

S2 Table. Primers applied for the tests

Name	Sequence	Variants queried	Purpose
B1_C_e-13_F	CCTTCTAACAGCTACCCTTCCA	BRCA1 c.4484+4dupA	cDNA primers for aberrant splicing detection
B1_C_e16_R	AGAAGGATCAGATTCAGGGTCA	BRCA1 c.4484+4dupA	cDNA primers for aberrant splicing detection qPCR
Rd_B1_ex14_F	GGT GCT AGC ATC TGT CTG TTG	BRCA1 c.4484+4dupA BRCA1 c.4358-31A>C	Tumor gDNA sequencing for LOH detection
Rd_B1_ex14_R	CCG CAC ATT TCT CAT GTT GTA	BRCA1 c.4484+4dupA BRCA1 c.4358-31A>C	Tumor gDNA sequencing for LOH detection
B1_C_e-16_REV_FAM	5' FAM _AGAAGGATCAGATTCAGGGTCA	BRCA1 c.4484+4dupA BRCA1 c.4358-31A>C	QMPSF
B1_exon_13-15_F	AAGCACATCAGAAAAAGGTC	BRCA1 c.4484+4dupA BRCA1 c.4358-31A>C	qPCR and QMPSF
B1_exon_14-15_F	ATAAAGAACCAGGAGTGGAAAGGTC	BRCA1 c.4484+4dupA BRCA1 c.4358-31A>C	qPCR and QMPSF
BRCA1_C_e-16_Rev2	TTTGTTGACCCTTTCTGTTGAAG	BRCA1 c.4484+4dupA	Measure of incomplete aberrant splicing allelic imbalance
B1_C-e-14_For	GTGTCTGCAGATAGTTCTACCAGTAA	BRCA1 c.4484+4dupA	Measure of incomplete aberrant splicing
B1_C_e-15_F	GTGGTACATGCACAGTTGCTCT	BRCA1 c.4484+4dupA	Allelic imbalance
B1_C_e-13_F	CCTTCTAACAGCTACCCTTCCA	BRCA1 c.4358-31A>C	cDNA primers for aberrant splicing detection
B1_C_e16_R	AGAAGGATCAGATTCAGGGTCAT	BRCA1 c.4358-31A>C	cDNA primers for aberrant splicing detection
B1_C_e-18_F	GTGTGTGAACGGACACTGAAAT	BRCA1 c.5407-10G>A	cDNA primers for aberrant splicing detection
Rd_B1_ex24_R	CCA GGA CAG TAG AAG GAC TGA AGA	BRCA1 c.5407-10G>A	cDNA primers for aberrant splicing detection
Rd_B1_ex23_F	CCA GTA GTC CTA CTT TGA CAC TTT GA	BRCA1 c.5407-10G>A	Tumor gDNA sequencing for LOH detection
Rd_B1_ex23_R	CCC ATG GAA ACA GTT CAT GTA TT	BRCA1 c.5407-10G>A	Tumor gDNA sequencing for LOH detection
B1_C_e-15_F (ex16-ra)	GTGGTACATGCACAGTTGCTCT	BRCA1 c.5407-10G>A	Allelic imbalance
B1_C_e-20_R (ex16-ra)	GACCTTGGTGGTTTCTTCCAT	BRCA1 c.5407-10G>A	Allelic imbalance

Rd_B1_ex11.11_F (ex13-ra)	CCA CCT CCA AGG TGT ATG AAG TAT	BRCA1 c.5407-10G>A	Allelic imbalance
B1_C_e-14_R (ex13-ra)	GGCTTATAGGGTATTCACTACTTTTCTG	BRCA1 c.5407-10G>A	Allelic imbalance
B1_exon20_F2_FAM:	5' FAM _AAACCACCAAGGTCCAAAGC	BRCA1 c.5407-10G>A	QMPSF
B1_exon_23_norm_R:	ACAATTGGGTGGACACCTGT	BRCA1 c.5407-10G>A	QMPSF
B1_exon_23_mut_R:	TGGACACCCTGGATCCTGT	BRCA1 c.5407-10G>A	QMPSF
B2_C_e-16_F	AGGAATACAGTTGGCTGATGGT	BRCA2 c.8487G>T BRCA2 c.8427C>T	cDNA primers for aberrant splicing detection
B2_C_e-22_R	CATTCAACATTTGCCTGTGATT	BRCA2 c.8487G>T BRCA2 c.8427C>T	cDNA primers for aberrant splicing detection
B2-C-e19_Rev	CCATCACTGAAAAGCGATGATA	BRCA2 c.8487G>T	Measure aberrant splicing completeness
B2-C-e13_For	TTCTTTAGAGCCGATTACCTGTG	BRCA2 c.8487G>T	Measure aberrant splicing completeness
B2_C_e17_For	GCTCTGTGTGACACTCCAGGT	BRCA2 c.8427C>T	Nested sequencing for aberrant splicing detection
B2_C_e18_For	ATGGAAAGGGATGACACAGC	BRCA2 c.8427C>T	Nested sequencing for aberrant splicing detection
Rd_B2_ex19_F	GGC AGT TCT AGA AGA ATG AAA ACT CT	BRCA2 c.8427C>T	Alleleic imbalance
Rd_B2_ex19_R	TGG GCA AGA GAC CGA AAC T	BRCA2 c.8427C>T	Alleleic imbalance
B2_C_e7_For	CCACCCACCCTTAGTTCTACTG	BRCA2 c.793G>A	cDNA primers for aberrant splicing detection
Rd_B2_ex10.01_R	GGT TCC ACT TCA GAT ACA AAT GAG TA	BRCA2 c.793G>A	cDNA primers for aberrant splicing detection

LOH, loss of heterozygosity; QMPSF, quantitative multiplex PCR of short fluorescent fragments; qPCR, quantitative real-time polymerase chain reaction.