**Twin Research and Human Genetics**

**Prevalence of germline *BAP1, CDKN2A* and *CDK4* mutations in a population-based sample of cutaneous melanoma cases**

**SUPPLEMENTARY MATERIAL**

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**Supplementary Table S1**

Primers for Sanger sequencing validation

|  |  |
| --- | --- |
| **Amplicon** | **Primer Sequence** |
| BAP1 exon 5 forward | TGGGTATTTGGTAGGTGCTTG |
| BAP1 exon 5 reverse | CCCGCAACTGCATCTAAAAAC |
| BAP1 exon 6 and 7 forward | TCCACCCATAGTCCTACCTGA |
| BAP1 exon 6 and 7 reverse | GGGCAATATGGTGTAGGGTGA |
| BAP1 exon 9 forward | TTCCAGATAGGCCCCTCATAC |
| BAP1 exon 9 reverse | GTGGTTAGCTGAAGCCCAGAT |
| BAP1 exon 11 forward | GGGGAGACTGTGAGCTTTTCT |
| BAP1 exon 11 reverse | ATCAGGCAGAGGAACCTAGCA |
| BAP1 exon 13 forward | TTCTGGGTACTGCTGGGTATG |
| BAP1 exon 13 reverse | GGACACTTTGTGGTCACTTGG |
| BAP1 exon 14 forward | CGTGTTGACTGCATACGCTAC |
| BAP1 exon 14 reverse | CCAATCTTCACACCAAAGTTCC |
| P16 exon 1 forward | ACGCACTCAAACACGCCTTTG |
| P16 exon 1 reverse | CAAACTTCGTCCTCCAGAGTC |
| P16 exon 2 forward | CAGAAGTTCGGAGGATATAATG |
| P16 exon 2 reverse | GGGCTGAACTTTCTGTGCTG |
| P14 forward | CCTCGCTTTCCTTTCTTCCT |
| P14 reverse | CGGTTATCTCCTCCTCCTCC |

**Supplementary Table S2:** Summary of cancers in high-risk families reported by Aitken et al., 1999

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| proband | gene | variant | ages of onset of CMM | other cancers in proband (age) | other cancers in family (age) |
| 001790 | CDKN2A (p16) | p.R24P | 28, 29, 36, 40 |  | MPM (4 additional cases), CMM (42), CMM (65), CMM (65), CMM (72), CMM (72), breast (84), bladder (43), uterus (76) |
| 006581 | CDKN2A (p16) | p.L32P | 30, 43, 51, 56 | pancreas (54) | CMM (33), MPM (3 cases), bronchogenic (59), bowel (50), lung (65) |
| 004197 | CDKN2A (p16) | p.G35A | 31, 31, 31, 32, 35 |  | CMM (39), CMM (40), CMM (41), CMM (45), CMM (51), CMM (54), CMM (70), breast (39), lymphoma (62), Hodgkin’s lymphoma (37), non-Hodgkin’s lymphoma (52) |
| 005526 | CDKN2A (p16) | p.M53I | 37 |  | CMM (34), CMM (53), MPM (2 cases), ovary (41), carcinomatosis (49), caecum (83), liver (56) |
| 003301 | CDKN2A (p16) | p.M53I | 26, +3 |  | CMM (23), CMM (26), CMM (27), CMM (52), MPM (9 cases), rectum (43), prostate (72), breast (43), breast (48), breast (56), bile duct (64) |
| 004478 | CDKN2A (p16) | p.D108N | 36, 38, 39, 46, 49, 58, 59, 63, 63, 64, 66, 66 |  | CMM (19), CMM (46), MPM (28) |
| 002622 | CDKN2A (p16) | 24 bp deletion | 28 |  | CMM (37), CMM (44) |
| 000397 | CDKN2A (p16) | 24 bp duplication | 38, 43, 43, 44, 44, 48, 49, 53 |  | CMM (27), CMM (30), CMM (33), CMM (40), CMM (41), MPM (2 cases), lung (60), carcinomatosis (79) |
| 002899 | CDKN2A (p16) | Nt46delC | 47 |  | CMM (27), CMM (34), CMM (40), MPM (3 cases), stomach (52) |

Note: CMM is cutaneous malignant melanoma; MPM is multiple primary melanomas