* | *TET1* | *ZBTB46* | *ZNF202* | *ZNF394* | *ZNF560* | *ZNF709* | ZSCAN21 |
| *AR* | *DMRTA1* | *FOXI1* | *HNF4A* | *LBX2* | *NFKBIZ* | *POU3F1* | *SMAD3* | *TFAM* | *ZBTB47* | *ZNF205* | *ZNF396* | *ZNF561* | *ZNF71* | ZSCAN22 |
| *ARGFX* | *DMRTA2* | *FOXI2* | *HNF4G* | *LEF1* | *NFX1* | *POU3F2* | *SMAD4* | *TFAP2A* | *ZBTB48* | *ZNF211* | *ZNF397* | *ZNF562* | *ZNF710* | ZSCAN23 |
| *ARID1A* | *DMRTB1* | *FOXI3* | *HOMEZ* | *LEUTX* | *NFXL1* | *POU3F3* | *SMAD5* | *TFAP2B* | *ZBTB49* | *ZNF212* | *ZNF398* | *ZNF563* | *ZNF711* | ZSCAN29 |
| *ARID2* | *DMRTC2* | *FOXJ1* | *HOPX* | *LHX1* | *NFYA* | *POU3F4* | *SMAD6* | *TFAP2C* | *ZBTB5* | *ZNF213* | *ZNF404* | *ZNF565* | *ZNF713* | ZSCAN30 |
| *ARID3A* | *DMTF1* | *FOXJ2* | *HOXA1* | *LHX2* | *NFYB* | *POU4F1* | *SMAD7* | *TFAP2D* | *ZBTB6* | *ZNF214* | *ZNF407* | *ZNF566* | *ZNF714* | ZSCAN4 |
| *ARID3B* | *DNAJC1* | *FOXJ3* | *HOXA10* | *LHX3* | *NFYC* | *POU4F2* | *SMAD9* | *TFAP2E* | *ZBTB7A* | *ZNF215* | *ZNF408* | *ZNF567* | *ZNF716* | ZSCAN5A |
| *ARID3C* | *DNAJC2* | *FOXK1* | *HOXA11* | *LHX4* | *NHLH1* | *POU4F3* | *SMARCA1* | *TFAP4* | *ZBTB7B* | *ZNF217* | *ZNF41* | *ZNF568* | *ZNF717* | ZSCAN5B |
| *ARID4B* | *DNMT1* | *FOXK2* | *HOXA13* | *LHX5* | *NHLH2* | *POU5F1* | *SMARCA5* | *TFCP2* | *ZBTB7C* | *ZNF219* | *ZNF410* | *ZNF569* | *ZNF718* | ZXDA |
| *ARID5A* | *DNMT3A* | *FOXL1* | *HOXA2* | *LHX6* | *NKX1-2* | *POU5F1B* | *SMARCC1* | *TFCP2L1* | *ZBTB8B* | *ZNF22* | *ZNF414* | *ZNF57* | *ZNF721* | ZXDB |
| *ARID5B* | *DNMT3B* | *FOXL2* | *HOXA3* | *LHX8* | *NKX2-1* | *POU5F2* | *SMARCC2* | *TFDP1* | *ZC3H4* | *ZNF221* | *ZNF415* | *ZNF571* | *ZNF724P* | ZXDC |
| *ARNT* | *DNMT3L* | *FOXM1* | *HOXA5* | *LHX9* | *NKX2-2* | *POU6F1* | *SMARCE1* | *TFE3* | *ZC3H6* | *ZNF222* | *ZNF416* | *ZNF572* | *ZNF726* | |
| *ARNT2* | *DPF2* | *FOXN1* | *HOXA6* | *LMX1A* | *NKX2-3* | *POU6F2* | *SNAI1* | *TFEB* | *ZC3H8* | *ZNF223* | *ZNF417* | *ZNF573* | *ZNF727* | |
| *ARNTL* | *DPRX* | *FOXN3* | *HOXA7* | *LMX1B* | *NKX2-4* | *PPARA* | *SNAI2* | *TFEC* | *ZEB1* | *ZNF224* | *ZNF418* | *ZNF574* | *ZNF729* | |
| *ARNTL2* | *DRGX* | *FOXN4* | *HOXA9* | *LRRFIP1* | *NKX2-5* | *PPARD* | *SNAI3* | *TGIF1* | *ZEB2* | *ZNF225* | *ZNF419* | *ZNF575* | *ZNF732* | |
| *ARX* | *DUX2* | *FOXO1* | *HOXB1* | *LRRFIP2* | *NKX2-8* | *PPARG* | *SNAPC4* | *TGIF2* | *ZFAT* | *ZNF226* | *ZNF420* | *ZNF576* | *ZNF733P* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *ASCL1* | *DUX4L2* | *FOXO3* | *HOXB13* | *LYAR* | *NKX3-1* | *PPP1R13L* | *SOHLH1* | *THAP1* | *ZFHX2* | *ZNF227* | *ZNF425* | *ZNF578* | *ZNF735* | |
| *ASCL2* | *DUXA* | *FOXO4* | *HOXB2* | *LYL1* | *NKX3-2* | *PRDM1* | *SOHLH2* | *THAP10* | *ZFHX3* | *ZNF229* | *ZNF426* | *ZNF579* | *ZNF736* | |
| *ASCL3* | *E2F1* | *FOXP1* | *HOXB3* | *MAD1L1* | *NKX6-1* | *PRDM10* | *SOX1* | *THAP11* | *ZFHX4* | *ZNF23* | *ZNF429* | *ZNF580* | *ZNF737* | |
| *ASCL4* | *E2F2* | *FOXP2* | *HOXB4* | *MAF* | *NKX6-3* | *PRDM13* | *SOX10* | *THAP2* | *ZFP1* | *ZNF230* | *ZNF43* | *ZNF581* | *ZNF74* | |
| *ATF1* | *E2F3* | *FOXP3* | *HOXB5* | *MAFA* | *NOBOX* | *PRDM14* | *SOX11* | *THAP3* | *ZFP14* | *ZNF233* | *ZNF430* | *ZNF582* | *ZNF740* | |
| *ATF2* | *E2F4* | *FOXP4* | *HOXB6* | *MAFB* | *NONO* | *PRDM15* | *SOX12* | *THAP4* | *ZFP2* | *ZNF234* | *ZNF431* | *ZNF583* | *ZNF746* | |
| *ATF3* | *E2F5* | *FOXQ1* | *HOXB8* | *MAFF* | *NOTO* | *PRDM16* | *SOX13* | *THAP5* | *ZFP28* | *ZNF235* | *ZNF432* | *ZNF585A* | *ZNF749* | |
| *ATF4* | *E2F6* | *FOXR1* | *HOXB9* | *MAFG* | *NPAS1* | *PRDM2* | *SOX17* | *THAP6* | *ZFP3* | *ZNF236* | *ZNF433* | *ZNF585B* | *ZNF75A* | |
| *ATF5* | *E2F7* | *FOXR2* | *HOXC10* | *MAFK* | *NPAS2* | *PRDM8* | *SOX18* | *THAP7* | *ZFP30* | *ZNF239* | *ZNF436* | *ZNF586* | *ZNF75D* | |
| *ATF6* | *E2F8* | *GABPA* | *HOXC11* | *MAX* | *NPAS3* | *PRDM9* | *SOX21* | *THAP8* | *ZFP36* | *ZNF24* | *ZNF438* | *ZNF587* | *ZNF76* | |
| *ATF6B* | *E4F1* | *GATA1* | *HOXC13* | *MAZ* | *NPAS4* | *PRKRIR* | *SOX3* | *THAP9* | *ZFP37* | *ZNF25* | *ZNF439* | *ZNF589* | *ZNF761* | |
| *ATF7* | *EBF1* | *GATA2* | *HOXC4* | *MECOM* | *NR0B1* | *PROP1* | *SOX30* | *THRA* | *ZFP41* | *ZNF250* | *ZNF44* | *ZNF592* | *ZNF763* | |
| *ATMIN* | *EBF2* | *GATA3* | *HOXC5* | *MEF2A* | *NR0B2* | *PROX2* | *SOX4* | *THRB* | *ZFP42* | *ZNF251* | *ZNF440* | *ZNF593* | *ZNF764* | |
| *ATOH1* | *EBF3* | *GATA4* | *HOXC8* | *MEF2C* | *NR1D1* | *PRRX1* | *SOX5* | *TLX1* | *ZFP57* | *ZNF252P* | *ZNF441* | *ZNF594* | *ZNF765* | |
| *ATOH8* | *EBF4* | *GATA5* | *HOXC9* | *MEF2D* | *NR1D2* | *PRRX2* | *SOX6* | *TLX2* | *ZFP62* | *ZNF253* | *ZNF442* | *ZNF595* | *ZNF766* | |
| *BACH1* | *EGR1* | *GATA6* | *HOXD1* | *MEFV* | *NR1H2* | *PSPC1* | *SOX7* | *TLX3* | *ZFP64* | *ZNF254* | *ZNF443* | *ZNF597* | *ZNF768* | |
| *BACH2* | *EGR2* | *GATAD1* | *HOXD11* | *MEIS1* | *NR1H3* | *PTF1A* | *SOX8* | *TONSL* | *ZFP69* | *ZNF256* | *ZNF444* | *ZNF599* | *ZNF77* | |
| *BARHL1* | *EGR3* | *GATAD2A* | *HOXD12* | *MEIS2* | *NR1H4* | *PTX3* | *SOX9* | *TOX* | *ZFP69B* | *ZNF257* | *ZNF445* | *ZNF600* | *ZNF770* | |
| *BARHL2* | *EGR4* | *GATAD2B* | *HOXD13* | *MEIS3P1* | *NR1I2* | *PURA* | *SP1* | *TOX2* | *ZFP82* | *ZNF260* | *ZNF446* | *ZNF605* | *ZNF771* | |
| *BARX1* | *EHF* | *GBX1* | *HOXD3* | *MEOX1* | *NR1I3* | *PURB* | *SP100* | *TOX3* | *ZFP90* | *ZNF263* | *ZNF449* | *ZNF606* | *ZNF772* | |
| *BARX2* | *ELF1* | *GBX2* | *HOXD8* | *MEOX2* | *NR2C1* | *PURG* | *SP110* | *TOX4* | *ZFP91* | *ZNF264* | *ZNF45* | *ZNF607* | *ZNF773* | |
| *BATF* | *ELF2* | *GCM1* | *HOXD9* | *MESP1* | *NR2C2* | *RAD54L* | *SP140* | *TP53* | *ZFP92* | *ZNF266* | *ZNF451* | *ZNF610* | *ZNF774* | |
| *BATF2* | *ELF3* | *GCM2* | *HSF2* | *MESP2* | *NR2E1* | *RARA* | *SP140L* | *TP63* | *ZFX* | *ZNF267* | *ZNF454* | *ZNF611* | *ZNF775* | |
| *BATF3* | *ELF5* | *GFI1* | *HSF4* | *MIER1* | *NR2E3* | *RARB* | *SP2* | *TP73* | *ZFY* | *ZNF268* | *ZNF460* | *ZNF613* | *ZNF776* | |
| *BBX* | *ELK1* | *GFI1B* | *HSF5* | *MIER2* | *NR2F1* | *RARG* | *SP3* | *TPRX1* | *ZGLP1* | *ZNF271* | *ZNF461* | *ZNF614* | *ZNF777* | |
| *BCL11A* | *ELK3* | *GLI1* | *ID1* | *MIER3* | *NR2F2* | *RAX* | *SP4* | *TPRXL* | *ZGPAT* | *ZNF273* | *ZNF462* | *ZNF615* | *ZNF778* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *BCL3* | *ELK4* | *GLI2* | *ID2* | *MITF* | *NR2F6* | *RAX2* | *SP5* | *TSC22D1* | *ZHX1* | *ZNF274* | *ZNF467* | *ZNF616* | *ZNF780A* | |
| *BCL6* | *ELMSAN1* | *GLI3* | *ID3* | *MIXL1* | *NR3C1* | *RBAK* | *SP6* | *TSC22D2* | *ZHX3* | *ZNF275* | *ZNF468* | *ZNF618* | *ZNF780B* | |
| *BCL6B* | *EMX1* | *GLI4* | *ID4* | *MKX* | *NR3C2* | *RBP1* | *SP7* | *TSC22D3* | *ZIC1* | *ZNF276* | *ZNF469* | *ZNF619* | *ZNF781* | |
| *BHLHA9* | *EMX2* | *GLIS1* | *IKZF1* | *MLX* | *NR4A1* | *RBP2* | *SP8* | *TSC22D4* | *ZIC2* | *ZNF277* | *ZNF470* | *ZNF621* | *ZNF782* | |
| *BHLHE22* | *EN1* | *GLIS2* | *IKZF2* | *MLXIP* | *NR4A2* | *RBPJ* | *SP9* | *TSHZ1* | *ZIC3* | *ZNF28* | *ZNF471* | *ZNF623* | *ZNF784* | |
| *BHLHE23* | *EN2* | *GLIS3* | *IKZF3* | *MLXIPL* | *NR4A3* | *RBPJL* | *SPDEF* | *TSHZ2* | *ZIC4* | *ZNF280A* | *ZNF479* | *ZNF624* | *ZNF785* | |
| *BHLHE41* | *EOMES* | *GMEB1* | *IKZF4* | *MNT* | *NR5A1* | *RC3H2* | *SPI1* | *TSHZ3* | *ZIC5* | *ZNF280B* | *ZNF48* | *ZNF625* | *ZNF786* | |
| *BLZF1* | *EPAS1* | *GMEB2* | *IKZF5* | *MNX1* | *NR5A2* | *RCOR1* | *SPIB* | *TWIST1* | *ZIK1* | *ZNF280C* | *ZNF480* | *ZNF626* | *ZNF787* | |
| *BNC1* | *ERG* | *GRHL1* | *INSM1* | *MSC* | *NR6A1* | *RCOR2* | *SPIC* | *UBP1* | *ZIM2* | *ZNF280D* | *ZNF483* | *ZNF627* | *ZNF788* | |
| *BNC2* | *ESR1* | *GRHL2* | *INSM2* | *MSX1* | *NRF1* | *RCOR3* | *SREBF1* | *UBTF* | *ZIM3* | *ZNF281* | *ZNF484* | *ZNF628* | *ZNF789* | |
| *BRPF1* | *ESR2* | *GRHL3* | *IRF1* | *MSX2* | *NRL* | *REL* | *SREBF2* | *UBTFL1* | *ZKSCAN1* | *ZNF282* | *ZNF485* | *ZNF629* | *ZNF79* | |
| *BSX* | *ESRRA* | *GSC* | *IRF2* | *MTF1* | *OAZ1* | *RELA* | *SRF* | *UNCX* | *ZKSCAN2* | *ZNF283* | *ZNF490* | *ZNF639* | *ZNF790* | |
| *CASZ1* | *ESRRB* | *GSC2* | *IRF3* | *MUM1* | *OLIG1* | *RELB* | *SSRP1* | *USF1* | *ZKSCAN3* | *ZNF284* | *ZNF491* | *ZNF641* | *ZNF791* | |
| *CDC5L* | *ESRRG* | *GSX1* | *IRF5* | *MXD3* | *OLIG2* | *REST* | *ST18* | *USF2* | *ZKSCAN4* | *ZNF285* | *ZNF492* | *ZNF644* | *ZNF792* | |
| *CDX1* | *ESX1* | *GTF3A* | *IRF6* | *MXD4* | *OLIG3* | *RFX1* | *STAT1* | *UTF1* | *ZKSCAN5* | *ZNF286A* | *ZNF493* | *ZNF646* | *ZNF793* | |
| *CDX2* | *ETF1* | *GZF1* | *IRF7* | *MXI1* | *ONECUT1* | *RFX2* | *STAT2* | *VAX1* | *ZNF10* | *ZNF286B* | *ZNF496* | *ZNF648* | *ZNF799* | |
| *CDX4* | *ETS1* | *HAND1* | *IRF8* | *MYB* | *ONECUT2* | *RFX3* | *STAT3* | *VAX2* | *ZNF100* | *ZNF287* | *ZNF497* | *ZNF649* | *ZNF8* | |
| *CEBPA* | *ETS2* | *HAND2* | *IRF9* | *MYBL1* | *ONECUT3* | *RFX4* | *STAT4* | *VDR* | *ZNF101* | *ZNF292* | *ZNF500* | *ZNF652* | *ZNF80* | |
| *CEBPB* | *ETV1* | *HBP1* | *IRX1* | *MYBL2* | *OSR1* | *RFX5* | *STAT5A* | *VENTX* | *ZNF107* | *ZNF296* | *ZNF502* | *ZNF653* | *ZNF800* | |
| *CEBPD* | *ETV3* | *HDX* | *IRX2* | *MYCL1* | *OSR2* | *RFX6* | *STAT5B* | *VEZF1* | *ZNF114* | *ZNF3* | *ZNF506* | *ZNF654* | *ZNF805* | |
| *CEBPE* | *ETV3L* | *HELT* | *IRX3* | *MYCN* | *OTP* | *RFX7* | *STAT6* | *VSX1* | *ZNF117* | *ZNF30* | *ZNF507* | *ZNF655* | *ZNF808* | |
| *CEBPG* | *ETV4* | *HELZ2* | *IRX4* | *MYNN* | *OTX1* | *RFX8* | *SUZ12* | *VSX2* | *ZNF124* | *ZNF300* | *ZNF510* | *ZNF658* | *ZNF81* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *CIC* | *ETV5* | *HES1* | *IRX5* | *MYOD1* | *OVOL1* | *RFXANK* | *T* | *WHSC1* | *ZNF131* | *ZNF302* | *ZNF512* | *ZNF662* | *ZNF812* | |
| *CLOCK* | *ETV6* | *HES2* | *IRX6* | *MYOG* | *OVOL2* | *RFXAP* | *TADA2A* | *WIZ* | *ZNF132* | *ZNF304* | *ZNF512B* | *ZNF664* | *ZNF813* | |
| *CNOT4* | *ETV7* | *HES3* | *ISL1* | *MYT1* | *OVOL3* | *RIT1* | *TADA2B* | *WT1* | *ZNF133* | *ZNF311* | *ZNF513* | *ZNF665* | *ZNF814* | |
| *CREB1* | *EVX1* | *HES4* | *ISL2* | *MYT1L* | *PATZ1* | *RLF* | *TAF1* | *YBX1* | *ZNF134* | *ZNF317* | *ZNF514* | *ZNF667* | *ZNF816* | |
| *CREB3* | *EVX2* | *HES5* | *ISX* | *MZF1* | *PAX1* | *RORA* | *TAL1* | *YBX2* | *ZNF135* | *ZNF319* | *ZNF516* | *ZNF668* | *ZNF827* | |
| *CREB3L1* | *FERD3L* | *HES6* | *JARID2* | *NANOG* | *PAX2* | *RORB* | *TBP* | *YY1* | *ZNF136* | *ZNF32* | *ZNF517* | *ZNF669* | *ZNF829* | |
| *CREB3L2* | *FEV* | *HES7* | *JDP2* | *NCOA1* | *PAX3* | *RORC* | *TBPL1* | *YY2* | *ZNF137P* | *ZNF320* | *ZNF518A* | *ZNF671* | *ZNF83* | |
| *CREB3L3* | *FEZF1* | *HESX1* | *JUN* | *NCOA2* | *PAX4* | *RREB1* | *TBPL2* | *ZBED1* | *ZNF138* | *ZNF322* | *ZNF518B* | *ZNF672* | *ZNF835* | |
| *CREB3L4* | *FEZF2* | *HEY1* | *JUNB* | *NCOA3* | *PAX5* | *RUNX1* | *TBR1* | *ZBED2* | *ZNF14* | *ZNF324* | *ZNF519* | *ZNF674* | *ZNF836* | |
| *CREB5* | *FGF4* | *HEY2* | *JUND* | *NCOR1* | *PAX6* | *RUNX2* | *TBX1* | *ZBED3* | *ZNF140* | *ZNF324B* | *ZNF521* | *ZNF676* | *ZNF837* | |
| *CREBL2* | *FIGLA* | *HEYL* | *KAT5* | *NCOR2* | *PAX7* | *RUNX3* | *TBX10* | *ZBED4* | *ZNF141* | *ZNF329* | *ZNF524* | *ZNF677* | *ZNF84* | |
| *CREBZF* | *FIZ1* | *HHEX* | *KAT6A* | *NEUROD1* | *PAX8* | *RXRA* | *TBX15* | *ZBED5* | *ZNF142* | *ZNF331* | *ZNF525* | *ZNF678* | *ZNF841* | |
| *CREM* | *FLI1* | *HIC1* | *KAT6B* | *NEUROD2* | *PAX9* | *RXRB* | *TBX18* | *ZBED6* | *ZNF143* | *ZNF333* | *ZNF526* | *ZNF680* | *ZNF844* | |
| *CRX* | *FOSB* | *HIC2* | *KAT7* | *NEUROD4* | *PBRM1* | *RXRG* | *TBX19* | *ZBTB1* | *ZNF146* | *ZNF334* | *ZNF527* | *ZNF681* | *ZNF845* | |
| *CSRNP1* | *FOSL1* | *HIF1A* | *KAT8* | *NEUROD6* | *PBX1* | *SALL1* | *TBX2* | *ZBTB11* | *ZNF148* | *ZNF335* | *ZNF528* | *ZNF682* | *ZNF846* | |
| *CSRNP2* | *FOSL2* | *HIF3A* | *KDM2A* | *NEUROG1* | *PBX3* | *SALL2* | *TBX20* | *ZBTB12* | *ZNF154* | *ZNF337* | *ZNF529* | *ZNF683* | *ZNF85* | |
| *CSRNP3* | *FOXA1* | *HINFP* | *KDM5B* | *NEUROG2* | *PBX4* | *SALL3* | *TBX21* | *ZBTB16* | *ZNF155* | *ZNF33A* | *ZNF532* | *ZNF684* | *ZNF853* | |
| *CTCF* | *FOXA2* | *HIVEP1* | *KDM5C* | *NFAT5* | *PCM1* | *SALL4* | *TBX22* | *ZBTB17* | *ZNF157* | *ZNF33B* | *ZNF534* | *ZNF687* | *ZNF860* | |
| *CTCFL* | *FOXA3* | *HIVEP2* | *KDM5D* | *NFATC1* | *PDX1* | *SATB1* | *TBX3* | *ZBTB2* | *ZNF16* | *ZNF34* | *ZNF536* | *ZNF689* | *ZNF876P* | |
| *CUX1* | *FOXB1* | *HIVEP3* | *KLF1* | *NFATC2* | *PEG3* | *SATB2* | *TBX4* | *ZBTB20* | *ZNF160* | *ZNF341* | *ZNF540* | *ZNF69* | *ZNF878* | |
| *CUX2* | *FOXB2* | *HKR1* | *KLF10* | *NFATC3* | *PGR* | *SCRT1* | *TBX5* | *ZBTB24* | *ZNF165* | *ZNF343* | *ZNF542* | *ZNF691* | *ZNF879* | |
| *CXXC1* | *FOXC1* | *HLF* | *KLF11* | *NFATC4* | *PHF5A* | *SCRT2* | *TBX6* | *ZBTB25* | *ZNF169* | *ZNF345* | *ZNF543* | *ZNF692* | *ZNF880* | |
| *DBP* | *FOXC2* | *HLX* | *KLF12* | *NFE2* | *PHOX2A* | *SEBOX* | *TCF12* | *ZBTB26* | *ZNF17* | *ZNF347* | *ZNF544* | *ZNF695* | *ZNF883* | |
| *DBX1* | *FOXD1* | *HMBOX1* | *KLF13* | *NFE2L1* | *PHOX2B* | *SFPQ* | *TCF15* | *ZBTB32* | *ZNF174* | *ZNF35* | *ZNF546* | *ZNF696* | *ZNF90* | |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| *DBX2* | *FOXD2* | *HMG20A* | *KLF14* | *NFE2L2* | *PHTF1* | *SHOX* | *TCF21* | *ZBTB33* | *ZNF175* | *ZNF350* | *ZNF547* | *ZNF697* | *ZNF91* | |
| *DDIT3* | *FOXD3* | *HMG20B* | *KLF15* | *NFE2L3* | *PHTF2* | *SHOX2* | *TCF23* | *ZBTB34* | *ZNF18* | *ZNF354A* | *ZNF548* | *ZNF699* | *ZNF92* | |
| *DEAF1* | *FOXD4* | *HMGA1* | *KLF16* | *NFIA* | *PITX1* | *SIM1* | *TCF25* | *ZBTB37* | *ZNF180* | *ZNF354B* | *ZNF549* | *ZNF7* | *ZNF93* | |
| *DLX1* | *FOXD4L1* | *HMGA2* | *KLF17* | *NFIB* | *PITX2* | *SIM2* | *TCF7* | *ZBTB38* | *ZNF181* | *ZNF354C* | *ZNF550* | *ZNF70* | *ZNF98* | |

| | | Primer sequence 5′→3′ | | | | | |
|---|---|---|---|---|---|---|---|
| GENE | Proband | SNV | Forward | Reverse | Product size (bp) | Amplification Conditions (°C) | Validation method |
| | | | | | | | |
| *PPP2R3A* | OX6136 | c.736delT: p.C246fs | CCCATCCTTTGGTTTACTGCGG AGTTCCTC | ACT GAT CAG TTC TGT TAA ATC TAG AGC CTC | 424 | 65 | Dideoxy seq |
| | OX4481 | Intron 12-13delATCA;(sp) | TACCTTAGCACCTCTTTTAAAC CTGTTGAA | TGT AAG GGG GTT TTA ATA CTC TGG AC | 324 | 65 | Dideoxy seq |
| | | | | | | | |
| *FHOD3* | OX4580 | c.A3527C: p.D1176A | GTTTTAATTTACAGTTCCCAAA GATTTATT | TAC CTG TTG ATT ATC CTT CTA TGG ACA A | 473 | 65 | Dideoxy seq |
| | OX5569 | c.C2354T: p.P785L | CACTTATTCTCATCGATTCTCTC ATGGAAGTGAG | TTG GCC ATG AGC TGG TCC CAG ATG TAA TC | 502 | 65 | Dideoxy seq |
| | | | | | | | |
| ZNF16 | OX797RB | c.T1277C: p.V426A | GCTGAGCTGTGGCTGAAGGCCT TCCCAC | AGC CTT ATG AGT GTA ATG ATT GTG GCA A | 301 | 65 | Dideoxy seq |
| | OX4580 | c.A970G: p.N324D | CTCCAGTGTGGACCCTCTGGTG CTTCCTCA | TGA TGA TTG TGG GAA AAC CTT CAG CC | 488 | 65 | Dideoxy seq |

Table 32 Primers and conditions used for validation of variants identified from exome sequencing 19 mutation-negative patients with coronal synostosis.