

Supplementary Table 1: KASP primers used to type mutation haplotype in *BRCA1* c.3331_3334delCAAG mutation carriers from Portugal and Brazil.

SNP	A1	A2	Cohort MAF	3450del4 (-) MAF	3450del (+) MAF	Difference in MAF	Chr17 coordinate	Distance from Mutation (Mb)	
rs936656	A	G	0.3399	0.3373	0.4535	0.1162	33433819	-7.810144	
			Primer_A1	GAAGGTGACCAAGTTCATGCTGGGAGGGTTTCCTGGAAGAATCA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGGAGGGTTTCCTGGAAGAATCG					
			Primer_C1	CCCTGTTGATGCCATCATCCCATT					
rs7211602	A	C	0.2764	0.2728	0.4318	0.159	36026400	-5.217563	
			Primer_A1	GAAGGTGACCAAGTTCATGCTGGCAACATTTTCAAGAGGGACATT G					
			Primer_A2	GAAGGTCGGAGTCAACGGATTAGGCAACATTTTCAAGAGGGACAT TT					
			Primer_C1	CCAGACTTCTCACTATCCTGGTCAT					
rs16966483	C	T	0.06497	0.06179	0.2045	0.14271	36740607	-4.503356	
			Primer_A1	GAAGGTGACCAAGTTCATGCTCCCTGTGGCTTGAATCTCGGA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTCCCTGTGGCTTGAATCTCGGG					
			Primer_C1	TTCTCCAAGGAACCTCAGTGTGCAA					
rs7217037	T	C	0.3657	0.3665	0.3295	-0.037	38223190	-3.020773	
			Primer_A1	GAAGGTGACCAAGTTCATGCTCACATATACAGTTTGTATGAACTA G					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGCTCACATATACAGTTTGTATGA ACTAA					
			Primer_C1	CCTCATCTGTAAATTGGGGATGATCATAT					
rs17558560	T	C	0.3096	0.3037	0.4318	0.1281	38936658	-2.307305	
			Primer_A1	GAAGGTGACCAAGTTCATGCTCATCCAGGACTCTTCGCAAACC					
			Primer_A2	GAAGGTCGGAGTCAACGGATTTCATCCAGGACTCTTCGCAAAC					
			Primer_C1	CTAAGGTTTGAACACGAGCTAGCGCTT					
rs11651246	G	T	0.1851	0.1802	0.3977	0.2175	40759936	-0.484027	
			Primer_A1	GAAGGTGACCAAGTTCATGCTAAGAGACCCAGTTAAAGGTGTCAC A					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGAGACCCAGTTAAAGGTGTCACC					
			Primer_C1	GCTAAAGGCTCTGGAAGCCACAAAT					
rs2229611	T	C	0.4103	0.415	0.2045	-0.2105	41063465	-0.180498	
			Primer_A1	GAAGGTGACCAAGTTCATGCTAAGAGATGTGGAGTCTTCGGTGT T					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGAGATGTGGAGTCTTCGGTGTTC					
			Primer_C1	GTAGTCCTCCTCAATCCCTGGCAT					
rs80357903	CAAG	-					41244214	0	
rs17599948	G	A	0.2452	0.2484	0.1023	-0.1461	41353409	0.109446	

			Primer_A1	GAAGGTGACCAAGTTCATGCTGGAAAATGCTGGAGCCTTGACTA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGGAAAATGCTGGAGCCTTGACTG					
			Primer_C1	CGGAAATGTCTAGCTTTTGGGGAGAA					
rs1107748	C	T	0.4158	0.4199	0.2386	-0.1813	41773813	0.52985	
			Primer_A1	GAAGGTGACCAAGTTCATGCTAAATGACAATAGTTCTTTCTAGAC TCAT					
			Primer_A2	GAAGGTCGGAGTCAACGGATTATGACAATAGTTCTTTCTAGACT CAC					
			Primer_C1	TCTTTACAAGGGCACTGCAGGCAAA					
rs8077889	C	A	0.1526	0.1456	0.4545	0.3089	41878165	0.634202	
			Primer_A1	GAAGGTGACCAAGTTCATGCTAATCATTTTTGCTCAGAGACAACC CA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTATCATTTTTGCTCAGAGACAACCC C					
			Primer_C1	CGTCACTGTAAACCATTCTGTCAAGAAT					
rs3826425	G	A	0.2363	0.2387	0.1279	-0.1108	42981653	1.73769	
			Primer_A1	GAAGGTGACCAAGTTCATGCTATGGTGGCCACTTTCCAGGACA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGGTGGCCACTTTCCAGGACG					
			Primer_C1	GAAGACATGGAGGTGCCCATAGTAT					
rs7214920	A	G	0.1215	0.118	0.2791	0.1611	45051128	3.807165	
			Primer_A1	GAAGGTGACCAAGTTCATGCTTGACAAGAATCTGCACTCAAGAGA TA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTGACAAGAATCTGCACTCAAGAGAT G					
			Primer_C1	GTCGCACGCATGCACACACATTTAA					
rs16957364	C	T	0.05983	0.05651	0.2045	0.14799	46555744	5.311781	
			Primer_A1	GAAGGTGACCAAGTTCATGCTATTTGCACACGGTGGCCACAA					
			Primer_A2	GAAGGTCGGAGTCAACGGATTATTTGCACACGGTGGCCACAG					
			Primer_C1	TGCCCAGCTGCACTGGTTTCTTAA					
rs16948048	G	A	0.2686	0.2641	0.4659	0.2018	47440465	6.196502	
			Primer_A1	GAAGGTGACCAAGTTCATGCTCCCTGTACCCTAAACACTTTCAAC					
			Primer_A2	GAAGGTCGGAGTCAACGGATTCTCCCTGTACCCTAAACACTTTCA AT					
			Primer_C1	GTAGCATTTATCCGAGTTCAGAGACAA					
rs9303562	C	T	0.3278	0.3273	0.3523	0.025	48857042	7.613079	
			Primer_A1	GAAGGTGACCAAGTTCATGCTAAATAGAAACACCTCTCAGCCCCG					
			Primer_A2	GAAGGTCGGAGTCAACGGATTACAAATAGAAACACCTCTCAGCCC A					
			Primer_C1	CCATTGATTATGACATGAACTTCTGAGGAA					

Supplementary Table 2: Summary of mutation carriers and genotype experiments

Country	Cases with mutation (<i>n</i>)	Cases with Affymetrix Array Data (<i>n</i>)	Cases with KASP Genotype Data (<i>n</i>)
Angola	1	1	0
Brazil	18	2	18
Colombia	32	32	3
Chile	10	10	0
Portugal	15	2	13
Spain	13	13	0