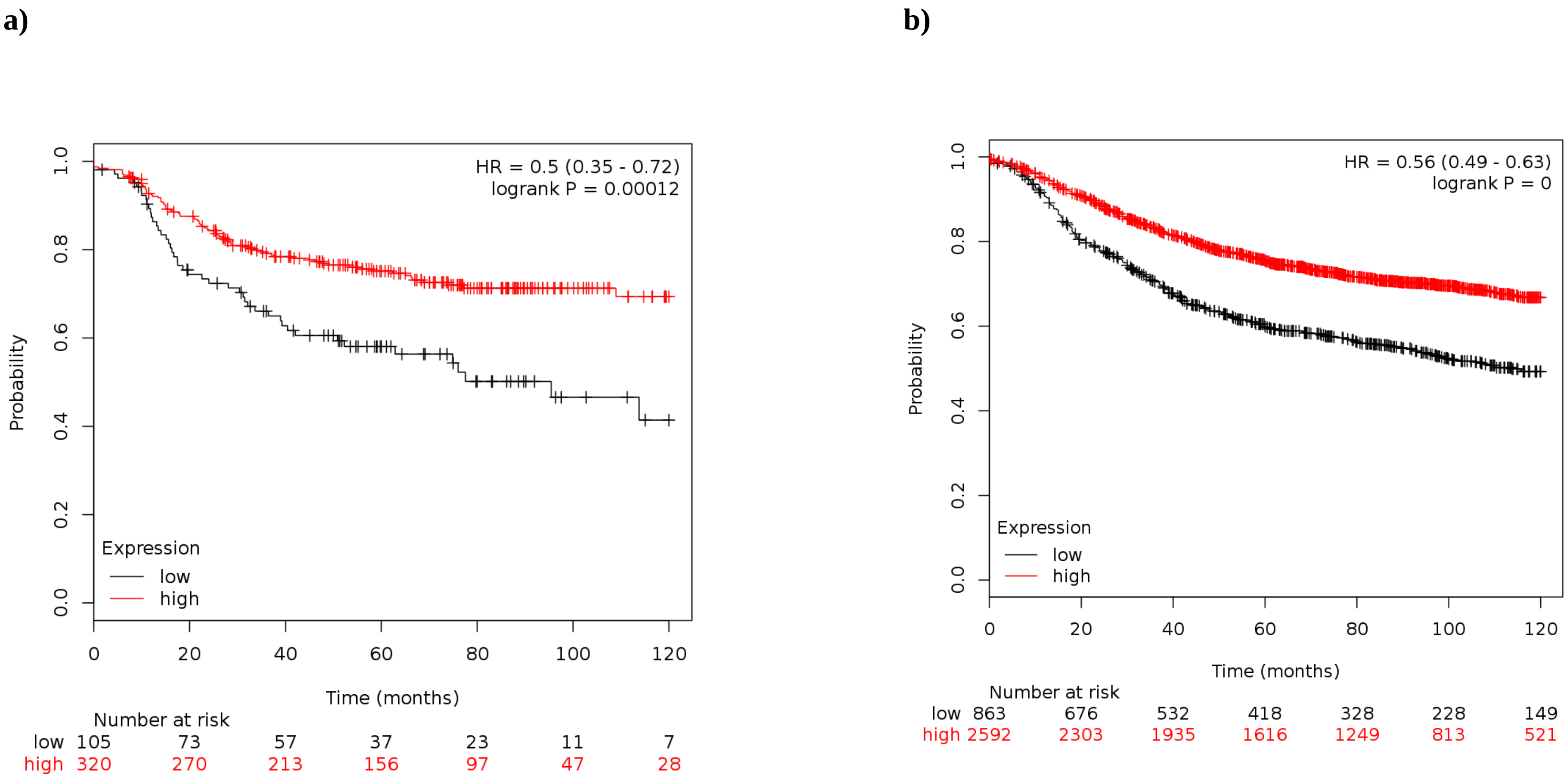
# The SNP rs6500843 in 16p13.3 is associated with survival specifically among chemotherapy-treated breast cancer patients

## Supplementary Material



**Supplementary Figure 1**: Expression of the *CCDC28A* gene is associated with relapse-free survival in breast cancer. Kaplan-Meier curves according to binarized *CCDC28A* gene expression in the KM-plotter database: **a**) cases treated with adjuvant chemotherapy, **b**) all cases.

**Supplementary Table 1:** Summarized SNP selection and survival analysis statistics from Stage I and Stage II analyses for all SNPs. Note that some Stage I SNPs are represented by several LD-tagged SNPs in Stage II and vice versa, and as a result appear as duplicates in the table. "iCOGS_SNP", "POSH_SNP" and "SUCCESS_SNP" indicate the SNPs matching to the selected SNP from HEBCS-GWS. In cases where the SNP is a tagging SNP, LD information is provided (r2 > 0.7 and D' > 0.8). "NA" indicates that no satisfactory match was found in that data set. The "Cat." column indicates one of the following Stage I selection criteria: Ia) 10 year overall survival (p < 10-4); Ib) 10 year breast cancer specific survival (p < 10-4); Ic) 5 year BDDM (breast cancer death or distant metastasis, p < 10-4); II) chemotherapy-treated cases (p < 10-4); III) chemotherapy-treated cases with homozygote-associated HR > 3.0 (p < 10-3).

| HEBCS_SNP | Cat. | S1_p_all | S1_p_chemo | R2 | D' | iCOGS_SNP | POSH_SNP | SUCCESS_SNP |
|---|---|---|---|---|---|---|---|---|
| rs10004399 | I | 3.42E-07 | 1.42E-06 | 1 | 1 | rs10004399 | rs10004399 | NA |
| rs10118669 | Ib | 3.99E-05 | 2.43E-02 | 1 | 1 | rs10118669 | rs10118669 | rs7854237 |
| rs11130876 | Ib | 5.59E-05 | 2.58E-03 | 1 | 1 | rs11130876 | rs11130876 | NA |
| rs1603867 | Ib | 2.49E-05 | 4.10E-03 | 0.775 | 0.926 | | | |
| rs6769168 | Ic | 2.03E-05 | 1.33E-03 | 0.739 | 0.889 | | | |
| rs11155012 | III | 1.21E-04 | 6.80E-04 | 1 | 1 | rs11155012 | rs11155012 | rs11155012 |
| rs2182424 | Ib | 1.59E-06 | 2.34E-03 | 1 | 1 | rs12569077 | rs2182424 | rs2182424 |
| rs12693085 | II | 4.13E-02 | 5.13E-06 | 1 | 1 | rs12693085 | rs12693085 | rs12693085 |
| rs13019468 | II | 2.62E-03 | 2.33E-06 | 1 | 1 | rs13019468 | rs13019468 | rs13019468 |
| rs10164900 | II | ns | 5.29E-05 | 0.732 | 0.897 | | | |
| rs11679968 | II | ns | 9.70E-06 | 0.732 | 0.897 | | | |
| rs13063401 | Ia | 1.57E-07 | 1.44E-03 | 1 | 1 | rs13063401 | rs13063401 | rs13063401 |
| rs6791223 | Ia | 1.44E-07 | 1.76E-03 | 0.93 | 1 | | | |
| rs10511205 | Ia | 3.13E-07 | 7.10E-03 | 0.866 | 1 | | | |
| rs13323146 | Ib | 4.17E-05 | 2.37E-04 | 1 | 1 | rs13323146 | rs13323146 | rs13323146 |
| rs10511205 | Ia | 3.13E-07 | 7.10E-03 | 1 | 1 | rs1515534 | rs10511205 | rs10511205 |
| rs6791223 | Ia | 1.44E-07 | 1.76E-03 | 0.931 | 1 | | | |
| rs13063401 | Ia | 1.57E-07 | 1.44E-03 | 0.866 | 1 | | | |
| rs1492872 | Ia | 6.07E-06 | 2.06E-04 | 1 | 1 | rs1602757 | rs1492872 | rs1492872 |
| rs1603867 | Ib | 2.49E-05 | 4.10E-03 | 1 | 1 | rs1603867 | rs1603867 | rs4686025 |
| rs11130876 | Ib | 5.59E-05 | 2.58E-03 | 0.775 | 0.926 | | | |
| rs1359280 | Ia | 4.55E-05 | 2.31E-03 | 1 | 1 | rs1657224 | rs1359280 | rs7071602 |
| rs2477017 | Ia | 4.74E-05 | 1.98E-03 | 0.962 | 1 | | | |
| rs2490806 | Ia | 4.77E-05 | 1.98E-03 | 0.962 | 1 | | | |
| rs16959669 | II | 1.05E-02 | 1.55E-05 | 1 | 1 | rs16959669 | rs16959669 | rs16959669 |
| rs1435737 | II | 1.26E-02 | 1.55E-05 | 1 | 1 | | | |
| rs7179135 | II | 1.03E-02 | 1.55E-05 | 1 | 1 | | | |
| rs17199739 | III | 3.12E-02 | 1.37E-04 | 0.832 | 1 | rs17794824 | rs996418 | rs17199690 |
| rs886108 | III | 3.12E-02 | 1.37E-04 | 0.832 | 1 | | | |
| rs182286 | Ib | 3.79E-05 | 1.36E-03 | 1 | 1 | rs182286 | rs182286 | rs182286 |
| rs6769168 | Ic | 2.03E-05 | 1.33E-03 | 0.806 | 0.961 | | | |
| rs1552329 | Ia | 1.10E-05 | 8.45E-03 | 1 | 1 | rs1893791 | rs1552329 | rs1893791 |
| rs2490806 | Ia | 4.77E-05 | 1.98E-03 | 0.781 | 1 | rs2476995 | rs2490806 | rs2490817 |
| rs2477017 | Ia | 4.74E-05 | 1.98E-03 | 0.781 | 1 | | | |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs1359280 | Ia | 4.55E-05 | 2.31E-03 | 0.752 | 1 | | | |
| rs2670927 | III | 2.22E-02 | 8.66E-04 | 1 | 1 | rs2670927 | NA | rs2670927 |
| rs4891218 | Ia | 3.67E-07 | 7.46E-03 | 0.847 | 0.945 | rs2959164 | rs4891218 | rs2959167 |
| rs4307960 | III | 8.47E-05 | 1.13E-04 | 1 | 1 | rs4307960 | rs4307960 | rs4307960 |
| rs4502225 | Ia | 3.97E-05 | 3.75E-03 | 1 | 1 | rs4502225 | rs4502225 | rs4502225 |
| rs4656879 | Ia | 1.67E-07 | 1.27E-03 | 1 | 1 | rs4656879 | rs4656879 | rs4656879 |
| rs4712523 | Ia | 4.83E-05 | 5.06E-03 | 1 | 1 | rs4712523 | rs4712523 | rs4712523 |
| rs4891218 | Ia | 3.67E-07 | 7.46E-03 | 1 | 1 | rs4891218 | rs4891218 | NA |
| rs9319675 | Ia | 6.10E-07 | 7.71E-03 | 0.8 | 1 | | | |
| rs487989 | III | ns | 1.97E-04 | 0.866 | 1 | rs526502 | rs487989 | NA |
| rs6077999 | II | ns | 8.48E-05 | 1 | 1 | rs6077999 | NA | rs6077999 |
| rs487989 | III | ns | 1.97E-04 | 0.929 | 1 | rs626802 | rs487989 | rs1858783 |
| rs11865240 | Ic | 6.98E-05 | 8.60E-03 | 0.72 | 0.923 | rs6500843 | rs6500843 | rs4786939 |
| rs6750002 | Ia | 1.72E-05 | 3.29E-03 | 1 | 1 | rs6750002 | rs6750002 | rs673811 |
| rs12474326 | Ia | 8.64E-05 | 6.67E-03 | 0.962 | 1 | | | |
| rs7080287 | Ic | 7.48E-05 | 3.28E-04 | 1 | 1 | rs7080287 | NA | rs7080287 |
| rs7821832 | III | 3.84E-04 | 2.08E-04 | 1 | 1 | rs7821832 | rs7821832 | rs7821832 |
| rs6997996 | III | 3.78E-04 | 1.62E-04 | 1 | 1 | | | |
| rs9319675 | Ia | 6.10E-07 | 7.71E-03 | 1 | 1 | rs9319675 | rs9319675 | rs4891220 |
| rs4891218 | Ia | 3.67E-07 | 7.46E-03 | 0.8 | 1 | | | |
| rs964519 | II | ns | 4.24E-08 | 1 | 1 | rs964519 | rs964519 | rs964519 |
| rs6769168 | Ic | 2.03E-05 | 1.33E-03 | 0.967 | 1 | rs9850025 | rs6769168 | rs9850025 |
| rs182286 | Ib | 3.79E-05 | 1.36E-03 | 0.843 | 1 | | | |
| rs11130876 | Ib | 5.59E-05 | 2.58E-03 | 0.771 | 0.893 | | | |
| rs9857979 | III | ns | 4.65E-04 | 1 | 1 | rs9857979 | rs9857979 | NA |
| rs1488688 | Ia | 6.34E-05 | 6.62E-03 | 1 | 1 | rs9889588 | rs1488688 | rs3829957 |
| rs1488688 | Ia | 6.34E-05 | 6.62E-03 | 0.877 | 1 | rs9910285 | rs1488688 | NA |
| rs9932176 | Ia | 8.12E-09 | 7.22E-03 | 1 | 1 | rs9932176 | rs9932176 | rs9932176 |

**Supplementary Table 2:** Tumor histopathological characteristics tabulated according to rs6500843 and rs11155012 genotype.

### a) rs6500843

| | | GG | (%) | AG | (%) | AA | (%) | p-value |
|---|---|---|---|---|---|---|---|---|
| **ER** | | | | | | | | |
| | negative | 1121 | (20.1%) | 1423 | (20.4%) | 458 | (20.8%) | 0.7874 |
| | positive | 4453 | (79.9%) | 5555 | (79.6%) | 1744 | (79.2%) | |
| **PgR** | | | | | | | | |
| | negative | 1535 | (31.6%) | 1879 | (30.7%) | 569 | (29.3%) | 0.1761 |
| | positive | 3323 | (68.4%) | 4250 | (69.3%) | 1371 | (70.7%) | |
| **N (node status)** | | | | | | | | |
| | negative | 3378 | (62.0%) | 4251 | (62.5%) | 1346 | (61.2%) | 0.5354 |
| | positive | 2069 | (38.0%) | 2549 | (37.5%) | 853 | (38.8%) | |
| **Grade** | | | | | | | | |
| | 1 | 1059 | (20.5%) | 1414 | (21.8%) | 438 | (21.6%) | 0.2988 |
| | 2 | 2413 | (46.7%) | 3022 | (46.6%) | 919 | (45.3%) | |
| | 3 | 1692 | (32.8%) | 2049 | (31.6%) | 673 | (33.2%) | |
| **T (tumor size category)[a]** | | | | | | | | |
| | 1 | 3499 | (63.3%) | 4419 | (64.1%) | 1419 | (65.4%) | 0.2325 |
| | 2 | 1791 | (32.4%) | 2156 | (31.3%) | 668 | (30.8%) | |
| | 3 | 236 | (4.3%) | 317 | (4.6%) | 82 | (3.8%) | |
| **HER2** | | | | | | | | |
| | negative | 2588 | (84.8%) | 3290 | (85.2%) | 1061 | (86.6%) | 0.3056 |
| | positive | 465 | (15.2%) | 570 | (14.8%) | 164 | (13.4%) | |

### b) rs11155012

| | | GG | (%) | AG | (%) | AA | (%) | p-value |
|---|---|---|---|---|---|---|---|---|
| **ER** | | | | | | | | |
| | negative | 1649 | (20.8%) | 1142 | (19.9%) | 203 | (19.4%) | 0.3486 |
| | positive | 6285 | (79.2%) | 4587 | (80.1%) | 843 | (80.6%) | |
| **PgR** | | | | | | | | |
| | negative | 2177 | (31.3%) | 1523 | (30.5%) | 268 | (28.9%) | 0.2867 |
| | positive | 4787 | (68.7%) | 3469 | (69.5%) | 660 | (71.1%) | |
| **N (node status)** | | | | | | | | |
| | negative | 4820 | (62.1%) | 3513 | (62.3%) | 611 | (61.0%) | 0.7177 |
| | positive | 2942 | (37.9%) | 2123 | (37.7%) | 391 | (39.0%) | |
| **Grade** | | | | | | | | |
| | 1 | 1558 | (21.2%) | 1114 | (21.0%) | 225 | (23.0%) | 0.4047 |
| | 2 | 3403 | (46.2%) | 2473 | (46.7%) | 464 | (47.3%) | |
| | 3 | 2397 | (32.6%) | 1711 | (32.3%) | 291 | (29.7%) | |
| **T (tumor size category)[a]** | | | | | | | | |
| | 1 | 5011 | (63.8%) | 3675 | (64.6%) | 620 | (61.9%) | 0.2855 |
| | 2 | 2483 | (31.6%) | 1787 | (31.4%) | 332 | (33.1%) | |
| | 3 | 357 | (4.5%) | 228 | (4.0%) | 50 | (5.0%) | |
| **HER2** | | | | | | | | |
| | negative | 3687 | (85.1%) | 2704 | (85.5%) | 519 | (84.8%) | 0.8655 |
| | positive | 644 | (14.9%) | 459 | (14.5%) | 93 | (15.2%) | |

**Supplementary Table 3:** Multivariate Cox proportional hazards model constructed to detect interaction between the SNP rs11155012 and treatment (anthracycline-based adjuvant chemotherapy).

**Model without interaction**

| | HR | 95% C.I. | p |
|---|---|---|---|
| rs11155012 | 1.162 | 1.05 - 1.29 | 3.90E-03 |
| Anthracycline | 0.993 | 0.81 - 1.22 | 9.50E-01 |
| Age | 1.05 | 1.04 - 1.06 | 0.00E+00 |
| Grade | 1.375 | 1.24 - 1.53 | 5.50E-09 |
| ER | 0.726 | 0.61 - 0.86 | 2.70E-04 |
| T | 1.535 | 1.37 - 1.72 | 9.70E-14 |
| N | 1.888 | 1.64 - 2.18 | 0.00E+00 |

*(Likelihood ratio test=526 on 10 df, p=0, n= 8495, number of events= 907)*

**Model with an interaction term (rs11155012:Anthracycline)**

| | HR | 95% C.I. | p |
|---|---|---|---|
| rs11155012 | 1.134 | 1.01 - 1.27 | 3.00E-02 |
| Anthracycline | 0.806 | 0.51 - 1.28 | 3.60E-01 |
| Age | 1.05 | 1.04 - 1.06 | 0.00E+00 |
| Grade | 1.374 | 1.23 - 1.53 | 5.70E-09 |
| ER | 0.728 | 0.61 - 0.86 | 3.00E-04 |
| T | 1.537 | 1.37 - 1.72 | 8.30E-14 |
| N | 1.887 | 1.63 - 2.18 | 0.00E+00 |
| rs11155012:Anthracycline | 1.141 | 0.88 - 1.48 | 3.20E-01 |

*(Likelihood ratio test=526 on 11 df, p=0, n= 8495, number of events= 907)*

Likelihood ratio test between models: chisq=0.98, df=1, p = 0.3222

**Supplementary Table 4:** Complete eQTL analysis results for all SNPs in the a) rs6500843 and b) rs11155012 locus. SNPs with r2 > 0.1 were included in the analysis.

**a) SNPs in the rs6500843 region**

| SNP | Chr | Pos | rsq | D' | probe | gene | g_chr | gene_pos | p_value | |
|---|---|---|---|---|---|---|---|---|---|---|
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1806818 | MCM3 | 6 | 52237346 | 5.94E-08 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2368718 | CENPM | 22 | 40664777 | 1.36E-07 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1668814 | CENPM | 22 | 40669561 | 5.13E-07 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1766658 | PKMYT1 | 16 | 2964552 | 1.25E-06 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1680955 | AURKA | 20 | 54378046 | 1.37E-06 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2222008 | KIFC1 | 6 | 33482588 | 1.59E-06 | * |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1806037 | TK1 | 0 | 73682076 | 1.70E-06 | * |
| rs7500396 | 16 | 6843128 | 0.104 | 0.358 | ILMN_1787237 | RELT | 11 | 72765380 | 2.30E-06 | * |
| rs11648981 | 16 | 6804820 | 0.255 | 0.531 | ILMN_1709484 | BLM | 15 | 89155580 | 2.93E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2265654 | UBE2C | 20 | 43877439 | 3.14E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2357438 | AURKA | 20 | 54378054 | 3.28E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2368721 | CENPM | 22 | 40672375 | 3.37E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2401436 | PKMYT1 | 16 | 2964004 | 3.66E-06 | |
| rs11648981 | 16 | 6804820 | 0.255 | 0.531 | ILMN_1673721 | EXO1 | 1 | 240119417 | 3.80E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1728934 | PRC1 | 15 | 89310505 | 3.86E-06 | |
| rs12446260 | 16 | 6748770 | 0.101 | 0.504 | ILMN_1681802 | GRK6 | 5 | 176800596 | 4.05E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1782743 | LETMD1 | 12 | 49739813 | 4.14E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_2344971 | FOXM1 | 12 | 2837420 | 4.19E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1678300 | KPNA2 | 17 | 1716044 | 4.35E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1740291 | POLQ | 3 | 122633050 | 4.48E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1716400 | FOXM1 | 12 | 2837461 | 4.74E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1665737 | UFD1L | 22 | 17817951 | 4.87E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1675266 | NAGPA | 16 | 5015039 | 6.18E-06 | |
| rs12445513 | 16 | 6843082 | 0.118 | 0.47 | ILMN_1729734 | MFSD4 | 1 | 203837106 | 6.27E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1810550 | BAIAP3 | 16 | 1339189 | 6.49E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1803124 | BIRC5 | 17 | 73733114 | 6.55E-06 | |
| rs6500842 | 16 | 6808287 | 0.541 | 0.87 | ILMN_1798654 | MCM6 | 2 | 136314638 | 6.59E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1786065 | UHRF1 | 19 | 4912680 | 6.96E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_2395236 | CHEK2 | 22 | 27420025 | 8.06E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1776490 | C17orf53 | 17 | 39595293 | 8.27E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1719232 | DGCR14 | 22 | 17502578 | 8.43E-06 | |
| rs7205424 | 16 | 6805109 | 0.255 | 0.531 | ILMN_1683450 | CDCA5 | 11 | 64601683 | 9.00E-06 | |
| rs8060556 | 16 | 6808512 | 0.215 | 1 | ILMN_1771852 | ZNF575 | 19 | 48731821 | 9.30E-06 | |

**b) SNPs in the rs11155012 region**

| SNP | Chr | Pos | rsq | D' | probe | gene | g_chr | gene_pos | p_value | |
|---|---|---|---|---|---|---|---|---|---|---|
| rs911081 | 6 | 139171906 | 0.123 | 0.857 | ILMN_2367157 | CHRFAM7A | 15 | 28459848 | 2.21E-08 | * |
| rs9495127 | 6 | 138918259 | 0.129 | 1 | ILMN_1760708 | CRYBB2 | 22 | 23955472 | 2.30E-07 | * |
| rs7742202 | 6 | 139115973 | 0.11 | 0.627 | ILMN_1688218 | RASGRP4 | 19 | 43602502 | 5.51E-07 | * |
| rs9495127 | 6 | 138918259 | 0.129 | 1 | ILMN_2323801 | MOCS1 | 6 | 39981286 | 5.60E-07 | * |
| rs6570277 | 6 | 139119982 | 0.119 | 0.641 | ILMN_1688218 | RASGRP4 | 19 | 43602502 | 6.50E-07 | * |
| rs9495127 | 6 | 138918259 | 0.129 | 1 | ILMN_1783185 | SOX21 | 13 | 94160237 | 7.02E-07 | * |
| rs9495127 | 6 | 138918259 | 0.129 | 1 | ILMN_2353276 | UPK3B | 7 | 75978966 | 1.65E-06 | * |
| rs9321678 | 6 | 139196016 | 0.147 | 1 | ILMN_2367157 | CHRFAM7A | 15 | 28459848 | 1.80E-06 | * |
| rs4321841 | 6 | 139118463 | 0.103 | 0.53 | ILMN_1732198 | UTS2 | 1 | 7836091 | 3.14E-06 | |
| rs4321841 | 6 | 139118463 | 0.103 | 0.53 | ILMN_1671818 | UTS2 | 1 | 7833477 | 3.42E-06 | |
| rs2698744 | 6 | 139227390 | 0.111 | 0.643 | ILMN_2062381 | LCOR | 10 | 98708585 | 6.17E-06 | |
| rs4538729 | 6 | 139117034 | 0.152 | 0.581 | ILMN_1671818 | UTS2 | 1 | 7833477 | 8.01E-06 | |

**Supplementary Table 5**: *Kaplan-Meier Plotter* survival analysis of statistically significant trans-eQTL genes associated with the rs6500843 and rs11155012 loci. The end point of the analysis is 10-year relapse-free survival, and the analysis was performed among cases treated with adjuvant chemotherapy (n=425), cases not treated with systemic therapy (n=1877) and all cases irrespective of treatment (n = 3455). To detect genes with potential interaction with treatment, tests for heterogeneity have been carried out between treated and untreated cases for each gene.

| SNP | Gene | HR | 95% C.I. | p-value | p(het)* |
|---|---|---|---|---|---|
| **Chemotherapy-treated cases (n = 425)** | | | | | |
| rs6500843 | *MCM3* | 1.43 | 0.99 - 2.06 | 0.058 | *ns* |
| rs6500843 | *CENPM* | 2.04 | 1.27 - 3.30 | 0.0027 | *ns* |
| rs6500843 | *PKMYT1* | 1.81 | 1.20 - 2.73 | 0.0044 | *ns* |
| rs6500843 | *AURKA* | 1.25 | 0.89 - 1.77 | 0.2 | 0.0209 |
| rs6500843 | *KIFC1* | 1.91 | 1.30 - 2.81 | 0.00077 | *ns* |
| rs6500843 | *TK1* | 1.58 | 1.10 - 2.27 | 0.013 | *ns* |
| rs11155012 | *CHRFAM7A* | 0.77 | 0.54 - 1.09 | 0.13 | *ns* |
| rs11155012 | *CRYBB2* | 1.18 | 0.82 - 1.70 | 0.37 | *ns* |
| rs11155012 | *RASGRP4* | *na* | *na* | *na* | *n/a* |
| rs11155012 | *MOCS1* | 1.36 | 0.91 - 2.05 | 0.13 | *ns* |
| rs11155012 | *SOX21* | 1.45 | 1.02 - 2.06 | 0.039 | 0.0239 |
| **Cases without adjuvant chemotherapy (n = 1877)** | | | | | |
| rs6500843 | *MCM3* | 1.46 | 1.23 - 1.74 | 1.20E-05 | *ns* |
| rs6500843 | *CENPM* | 1.91 | 1.61 - 2.27 | 5.90E-14 | *ns* |
| rs6500843 | *PKMYT1* | 1.65 | 1.38 - 1.96 | 2.10E-08 | *ns* |
| rs6500843 | *AURKA* | 2.34 | 1.91 - 2.88 | 0.00E+00 | 0.0209 |
| rs6500843 | *KIFC1* | 2.02 | 1.70 - 2,4 | 3.30E-16 | *ns* |
| rs6500843 | *TK1* | 1.79 | 1.50 - 2.12 | 1.90E-11 | *ns* |
| rs11155012 | *CHRFAM7A* | 0.85 | 0.70 - 1.03 | 0.093 | *ns* |
| rs11155012 | *CRYBB2* | 0.79 | 0.67 - 0.95 | 0.0097 | *ns* |
| rs11155012 | *RASGRP4* | *na* | *na* | *na* | *n/a* |
| rs11155012 | *MOCS1* | 0.79 | 0.66 - 0.94 | 0.0089 | *ns* |
| rs11155012 | *SOX21* | 0.82 | 0.69 - 0.98 | 0.029 | 0.0239 |
| **All cases (n = 3455)** | | | | | |
| rs6500843 | *MCM3* | 1.31 | 1.16 - 1.49 | 2.60E-05 | - |
| rs6500843 | *CENPM* | 1.72 | 1.50 - 1.98 | 6.80E-15 | - |
| rs6500843 | *PKMYT1* | 1.34 | 1.18 - 1.52 | 5.90E-06 | - |
| rs6500843 | *AURKA* | 2.44 | 2.06 - 2.89 | 0 | - |
| rs6500843 | *KIFC1* | 1.69 | 1.49 - 1.91 | 0 | - |
| rs6500843 | *TK1* | 1.55 | 1.38 - 1.75 | 7.10E-13 | - |
| rs11155012 | *CHRFAM7A* | 0.85 | 0.73 - 0.97 | 0.018 | - |
| rs11155012 | *CRYBB2* | 0.66 | 0.58 - 0.75 | 2.50E-11 | - |
| rs11155012 | *RASGRP4* | *na* | *na* | *na* | - |
| rs11155012 | *MOCS1* | 0.61 | 0.54 - 0.69 | 8.90E-16 | - |
| rs11155012 | *SOX21* | 0.62 | 0.55 - 0.70 | 3.40E-14 | - |

* Z-test for heterogeneity between treated and untreated, BH-adjusted for multiple testing

**Supplementary Table 6:** Summary of the HaploReg analysis results for the rs11155012 locus. The rs11155012 SNP is shown in yellow. Linked promoter (green) and enhancer (orange) sites have been marked in the table.

| rsID | chr | pos | r2 | D' | Promoter_ENCODE | Enhancer_ENCODE | DNAse_evidence | Protein_binding_evidence | Binding_motif_evidence | GENCODE_name |
|---|---|---|---|---|---|---|---|---|---|---|
| rs13209413 | 6 | 139050249 | 0.2 | 0.5 | . | . | . | . | . | RP11-390P2.4 |
| rs10457675 | 6 | 139050948 | 0.2 | 0.51 | . | . | . | . | Yes | RP11-390P2.4 |
| rs10457676 | 6 | 139051550 | 0.2 | 0.51 | . | . | . | . | Yes | RP11-390P2.4 |
| rs5880399 | 6 | 139054508 | 0.2 | 0.52 | . | . | . | . | Yes | RP11-390P2.4 |
| rs6938663 | 6 | 139055240 | 0.2 | 0.52 | . | . | . | . | Yes | RP11-390P2.4 |
| rs10457677 | 6 | 139055546 | 0.2 | 0.52 | . | . | . | . | Yes | RP11-390P2.4 |
| rs12206431 | 6 | 139056090 | 0.2 | 0.52 | . | . | . | . | Yes | RP11-390P2.4 |
| rs12194617 | 6 | 139063878 | 0.3 | 0.53 | . | . | . | . | Yes | CCDC28A |
| rs12200534 | 6 | 139069323 | 0.2 | 0.52 | . | . | . | . | Yes | CCDC28A |
| rs139233693 | 6 | 139071974 | 0.2 | 0.64 | . | . | . | . | Yes | CCDC28A |
| rs73774898 | 6 | 139072232 | 0.3 | 0.65 | . | . | . | . | Yes | CCDC28A |
| rs12194832 | 6 | 139073407 | 0.3 | 0.66 | . | . | Yes | Yes | Yes | CCDC28A |
| rs201825583 | 6 | 139089068 | 0.3 | 0.69 | . | Huvec,7_Weak_Enhancer | . | . | Yes | CCDC28A |
| rs200050818 | 6 | 139089069 | 0.3 | 0.69 | . | Huvec,7_Weak_Enhancer | . | . | Yes | CCDC28A |
| rs11154998 | 6 | 139089070 | 0.3 | 0.69 | . | Huvec,7_Weak_Enhancer | . | . | Yes | CCDC28A |
| rs12210412 | 6 | 139091791 | 0.3 | 0.7 | . | . | . | . | Yes | CCDC28A |
| rs11155000 | 6 | 139099401 | 0.3 | 0.7 | . | . | . | . | Yes | CCDC28A |
| rs7753418 | 6 | 139104107 | 0.2 | 0.57 | . | . | . | . | . | CCDC28A |
| rs6570281 | 6 | 139105519 | 0.2 | 0.53 | . | . | . | . | Yes | CCDC28A |
| rs7772915 | 6 | 139105637 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs7773593 | 6 | 139105923 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs3763185 | 6 | 139106599 | 0.3 | 0.71 | . | . | . | . | Yes | CCDC28A |
| rs3763186 | 6 | 139106722 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs3763187 | 6 | 139106787 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs7746303 | 6 | 139108026 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs7751745 | 6 | 139109125 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |

| rsID | chr | pos | r2 | D' | Promoter_ENCODE | Enhancer_ENCODE | DNAse_evidence | Protein_binding_evidence | Binding_motif_evidence | GENCODE_name |
|---|---|---|---|---|---|---|---|---|---|---|
| rs2327900 | 6 | 139109784 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs2327901 | 6 | 139110033 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs11155003 | 6 | 139110345 | 0.2 | 0.54 | . | . | . | . | . | CCDC28A |
| rs9495249 | 6 | 139110841 | 0.2 | 0.53 | . | . | . | . | Yes | CCDC28A |
| rs9495252 | 6 | 139111372 | 0.2 | 0.54 | . | . | . | . | Yes | CCDC28A |
| rs12205733 | 6 | 139112228 | 0.3 | 0.71 | . | . | . | . | Yes | CCDC28A |
| rs7773485 | 6 | 139113098 | 0.2 | 0.58 | . | . | . | . | Yes | CCDC28A |
| rs9321673 | 6 | 139113392 | 0.2 | 0.58 | . | . | . | . | Yes | CCDC28A |
| rs34409315 | 6 | 139113636 | 0.2 | 0.58 | . | . | . | . | Yes | CCDC28A |
| rs139961983 | 6 | 139113869 | 0.3 | 0.71 | . | . | Yes | . | Yes | CCDC28A |
| rs201682274 | 6 | 139114731 | 0.2 | 0.57 | . | . | . | . | Yes | CCDC28A |
| rs2327902 | 6 | 139114732 | 0.2 | 0.61 | . | . | . | . | Yes | CCDC28A |
| rs5880402 | 6 | 139115297 | 0.2 | 0.57 | . | . | . | . | Yes | CCDC28A |
| rs1474962 | 6 | 139115518 | 0.2 | 0.58 | . | . | . | . | . | CCDC28A |
| rs1474963 | 6 | 139115882 | 0.2 | 0.58 | . | . | . | . | Yes | ECT2L |
| rs12198968 | 6 | 139120627 | 0.5 | 0.75 | . | . | . | . | Yes | ECT2L |
| rs79981479 | 6 | 139120787 | 0.5 | 0.77 | . | . | . | . | Yes | ECT2L |
| rs10457678 | 6 | 139122240 | 0.5 | 0.77 | . | . | Yes | Yes | . | ECT2L |
| rs9321674 | 6 | 139122341 | 0.5 | 0.77 | . | . | . | . | Yes | ECT2L |
| rs12664876 | 6 | 139132840 | 0.4 | 0.73 | . | . | . | . | Yes | ECT2L |
| rs9403003 | 6 | 139134041 | 0.3 | 0.58 | . | . | . | . | Yes | ECT2L |
| rs746865 | 6 | 139134103 | 0.3 | 0.58 | . | . | . | . | Yes | ECT2L |
| rs746864 | 6 | 139134112 | 0.3 | 0.58 | . | . | . | . | Yes | ECT2L |
| rs4895548 | 6 | 139134754 | 0.2 | -1 | . | HepG2,4_Strong_Enhancer | . | . | Yes | ECT2L |
| rs1321356 | 6 | 139134978 | 0.2 | -1 | . | HepG2,4_Strong_Enhancer;HMEC,7_Weak_Enhancer;NHEK,7_Weak_Enhancer | . | . | . | ECT2L |
| rs9389619 | 6 | 139135077 | 0.7 | 0.97 | . | HepG2,4_Strong_Enhancer;HMEC,7_Weak_Enhancer;NHEK,7_Weak_Enhancer | . | . | Yes | ECT2L |

| **rsID** | **chr** | **pos** | **r2** | **D'** | **Promoter_ENCODE** | **Enhancer_ENCODE** | **DNAse_evidence** | **Protein_binding_evidence** | **Binding_motif_evidence** | **GENCODE_name** |
|---|---|---|---|---|---|---|---|---|---|---|
| rs9403004 | 6 | 139135392 | 0.2 | -0.9 | . | HepG2,4_Strong_Enhancer;NHEK,7_Weak_Enhancer;Huvec,5_Strong_Enhancer;HMEC,6_Weak_Enhancer | Yes | Yes | Yes | ECT2L |
| rs9376372 | 6 | 139135990 | 0.2 | -1 | . | HepG2,4_Strong_Enhancer;NHEK,7_Weak_Enhancer;HMEC,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs11155008 | 6 | 139137500 | 1 | 0.99 | . | NHEK,7_Weak_Enhancer;HMEC,5_Strong_Enhancer | Yes | Yes | Yes | ECT2L |
| rs11155009 | 6 | 139137778 | 1 | 1 | . | HMEC,5_Strong_Enhancer;NHEK,5_Strong_Enhancer | . | . | Yes | ECT2L |
| rs4896407 | 6 | 139137868 | 0.2 | -0.9 | . | HMEC,5_Strong_Enhancer;NHEK,5_Strong_Enhancer | . | . | Yes | ECT2L |
| rs6570288 | 6 | 139139589 | 0.2 | -1 | . | Huvec,5_Strong_Enhancer;HepG2,6_Weak_Enhancer;GM12878,6_Weak_Enhancer;NHEK,4_Strong_Enhancer;HSMM,7_Weak_Enhancer;H1,7_Weak_Enhancer;HMEC,4_Strong_Enhancer | . | . | Yes | ECT2L |
| rs12055486 | 6 | 139140446 | 0.3 | -1 | . | Huvec,6_Weak_Enhancer;GM12878,6_Weak_Enhancer;HMEC,5_Strong_Enhancer;NHEK,5_Strong_Enhancer | Yes | . | Yes | ECT2L |
| rs35219429 | 6 | 139140679 | 1 | 1 | . | Huvec,6_Weak_Enhancer;GM12878,6_Weak_Enhancer;HMEC,5_Strong_Enhancer;NHEK,5_Strong_Enhancer | Yes | . | Yes | ECT2L |
| rs6570290 | 6 | 139140891 | 0.6 | 0.98 | . | NHEK,5_Strong_Enhancer;HMEC,7_Weak_Enhancer;GM12878,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs9321676 | 6 | 139141089 | 0.2 | 1 | . | NHEK,5_Strong_Enhancer;GM12878,7_Weak_Enhancer;HMEC,5_Strong_Enhancer | . | . | Yes | ECT2L |
| rs9321677 | 6 | 139141137 | 0.2 | 1 | . | NHEK,5_Strong_Enhancer;GM12878,7_Weak_Enhancer;HMEC,5_Strong_Enhancer;HSMM,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs9389621 | 6 | 139141801 | 0.3 | -1 | . | NHEK,5_Strong_Enhancer;GM12878,7_Weak_Enhancer;HMEC,5_Strong_Enhancer | . | . | Yes | ECT2L |
| rs11155010 | 6 | 139142400 | 0.6 | 0.99 | HepG2,3_Poised_Promoter | GM12878,7_Weak_Enhancer;HMEC,5_Strong_Enhancer;NHEK,7_Weak_Enhancer | Yes | . | Yes | ECT2L |

| rsID | chr | pos | r2 | D' | Promoter_ENCODE | Enhancer_ENCODE | DNAse_evidence | Protein_binding_evidence | Binding_motif_evidence | GENCODE_name |
|---|---|---|---|---|---|---|---|---|---|---|
| rs12193938 | 6 | 139142855 | 1 | 1 | HepG2,2_Weak_Promoter | HMEC,7_Weak_Enhancer | Yes | Yes | Yes | ECT2L |
| rs12193993 | 6 | 139142909 | 1 | 1 | HepG2,2_Weak_Promoter | . | Yes | Yes | Yes | ECT2L |
| rs998035 | 6 | 139142941 | 0.6 | 0.99 | HepG2,2_Weak_Promoter | . | Yes | Yes | Yes | ECT2L |
| rs7773177 | 6 | 139143088 | 1 | 1 | HepG2,2_Weak_Promoter | . | . | . | Yes | ECT2L |
| rs6570291 | 6 | 139143274 | 1 | 1 | HepG2,2_Weak_Promoter | . | . | . | Yes | ECT2L |
| rs10872493 | 6 | 139147421 | 0.6 | 0.98 | . | . | . | . | Yes | ECT2L |
| rs10872494 | 6 | 139148135 | 0.6 | 0.96 | . | . | . | . | Yes | ECT2L |
| rs11155012 | 6 | 139151784 | 1 | 1 | . | . | . | . | Yes | ECT2L |
| rs11155013 | 6 | 139151838 | 0.9 | 0.99 | . | . | . | . | Yes | ECT2L |
| rs55747302 | 6 | 139153882 | 0.6 | 0.97 | . | . | . | . | Yes | ECT2L |
| rs9389625 | 6 | 139154056 | 0.4 | 0.98 | . | . | . | . | Yes | ECT2L |
| rs12205070 | 6 | 139154247 | 1 | 1 | . | . | . | . | Yes | ECT2L |
| rs10872495 | 6 | 139155677 | 1 | 0.99 | . | . | . | . | Yes | ECT2L |
| rs1529152 | 6 | 139155855 | 0.3 | -1 | . | . | Yes | . | Yes | ECT2L |
| rs9285487 | 6 | 139156267 | 0.3 | -1 | . | . | . | . | Yes | ECT2L |
| rs34665665 | 6 | 139156282 | 1 | 0.99 | . | . | . | . | Yes | ECT2L |
| rs6916561 | 6 | 139157259 | 0.3 | -1 | . | . | . | . | Yes | ECT2L |
| rs12207788 | 6 | 139157314 | 1 | 0.99 | . | . | . | . | Yes | ECT2L |
| rs71766787 | 6 | 139157807 | 0.3 | -1 | . | . | Yes | . | Yes | ECT2L |
| rs4896408 | 6 | 139158129 | 0.3 | -1 | . | . | Yes | . | Yes | ECT2L |
| rs4896409 | 6 | 139158192 | 0.3 | -1 | . | . | Yes | . | Yes | ECT2L |
| rs10872496 | 6 | 139158676 | 1 | 0.99 | . | . | Yes | . | Yes | ECT2L |
| rs3886649 | 6 | 139158791 | 0.3 | -1 | . | . | . | . | Yes | ECT2L |
| rs2017360 | 6 | 139159206 | 0.3 | -1 | . | . | . | . | Yes | ECT2L |
| rs9321680 | 6 | 139159382 | 0.3 | -1 | . | . | . | . | Yes | ECT2L |

| rsID | chr | pos | r2 | D' | Promoter_ENCODE | Enhancer_ENCODE | DNAse_evidence | Protein_binding_evidence | Binding_motif_evidence | GENCODE_name |
|---|---|---|---|---|---|---|---|---|---|---|
| rs9385842 | 6 | 139178400 | 0.3 | 0.66 | . | . | . | . | Yes | ECT2L |
| rs6899686 | 6 | 139179061 | 0.3 | 0.67 | . | . | Yes | Yes | Yes | ECT2L |
| rs11155014 | 6 | 139180152 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs10872497 | 6 | 139180200 | 0.3 | 0.65 | . | . | . | . | Yes | U6 |
| rs10872498 | 6 | 139180508 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs10457679 | 6 | 139180868 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs71754397 | 6 | 139181653 | 0.3 | 0.64 | . | . | . | . | Yes | ECT2L |
| rs11155015 | 6 | 139182263 | 0.2 | 0.51 | . | . | . | . | . | ECT2L |
| rs11155016 | 6 | 139182265 | 0.3 | 0.51 | . | . | . | . | Yes | ECT2L |
| rs2038444 | 6 | 139182617 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs2038445 | 6 | 139182742 | 0.3 | 0.64 | . | . | . | . | Yes | ECT2L |
| rs2038446 | 6 | 139182979 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs12204850 | 6 | 139183879 | 0.3 | 0.65 | . | HSMM,7_Weak_Enhancer | Yes | . | Yes | ECT2L |
| rs12205077 | 6 | 139184362 | 0.3 | 0.65 | . | HSMM,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs12205026 | 6 | 139184382 | 0.3 | 0.65 | . | HSMM,7_Weak_Enhancer | Yes | . | Yes | ECT2L |
| rs4896412 | 6 | 139184478 | 0.3 | 0.65 | . | HSMM,7_Weak_Enhancer | Yes | . | Yes | ECT2L |
| rs4896413 | 6 | 139184640 | 0.3 | 0.65 | . | HSMM,6_Weak_Enhancer | Yes | Yes | Yes | ECT2L |
| rs12211862 | 6 | 139185884 | 0.3 | 0.65 | . | HSMM,5_Strong_Enhancer | Yes | . | Yes | ECT2L |
| rs12198757 | 6 | 139186457 | 0.3 | 0.65 | . | HSMM,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs12200274 | 6 | 139186751 | 0.3 | 0.65 | . | . | . | . | . | ECT2L |
| rs12200387 | 6 | 139186961 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs12213988 | 6 | 139187231 | 0.3 | 0.63 | . | . | . | . | Yes | ECT2L |
| rs12215577 | 6 | 139187656 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs12215681 | 6 | 139187866 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs10457680 | 6 | 139190959 | 0.3 | 0.65 | . | NHEK,6_Weak_Enhancer;HMEC,7_Weak_Enhancer;GM12878,5_Strong_Enhancer | Yes | . | Yes | ECT2L |
| rs11155017 | 6 | 139192425 | 0.3 | 0.65 | . | GM12878,7_Weak_Enhancer | . | . | . | ECT2L |
| rs9495277 | 6 | 139193041 | 0.3 | 0.65 | . | GM12878,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs10457681 | 6 | 139193565 | 0.3 | 0.65 | . | GM12878,7_Weak_Enhancer | Yes | . | Yes | ECT2L |

| rsID | chr | pos | r2 | D' | Promoter_ENCODE | Enhancer_ENCODE | DNAse_evidence | Protein_binding_evidence | Binding_motif_evidence | GENCODE_name |
|---|---|---|---|---|---|---|---|---|---|---|
| rs10872499 | 6 | 139193603 | 0.3 | 0.65 | . | GM12878,7_Weak_Enhancer | Yes | . | Yes | ECT2L |
| rs10872500 | 6 | 139193673 | 0.3 | 0.65 | . | GM12878,7_Weak_Enhancer | . | . | Yes | ECT2L |
| rs9484210 | 6 | 139194792 | 0.3 | 0.65 | . | . | . | . | Yes | ECT2L |
| rs1157388 | 6 | 139195283 | 0.3 | 0.64 | . | . | Yes | . | Yes | ECT2L |

**Supplementary Table 7:** Breast Cancer Association Consortium (BCAC) studies genotyped on the iCOGS chip and represented in this work.

| Study Acronym | Study | Country | Cases[a] | Events[a] | Study design |
|---|---|---|---|---|---|
| ABCFS | Australian Breast Cancer Family Study | Australia | 671 | 160 | 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. |
| ABCS | Amsterdam Breast Cancer Study | Netherlands | 443 | 60 | Breast cancer patients diagnosed before age 50 in 2003-2009 at the NKI-AVL; and (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. |
| BBCC | Bavarian Breast Cancer Cases and Controls | Germany | 248 | 16 | Consecutive, unselected cases with invasive breast cancer recruited at the University Breast Centre, Franconia in Northern Bavaria from 2002-2010. |
| HEBCS | Helsinki Breast Cancer Study | Finland | 1451 | 199 | (1) Consecutive cases (883) from the Department of Oncology, Helsinki University Hospital 1997-8 and 2000, (2) Consecutive cases (986) from the Department of Surgery, Helsinki University Hospital 2001–2004, (3) Familial breast cancer patients (536) from the Helsinki University Hospital, Departments of Oncology and Clinical Genetics (1995-). |
| KARBAC | Karolinska Breast Cancer  Study | Sweden | 409 | 106 | 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. |
| KBCP | Kuopio Breast Cancer Project | Finland | 410 | 154 | 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. |
| kConFab/AOCS | Kathleen Cuningham Foundation Consortium for Research into Familial Breast Cancer / Australian Ovarian Cancer Study | Australia | 183 | 16 | 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. |
| LMBC | Leuven Multidisciplinary Breast Centre | Belgium | 2074 | 67 | All patients diagnosed with breast cancer and seen in the Multidisciplinary Breast Centre in Leuven (Gashuisberg) since June 2007 plus retrospective collection of cases diagnosed since 2000. |
| MARIE | Mammary Carcinoma Risk Factor Investigation | Germany | 1635 | 244 | 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. |

| Study Acronym | Study | Country | Cases[a] | Events[a] | Study design |
|---|---|---|---|---|---|
| MCBCS | Mayo Clinic Breast Cancer Study | USA | 1198 | 100 | Incident cases residing in 6 states (MN, WI, IA, IL, ND, SD) seen at the Mayo Clinic in Rochester, MN from 2002-5 |
| OFBCR | Ontario Familial Breast Cancer Registry | Canada | 686 | 80 | 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. |
| PBCS | NCI Polish Breast Cancer Study | Poland | 389 | 35 | Incident cases from 2000-2003 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. |
| RBCS | Rotterdam Breast Cancer Study | Netherlands | 529 | 84 | Familial breast cancer patients selected from the clinical genetics centre at Erasmus Medical Centre between 1994-2005. |
| SASBAC | Singapore and Sweden Breast Cancer Study | Sweden | 1146 | 155 | Women diagnosed in Sweden aged 50-74 in 1993-1995. |
| SEARCH | Study of Epidemiology & Risk Factors in Cancer Heredity | UK | 6233 | 721 | Identified through the Eastern Cancer Registration and Information Centre: (I) prevalent Cases; diagnosed 1991-1996; under 55 years of age at diagnosis; recruited 1996-2002 (ii) incidence cases; diagnosed since 1996; under 70 years of age at diagnosis; recruited 1996-present. |
| SKKDKFZS[b] | Städtisches Klinikum Karlsruhe Deutsches Krebsforschungszentrum Study | Germany | 123 | 21 | Women diagnosed with primary in situ or invasive breast cancer at the Städtisches Klinikum Karlsruhe from March 1993 to July 2005. Cases were 21-93 years of age. |

[a] Counts indicate cases eligible for the current study (treatment and follow-up information available, follow-up time > recruitment latency).

[b] While SKKDKFZS is a BCAC study, this data set was genotyped as part of the Triple Negative Consortium (TNBCC).