Supplementary Material

Supplementary Methods

Study descriptions

Amsterdam Breast Cancer Study (ABCS): 67 triple negative breast cancer cases from the Netherlands Cancer Insitute – Antoni van Leeuwenhoek hospital were included. Breast cancer cases were obtained from a hospital-based consecutive case series of operable cases with invasive mammary carcinoma aged <50 years period 1995-2002 and enriched with familial breast cancer cases of all ages, counseled from 1995 to 2007.

Australia Breast Cancer Tissue Bank (ABCTB): 166 triple negative breast cancer cases from the ABCTB tumor bank were included. Breast cancer cases were collected from six hospitals in New South Wales, Australia: Royal Prince Alfred Hospital, Westmead Hospital, Royal North Shore Hospital, St. Vincent's Hospital, Hunter Area Hospitals, and Port Macquarie beginning in 2006.

Bavarian Breast Cancer Cases and Controls (BBCC): 332 triple negative breast cancer cases were included from this consecutive series of cases with invasive breast cancer recruited at the University Breast Centre, Franconia in Northern Bavaria, Germany from 2002-2006. Cases were between 22-96 years of age. Controls were population-based unaffected women from the same geographical area.

British Breast Cancer Study (BBCS): 57 triple negative breast cancer cases and 63 controls from this study of cancer registry and National Cancer Research network

(NCRN) based cases were included. Cases were identified from English & Scottish Cancer Registries and oncological departments, including all breast cancer cases who developed a first primary before age 65 in 1971 or later and who subsequently developed a second primary cancer and unilateral breast cancer cases diagnosed before age 70 in 1971 or later. Cases were between 24-76 years of age. Population-based controls were identified as a friend, sister-in-law, daughter-in-law or other non-blood relative of cases.

Breast Cancer in Galway Genetic Study (BIGGS): 29 triple negative cases and 67 controls were included from this study of hospital-based breast cancer cases and population-based controls. Cases were unselected breast cancers recruited from University College Hospital Galway and surrounding hospitals in the West of Ireland since 2001 with an age range of 24-90 years. Controls were 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 (same catchment area as cases) during the period 2001-2008.

Cancer Genetic Markers of Susceptibility (CGEMS): 1,126 controls were included in this study. The Nurses' Health Study (NHS) is a longitudinal study of 121,700 women enrolled in 1976. The CGEMS nested case-control study is derived from 32,826 participants who provided a blood sample between 1989 and 1990 and were free of diagnosed breast cancer at blood collection and followed for incident disease until June 1, 2004. Controls were not diagnosed with breast cancer during follow-up, and were matched to cases based on age at diagnosis, blood collection variables (time of day,

season, and year of blood collection, as well as recent (<3 months) use of postmenopausal hormones), ethnicity (all cases and controls are self-reported Caucasians), and menopausal status (all cases were postmenopausal at diagnosis) [8].

DEMOKRITOS: : 273 triple negative breast cancer cases from an unselected breast tumor series and 85 regional controls in Athens, Greece were included in this study. Cases were enrolled from 1997 until 2010 in several major hospitals covering most geographical areas of Greece, such as Athens metropolitan area, Thessaloniki, Ioannina, Patras, and Crete (Chania), in collaboration with the Hellenic Cooperative Oncology Group (HECOG). Cases had an age range of 20-87 years. Controls were population-based unaffected women of the same age range.

Dana Farber Cancer Institute (DFCI): 300 triple negative breast cancers with available DNA from residual bloods from an unselected series of breast tumors from the Dana Farber Cancer Institute were included in this study.

Fox Chase Cancer Center (FCCC): Peripheral blood DNA from 159 triple negative breast cancer cases and 159 unaffected matched controls from the Fox Chase Cancer Center were included in this study. Cases were between 28-80 years of age at diagnosis. Comprehensive clinical data including histology, staging, treatment and outcomes was provided for all cases. Controls were healthy females with no personal cancer history matched geographically and by gender, race and age.

Gene Environment Interaction and Breast Cancer in Germany (GENICA): 60 triple negative cases and 64 controls from this population-based case control study of breast cancer in the Greater Bonn area of Germany were included. Cases were incident breast cancer cases enrolled between 2000 and 2004 from the Greater Bonn area (by of the hospitals within the study region), all of which were enrolled within 6 months of diagnosis. Cases were between 23-80 years of age. Controls were selected from population registries from 31 communities in the greater Bonn area and matched to cases in 5-year age classes between 2001 and 2004.

Helsinki Breast Cancer Study (HEBCS): 85 triple negative cases from this hospital-based case-control study in Southern Finland were included in this study. Cases were consecutive breast cancer cases from the 1) Department of Oncology, Helsinki University Central Hospital 1997-8 and 2000, 2) consecutive cases from the Department of Surgery, Helsinki University Central Hospital 2001 – 2004, or 3) Familial breast cancer patients from the Helsinki University Central Hospital, Departments of Oncology and Clinical Genetics (from 1995). Cases were between 22 and 96 years of age. The population allele and genotype frequencies were obtained from the Finnish Genome Centre on 221 healthy population controls in the NordicDB, a Nordic pool and portal for genome-wide control data [25].

Karolinska Breast Cancer Study (KARBAC): 27 triple negative cases and 26 controls from this Swedish case-control study consisting of population- and hospital-based cases and geographically matched controls were included in these analyses. Cases were

consecutive cases from Department of Oncology, Huddinge & Söder Hospital, Stockholm collected between 1998-2000. Controls were blood donors of mixed gender from the same geographical region. Excess material was received from all blood donors over a three month period in 2004 (approximately 3000) and DNA was extracted from a random sample of 1500 subjects.

Kuopio Breast Cancer Project (KBCP): 34 triple negative breast cancers were included from this hospital-based prospective clinical cohort were included. Cases were women in the cohort seen at Kuopio University Hospital between 1990 and 1995 because of breast lump, mammographic abnormality, or other breast symptom and who were found to have breast cancer.

Cooperative Health Research in the Region of Augsburg (KORA): In total, four population based health surveys have been conducted between 1984 and 2000 with 18.000 participants in the age of 25 to 74 years, and a biological specimen bank was established in order to enable the researchers to perform epidemiologic research with respect to molecular and genetic factors. The KORA study center conducts regular follow-up investigations and has collected a wealth of information on sociodemography, general medical history, environmental factors, smoking, nutrition, alcohol consumption, and various laboratory parameters. Follow-up activities include address inquiry for all participants (incl. assessment of vital status and cause of death), postal questionnaires focusing on chronic diseases, and complete follow-up studies with interviews and physical examination. 215 controls from this cohort study were included.

Leuven Multidisciplinary Breast Centre (LMBC): 59 triple negative cases and 89 controls from this hospital-based case control study in Leuven, Belgium were included. Cases included 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. Healthy controls (blood donors) were collected at the Red Cross located in Gasthuisberg hospital (Oct-2007-March 2008).

Mammary Carcinoma Risk Factor Investigation (MARIE): 273 triple negative cases and 182 controls from this population-based case-control study of breast cancer in Northern and Southern Germany were included in this analysis. Cases from this study were incident and prevalent cases diagnosed from 2001-2005 in the study region of Hamburg in Northern Germany and from 2002-2005 in the study region of Rhein-Neckar-Karlsruhe in Southern Germany. Controls 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.

Mayo Clinic Breast Cancer Study (MCBCS): 154 triple negative cases and 155 matched controls from this clinic-based breast cancer case-control study at the Mayo Clinic were included in this study. Subjects were enrolled between February 1, 2001 and June 30, 2005. Cases were comprised of Caucasian women with primary invasive breast cancer ascertained with 6 months of diagnosis. Controls were comprised of Caucasian women visiting the Mayo Clinic for general medical exams in the Department of Internal

Medicine with no prior history of cancer. Controls were frequency matched to cases on region of residence, race, and 5-year age group.

Melbourne Collaborative Cohort Study (MCCS): 41 triple negative breast cancer cases and 59 controls from this population-based prospective cohort study were included in this report. Incident cases of breast cancer were diagnosed within the Melbourne Collaborative Cohort Study in Melbourne, Australia during the follow-up from baseline (1990-1994) to 2008 of the 24469 participating women, and controls were randomly sampled from the initial cohort among members not diagnosed with breast cancer at the end of follow-up.

Oulu Breast Cancer Study (OBCS): 67 triple negative cases and 88 controls from this Finnish hospital-based case-control study were included. Cases were consecutive incident cases diagnosed at the Oulu University Hospital between 2000 and 2004. Controls were healthy, consecutive, anonymous, female Finnish Red-Cross blood donors recruited in 2002 from the same geographical region in Northern Finland.

Prospective Study of Outcomes in Sporadic Versus Hereditary Breast Cancer (POSH): 274 cases of triple negative breast cancer from this prospective cohort study in the United Kingdom were included. Cases were aged 40 or younger at breast cancer diagnosis, recruited across the UK, and diagnosed between January 2000 and December 2007.

Australian Twin Cohort study from the Queensland Institute of Medical Research (QIMR): Australian controls were unselected parents of adolescent twins taking part in studies of melanoma risk factors and cognition. Both mothers and fathers were collected but only mothers were used in this analysis. No phenotype data were collected for parents but a blood DNA sample was collected and typed using the Illumina 610K array. All controls were of northern European ancestry, mainly British Isles. Phenotypic and genotypic data collection was approved by the Queensland Institute of Medical Research (QIMR) Ethics Committee and informed consent was obtained from all participants.

RPCI: Data and specimens from 139 women with TN breast cancer and 137 healthy controls were obtained from the Data Bank and Biorepository (DBBR) at Roswell Park Cancer Institute (RPCI). The DBBR, as previously described (40), is a comprehensive data and sample bank containing pretreatment biospecimens that are rigorously collected and processed, with comprehensive clinical and epidemiologic data. Briefly, patients newly diagnosed with cancer at RPCI are invited to participate during their initial visit with the surgical oncologist. After consent, blood samples are collected (prior to any treatment for breast cancer, including surgery) in phlebotomy when specimens for clinical measures are drawn, transported to the laboratory through a pneumatic tube system, and processed within one hour of blood draw. Specimens are maintained in liquid nitrogen until analysis. The average time interval between the time of diagnosis and the time of blood draw for the women in our study was 27 days. Data on tumor characteristics, including ER, PR and HER2 status are obtained from the Pathology Resource Network and matched to cases by an honest broker by medical record numbers.

Healthy controls are identified from family members, friends or visitors (n=35, 6%) of patients with cancer other than breast, employee volunteers, and women recruited from community events such as Susan G. Komen Foundation sponsored Western New York Race for the Cure.

Sheffield Breast Cancer Study (SBCS): 46 triple negative cases and 53 controls from this hospital-based case-control study of breast cancer study were included. The study consists of women with pathologically confirmed breast cancer recruited from surgical outpatient clinics at the Royal Hallamshire Hospital, Sheffield, 1998 – 2005 and unselected women attending the Sheffield Mammography Screening Service between Sep 2000 - Aug 2004 if their mammograms showed no evidence of a breast lesion. Cases are a mixture of prevalent and incident disease.

Study of Epidemiology and Risk factors in Cancer Heredity (SEARCH): 202 cases of triple negative breast cancer from this population-based case-control study of breast cancer in the UK were included. Two groups of cases were identified through the East Anglican Cancer Registry: 1) prevalent cases diagnosed age <55 from 1991-6 and alive when study started in 1996; 2) incident cases diagnosed age < 70 diagnosed after 1996.

Städtisches Klinikum Karlsruhe and Deutsches Krebsforschungszentrum Breast Cancer Study (SKKDKFZS): 142 triple negative breast cancer cases were included from this breast cancer case cohort study. The study consists of women with pathologically confirmed breast cancer recruited at the Städtisches Klinikum Karlruhe, Karlsruhe,

Germany from 1993 - 2005. Cases were between 21-93 years of age. Controls were from an unselected series of unaffected women from the same geographical area.

Wellcome Trust Case Control Consortium (WTCCC): 1,374 controls were included from the 1958 Birth Cohort Controls (58BC) from the WTCCC. The 1958 Birth Cohort (also known as the National Child Development Study) includes all births in England, Wales and Scotland, during one week in 1958. From an original sample of over 17,000 births, survivors were followed up at ages 7, 11, 16, 23, 33 and 42 yrs. In a biomedical examination at 44-45 yrs, 9,377 cohort members were visited at home providing 7,692 blood samples with consent for future Epstein–Barr virus (EBV)-transformed cell lines. DNA samples extracted from 1,500 cell lines of self-reported white ethnicity and representative of gender and each geographical region were selected for use as controls.

Supplementary Table 1. Age distributions and years of diagnosis by study site

Study	Country	Age range (mean)		Years of diagnosis
		Cases	Controls	
ABCS	Netherlands	26 - 62 (39)	NA	1995 - 2007
ABCTB	Australia	28 - 91 (57)	NA	2006 - 2009
BBCC	Germany	27 - 88 (53)	NA	2002 - 2006
BBCS	U.K.	48 - 75 (59)	42 - 67 (60)	1971 - 2009
BIGGS	Ireland	31 - 70 (53)	60 - 60 (60)	2001 - 2008
CGEMS	U.S.A.	NA	54 - 75 (66)	NA
DEMOKRITOS	Greece	21 - 79 (53)	34 - 82 (50)	1997 - 2010
DFCI	U.S.A.	27 - 79 (49)	NA	1990- 2008
FCCC	U.S.A.	29 - 81 (53)	28 - 81 (53)	1995 - 2008
GENICA	Germany	25 - 79 (53)	24 - 79 (54)	2000 -2004
HEBCS	Finland	27 - 80 (53)	18 - 80 (58)	1995 - 2004
KARBAC	Sweden	48 - 88 (62)	48 - 85 (62)	1998 - 2000
KBCP	Finland	30 - 87 (57)	NA	1990 - 1995
KORA	Germany	NA	31 - 81 (60)	NA
LMBC	Germany	32 - 84 (54)	24 - 66 (50)	2000 - 2008
MARIE	Germany	50 - 75 (61)	50 - 76 (61)	2001 - 2005
MCBCS	U.S.A.	25 - 85 (52)	24 - 85 (52)	2001 - 2005
MCCS	Australia	25 - 85 (52)	42 - 70 (57)	1990 - 1994
OBCS	Finland	28 - 90 (57)	50 - 66 (56)	2000 - 2004
POSH	U.K.	22 - 41 (36)	NA	2000 - 2007
QIMR	Australia	NA	29 - 72 (45)	NA
RPCI	U.S.A.	28 - 92 (56)	27 - 92 (55)	2006 - 2010
SBCS	U.K.	31 - 93 (60)	46 - 81 (60)	1998 - 2004
SEARCH	U.K.	29 - 68 (50)	NA	1991- 2010
SKKDKFZS	Germany	22 - 87 (59)	46 - 68 (58)	1993 - 2005
WTCCC	U.K.	NA	52 - 52 (52)	NA

Supplementary Table 2. Criteria used to define ER, PR, and HER2 negative status by study site

Study	ER	PR	HER2
ABCS	***Negative***: <10% cells stained positive ((Neomarkers, 1D5 and 6F11 clones). ***Data source***: TMA.	***Negative***: <10% cells stained positive (ImmunoLogic, PR-1 clone). ***Data source***: TMA.	***Negative***: Score 0, 1+ or 2+ (NeoMarkers, 3B5 and 23 clones). ***Data source***: TMA.
ABCTB	***Negative***: Staining of any intensity in <1% of cells. ***Data source***: TMA.	***Negative***: Staining of any intensity in <1% of cells. ***Data source***: TMA.	***Negative***: <4 copies of HER2 gene per cell. ***Data source***: Single probe for HER2 SISH (N.B. some of the older cases were done by FISH).
BBCC	***Negative***: <9% of the cells stained positive (1D5; 1:200; monoclonal mouse IgG 1k; Dako, Denmark). ***Data source***: Whole sections of FFPE tissue.	***Negative***: <9% of the cells stained positive (PgR 636; 1:200; monoclonal mouse IgG; Dako, Denmark). ***Data source***: Whole sections of FFPE tissue.	***Negative***: DAKO score 0 or 1+. ***Data source***: Whole sections of FFPE tissue.
BBCS	***Negative***: Quick (Allred) score (intensity & proportion) 0-2. ***Data source***: Data extracted from clinical notes.	***Negative***: Quick (Allred) score (intensity & proportion) 0-2. ***Data source***: Data extracted from clinical notes.	***Negative***: IHC scores 0 or 1+; FISH ratio <2.0 ***Data source***: Data extracted from clinical notes.
BIGGS	***Negative***: Allred score=0-2 (score range 0-8). ***Data source***: Data from hospital pathology reports.	***Negative***: Allred score=0-2 (score range 0-8). ***Data source***: Data from hospital pathology reports.	***Negative***: IHC scores 0 or 1+. ***Data source***: Data from hospital pathology reports.
DEMOKRITOS	***Negative***: <1% immunoreactive nuclei (clone 6F11, Leica BioSystems, Newcastle, UK). ***Data source***: Central pathology review on TMA for samples initially evaluated before 2004 (30% of the samples) or abstracted from medical records for newer samples.	***Negative***: <1% immunoreactive nuclei (clone 1A6, Leica BioSystems, Newcastle, UK). ***Data source***: Central pathology review on TMA for samples initially evaluated before 2004 (30% of the samples) or abstracted from medical records for newer samples.	***Negative***: Score 0 or 1+ (clone PL, Dako, Glostrup, Denmark). ***Data source***: Central pathology review on TMA for samples initially evaluated before 2004 (30% of the samples) or abstracted from medical records for newer samples.

DFCI	***Negative***: <1% of cells stained positive (Dako, 1D5). ***Data source***: Whole sections of FFPE tissue.	***Negative***: <1% of cells stained positive (Dako, PgR 636). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Negative=0 or 1+ staining, or 2+ IHC (Dako A0485)and FISH not amplified (ratio < 2). ***Data source***: Whole sections of FFPE tissue.
FCCC	***Negative***: No nuclear staining (Novocastra, 6F11/2 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: No nuclear staining (Dako, PgR 636 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Score 0 or 1+ (Dako, HercepTest™). ***Data source***: Whole sections of FFPE tissue.
GENICA	***Negative***: Number of cells x intensity (german immuno reactive score) 0-2 (Dako, 1D5 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Number of cells x intensity (german immuno reactive score) 0-2 (Dako, PgR 636 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Score 0 or 1+ (Dako, HercepTest™). ***Data source***: Whole sections of FFPE tissue.
HEBCS	***Negative***: <10% cells stained positive (Novocastra). ***Data source***: Abstracted from medical records	***Negative***: <10% cells stained positive (Dako). ***Data source***: Abstracted from medical records	***Negative***: IHC score 0 or 1+ (IHC Novocastra Zymed, NCL-BC11 ErbB2 probe). ***Data source***: TMA.
KARBAC	***Negative***: <0.05 fmol/µg DNA (quantitative method, cytosol assay) or ≥10% stained (IHC). ***Data source***: Abstracted from medical records	***Negative***: <0.05 fmol/µg DNA (quantitative method, cytosol assay) or ≥10% stained (IHC). ***Data source***: Abstracted from medical records	Negative: 0 or 1+ with IHC, 2+ with IHC and FISH-negative. ***Data source***: Abstracted from medical records.
KBCP	***Negative***: Intensity score (0.1,2,3)*percentage score (0,1,2,3)= 0-2 (score range 0-6) (Abbot, ER_ICA kit). ***Data source***: Whole sections of FFPE tissue; abstracted from medical records.	***Negative***: Intensity score (0.1,2,3)*percentage score (0,1,2,3)= 0-2 (score range 0-6) (Abbot, PR_ICA kit). ***Data source***: Whole sections of FFPE tissue; abstracted from medical records.	***Negative***: Intensity scores 0 = no staining, Scoring of % cells stained: 0-10 %=negative ***Data source***: Hospital registry.
LMBC	***Negative***: Quick score 0-2, corresponding to no or weak staining.	***Negative***: Quick score 0-2, corresponding to no or weak staining.	***Negative***: IHC score 0 or 1+.
MARIE	***Negative***: <10% tumor nucei stained with intensity score <=1 (Dako, ID5 clone). ***Data source***: abstracted from medical records.	***Negative***: <10% tumor nucei stained with intensity score <=1 (Dako, PgR 636 clone). ***Data source***: abstracted from medical records.	***Negative***: Score 0, 1+ or 2 + and FISH negative (Dako A0485, cerB2 clone). ***Data source***: abstracted from medical records.

MCBCS	***Negative***: No nuclear staining (Novocastra, 6F11/2 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: No nuclear staining (Dako, PgR 636 clone). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Score 0 or 1+ (Dako, HercepTest™). ***Data source***: Whole sections of FFPE tissue.
MCCS	***Negative***: No positive nuclei with intensity score >=1 (Neomarkers RM9101, SP1 clone) ***Data source***: Whole sections of FFPE tissue; abstracted from medical records.	***Negative***: No positive nuclei with intensity score >=1 (Dako M3569, PgR 636 clone). ***Data source***: Whole sections of FFPE tissue; abstracted from medical records.	***Negative***: Score 0 or 1+ (Dako A0485, CerB2 clone). ***Data source***: Whole sections of FFPE tissue.
OBCS	***Negative***: <1% tumor nucei stained (DAKO, monoclonal, clone 1D5). ***Data source***: Whole sections of FFPE tissue.	***Negative***: <1% tumor nucei stained (DAKO, monoclonal, clone PgR636). ***Data source***: Whole sections of FFPE tissue.	***Negative***: Completely negative membranous immunostaining (DAKO, polyclonal). ***Data source***: Whole sections of FFPE tissue.
POSH	***Negative***: Allred or equivalent system scores of <3. ***Data source***: Data abstracted from clinical pathology reports.	***Negative***: Allred or equivalent system scores of <3. ***Data source***: Data abstracted from clinical pathology reports.	***Negative***: Score 0 or 1+. ***Data source***: Data abstracted from clinical pathology reports.
RPCI	***Negative***: Allred score <3 (score range 0-8).	***Negative***: Allred score <3 (score range 0-8).	***Negative***: Score 0 or 1+.
SBCS	***Negative***: Intensity score (0,1,2,3) * percentage of cells stained (0-100%) <50 (total score range 0-300) (Vector, 6F11/2 clone). ***Data source***: TMA; abstracted from medical records.	***Negative***: Intensity score (0,1,2,3) * percentage of cells stained (0-100%) <50 (total score range 0-300) (Vector, 1A6 clone). ***Data source***: TMA.	***Negative***: Score 0 or 1+ (Dako, HercepTest™K5204). ***Data source***: TMA.
SEARCH	***Negative***: Allred score <3 (score range 0-8) (Novocastra, 6F11/2 clone). ***Data source***: TMA; abstracted from medical records.	***Negative***: Allred score <3 (score range 0-8) (Dako, PgR 636 clone). ***Data source***: TMA; abstracted from medical records.	***Negative***: Score 0 or 1+ (Dako, HercepTest™K5204). ***Data source***: TMA; abstracted from medical records.
SKKDKFZS	***Negative***: Remmele Score <3. If the Remmele Score was not available, the status was based on the biochemical analysis with <20 fmol/mg protein being negative. ***Data source***: Whole sections of FFPE tissue.	***Negative***: Remmele Score <3. If the Remmele Score was not available, the status was based on the biochemical analysis with <20 fmol/mg protein being negative. ***Data source***: Whole sections of FFPE tissue.	***Negative***: Score 0 or 1+ (Dako A0485, CerB2 clone). ***Data source***: Whole sections of FFPE tissue.

Supplementary Table 3. Allele frequencies for 22 SNPs with available genotype data

SNP	Gene/Locus	Locus	Position (bp)	Minor (tested) Allele	Reference Allele	iPLEX						Combined					
						Total		Cases		Controls		Total		Cases		Controls	
						N	MAF	N	MAF	N	MAF	N	MAF	N	MAF	N	MAF
rs11249433	*1p11.2*	1p11.2	120982136	G	A	4092	0.40	2707	0.40	1385	0.41	7944	0.40	2976	0.40	4968	0.41
rs17468277	*ALS2CR12*	2q33.1	201862445	T	C	4092	0.13	2707	0.12	1385	0.13	7956	0.13	2979	0.12	4977	0.14
rs13387042	*2q35*	2q35	217614077	G	A	4089	0.47	2705	0.47	1384	0.47	7953	0.48	2977	0.47	4976	0.49
rs4973768	*SLC4A7: NEK10*	3p24	2739101	A	G	4070	0.48	2688	0.49	1382	0.47	7934	0.48	2960	0.49	4974	0.47
rs10941679	*MRPS30:FGF10*	5p12	44742255	G	A	4090	0.26	2705	0.26	1385	0.25	4090	0.26	2705	0.26	1385	0.25
rs889312	*MAP3K1*	5q11.2	56067641	C	A	4092	0.29	2707	0.29	1385	0.28	5601	0.29	2844	0.29	2757	0.28
rs2046210	*ESR1*	6q25.1	151990059	A	G	4092	0.38	2707	0.40	1385	0.34	4092	0.38	2707	0.40	1385	0.34
rs12662670	*ESR1*	6q25.1	151960549	G	T	4092	0.092	2707	0.10	1385	0.07	5466	0.09	2707	0.10	2759	0.076
rs13281615	*8q24*	8q24.21	128424800	G	A	4092	0.42	2707	0.42	1385	0.42	6254	0.42	2841	0.42	3413	0.41
rs1011970	*CDKN2BAS:CDKN2A: CDKN2B*	9p21.3	22052134	T	G	4092	0.18	2707	0.19	1385	0.17	7956	0.18	2979	0.19	4977	0.18
rs865686	*LOC100128657*	9q31.2	109928299	G	T	4092	0.37	2707	0.37	1385	0.37	7950	0.37	2979	0.37	4971	0.37
rs2380205	*ANKRD16:FBXO18*	10p15.1	5926740	T	C	4092	0.43	2707	0.43	1385	0.44	7953	0.43	2979	0.43	4974	0.43
rs10509168	*ZNF365*	10q21.2	63927834	T	C	4092	0.47	2707	0.46	1385	0.47	7956	0.47	2980	0.47	4976	0.47
rs704010	*ZMIZ1*	10q22.3	80511154	T	C	4062	0.38	2692	0.38	1370	0.38	7927	0.38	2964	0.38	4963	0.38
rs2981582	*FGFR2*	10q26	123342307	T	C	4092	0.40	2707	0.40	1385	0.41	5463	0.41	2707	0.40	2756	0.41
rs3817198	*LSP1*	11p15.5	1865582	C	T	4092	0.32	2707	0.32	1385	0.32	7685	0.32	2929	0.32	4756	0.32
rs614367	*MYEOV:CCND1*	11q13	69037945	T	C	4092	0.15	2707	0.15	1385	0.14	7675	0.15	2926	0.15	4749	0.15
rs999737	*RAD51L1*	14q24.1	68104435	T	C	4091	0.22	2706	0.21	1385	0.23	7955	0.23	2978	0.21	4977	0.24
rs3803662	*TOX3*	16q12.1	51143842	A	G	4092	0.29	2707	0.31	1385	0.27	7953	0.28	2980	0.30	4973	0.27
rs6504950	*COX11*	17q23.2	50411470	A	G	4092	0.28	2707	0.28	1385	0.29	4092	0.28	2707	0.28	1385	0.29
rs8170	*C19orf62:ANKLE1*	19p13.11	17250704	T	C	4092	0.21	2707	0.22	1385	0.17	7957	0.20	2979	0.22	4978	0.19
rs8100241	*C19orf62:ANKLE1*	19p13.11	17253894	A	G	4092	0.49	2707	0.47	1385	0.53	7300	0.50	2980	0.47	4320	0.51

Supplementary Table 4. Study subjects by site and membership in the Breast Cancer Association Consortium (BCAC)

Study Site	Country	All subjects			Non-BCAC			BCAC		
		Cases	Controls	Total	Cases	Controls	Total	Cases	Controls	Total
ABCS	Netherlands	67	0	67	31	0	31	36	0	36
ABCTB	Australia	154	0	154	154	0	154	0	0	0
BBCC	Germany	309	0	309	161	0	161	148	0	148
BBCS	U.K.	57	58	115	52	46	98	5	12	17
BIGGS	Ireland.	29	67	96	0	0	0	29	67	96
CGEMS	U.S.A	0	1126	1126	0	1126	1126	0	0	0
DEMOKRITOS	Greece	273	85	358	273	85	358	0	0	0
DFCI	U.S.A	271	0	271	271	0	271	0	0	0
FCCC	U.S.A	150	156	306	150	156	306	0	0	0
GENICA	Germany	57	64	121	0	0	0	57	64	121
HEBCS	Finland	85	221	306	0	0	0	85	221	306
KARBAC	Sweden	27	26	53	27	8	35	0	18	18
KBCP	Finland	34	0	34	34	0	34	0	0	0
KORA	Germany	0	215	215	0	215	215	0	0	0
LMBC	Germany	59	89	148	3	0	3	56	89	145
MARIE	Germany	273	182	455	102	0	102	171	182	353
MCBCS	U.S.A	151	155	306	6	9	15	145	146	291
MCCS	Australia	32	59	91	1	59	60	31	0	31
OBCS	Finland	67	88	155	0	0	0	67	88	155
POSH	U.K.	271	0	271	271	0	271	0	0	0
QIMR	Australia	0	657	657	0	657	657	0	0	0
RPCI	U.S.A	139	137	276	139	137	276	0	0	0
SBCS	U.K.	43	53	96	0	0	0	43	53	96
SEARCH	U.K.	202	0	202	2	0	2	200	0	200
SKKDKFZS	Germany	142	166	308	142	166	308	0	0	0
WTCCC	U.K.	0	1374	1374	0	1374	1374	0	0	0
Total		**2892**	**4978**	**7870**	**1819**	**4038**	**5857**	**1073**	**940**	**2013**

Supplementary Table 5. Breast cancer susceptibility SNP (n=22) associations with triple negative breast cancer with and without BCAC subjects

SNP	Gene/Locus	Chromosome	Position (bp)	Tested Allele	BCAC included	iPlex				Combined			
						Cases	Controls	P-trend	OR (95% CI)	Cases	Controls	P-trend	OR (95% CI)
rs11249433	1p11.2	1p11.2	120982136	G	N	1719	666	0.49	0.96 (0.84-1.09)	1916	4249	0.15	0.94 (0.87-1.02)
rs11249433	1p11.2	1p11.2	120982136	G	Y	2707	1385	0.54	0.97 (0.88-1.07)	2976	4968	0.27	0.96 (0.90-1.03)
rs17468277	*ALS2CR12*	2q33.1	201862445	T	N	1719	666	0.081	0.84 (0.69-1.02)	1919	4258	0.003	0.83 (0.73-0.94
rs17468277	*ALS2CR12*	2q33.1	201862445	T	Y	2707	1385	0.16	0.90 (0.78-1.04)	2979	4977	0.005	0.87 (0.78-0.96)
rs13387042	2q35	2q35	217614077	G	N	1717	665	0.15	0.91 (0.80-1.03)	1917	4257	0.11	0.94 (0.86-1.02)
rs13387042	2q35	2q35	217614077	G	Y	2705	1384	0.92	0.99 (0.91-1.09)	2977	4976	0.26	0.96 (0.90-1.03)
rs4973768	*SLC4A7:NEK10*	3p24	2739101	A	N	1706	664	0.39	1.06 (0.93-1.21)	1906	4256	0.57	1.02 (0.94-1.11)
rs4973768	*SLC4A7:NEK10*	3p24	2739101	A	Y	2688	1382	0.21	1.06 (0.97-1.17)	2960	4974	0.24	1.04 (0.97-1.12)
rs10941679	*FGF10*	5p12	44742255	G	N	1717	666	0.45	1.06 (0.91-1.22)	1717	666	0.45	1.06 (0.91-1.22)
rs10941679	*FGF10*	5p12	44742255	G	Y	2705	1385	0.43	1.04 (0.94-1.16)	2705	1385	0.43	1.04 (0.94-1.16)
rs889312	*MAP3K1*	5q11.2	56067641	C	N	1719	666	0.36	1.07 (0.93-1.23)	1783	2038	0.38	1.05 (0.94-1.18)
rs889312	*MAP3K1*	5q11.2	56067641	C	Y	2707	1385	0.20	1.07 (0.97-1.19)	2844	2757	0.13	1.07 (0.98-1.17)
rs2046210	*ESR1*	6q25.1	151990059	A	N	1719	666	4.03×10^{-5}	1.32 (1.16-1.51)	1719	666	4.03×10^{-5}	1.32 (1.16-1.51)
rs2046210	*ESR1*	6q25.1	151990059	A	Y	2707	1385	4.38×10^{-7}	1.29 (1.17-1.42)	2707	1385	4.38×10^{-7}	1.29 (1.17-1.42)
rs12662670	*ESR1*	6q25.1	151960549	G	N	1719	666?	0.002	1.45 (1.15-1.84)	1719	2040	0.002	1.33 (1.11-1.60)
rs12662670	*ESR1*	6q25.1	151960549	G	Y	2707	1385	3.52×10^{-4}	1.37 (1.15-1.62)	2707	2759	1.13×10^{-4}	1.33 (1.15-1.53)
rs13281615	8q24	8q24.21	128424800	G	N	1719	666	0.11	0.90 (0.79-1.02)	1782	2694	0.40	0.96 (0.87-1.06)
rs13281615	8q24	8q24.21	128424800	G	Y	2707	1385	0.70	0.98 (0.89-1.08)	2841	3413	0.79	0.99 (0.92-1.07)
rs1011970	*CDKN2BAS:CDKN2A:CDKN2B*	9p21.3	22052134	T	N	1719	666	0.002	1.32 (1.11-1.58)	1919	4258	0.067	1.10 (0.99-1.23)
rs1011970	*CDKN2BAS:CDKN2A:CDKN2B*	9p21.3	22052134	T	Y	2707	1385	0.024	1.16 (1.02-1.31)	2979	4977	0.13	1.07 (0.98-1.17)
rs865686	*LOC100128657*	9q31.2	109928299	G	N	1719	666	0.30	1.07 (0.94-1.23)	1919	4252	0.25	1.05 (0.97-1.14)
rs865686	*LOC100128657*	9q31.2	109928299	G	Y	2707	1385	0.96	1.00 (0.91-1.1)	2979	4971	0.65	1.02 (0.95-1.09)
rs2380205	*ANKRD16:FBXO18*	10p15.1	5926740	T	N	1719	666	0.52	1.04 (0.92-1.19)	1919	4255	0.91	1.00 (0.92-1.08)
rs2380205	*ANKRD16:FBXO18*	10p15.1	5926740	T	Y	2707	1385	0.94	1.00 (0.91-1.1)	2979	4974	0.71	0.99 (0.92-1.06)
rs10509168	*ZNF365*	10q21.2	63927834	T	N	1719	666	0.71	1.02 (0.90-1.17)	1919	4257	0.26	1.05 (0.97-1.14)
rs10509168	*ZNF365*	10q21.2	63927834	T	Y	2707	1385	0.88	0.99 (0.90-1.09)	2980	4976	0.79	1.01 (0.94-1.08)
rs704010	*ZMIZ1*	10q22.3	80511154	T	N	1711	657	0.87	1.01 (0.89-1.15)	1911	4250	0.99	1.00 (0.92-1.09)
rs704010	*ZMIZ1*	10q22.3	80511154	T	Y	2692	1370	0.99	1.00 (0.91-1.1)	2964	4963	0.80	0.99 (0.93-1.06)

rs2981582	*FGFR2*	10q26	123342307	T	N	1719	666	0.026	0.86 (0.75-0.98)	1719	2037	0.006	0.86 (0.77-0.96)
rs2981582	*FGFR2*	10q26	123342307	T	Y	2707	1385	0.64	0.98 (0.89-1.08)	2707	2756	0.24	0.95 (0.88-1.03)
rs3817198	*LSP1*	11p15.5	1865582	C	N	1719	666	0.74	0.98 (0.85-1.12)	1868	4037	0.70	0.98 (0.90-1.07)
rs3817198	*LSP1*	11p15.5	1865582	C	Y	2707	1385	0.68	1.02 (0.92-1.13)	2929	4756	0.49	1.03 (0.95-1.10)
rs614367	*MYEOV:CCND1*	11q13	69037945	T	N	1719	666	0.14	1.15 (0.95-1.38)	1865	4030	0.29	1.06 (0.95-1.19)
rs614367	*MYEOV:CCND1*	11q13	69037945	T	Y	2707	1385	0.12	1.12 (0.97-1.28)	2926	4749	0.17	1.07 (0.97-1.18)
rs999737	*RAD51L1*	14q24.1	68104435	T	N	1718	666	0.10	0.88 (0.76-1.03)	1918	4258	0.001	0.85 (0.78-0.94)
rs999737	*RAD51L1*	14q24.1	68104435	T	Y	2706	1385	0.053	0.90 (0.80-1.00)	2978	4977	2.96×10^{-4}	0.86 (0.80-0.93)
rs3803662	*TOX3*	16q12.1	51143842	A	N	1719	666	0.06	1.15 (0.99-1.32)	1919	4254	2.91×10^{-4}	1.18 (1.08-1.29)
rs3803662	*TOX3*	16q12.1	51143842	A	Y	2707	1385	8.25×10^{-4}	1.20 (1.08-1.33)	2980	4973	3.66×10^{-5}	1.17 (1.09-1.26)
rs6504950	*COX11*	17q23.2	50411470	A	N	1719	666	0.55	0.96 (0.83-1.11)	1719	666	0.55	0.96 (0.83-1.11)
rs6504950	*COX11*	17q23.2	50411470	A	Y	2707	1385	0.54	0.97 (0.87-1.07)	2707	1385	0.54	0.97 (0.87-1.07)
rs8170	*C19orf62:ANKLE1*	19p13.11	17250704	T	N	1719	666	2.65×10^{-4}	1.36 (1.15-1.61)	1919	4259	1.03×10^{-5}	1.25 (1.13-1.38)
rs8170	*C19orf62:ANKLE1*	19p13.11	17250704	T	Y	2707	1385	7.30×10^{-8}	1.40 (1.24-1.58)	2979	4978	2.25×10^{-8}	1.27 (1.17-1.38)
rs8100241	*C19orf62:ANKLE1*	19p13.11	17253894	A	N	1719	666	7.18×10^{-4}	0.79 (0.69-0.91)	1919	3601	7.92×10^{-6}	0.82 (0.76-0.90)
rs8100241	*C19orf62:ANKLE1*	19p13.11	17253894	A	Y	2707	1385	1.81×10^{-6}	0.79 (0.71-0.87)	2980	4320	8.66×10^{-7}	0.84 (0.78-0.90)