## Supplementary Tables

**Supplementary Table 1.** Genotype specific ORs among women of European ancestry, and evidence for heterogeneity in the per-allele OR among studies, in the BCAC replication.

| SNP | Chromosome Position | Het OR (95%CI) | HomOR (95%CI) | Heterogeneity P |
|---|---|---|---|---|
| rs10771399 | 12p11 28046347[1] | 0.85 (0.82-0.88) | 0.72 (0.64-0.82) | 0.09 |
| | | | | |
| rs1292011 | 12q24 114320905 | 0.93 (0.90-0.96) | 0.85 (0.82-0.89) | 0.10 |
| | | | | |
| rs2823093 | 21q21 15442703 | 0.94 (0.91-0.97) | 0.87 (0.83-0.92) | 0.002 |
| | | | | |

[1] Build 36

**Supplementary Table 2.**
Summary results for all SNPs typed in the BCAC replication stage.

| SNP | Chr. | Position[1] | Alleles | AF[2] | UK2 | | | BBCS | | | Other GWAS | | | Combined *P* |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | Per-allele OR | (95%CI) | *P* | Per-allele OR | (95%CI) | *P* | Per-allele OR | (95%CI) | *P* | |
| rs3738863 | 2 | 30311065 | GA | 0.732 | 1.17 | (1.09-1.25) | $1.6x10^{-5}$ | 1.08 | (0.99-1.18) | 0.089 | 1.03 | (0.96-1.10) | 0.39 | 0.00059 |
| rs6734368 | 2 | 58422570 | GA | 0.81 | 1.14 | (1.06-1.24) | 0.00087 | 1.06 | (0.96-1.17) | 0.27 | 0.93 | (0.87-1.00) | 0.044 | 0.47 |
| rs1028246/ rs1403400[$] | 2 | 114319652 | TC | 0.915 | 1.22 | (1.09-1.36) | 0.00044 | 1.14 | (0.98-1.32) | 0.090 | 1.02 | (0.92-1.14) | 0.64 | 0.0047 |
| rs13400898 | 2 | 154583859 | TC | 0.062 | 0.82 | (0.72-0.94) | 0.0033 | 0.97 | (0.83-1.14) | 0.71 | 1.12 | (0.99-1.27) | 0.081 | 0.50 |
| rs10469689 | 2 | 163968309 | TC | 0.058 | 0.81 | (0.71-0.93) | 0.0024 | 0.99 | (0.84-1.16) | 0.87 | 0.93 | (0.83-1.03) | 0.17 | 0.0083 |
| rs11711782 | 3 | 149160340 | GA | 0.119 | 0.99 | (0.90-1.09) | 0.81 | 0.90 | (0.80-1.03) | 0.12 | 0.96 | (0.89-1.05) | 0.38 | 0.23 |
| rs9884706 | 4 | 32275985 | TG | 0.844 | 1.16 | (1.07-1.27) | 0.00048 | 1.13 | (1.01-1.26) | 0.031 | 1.03 | (0.96-1.11) | 0.40 | 0.0019 |
| rs2166278 | 4 | 32286120 | TC | 0.844 | 1.16 | (1.07-1.26) | 0.00058 | 1.13 | (1.01-1.26) | 0.033 | 1.03 | (0.96-1.11) | 0.43 | 0.0023 |
| rs4403040 | 4 | 87257505 | TC | 0.357 | 1.11 | (1.05-1.19) | 0.00088 | 1.05 | (0.97-1.15) | 0.23 | 0.97 | (0.91-1.03) | 0.29 | 0.14 |
| rs9761051 | 4 | 120233038 | CA | 0.147 | 1.16 | (1.07-1.27) | 0.00056 | 1.12 | (1.01-1.26) | 0.041 | 0.97 | (0.90-1.05) | 0.50 | 0.042 |
| rs10940235 | 5 | 49672510 | TG | 0.541 | 1.04 | (0.97-1.10) | 0.26 | 1.12 | (1.03-1.21) | 0.0060 | 1.05 | (0.99-1.11) | 0.080 | 0.0060 |
| rs820848 | 5 | 74000416 | GA | 0.711 | 1.13 | (1.06-1.21) | 0.00034 | 1.04 | (0.93-1.16) | 0.50 | 1.00 | (0.93-1.08) | 0.92 | 0.022 |
| rs187727 | 5 | 75439941 | GA | 0.277 | 1.12 | (1.05-1.20) | 0.00084 | 1.09 | (1.00-1.19) | 0.059 | 0.99 | (0.93-1.06) | 0.86 | 0.029 |
| rs4704018 | 5 | 84299468 | TC | 0.624 | 1.12 | (1.05-1.19) | 0.00041 | 1.08 | (0.99-1.17) | 0.075 | 0.98 | (0.93-1.04) | 0.49 | 0.044 |
| rs11241138 | 5 | 111169939 | CA | 0.29 | 0.90 | (0.84-0.96) | 0.0013 | 0.90 | (0.82-0.99) | 0.029 | 0.99 | (0.93-1.05) | 0.65 | 0.0095 |
| rs13160298 | 5 | 157728895 | GA | 0.642 | 0.98 | (0.92-1.04) | 0.52 | 1.06 | (0.97-1.15) | 0.19 | 1.06 | (1.00-1.13) | 0.037 | 0.15 |
| rs12203592 | 6 | 341321 | TC | 0.772 | 0.95 | (0.88-1.03) | 0.20 | 0.91 | (0.74-1.11) | 0.35 | 1.02 | (0.92-1.14) | 0.66 | 0.39 |
| rs9467504 | 6 | 25522304 | GA | 0.837 | 0.84 | (0.77-0.91) | $2.1x10^{-5}$ | 0.91 | (0.81-1.02) | 0.12 | 1.00 | (0.93-1.08) | 0.95 | 0.0029 |
| rs12333016 | 6 | 107205884 | GA | 0.9 | 0.83 | (0.75-0.92) | 0.00037 | 1.01 | (0.88-1.15) | 0.93 | 1.08 | (0.98-1.18) | 0.10 | 0.45 |
| rs9401003 | 6 | 117824996 | GA | 0.781 | 0.89 | (0.83-0.96) | 0.0026 | 1.00 | (0.90-1.10) | 0.94 | 0.91 | (0.85-0.98) | 0.011 | 0.00065 |
| rs2189630 | 7 | 8695731 | TG | 0.655 | 0.89 | (0.84-0.95) | 0.00050 | 0.92 | (0.85-1.00) | 0.044 | 1.04 | (0.98-1.11) | 0.22 | 0.085 |
| rs7810604 | 7 | 11387506 | TC | 0.371 | 0.89 | (0.84-0.95) | 0.00038 | 0.87 | (0.80-0.94) | 0.00064 | 0.98 | (0.93-1.04) | 0.60 | 0.00081 |
| rs1981576 | 7 | 82057361 | TC | 0.087 | 0.81 | (0.72-0.90) | 0.00015 | 0.91 | (0.77-1.07) | 0.25 | 1.05 | (0.95-1.16) | 0.34 | 0.10 |
| rs1011692 | 7 | 121347247 | TC | 0.123 | 0.85 | (0.78-0.94) | 0.00076 | 0.95 | (0.84-1.06) | 0.35 | 1.07 | (0.99-1.16) | 0.084 | 0.46 |
| rs2617076 | 8 | 4432582 | CA | 0.4 | 1.11 | (1.04-1.18) | 0.0016 | 0.99 | (0.91-1.07) | 0.78 | 0.99 | (0.94-1.05) | 0.83 | 0.14 |
| rs4872360 | 8 | 22084613 | TC | 0.259 | 0.84 | (0.78-0.90) | $2.1x10^{-6}$ | 0.91 | (0.83-1.00) | 0.054 | 0.99 | (0.93-1.06) | 0.86 | 0.00099 |
| rs2211914 | 8 | 102738843 | GA | 0.612 | 0.99 | (0.93-1.05) | 0.73 | 0.93 | (0.86-1.01) | 0.084 | 0.93 | (0.88-0.99) | 0.013 | 0.012 |
| rs4268201[+] | 9 | 36878148 | TG | 0.862 | 0.86 | (0.78-0.93) | 0.00040 | 0.96 | (0.86-1.08) | 0.50 | 0.99 | (0.91-1.07) | 0.72 | 0.020 |
| rs1952461 | 9 | 78412404 | GA | 0.047 | 1.18 | (0.96-1.45) | 0.12 | 1.02 | (0.72-1.45) | 0.90 | 0.91 | (0.76-1.08) | 0.27 | 0.92 |
| rs1932658 | 9 | 86005570 | GA | 0.727 | 0.87 | (0.81-0.93) | $8.3x10^{-5}$ | 0.96 | (0.88-1.05) | 0.42 | 0.97 | (0.91-1.03) | 0.36 | 0.0029 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| rs12350727 | 9 | 93304437 | GA | 0.942 | 0.80 | (0.70-0.91) | 0.00065 | 1.05 | (0.88-1.26) | 0.60 | 0.96 | (0.86-1.08) | 0.55 | 0.039 |
| rs4455975 | 9 | 128423020 | GA | 0.449 | 1.07 | (1.00-1.13) | 0.044 | 1.08 | (1.00-1.17) | 0.061 | 1.08 | (1.02-1.14) | 0.0091 | 0.00056 |
| rs7074055 | 10 | 12665442 | GA | 0.769 | 0.90 | (0.83-0.96) | 0.0028 | 1.02 | (0.91-1.13) | 0.75 | 1.00 | (0.93-1.07) | 0.89 | 0.098 |
| rs4746065 | 10 | 72183963 | CA | 0.227 | 0.88 | (0.82-0.95) | 0.00074 | 0.91 | (0.82-1.00) | 0.039 | 1.06 | (0.99-1.13) | 0.083 | 0.20 |
| rs7116850 | 11 | 27602106 | GA | 0.731 | 1.15 | (1.07-1.23) | $8.4x10^{-5}$ | 1.10 | (1.00-1.20) | 0.042 | 0.98 | (0.92-1.04) | 0.47 | 0.028 |
| rs11604821 | 11 | 69061318 | GA | 0.665 | 0.87 | (0.82-0.93) | $4.6x10^{-5}$ | 0.92 | (0.85-1.00) | 0.054 | 1.02 | (0.96-1.08) | 0.47 | 0.022 |
| rs2284424 | 12 | 13880137 | GA | 0.296 | 1.12 | (1.05-1.20) | 0.00083 | 1.03 | (0.95-1.12) | 0.50 | 1.01 | (0.95-1.07) | 0.84 | 0.034 |
| rs10771399 | 12 | 28046347 | GA | 0.893 | 1.27 | (1.15-1.41) | $3.1x10^{-6}$ | 1.19 | (1.05-1.36) | 0.0079 | 1.21 | (1.10-1.33) | $5.7x10^{-5}$ | $5.7x10^{-10}$ |
| rs1975930 | 12 | 28053015 | TC | 0.107 | 0.79 | (0.71-0.87) | $2.9x10^{-6}$ | 0.84 | (0.74-0.96) | 0.0081 | 0.83 | (0.75-0.91) | $5.4x10^{-5}$ | $5.3x10^{-10}$ |
| rs1292011 | 12 | 114320905 | GA | 0.586 | 1.14 | (1.07-1.21) | $5.8x10^{-5}$ | 1.05 | (0.97-1.14) | 0.23 | 1.10 | (1.04-1.17) | 0.00083 | $9.5x10^{-7}$ |
| rs7955262 | 12 | 114691345 | TC | 0.103 | 1.22 | (1.10-1.35) | 0.00012 | 1.06 | (0.92-1.22) | 0.43 | 1.02 | (0.93-1.12) | 0.62 | 0.0054 |
| rs9586525 | 13 | 103922680 | TC | 0.185 | 1.16 | (1.07-1.25) | 0.00026 | 1.03 | (0.92-1.14) | 0.63 | 1.01 | (0.94-1.08) | 0.86 | 0.021 |
| rs3784194 | 14 | 31993377 | TC | 0.827 | 0.94 | (0.87-1.02) | 0.12 | 0.95 | (0.85-1.05) | 0.30 | 0.96 | (0.90-1.04) | 0.33 | 0.066 |
| rs1263441 | 14 | 82695911 | GA | 0.715 | 1.14 | (1.06-1.22) | 0.00023 | 1.03 | (0.95-1.13) | 0.45 | 0.99 | (0.93-1.05) | 0.64 | 0.067 |
| rs10484150 | 14 | 82707696 | TC | 0.715 | 1.14 | (1.06-1.22) | 0.00018 | 1.04 | (0.95-1.13) | 0.43 | 0.98 | (0.93-1.05) | 0.61 | 0.063 |
| rs2277509 | 14 | 90819348 | CA | 0.697 | 1.13 | (1.06-1.21) | 0.00035 | 1.00 | (0.92-1.09) | 0.98 | 0.99 | (0.93-1.05) | 0.78 | 0.085 |
| rs3101649 | 15 | 25607717 | GA | 0.062 | 1.22 | (1.08-1.38) | 0.0020 | 1.08 | (0.85-1.36) | 0.54 | 1.05 | (0.93-1.18) | 0.45 | 0.010 |
| rs12443310 | 15 | 52387367 | TC | 0.911 | 0.82 | (0.74-0.91) | 0.00024 | 0.88 | (0.76-1.01) | 0.075 | 0.95 | (0.86-1.06) | 0.37 | 0.0013 |
| rs1873062 | 17 | 3292880 | TC | 0.19 | 0.86 | (0.80-0.93) | 0.00022 | 0.91 | (0.82-1.00) | 0.051 | 0.96 | (0.90-1.03) | 0.29 | 0.00095 |
| rs1990236 | 17 | 11806187 | GA | 0.189 | 1.16 | (1.07-1.25) | 0.00025 | 1.12 | (1.01-1.24) | 0.032 | 1.10 | (1.03-1.18) | 0.0082 | $6.6x10^{-6}$ |
| rs231020 | 17 | 15103114 | TC | 0.52 | 0.88 | (0.83-0.93) | $3.7x10^{-5}$ | 0.95 | (0.88-1.03) | 0.18 | 1.05 | (0.99-1.11) | 0.088 | 0.14 |
| rs1526123 | 17 | 41139123 | TC | 0.463 | 0.90 | (0.84-0.95) | 0.00047 | 0.89 | (0.82-0.97) | 0.0063 | 0.95 | (0.90-1.01) | 0.11 | $8.8x10^{-5}$ |
| rs7206949 | 17 | 41602794 | CA | 0.396 | 1.14 | (1.07-1.21) | $6.4x10^{-5}$ | 1.30 | (1.13-1.49) | 0.00021 | 0.98 | (0.92-1.05) | 0.55 | 0.0078 |
| rs2668632* | 17 | 41675143 | AG | 0.758 | 1.13 | (1.05-1.22) | 0.0011 | | | | | | | 0.0011 |
| rs2532348* | 17 | 41696030 | GA | 0.762 | 1.13 | (1.05-1.22) | 0.0011 | | | | | | | 0.0011 |
| rs199523* | 17 | 42203685 | CA | 0.743 | 1.15 | (1.07-1.24) | .00011 | | | | | | | .00011 |
| rs4968451 | 17 | 57282089 | CA | 0.842 | 0.84 | (0.77-0.91) | $1.9x10^{-5}$ | 0.90 | (0.81-1.00) | 0.055 | 0.99 | (0.92-1.06) | 0.72 | 0.0022 |
| rs4522464 | 17 | 57553912 | TG | 0.135 | 1.20 | (1.10-1.31) | $5.0x10^{-5}$ | 1.14 | (1.00-1.30) | 0.047 | 0.99 | (0.92-1.08) | 0.87 | 0.0070 |
| rs2233768 | 17 | 64656171 | TC | 0.949 | 0.81 | (0.71-0.93) | 0.0035 | 0.94 | (0.78-1.13) | 0.52 | 1.05 | (0.93-1.18) | 0.46 | 0.23 |
| rs12606686 | 18 | 9032622 | GA | 0.082 | 1.18 | (1.06-1.32) | 0.0036 | 0.91 | (0.77-1.07) | 0.23 | 1.03 | (0.92-1.15) | 0.63 | 0.096 |
| rs1175745 | 18 | 22841047 | CA | 0.601 | 1.07 | (1.01-1.14) | 0.028 | 1.14 | (1.05-1.23) | 0.0018 | 1.05 | (1.00-1.12) | 0.066 | 0.00055 |
| rs3886058 | 18 | 33702576 | GA | 0.44 | 0.95 | (0.89-1.01) | 0.097 | 0.92 | (0.85-1.00) | 0.057 | 0.99 | (0.93-1.05) | 0.68 | 0.084 |
| rs8098165 | 18 | 65319547 | TC | 0.685 | 1.03 | (0.96-1.10) | 0.41 | 1.01 | (0.93-1.10) | 0.73 | 1.09 | (1.02-1.15) | 0.0057 | 0.011 |
| rs4536550 | 18 | 69099471 | GA | 0.303 | 1.01 | (0.94-1.08) | 0.82 | 1.12 | (1.02-1.21) | 0.013 | 0.99 | (0.93-1.05) | 0.70 | 0.54 |
| rs2163823 | 19 | 43684018 | GA | 0.247 | 0.90 | (0.83-0.96) | 0.0027 | 1.06 | (0.96-1.16) | 0.23 | 0.93 | (0.87-0.99) | 0.020 | 0.0038 |
| rs6027564 | 20 | 58389556 | GA | 0.901 | 1.15 | (1.03-1.27) | 0.0095 | 0.99 | (0.86-1.15) | 0.94 | 1.03 | (0.94-1.13) | 0.55 | 0.11 |
| rs2823093 | 21 | 15442703 | GA | 0.262 | 0.96 | (0.89-1.03) | 0.21 | 0.83 | (0.76-0.92) | 0.00013 | 0.91 | (0.85-0.97) | 0.0032 | $9.5x10^{-5}$ |
| rs2837766 | 21 | 40950152 | TC | 0.615 | 1.12 | (1.05-1.19) | 0.00036 | 1.12 | (1.03-1.21) | 0.0068 | 1.00 | (0.94-1.06) | 0.94 | 0.0067 |
| rs7285871 | 22 | 16242268 | TC | 0.437 | 0.89 | (0.84-0.95) | 0.00042 | 0.90 | (0.83-0.98) | 0.012 | 1.00 | (0.94-1.06) | 0.96 | 0.0055 |
| rs1541326 | 22 | 18034452 | GA | 0.292 | 1.16 | (1.08-1.24) | $1.4x10^{-5}$ | 1.01 | (0.92-1.10) | 0.89 | 0.99 | (0.93-1.06) | 0.72 | 0.024 |
| rs547043 | X | 150007811 | GA | 0.448 | 1.18 | (1.10-1.28) | $1.3x10^{-5}$ | 1.20 | (1.09-1.31) | $8.6x10^{-5}$ | 1.03 | (0.97-1.10) | 0.31 | $7.9x10^{-5}$ |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| rs5970292 | X | 151231436 | GA | 0.644 | 0.85 | (0.79-0.92) | $2.5x10^{-5}$ | 0.91 | (0.83-1.00) | 0.060 | 0.93 | (0.87-0.99) | 0.020 | $2.1x10^{-5}$ |
| rs6627588 | X | 151264915 | CA | 0.274 | 1.20 | (1.11-1.31) | $7.5x10^{-6}$ | 1.18 | (1.05-1.32) | 0.0065 | 1.12 | (1.05-1.20) | 0.0014 | $1.1x10^{-7}$ |

(b) GWAS+BCAC replication

| SNP | Chr. | Position[1] | Alleles | AF[2] | All GWAS | | | BCAC | | | Combined *P* |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | Per-allele OR | (95%CI) | *P* | Per-allele OR | (95%CI) | *P* | |
| rs3738863 | 2 | 30311065 | GA | 0.732 | 1.08 | (1.03-1.13) | 0.00059 | 1.00 | (0.98-1.02) | 0.87 | 0.15 |
| rs6734368 | 2 | 58422570 | GA | 0.81 | 1.02 | (0.97-1.07) | 0.47 | 1.02 | (0.99-1.05) | 0.12 | 0.084 |
| rs1028246/ rs1403400[$] | 2 | 114319652 | TC | 0.915 | 1.11 | (1.03-1.19) | 0.0047 | 1.04 | (0.98-1.11) | 0.20 | 0.0043 |
| rs13400898 | 2 | 154583859 | TC | 0.062 | 0.97 | (0.89-1.06) | 0.50 | 0.98 | (0.91-1.07) | 0.68 | 0.45 |
| rs10469689 | 2 | 163968309 | TC | 0.058 | 0.90 | (0.83-0.97) | 0.0083 | 1.02 | (0.98-1.06) | 0.30 | 0.77 |
| rs11711782 | 3 | 149160340 | GA | 0.119 | 0.96 | (0.91-1.02) | 0.23 | 1.00 | (0.97-1.03) | 0.76 | 0.41 |
| rs9884706 | 4 | 32275985 | TG | 0.844 | 1.09 | (1.03-1.15) | 0.0019 | 1.01 | (0.96-1.06) | 0.76 | 0.018 |
| rs2166278 | 4 | 32286120 | TC | 0.844 | 1.09 | (1.03-1.14) | 0.0023 | 1.01 | (0.96-1.06) | 0.82 | 0.024 |
| rs4403040 | 4 | 87257505 | TC | 0.357 | 1.03 | (0.99-1.07) | 0.14 | 1.03 | (1.01-1.05) | 0.0067 | 0.0020 |
| rs9761051 | 4 | 120233038 | CA | 0.147 | 1.06 | (1.00-1.12) | 0.042 | 0.99 | (0.94-1.04) | 0.63 | 0.29 |
| rs10940235 | 5 | 49672510 | TG | 0.541 | 1.06 | (1.02-1.10) | 0.0060 | 1.03 | (1.01-1.05) | 0.0016 | $6.0x10^{-5}$ |
| rs820848 | 5 | 74000416 | GA | 0.711 | 1.06 | (1.01-1.11) | 0.022 | 1.00 | (0.98-1.02) | 0.92 | 0.30 |
| rs187727 | 5 | 75439941 | GA | 0.277 | 1.05 | (1.01-1.10) | 0.029 | 1.01 | (0.97-1.05) | 0.68 | 0.069 |
| rs4704018 | 5 | 84299468 | TC | 0.624 | 1.04 | (1.00-1.08) | 0.044 | 1.00 | (0.97-1.04) | 0.85 | 0.13 |
| rs11241138 | 5 | 111169939 | CA | 0.29 | 0.94 | (0.91-0.99) | 0.0095 | 0.99 | (0.97-1.01) | 0.39 | 0.051 |
| rs13160298 | 5 | 157728895 | GA | 0.642 | 1.03 | (0.99-1.07) | 0.15 | 0.98 | (0.93-1.02) | 0.21 | 0.63 |
| rs12203592 | 6 | 341321 | TC | 0.772 | 0.97 | (0.91-1.04) | 0.39 | 1.01 | (0.97-1.04) | 0.73 | 0.90 |
| rs9467504 | 6 | 25522304 | GA | 0.837 | 0.92 | (0.87-0.97) | 0.0029 | 1.01 | (0.96-1.06) | 0.71 | 0.082 |
| rs12333016 | 6 | 107205884 | GA | 0.9 | 0.98 | (0.91-1.04) | 0.45 | 1.02 | (0.96-1.09) | 0.43 | 0.96 |
| rs9401003 | 6 | 117824996 | GA | 0.781 | 0.92 | (0.88-0.97) | 0.00065 | 1.00 | (0.96-1.05) | 0.91 | 0.022 |
| rs2189630 | 7 | 8695731 | TG | 0.655 | 0.96 | (0.92-1.01) | 0.085 | 1.01 | (0.98-1.03) | 0.60 | 0.75 |
| rs7810604 | 7 | 11387506 | TC | 0.371 | 0.93 | (0.90-0.97) | 0.00081 | 1.00 | (0.97-1.02) | 0.65 | 0.052 |
| rs1981576 | 7 | 82057361 | TC | 0.087 | 0.94 | (0.88-1.01) | 0.10 | 1.00 | (0.94-1.06) | 0.90 | 0.24 |
| rs1011692 | 7 | 121347247 | TC | 0.123 | 0.98 | (0.93-1.04) | 0.46 | 0.99 | (0.90-1.08) | 0.81 | 0.45 |
| rs2617076 | 8 | 4432582 | CA | 0.4 | 1.03 | (0.99-1.07) | 0.14 | 1.00 | (0.96-1.04) | 1.0 | 0.31 |
| rs4872360 | 8 | 22084613 | TC | 0.259 | 0.93 | (0.89-0.97) | 0.00099 | 0.99 | (0.97-1.02) | 0.63 | 0.053 |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| rs2211914 | 8 | 102738843 | GA | 0.612 | 0.95 | (0.91-0.99) | 0.012 | 1.00 | (0.96-1.04) | 0.88 | 0.066 |
| rs4268201 | 9 | 36878148 | TG | 0.862 | 0.94 | (0.89-0.99) | 0.020 | 0.99 | (0.96-1.02) | 0.54 | 0.098 |
| rs1952461 | 9 | 78412404 | GA | 0.047 | 0.99 | (0.87-1.13) | 0.92 | 0.98 | (0.91-1.05) | 0.51 | 0.53 |
| rs1932658 | 9 | 86005570 | GA | 0.727 | 0.94 | (0.90-0.98) | 0.0029 | 1.02 | (0.99-1.05) | 0.12 | 0.79 |
| rs12350727 | 9 | 93304437 | GA | 0.942 | 0.92 | (0.84-1.00) | 0.039 | 1.00 | (0.93-1.08) | 0.96 | 0.15 |
| rs4455975 | 9 | 128423020 | GA | 0.449 | 1.07 | (1.03-1.11) | 0.00056 | 1.02 | (0.98-1.06) | 0.30 | 0.0017 |
| rs7074055 | 10 | 12665442 | GA | 0.769 | 0.96 | (0.92-1.01) | 0.098 | 0.99 | (0.95-1.04) | 0.68 | 0.15 |
| rs4746065 | 10 | 72183963 | CA | 0.227 | 0.97 | (0.93-1.02) | 0.20 | 1.01 | (0.99-1.04) | 0.24 | 0.66 |
| rs7116850 | 11 | 27602106 | GA | 0.731 | 1.05 | (1.01-1.10) | 0.028 | 0.97 | (0.93-1.01) | 0.11 | 0.73 |
| rs11604821 | 11 | 69061318 | GA | 0.665 | 0.95 | (0.91-0.99) | 0.022 | 0.97 | (0.93-1.01) | 0.093 | 0.0053 |
| rs2284424 | 12 | 13880137 | GA | 0.296 | 1.05 | (1.00-1.09) | 0.034 | 1.03 | (0.99-1.07) | 0.17 | 0.014 |
| rs10771399 | 12 | 28046347 | GA | 0.893 | 1.22 | (1.15-1.31) | $5.7\times10^{-10}$ | 1.18 | (1.14-1.21) | $3.3\times10^{-27}$ | $2.7\times10^{-35}$ |
| rs1975930 | 12 | 28053015 | TC | 0.107 | 0.82 | (0.77-0.87) | $5.3\times10^{-10}$ | 0.83 | (0.79-0.87) | $1.4\times10^{-15}$ | $5.7\times10^{-24}$ |
| rs1292011 | 12 | 114320905 | GA | 0.586 | 1.10 | (1.06-1.15) | $9.5\times10^{-7}$ | 1.08 | (1.06-1.10) | $6.2\times10^{-14}$ | $4.8\times10^{-19}$ |
| rs7955262 | 12 | 114691345 | TC | 0.103 | 1.10 | (1.03-1.17) | 0.0054 | 1.04 | (0.98-1.10) | 0.18 | 0.0042 |
| rs9586525 | 13 | 103922680 | TC | 0.185 | 1.06 | (1.01-1.12) | 0.021 | 1.03 | (0.98-1.08) | 0.27 | 0.016 |
| rs3784194 | 14 | 31993377 | TC | 0.827 | 0.95 | (0.91-1.00) | 0.066 | 1.03 | (1.00-1.05) | 0.034 | 0.30 |
| rs1263441 | 14 | 82695911 | GA | 0.715 | 1.04 | (1.00-1.09) | 0.067 | 1.06 | (0.99-1.14) | 0.089 | 0.015 |
| rs10484150 | 14 | 82707696 | TC | 0.715 | 1.04 | (1.00-1.09) | 0.063 | 1.05 | (0.92-1.21) | 0.48 | 0.047 |
| rs2277509 | 14 | 90819348 | CA | 0.697 | 1.04 | (0.99-1.08) | 0.085 | 1.04 | (1.00-1.08) | 0.060 | 0.011 |
| rs3101649 | 15 | 25607717 | GA | 0.062 | 1.12 | (1.03-1.22) | 0.010 | 1.00 | (0.96-1.04) | 0.98 | 0.28 |
| rs12443310 | 15 | 52387367 | TC | 0.911 | 0.89 | (0.83-0.96) | 0.0013 | 1.04 | (1.00-1.08) | 0.043 | 0.85 |
| rs1873062 | 17 | 3292880 | TC | 0.19 | 0.92 | (0.88-0.97) | 0.00095 | 0.97 | (0.93-1.02) | 0.19 | 0.0013 |
| rs1990236 | 17 | 11806187 | GA | 0.189 | 1.12 | (1.07-1.18) | $6.6\times10^{-6}$ | 1.02 | (0.99-1.05) | 0.12 | 0.00052 |
| rs231020 | 17 | 15103114 | TC | 0.52 | 0.97 | (0.93-1.01) | 0.14 | 0.99 | (0.96-1.03) | 0.77 | 0.22 |
| rs1526123 | 17 | 41139123 | TC | 0.463 | 0.92 | (0.89-0.96) | $8.8\times10^{-5}$ | 0.99 | (0.96-1.03) | 0.76 | 0.0038 |
| rs7206949 | 17 | 41602794 | CA | 0.396 | 1.06 | (1.02-1.11) | 0.0078 | 1.01 | (0.98-1.05) | 0.48 | 0.024 |
| rs2668632* | 17 | 41675143 | AG | 0.758 | 1.13 | (1.05-1.22) | 0.0011 | 1.05 | (1.01-1.10) | 0.028 | 0.00031 |
| rs2532348* | 17 | 41696030 | GA | 0.762 | 1.13 | (1.05-1.22) | 0.0011 | 1.08 | (1.04-1.12) | $7.8\times10^{-5}$ | $5.8\times10^{-7}$ |
| rs199523* | 17 | 42203685 | CA | 0.743 | 1.15 | (1.07-1.24) | .00011 | 1.12 | (1.03-1.21) | 0.0063 | $2.6\times10^{-6}$ |
| rs4968451 | 17 | 57282089 | CA | 0.842 | 0.92 | (0.88-0.97) | 0.0022 | 0.98 | (0.93-1.03) | 0.34 | 0.0048 |
| rs4522464 | 17 | 57553912 | TG | 0.135 | 1.08 | (1.02-1.15) | 0.0070 | 1.02 | (0.97-1.08) | 0.38 | 0.014 |
| rs2233768 | 17 | 64656171 | TC | 0.949 | 0.95 | (0.87-1.03) | 0.23 | 1.05 | (0.96-1.14) | 0.29 | 0.93 |
| rs12606686 | 18 | 9032622 | GA | 0.082 | 1.07 | (0.99-1.15) | 0.096 | 0.98 | (0.94-1.02) | 0.39 | 0.95 |
| rs1175745 | 18 | 22841047 | CA | 0.601 | 1.07 | (1.03-1.12) | 0.00055 | 1.00 | (0.97-1.04) | 0.80 | 0.010 |
| rs3886058 | 18 | 33702576 | GA | 0.44 | 0.97 | (0.93-1.00) | 0.084 | 0.99 | (0.97-1.01) | 0.60 | 0.20 |
| rs8098165 | 18 | 65319547 | TC | 0.685 | 1.05 | (1.01-1.10) | 0.011 | 0.98 | (0.95-1.00) | 0.048 | 0.64 |
| rs4536550 | 18 | 69099471 | GA | 0.303 | 1.01 | (0.97-1.06) | 0.54 | 1.01 | (0.97-1.05) | 0.74 | 0.51 |
| rs2163823 | 19 | 43684018 | GA | 0.247 | 0.94 | (0.89-0.98) | 0.0038 | 1.01 | (0.96-1.05) | 0.82 | 0.054 |
| rs6027564 | 20 | 58389556 | GA | 0.901 | 1.06 | (0.99-1.13) | 0.11 | 1.08 | (1.02-1.15) | 0.010 | 0.0029 |
| rs2823093 | 21 | 15442703 | GA | 0.262 | 0.92 | (0.88-0.96) | $9.5\times10^{-5}$ | 0.94 | (0.92-0.96) | $1.7\times10^{-9}$ | $1.1\times10^{-12}$ |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| rs2837766 | 21 | 40950152 | TC | 0.615 | 1.06 | (1.02-1.10) | 0.0067 | 0.97 | (0.94-1.01) | 0.18 | 0.38 |
| rs7285871 | 22 | 16242268 | TC | 0.437 | 0.94 | (0.91-0.98) | 0.0055 | 0.98 | (0.95-1.02) | 0.40 | 0.013 |
| rs1541326 | 22 | 18034452 | GA | 0.292 | 1.05 | (1.01-1.10) | 0.024 | 0.97 | (0.95-0.99) | 0.012 | 0.22 |
| rs547043 | X | 150007811 | GA | 0.448 | 1.10 | (1.05-1.15) | $7.9x10^{-5}$ | 1.02 | (0.98-1.05) | 0.40 | 0.0018 |
| rs5970292 | X | 151231436 | GA | 0.644 | 0.90 | (0.86-0.95) | $2.1x10^{-5}$ | 1.02 | (1.00-1.04) | 0.13 | 0.71 |
| rs6627588 | X | 151264915 | CA | 0.274 | 1.15 | (1.09-1.21) | $1.1x10^{-7}$ | 1.03 | (0.99-1.07) | 0.20 | $2.0x10^{-5}$ |

[1] Build 36
[2] Frequency of the 2nd listed allele
[*] SNPs not on HapMap 2, GWAS results from genotyped data (UK2 only)
[+] SNP not on HapMap 2, GWAS results imputed from HapMap3
$ GWAS results for SNP rs1028246, surrogate rs1403400 typed in the BCAC replication

**Supplementary Table 3.** Estimated per-allele ORs for Asian women from BCAC.

| SNP | Chromosome Position | MAF | Per-allele OR (95%CI) | P | P for difference with OR for European women |
|---|---|---|---|---|---|
| rs10771399 | 12p11 28046347 | 0.11 | 0.85 (0.77-0.94) | 0.002 | 0.93 |
| rs1292011 | 12q24 114320905 | 0.41 | 0.95 (0.85-1.07) | 0.43 | 0.59 |
| rs2823093 | 21q21 15442703 | 0.26 | 1.14 (0.93-1.40) | 0.20 | 0.058 |

**Supplementary Table 4.** Estimated per-allele ORs by ER-status, DCIS vs invasive disease, and age at diagnosis from the BCAC replication.

(a) Estimated ORs by ER-status

| SNP | Chromosome Position | No. Controls | No. ER+ cases | ER+ Per-allele OR (95%CI) | P | No. ER- cases | ER- Per-allele OR (95%CI) | P | P-diff |
|---|---|---|---|---|---|---|---|---|---|
| rs10771399 | 12p11 28046347 | 48,368 | 24,775 | 0.87 (0.84-0.90) | $4.5x10^{-14}$ | 7,122 | 0.85 (0.80-0.90) | $6.3x10^{-8}$ | .46 |
| | | | | | | | | | |
| rs1292011 | 12q24 114320905 | 40,739 | 18,394 | 0.90 (0.87-0.92) | $2.0x10^{-15}$ | 5,167 | 0.99 (0.94-1.03) | .55 | .0001 |
| | | | | | | | | | |
| rs2823093 | 21q21 15442703 | 46,063 | 21,409 | 0.93 (0.90-0.95) | $4.6x10^{-8}$ | 6,093 | 0.99 (0.95-1.04) | .72 | .02 |
| | | | | | | | | | |

(b) Estimated ORs for DCIS.

| SNP | No. controls | No. DCIS cases | Chromosome Position | Per-allele OR (95%CI) | P (vs.controls) | P (vs.invasive disease) |
|---|---|---|---|---|---|---|
| rs10771399 | 33,767 | 2,148 | 12p11 28046347 | 0.89 (0.80-0.99) | .026 | 0.50 |
| rs1292011 | 28,588 | 1,566 | 12q24 114320905 | 0.92 (0.86-1.00) | .043 | 0.80 |
| rs2823093 | 31,543 | 1,881 | 21q21 15442703 | 0.94 (0.87-1.01) | .11 | 0.39 |

(c) Estimated per-allele ORs by age-group.

| SNP | Age group | | | | P-trend |
|---|---|---|---|---|---|
| | <40 | 40-49 | 50-59 | 60+ | |
| rs10771399 | 0.83 (0.75-0.92) | 0.81 (0.75-0.87) | 0.86 (0.81-0.91) | 0.88 (0.84-0.93) | .31 |
| | | | | | |
| rs1292011 | 0.96 (0.90-1.04) | 0.91 (0.87-0.96) | 0.93 (0.89-0.96) | 0.92 (0.89-0.95) | .25 |
| | | | | | |
| rs2823093 | 0.96 (0.90-1.04) | 0.94 (0.90-0.99) | 0.92 (0.89-0.96) | 0.94 (0.91-0.97) | .58 |
| | | | | | |

**Supplementary Table 5.** Estimated per-allele ORs among studies in which cases were selected or unselected for family history/bilaterality from the BCAC replication.

| SNP | Unselected studies | | Selected studies | | P diff |
|---|---|---|---|---|---|
| | Per-allele OR (95%CI) | P | Per-allele OR (95%CI) | P | |
| rs10771399 | 0.86 (0.83-0.89) | $8.6x10^{-21}$ | 0.79 (0.73-0.85) | $4.4x10^{-10}$ | .027 |
| rs1292011 | 0.92 (0.90-0.95) | $4.1x10^{-21}$ | 0.92 (0.87-0.97) | .0013 | .80 |
| rs2823093 | 0.94 (0.92-0.96) | $3.9x10^{-8}$ | 0.91 (0.86-0.97) | .0017 | .31 |

**Supplementary Table 6.** Genome-wide association studies contributing to the current analysis.

| Study | Country | Case ascertainment | Control ascertainment | Genotyping platform | Cases[1] | Controls[1] | Reference |
|---|---|---|---|---|---|---|---|
| ABCFS/kConFab | Australia | Recruitment through cancer registries in Victoria and New South Wales | Recruitment from the electoral rolls in Melbourne and Sydney matched to cases by age in-5 year categories | Illumina 610k | 282 | 285 | [1] |
| BBCS | UK | Recruitment through cancer registries in UK, predominantly bilateral cases | WTCCC2: 1958 Birth Cohort + UK National Blood Service | Illumina 370k (cases) Illumina 1.2M (controls) | 1609 | 5190 | [2] |
| CGEMS | USA | Postmenopausal cases from Nurses Health Study | Individually matched controls from Nurses Health Study | Illumina 550k | 1127 | 1130 | [3] |
| GC-HBOC | Germany | BRCA1/2 mutation negative cases from University Clinics in Cologne and Munich | KORA (Cooperative Health Research in the Region Augsburg) | Affymetrix 5.0k (cases) Affymetrix 6.0k (controls) | 634 | 477 | [4] |
| MARIE | Germany | Random sample of cases from the MARIE study, but restricted to | KORA (Cooperative Health Research in the | Illumina 370k (cases) Illumina 550k (controls) | 708 | 470 | [5] |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | | ductal and lobular carcinomas and oversampled for lobular (about 2:1) | Region Augsburg) | | | | |
| HEBCS | Finland | Unselected cases plus additional familial cases from Helsinki University Central Hospital | Healthy Population Controls from Finnish Genome Centre (NordicDB) | Illumina 550k + 610k (cases) Illumina 370k (controls) | 810 | 1012 | [6, 7] |
| SASBAC | Sweden | Population-based case control study of postmenopausal women | Population-based controls frequency matched by age to cases | Illumina 317k+240k (cases) Illumina 550k (controls) | 790 | 756 | [7] |
| UK2 | UK | UK cancer genetics clinics + oncology clinicas | WTCCC2: 1958 Birth Cohort + UK National Blood Service | Illumina 670k (cases) Illumina 1.2M (controls) | 3628 | 5190 | [8] |
| DFBBCS | Netherlands | BRCA1/2 mutation negative familial bilateral breast cancer patients selected from five clinical genetics centers; Erasmus University Medical | Controls were from the Rotterdam study, and are 55 years or older at the time of inclusion. For this study females were selected and | Illumina 610k (cases) Illumina 550k (controls) | 464 | 3255 | [9] |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | | Center/Daniel den Hoed, The Netherlands Cancer Institute, Leiden University Medical Center, University Medical Center Utrecht, and VU University Medical Center. | breast cancer cases were excluded. | | | | |

[1]Final numbers used in the analysis, after QC.

**Supplementary Table 7.** Participating studies in the BCAC replication phase.

| Study Acronym | Study Name [Reference] | Country | Recruitment base | |
|---|---|---|---|---|
| | | | Cases | Controls |
| **STUDIES OF WHITE EUROPEAN WOMEN** | | | | |
| ABCFR | Australian Breast Cancer Family Study [1] | Australia | Cancer registries in Victoria and New South Wales (1992-1999): all cases from Melbourne and Sydney diagnosed before age 40 plus a random sample of those diagnosed at ages 40-59 | Identified between 1992-1999 from the electoral rolls in Melbourne and Sydney (enrolling to vote is compulsory); frequency matched to cases by age in-5 year categories |
| ABCS | Amsterdam Breast Cancer Study [10] | Netherlands | (ABCS-F) All non-BRCA1/2 breast cancer cases from the family cancer clinic of the NKI-AVL tested in the period 1995-2009; all ages and diagnosed with breast cancer in 1965-2008.. | Randomly selected women from population-based prospective cohort studies, aged <50 at baseline (1987-1991 and 1993-1997) and from the same areas as cases. |
| BBCC | Bavarian Breast Cancer Cases and Controls [11] | Germany | Consecutive, unselected cases with invasive breast cancer recruited at the University Breast Centre, Franconia in Northern Bavaria from 2002-2006 | Healthy women aged 55 or older with no diagnosis of cancer. Invited by a newspaper advertisement in Northern Bavaria between 2002-2006 |
| BBCS | British Breast Cancer Study [2] | U.K. | (i) English & Scottish Cancer Registries: all breast cancer cases who developed a first primary before age 66 in 1971 or later and who subsequently developed a second primary cancer<br>(ii) Breast Cancer Clinics: all breast cancer cases who developed a first primary before age 71 in 1967 or later and who either subsequently developed a second primary or had at least two affected female first-degree relatives.<br>All recruited from 2001-2008. | A friend, sister-in-law, daughter-in-law or other non-blood relative of cases, recruited from 2001-2008 |
| BIGGS | Breast Cancer in Galway Genetic Study [12] | Ireland | Unselected cases recruited from University College Hospital Galway and surrounding hospitals in the West of Ireland since 2001 | Women > 60 years with no personal history of any cancer and no family history of breast or ovarian cancer identified from retirement groups in the West of Ireland between 2001-2008. |
| BSUCH | Breast Cancer Study of the University Clinic Heidelberg [13] | Germany | All cases diagnosed with breast cancer in 2007-2009 at the University Women`s Clinic Heidelberg | Female blood donors recruited in 2007- 2009 at the Institute of Transfusion Medicine & Immunology, Mannheim. |

| | | | | |
|---|---|---|---|---|
| CECILE | CECILE Breast cancer study [14] | France | All cases diagnosed with breast cancer in 2005-2007 among women <75 years of age residing in the *départements* of Ille-et-Vilaine and Côte d'Or . Cases were recruited from the main cancer treatment center (Centre Eugène-Marquis in Rennes and Centre Georges-François-Leclerc in Dijon) and from other private or public hospitals in each area. | General population control women residing in the same areas as the cases (Ille-et-Vilaine and Côte d'Or). Controls were frequency-matched to the cases by 5-year age groups. They were recruited in 2005-2007 using a random digit dialing procedure and quotas by socioeconomic status to reflect the distribution by SES of the population in each area. |
| CGPS | Copenhagen General Population Study [15] | Denmark | Consecutive, incident cases from one hospital with centralized care for a population of 400,000 women in Copenhagen (2001-present) | Women with no history of breast cancer residing in the same region as cases identified from the Copenhagen General Population Study (2003-2007) |
| CNIO-BCS | Spanish National Cancer Centre Breast Cancer Study [16] | Spain | (i) consecutive breast cancer patients from three public hospitals, two in Madrid and one in Oviedo; (ii) cases with at least one affected first degree relative recruited through the CNIO family cancer clinic in Madrid (2000-2005) | Women attending the Menopause Research Centre, Madrid and female members of the College of Lawyers attending a free, targeted medical check-up in Madrid, all free of breast cancer and all in Madrid between 2000-2005 |
| CTS | California Teachers Study [17] | USA | Nested case-control study conducted within a cohort of California teachers (113,590) who were under age 80 years at baseline, had no prior history of invasive or *in situ* breast cancer. Cases are women newly diagnosed with a histologically confirmed invasive primary adenocarcinoma of the breast at age 80 years or younger from 1998 to 2008. | Controls are a probability sample of at-risk cohort members, frequency matched to cases on age at baseline (5-year age groups), self-reported race/ethnicity (white, African American, Latina,Asian, other), and broad geographic region within California Controls were selected without replacement, using an assigned reference date. |
| ESTHER | ESTHER Breast Cancer Study [18] | Germany | Breast cancer cases in all hospitals in the state of Saarland, from 2001-2003 (ESTHER) and 1996-1998 (VERDI) | Random sample of women a routine health check-up in Saarland, in 2000-2002; frequency matched to cases by age in-5 year categories |
| GC-HBOC | German Familial Breast Cancer Study [4] | Germany | Index patients from German breast cancer families; BRCA1/2 mutation free, collected 1996-2007 via Institute of Human Genetics, University Heidelberg Department of Gynaecology & Obstetrics, Cologne & Department of Gynaecology and Obstetrics at the Ludwig-Maximilians-University, Munich | Female blood donors recruited in 2004 & 2007 at the Institute of Transfusion Medicine & Immunology, Mannheim. |
| GENICA | Gene Environment | Germany | Incident breast cancer cases enrolled at hospitals | Random address sample selected in 2001-2004 from |

| | | | | |
|---|---|---|---|---|
| | Interaction & Breast Cancer in Germany [19, 20] | | in the Greater Bonn area between 2000-2004 | 31 population registries in the greater Bonn area; frequency matched to cases on year of birth in 5-year categories |
| GESBC | Genetic Epidemiology Study of Breast Cancer by Age 50 [21] | Germany | All incident cases diagnosed <50 years of age in 1992-5 in two regions: Rhein-Neckar-Odenwald and Freiburg, by surveying the 38 clinics serving these regions | Selected from random lists of residents of the study regions supplied by population registries; two controls were selected for each case, matched by age and study region. Recruitment was carried out 1992-1998. |
| HABCS | Hannover Breast Cancer Study [22] | Germany | Cases who received radiotherapy for breast cancer at Hannover Medical School 1995-2003 | Female blood bank donors at Hannover Medical School, collected in 2005 |
| HEBCS | Helsinki Breast Cancer Study | Finland | (1) Consecutive cases (883) from the Department of Oncology, Helsinki University Central Hospital 1997-8 and 2000, (2) Consecutive cases (986) from the Department of Surgery, Helsinki University Central Hospital 2001 – 2004, (3) Familial breast cancer patients (536) from the Helsinki University Central Hospital, Departments of Oncology and Clinical Genetics (1995-) | Healthy females from the same geographical region in Southern Finland in 2003. |
| HMBCS | Hannover-Minsk Breast Cancer Study [23] | Belarus | Cases from the Byelorussian Institute for Oncology and Medical Radiology Aleksandrov N.N. in Minsk or at one of 5 regional oncology centers in Gomel, Mogilev, Grodno, Brest or Vitebsk (2002-2008) | Women attending general medical examination at gynecology clinics in Gomel, Mogilev, Grodno, Brest or Vitebsk; women attending the Institute for Inherited Diseases in Minsk; female blood donors in Minsk; healthy relatives of cases (2002-2008) |

| | | | | |
|---|---|---|---|---|
| HUBCS | Hannover-Ufa Breast Cancer Study [23] | Russia | Consecutive cancer patients diagnosed at two participating oncological centers in Bashkorstostan and Siberia between 2000-2008 | Healthy volunteers selected from poulation studies in the same geographical regions during 2002-2008. |
| KARBAC | Karolinska Breast Cancer Study | Sweden | 1. Familial cases from Department of Clinical Genetics, Karolinska University Hospital , Stockholm. 2. Consecutive cases from Department of Oncology, Huddinge & Söder Hospital, Stockholm 1998-2000 | Blood donors of mixed gender from same geographical region. Excess material was received from all blood donors over a 3 month period in 2004 (approximately 3000) and DNA was extracted from a random sample of 1500 |
| KBCP | Kuopio Breast Cancer Project [24] | Finland | Women seen at Kuopio University Hospital between 1990-1995 because of a breast lump, mammographic abnormality, or other breast symptom and who were found to have breast cancer | Selected from the National Population Register between 1990-1995; age and long-term area-of-residence matched to cases |
| kConFab/ AOCS | Kathleen Cuningham Foundation Consortium for Research into Familial Breast Cancer / Australian Ovarian Cancer Study [25] | Australia | Index (youngest affected) cases from *BRCA1*- and *BRCA2*-mutation-negative multiple-case breast and breast-ovarian families recruited though family cancer clinics from across Australia and New Zealand from 1998-present | Identified from the electoral rolls from across Australia as part of the Australian Ovarian Cancer Study in 2002-2006 |
| LMBC | Leuven Multidisciplinary Breast Centre [26] | Belgium | All patients diagnosed with breast cancer and seen in the Multidisciplinary Breast Center in Leuven (Gashuisberg) since June 2007 plus retrospective collection of cases diagnosed since 2000 | Blood donors at Gasthuisberg Hospital (200-2008) |
| MARIE | Mammary Carcinoma Risk Factor Investigation [5] | Germany | Incident cases diagnosed from 2001-2005 in the study region Hamburg in Northern Germany, and from 2002-2005 in the study region Rhein-Neckar-Karlsruhe in Southern Germany. | 2 controls per case were randomly drawn from population registries and frequency matched by birth year and study region to the case. Controls were recruited from 2002 to 2006. |
| MBCSG | Milan Breast Cancer Study Group [27] | Italy | Familial and/or early onset breast cancer patients (aged 22-87) negative for mutations in *BRCA1* and *BRCA2*, ascertained at two large cancer centers in Milan from 2000-present | Female blood donors recruited at two centres in Milan from 2004-present and 2007-present |
| MCBCS | Mayo Clinic Breast Cancer Study [28] | U.S.A. | Incident cases residing in 6 states (MN, WI, IA, IL, ND, SD) seen at the Mayo Clinic in Rochester, MN from 2002-2010 | Women presenting for general medical examination at the Mayo Clinic from 2002-2010; frequency matched to cases on age, ethnicity and county/state |

| | | | | |
|---|---|---|---|---|
| MCCS | Melbourne Collaborative Cohort Study [29] | Australia | Incident cases from the cohort of 24,469 women, diagnosed during the follow-up from baseline (1990-1994) to 2008 | Random sample of the initial cohort |
| NBCS | Norwegian Breast Cancer Study [30] | Norway | Incidence cases from three different hospitals: Ullevål Univ. Hospital 1990-94, Norwegian Radium Hospital 1975-1986 and 1995-1998, Haukeland Univ. Hospital 1992-2001 | Women residing in Tromsø and Bergen who attended the Norwegian Breast Cancer Screening Program. |
| NC-BCFR | Northern California Breast Cancer Family Registry [31] | U.S.A. | Incident cases aged <65 years identified through the SEER cancer registry of the Greater San Francisco Bay Area from 1995-2003. All cases likely at increased genetic risk were selected; 2.5% of white cases not meeting these criteria were randomly sampled | Identified through random digit dialling conducted in the same geographic region from 1999-2000; frequency matched to cases diagnosed from 1995-1998 on 5-year age group and race/ethnicity |
| OBCS | Oulu Breast Cancer Study [32] | Finland | Consecutive incident cases diagnosed at the Oulu University Hospital between 2000-2004 | Female blood donors recruited in 2002 from the same geographical region in Northern Finland |
| OFBCR | Ontario Familial Breast Cancer Registry [31] | Canada | Invasive cases aged 20-54 years identified from the Ontario Cancer Registry from 1996-1998. All those at high genetic risk were eligible; random samples of women not meeting these criteria were also asked to participate. | Identified by calling randomly selected residential telephone numbers in the same geographical region from 1998-2001; frequency matched to cases by age in 5 year categories |
| ORIGO | Leiden University Medical Centre Breast Cancer Study [33, 34] | Netherlands | Consecutive case patients diagnosed 1996–2006 in 2 hospitals in South–West Netherlands (Leiden & Rotterdam). No selection for family history; Rotterdam case patients selected for diagnosis aged <70. Case patients with in situ carcinomas eligible. | (1) Blood bank healthy donors from Southwest Netherlands recruited in 1996, 2000 or 2007; (2) People who married a person who was part of a family with high breast cancer risk (BRCA1/2/x). From the Southwest of the Netherlands, recruited 1990–1996; (3) Females tested at the local clinical genetics department for familial diseases, excluding familial cancer syndromes (no mutation found in gene(s) related to the disease being tested), recruited 1995–2007. |
| PBCS | NCI Polish Breast Cancer Study [35] | Poland | Incident cases identified through a rapid identification system in participating hospitals covering ~ 90% of all eligible cases, and cancer registries in Warsaw and Łódź covering 100% of all eligible cases (2000-2003) | Randomly selected from population lists of all residents of Poland from 2000-2003, stratified and frequency matched to cases on city and age in 5-year categories |

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| RBCS | Rotterdam Breast Cancer Study [36] | Netherlands | Familial breast cancer patients selected from the clinical genetics center at Erasmus Medical Center between 1994-2005 | Spouses or mutation-negative siblings of heterozygous Cystic Fibrosis mutation carriers selected from the clinical genetics centre at Erasmus Medical Center between 1996-2006 |
| SASBAC | Singapore and Sweden Breast Cancer Study [37] | Sweden | Women diagnosed in Sweden aged 50-74 in 1993-1995 | Population-based controls frequency matched by age to the caases |
| SBCS | Sheffield Breast Cancer Study [38] | U.K. | Women with breast cancer recruited in 1998-2005 at surgical outpatient clinics at the Royal Hallamshire Hospital, Sheffield | Unselected women attending the Sheffield Mammography Screening Service in 2000-2004 with no evidence of a breast lesion |
| SEARCH | Study of Epidemiology & Risk Factors in Cancer Heredity [39] | U.K. | Identified through the East Anglian Cancer Registry: (i) 1991-1996: alive, prevalent cases diagnosed before age 55; (ii) since 1996: incident cases diagnosed before age 70 diagnosed after 1996 | (a) Women from the same geographic region selected from the EPIC-Norfolk cohort study, 1992-1994 (b) women attending GP practices, frequency matched to cases by age and geographic region (2003-present) |

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|---|---|---|---|---|
| SZBCS | IHCC-Szczecin Breast Cancer Study [40] | Poland | Prospectively ascertained cases of invasive breast cancer patients diagnosed at the Regional Oncology Hospital (2002-2003 and 2006-2007) or the University Hospital (2002-2007), both in Szczecin, West Pomerania, Poland. | Selected from a population-based study of the 1.3 million inhabitants of West Pomerania (2003-2004); matched to cases for year of birth, sex and region |
| UCIBCS | UCI Breast Cancer Study [41] | U.S.A. | All cases diagnosed in Orange County, California from 1994-1995; ascertained through the population-based Cancer Surveillance Program of Orange County California (CSPOC) | Recruited from 1998-2003 using random digit dialling among Orange County residents; frequency matched to cases by age and race/ethnicity |
| UKBGS | Breakthrough Generations Study | UK | All cohort members who had had breast cancer or in situ breast cancer before entry into the Breakthrough Generations Study (cohort of >100,000 women followed up for breast cancer, recruited from the UK during 2003-2011). | Women who had not had breast cancer or in situ breast cancer before entry into the cohort study selected by 1:1 matching to cases on date of birth, year of entry in to the study (2003-2009) source of recruitment, blood sample and ethnicity. |
| **STUDIES OF ASIAN WOMEN** | | | | |
| ACP | Asian Cancer Project | Thailand | Cases from oncology centres in Thailand that underwent biopsy and had been pathologically diagnosed as having breast cancer. | Hospital based controls are women who were admitted to the same hospital as the cases with diseases not related to cancer or metabolic syndromes such as diabetes, heart diseases or conditions related to gynaecology. |
| SEBCS | Seoul Breast Cancer Study [42] | South Korea | Consecutive, incident, cases from 2 hospitals in Seoul recruited between 2001-2005 | Women from same catchment area and participating in annual health check-up (2001-2005) |
| TWBCS | Taiwanese Breast Cancer Study [43] | Taiwan | Incident cases diagnosed & treated at 2 major teaching hospitals in Taiwan between 2002-2005 | Randomly selected women attending a health examination at same hospitals between 2002-2005 |

# Supplementary Note

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