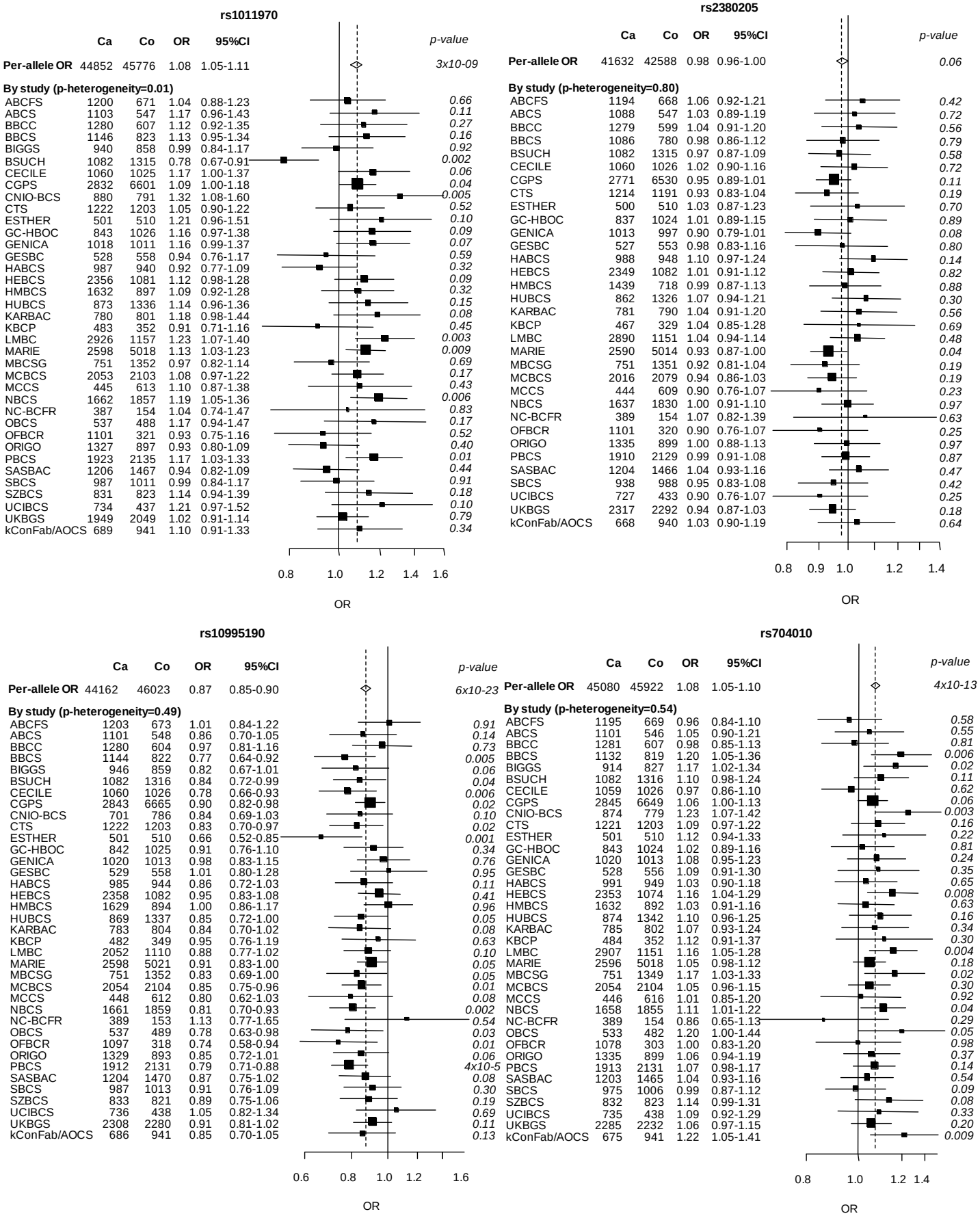
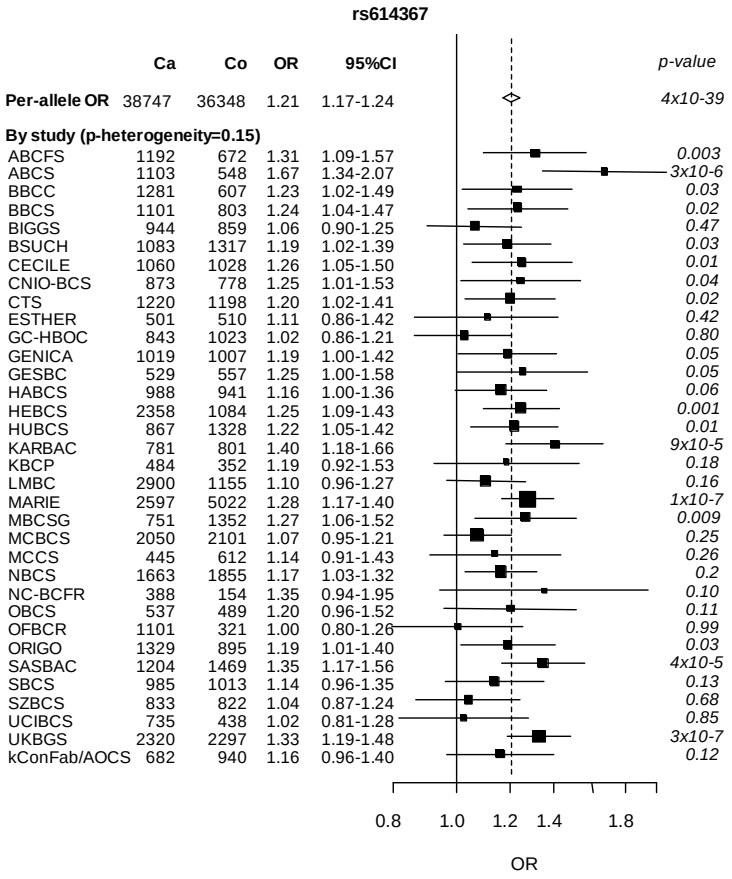
Supp. Figure S1. Forest plots of individual BCAC studies of the 5 variants in European women



Supp. Figure S1 (cont'd)



Supp. Table S1. Study description and genotyping method

Study	Acronym	Country	Case Definition and Ascertainment	Control Definition and Ascertainment	Age Range for Cases / Controls	Max. number of Cases / Controls included in analysis		Genotyping method[3]
						European[1]	Asian[2]	
Australian Breast Cancer Family Study [Dite et al., 2003]	ABCFS	Australia	All cases diagnosed < age 40 plus a random sample of those diagnosed ages 40-59 from cancer registries in Victoria and New South Wales, plus a limited number diagnosed aged 60-69; cases living in Melbourne recruited from 1992-99 and in Sydney from 1993-98.	Identified from the electoral rolls in Melbourne from 1992-98 and Sydney from 1993-99. Frequency matched to cases by age in 5 year categories.	23-69 / 20-68	1205 / 673	58 / 16	T
Amsterdam Breast Cancer Study [Schmidt.M.K. et al., 2007]	ABCS	Netherlands	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 in1965-2008.	Random women <50 years of age at baseline from 2 population-based prospective studies - the Monitoring Project on Cardiovascular Risk Factors (1987-1991) and the Monitoring Project on Chronic Disease Risk Factors (1993-1997). These studies were run by National Institute for Public Health and the Environment, The Netherlands. Controls are selected from the same catchment area as cases.	23-79 / 20-49	1103 / 548		M
Asia Cancer Program	ACP	Thailand	Women who underwent biopsy and have been pathologically diagnosed as having breast cancer. Aged less than 71 years of age	Women aged less than 71 years of age without cancer history of any kinds. Women who attend the out patient clinic under the minor injuries such as cuts, broken bones. Women who are institutionalized at the hospital with diseases not related to cancer or metabolic syndromes such as diabetes, heart diseases or conditions related to gynaecology and are well enough to give information to researchers."	21-70 / 18-70		317 / 558	M
Bavarian Breast Cancer Cases and Controls [Fasching et al., 2008; Schrauder et al., 2008]	BBCC	Germany	Consecutive, unselected cases with invasive breast cancer recruited at the University Breast Centre, Franconia in Northern Bavaria during 2002-2006.	Healthy women with no diagnosis of cancer aged 55 or older. Invited by a newspaper advertisement in Northern Bavaria, and recruited during 2002-2006	22-96 / 18-100	1281 / 607		M

British Breast Cancer Study [Fletcher et al., 2006]	BBCS	UK	1) English & Scottish Cancer Registries: all breast cancer cases who developed a first primary before age 65 in 1971 or later and who subsequently developed a second primary cancer. 2) Unilateral breast cancer cases diagnosed before age 70 in 1971 or later.	1) A friend, sister-in-law, daughter-in-law or other non-blood relative of cases. Recruitment of cases and controls began in January 2001.	26-77 / 21-81	1147 / 825	M
Breast Cancer in Galway Genetic Study [Colleran et al., 2010; McInerney et al., 2009]	BIGGS	Ireland	Unselected cases recruited from West of Ireland since 2001. Cases were recruited from University College Hospital Galway and surrounding hospitals	Women > 60 years with no personal history of any cancer and no family History of breast or ovarian cancer were identified from retirement groups in the West of Ireland (same catchment area as cases) during the period 2001-2008.	24-90 / 25-96	1147 / 859	T
Breast Cancer Study of the University Clinic of Heidelberg	BSUCH	Germany	All cases diagnosed with breast cancer in 2007-2009 at the University Women`s Clinic Heidelberg	Healthy, unrelated, ethnically matched female blood donors recruited in 2007 & 2009 by German Red Cross Blood Service of Baden-Württemberg-Hessen, Institute of Transfusion Medicine & Immunology, Mannheim.	25-89 / 30-69	1083 / 1317	M
CECILE Breast Cancer Study [Villeneuve et al., 2011]	CECILE	France	All cases diagnosed with breast cancer in 2005-2007 among women <75 years of age and residing in Ille-et-Vilaine (Rennes) or in Côte d'Or (Dijon) at diagnosis. Cases were recruited from the main cancer treatment center (Centre Eugène-Marquis in Rennes and Centre Georges-François-Leclerc in Dijon) and from private or public hospitals in each area.	General population control women residing in the same areas. Controls were recruited in 2005-2007 using a random digit dialing procedure and quotas by socioeconomic status, and were frequency-matched to the cases by 5-year age groups.	25-74 / 25-74	1060 / 1028	F
Copenhagen General Population Study [Weischer et al., 2007]	CGPS	Denmark	Consecutive, incident cases from 1 hospital with centralized care for a population of 400,000 women from 2001 to the present.	Community controls residing in the same region as cases and with no history of breast cancer were identified from the Copenhagen General Population Study recruited 2003-2007. All controls were known to still be breast cancer-free at the end of 2007.	24-95 / 20-91	2848 / 6678	T
Spanish National Cancer Centre Breast Cancer Study [Milne et al., 2006]	CNIO-BCS	Spain	Two groups of cases: 1) 574 consecutive breast cancer patients, unselected for family history, from 3 public hospitals, 2 in Madrid and one in Oviedo, from 2000 to 2005. 2) 291 cases with at least one first degree relative also affected with breast cancer, recruited through the CNIO family cancer clinic in Madrid from 2000 to 2004.	Women attending the Menopause Research Centre between 2000 and 2004 and female members of the College of Lawyers attending a free, targeted medical check-up in 2005, all free of breast cancer and all in Madrid	23-88 / 26-86	882 / 791	M

California Teachers Study	CTS	USA	Prospective cohort study: nested case-control	Prospective cohort study: nested case-control	32-83 / 26-77	1222 / 1203	41 / 30	M
ESTHER Breast Cancer Study [Widschwendter et al., 2008]	ESTHER	Germany	Statewide recruitment of breast cancer cases in all hospitals in Saarland/Germany in 2001-2003	Statewide recruitment of participants of a routine health check-up in Saarland/Germany in 2000-2002. A stratified random sample, matched to the cases by five year age groups, was selected as controls.	30-79 / 49-75	501 / 510		M
German Consortium for Hereditary Breast & Ovarian Cancer [Frank et al., 2006]	GC-HBOC	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; Germany.	Healthy, unrelated, ethnically matched female blood donors recruited in 2004 & 2007 by German Red Cross Blood Service of Baden-Württemberg-Hessen, Institute of Transfusion Medicine & Immunology, Mannheim.	19-87 / 19-68	844 / 1026		M
Gene Environment Interaction and Breast Cancer in Germany [Justenhoven et al., 2008; Pesch et al., 2005]	GENICA	Germany	Incident breast cancer cases enrolled between 2000 and 2004 from the Greater Bonn area (by of the hospitals within the study region); all enrolled within 6 months of diagnosis	Selected from population registries from 31 communities in the greater Bonn area; matched to cases in 5-year age classes between 2001 and 2004	23-80 / 24-80	1020 / 1013		M
Genetic Epidemiology Study of Breast Cancer by Age 50 [Chang-Claude et al., 2000]	GESBC	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.	24-50 / 24-52	529 / 558		M
Hannover Breast Cancer Study [Dork et al., 2001]	HABCS	Germany	Cases who received radiotherapy for breast cancer at Hannover Medical School between 1997-2003, unselected for age or family history	Anonymous female blood bank donors at Hannover Medical School, collected from 8/2005-12/2005, with known age and ethnic background	25-91 / 18-68	994 / 950		T

Helsinki Breast Cancer Study [Kilpivaara et al., 2005; Syrjakoski et al., 2000]	HEBCS	Finland	(1) Consecutive cases (883) from the Department of Oncology, Helsinki University Central Hospital 1997-1998 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 (from 1995)	Healthy females from the same geographical region in Southern Finland in 2003.	22-96 / 18-66	2358 / 1084	M
Hannover-Minsk Breast Cancer Study [Bogdanova et al., 2009]	HMBCS	Belarus	Ascertainment at 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 through the years 2002-2008.	Controls from the same population aged 18-72 years. Healthy (without personally history of cancer) female probands recruited from the same geographical regions as cases during the years 2002-2008. About 75% of controls were women invited for general medical examination at five regional gynecology clinics (in Gomel, Mogilev, Grodno, Brest or Vitebsk) and cancer-free volunteers ascertained at the Institute for Inherited Diseases in Minsk; 20% were cancer-free female blood bank donors recruited at Republic Blood Bank, Minsk, Belarus; finally 5% of controls were healthy cancer-free relatives of some breast cancer patients.	16-82 / 18-72	1643 / 905	T
Hannover-Ufa Breast Cancer Study [Bogdanova et al., 2009]	HUBCS	Russia	Consecutive Russian breast cancer patients aged 24-86 years ascertained at one of the two participating oncological centers in Bashkorstostan and Siberia through the years 2000-2008	Population controls aged 18-84 years recruited from a population study of different populations of Russia. Healthy volunteers (without any malignancy) were selected from the same geographical regions during the years 2002-2008.	25-85 / -	877 / 1348	T
Karolinska Breast Cancer Study [Lindblom et al., 1992; Margolin et al., 2004]	KARBAC	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	24-88 / -	785 / 805	T
Kuopio Breast Cancer Project [Hartikainen et al., 2005; Hartikainen et al., 2006]	KBCP	Finland	Women seen at Kuopio University Hospital between 1990 and 1995 because of breast lump, mammographic abnormality, or other breast symptom who were found to have breast cancer	Age and long-term area-of-residence matched controls selected from the National Population Register and interviewed in parallel with the cases	23-92 / 27-77	485 / 353	M

Kathleen Cuningham Foundation Consortium for research into Familial Breast Cancer/Australian Ovarian Cancer Study [Beesley et al., 2007; Mann et al., 2006]	KConFab/AOCS	Australia and New Zealand	Cases were from multiple-case breast and breast-ovarian families recruited though family cancer clinics from across Australia and New Zealand from 1998 to the present. Cases were selected for inclusion in BCAC studies if (i) family was negative for mutations in BRCA1 and BRCA2 (ii) case was the index for the family, defined as youngest breast cancer affected family member.	Female controls were ascertained by the Australian Ovarian Cancer Study identified from the electoral rolls from all over Australia from 2002-2006.	17-78 / 20-83	691 / 941	M
Leuven Multidisciplinary Breast Centre [De et al., 2008; Neven et al., 2008]	LMBC	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	Healthy controls (blood donors) collected at the Red Cross and located in Gasthuisberg hospital (Oct-2007-March 2008)	22-94 / 19-66	2930 / 1158	M
Mammary Carcinoma Risk Factor Investigation [Flesch-Janys et al., 2008]	MARIE	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.	50-75 / 49-75	2600 / 5023	M
Milan Breast Cancer Study Group [Catucci et al., 2009; De et al., 2009]	MBCSG	Italy	Familial and/or early onset breast cancer patients (aged 22-87) negative for mutations in BRCA genes, ascertained in two large cancer centres in Milan from 2000 to date.	Healthy blood donors aged 18-71 years, retruited at two blood centres in Milan from 2004 (centre 1) and 2007 (centre 2) to date	21-80 / 18-71	751 / 1352	M
Mayo Clinic Breast Cancer Study [Olson et al., 2007]	MCBCS	USA	Incident cases residing in 6 states (MN, WI, IA, IL, ND, SD) seen at the Mayo Clinic in Rochester, MN from 2002-2005	Women without cancer presenting for general medical examination at the Mayo Clinic. Controls were recruited concurrently with cases and were frequency matched to cases on age, ethnicity and county/state	22-93 / 25-86	2054 / 2104	T
Melbourne Collaborative Cohort Study [Giles and English, 2002]	MCCS	Australia	Incident cases diagnosed within the Melbourne Collaborative Cohort Study during the follow-up from baseline (1990-1994) to 2008 of the 24469 participating women	Random sample of the initial cohort	37-80 / 38-70	450 / 617	T

Norwegian Breast Cancer Study [Nordgard et al., 2008]	NBCS	Norway	Incidence cases from three different hospitals: 1) Cases (114) mean age 64 (28-92) at Ullevål Univ. Hospital 1990-94, 2) cases (182) mean age 59 (26-75) referred to Norwegian Radium Hospital 1975-1986, 3) cases (124), mean age 56 (29-82)) with stage I or II disease, in the Oslo micro-metastases study at Norwegian Radium Hospital between 1995-1998, 4) cases (71) mean age 67 (37–82) with locally advanced disease at Haukeland Univ. Hospital.	Control subjects were healthy women, age 55-71, residing in Tromsø (440), and Bergen (109) attending the Norwegian Breast Cancer Screening Program.	26-90 / 22-75	1664 / 1862		M
Northern California Breast Cancer Family Registry [John et al., 2004]	NC-BCFR	USA	Cases included those enrolled in the NC-BCFR as part of Phase I and II recruitment. Incident cases aged <65 years diagnosed between 1995 and 2003 were identified through the SEER cancer registry of the Greater San Francisco Bay Area. All cases likely at increased genetic risk were eligible to enroll in the BCFR (dx at age <35 yrs, personal history of ovarian or childhood cancer, bilateral breast cancer with 1st dx at age <50, family history of breast or ovarian cancer in first-degree relatives). Cases not meeting these criteria were randomly sampled (2.5% of whites, 30% of African Americans, 28% of Hispanics, 38% of Asian Americans).	Controls were identified through random digit dialing conducted from 1999-2000 in the same geographic region. Controls were frequency matched to cases on 5-year age group and race/ethnicity, at a ratio of 1 control per 2 cases.	22-65 / 19-66	398 / 154	460 / 61	T
Oulu Breast Cancer Study [Erkko et al., 2007]	OBCS	Finland	Consecutive incident cases diagnosed at the Oulu University Hospital between 2000 and 2004.	Healthy, consecutive, anonymous, female Finnish Red-Cross blood donors recruited in 2002 from the same geographical region in Northern Finland.	28-92 / 18-66	537 / 489		M

Ontario Familial Breast Cancer Registry[John et al., 2004]	OFBCR	Canada	Cases were identified from the Ontario Cancer Registry which registers >97% of all cases residing in the province at the time of diagnosis. Between 1 Jan1996-31 Dec 1998 all cases diagnosed with invasive breast cancer aged 20-54 years who met the OFBCR definition for high genetic risk (family history of specific cancers particularly breast and ovarian, early onset disease, Ashkenazi ethnicity or a diagnosis of multiple breast cancer) were asked to participate by completing risk factor questionnaires and providing a blood sample. A 25% random sample of individuals in this age category who did not meet the OFBCR definition, 35% of those aged 55-69 at high risk and 8.75% aged 55-69 at low risk were also asked to participate. All males between 20-79 years were invited to participate. During 2001-2005, enrollment was limited to those individuals who met high risk criteris. This multi-step sampling scheme enriched the population for genetically predisposed individuals, which was an objective of the Ontario Familial Breast Cancer Registry.	Unrelated, unaffected population controls were recruited between 1998-2001 by calling randomly selected residential telephone numbers throughout the same geographical region. Eligible controls were women with no history of breast cancer and were frequency-matched by 5-year age group to the expected age distribution of cases. Approximately, 65% of identified eligible women returned questionnaires, and 63% of these donated a blood specimen.	22-81 / 26-69	1101 / 321	120 / 15	T
Leiden University Medical Centre Breast Cancer Study [de Bock et al., 2004; Huijts et al., 2007]	ORIGO	Netherlands	Consecutive cases diagnosed 1996-2006 in 2 hospitals of South-West Netherlands (Leiden & Rotterdam). No selection for family history; Rotterdam cases selected for diagnosis aged <70. Cases with in situ carcinomas eligible.	Three groups of controls: (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.	22-80 / 22-80	1337 / 899		T
NCI Polish Breast Cancer Study[Garcia-Closas et al., 2006]	PBCS	Poland	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 Lodz covering 100% of all eligible cases	Randomly selected from population lists of all residents of Poland, stratified and frequency matched to cases by case city and age in 5 year categories. Recruited 2000-2003.	27-75 / 24-75	1926 / 2141		T

Singapore and Sweden Breast Cancer Study [Wedren et al., 2004]	SASBAC	Sweden	Incident cases from October 1993 to March 1995 identified via the 6 regional cancer registries in Sweden, to which reporting is mandatory.	Controls were randomly selected from the total population registry in 5-year age groups to match the expected age-frequency distribution among cases. Patients and controls were recruited from Oct 1993 through April 1995.	50-75 / 49-76	1206 / 1472		M
Sheffield Breast Cancer Study [MacPherson et al., 2004; Rafii et al., 2002]	SBCS	UK	Women with pathologically confirmed breast cancer recruited from surgical outpatient clinics at the Royal Hallamshire Hospital, Sheffield, 1998 – 2005; cases are a mixture of prevalent and incident disease	Unselected women attending the Sheffield Mammography Screening Service between Sep 2000 - Aug 2004, if their mammograms showed no evidence of a breast lesion	28-92 / 45-78	987 / 1013		M
Seoul Breast Cancer Study [Han et al., 2008; Lee et al., 2005]	SEBCS	Korea	Consecutive, incident, cases from 2 hospitals in Seoul recruited 2001-2005	Healthy community controls from same catchment area and participating in annual health check-up, 2001-2005.	19-82 / 18-86		2083 / 323	T
IHCC-Szczecin Breast Cancer Study [Jakubowska et al., 2009; Lubinski et al., 2009]	SZBCS	Poland	Prospectively ascertained cases of invasive breast cancer patients diagnosed at the Regional Oncology Hospital (Szczecin) in the years 2002, 2003, 2006 and 2007 or the University Hospital from 2002 to 2007 in Szczecin, West-Pomerania, Poland. Patients with pure intraductal or intralobular cancer were excluded (DCIS or LCIS) but patients with DCIS with micro-invasion were included.	Unaffected, matched to cases for year of birth, sex and region; from families with negative cancer family history; controls were part of a population-based study of the 1.3 million inhabitants of West Pomerania performed in 2003 and 2004 designed to identify familial aggregations of cancer by our centre	25-88 / 26-91	833 / 824		T
Taiwanese Breast Cancer Study [Ding et al., 2009; Hsu et al., 2007]	TWBCS	Taiwan	Incident cases diagnosed & treated at 2 major teaching hospitals in Taiwan. [between March 2002 and August 2005]	Controls cancer-free individuals, randomly selected from women attending health exam. at same hospital during study period. Underwent 1-day health examination - any showing evidence cancer excluded.	18-85 / 25-81		886 / 834	T
UCI Breast Cancer Study [nton-Culver et al., 2000; Ziogas et al., 2000]	UCIBCS	USA	All cases diagnosed in Orange County, California, during one-year period beginning March 1, 1994. Ascertained through the population-based Cancer Surveillance Program of Orange County California (CSPOC)	Female controls under age 75 years without history of cancer recruited using random digit dialing among Orange County residents & frequency matched to cases by age & race/ethnicity. Recruited from 1998-2003	24-90 / 20-75	736 / 438	44 / 14	M
UK Breakthrough Generations Study	UKBGS	UK	All members who had had breast cancer before entry into the Breakthrough Generations Study (cohort of 100,000+ women followed up for breast cancer, recruited from the UK during 2003-2009).	Women who had not had breast cancer before entry into the cohort study, 1:1 matched to cases on date of birth, year of entry into the study (2003-2009), source of recruitment, blood sample and ethnicity	24-84 / 26-86	2326 / 2310		T

[1]Subjects that did not declared themselves as non-European
[2]Subjects that declared themselves as Asian (studies with less than 10 cases or 10 controls were excluded)
[3]Genotyping platforms: M: MALDI TOF MS; T: TaqMan; F: Fluidigm

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