

rs2735383, located at a microRNA binding site in the 3'UTR of *NBS1*, is not associated with breast cancer risk

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Supplementary Data

Supplementary Table S1. Subgroup analysis of *NBS1* rs2735383 under a dominant genetic model and breast cancer risk in the European BCAC studies.

Subgroup	N Controls	N Cases	MAF Controls	MAF Cases	OR (95% CI)*	P-value*
Age						
≤ 50 years	13,055	13,362	33.76%	33.41%	0.995 (0.944-1.050)	0.995
> 50 years	26,987	28,553	33.28%	33.38%	1.006 (0.971-1.042)	0.742
Age at menarche						
≤ 13 years	14,312	13,843	33.72%	33.13%	0.961 (0.916-1.008)	0.106
> 13 years	8,964	8,095	32.65%	33.63%	1.065 (1.002-1.132)	0.044
Age at menopause						
≤ 50 years	5,571	7,288	32.79%	33.43%	1.036 (0.962-1.115)	0.349
> 50 years	3,366	4,262	33.50%	33.24%	0.977 (0.888-1.074)	0.628
Menopausal status						
Premenopausal	8,974	7,412	33.66%	33.07%	0.969 (0.908-1.033)	0.332
Postmenopausal	19,648	17,353	33.39%	33.56%	1.011 (0.970-1.054)	0.608
Number of full-term pregnancies						
≤ 2	21,008	19,722	33.53%	33.20%	0.999 (0.959-1.041)	0.974
> 2	8,258	7,327	33.26%	33.44%	1.017 (0.952-1.086)	0.624
Breast feeding						
No	6,849	6,805	33.36%	33.25%	0.991 (0.923-1.064)	0.810
Yes	11,947	12,709	33.68%	33.28%	0.988 (0.939-1.041)	0.657
Family history						
1 st degree relative with BC	23,648	4,119	33.21%	32.68%	0.999 (0.931-1.073)	0.985
Receptor status						
ER-positive	39,699	25,959	33.47%	33.40%	1.006 (0.973-1.039)	0.740
ER-negative	39,618	6,774	33.42%	32.90%	0.997 (0.943-1.053)	0.904
Triple negative	30,696	2,712	33.10%	32.04%	0.987 (0.901-1.080)	0.770

N, number; MAF, minor allele frequency; OR, odds ratio; CI, confidence interval; ER, estrogen receptor; BC, breast cancer.

*Adjusted for age, study and nine principal components.

Supplementary Table S2. Subgroup analysis of *NBS1* rs2735383 under an additive genetic model and breast cancer risk in the European BCAC studies.

Subgroup	N Controls	N Cases	MAF Controls	MAF Cases	OR (95% CI)*	P-value*
Age						
≤ 50 years	13,055	13,362	33.76%	33.41%	0.992 (0.954-1.032)	0.699
> 50 years	26,987	28,553	33.28%	33.38%	1.009 (0.983-1.036)	0.495
Age at menarche						
≤ 13 years	14,312	13,843	33.72%	33.13%	0.975 (0.941-1.010)	0.163
> 13 years	8,964	8,095	32.65%	33.63%	1.053 (1.006-1.102)	0.027
Age at menopause						
≤ 50 years	5,571	7,288	32.79%	33.43%	1.024 (0.969-1.082)	0.398
> 50 years	3,366	4,262	33.50%	33.24%	0.986 (0.918-1.058)	0.686
Menopausal status						
Premenopausal	8,974	7,412	33.66%	33.07%	0.979 (0.933-1.026)	0.374
Postmenopausal	19,648	17,353	33.39%	33.56%	1.008 (0.977-1.039)	0.635
Number of full-term pregnancies						
≤ 2	21,008	19,722	33.53%	33.20%	1.001 (0.971-1.031)	0.969
> 2	8,258	7,327	33.26%	33.44%	1.016 (0.968-1.067)	0.518
Breast feeding						
No	6,849	6,805	33.36%	33.25%	0.992 (0.942-1.046)	0.778
Yes	11,947	12,709	33.68%	33.28%	0.989 (0.952-1.027)	0.560
Family history						
1 st degree relative with BC	23,648	4,119	33.21%	32.68%	0.997 (0.946-1.052)	0.923
Receptor status						
ER-positive	39,699	25,959	33.47%	33.40%	1.008 (0.983-1.033)	0.533
ER-negative	39,618	6,774	33.42%	32.90%	0.996 (0.956-1.038)	0.856
Triple negative	30,696	2,712	33.10%	32.04%	0.988 (0.923-1.057)	0.731

N, number; MAF, minor allele frequency; OR, odds ratio; CI, confidence interval; ER, estrogen receptor; BC, breast cancer.

*Adjusted for age, study and nine principal components.

Supplementary Table S4. PCR primers for amplification of the 3'UTR of *NBS1* and the *LRR4* gene.

Gene	Forward primer	Reverse primer
<i>NBS1</i>	5'-TTGCCTAGCTACTTGGTTC-3'	5'-ATGGTCTTCAGTTTCTAGTAC-3'
<i>LRR4</i>	5'-TGGGAGAGTCGTAGCTTTC-3'	5'-CGATGGGGTTGTTGCCAAG-3'

Supplementary Table S5. Clinical and demographic characteristics of breast cancer cases in the European BCAC studies.

Subgroup	N cases	rs2735383 GG+GC		rs2735383 CC		P-value
Age						0.64
≤ 50 years	13,362	11,866	(88.80%)	1,496	(11.20%)	
> 50 years	28,553	25,312	(88.65%)	3,241	(11.35%)	
Age at menarche						1
≤ 13 years	13,843	12,267	(88.62%)	1,576	(11.38%)	
> 13 years	8,095	7,173	(88.61%)	922	(11.39%)	
Age at menopause						0.70
≤ 50 years	7,288	6,464	(88.69%)	824	(11.31%)	
> 50 years	4,262	3,790	(88.93%)	472	(11.07%)	
Menopausal status						1
Premenopausal	7,412	6,570	(88.64%)	842	(11.36%)	
Postmenopausal	17,353	15,392	(88.70%)	1,961	(11.30%)	
Number of full-term pregnancies						0.59
≤ 2	19,722	17,518	(88.82%)	2,204	(11.18%)	
> 2	7,327	6,491	(88.59%)	836	(11.41%)	
Breast feeding						1
No	6,805	6,041	(88.77%)	764	(11.23%)	
Yes	12,709	11,278	(88.74%)	1,431	(11.26%)	
Family history						0.42
No	19,405	17,200	(88.64%)	2,205	(11.36%)	
1 st degree relative with BC	4,119	3,669	(89.08%)	450	(10.92%)	
ER status						0.32
ER positive	25,959	23,003	(88.61%)	2,956	(11.39%)	
ER negative	6,774	6,032	(89.05%)	742	(10.95%)	
TN status						0.14
Non-TN	15,192	13,478	(88.72%)	1,714	(11.28%)	
TN	2,712	2,432	(89.68%)	280	(10.32%)	

N, number; BC, breast cancer; ER, estrogen receptor; TN, triple negative.