

BRCA1

Chr:Pos	Identifier	HGVS c	HGVS p	Gene	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
17:41246481	rs1799950	NM_007294.3:c.1067A>G	NP_009225.1:p.Gln356Arg	BRCA1	4	0	0.1	4998	176	0.0440706	Yes	Benign
17:41245471	rs4986850	NM_007294.3:c.2077G>A	NP_009225.1:p.Asp693Asn	BRCA1	5	0	0.125	6379	257	0.0568148	Yes	Benign
17:41244435	rs16941	NM_007294.3:c.3113A>G	NP_009225.1:p.Glu1038Gly	BRCA1	10	2	0.35	26322	7641	0.342866	Yes	Benign
17:41244000	rs16942	NM_007294.3:c.3548A>G	NP_009225.1:p.Lys1183Arg	BRCA1	10	2	0.35	26614	7768	0.349005	Yes	Benign
17:41223094	rs1799966	NM_007294.3:c.4837A>G	NP_009225.1:p.Ser1613Gly	BRCA1	10	2	0.35	26717	7852	0.349572	Yes	Benign
17:41244936	rs799917	NM_007294.3:c.2612C>T	NP_009225.1:p.Pro871Leu	BRCA1	11	2	0.375	26590	11586	0.41005	Yes	Benign
17:41246664	.	NM_007294.3:c.884A>G	NP_009225.1:p.Asp295Gly	BRCA1	1	0	0.025	8	0	6.59207E-05	Yes	Uncertain Significance
17:41244429	rs4986852	NM_007294.3:c.3119G>A	NP_009225.1:p.Ser1040Asn	BRCA1	1	0	0.025	1566	16	0.0131668	Yes	Benign
17:41226387	rs28897691	NM_007294.3:c.4636G>A	NP_009225.1:p.Asp1546Asn	BRCA1	1	0	0.025	16	0	0.00013182	Yes	Benign

BRCA2

Chr:Pos	Identifier	HGVS c	HGVS p	Gene Names	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
13:32929387	rs169547	NM_000059.3:c.7397T>C	NP_000050.2:p.Val2466Ala	BRCA2	0	20	1	718	59912	0.993718	Yes	Benign
13:32906729	rs144848	NM_000059.3:c.1114A>C	NP_000050.2:p.Asn372His	BRCA2	6	1	0.2	23687	4939	0.27793	Yes	Benign
13:32911463	rs1799944	NM_000059.3:c.2971A>G	NP_000050.2:p.Asn991Asp	BRCA2	4	0	0.1	5880	274	0.0534091	Yes	Benign
13:32906480	rs766173	NM_000059.3:c.865A>C	NP_000050.2:p.Asn289His	BRCA2	4	0	0.1	5691	271	0.0517837	Yes	Benign
13:32931967	.	NM_000059.3:c.7706G>A	NP_000050.2:p.Gly2569Asp	BRCA2	1	0	0.025	.	.	.	Yes	Uncertain Significance
13:32913804	rs80358755	NM_000059.3:c.5312G>A	NP_000050.2:p.Gly1771Asp	BRCA2	1	0	0.025	35	1	0.000306988	Yes	Benign
13:32911547	rs55638633	NM_000059.3:c.3055C>G	NP_000050.2:p.Leu1019Val	BRCA2	1	0	0.025	20	0	0.000166323	Yes	Benign

MLH1

Chr:Pos	Identifier	HGVS c	HGVS p	Gene	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
3:37053568	rs1799977	NM_000249.3:c.655A>G	NP_000240.1:p.Ile219Val	MLH1	7	0	0.175	20186	4003	0.232538	Yes	Benign
3:37083806	.	NM_000249.3:c.1715G>T	NP_000240.1:p.Gly572Val	MLH1	1	0	0.025	.	.	.	NM_000249.3(MLH1):c.1714G>A (p.Gly572Ser) same amino acid different nucleotide	Uncertain Significance
3:37061893	rs63751049	NM_000249.3:c.977T>C	NP_000240.1:p.Val326Ala	MLH1	1	0	0.025	64	0	0.000527313	Yes	Uncertain Significance
3:37038152	.	NM_000249.3:c.159G>T	NP_000240.1:p.Glu53Asp	MLH1	2	0	0.05	.	.	.	NM_000249.3(MLH1):c.157G>A (p.Glu53Lys) same amino acid different nucleotide	Uncertain Significance

MSH2

Chr:Pos	Identifier	HGVS c	HGVS p	Gene	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
2:47641560	rs11309117	NM_00251.2:c.942+3_942+6del	.	MSH2	18	1	0.5	107	1	0.00750172	NM_000251.2(MSH2):c.942+3_942+7delAAAAA (nucleotide A homopolymer)	Benign
2:47698167	.	NM_000251.2:c.1725T>A	NP_000242.1:p.Asp575Glu	MSH2	1	0	0.025	.	.	.	No	Uncertain Significance
2:47643457	rs4987188	NM_000251.2:c.965G>A	NP_000242.1:p.Gly322Asp	MSH2	1	0	0.025	1671	19	0.0141058	Yes	Benign
2:47630353	rs17217716	NM_000251.2:c.23C>T	NP_000242.1:p.Thr8Met	MSH2	1	0	0.025	164	0	0.00359996	Yes	Benign

MSH6

Chr:Pos	Identifier	HGVS c	HGVS p	Gene	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
2:48010488	rs1042821	NM_000179.2:c.116G>A	NP_000170.1:p.Gly39Glu	MSH6	9	1	0.275	15722	1894	0.21409	Yes	Benign
2:48025764	rs1800937	NM_000179.2:c.642C>T	NP_000170.1:p.=	MSH6	1	0	0.025	7770	481	0.0734955	Yes	Benign

PMS2 - EPCAM

Chr:Pos	Identifier	HGVS c	HGVS p	Gene	Armenian Cohort			exAC			Clinvar Entry	Clinical Significance
					# Hets	# of Homz	alt Allele Freq	# Hets	# Homz	alt Allele Freq		
7:6026942	rs1805323	NM_000535.5:c.1454C>A	NP_000526.1:p.Thr485Lys	PMS2	7	0	0.175	7919	901	0.0800886	Yes	Benign
7:6026865	rs2228007	NM_000535.5:c.1531A>G	NP_000526.1:p.Thr511Ala	PMS2	1	0	0.025	2915	62	0.0251377	Yes	Benign
7:6026775	rs2228006	NM_000535.5:c.1621A>G	NP_000526.1:p.Lys541Glu	PMS2	8	12	0.8	14556	44055	0.851392	Yes	Benign
7:6026988	rs1805321	NM_000535.5:c.1408C>T	NP_000526.1:p.Pro470Ser	PMS2	15	0	0.375	28407	9186	0.385393	Yes	Benign
7:6045627	rs10254120	NM_000535.5:c.59G>A	NP_000526.1:p.Arg20Gln	PMS2	2	0	0.05	8183	410	0.0781768	Yes	Benign
7:6013049	.	NM_000535.5:c.2570G>C	NP_000526.1:p.Gly857Ala	PMS2	9	0	0.225	20102	2453	0.370208	Yes	Benign
7:6026708	rs63750668	NM_000535.5:c.1688G>T	NP_000526.1:p.Arg563Leu	PMS2	1	0	0.025	699	2	0.00581338	Yes	Benign
7:6026607	rs1805318	NM_000535.5:c.1789A>T	NP_000526.1:p.Thr597Ser	PMS2	1	0	0.025	990	3	0.00820915	Yes	Benign
2:47601106	rs1126497	NM_002354.2:c.344T>C	NP_002345.2:p.Met115Thr	EPCAM	14	2	0.45	27668	17704	0.519786	Yes	Benign