

Supplementary table 1. Contributing studies.

Consortium	Country-stratum	Study	Acronym	Country	Study design	Study subjects		Pathology data to support imputation		
						BRCA1	BRCA2	BRCA1/2	non-carriers	
CIMBA	Australia	Kathleen Cuninghams Consortium for Research into Familial Breast Cancer	KCONFAB	Australia	Cancer clinic based study	315	246	64		
		Victorian Familial Cancer Trials Group	VFCTG	Australia	Cancer clinic based study	67		52		
	Austria	General Hospital Vienna	MUV	Austria	Cancer clinic based study	189	99	58		
	Denmark	Copenhagen Breast Cancer Study	CBCS	Denmark	Cancer clinic based study	109	68	24		
		Odense University Hospital	OUIH	Denmark	Cancer clinic based study	169	135	41		
	Finland	Helsinki Breast Cancer Study	HEBCS	Finland	Cancer clinic based study	48	56	13		
	Germany	German Familial Breast Group	GC-HBOC	Germany	Cancer clinic based study	398	231	1149		
	Netherlands	Hereditary Breast and Ovarian cancer study the Netherlands	HEBON	Netherlands	Cancer clinic based study	274	103	38		
	NorthAmerica	Ontario site of the Breast Cancer Family Registry / Ontario Cancer Genetics Network	BCFR-ON/OCGN	Canada	Cancer clinic and population based study		45	77		
		City of Hope Cancer Center	COH	USA	Cancer clinic based study	94		6		
		Dana Farber Cancer Institute	DFCI	USA	Cancer clinic based study	58	43	14		
		University of Pennsylvania	UPENN	USA	Cancer clinic based study	101		145		
		University of Texas MD Anderson Cancer Center	UTMDACC	USA	Cancer clinic based study	61		38		
	Poland	International Hereditary Cancer Centre	IHCC	Poland	Cancer clinic based study	244		10		
	Sweden	Swedish Breast Cancer Study	SWE-BRCA	Sweden	Cancer clinic based study	83		48		
	Iberia	Institut Català d'Oncologia	ICO	Spain	Cancer clinic based study	78	86	86		
		Portuguese Oncology Institute-Porto Breast Cancer Study	IPOBCS	Portugal	Cancer clinic based study		81	43		
	UK	Victorian Familial Cancer Trials Group	EMBRACE	UK	Cancer clinic based study	604	620	232		
	BCAC	Australia	Kathleen Cuninghams Foundation Consortium for research into Familial Breast Cancer / Australian Ovarian Cancer Study	KCONFAB/AOCS	Australia and New Zealand	Clinic-based recruitment of familial breast cancer patients		9	2	1002
			Melbourne Collaborative Cohort Study	MCCS	Australia	Prospective cohort study: nested case-control study	3	7	1	652
CentralEurope		Breast Cancer Study of the University of Heidelberg	BSUCH	Germany	Hospital-based cases	5	5		176	
		CECILE Breast Cancer Study	CECILE	France	Population-based case-control study	7	12	3	833	
		German Consortium for Hereditary Breast & Ovarian Cancer	GC-HBOC	Germany	Clinic-based case study and prospective cohort study		4	16	2099	
		Gene Environment Interaction and Breast Cancer in Germany	GENICA	Germany	Population-based case-control study	9	15		691	
		Genetic Epidemiology Study of Breast Cancer by Age 50	GESBC	Germany	Population-based study of women <50 years	23	17		510	
		Mammary Carcinoma Risk Factor Investigation	MARIE	Germany	Population-based case-control study	9	27	1	1999	
		Städtisches Klinikum Karlsruhe Deutsches Krebsforschungszentrum Study	SKDKFZS	Germany	Hospital-based breast cancer cohort	12	16	1	700	
Denmark		Copenhagen General Population Study	CGPS	Denmark	Population-based case-control study	34	24	4	1893	
Finland		Helsinki Breast Cancer Study	HEBCS	Finland	Hospital-based case-control study, plus additional familial cases	8	13	1	1499	
		Kuopio Breast Cancer Project	KBCP	Finland	Population-based prospective clinical cohort	3	3	5	404	
Netherlands		Amsterdam Breast Cancer Study	ABCS	Netherlands	Hospital-based consecutive cases	27	26	26	962	
		Amsterdam Breast Cancer Study - Familial	ABCS-F	Netherlands	Clinical Genetic Center-based cases				119	
		Rotterdam Breast Cancer Study	RBCS	Netherlands	Hospital-based case-control study, Rotterdam area	5	2		894	
NorthAmerica		Ontario Familial Breast Cancer Registry	OFBCR	Canada	Population-based familial case-control study	1	3		354	
		The Prostate, Lung, Colorectal and Ovarian (PLCO) Cancer Screening Trial	PLCO	USA	Prospective cohort study: nested case-control	2	15	1	1188	
		Utah Breast Cancer Study	UBCS	USA	Pedigrees including multiple cases; Hospital-based cases; Population-based cases	5	7	5	486	
Poland		NCI Polish Breast Cancer Study	PBCS	Poland	Population-based case-control study	52	29	6	1460	
Scandinavia		Karolinska Breast Cancer Study	KARBAC	Sweden	Population and hospital-based cases	2	4		285	
	Karolinska Mammography Project for Risk Prediction of Breast Cancer - Cohort Study	KARMA	Sweden	Cohort study	13	19	2	2425		
	Norwegian Breast Cancer Study	NBCS	Norway	Hospital-based case-control study	1	3		424		
	Singapore and Sweden Breast Cancer Study	SASBAC	Sweden	Population-based case-control study	7	3		903		
SouthernEurope	Breast Oncology Galicia Network	BREGGAN	Spain	Population-based case-control	8	10	4	489		
	Crete Cancer Genetics Program	CCGP	Greece	Hospital-based case-control study	8	2		387		
	Milan Breast Cancer Study Group	MBCSG	Italy	Clinic-based familial/early onset breast cancer patients	2	2		425		
UK	Predicting the Risk Of Cancer At Screening Study	PROCAS	UK	Population based study	4		1	322		
	Study of Epidemiology and Risk factors in Cancer Heredity	SEARCH	UK	Population-based case-control study	72	151	47	9331		