|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Supplement Table 1. List of all SNPs assessed | | | | | | |  |  | |  |  | |  | | |  | |  | |
|  |  | | | **Chromosome** | **Gene** |  | |  | **Major/ Minor** | | | **MAF**1 | | | **FDR HWE *p*** | | | |
| **Gene** | | **Alias** | | **Location** | **PARTP** | **dbSNP ID** | | **Coordinate** | | **Allele** | **NHW** | | | **HISP** | **NHW** | | **HISP** | |
| *ACVR1* | | *ACTRI, ACVR1A, ALK2,SKR1,* | | 2q23-q24 | 0.87 | rs2033962 | | 158638441 | | G/T | 0.17 | | | 0.14 | 0.96 | | 0.68 | |
|  | |  |  | rs4380178 | | 158668445 | | G/A | 0.14 | | | 0.13 | 1.00 | | 0.63 | |
|  | | *ACVRLK2,* | |  |  | rs1220134 | | 158603556 | | T/A | 0.29 | | | 0.20 | 0.96 | | 0.81 | |
|  | | *FOP, TSRI* | |  |  | rs920522 | | 158705824 | | T/C | 0.07 | | | 0.08 | 0.96 | | 0.78 | |
|  | |  | |  |  | rs2165436 | | 158726896 | | C/T | 0.15 | | | 0.14 | 0.96 | | 0.76 | |
|  | |  | |  |  | rs6437117 | | 158723183 | | A/T | 0.15 | | | 0.12 | 0.89 | | 0.49 | |
|  | |  | |  |  | rs17182166 | | 158596197 | | G/T | 0.17 | | | 0.07 | 0.86 | | 0.68 | |
|  | |  | |  |  | rs1146035 | | 158633411 | | G/T | 0.18 | | | 0.13 | 0.96 | | 0.74 | |
|  | |  | |  |  | rs17798043 | | 158663277 | | C/T | 0.08 | | | 0.04 | 0.97 | | 0.002 | |
|  | |  | |  |  | rs10497191 | | 158667217 | | C/T | 0.13 | | | 0.14 | 0.98 | | 0.98 | |
|  | |  | |  |  | rs10497192 | | 158671700 | | T/C | 0.26 | | | 0.27 | 1.00 | | 0.94 | |
|  | |  | |  |  | rs4233672 | | 158691926 | | G/A | 0.17 | | | 0.18 | 0.96 | | 0.92 | |
|  | |  | |  |  | rs10933443 | | 158694121 | | T/C | 0.25 | | | 0.25 | 0.96 | | 0.64 | |
|  | |  | |  |  | rs2883605 | | 158696529 | | G/T | 0.09 | | | 0.05 | 0.96 | | 0.87 | |
|  | |  | |  |  | rs10497193 | | 158709678 | | A/G | 0.17 | | | 0.14 | 1.00 | | 0.59 | |
|  | |  | |  |  | rs4664901 | | 158712538 | | T/C | 0.23 | | | 0.22 | 0.96 | | 0.97 | |
| *ACVR2A* | | *ACTRII,* | | 2q22.3 | 0.80 | rs1014064 | | 148612154 | | A/G | 0.31 | | | 0.44 | 0.96 | | 0.95 | |
|  | | *ACVR2* | |  |  | rs2161983 | | 148649386 | | C/T | 0.31 | | | 0.44 | 0.96 | | 0.81 | |
|  | |  | |  |  | rs3768687 | | 148672020 | | G/A | 0.31 | | | 0.44 | 0.96 | | 0.83 | |
|  | |  | |  |  | rs3768688 | | 148671101 | | C/T | 0.42 | | | 0.40 | 0.62 | | 0.59 | |
|  | |  | |  |  | rs1424941 | | 148643118 | | G/A | 0.21 | | | 0.10 | 0.98 | | 0.81 | |
|  | |  | |  |  | rs10497025 | | 148662202 | | C/G | 0.25 | | | 0.20 | 0.96 | | 0.57 | |
| *ACVR2B* | | *ACTRIIB,* | | 3p22 | 0.82 | rs928813 | | 38528537 | | G/T | 0.57 | | | 0.34 | 0.96 | | 0.20 | |
|  | | *MGC116908,* | |  |  | rs2276541 | | 38512779 | | A/G | 0.38 | | | 0.49 | 0.96 | | 0.84 | |
|  | | *ActR-IIB* | |  |  | rs503327 | | 38524363 | | G/A | 0.11 | | | 0.05 | 0.91 | | 0.94 | |
| *ACVRL1* | | *HHT, HHT2,* | | 12q11-q14 | 0.91 | rs7956340 | | 52302443 | | T/G | 0.08 | | | 0.24 | 0.96 | | 0.68 | |
|  | | *ALK-1, ALK1,* | |  |  | rs11169953 | | 52304399 | | C/T | 0.32 | | | 0.29 | 0.98 | | 0.59 | |
|  | | *ACVRLK1,TSR-I,* | |  |  | rs706819 | | 52315923 | | G/A | 0.26 | | | 0.50 | 1.00 | | 0.86 | |
|  | | *ORW2, SKR3,* | |  |  | rs2641534 | | 52317973 | | C/T | 0.12 | | | 0.14 | 0.97 | | 0.27 | |
| *BMP1* | | *FLJ44432,* | | 8p21 | 0.21 | rs7592 | | 22021883 | | G/A | 0.33 | | | 0.29 | 0.96 | | 0.59 | |
|  | | *PCOLC,* | |  |  | rs7812993 | | 22025644 | | A/G | 0.22 | | | 0.20 | 0.96 | | 0.63 | |
|  | | *PCP,* | |  |  | rs4872360 | | 22028668 | | T/C | 0.27 | | | 0.23 | 0.96 | | 0.91 | |
|  | | *TLD* | |  |  | rs12114940 | | 22048790 | | T/G | 0.41 | | | 0.48 | 0.96 | | 0.88 | |
|  | |  | |  |  | rs3924231 | | 22049780 | | T/C | 0.17 | | | 0.17 | 0.86 | | 0.43 | |
|  | |  | |  |  | rs3924229 | | 22049892 | | T/C | 0.11 | | | 0.07 | 0.91 | | 0.75 | |
|  | |  | |  |  | rs4075478 | | 22053123 | | T/C | 0.36 | | | 0.54 | 0.62 | | 0.52 | |
|  | |  | |  |  | rs3857979 | | 22055348 | | C/T | 0.53 | | | 0.40 | 0.89 | | 0.30 | |
|  | |  | |  |  | rs11775186 | | 22058122 | | T/G | 0.10 | | | 0.05 | 0.96 | | 0.23 | |
|  | |  | |  |  | rs13257482 | | 22059606 | | G/A | 0.22 | | | 0.24 | 0.98 | | 0.06 | |
| *BMP2* | | *BMP2A* | | 20p12 | 0.04 | rs1979855 | | 6747607 | | T/C | 0.18 | | | 0.10 | 0.89 | | 0.63 | |
|  | |  | |  |  | rs7270163 | | 6751316 | | A/G | 0.11 | | | 0.17 | 0.99 | | 0.85 | |
|  | |  | |  |  | rs1005464 | | 6756148 | | G/A | 0.23 | | | 0.22 | 1.00 | | 0.61 | |
|  | |  | |  |  | rs15705 | | 6759980 | | A/C | 0.23 | | | 0.14 | 0.96 | | 0.27 | |
|  | |  | |  |  | rs3178250 | | 6760201 | | T/C | 0.21 | | | 0.13 | 0.96 | | 0.45 | |
|  | |  | |  |  | rs235770 | | 6761765 | | C/T | 0.36 | | | 0.31 | 0.96 | | 0.95 | |
| *BMP4* | | *BMP2B,* | | 14q22-q23 | 0.02 | rs17563 | | 54417522 | | T/C | 0.56 | | | 0.31 | 0.86 | | 0.06 | |
|  | | *BMP2B1* | |  |  | rs762642 | | 54423053 | | T/G | 0.41 | | | 0.42 | 0.98 | | 0.76 | |
|  | | *ZYME* | |  |  | rs2761887 | | 54425052 | | A/C | 0.44 | | | 0.43 | 0.96 | | 0.69 | |
|  | |  | |  |  | rs4898820 | | 54427057 | | T/G | 0.47 | | | 0.49 | 0.96 | | 0.81 | |
| *BMP6* | | *VGR, VGR1* | | 6p24-p23 | 0.14 | rs267190 | | 7842121 | | T/G | 0.58 | | | 0.35 | 1.00 | | 0.02 | |
|  | |  | |  |  | rs267204 | | 7860158 | | A/G | 0.22 | | | 0.13 | 0.96 | | 0.40 | |
|  | |  | |  |  | rs267205 | | 7860317 | | A/G | 0.25 | | | 0.53 | 0.86 | | 0.06 | |
|  | |  | |  |  | rs270398 | | 7765840 | | C/A | 0.19 | | | 0.19 | 0.96 | | 0.96 | |
|  | |  | |  |  | rs10498671 | | 7797591 | | T/C | 0.18 | | | 0.17 | 0.97 | | 0.69 | |
|  | |  | |  |  | rs1225929 | | 7874233 | | A/T | 0.61 | | | 0.33 | 0.96 | | 0.09 | |
|  | |  | |  |  | rs11759532 | | 7791277 | | C/A | 0.40 | | | 0.51 | 1.00 | | 0.43 | |
|  | |  | |  |  | rs267806 | | 7819895 | | C/T | 0.28 | | | 0.58 | 0.97 | | 0.01 | |
|  | |  | |  |  | rs9505293 | | 7870593 | | G/T | 0.12 | | | 0.13 | 0.82 | | 0.74 | |
|  | |  | |  |  | rs270413 | | 7749643 | | T/C | 0.50 | | | 0.38 | 0.96 | | 0.77 | |
|  | |  | |  |  | rs13196371 | | 7748785 | | G/C | 0.16 | | | 0.21 | 0.96 | | 0.59 | |
|  | |  | |  |  | rs2326994 | | 7810021 | | A/G | 0.29 | | | 0.14 | 0.98 | | 0.07 | |
|  | |  | |  |  | rs3812163 | | 7725760 | | T/A | 0.46 | | | 0.26 | 0.98 | | 0.61 | |
|  | |  | |  |  | rs9505276 | | 7751449 | | A/G | 0.18 | | | 0.19 | 0.52 | | 0.68 | |
|  | |  | |  |  | rs11243204 | | 7772699 | | A/G | 0.23 | | | 0.23 | 0.81 | | 0.78 | |
|  | |  | |  |  | rs1107495 | | 7726057 | | A/G | 0.19 | | | 0.43 | 0.98 | | 0.0004 | |
|  | |  | |  |  | rs270417 | | 7729614 | | T/C | 0.28 | | | 0.18 | 0.96 | | 0.02 | |
|  | |  | |  |  | rs6910759 | | 7733862 | | A/G | 0.46 | | | 0.25 | 0.96 | | 0.71 | |
|  | |  | |  |  | rs199205 | | 7736417 | | C/G | 0.20 | | | 0.19 | 0.96 | | 0.58 | |
|  | |  | |  |  | rs2068361 | | 7736743 | | G/A | 0.25 | | | 0.36 | 0.97 | | 0.62 | |
|  | |  | |  |  | rs911749 | | 7738959 | | G/A | 0.23 | | | 0.33 | 1.00 | | 0.01 | |
|  | |  | |  |  | rs11964227 | | 7820669 | | G/A | 0.43 | | | 0.29 | 0.96 | | 0.73 | |
|  | |  | |  |  | rs12215656 | | 7829209 | | G/A | 0.15 | | | 0.05 | 0.93 | | 0.68 | |
| *BMP7* | | *OP-1* | | 20q13 | 0.58 | rs1475000 | | 55792997 | | A/G | 0.41 | | | 0.40 | 0.96 | | 0.48 | |
|  | |  | |  |  | rs6127978 | | 55819173 | | A/G | 0.14 | | | 0.28 | 0.62 | | 0.51 | |
|  | |  | |  |  | rs12438 | | 55743905 | | A/G | 0.51 | | | 0.41 | 0.96 | | 0.64 | |
|  | |  | |  |  | rs4811822 | | 55778969 | | T/C | 0.50 | | | 0.46 | 0.86 | | 0.26 | |
|  | |  | |  |  | rs162315 | | 55808880 | | G/A | 0.22 | | | 0.33 | 0.86 | | 0.78 | |
|  | |  | |  |  | rs6127983 | | 55824475 | | T/C | 0.34 | | | 0.48 | 0.96 | | 0.52 | |
|  | |  | |  |  | rs6123674 | | 55762947 | | A/G | 0.41 | | | 0.35 | 0.91 | | 0.38 | |
|  | |  | |  |  | rs17404303 | | 55803687 | | C/T | 0.43 | | | 0.29 | 0.96 | | 0.63 | |
|  | |  | |  |  | rs162317 | | 55812278 | | G/A | 0.41 | | | 0.40 | 0.96 | | 0.59 | |
|  | |  | |  |  | rs3787380 | | 55754110 | | T/C | 0.39 | | | 0.52 | 1.00 | | 0.29 | |
|  | |  | |  |  | rs17480735 | | 55745485 | | G/A | 0.08 | | | 0.04 | 0.96 | | 0.90 | |
|  | |  | |  |  | rs13037653 | | 55754239 | | T/C | 0.07 | | | 0.03 | 0.82 | | 0.92 | |
|  | |  | |  |  | rs6064508 | | 55760567 | | T/C | 0.36 | | | 0.30 | 0.98 | | 0.62 | |
|  | |  | |  |  | rs7273197 | | 55812058 | | C/T | 0.33 | | | 0.24 | 0.96 | | 0.89 | |
|  | |  | |  |  | rs6014949 | | 55752810 | | G/A | 0.51 | | | 0.42 | 0.96 | | 0.60 | |
|  | |  | |  |  | rs3787382 | | 55755170 | | C/T | 0.19 | | | 0.23 | 0.96 | | 0.83 | |
|  | |  | |  |  | rs6025446 | | 55786811 | | A/G | 0.41 | | | 0.51 | 0.96 | | 0.01 | |
|  | |  | |  |  | rs12481628 | | 55787622 | | A/G | 0.43 | | | 0.43 | 0.86 | | 0.57 | |
|  | |  | |  |  | rs172983 | | 55800039 | | G/A | 0.10 | | | 0.25 | 0.89 | | 0.82 | |
|  | |  | |  |  | rs2180780 | | 55800721 | | C/G | 0.44 | | | 0.42 | 0.96 | | 0.78 | |
|  | |  | |  |  | rs6127973 | | 55806280 | | G/A | 0.13 | | | 0.15 | 0.97 | | 0.98 | |
|  | |  | |  |  | rs6025468 | | 55830151 | | A/G | 0.20 | | | 0.20 | 0.86 | | 0.38 | |
|  | |  | |  |  | rs6070036 | | 55831445 | | G/T | 0.11 | | | 0.06 | 0.68 | | 0.71 | |
|  | |  | |  |  | rs6014967 | | 55836028 | | G/A | 0.21 | | | 0.31 | 0.96 | | 0.95 | |
| *BMPR1A* | | *ACVRLK3,* | | 10q22.3 | 0.66 | rs6586034 | | 88524209 | | T/G | 0.42 | | | 0.49 | 0.96 | | 0.71 | |
|  | | *ALK3,* | |  |  | rs7088641 | | 88532510 | | T/C | 0.30 | | | 0.37 | 0.97 | | 0.74 | |
|  | | *CD292* | |  |  | rs2168730 | | 88567036 | | A/G | 0.24 | | | 0.35 | 0.96 | | 0.57 | |
|  | |  | |  |  | rs12415784 | | 88572727 | | T/C | 0.16 | | | 0.28 | 1.00 | | 0.35 | |
|  | |  | |  |  | rs7895217 | | 88588739 | | T/A | 0.36 | | | 0.43 | 0.96 | | 0.50 | |
|  | |  | |  |  | rs2883420 | | 88611122 | | T/C | 0.38 | | | 0.45 | 0.96 | | 0.69 | |
|  | |  | |  |  | rs4934275 | | 88617392 | | T/C | 0.13 | | | 0.30 | 0.96 | | 0.92 | |
|  | |  | |  |  | rs10887668 | | 88662216 | | A/C | 0.11 | | | 0.08 | 0.86 | | 0.68 | |
|  | |  | |  |  | rs12765929 | | 88682555 | | G/T | 0.27 | | | 0.37 | 0.96 | | 0.46 | |
| *BMPR1B* | | *ALK-6,* | | 4q22-q24 | 0.44 | rs10049681 | | 95684380 | | T/C | 0.27 | | | 0.53 | 0.96 | | 0.71 | |
|  | | *ALK6,* | |  |  | rs12508087 | | 95692641 | | T/A | 0.24 | | | 0.19 | 0.96 | | 0.98 | |
|  | | *CDw293* | |  |  | rs9307147 | | 95705451 | | A/G | 0.45 | | | 0.29 | 0.89 | | 0.36 | |
|  | |  | |  |  | rs4490463 | | 95732345 | | A/G | 0.42 | | | 0.30 | 0.93 | | 0.12 | |
|  | |  | |  |  | rs2214395 | | 95754840 | | A/G | 0.16 | | | 0.11 | 0.96 | | 0.96 | |
|  | |  | |  |  | rs2719176 | | 95784645 | | C/G | 0.39 | | | 0.43 | 0.93 | | 0.83 | |
|  | |  | |  |  | rs17616243 | | 95792516 | | C/T | 0.15 | | | 0.28 | 0.93 | | 0.06 | |
|  | |  | |  |  | rs6849425 | | 95855547 | | C/T | 0.22 | | | 0.19 | 0.97 | | 0.72 | |
|  | |  | |  |  | rs7698964 | | 95856974 | | G/A | 0.12 | | | 0.07 | 0.97 | | 0.45 | |
|  | |  | |  |  | rs17022671 | | 95866073 | | G/A | 0.25 | | | 0.15 | 0.96 | | 0.57 | |
|  | |  | |  |  | rs7661049 | | 95883027 | | G/A | 0.53 | | | 0.39 | 0.96 | | 0.38 | |
|  | |  | |  |  | rs2120834 | | 95896344 | | G/C | 0.37 | | | 0.28 | 0.98 | | 0.19 | |
|  | |  | |  |  | rs4145993 | | 95916279 | | C/T | 0.20 | | | 0.15 | 0.96 | | 0.10 | |
|  | |  | |  |  | rs7694043 | | 95938768 | | C/T | 0.36 | | | 0.25 | 0.96 | | 0.89 | |
|  | |  | |  |  | rs13134042 | | 95959027 | | G/A | 0.21 | | | 0.25 | 0.93 | | 0.85 | |
|  | |  | |  |  | rs1863652 | | 95991417 | | C/T | 0.34 | | | 0.40 | 0.96 | | 0.59 | |
|  | |  | |  |  | rs3796442 | | 96054027 | | C/A | 0.09 | | | 0.18 | 0.66 | | 0.44 | |
|  | |  | |  |  | rs3821968 | | 96072245 | | C/T | 0.11 | | | 0.28 | 1.00 | | 0.10 | |
| *BMPR2* | | *BMPR-II,* | | 2q33-q34 | 0.47 | rs1980153 | | 203246340 | | A/T | 0.11 | | | 0.17 | 0.96 | | 0.56 | |
|  | | *BMPR3,* | |  |  | rs4303700 | | 203246411 | | G/A | 0.24 | | | 0.13 | 0.99 | | 0.53 | |
|  | | *BMR2,* | |  |  | rs4675278 | | 203334572 | | G/A | 0.27 | | | 0.34 | 0.98 | | 0.85 | |
|  | | *BRK-3,* | |  |  | rs12477602 | | 203338245 | | G/A | 0.13 | | | 0.15 | 0.96 | | 0.29 | |
|  | | *PPH1,* | |  |  | rs6751210 | | 203352937 | | A/G | 0.49 | | | 0.44 | 0.96 | | 0.81 | |
|  | | *T-ALK,* | |  |  | rs12621870 | | 203358214 | | T/C | 0.23 | | | 0.21 | 0.86 | | 0.72 | |
|  | | *TRG10* | |  |  | rs1199496 | | 203408736 | | A/T | 0.29 | | | 0.35 | 0.96 | | 0.67 | |
|  | |  | |  |  | rs17199235 | | 203419859 | | A/G | 0.12 | | | 0.06 | 0.73 | | 0.88 | |
| *GDF10* | | *BMP-3b,* | | 10q11.22 | 0.06 | rs7093975 | | 48424759 | | C/T | 0.27 | | | 0.23 | 0.62 | | 0.82 | |
|  | | *BMP3B* | |  |  | rs762454 | | 48427246 | | A/G | 0.32 | | | 0.23 | 0.96 | | 0.08 | |
|  | |  | |  |  | rs11598444 | | 48430867 | | G/A | 0.14 | | | 0.07 | 0.68 | | 0.74 | |
|  | |  | |  |  | rs2853838 | | 48431110 | | C/A | 0.20 | | | 0.21 | 0.93 | | 0.92 | |
|  | |  | |  |  | rs1902725 | | 48436242 | | G/A | 0.21 | | | 0.15 | 0.68 | | 0.81 | |
|  | |  | |  |  | rs1902724 | | 48438147 | | A/C | 0.32 | | | 0.31 | 0.78 | | 0.75 | |
| *MSTN* | | *GDF8* | | 2q32.2 | 0.76 | rs3791783 | | 190924163 | | T/C | 0.21 | | | 0.23 | 0.86 | | 0.94 | |
| *RUNX1* | | *AML1,* | | 21q22.3 | 0.002 | rs7279383 | | 36224963 | | C/G | 0.19 | | | 0.11 | 0.89 | | 0.74 | |
|  | | *AML1-EVI-1,* | |  |  | rs2268288 | | 36232671 | | T/C | 0.20 | | | 0.12 | 1.00 | | 0.84 | |
|  | | *AMLCR1,* | |  |  | rs2252585 | | 36241929 | | T/C | 0.27 | | | 0.43 | 0.96 | | 0.42 | |
|  | | *CBFA2,* | |  |  | rs11701453 | | 36338916 | | G/C | 0.20 | | | 0.15 | 0.96 | | 0.81 | |
|  | | *EVI-1,* | |  |  | rs8127225 | | 36364765 | | T/C | 0.13 | | | 0.27 | 0.96 | | 0.75 | |
|  | | *PEBP2aB* | |  |  | rs1474479 | | 36405666 | | G/A | 0.38 | | | 0.17 | 0.78 | | 0.40 | |
|  | |  | |  |  | rs1883066 | | 36412156 | | G/C | 0.12 | | | 0.07 | 0.96 | | 0.91 | |
|  | |  | |  |  | rs7279123 | | 36415087 | | C/T | 0.26 | | | 0.18 | 0.96 | | 0.69 | |
| *RUNX2* | | *RP1-166H4.1,* | | 6p21 | 0.20 | rs1321075 | | 45391943 | | C/A | 0.16 | | | 0.35 | 0.96 | | 0.85 | |
|  | | *AML3,* | |  |  | rs17209895 | | 45402445 | | T/C | 0.27 | | | 0.14 | 0.96 | | 0.08 | |
|  | | *CBFA1,* | |  |  | rs2677108 | | 45403774 | | T/C | 0.41 | | | 0.54 | 0.97 | | 0.72 | |
|  | | *CCD, CCD1,* | |  |  | rs2819854 | | 45404528 | | T/C | 0.51 | | | 0.47 | 0.96 | | 0.43 | |
|  | | *MGC120022,* | |  |  | rs2790093 | | 45437484 | | A/G | 0.33 | | | 0.31 | 0.86 | | 0.68 | |
|  | | *MGC120023,* | |  |  | rs9463090 | | 45453345 | | G/A | 0.21 | | | 0.18 | 0.86 | | 0.82 | |
|  | | *OSF2PEA2aA,* | |  |  | rs2396441 | | 45467765 | | C/T | 0.50 | | | 0.49 | 0.86 | | 0.11 | |
|  | | *PEBP2A1,* | |  |  | rs1316330 | | 45469626 | | G/T | 0.25 | | | 0.16 | 0.66 | | 0.72 | |
|  | | *PEBP2A2,* | |  |  | rs7750470 | | 45473256 | | T/C | 0.19 | | | 0.20 | 0.97 | | 0.84 | |
|  | | *PEBP2aA,* | |  |  | rs6930053 | | 45488758 | | C/T | 0.41 | | | 0.30 | 0.89 | | 0.99 | |
|  | | *PEBP2aA1* | |  |  | rs12208240 | | 45501937 | | G/A | 0.08 | | | 0.12 | 0.96 | | 0.95 | |
|  | |  | |  |  | rs12209785 | | 45506122 | | A/G | 0.25 | | | 0.28 | 0.59 | | 0.52 | |
|  | |  | |  |  | rs10948238 | | 45511541 | | C/T | 0.39 | | | 0.39 | 0.68 | | 0.32 | |
|  | |  | |  |  | rs13201287 | | 45511945 | | G/A | 0.25 | | | 0.30 | 0.62 | | 0.64 | |
|  | |  | |  |  | rs12333172 | | 45512215 | | C/T | 0.20 | | | 0.16 | 1.00 | | 0.52 | |
|  | |  | |  |  | rs1200428 | | 45518202 | | C/A | 0.22 | | | 0.28 | 0.62 | | 0.95 | |
|  | |  | |  |  | rs598953 | | 45520030 | | T/A | 0.37 | | | 0.43 | 0.93 | | 0.71 | |
| *RUNX3* | | *RP3-398I9.1,* | | 1p36 | 0.56 | rs2236850 | | 25240341 | | T/C | 0.44 | | | 0.40 | 0.96 | | 0.59 | |
|  | | *AML2,* | |  |  | rs9438876 | | 25241116 | | A/G | 0.54 | | | 0.42 | 0.62 | | 0.80 | |
|  | | *CBFA3,* | |  |  | rs7517302 | | 25254317 | | T/C | 0.43 | | | 0.37 | 0.96 | | 0.88 | |
|  | | *FLJ34510,* | |  |  | rs906296 | | 25264658 | | C/G | 0.23 | | | 0.18 | 0.96 | | 0.39 | |
|  | | *MGC16070,* | |  |  | rs7551188 | | 25273200 | | C/T | 0.54 | | | 0.47 | 0.98 | | 0.21 | |
|  | | *PEBP2aC* | |  |  | rs6688058 | | 25274998 | | G/A | 0.13 | | | 0.14 | 0.96 | | 0.61 | |
|  | |  | |  |  | rs11249206 | | 25277982 | | T/C | 0.51 | | | 0.35 | 0.96 | | 0.81 | |
|  | |  | |  |  | rs4478762 | | 25281015 | | G/A | 0.11 | | | 0.12 | 0.62 | | 0.82 | |
| *SMAD1* | | *BSP1,* | | 4q31 | 0.18 | rs6537355 | | 146402592 | | A/G | 0.12 | | | 0.10 | 0.96 | | 0.97 | |
|  | | *JV4-1, JV41,* | |  |  | rs2118438 | | 146428384 | | G/A | 0.20 | | | 0.39 | 0.96 | | 0.05 | |
|  | | *MADH1,* | |  |  | rs714195 | | 146445680 | | A/G | 0.56 | | | 0.38 | 0.97 | | 0.30 | |
|  | | *MADR1* | |  |  | rs12505085 | | 146481705 | | A/G | 0.25 | | | 0.21 | 0.62 | | 0.85 | |
| *SMAD2* | | *JV18, JV18-1,* | | 18q21.1 | 0.16 | rs1792689 | | 45368587 | | C/T | 0.12 | | | 0.07 | 1.00 | | 0.62 | |
|  | | *MADH2, MADR2,* | |  |  | rs1792658 | | 45382605 | | A/C | 0.20 | | | 0.42 | 0.98 | | 0.17 | |
|  | | *MGC22139,* | |  |  | rs1787199 | | 45404603 | | A/T | 0.47 | | | 0.37 | 0.93 | | 0.74 | |
|  | | *MGC34440,* | |  |  | rs17814648 | | 45413714 | | C/T | 0.05 | | | 0.05 | 0.97 | | 0.68 | |
|  | | *hMAD-2, hSMAD2* | |  |  | rs4940086 | | 45446307 | | T/C | 0.34 | | | 0.30 | 0.96 | | 0.92 | |
| *SMAD3* | | *DKFZP586N0721,* | | 15q22.33 | 0.82 | rs12904944 | | 67361774 | | G/A | 0.34 | | | 0.19 | 0.96 | | 0.26 | |
|  | | *DKFZp686J10186,* | |  |  | rs1498506 | | 67367634 | | A/C | 0.46 | | | 0.33 | 0.96 | | 0.88 | |
|  | | *HSPC193,* | |  |  | rs12901071 | | 67370389 | | A/G | 0.34 | | | 0.15 | 0.86 | | 0.68 | |
|  | | *HsT17436,* | |  |  | rs4776881 | | 67376000 | | T/C | 0.45 | | | 0.35 | 0.89 | | 0.16 | |
|  | | *JV15-2,* | |  |  | rs12907997 | | 67380609 | | C/T | 0.50 | | | 0.36 | 0.93 | | 0.47 | |
|  | | *MADH3,* | |  |  | rs7176870 | | 67388553 | | A/G | 0.43 | | | 0.30 | 0.96 | | 0.53 | |
|  | | *MGC60396* | |  |  | rs9972423 | | 67391805 | | T/A | 0.37 | | | 0.29 | 0.89 | | 0.73 | |
|  | |  | |  |  | rs4776890 | | 67393045 | | T/G | 0.41 | | | 0.23 | 0.72 | | 0.88 | |
|  | |  | |  |  | rs2118611 | | 67401466 | | A/G | 0.20 | | | 0.46 | 0.99 | | 0.03 | |
|  | |  | |  |  | rs11071933 | | 67401746 | | C/G | 0.32 | | | 0.57 | 0.96 | | 0.76 | |
|  | |  | |  |  | rs11637581 | | 67412049 | | C/T | 0.28 | | | 0.12 | 0.96 | | 0.60 | |
|  | |  | |  |  | rs7163381 | | 67414055 | | G/A | 0.26 | | | 0.44 | 0.96 | | 0.73 | |
|  | |  | |  |  | rs4776892 | | 67414237 | | A/T | 0.18 | | | 0.37 | 0.96 | | 0.98 | |
|  | |  | |  |  | rs1992215 | | 67418937 | | T/C | 0.33 | | | 0.34 | 0.85 | | 0.95 | |
|  | |  | |  |  | rs991157 | | 67419013 | | G/A | 0.31 | | | 0.15 | 0.89 | | 0.68 | |
|  | |  | |  |  | rs121021712 | | 67425033 | | C/T | 0.16 | | | 0.23 | 0.04 | | 2.6E-13 | |
|  | |  | |  |  | rs4147358 | | 67428014 | | C/A | 0.22 | | | 0.43 | 0.96 | | 0.61 | |
|  | |  | |  |  | rs2118610 | | 67428334 | | G/A | 0.46 | | | 0.23 | 0.96 | | 0.41 | |
|  | |  | |  |  | rs2414937 | | 67432676 | | G/C | 0.19 | | | 0.26 | 1.00 | | 0.74 | |
|  | |  | |  |  | rs12443188 | | 67433941 | | T/A | 0.23 | | | 0.28 | 0.62 | | 0.95 | |
|  | |  | |  |  | rs12915039 | | 67434348 | | A/C | 0.23 | | | 0.28 | 0.54 | | 0.86 | |
|  | |  | |  |  | rs745103 | | 67435075 | | C/T | 0.55 | | | 0.42 | 0.96 | | 0.42 | |
|  | |  | |  |  | rs12439792 | | 67437385 | | T/A | 0.09 | | | 0.37 | 0.97 | | 0.66 | |
|  | |  | |  |  | rs17293443 | | 67437863 | | T/C | 0.23 | | | 0.12 | 0.96 | | 0.70 | |
|  | |  | |  |  | rs893473 | | 67438091 | | C/T | 0.19 | | | 0.45 | 0.97 | | 0.94 | |
|  | |  | |  |  | rs750766 | | 67438586 | | G/A | 0.47 | | | 0.40 | 0.78 | | 1.00 | |
|  | |  | |  |  | rs2289263 | | 67439207 | | A/C | 0.47 | | | 0.39 | 0.82 | | 0.94 | |
|  | |  | |  |  | rs1866317 | | 67441764 | | C/G | 0.12 | | | 0.06 | 0.96 | | 0.83 | |
|  | |  | |  |  | rs731874 | | 67446831 | | G/A | 0.29 | | | 0.13 | 0.62 | | 0.50 | |
|  | |  | |  |  | rs4601989 | | 67451954 | | C/T | 0.22 | | | 0.30 | 0.62 | | 0.93 | |
|  | |  | |  |  | rs11639295 | | 67460757 | | C/T | 0.31 | | | 0.44 | 0.97 | | 0.59 | |
|  | |  | |  |  | rs7183244 | | 67461311 | | C/T | 0.40 | | | 0.18 | 0.74 | | 0.62 | |
|  | |  | |  |  | rs16950687 | | 67464013 | | A/G | 0.28 | | | 0.37 | 0.93 | | 0.25 | |
|  | |  | |  |  | rs12708492 | | 67467541 | | T/C | 0.52 | | | 0.43 | 1.00 | | 0.76 | |
|  | |  | |  |  | rs1470003 | | 67469335 | | G/C | 0.47 | | | 0.24 | 0.96 | | 0.18 | |
|  | |  | |  |  | rs7181556 | | 67476768 | | C/T | 0.25 | | | 0.36 | 0.89 | | 0.06 | |
|  | |  | |  |  | rs7173811 | | 67479051 | | T/C | 0.52 | | | 0.27 | 0.86 | | 0.10 | |
|  | |  | |  |  | rs3825977 | | 67481248 | | C/T | 0.20 | | | 0.33 | 0.96 | | 0.02 | |
|  | |  | |  |  | rs11629568 | | 67482368 | | T/G | 0.45 | | | 0.22 | 0.49 | | 0.04 | |
|  | |  | |  |  | rs3743343 | | 67486775 | | T/C | 0.24 | | | 0.36 | 0.96 | | 0.43 | |
|  | |  | |  |  | rs1052488 | | 67486847 | | T/C | 0.23 | | | 0.36 | 0.96 | | 0.46 | |
| *SMAD4* | | *DPC4, JIP,* | | 18q21.1 | 0.55 | rs10502913 | | 48568271 | | G/A | 0.23 | | | 0.29 | 1.00 | | 0.77 | |
|  | | *MADH4* | |  |  | rs8096092 | | 48581601 | | C/A | 0.38 | | | 0.40 | 0.97 | | 0.76 | |
| *SMAD6* | | *HsT17432,* | | 15q21-q22 | 0.51 | rs2439385 | | 67026861 | | C/A | 0.30 | | | 0.16 | 0.97 | | 0.70 | |
|  | | *MADH6, MADH7* | |  |  |  | |  | |  |  | | |  |  | |  | |
| *SMAD7* | | *FLJ16482,* | | 18q21.1 | 0.76 | rs12953717 | | 46453929 | | C/T | 0.43 | | | 0.28 | 0.82 | | 0.24 | |
|  | | *MADH7,* | |  |  | rs12456328 | | 46457096 | | C/T | 0.13 | | | 0.06 | 0.96 | | 0.24 | |
|  | | *MADH8* | |  |  | rs4464148 | | 46459032 | | T/C | 0.31 | | | 0.19 | 0.86 | | 0.50 | |
|  | |  | |  |  | rs2337107 | | 46459323 | | G/A | 0.40 | | | 0.40 | 0.96 | | 0.80 | |
|  | |  | |  |  | rs2337106 | | 46460903 | | G/C | 0.52 | | | 0.48 | 0.93 | | 0.94 | |
|  | |  | |  |  | rs7238442 | | 46461786 | | T/C | 0.45 | | | 0.45 | 0.54 | | 0.89 | |
|  | |  | |  |  | rs4939832 | | 46465665 | | A/G | 0.24 | | | 0.29 | 0.98 | | 0.83 | |
|  | |  | |  |  | rs3764482 | | 46468946 | | C/T | 0.20 | | | 0.10 | 0.96 | | 0.50 | |
|  | |  | |  |  | rs1316447 | | 46472676 | | C/T | 0.19 | | | 0.12 | 0.94 | | 0.84 | |
|  | |  | |  |  | rs3736242 | | 46474746 | | G/A | 0.24 | | | 0.25 | 0.97 | | 0.99 | |
| *TGFB1* | | *CED,* | | 19q13.1 | 0.08 | rs1800469 | | 41860296 | | C/T | 0.32 | | | 0.46 | 0.62 | | 0.82 | |
|  | | *DPD1, LAP,* | |  |  | rs4803455 | | 41851509 | | C/A | 0.49 | | | 0.35 | 0.96 | | 0.63 | |
|  | | *TGFB, TGFbeta* | |  |  |  | |  | |  |  | | |  |  | |  | |
| *TGFB2* | | *MGC116892,* | | 1q41 | 0.66 | rs6604609 | | 218568482 | | T/A | 0.06 | | | 0.12 | 0.96 | | 0.96 | |
|  | | *TGF-beta2* | |  |  |  | |  | |  |  | | |  |  | |  | |
| *TGFBR1* | | *AAT5,* | | 9q22 | 0.21 | rs6478974 | | 101874403 | | T/A | 0.47 | | | 0.35 | 0.89 | | 0.94 | |
|  | | *ACVRLK4,* | |  |  | rs1571590 | | 101883808 | | A/G | 0.20 | | | 0.10 | 0.97 | | 0.47 | |
|  | | *ALK-5, ALK5,* | |  |  | rs1013186 | | 101884337 | | G/A | 0.20 | | | 0.11 | 0.97 | | 0.60 | |
|  | | *LDS1A, LDS2A,* | |  |  | rs11568785 | | 101905834 | | A/G | 0.08 | | | 0.04 | 0.96 | | 0.67 | |
|  | | *SKR4, TGFR-1* | |  |  | rs10733710 | | 101907424 | | G/A | 0.23 | | | 0.35 | 0.97 | | 0.12 | |
| *TGFBR2* | | *AAT3, FAA3,* | | 3p22 | 0.14 | rs3773644 | | 30712344 | | C/T | 0.40 | | | 0.27 | 0.96 | | 0.35 | |
|  | | *LDS1B, LDS2B,* | |  |  |  | |  | |  |  | | |  |  | |  | |
|  | | MFS2, RIIC, | | |  |  | |  | |  |  | | |  |  | |  | |
|  | | *TAAD2,* |  | |  |  | |  | |  |  | | |  |  | |  | |
|  | | *TGFR-2, TGFbeta-RII* |  | |  |  | |  | |  |  | | |  |  | |  | |
| *TGFBR3* | | *BGCAN,* | 1p33-p32 | | 0.04 | rs4650272 | | 92168649 | | T/A | 0.18 | | | 0.24 | 0.89 | | 0.49 | |
|  | | *betaglycan* |  | |  | rs284185 | | 92233820 | | A/T | 0.41 | | | 0.49 | 0.96 | | 0.66 | |
|  | |  |  | |  | rs284200 | | 92239336 | | C/T | 0.38 | | | 0.42 | 0.96 | | 0.62 | |
|  |  | |  | |  | rs6678564 | | 92257650 | | G/C | 0.09 | | | 0.08 | 0.68 | | 0.71 | |
|  |  | |  | |  | rs10493856 | | 92334749 | | A/G | 0.39 | | | 0.47 | 0.96 | | 0.78 | |

1Minor Allele Frequency (MAF) and FDR-adjusted Hardy-Weinberg Equilibrium (FDR HWE) based on control population.

2TagSNP is out of HWE (HWE p<0.003 among 0-28% and 29-100% NA ancestry groups) and is excluded from analysis.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Supplement Table 2. Breast cancer survival by ER/PR tumor status at time of diagnosis | | | | | | | | | | | | | | |
|  |  | ER+/PR+ | | | | ER+/PR- | | | | ER-/PR- | | | |  |
|  |  | Death/ Person Years | HR1 | (95% CI) | | Death/ Person Years | HR | (95% CI) | | Death/ Person Years | HR | (95% CI) | | Interaction P2 |
| **P ARTP:** | | 0.34 |  |  |  | 0.16 |  |  |  | 0.58 |  |  |  |  |
| ***BMP6*** | |  |  |  |  |  |  |  |  |  |  |  |  |  |
| (rs3812163) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.03 |
|  | TT | 36 / 4476 | 1.00 |  |  | 3 / 729 | 1.00 |  |  | 30 / 1302 | 1.00 |  |  |  |
|  | TA | 41 / 4865 | 1.15 | (0.73, | 1.82) | 15 / 888 | 5.60 | (1.45, | 21.63) | 24 / 1528 | 0.76 | (0.43, | 1.32) |  |
|  | AA | 15 / 1604 | 1.63 | (0.86, | 3.09) | 5 / 309 | 5.67 | (1.17, | 27.49) | 7 / 395 | 0.75 | (0.31, | 1.79) |  |
|  | P-value |  | 0.14 |  |  |  | 0.03 |  |  |  | 0.51 |  |  |  |
| (rs1107495) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.02 |
|  | AA | 57 / 6082 | 1.00 |  |  | 19 / 1137 | 1.00 |  |  | 26 / 1670 | 1.00 |  |  |  |
|  | AG/GG | 35 / 4881 | 0.75 | (0.48, | 1.17) | 4 / 789 | 0.29 | (0.08, | 0.99) | 35 / 1532 | 1.46 | (0.86, | 2.49) |  |
|  | P-value |  | 0.20 |  |  |  | 0.05 |  |  |  | 0.16 |  |  |  |
| (rs6910759) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.24 |
|  | AA | 40 / 4111 | 1.00 |  |  | 4 / 698 | 1.00 |  |  | 20 / 1305 | 1.00 |  |  |  |
|  | AG | 37 / 5131 | 0.80 | (0.50, | 1.29) | 12 / 873 | 2.88 | (0.80, | 10.38) | 33 / 1494 | 1.61 | (0.90, | 2.88) |  |
|  | GG | 15 / 1721 | 1.21 | (0.64, | 2.29) | 7 / 354 | 4.15 | (1.02, | 16.86) | 8 / 427 | 1.18 | (0.50, | 2.76) |  |
|  | P-value |  | 0.55 |  |  |  | 0.05 |  |  |  | 0.71 |  |  |  |
| ***RUNX2*** | | 0.06 |  |  |  | 0.07 |  |  |  | 0.30 |  |  |  |  |
| (rs1321075) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.03 |
|  | CC | 58 / 6324 | 1.00 |  |  | 9 / 1142 | 1.00 |  |  | 29 / 1954 | 1.00 |  |  |  |
|  | CA | 27 / 3931 | 0.70 | (0.44, | 1.13) | 11 / 676 | 3.06 | (1.15, | 8.17) | 29 / 960 | 1.94 | (1.11, | 3.38) |  |
|  | AA | 7 / 695 | 1.04 | (0.46, | 2.36) | 3 / 107 | 7.62 | (1.31, | 44.25) | 3 / 311 | 0.72 | (0.21, | 2.49) |  |
|  | P-value |  | 0.92 |  |  |  | 0.02 |  |  |  | 0.61 |  |  |  |
| (rs2819854) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.04 |
|  | TT | 20 / 2656 | 1.00 |  |  | 5 / 402 | 1.00 |  |  | 23 / 868 | 1.00 |  |  |  |
|  | TC | 47 / 5618 | 1.19 | (0.70, | 2.03) | 13 / 1009 | 0.47 | (0.14, | 1.57) | 28 / 1502 | 0.85 | (0.48, | 1.50) |  |
|  | CC | 25 / 2679 | 1.48 | (0.82, | 2.69) | 5 / 514 | 0.39 | (0.10, | 1.49) | 10 / 856 | 0.49 | (0.23, | 1.04) |  |
|  | P-value |  | 0.20 |  |  |  | 0.17 |  |  |  | 0.06 |  |  |  |
| (rs2396441) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.02 |
|  | CC | 15 / 2782 | 1.00 |  |  | 8 / 504 | 1.00 |  |  | 16 / 883 | 1.00 |  |  |  |
|  | CT | 46 / 5296 | 1.62 | (0.90, | 2.92) | 11 / 961 | 0.65 | (0.23, | 1.82) | 39 / 1583 | 1.05 | (0.58, | 1.92) |  |
|  | TT | 31 / 2885 | 2.10 | (1.13, | 3.93) | 4 / 460 | 0.53 | (0.15, | 1.87) | 6 / 760 | 0.38 | (0.15, | 0.99) |  |
|  | P-value |  | 0.02 |  |  |  | 0.33 |  |  |  | 0.05 |  |  |  |
| (rs6930053) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.04 |
|  | CC | 24 / 4311 | 1.00 |  |  | 8 / 698 | 1.00 |  |  | 30 / 1431 | 1.00 |  |  |  |
|  | CT | 51 / 5097 | 1.76 | (1.08, | 2.88) | 13 / 983 | 0.99 | (0.38, | 2.58) | 26 / 1425 | 0.77 | (0.45, | 1.31) |  |
|  | TT | 17 / 1546 | 2.11 | (1.12, | 3.98) | 2 / 244 | 0.59 | (0.12, | 2.93) | 5 / 370 | 0.61 | (0.23, | 1.60) |  |
|  | P-value |  | 0.02 |  |  |  | 0.52 |  |  |  | 0.32 |  |  |  |
| (rs10948238) | | |  |  |  |  |  |  |  |  |  |  |  | 0.04 |
|  | CC | 39 / 3961 | 1.00 |  |  | 4 / 656 | 1.00 |  |  | 12 / 1083 | 1.00 |  |  |  |
|  | CT | 43 / 5186 | 0.72 | (0.46, | 1.12) | 14 / 992 | 1.83 | (0.58, | 5.76) | 39 / 1492 | 2.38 | (1.23, | 4.60) |  |
|  | TT | 10 / 1802 | 0.46 | (0.23, | 0.93) | 4 / 272 | 2.03 | (0.44, | 9.41) | 10 / 650 | 1.47 | (0.63, | 3.45) |  |
|  | P-value |  | 0.03 |  |  |  | 0.36 |  |  |  | 0.37 |  |  |  |
| (rs598953) | |  |  |  |  |  |  |  |  |  |  |  |  | 0.24 |
|  | TT | 44 / 4088 | 1.00 |  |  | 8 / 755 | 1.00 |  |  | 21 / 1200 | 1.00 |  |  |  |
|  | TA | 40 / 5274 | 0.59 | (0.38, | 0.92) | 11 / 930 | 1.04 | (0.40, | 2.68) | 36 / 1581 | 1.30 | (0.75, | 2.26) |  |
|  | AA | 8 / 1601 | 0.41 | (0.19, | 0.88) | 4 / 241 | 1.08 | (0.27, | 4.29) | 4 / 445 | 0.54 | (0.18, | 1.62) |  |
|  | P-value |  | 0.02 |  |  |  | 0.91 |  |  |  | 0.27 |  |  |  |
| 1Hazard Ratios (HR) and 95% Confidence Intervals (CI) among primary invasive cases adjusted for age, study center, BMI during referent year, parity, genetic ancestry, and SEER summary stage. | | | | | | | | | | | | | | |
| 2P value based on 2-df Wald test treating ER/PR status as categorical. None of the interaction p values remained significant after adjustment for multiple comparisons | | | | | | | | | | | | |  |  |