Table S1. Triple Negative Breast Cancer Consortium (TNBCC) studies

Stage	Study Abbreviation	Full Name	Platform	Country	Cases	Controls
Stage 1	ABCTB	Australian Breast Cancer Tissue Bank	Illumina 660-Quad	Australia	144	
	BBCC	Bavarian Breast Cancer Cases and Controls	Illumina 660-Quad	Germany	218	
	CGEMS	Cancer Genetic Markers of Susceptibility	Illumina 550 v.1	USA		947
	DFCI	Harvard Breast Cancer SPORE Blood Repository	Illumina 660-Quad	USA	246	
	FCCC	Fox Chase Cancer Center	Illumina 660-Quad	USA	120	
	GENICA	Gene Environment Interaction and Breast Cancer in Germany	Illumina 660-Quad	Germany	26	
	HEBCS	Helsinki Breast Cancer Study	Illumina HumanHap 550k DUO/ Illumina CNV370-Duo	Finland	83	219
	KORA	Cooperative Health Research in the Region of Augsburg	Illumina 550	Germany		215
	MARIE	Mammary Carcinoma Risk Factor Investigation	Illumina 660-Quad/ Illumina CNV370	Germany	148	
	MCBCS	Mayo Clinic Breast Cancer Study	Illumina 660-Quad	USA	147	
	MCCS	Melbourne Collaborative Cohort Study	Illumina 660-Quad	Australia	39	
	POSH	Prospective Study of Outcomes in Sporadic Versus Hereditary Breast Cancer	Illumina 660-Quad	UK	266	
	QIMR	Australian Twin Cohort study from the Queensland Institute of Medical Research	Illumina 610-Quad	Australia		650
	SBCS	Sheffield Breast Cancer Study	Illumina 660-Quad	UK	42	
	WTCCC	Wellcome Trust Case Control Consortium	Illumina 1.2M	UK		1368
				TOTAL	**1529**	**3399**
Stage 2	CTS	California Teachers Study	iCOGS	USA	68	71
	DEMOKRITOS	Demokritos	iCOGS	Greece	526	304
	FCCC	Fox Chase Cancer Center	iCOGS	USA	4	137
	GENICA	Gene Environment Interaction and Breast Cancer in Germany	iCOGS	Germany	33	30
	KUMC	Kansas University Medical Center	iCOGS	USA	74	
	MCBCS	Mayo Clinic Breast Cancer Study	iCOGS	USA	53	
	NBCS	Norwegian Breast Cancer Study	iCOGS	Norway	22	70
	NBHS	The Nashville Breast Health Study	iCOGS	USA	125	118
	OSU	Ohio State University	iCOGS	USA	276	279
	RPCI	Roswell Park Cancer Institute	iCOGS	USA	136	132
	SBCS	Sheffield Breast Cancer Study	iCOGS	UK	3	
	SKKDKFZS	Städtisches Klinikum Karlsruhe and Deutsches Krebsforschungszentrum Breast Cancer Study	iCOGS	Germany	136	168
	SUCCESS C	Simultaneous Study of Docetaxel Based Anthracycline Free Adjuvant Treatment Evaluation, as well as Life Style Intervention Strategies	iCOGS	Germany	605	
	WASHU	Washington University	iCOGS	USA	87	
				TOTAL	**2148**	**1309**

Table S2. TN subjects with DASL and SNP data

	Sample type	Total	Post-QC samples		Excluding ER+ samples	
			All	**SNP data**	**All**	**SNP data**
ABCTB	10 µm sections	101	97	86	95	84
Demokritos	10 µm sections	139	137	117	127	109
HEBCS	10 µm sections	92	89	48	79	43
KBCP*	1 mm cores	40	37	35	32	30
MCBCS	10 µm sections	31	30	28	29	27
MCCS	10 µm sections	23	23	16	22	15
NBHS	10 µm sections	18	16	15	16	15
POSH	1 mm cores	121	107	106	104	103
SBCS	10 µm sections	36	34	33	32	32
SKK	10 µm sections	101	98	94	60	58
		702	**668**	**578**	**596**	**516**

Table S3. 78 Known breast cancer susceptibility variants

Locus	SNP	Platform	Proxy	Chr.	Postion36	Alleles	AF2	Source
PEX14	rs616488	GWAS +iCOGS		1	10488802	A/G	0.33	(1)
1p13.2	rs11552449	N/A	rs3761936	1	114249912	C/T	0.17	(1)
1p11.2	rs11249433	GWAS +iCOGS		1	120982136	A/G	0.41	(2)
LGR6	rs6678914	GWAS +iCOGS		1	200453799	G/A	0.41	(3)
MDM4	rs4245739	GWAS +iCOGS		1	202785465	A/C	0.26	(3)
2p24.1	rs12710696	GWAS +iCOGS		2	19184284	C/T	0.36	(3)
2q14.2	rs4849887	GWAS +iCOGS		2	120961592	C/T	0.098	(1)
2q31.1	rs2016394	GWAS +iCOGS		2	172681217	G/A	0.48	(1)
CDCA7	rs1550623	GWAS +iCOGS		2	173921140	A/G	0.16	(1)
CASP8	rs1045485	GWAS +iCOGS		2	201857834	C/G	0.13	(4)
2q35	rs13387042	GWAS +iCOGS		2	217614077	A/G	0.47	(5)
2q35	rs16857609	GWAS +iCOGS		2	218004753	C/T	0.26	(1)
3p26.2	rs6762644	GWAS +iCOGS		3	4717276	A/G	0.4	(1)
SLC4A7	rs4973768	GWAS +iCOGS		3	27391017	C/T	0.48	(6)
TGFBR2	rs12493607	GWAS +iCOGS		3	30657943	G/C	0.35	(1)
TET2	rs9790517	GWAS +iCOGS		4	106304227	C/T	0.23	(1)
ADAM29	rs6828523	GWAS +iCOGS		4	176083001	C/A	0.13	(1)
TERT	rs10069690	GWAS +iCOGS		5	1332790	C/T	0.27	(7)
TERT	rs7705526	N/A	N/A	5	1338974	C/A	0.33	(8)
TERT	rs2736108	iCOGS	N/A	5	1350488	C/T	0.29	(8)
5p12	rs10941679	GWAS +iCOGS		5	44742255	A/G	0.27	(9)
MAP3K1	rs889312	GWAS +iCOGS		5	56067641	A/C	0.29	(10)
RAB3C	rs10472076	GWAS +iCOGS		5	58219818	T/C	0.38	(1)
PDE4D	rs1353747	GWAS +iCOGS		5	58373238	T/G	0.095	(1)
EBF1	rs1432679	GWAS +iCOGS		5	158176661	T/C	0.43	(1)
FOXQ1	rs11242675	GWAS +iCOGS		6	1263878	T/C	0.39	(1)
RANBP1	rs204247	GWAS +iCOGS		6	13830502	A/G	0.43	(1)
6q14.1	rs17529111	GWAS +iCOGS		6	82185105	T/C	0.22	(1)
ESR1	rs3757318	GWAS +iCOGS		6	151955806	G/A	0.07	(11)
ESR1	rs2046210	GWAS +iCOGS		6	151990059	G/A	0.35	(12)
7q35	rs720475	GWAS +iCOGS		7	143705862	G/A	0.25	(1)
8p21.1	rs9693444	GWAS +iCOGS		8	29565535	C/A	0.32	(1)
8q21.11	rs6472903	GWAS +iCOGS		8	76392856	T/G	0.18	(1)
HNF4G	rs2943559	GWAS +iCOGS		8	76580492	A/G	0.07	(1)
8q24	rs13281615	GWAS +iCOGS		8	128424800	A/G	0.42	(10)
8q24.21	rs11780156	GWAS +iCOGS		8	129263823	C/T	0.16	(1)
CDKN2A/B	rs1011970	GWAS +iCOGS		9	22052134	G/T	0.17	(11)
9q31.2	rs10759243	GWAS +iCOGS		9	109345936	C/A	0.39	(1)
9q31	rs865686	GWAS +iCOGS		9	109928299	T/G	0.37	(13)
ANKRD16	rs2380205	GWAS +iCOGS		10	5926740	C/T	0.44	(11)
DNAJC1	rs7072776	GWAS +iCOGS		10	22072948	G/A	0.29	(1)
DNAJC1	rs11814448	GWAS +iCOGS		10	22355849	A/C	0.02	(1)
ZNF365	rs10995190	GWAS +iCOGS		10	63948688	G/A	0.15	(11)

ZMIZ1	rs704010	GWAS +iCOGS		10	80511154	C/T	0.39	(11)
TCF7L2	rs7904519	GWAS +iCOGS		10	114763917	A/G	0.46	(1)
10q26.12	rs11199914	GWAS +iCOGS		10	123083891	C/T	0.32	(1)
FGFR2	rs2981579	GWAS +iCOGS		10	123327325	G/A	0.43	(11)
FGFR2	rs2981582	GWAS +iCOGS		10	123342307	G/A	0.41	(10)
LSP1	rs3817198	GWAS +iCOGS		11	1865582	T/C	0.32	(10)
11q13.1	rs3903072	GWAS +iCOGS		11	65339642	G/T	0.47	(1)
CCDN1	rs614367	GWAS +iCOGS		11	69037945	C/T	0.16	(11)
CCND1	rs554219	GWAS +iCOGS		11	69040823	C/G	0.14	(14)
11q24.3	rs11820646	GWAS +iCOGS		11	128966381	C/T	0.41	(1)
CCND1	rs75915166	N/A	N/A	11	69379161	A/C	0.31	(14)
12p13.1	rs12422552	GWAS +iCOGS		12	14305198	G/C	0.26	(1)
PTHLH	rs10771399	GWAS +iCOGS		12	28046347	A/G	0.11	(15)
NTN4	rs17356907	GWAS +iCOGS		12	94551890	A/G	0.3	(1)
12q24	rs1292011	GWAS +iCOGS		12	114320905	A/G	0.41	(15)
BRCA2	rs11571833	GWAS +iCOGS		13	31870626	A/T	0.008	(1)
PAX9	rs2236007	GWAS +iCOGS		14	36202520	G/A	0.21	(1)
RAD51L1	rs2588809	GWAS +iCOGS		14	67730181	C/T	0.16	(1)
RAD51L1	rs999737	GWAS +iCOGS		14	68104435	C/T	0.22	(2)
CCDC88C	rs941764	GWAS +iCOGS		14	90910822	A/G	0.34	(1)
TOX3	rs3803662	GWAS +iCOGS		16	51143842	G/A	0.29	(10)
FTO	rs17817449	GWAS +iCOGS		16	52370868	T/G	0.4	(1)
FTO	rs11075995	GWAS +iCOGS		16	52412792	T/A	0.24	(3)
CDYL2	rs13329835	GWAS +iCOGS		16	79208306	A/G	0.22	(1)
COX11	rs6504950	GWAS +iCOGS		17	50411470	G/A	0.27	(6)
18q11.2	rs527616	GWAS +iCOGS		18	22591422	G/C	0.38	(1)
CHST9	rs1436904	GWAS +iCOGS		18	22824665	T/G	0.4	(1)
MERIT40	rs8170	GWAS +iCOGS		19	17250704	G/A	0.19	(16)
MERIT40	rs2363956	GWAS +iCOGS		19	17255124	G/T	0.49	(16)
SSBP4	rs4808801	GWAS +iCOGS		19	18432141	A/G	0.35	(1)
19q13.31	rs3760982	GWAS +iCOGS		19	48978353	G/A	0.46	(1)
RALY	rs2284378	N/A	rs9753679	20	32051756	C/T	0.28	(17)
NRIP1	rs2823093	GWAS +iCOGS		21	15442703	G/A	0.26	(15)
22q12.2	rs132390	iCOGS	N/A	22	27951477	T/C	0.036	(1)
MKL1	rs6001930	iCOGS	rs6001913	22	39206180	T/C	0.11	(1)

Table S4. SNPs associated with TNBC in 2-stage GWAS

SNP	G/I	Chr.	Position	Locus	Allele	OR	95% CI	P-value
rs4245739	I	1	202785465	*MDM4*	C	1.19	1.11-1.29	4.0×10^{-06}
rs3757318	G	6	151955806	*ESR1*	A	1.33	1.17-1.51	9.2×10^{-06}
rs10484919	G	6	152016115	*ESR1*	A	1.31	1.16-1.47	5.7×10^{-06}
rs2619434	G	12	28056724	*PTHLH*	A	0.84	0.77-0.91	1.0×10^{-05}
rs8170	G	19	17250704	19p13.1	A	1.26	1.16-1.37	1.3×10^{-07}

Table S5. SNPs associated with TNBC ($p<1\times10^{-3}$) in 2-stage GWAS, excluding known 78 loci

SNP	G/I	Chr.	Position	Genes	Allele	MAF	OR	95% CI	p-value
rs9761827	G	4	138635961	PCDH18	A	0.38	1.17	(1.09-1.26)	1.1x10-5
rs4425715	G	7	54233081	HPVC1	G	0.33	1.17	(1.09-1.26)	1.7 x10-5
rs1353868	G	3	174143933	SPATA16	A	0.36	1.17	(1.09-1.25)	2.6 x10-5
rs3855959	G	1	46406461	PIK3R3:TSPAN1:POMGNT1:C1orf190	A	0.40	0.86	(0.80-0.92)	3.0 x10-5
rs3810295	G	19	51830486	CALM3:PTGIR:GNG8:DACT3:PRKD2	A	0.13	1.24	(1.12-1.37)	4.3 x10-5
rs9257181	G	6	28862499	TRNAA-UGC:TRNAF-GAA:TRNAA-AGC:NOL5BP	A	0.28	1.17	(1.08-1.26)	4.9 x10-5
rs230310	G	1	40080306	TRIT1	A	0.23	1.18	(1.09-1.28)	6.0 x10-5
rs4717599	G	7	70607962	WBSCR17	G	0.27	0.85	(0.79-0.92)	6.6 x10-5
rs7020507	G	9	1705820	SMARCA2	G	0.14	0.81	(0.74-0.90)	6.9 x10-5
rs7790719	G	7	3684577	SDK1	A	0.28	0.86	(0.80-0.93)	8.0 x10-5

Table S6. 74 known breast cancer susceptibility loci and risk of TNBC compared to ER-negative and overall breast cancer risk estimates from BCAC

SNP	G/I	Chr.	Position	Locus	Allele	TN OR	TN 95% CI	TN P-value	ER-negative (1) OR	ER-negative (1) 95% CI	ER-negative (1) P-value	Overall (1) OR	Overall (1) 95% CI	Overall (1) P-value
rs616488	G	1	10488802	PEX14	G	0.91	(0.85-0.98)	9.73×10^{-03}	0.90	(0.87-0.94)	4.44×10^{-07}	0.94	(0.92-0.96)	2.13×10^{-08}
rs11249433	G	1	120982136	1p11.2	G	1.03	(0.96-1.10)	0.49	1.00	(0.96-1.04)	0.97	1.09	(1.07-1.11)	7.66×10^{-19}
rs6678914	G	1	200453799	LGR6	A	0.90	(0.84-0.97)	3.31×10^{-03}	0.92	(0.89-0.96)	2.83×10^{-05}	0.99	(0.97-1.01)	0.43
rs4245739	I	1	202785465	MDM4	C	1.19	(1.11-1.29)	4.00×10^{-06}	1.16	(1.11-1.20)	4.30×10^{-12}	1.03	(1.01-1.05)	7.03×10^{-03}
rs12710696	I	2	19184284	2p24.1	A	1.11	(1.04-1.19)	3.51×10^{-03}	1.10	(1.06-1.14)	8.56×10^{-07}	1.04	(1.02-1.06)	1.08×10^{-04}
rs4849887	G	2	120961592	2q14.2	A	0.89	(0.79-1.00)	0.041	0.91	(0.86-0.97)	5.94×10^{-03}	0.91	(0.88-0.94)	8.23×10^{-09}
rs2016394	G	2	172681217	2q31.1	A	1.10	(1.03-1.18)	6.90×10^{-03}	0.99	(0.96-1.03)	0.77	0.95	(0.93-0.97)	3.02×10^{-07}
rs1550623	G	2	173921140	CDCA7	G	0.94	(0.85-1.03)	0.16	0.95	(0.90-1.00)	0.046	0.94	(0.92-0.97)	2.08×10^{-05}
rs1045485	I	2	201857834	CASP8	C	1.00	(0.90-1.11)	0.99	0.97	(0.91-1.02)	0.22	0.97	(0.94-1.00)	0.037
rs13387042	G	2	217614077	2q35	G	0.93	(0.87-1.00)	0.049	0.96	(0.92-0.99)	0.021	0.88	(0.86-0.89)	3.04×10^{-41}
rs16857609	I	2	218004753	2q35	A	1.08	(1.00-1.16)	0.060	1.08	(1.03-1.12)	3.36×10^{-04}	1.08	(1.05-1.10)	7.23×10^{-12}
rs6762644	G	3	4717276	3p26.2	G	0.97	(0.90-1.04)	0.38	1.02	(0.98-1.06)	0.32	1.07	(1.04-1.09)	1.83×10^{-10}
rs4973768	G	3	27391017	SLC4A7	A	1.06	(0.99-1.14)	0.075	1.05	(1.01-1.09)	0.011	1.10	(1.08-1.12)	1.36×10^{-21}
rs12493607	I	3	30657943	TGFBR2	C	1.00	(0.93-1.07)	0.89	1.01	(0.97-1.05)	0.52	1.06	(1.04-1.08)	6.86×10^{-08}
rs9790517	I	4	106304227	TET2	A	1.00	(0.92-1.09)	0.94	1.03	(0.98-1.07)	0.22	1.05	(1.03-1.07)	2.71×10^{-05}
rs6828523	I	4	176083001	ADAM29	A	0.84	(0.75-0.93)	1.33×10^{-03}	1.01	(0.96-1.07)	0.66	0.89	(0.87-0.92)	1.22×10^{-13}
rs10069690	I	5	1332790	TERT	A	1.24	(1.14-1.34)	1.43×10^{-07}	1.16	(1.11-1.21)	1.69×10^{-12}	1.06	(1.04-1.09)	2.83×10^{-08}
rs2736108[a]	G	5	1350488	TERT	T	0.77	(0.69-0.87)	8.33×10^{-6}	0.89[b]	(0.83-0.93)	1.41×10^{-8}	0.94[b]	(0.92-0.95)	6.73×10^{-9}
rs10941679	I	5	44742255	5p12	G	1.02	(0.94-1.11)	0.59	1.04	(1.00-1.08)	0.080	1.13	(1.11-1.16)	3.57×10^{-29}
rs889312	G	5	56067641	MAP3K1	C	1.01	(0.94-1.09)	0.76	1.05	(1.01-1.10)	0.011	1.12	(1.10-1.15)	3.56×10^{-27}
rs10472076	I	5	58219818	RAB3C	G	0.96	(0.89-1.03)	0.24	1.05	(1.02-1.10)	5.87×10^{-03}	1.05	(1.03-1.07)	8.35×10^{-07}
rs1353747	G	5	58373238	PDE4D	C	1.01	(0.90-1.14)	0.89	0.91	(0.86-0.98)	6.65×10^{-03}	0.92	(0.89-0.95)	1.29×10^{-06}
rs1432679	G	5	158176661	EBF1	G	1.10	(1.02-1.17)	8.62×10^{-03}	1.08	(1.04-1.12)	2.36×10^{-05}	1.07	(1.05-1.09)	3.29×10^{-12}
rs11242675	G	6	1263878	FOXQ1	G	1.00	(0.93-1.07)	0.98	0.94	(0.90-0.98)	1.54×10^{-03}	0.95	(0.93-0.97)	4.29×10^{-08}
rs204247	G	6	13830502	RANBP1	G	1.03	(0.96-1.11)	0.36	1.01	(0.97-1.05)	0.58	1.05	(1.03-1.07)	2.67×10^{-07}
rs17529111	I	6	82185105	6q14.1	G	1.04	(0.96-1.13)	0.31	1.04	(1.00-1.09)	0.054	1.06	(1.04-1.09)	3.19×10^{-07}
rs17530068	G	6	82249828	6q14	G	1.07	(0.99-1.16)	0.093	1.05	(1.00-1.09)	0.034	1.06	(1.03-1.08)	1.97×10^{-06}
rs3757318	G	6	151955806	ESR1	A	1.33	(1.17-1.51)	9.25×10^{-06}	1.22	(1.14-1.31)	3.95×10^{-09}	1.16	(1.12-1.20)	1.09×10^{-15}
rs2046210	I	6	151990059	ESR1	A	1.16	(1.08-1.24)	5.26×10^{-05}	1.16	(1.12-1.21)	2.36×10^{-14}	1.08	(1.06-1.10)	1.38×10^{-14}
rs720475	G	7	143705862	7q35	A	1.02	(0.94-1.10)	0.62	0.99	(0.95-1.03)	0.58	0.94	(0.92-0.96)	2.49×10^{-08}
rs9693444	G	8	29565535	8p21.1	A	1.07	(0.99-1.15)	0.087	1.09	(1.05-1.13)	2.25×10^{-05}	1.07	(1.05-1.09)	4.61×10^{-11}
rs6472903	I	8	76392856	8q21.11	C	0.98	(0.90-1.08)	0.70	0.93	(0.89-0.98)	3.94×10^{-03}	0.91	(0.89-0.93)	3.08×10^{-13}
rs2943559	I	8	76580492	HNF4G	G	1.10	(0.97-1.24)	0.13	1.08	(1.01-1.16)	0.030	1.13	(1.09-1.17)	3.31×10^{-11}
rs13281615	G	8	128424800	8q24	G	1.01	(0.95-1.09)	0.71	1.02	(0.98-1.06)	0.28	1.10	(1.08-1.12)	1.87×10^{-20}

						OR	CI	p	OR	CI	p	OR	CI	p
rs11780156	G	8	129263823	8q24.21	A	1.03	(0.95-1.13)	0.47	1.06	(1.01-1.11)	0.024	1.07	(1.05-1.10)	3.06×10^{-08}
rs1011970	G	9	22052134	*CDKN2A/B*	A	1.08	(0.99-1.18)	0.075	1.12	(1.06-1.17)	6.58×10^{-06}	1.05	(1.03-1.08)	4.04×10^{-05}
rs10759243	I	9	109345936	9q31.2	A	1.00	(0.93-1.08)	0.97	1.01	(0.97-1.05)	0.70	1.05	(1.03-1.08)	1.02×10^{-06}
rs865686	G	9	109928299	9q31	C	1.03	(0.96-1.11)	0.41	0.98	(0.95-1.02)	0.35	0.90	(0.88-0.91)	6.25×10^{-28}
rs2380205	G	10	5926740	*ANKRD16*	A	1.00	(0.94-1.07)	0.92	1.00	(0.96-1.04)	0.91	0.98	(0.96-1.00)	0.077
rs7072776	G	10	22072948	*DNAJC1*	A	0.96	(0.89-1.03)	0.24	0.94	(0.90-0.98)	3.94×10^{-03}	1.07	(1.05-1.09)	8.98×10^{-10}
rs10995190	G	10	63948688	*ZNF365*	A	0.93	(0.85-1.03)	0.16	0.87	(0.83-0.92)	2.52×10^{-07}	0.86	(0.84-0.88)	6.15×10^{-29}
rs704010	G	10	80511154	*ZMIZ1*	A	1.04	(0.97-1.12)	0.27	1.03	(0.99-1.07)	0.092	1.08	(1.06-1.10)	2.96×10^{-15}
rs7904519	G	10	114763917	*TCF7L2*	G	1.12	(1.05-1.20)	9.95×10^{-04}	1.06	(1.02-1.10)	3.18×10^{-03}	1.06	(1.04-1.08)	1.25×10^{-09}
rs11199914	G	10	123083891	10q26.12	A	1.04	(0.97-1.12)	0.28	1.02	(0.98-1.06)	0.35	0.95	(0.93-0.97)	1.44×10^{-06}
rs2981579	G	10	123327325	*FGFR2*	A	0.99	(0.93-1.06)	0.81	1.03	(0.99-1.07)	0.12	1.27	(1.24-1.29)	5.90×10^{-129}
rs2981582	I	10	123342307	*FGFR2*	A	0.98	(0.92-1.05)	0.61	1.02	(0.98-1.06)	0.27	1.26	(1.23-1.28)	1.71×10^{-117}
rs3817198	G	11	1865582	*LSP1*	G	1.06	(0.99-1.14)	0.10	1.06	(1.02-1.10)	5.81×10^{-03}	1.07	(1.05-1.09)	5.39×10^{-10}
rs3903072	I	11	65339642	11q13.1	A	0.92	(0.86-0.99)	0.024	0.97	(0.93-1.01)	0.099	0.94	(0.93-0.96)	2.89×10^{-09}
rs614367	G	11	69037945	*CCDN1*	A	1.02	(0.92-1.12)	0.75	1.02	(0.97-1.08)	0.41	1.21	(1.18-1.24)	5.21×10^{-48}
rs554219	I	11	69040823	*CCND1*	G	0.94	(0.85-1.04)	0.20	1.02	(0.96-1.08)	0.49	1.27	(1.23-1.30)	3.72×10^{-62}
rs11820646	I	11	128966381	11q24.3	A	0.92	(0.86-0.98)	0.016	0.96	(0.92-1.00)	0.028	0.95	(0.93-0.97)	2.44×10^{-07}
rs12422552	I	12	14305198	12p13.1	C	1.13	(1.04-1.21)	2.70×10^{-03}	1.04	(1.00-1.08)	0.080	1.05	(1.03-1.07)	2.47×10^{-05}
rs10771399	I	12	28046347	*PTHLH*	G	0.72	(0.64-0.80)	1.55×10^{-08}	0.83	(0.78-0.89)	3.35×10^{-09}	0.85	(0.83-0.88)	5.31×10^{-25}
rs17356907	G	12	94551890	*NTN4*	G	0.90	(0.84-0.97)	7.55×10^{-03}	0.94	(0.90-0.98)	2.27×10^{-03}	0.91	(0.89-0.93)	1.20×10^{-18}
rs1292011	G	12	114320905	12q24	G	1.08	(1.01-1.16)	0.035	0.98	(0.94-1.02)	0.31	0.92	(0.90-0.94)	6.19×10^{-17}
rs11571833	I	13	31870626	*BRCA2*	T	1.44	(1.05-1.96)	0.023	1.44	(1.20-1.71)	5.88×10^{-05}	1.26	(1.14-1.39)	5.36×10^{-06}
rs2236007	I	14	36202520	*PAX9*	A	0.99	(0.91-1.07)	0.75	0.96	(0.92-1.01)	0.096	0.93	(0.90-0.95)	1.69×10^{-10}
rs2588809	I	14	67730181	*RAD51L1*	A	0.91	(0.83-1.00)	0.041	1.01	(0.96-1.06)	0.78	1.08	(1.05-1.11)	4.71×10^{-09}
rs999737	G	14	68104435	*RAD51L1*	A	0.95	(0.88-1.03)	0.22	0.95	(0.91-0.99)	0.015	0.92	(0.90-0.94)	3.73×10^{-13}
rs941764	I	14	90910822	*CCDC88C*	G	1.03	(0.95-1.10)	0.50	1.03	(0.99-1.07)	0.091	1.06	(1.04-1.09)	1.02×10^{-09}
rs3803662	G	16	51143842	*TOX3*	A	1.09	(1.01-1.17)	0.022	1.14	(1.10-1.19)	1.16×10^{-10}	1.24	(1.21-1.27)	1.38×10^{-88}
rs17817449	I	16	52370868	*FTO*	C	0.99	(0.92-1.06)	0.68	0.91	(0.87-0.94)	5.07×10^{-07}	0.93	(0.91-0.95)	1.41×10^{-12}
rs11075995	I	16	52412792	*FTO*	A	1.08	(1.00-1.17)	0.065	1.11	(1.06-1.16)	2.13×10^{-06}	1.04	(1.02-1.07)	1.19×10^{-04}
rs13329835	G	16	79208306	*CDYL2*	G	1.03	(0.95-1.11)	0.51	1.02	(0.98-1.07)	0.30	1.08	(1.06-1.11)	1.48×10^{-11}
rs6504950	I	17	50411470	*COX11*	A	0.96	(0.89-1.04)	0.33	0.97	(0.93-1.01)	0.16	0.94	(0.92-0.96)	2.27×10^{-09}
rs527616	I	18	22591422	18q11.2	C	0.95	(0.88-1.02)	0.14	0.98	(0.94-1.02)	0.24	0.95	(0.93-0.97)	2.53×10^{-07}
rs1436904	G	18	22824665	*CHST9*	C	0.99	(0.93-1.07)	0.84	1.00	(0.96-1.04)	0.86	0.95	(0.94-0.97)	3.27×10^{-06}
rs8170	G	19	17250704	19p13.1	A	1.26	(1.16-1.37)	1.26×10^{-07}	1.14	(1.09-1.19)	1.26×10^{-08}	1.04	(1.01-1.06)	2.74×10^{-03}
rs2363956	G	19	17255124	19p13.1	C	0.82	(0.77-0.88)	2.33×10^{-08}	1.13	(1.09-1.17)	1.38×10^{-10}	1.03	(1.01-1.05)	1.86×10^{-03}
rs4808801	G	19	18432141	*SSBP4*	G	1.03	(0.96-1.11)	0.40	0.92	(0.88-0.95)	1.88×10^{-05}	0.93	(0.91-0.95)	4.70×10^{-13}
rs3760982	G	19	48978353	19q13.31	A	0.99	(0.93-1.06)	0.85	1.04	(1.00-1.08)	0.026	1.06	(1.04-1.08)	1.68×10^{-08}
rs2823093	G	21	15442703	*NRIP1*	A	1.04	(0.96-1.12)	0.35	0.97	(0.93-1.02)	0.21	0.92	(0.90-0.95)	1.57×10^{-12}
rs132390[a]	G	22	27951477	22q12.2	C	1.16	(0.89-1.52)	0.28	1.08	(0.98-1.19)	0.11	1.12	(1.07-1.18)	3.1×10^{-9}
rs6001930[a]	G	22	39206180	*MLK1*	C	1.21	(1.02-1.43)	0.025	1.10	(1.04-1.17)	1.1×10^{-3}	1.12	(1.09-1.16)	8.8×10^{-19}

a Genotyped in stage 2 only on the iCOGS platform (2,148 cases, 1,309 controls)

b Overall and ER-negative breast cancer risk results for rs2736108 from Bojesen, et al. (8)

Table S7. Additional significant SNPs in the known breast cancer susceptibility loci

Risk SNP	Reported SNP	R2 with reported SNP	G/I	Locus	Chr.	Position	Allele	OR	95% CI	P-value
a) SNPs in regions where reported SNP has p<0.05										
rs9397437	rs2046210; rs3757318	0.11; 0.38	I	*ESR1*	6	151994025	A	1.42	(1.25-1.61)	8.9×10^{-8}
rs620405	rs616488	0.73	G	*PEX14*	1	10477381	A	0.86	(0.80-0.93)	1.0×10^{-4}
b) SNPs in regions where reported SNP has p>0.05										
rs3731711	rs1045485	0.93	I	*CASP8*	2	201921306	G	0.84	(0.76-0.92)	1.4×10^{-4}
c) SNPs in regions where reported SNP not genotyped										
rs6142050	rs2284378	0.56	G	*RALY*	20	31990789	G	1.11	(1.03-1.19)	3.8×10^{-3}

Table S8. Cis-eQTL associations with known TN risk SNPs

eQTL SNP	eQTL gene	eQTL probe	chr	pos	t.stat	p.value	Risk locus
rs620405	*UBIAD1*	ILMN_1651872	1	10477381	-3.13	1.85E-03	*PEX14*
rs620405	*DFFA*	ILMN_2385220	1	10477381	-2.87	4.29E-03	*PEX14*
rs620405	*PGD*	ILMN_1794165	1	10477381	2.39	1.70E-02	*PEX14*
rs620405	*CASZ1*	ILMN_2340202	1	10477381	-2.28	2.29E-02	*PEX14*
rs620405	*CLSTN1*	ILMN_1720181	1	10477381	-2.14	3.30E-02	*PEX14*
rs620405	*C1orf200*	ILMN_1703119	1	10477381	-1.98	4.80E-02	*PEX14*
rs616488	*UBIAD1*	ILMN_1651872	1	10488802	2.67	7.72E-03	*PEX14*
rs616488	*CTNNBIP1*	ILMN_1688103	1	10488802	-2.30	2.20E-02	*PEX14*
rs616488	*CASZ1*	ILMN_2340202	1	10488802	2.08	3.76E-02	*PEX14*
rs6678914	*LGR6*	ILMN_1662362	1	200453799	2.16	3.09E-02	*LGR6*
rs3795598	*CHI3L1*	ILMN_1772289	1	200463784	-2.25	2.48E-02	*LGR6*
rs4245739	*LRRN2*	ILMN_1781841	1	202785465	-2.46	1.41E-02	*MDM4*
rs4245739	*NUAK2*	ILMN_1789793	1	202785465	-2.35	1.93E-02	*MDM4*
rs4245739	*REN*	ILMN_1742272	1	202785465	-2.06	3.99E-02	*MDM4*
rs4849887	*SCTR*	ILMN_1772537	2	120961592	-1.96	5.00E-02	2q14.2
rs2016394	*DYNC1I2*	ILMN_1773847	2	172681217	-2.85	4.51E-03	2q31.1
rs2016394	*ZAK*	ILMN_1698803	2	172681217	-2.46	1.40E-02	2q31.1
rs3731711	*AOX2P*	ILMN_1789676	2	201921306	-2.12	3.41E-02	*CASP8*
rs13387042	*TNS1*	ILMN_1807919	2	217614077	2.60	9.59E-03	2q35
rs10069690	*ZDHHC11*	ILMN_1694514	5	1332790	1.98	4.77E-02	*TERT*
rs1432679	*RNF145*	ILMN_1710906	5	158176661	2.47	1.39E-02	*EBF1*
rs9397437	*ZBTB2*	ILMN_1766247	6	151994025	2.04	4.14E-02	*ESR1*
rs2807985	*MLLT10*	ILMN_1743538	10	22270480	2.01	4.47E-02	*DNAJC1*
rs7904519	*ZDHHC6*	ILMN_2046003	10	114763917	-1.97	4.99E-02	*TCF7L2*
rs3903072	*CTSW*	ILMN_1794364	11	65339642	2.63	8.79E-03	11q13.1
rs3903072	*SART1*	ILMN_1680145	11	65339642	2.50	1.27E-02	11q13.1
rs3903072	*ACTN3*	ILMN_1665691	11	65339642	-2.32	2.09E-02	11q13.1
rs3903072	*SCYL1*	ILMN_1731991	11	65339642	-2.03	4.31E-02	11q13.1
rs3903072	*EHD1*	ILMN_1651832	11	65339642	2.00	4.65E-02	11q13.1
rs3903072	*CCDC85B*	ILMN_1657332	11	65339642	-1.97	4.96E-02	11q13.1
rs3903072	*C11orf85*	ILMN_2182850	11	65339642	1.97	4.99E-02	11q13.1
rs11820646	*ST14*	ILMN_1699887	11	128966381	3.08	2.20E-03	11q24.3
rs11820646	*APLP2*	ILMN_2081465	11	128966381	2.91	3.76E-03	11q24.3
rs11820646	*NFRKB*	ILMN_1718990	11	128966381	2.46	1.41E-02	11q24.3
rs11820646	*APLP2*	ILMN_1710482	11	128966381	2.43	1.56E-02	11q24.3
rs12422552	*GRIN2B*	ILMN_3307714	12	14305198	2.83	4.88E-03	12p13.1
rs12422552	*C12orf36*	ILMN_1755414	12	14305198	2.10	3.58E-02	12p13.1
rs11055891	*PDE6H*	ILMN_1702965	12	14312379	-2.58	1.00E-02	12p13.1
rs10771399	*REP15*	ILMN_1665884	12	28046347	-2.79	5.48E-03	*PTHLH*
rs17356907	*VEZT*	ILMN_2141398	12	94551890	-2.32	2.05E-02	*NTN4*
rs10850494	*TBX5*	ILMN_2282379	12	114311094	2.02	4.44E-02	12q24
rs2588809	*ZFYVE26*	ILMN_1798061	14	67730181	-2.07	3.87E-02	*RAD51L1*
rs8170	*PLVAP*	ILMN_2194577	19	17250704	-2.08	3.84E-02	19p13.1
rs2363956	*IL12RB1*	ILMN_1699908	19	17255124	-2.42	1.57E-02	19p13.1
rs2363956	*GTPBP3*	ILMN_1686587	19	17255124	-2.15	3.18E-02	19p13.1
rs1864112	*CPAMD8*	ILMN_1726250	19	17309960	2.38	1.78E-02	19p13.1

rs6142050	*PXMP4*	ILMN_3249742	20	31990789	-2.65	8.28E-03	*RALY*
rs6142050	*PXMP4*	ILMN_1771728	20	31990789	-2.19	2.88E-02	*RALY*
rs6142050	*PXMP4*	ILMN_3250812	20	31990789	-2.18	3.01E-02	*RALY*
rs6001913	*SLC25A17*	ILMN_1737312	22	39166699	3.02	2.64E-03	*MKL1*
rs6001913	*TNRC6B*	ILMN_1726786	22	39166699	2.98	3.00E-03	*MKL1*

Table S9. Functional annotation by Rhie, et al. (2013) in TN risk loci

SNP	Chr.	Positon	Locus	Allele	Description	Nearest gene	Number of SNPs in region (R2>0.5) overlapping with feature (18)		
							TSS	Enhancer	Exon
rs2046210	6	151990059	ESR1	A	intergenic	C6orf97	1	4	1
rs10771399	12	28046347	PTHLH	G	intergenic	PTHLH	1	62	
rs3803662	16	51143842	TOX3	A	intergenic	TOX3	1		
rs6678914	1	200453799	LGR6	A	intron	LGR6	2	10	1
rs2363956	19	17255124	19p13.1	C	exon (missense)	ANKLE1	2	2	2
rs3903072	11	65339642	11q13.1	A	intergenic	SNX32; OVOL1	2	11	3
rs2016394	2	172681217	2q31.1	A	intergenic	CDCA7	3		
rs4245739	1	202785465	MDM4	C	intron (3'utr)	MDM4	8	21	
rs616488	1	10488802	PEX14	G	intron	PEX14		16	1
rs1432679	5	158176661	EBF1	G	intron	EBF1			1
rs11571833	13	31870626	BRCA2	T	exon (nonsense)	BRCA2			
rs11820646	11	128966381	11q24.3	A	intergenic	CCND1		1	
rs17356907	12	94551890	NTN4	G	intergenic	NTN4		2	
rs4849887	2	120961592	2q14.2	A	intergenic	INHBB		3	
rs1292011	12	114320905	12q24	G	intergenic	MED13L		5	
rs12422552	12	14305198	12p13.1	C	intergenic	ATF7IP		9	
rs12710696	2	19184284	2p24.1	A	intergenic	OSR1		15	
rs13387042	2	217614077	2q35	G	intergenic	TNP1			
rs10069690	5	1332790	TERT	A	intron	TERT		1	
rs7904519	10	114763917	TCF7L2	G	intron	TCF7L2		36	
rs2588809	14	67730181	RAD51L1	A	intron	RAD51B		39	
rs6001930	22	39206180	MLK1	C	intron	MLK1		88	
rs6828523	4	176083001	ADAM29	A	intron	ADAM29			
Rs3757315	6	151955806	ESR1	A	intron	C6orf97			

Table S10. Cis-eQTL associations with SNPs in TN risk loci

Chr.	eQTL SNP	eQTL gene	t-statistic	eQTL p-value	Locus
1	rs11586387	*KLHDC8A*	-3.75	2.0E-04	*MDM4*
2	rs11892687	*IGFBP2*	-3.61	3.3E-04	2q35
2	rs7589722	*IGFBP2*	3.46	5.8E-04	2q35
2	rs10490444	*IGFBP2*	-3.48	5.5E-04	2q35
2	rs7579388	*PECR*	3.35	8.7E-04	2q35
2	rs6738142	*HAT1*	0.24	9.11×10^{-6}	2q31.1
2	rs2008518	*ZAK*	-0.074	9.36×10^{-5}	2q31.1
2	rs13016963	*ALS2CR12*	3.39	7.5E-04	*CASP8*
2	rs9288316	*ALS2CR12*	-3.44	6.3E-04	*CASP8*
2	rs1035142	*ALS2CR12*	3.38	7.8E-04	*CASP8*
2	rs1045494	*FZD7*	3.34	9.0E-04	*CASP8*
5	rs4246742	*SLC9A3*	3.38	7.7E-04	*TERT*
5	rs4246742	*SLC12A7*	3.33	9.4E-04	*TERT*
5	rs4246742	*SLC9A3*	3.38	7.7E-04	*TERT*
5	rs4246742	*SLC12A7*	3.33	9.4E-04	*TERT*
6	rs1871859	*AKAP12*	-3.92	1.0E-04	*ESR1*
10	rs7085532	*ACSL5*	3.64	3.0E-04	*TCF7L2*
10	rs17746916	*LOC143188*	3.60	3.5E-04	*TCF7L2*
10	rs290488	*ZDHHC6*	3.64	3.0E-04	*TCF7L2*
11	rs10896050	*SNX32*	3.78	1.8E-04	11q13.1
11	rs630303	*CTSW*	3.55	4.3E-04	11q13.1
11	rs656040	*CTSW*	-3.55	4.3E-04	11q13.1
11	rs11227332	*CTSW*	3.85	1.3E-04	11q13.1
11	rs665306	*CTSW*	-3.49	5.2E-04	11q13.1
11	rs11227306	*CTSW*	-3.45	6.1E-04	11q13.1
11	rs622614	*CTSW*	-5.61	3.3E-08	11q13.1
11	rs13817	*CTSW*	3.57	3.9E-04	11q13.1
11	rs10896050	*CTSW*	-3.90	1.1E-04	11q13.1
11	rs10896050	*MRPL11*	4.14	4.0E-05	11q13.1
12	rs11067547	*TBX3*	3.44	6.3E-04	12q24
12	rs2347230	*PTHLH*	0.47	5.67×10^{-5}	*PTHLH*
12	rs10843001	*PTHLH*	0.39	7.28×10^{-5}	*PTHLH*
12	rs16932270	*PPFIBP1*	-0.41	5.30×10^{-6}	*PTHLH*
12	rs10777711	*VEZT*	-3.38	7.8E-04	*NTN4*
12	rs7963386	*VEZT*	-3.60	3.5E-04	*NTN4*
13	rs206119	*B3GALTL*	-3.53	4.6E-04	*BRCA2*
13	rs9567670	*KL*	3.40	7.3E-04	*BRCA2*
14	rs10137893	*EXD2*	3.63	3.2E-04	*RAD51L1*
19	rs17533903	*NR2F6*	-0.34	6.45×10^{-5}	19p13.1
19	rs17454516	*FAM32A*	-0.16	6.51×10^{-5}	19p13.1
19	rs17533903	*SLC35E1*	-0.39	6.20×10^{-5}	19p13.1

Table S11. Linkage disequilibrium (R^2 >0.1) between eQTL SNPs in TN risk loci and candidate functional SNPs in exons identified by Rhie, et al.

Chr.	eQTL SNP	eQTL gene	Locus	Exon SNP	R^2 with eQTL SNP	Gene (exon)	Result
11	rs10896050	*SNX32, CTSW, MRPL11*	11q13.1	rs637571	0.174	*FOSL1*	synonomous
				rs1058068	0.124	*FOSL1*	synonomous
				rs633800	0.137	*EFEMP2*	synonomous
11	rs11227306	*CTSW*	11q13.1	rs637571	0.272	*FOSL1*	synonomous
				rs1058068	0.299	*FOSL1*	synonomous
				rs633800	0.467	*EFEMP2*	synonomous
11	rs11227332	*CTSW*	11q13.1	rs637571	0.303	*FOSL1*	synonomous
				rs1058068	0.236	*FOSL1*	synonomous
				rs633800	0.241	*EFEMP2*	synonomous
11	rs13817	*CTSW*	11q13.1	rs637571	0.188	*FOSL1*	synonomous
				rs1058068	0.278	*FOSL1*	synonomous
				rs633800	0.458	*EFEMP2*	synonomous
11	rs622614	*CTSW*	11q13.1	rs633800	0.219	*EFEMP2*	synonomous
				rs1058068	0.122	*FOSL1*	synonomous
11	rs630303	*CTSW*	11q13.1	rs637571	0.188	*FOSL1*	synonomous
				rs1058068	0.278	*FOSL1*	synonomous
				rs633800	0.458	*EFEMP2*	synonomous
11	rs656040	*CTSW*	11q13.1	rs637571	0.211	*FOSL1*	synonomous
				rs1058068	0.254	*FOSL1*	synonomous
				rs633800	0.422	*EFEMP2*	synonomous
11	rs665306	*CTSW*	11q13.1	rs637571	0.188	*FOSL1*	synonomous
				rs1058068	0.278	*FOSL1*	synonomous
				rs633800	0.458	*EFEMP2*	synonomous

Table S12. Linkage disequilibrium (R^2 >0.1) between eQTL SNPs in TN risk loci and candidate functional SNPs in transcroiption start sites identified by Rhie, et al.

Chr.	eQTL SNP	eQTL gene	Locus	TSS snp	R^2 with eQTL SNP
11	rs10896050	SNX32, CTSW, MRPL11	11q13.1	rs633800	0.137
				rs10896064	0.2
11	rs11227306	CTSW	11q13.1	rs633800	0.467
				rs10896064	0.317
11	rs11227332	CTSW	11q13.1	rs633800	0.241
				rs10896064	0.256
11	rs13817	CTSW	11q13.1	rs633800	0.458
				rs10896064	0.478
11	rs622614	CTSW	11q13.1	rs633800	0.219
				rs10896064	0.228
11	rs630303	CTSW	11q13.1	rs633800	0.458
				rs10896064	0.478
11	rs656040	CTSW	11q13.1	rs633800	0.422
				rs10896064	0.516
11	rs665306	CTSW	11q13.1	rs633800	0.458
				rs10896064	0.478

Table S13. Linkage disequilibrium (R^2 >0.1) between eQTL SNPs in TN risk loci and candidate functional SNPs in enhancers identified by Rhie, et al.

Chr.	eQTL SNP	eQTL gene	Locus	Enhancer SNP	R^2 with eQTL SNP
11	rs10896050	SNX32, CTSW, MRPL11	11q13.1	rs10160792	0.102
				rs1058068	0.124
				rs11227309	0.133
				rs11227311	0.133
				rs526631	0.105
				rs637571	0.174
				rs677029	0.124
				rs689274	0.112
11	rs630303	CTSW	11q13.1	rs10160792	0.198
				rs1058068	0.278
				rs11227309	0.443
				rs11227311	0.443
				rs1151523	0.218
				rs526631	0.244
				rs634534	0.218
				rs637571	0.188
				rs677029	0.235
				rs689274	0.248
11	rs656040	CTSW	11q13.1	rs10160792	0.175
				rs1058068	0.254
				rs11227309	0.427
				rs11227311	0.427
				rs1151523	0.198
				rs526631	0.222
				rs634534	0.198
				rs637571	0.211
				rs677029	0.212
				rs689274	0.227
11	rs11227332	CTSW	11q13.1	rs10160792	0.218
				rs1058068	0.236
				rs11227309	0.233
				rs11227311	0.233
				rs1151523	0.197
				rs526631	0.215
				rs634534	0.197
				rs637571	0.303
				rs677029	0.239
				rs689274	0.219
11	rs665306	CTSW	11q13.1	rs10160792	0.198
				rs1058068	0.278
				rs11227309	0.443
				rs11227311	0.443
				rs1151523	0.218
				rs526631	0.244
				rs634534	0.218

				rs637571	0.188
				rs677029	0.235
				rs689274	0.248
11	rs11227306	CTSW	11q13.1	rs10160792	0.275
				rs1058068	0.299
				rs11227309	0.45
				rs11227311	0.45
				rs1151523	0.268
				rs526631	0.305
				rs634534	0.268
				rs637571	0.272
				rs677029	0.35
				rs689274	0.309
11	rs622614	CTSW	11q13.1	rs10160792	0.107
				rs1058068	0.122
				rs11227309	0.212
				rs11227311	0.212
				rs526631	0.105
				rs677029	0.122
				rs689274	0.111
11	rs13817	CTSW	11q13.1	rs10160792	0.198
				rs1058068	0.278
				rs11227309	0.443
				rs11227311	0.443
				rs1151523	0.218
				rs526631	0.244
				rs634534	0.218
				rs637571	0.188
				rs677029	0.235
				rs689274	0.248

Table S14. Comparison of ORs for a subset of TNBCC subjects with expression data, stratified by DASL-defined ER status

SNP	G/I	Chr.	Position	Locus	Allele	Overall TN 3,677 cases 4,708 controls		TN with DASL 578 cases 4,638 controls			TN excluding ER+ 516 cases 4,638 controls			DASL-defined ER+ 62 cases 4,638 controls		
						OR	P-value	OR	95% CI	P-value	OR	95% CI	P-value	OR	95% CI	P-value
rs616488	G	1	10488802	*PEX14*	G	0.91	9.7×10^{-3}	0.99	(0.86-1.13)	0.85	0.99	(0.85-1.14)	0.84	1.00	(0.67-1.49)	0.99
rs6678914	G	1	200453799	*LGR6*	A	0.90	3.3×10^{-3}	0.97	(0.85-1.12)	0.7	0.96	(0.84-1.11)	0.62	1.07	(0.73-1.57)	0.74
rs4245739	I	1	202785465	*MDM4*	C	1.19	4.0×10^{-6}	1.19	(1.03-1.38)	0.017	1.16	(0.99-1.35)	0.061	1.56	(1.05-2.33)	0.029
rs12710696	I	2	19184284	2p24.1	A	1.11	3.5×10^{-3}	1.07	(0.93-1.23)	0.34	1.08	(0.94-1.25)	0.27	0.92	(0.62-1.36)	0.68
rs4849887	G	2	120961592	2q14.2	A	0.89	0.041	0.93	(0.75-1.15)	0.5	0.97	(0.78-1.22)	0.77	0.72	(0.42-1.27)	0.26
rs2016394	G	2	172681217	2q31.1	A	1.10	6.9×10^{-3}	1.13	(0.99-1.29)	0.074	1.12	(0.97-1.29)	0.11	1.21	(0.83-1.74)	0.32
rs3731711	I	2	201921306	*CASP8*	G	0.84	$1.4 \times 10-4$	0.94	(0.79-1.12)	0.51	0.92	(0.76-1.11)	0.38	1.02	(0.62-1.69)	0.93
rs13387042	G	2	217614077	2q35	G	0.93	0.049	0.92	(0.80-1.04)	0.19	0.93	(0.81-1.07)	0.29	0.85	(0.59-1.22)	0.38
rs6828523	I	4	176083001	*ADAM29*	A	0.84	1.3×10^{-3}	0.88	(0.71-1.08)	0.22	0.94	(0.75-1.17)	0.56	0.45	(0.20-1.00)	0.049
rs10069690	I	5	1332790	*TERT*	A	1.24	1.4×10^{-7}	1.27	(1.08-1.48)	3.1×10^{-3}	1.32	(1.12-1.56)	8.4×10^{-4}	0.91	(0.57-1.45)	0.69
rs2735845	I	5	1353584	*TERT*	G	0.80	2.5×10^{-7}	0.93	(0.80-1.09)	0.39	0.95	(0.81-1.12)	0.54	0.71	(0.44-1.15)	0.16
rs1432679	G	5	158176661	*EBF1*	G	1.10	8.6×10^{-3}	1.06	(0.93-1.22)	0.36	1.05	(0.91-1.21)	0.51	1.30	(0.89-1.90)	0.17
rs3757318	G	6	151955806	*ESR1*	A	1.33	9.2×10^{-6}	1.57	(1.25-1.98)	1.2×10^{-4}	1.58	(1.25-2.01)	1.5×10^{-4}	1.48	(0.75-2.92)	0.26
rs2046210	I	6	151990059	*ESR1*	A	1.16	5.3×10^{-5}	1.25	(1.09-1.43)	1.5×10^{-3}	1.22	(1.06-1.41)	6.8×10^{-3}	1.54	(1.04-2.27)	0.031
rs12525163	I	6	152081984	*ESR1*	C	1.15	4.9×10^{-4}	1.08	(0.93-1.25)	0.31	1.1	(0.94-1.28)	0.24	0.94	(0.61-1.46)	0.78
rs7904519	G	10	114763917	*TCF7L2*	G	1.12	9.9×10^{-4}	1.10	(0.97-1.26)	0.15	1.09	(0.95-1.26)	0.2	1.17	(0.80-1.71)	0.43
rs3903072	I	11	65339642	11q13.1	A	0.92	0.024	0.95	(0.83-1.08)	0.42	0.95	(0.82-1.09)	0.43	0.97	(0.66-1.43)	0.88
rs11820646	I	11	128966381	11q24.3	A	0.92	0.016	0.91	(0.79-1.04)	0.17	0.88	(0.77-1.02)	0.084	1.17	(0.80-1.72)	0.42
rs12422552	I	12	14305198	12p13.1	C	1.13	2.7×10^{-3}	1.15	(0.99-1.34)	0.06	1.16	(0.99-1.36)	0.059	1.07	(0.70-1.64)	0.74
rs10771399	I	12	28046347	*PTHLH*	G	0.72	1.5×10^{-8}	0.77	(0.61-0.96)	0.022	0.74	(0.58-0.94)	0.015	1.01	(0.57-1.80)	0.97
rs17356907	G	12	94551890	*NTN4*	G	0.90	7.5×10^{-3}	1.15	(0.93-1.22)	0.061	1.16	(0.99-1.35)	0.066	1.14	(0.74-1.75)	0.56
rs1292011	G	12	114320905	12q24	G	1.08	0.035	1.06	(0.93-1.22)	0.4	1.03	(0.90-1.19)	0.64	1.25	(0.84-1.89)	0.27
rs11571833	I	13	31870626	*BRCA2*	T	1.44	0.023	1.62	(0.92-2.86)	0.094	1.70	(0.96-3.03)	0.07	1.01	(0.15-6.72)	0.99
rs2588809	I	14	67730181	*RAD51L1*	A	0.91	0.041	0.87	(0.72-1.05)	0.14	0.85	(0.70-1.04)	0.11	1.00	(0.61-1.65)	1
rs3803662	G	16	51143842	*TOX3*	A	1.09	0.022	1.06	(0.91-1.22)	0.46	1.07	(0.92-1.25)	0.38	0.9	(0.59-1.37)	0.62
rs8170	G	19	17250704	19p13.1	A	1.26	1.3×10^{-7}	1.22	(1.04-1.44)	0.017	1.26	(1.06-1.49)	7.3×10^{-3}	1.03	(0.62-1.71)	0.9
rs2363956	G	19	17255124	19p13.1	C	0.82	2.3×10^{-8}	0.83	(0.72-0.94)	4.8×10^{-3}	0.82	(0.71-0.94)	5.4×10^{-3}	0.81	(0.56-1.17)	0.26
rs1864112	I	19	17309960	19p13.1	A	0.84	5.5×10^{-6}	0.81	(0.70-0.94)	7.1×10^{-3}	0.79	(0.67-0.93)	4.7×10^{-3}	0.9	(0.59-1.36)	0.61
rs6142050	G	20	31990789	*RALY*	G	1.11	3.8×10^{-3}	1.11	(0.97-1.27)	0.14	1.11	(0.96-1.28)	0.14	1.07	(0.72-1.59)	0.73
rs6001913	G	22	39166699	*MKL1*	A	1.20	1.8×10^{-3}	1.46	(1.17-1.82)	6.6×10^{-4}	1.45	(1.16-1.82)	1.3×10^{-3}	1.5	(0.82-2.77)	0.19

Table S15. Polygenic risk score and TNBC risk using the first quintile as the reference

PRS Quintile	74 SNPs				27 SNPs			
	Quintile definitions	OR	95% CI	p-value	Quintile definitions	OR	95% CI	p-value
1	PRS≤0.24	1.00	--	--	PRS≤-0.57	1.00	--	--
2	0.24<PRS≤0.58	1.53	1.29-1.81	1.1×10^{-6}	-0.57<PRS≤-0.26	1.43	1.21-1.69	2.8×10^{-5}
3	0.58<PRS≤0.86	1.97	1.68-2.32	9.9×10^{-16}	-0.26<PRS≤0.039	1.91	1.63-2.25	3.9×10^{-15}
4	0.86<PRS≤1.24	2.54	2.17-2.97	1.3×10^{-29}	0.039<PRS≤0.40	2.62	2.24-3.06	1.4×10^{-33}
5	1.24<PRS	4.03	3.46-4.70	4.8×10^{-69}	0.40<PRS	4.08	3.50-4.75	2.5×10^{-74}

Figure S1. Association between 19p13.1 variants (n=170) and TN breast cancer risk
a) TNBC associations in a 250kb region

b) Adjusted for rs8100241

Figure S2. Association between *ESR1* variants (n=448) and TN breast cancer risk
a) TNBC associations in a 250kb region
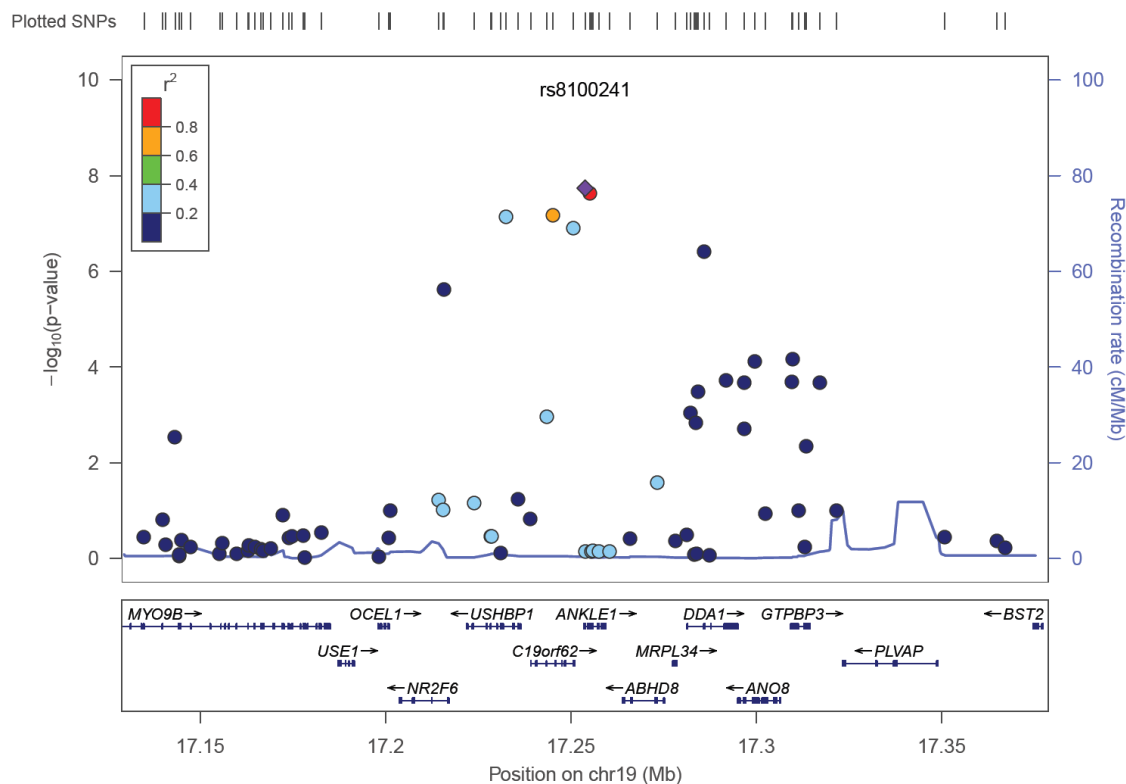
b) Adjusted for rs9397437
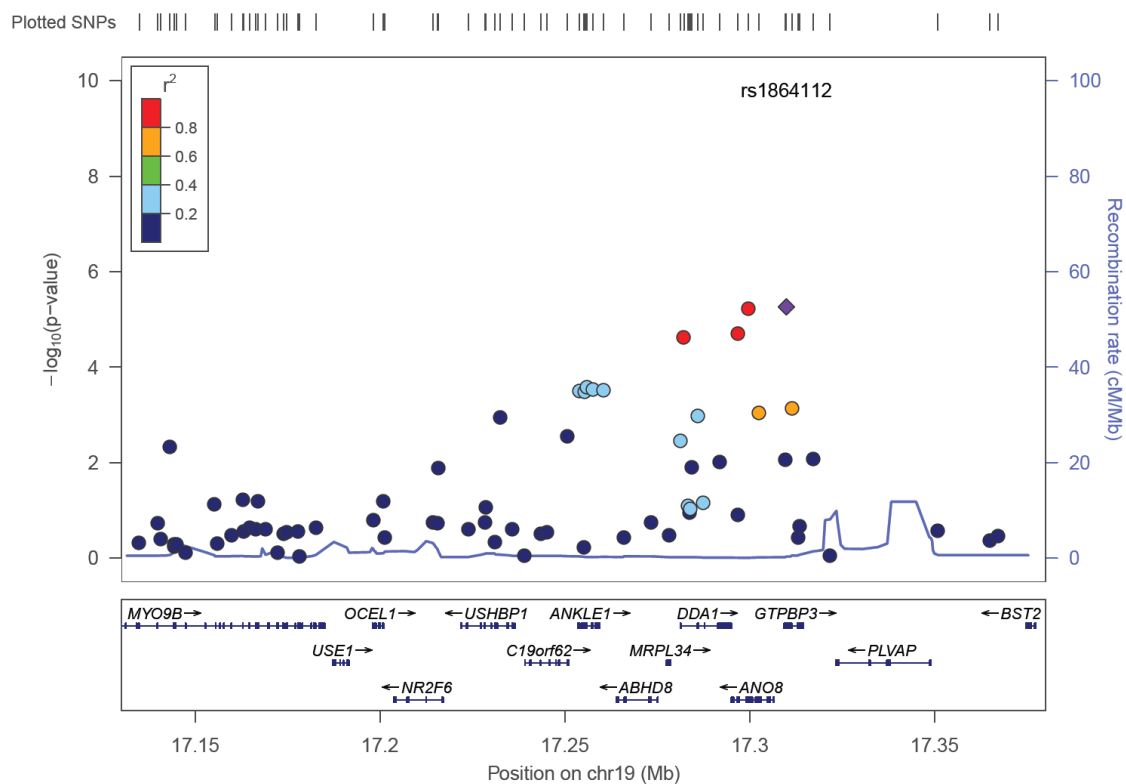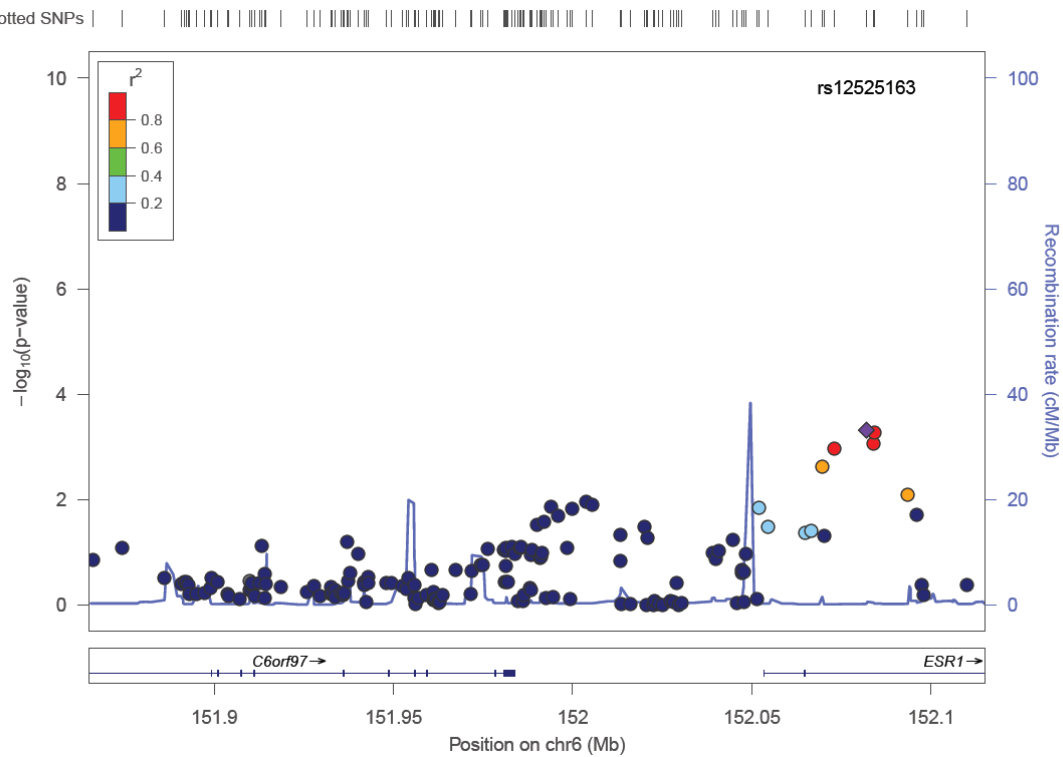
Figure S3. ROC curves for TN breast cancer risk by 74-SNP and 30-SNP PRS

Figure S4. Cumulative risk of TNBC stratified by a 30-SNP polygenic risk score

Cumulative risk of TNBC by age
Polygenic risk score with 30 SNPs

* 5th quintile (PRS>0.40)
○ 4th quintile (0.039<PRS<=0.40)
● 3rd quintile (−0.26<PRS<=0.039)
+ 2nd quintile (−0.57<PRS<=−0.26)
▽ 1st quintile (PRS<=−0.57)
— Population−based risk

Cumulative risk

Age (years)
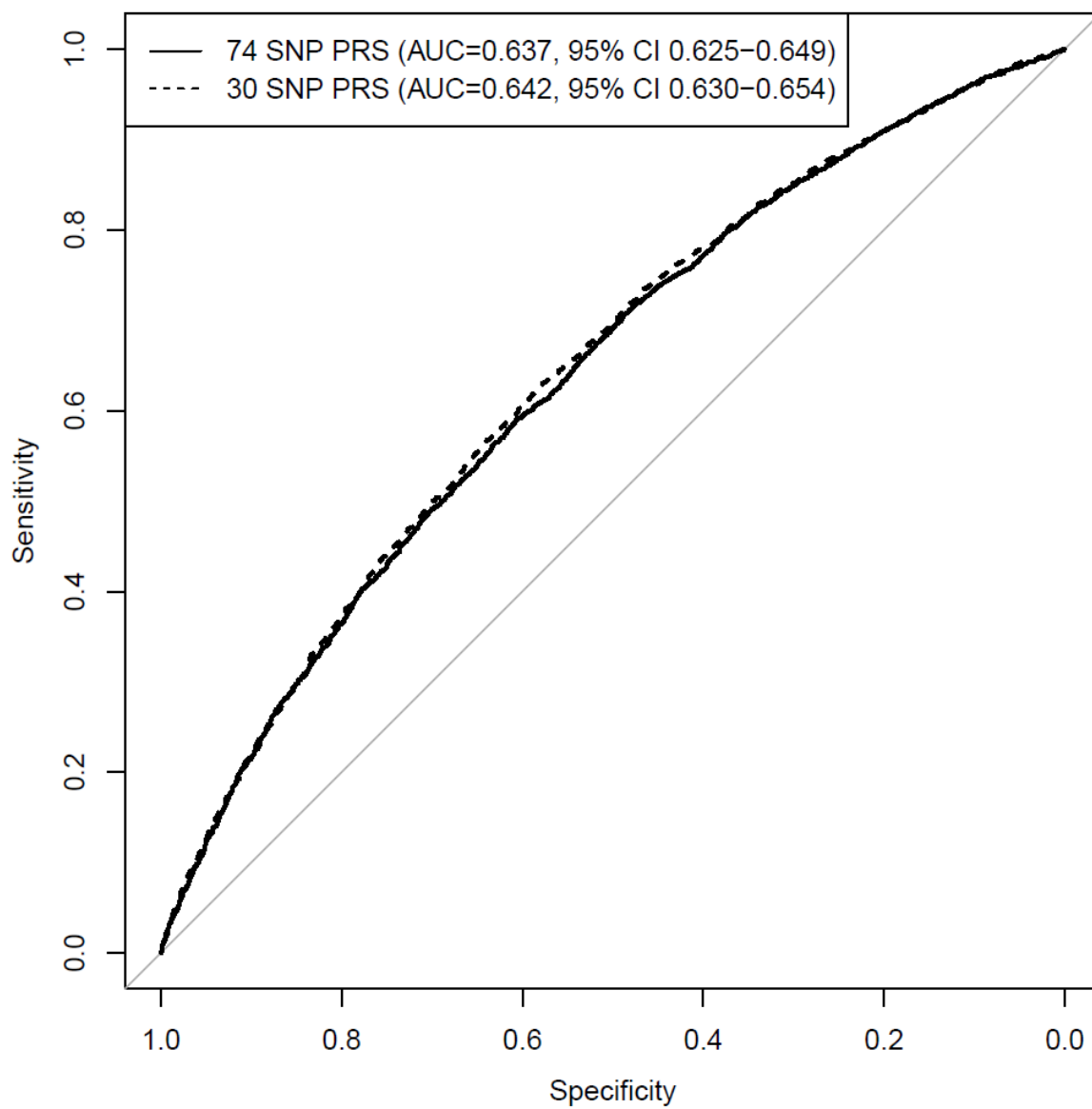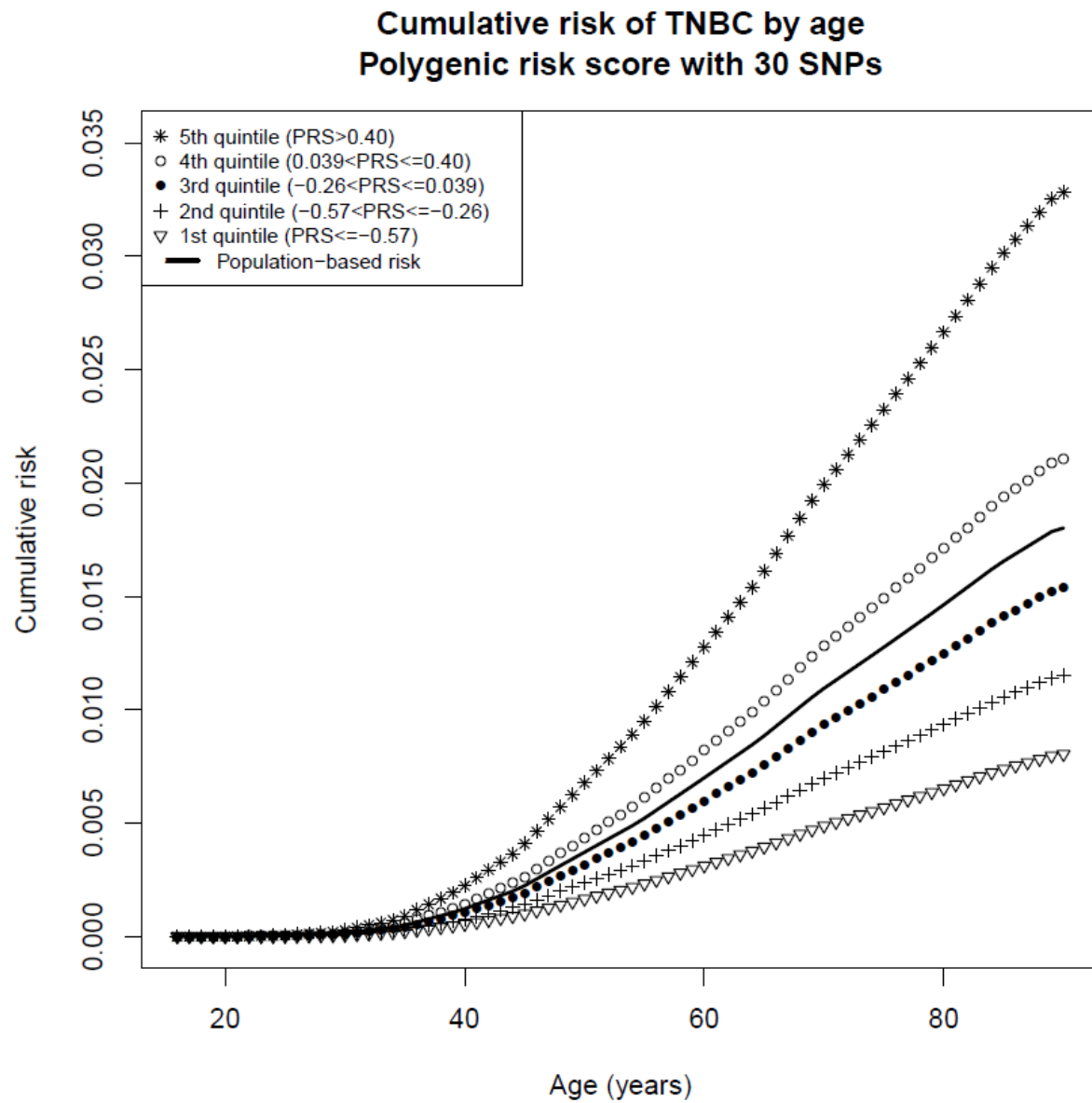
Figure legends

Figure S1. Association between 19p13.1 variants (n=170) and TN breast cancer risk

a) The association between 170 variants from the combined 19p13.1 analyses in stages 1 and 2 is

shown. The most significant SNP (rs8100241) is shown as the purple diamond (p=1.8 x 10^{-8}).

The remaining variants are shown as circles, colored by the degree of linkage disequilibrium (R^2)

between each SNP and rs8100241. The continuous blue line represents the recombination rate

(cM/Mb). b) The association between 19p13.1 variants adjusted for rs8100241 is shown. The

most significant SNP after adjustment for rs8100241 (rs1864112) is shown as the purple

diamond (p=5.5 x 10^{-6}).

Figure S2. Association between *ESR1* variants (n=448) and TN breast cancer risk

a) The association between 448 variants from the combined *ESR1* analyses in stages 1 and 2 is

shown. The most significant SNP (rs9397437) is shown as the purple diamond (p=8.9 x 10^{-8}).

The remaining variants are shown as circles, colored by the degree of linkage disequilibrium (R^2)

between each SNP and rs9397437. The continuous blue line represents the recombination rate

(cM/Mb). b) The association between *ESR1* variants adjusted for rs9397437is shown. The most

significant SNP after adjustment for rs9397437 (rs12525163) is shown as the purple diamond

(p=4.9 x 10^{-4}).

Figure S3. ROC curves for TN breast cancer risk by 74-SNP and 30-SNP PRS

Receiver operating characteristic (ROC) curves are shown for the 74-SNP PRS (solid black line)

and the 30-SNP PRS (dashed black line). The area under the curve (AUC) for the 74-SNP PRS

was 0.637 (95% CI 0.625-0.649) while the AUC for the 30-SNP PRS was 0.642 (95% CI 0.630-0.654).

Figure S4. Cumulative risk of TNBC stratified by a 30-SNP polygenic risk score

The effect of the 30-SNP polygenic risk score (PRS) on cumulative risk of triple negative (TN) breast cancer among Caucasian women, stratified by PRS quintile, is shown. The population-based cumulative risk curve is shown as a solid black line, and the first through fifth quintile-specific cumulative risk estimates are presented according to labels.

The Triple-Negative Breast Cancer Consortium (TNBCC)

Australia Breast Cancer Tissue Bank (ABCTB): 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): This is a 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.

California Teachers Study (CTS): Breast cancer cases from the CTS cohort, composed of women who were active or retired California teachers or administrators at the time the cohort was established in 1995. Cancer outcomes were identified through annual linkage with the California Cancer Registry (CCR). Unaffected individuals from the CTS cohort were sampled for controls.

Cancer Genetic Markers of Susceptibility (CGEMS): 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).

Dana Farber Cancer Institute (DFCI): Cases were obtained from an unselected series of breast tumors patients from the Dana Farber Cancer Institute. DNA samples from residual bloods from triple negative breast cancer patients were genotyped.

DEMOKRITOS: 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.

Fox Chase Cancer Center (FCCC): Cases were seen at FCCC and 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. DNA was obtained from peripheral blood samples.

Gene Environment Interaction and Breast Cancer in Germany (GENICA): This is a population-based case-control study of breast cancer in the Greater Bonn area of Germany. Cases were incident breast cancer cases enrolled between 2000 and 2004 (reported from 14 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.

University of Kansas Medical Center (KUMC): Cases were obtained from an unselected series of breast tumors patients from the University of Kansas Medical Center. DNA samples from residual bloods from triple negative breast cancer patients were genotyped.

Helsinki Breast Cancer Study (HEBCS): Cases from this hospital-based case-control study in Southern Finland 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 (19).

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 between 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.

Mammary Carcinoma Risk Factor Investigation (MARIE): This is a population-based case-control study of breast cancer in Northern and Southern Germany. 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): This is a clinic-based breast cancer case-control study at the Mayo Clinic. 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): 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 24,469 participating women, and controls were randomly sampled from the initial cohort among members not diagnosed with breast cancer at the end of follow-up.

Norwegian Breast Cancer Study (NBCS): Cases were comprised of 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 University Hospital. Control subjects were healthy women, age 55-71, residing in Tromsø (440), and Bergen (109) attending the Norwegian Breast Cancer Screening Program.

The Nashville Breast Health Study (NBHS): The NBHS is a population-based case-control study of breast cancer conducted in Tennessee. The study was initiated in 2001 to recruit patients with invasive breast cancer or ductal carcinoma in situ between the ages of 25 and 75 years. Cases were identified from participating hospitals in and around the Nashville Metropolitan area as well as from the Tennessee Cancer Registry (TCR). Diagnosis and tumor pathology were confirmed via medical record abstraction and ascertainment from the TCR. Controls were recruited through random digit dialing.

Ohio State University (OSU): Cases were obtained from an unselected series of breast tumors patients from the Ohio State University Stefanie Spielman Breast Bank. DNA samples isolated

from blood of triple negative breast cancer patients were genotyped. Controls were selected from the Columbus Area Control Sample Bank and were frequency matched for age and ethnicity to the cases.

Prospective Study of Outcomes in Sporadic Versus Hereditary Breast Cancer (POSH): Cases from this prospective cohort study in the United Kingdom 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): Two cohorts of Australian twins and their families (parents, children, spouses and siblings), were recruited to a Health and Lifestyle study in 1988 and 1990. The total number of participants was over 27,000, with an age range of 17 to 96 ($M = 39.7$, SD = 15.3). Phenotypic data were available for 20,464 individuals, of which 5117 (1727 males and 3390 females) from 2567 independent families were genotyped. 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.

Sheffield Breast Cancer Study (SBCS): This is a hospital-based case-control study of breast cancer. 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.

Städtisches Klinikum Karlsruhe and Deutsches Krebsforschungszentrum Breast Cancer Study (SKKDKFZS): This breast cancer case cohort 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 for the subgroup of TN breast cancer cases were from an unselected series of unaffected women from the same geographical area.

Simultaneous Study of Docetaxel Based Anthracycline Free Adjuvant treatment Evaluation, as well as Life Style Intervention Strategies (SUCCESS C): is a prospectively randomized trial for high risk breast cancer patients without metastases. All patients had to be at least 18 years of age, HER2 negative with an otherwise high risk of recurrence. A total of 3642 patients were recuited from March 2009 to August 2011. Of 3256 patients whole blood samples could be collected, of which 742 were from patients with triple negative tumors.

Washington University Young Women's Breast Cancer Study (WASHU): This breast cancer case cohort study consists of women with pathologically confirmed breast cancer identified through the Young Women's Breast Cancer Program at Washington University Siteman Cancer Center.

Wellcome Trust Case Control Consortium (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.

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