## Table 1S: Primer Sequences for Polymorphisms in the PKLR Region

| POSITION Relative to PKLR | GENE | RSNUMBER | UCSC Mar2006 build 126 | Primer direction | Primer sequence |
|---|---|---|---|---|---|
| (-)202210 | ASH1L | rs1360554 | 153738595 | Forward | CACGACGTTGTAAAACGACATAGTAAATTAGGACTTGGCTG |
| | | | | Reverse | TGCTGCTTAATGTAAATACGGATCTT |
| | | | | Sequence | AATACTGATTATTAAAAGAA |
| (-)162756 | ASH1L | rs12724079 | 153700566 | Forward | CGCCATAGCTTTTGCAGAGT |
| | | | | Reverse | CACGACGTTGTAAAACGACACATCCTTCCAGGCAGATCTAA |
| | | | | Sequence | CTCCATCATGTGACAAAT |
| (-)143582 | ASH1L | rs12730906 | 153681392 | Forward | CACGACGTTGTAAAACGACTTTTTAAATGGTCCTTGCTTTTT |
| | | | | Reverse | CACTGACCCTGTTTCATACCAC |
| | | | | Sequence | AAAATCTTTAAGACCTTGGG |
| (-)142118 | ASH1L | rs1325908 | 153679928 | Forward | CGGTAGTCTAGGACATGATGCTGC |
| | | | | Reverse | CACGACGTTGTAAAACGACCTTCTCTGAGGCTGTGGATGACAG |
| | | | | Sequence | TTGAGTTTTGAATAATTTAC |
| (-)141744 | ASH1L | rs11264372 | 153679554 | Forward | TTCACCAAAATCACTTTTTATCTCC |
| | | | | Reverse | CACGACGTTGTAAAACGACTGTACCCATTTTAGGATTCTCTTTT |
| | | | | Sequence | CATTAACACGTAGTTGAAGA |
| (-)39257 | ASH1L | rs12748814 | 153577067 | Forward | ACAGGCCCACTAAAAGAAGACTCA |
| | | | | Reverse | CACGACGTTGTAAAACGACAGGCTTTTTCCTGGTATTTTTTCT |
| | | | | Sequence | ACAAAACGTATAATTGGC |
| (-)19401 | c1orf104 | rs16836822 | 153557211 | C__27848527_10 | CAAAAGCTTAGGATATATCAAACCC[G/T]CTAGGTGGGTGACAAAAGACGCTAA |
| (-)18359 | FDPS | rs11264361 | 153556169 | Forward | CACGACGTTGTAAAACGACGAAGATCCTGCTGGAGATGG |
| | | | | Reverse | GTCCCCAAAGAGGTCAAGGTA |
| | | | | Sequence | AGCAAAAGAGGAAGGATG |
| (-)11643 | FDPS | rs11264359 | 153549453 | Forward | CACGACGTTGTAAAACGACGACAGAGGTCTTCCAAAGGCTATTC |
| | | | | Reverse | GTGTCAGTGAGCTGCCAGTTTT |
| | | | | Sequence | GAGCTGCCAGTTTTCAAT |
| (-)8296 | FDPS | rs2297480 | 153546106 | C___2737970_10 | AGGGAGGGGCACTCTGGGCTAAGGC[G/T]GGGGTGGGAGTTATCTGGGGAGCTG |
| (-)3774 | PKLR | rs12032720 | 153541584 | Forward | CACGACGTTGTAAAACGACAGATCTGGGGTAATGCACTATCCG |
| | | | | Reverse | GAATGGGCAGGGTGTTGGT |
| | | | | Sequence | GGCAGGGTGTTGGTA |
| (-)2683 | PKLR | rs4971072 | 153540493 | Forward | CACGACGTTGTAAAACGACGTCGTGGCCCATAGTGAAGATA |
| | | | | Reverse | CGGGAGGGACCTAAACCAT |
| | | | | Sequence | TCCATCCCCTGGGTA |
| 1410 | PKLR | rs3020781 | 153536400 | Forward | CCACACAGGGCCTATTGGTA |

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|---|---|---|---|---|---|
| | | | | Reverse | Biotinylated-ACTGATCTACTCCCAGGGCC |
| | | | | Sequence | TTTGACACAGTCATTCATT |
| 6009 | PKLR | rs2071053 | 153531801 | Forward | GACACCAAGGGACCGGAGAT |
| | | | | Reverse | Biotinylated-GATGTGAGTTCTGAGCCCCG |
| | | | | Sequence | CCGGAAAGGCGGC |
| 8573 | PKLR | rs4620533 | 153529237 | Forward | TCCAACCCCATGAAGAACAAAT |
| | | | | Reverse | CACGACGTTGTAAAACGACCCCACAGAGCCTACCAATACTGAGG |
| | | | | Sequence | CCTGTTCACGAGGAAAGA |
| 10803 | PKLR | rs1052176 | 153527007 | Forward | CTGGTGATTGTGGTGACAGG |
| | | | | Reverse | CACGACGTTGTAAAACGACAGGAAGGGATGGGGTACAAG |
| | | | | Sequence | CGGCTACACCAACATC |
| 10836 | PKLR | rs1052177 | 153526974 | Forward | CTGGTGATTGTGGTGACAGG |
| | | | | Reverse | CACGACGTTGTAAAACGACAGGAAGGGATGGGGTACAAG |
| | | | | Sequence | TATCCTGAGACGCCCCTCCC |
| 11090 | PKLR | rs932972 | 153526720 | Forward | CACGACGTTGTAAAACGACAAAGACTCAAGGCATCTTAGGG |
| | | | | Reverse | CCATCTCCATTTCACACAGGTCC |
| | | | | Sequence | ACAGCACCAATAGTACATTC |
| 11863 | HCN3 | rs8847 | 153525947 | Forward | CACGACGTTGTAAAACGACTTGCCCAAACCCATCAGC |
| | | | | Reverse | ATTCCCTTATTCCTGCACCAACAGC |
| | | | | Sequence | AACAAAACTCTAAAGGTAAA |
| 12434 | HCN3 | rs3814319 | 153525376 | Forward | GGTCTGGTTATCTTCCTGTTCCTG |
| | | | | Reverse | CACGACGTTGTAAAACGACGGTAAAAGTGGATACCCACACTCAG |
| | | | | Sequence | ATAGCCAGGTACTGCCA |
| 13695 | HCN3 | rs11264355 | 153524116 | Forward | GCTGCTCCGGTTCATAGATTATACTACTGACTCCCATCCCCTACC |
| | | | | Reverse | TGTACCGGGAGGGAGAAGTTAAG |
| | | | | Sequence | TGTTGGATATCAAAGAGAATG |
| 15280 | HCN3 | rs11264353 | 113522530 | RE_Forward | CCTACTTTGGGGGTCAGCA |
| | | | | RE_Reverse | CAACCTGGCTGTGCATTAGA |
| 15349 | HCN3 | rs11264352 | 153522461 | Forward | CACGACGTTGTAAAACGACCCTCACCGATGGATCCTACTTT |
| | | | | Reverse | AGCTCTGTGATTCTGCAGCGTACT |
| | | | | Sequence | GTGATTCTGCAGCGTACT |
| 15576 | HCN3 | novel | 153522688 | Forward | CACGACGTTGTAAAACGACCCCCTGTCCAGGAGATCATTA |
| | | | | Reverse | AAAGTAGGATCCATCGGTGAGG |
| | | | | Sequence | CCCGGCTGGAAGACCTCAA |
| 16727 | HCN3 | novel | 153521083 | Forward | CACGACGTTGTAAAACGACAGTATTCCCATGCCCTGTTCAAG |

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|---|---|---|---|---|---|
| | | | | Reverse | TGTCCCTGCTGACCTTCTCC |
| | | | | Sequence | GGCGTAGCATGTGGCACC |
| 17603 | HCN3 | rs7520184 | 153520207 | Forward | CACGACGTTGTAAAACGACTGTGGTTCTGGGCACATGACT |
| | | | | Reverse | GTCATTTGTGTGGAGTTTTGTGGCA |
| | | | | Sequence | CATGTATAGCTCATGCAAAG |
| 22191 | HCN3 | rs7551854 | 153515621 | Forward | CCGGACTTGAGGAAGTTTCTTT |
| | | | | Reverse | CACGACGTTGTAAAACGACGGAACTGTGTGAAGCCCTCAT |
| | | | | Sequence | CTCCTGAGTTGACACTGG |
| 22611 | HCN3 | rs7549276 | 153515199 | Forward | CACGACGTTGTAAAACGACGGAGCCTCGAAATACAGTTGT |
| | | | | Reverse | GAGAGTGAGATCCACAGGAGACTA |
| | | | | Sequence | TGGGAACGAGGCAGCAAA |
| 23073 | HCN3 | rs11264349 | 153514737 | Forward | CACGACGTTGTAAAACGACAAGTATCGGGGCCAGTCTGA |
| | | | | Reverse | CTTCACCACTAGGGACAGCG |
| | | | | Sequence | TTTTTCCTGGTAGTCTGT |
| 23479 | HCN3 | rs12749306 | 153514331 | Forward | CACGACGTTGTAAAACGACGGAGGGCAGGGTACATCAAT |
| | | | | Reverse | GCTAGACTGGTGCAAGTGACTCTC |
| | | | | Sequence | GTCCCGGTGTCGCTG |
| 23599 | HCN3 | novel | 153514211 | Forward | GCTGCTCCGGTTCATAGATTGGTACCTGATGGCAACAG |
| | | | | Reverse | AATACCGGAAGTCGCTGTAG |
| | | | | Sequence | CTCCTGCTCGATTTCCACTG |
| 27094 | CLK2 | rs12563994 | 153510716 | Forward | CACGACGTTGTAAAACGACAGTGCAATGGCCCGATCT |
| | | | | Reverse | GGTGGCTTACATAATGTGTGA |
| | | | | Sequence | AGGTAGGAGAATCGCTTGAAC |
| 29825 | CLK2 | rs2236863 | 153507985 | Forward | TTCTATCCTTATTTTCTTCAAACACAGC |
| | | | | Reverse | TCTGCTGCTCCGGTTCATAGATTTGGCAGTCTCCATCGGTCGTC |
| | | | | Sequence | TTGAAACACAGCTCCATGAA |
| 31562 | CLK2 | rs2361543 | 153506248 | Forward | ACCTGTGACAAGGTCCTGCCTTA |
| | | | | Reverse | TCTGCTGCTCCGGTTCATAGATTTGCCTTTTCAGAGGTTTTGGTGAC |
| | | | | Sequence | GCTAAGAAAAACTGTTCTCA |
| 33982 | CLK2 | rs4971070 | 153503828 | Forward | CGGATTTAGGATGGACAGTTTGATGATT |
| | | | | Reverse | CACGACGTTGTAAAACGACCCAGCCCAAGTTTCAAAATATCT |
| | | | | Sequence | GTTTTTGAACTATTTAGAATGA |
| 34353 | CLK2 | novel | 153503457 | Forward | TGGTAGAGGCTACTCCTTA |
| | | | | Reverse | TGGTAGAGGCTACTCCTTA |
| | | | | Sequence | TTAGTACCATTCTTCTTGGGTCT |

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|---|---|---|---|---|---|
| 35693 | CLK2 | rs1078699 | 153502117 | Forward | CACGACGTTGTAAAACGACGAAAGTTATCTGAAGTCCCCTGT |
| | | | | Reverse | TCCTCCCACATTTTCTCCCTGTCTG |
| | | | | Sequence | TAAATAGGTTGAAAGCGAGA |
| 36365 | CLK2 | rs1076556 | 153501445 | Forward | GCCTTCCAAGCCCTGG |
| | | | | Reverse | CACGACGTTGTAAAACGACCATATTTTTAGAGAAACTCCTTTGC |
| | | | | Sequence | GAAGACTACACTCAAGTGCC |
| 37782 | CLK2 | novel | 153500028 | GEL-Forward | GCCCCCGCCGCAGAGATCAAAGCAATGTCCTGGATTC |
| | | | | GEL-Reverse | CGACTGATATCCCGACTGGAG |
| 39112 | SCAMP3 | rs1046188 | 153498698 | RE-Forward | TCCATCTCGGGAGACTAGGAAG |
| | | | | RE-Reverse | ACCTCAAATCCATCATCCAACACTGC |
| 39641 | SCAMP3 | rs2242576 | 153498169 | RE-Forward | TTGTGACATTTGATCCTGGGAAGAGCC |
| | | | | RE-Reverse | CTCCAACCAGCCCCAACCT |
| 40754 | SCAMP3 | novel | 153494076 | Forward | ATTCCTGTGTTATGAGGGACTAGATG |
| | | | | Reverse | CACGACGTTGTAAAACGACCCTGAGTGCTGTATGAGCCATAGTT |
| | | | | Sequence | CAGCCACCACCAGCCTAT |
| 41055 | SCAMP3 | rs3180018 | 153496775 | Forward | AGTTGGACCGAAGGGAGCGAGA |
| | | | | Reverse | CACGACGTTGTAAAACGACCACCCAGCCGAGCCCAGG |
| | | | | Sequence | CTGCAGCATGTTGCCATGGG |
| 45338 | SCAMP3 | novel | 153489492 | Forward | TCCTCCGTAGTGCTCCCAATCCTAT |
| | | | | Reverse | CACGACGTTGTAAAACGACGAAGCAACAGCCGTGGCACA |
| | | | | Sequence | CACCTCTCACACGCC |
| 46885 | C1ORF2 | rs2242577 | 153490925 | Forward | CACGACGTTGTAAAACGACGACACAATGCCAACCACCC |
| | | | | Reverse | ACTCCTGGTCCTCTGCCTCCTTA |
| | | | | Sequence | TGCATCCTGTGGTGAGGG |

Primer sequences are provided for amplification and sequencing primers. Unless otherwise noted, assays were performed by Pyrosequencing. Other assays were restriction digest (RE) or size separation on sodium dodecyl sulfate/polyacrylamide gels (GEL). Primers assayed by Taqman used Assay by Design (Applied Biosystems, Inc); sequence of polymorphism is provided.