Supplementary Table 4. Prevalence and Odds Ratios for breast cancer risk of LoF variants in all studied genes between selected Greek breast cancer patients and Reference Controls from ExAC and FLOSSIES.

	Demokritos Cases					Controls (ExAC/FLOSSIES)					Cancer Risk (ExAC/FLOSSIES)		
Gene	**Truncating**	**Missense**	**Total**	**Cases, No**	**Freq (%)**	**Truncating**	**Missense**	**Total**	**Controls, No**	**Freq (%)**	**Odds Ratio**	**95% CI**	***p*-value**
ATM	13	3	16	1355	1.18	78/14	15/7	93/21	26862/7325	0.35/0.29	3.41/4.12	1.87-5.86/2-8.3	**$6.12x10^{-5}$/$2.45x10^{-5}$**
BLM	1	0	1	1355	0.07	50/NA	0/NA	50/NA	24941/NA	0.20/NA	0.37/NA	0.009-2.15/NA	0.616/NA
BRIP1	1	0	1	1355	0.07	52/11	0/0	52/11	26396/7325	0.20/0.15	0.37/0.49	0.009-2.19/0.01-3.39	0.616/0.789
CDKN2A	0	1	1	1355	0.07	1/NA	4/NA	5/NA	23419/NA	0.02/NA	3.46/NA	0.07-30.9/NA	0.466/NA
CHEK2	7	12	19	1355	1.40	155/28	63/11	218/39	25819/7325	0.84/0.53	1.66/2.63	0.98-2.67/1.44-4.68	0.11/**0.003**
CHEK2 miss	0	12	12	1355	0.89	0/0	63/11	63/11	26974/7325	0.23/0.15	3.79/5.9	1.86-7.12/2.38-14.78	**$1.2x10^{-3}$/$2.45x10^{-4}$**
CHEK2^	NA	12	12	1355	0.89	NA	233/46	233/46	26974/7325	0.85/0.63	1.02/1.40	0.52-1.83/0.67-2.69	0.88/0.28
DICER1	1	0	1	1355	0.07	3/NA	0/NA	3/NA	27171/NA	0.01/NA	6.68/NA	0.13-83.28/NA	0.307/NA
ERCC3	1	0	1	1355	0.07	63/NA	0/NA	63/NA	27164/NA	0.23/NA	0.32/NA	0.008-1.84/NA	0.539/NA
FANCC	1	0	1	1355	0.07	16/NA	24/NA	40/NA	26912/NA	0.15/NA	0.5/NA	0.012-2.93/NA	0.751/NA
FANCL	1	0	1	1355	0.07	16/NA	0/NA	16/NA	26795/NA	0.06/NA	1.24/NA	0.029-7.97/NA	0.642/NA
FANCM	2	0	2	1355	0.15	167/40	0/0	167/40	26461/7325	0.63/0.55	0.23/0.27	0.03-0.86/0.03-1.04	**0.044**/0.072
MLH1	1	0	1	1355	0.07	5/NA	2/NA	7/NA	27157/NA	0.03/NA	2.86/NA	0.06-22.31/NA	0.494/NA
MSH6	3	0	3	1355	0.22	15/NA	4/NA	19/NA	26990/NA	0.07/NA	3.14/NA	0.59-10.70/NA	0.159/NA
NBN	3	0	3	1355	0.22	17/14	0/0	17/14	25479/7325	0.07/0.19	3.32/1.16	0.62-11.49/0.21-4.16	0.155/0.789
NF1	1	0	1	1355	0.07	9/NA	2/NA	11/NA	25350/NA	0.04/NA	1.7/NA	0.04-11.72/NA	0.616/NA
PALB2	7	0	7	1355	0.52	17/6	0/0	17/6	26435/7325	0.06/0.08	8.03/6.3	2.81-20.42/1.81-22.75	**$7.16x10^{-3}$/0.003**
PMS2	2	0	2	1355	0.15	25/NA	10/NA	35/NA	25884/NA	0.14/NA	1.09/NA	0.13-4.26/NA	0.751/NA
PTEN	2	0	2	1355	0.15	1/0	0/0	1/0	27173/7325	0.00/0.00	40.09/Inf	2.08-2323.35/-	**0.017/0.036**
RAD51C	5	2	7	1355	0.52	19/2	3/1	22/3	26361/7325	0.08/0.04	6.19/12.6	2.23-15.03/2.87-75.64	**$0.002/4.73x10^{-4}$**
RAD51D	4	0	4	1355	0.30	5/2	0/1	5/3	26787/7325	0.02/0.04	15.81/7.2	3.13-73.54/1.22-49.3	**0.002/0.022**
SLX4	1	0	1	1355	0.07	36/14	0/0	36/14	26862/7325	0.13/0.19	0.55/0.39	0.01-3.28/0.01-2.54	1/0.616
TP53	2	6	7	1355	0.52	1/0	28/3	29/3	26889/7325	0.11/0.04	4.79/12.61	1.77-11.2/2.87-75.64	**$0.001/4.73x10^{-4}$**

Notes: CNV carriers, double LoF variant carriers and the low risk *CHEK2* variants: p.(Ile157Thr) & p.(Ser428Phe) were excluded from the analysis. *ExAC data extracted were non-TCGA/non-Finnish. FLOSSIES data extracted were on European-Americans.*

Abbreviations used: ExAC, Exome Aggregation Consortium; Freq, Variant Observed Frequency; miss, missense; No, number; NA, Not Applicable. ^*CHEK2* p.I157T only.