

**A**

Wild-type BRCA1

Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ACTTAAAGAA TTTGTCAA~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

701 KLTNAPGSFTKCSNTSELKEFVNPSLPREEKEEKLETVKVSNNAEDPKDL 750

SUM149PT cells sg\_GFP

2288delT: fs&gt;735X

Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ACTTAAAGAA TTTGTCAA~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

701 KLTNAPGSFTKCSNTSELKEFVN~~LA~~PQEKKKKRN STOP**Clone 3**

SUM149PT cells sg\_exon11, (IN-FRAME): deletion and reversion to long-form

2247del42: T710\_N723del14

Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ~~ACTTAAAGAA TTTGTCAA~~~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

701 KLTNAPGSFTKCSNTSELKEFVNPSLPREEKEEKLETVKVSNNAEDPKDL IN-FRAME

**Clone 4**

SUM149PT cells sg\_exon11, (IN-FRAME): deletion and reversion to long-form

2269del8; 2288delT: E717\_K719del3; EFVN720\_723GICQ

Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ~~ACTTAAAGAA TTTGTCAA~~~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

KLTNAPGSFTKCSNTSELK~~EL~~QPSLPREEKEEKLETVKVSNNAEDPKDL IN-FRAME**B****Clone 1**

SUM149PT cells sg\_exon11, (OUT-FRAME): out of frame deletion mutation

2271delC; 2288delT; fs&gt;724X

Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ~~ACTTAAAGAA TTTGTCAA~~~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

KLTNAPGSFTKCSNTSELK~~EL~~ST STOP**Clone 2**

SUM149PT cells sg\_exon11, (OUT-FRAME): out of frame insertion mutation

2270insA; 2288delT: fs&gt;719X

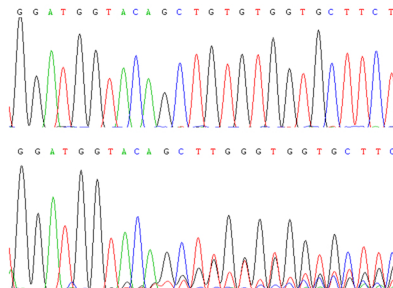
Genomic DNA:

2101 AAGTTAACAA ATGCACCTGG TTCTTTTACT AAGTGTCAA ATACCAGTGA  
 2151 ACTTAAAGAA TTTGTCAA~~TC~~ CTAGCCTTCC AAGAGAAGAA AAAGAAGAGA  
 2201 AACTAGAAAC AGTTAAAGTG TCTAATAATG CTGAAGACCC CAAAGATCTC

Peptide:

KLTNAPGSFTKCSNTSE~~EL~~ STOP

T-2288delT  
 Deletion  
 Insertion  
 Missense

**C****D**

SG\_GFP

SG\_EXON22