

# Supplementary Materials: A Novel Pathogenic *BRCA1* Splicing Variant Produces Partial Intron Retention in the Mature Messenger RNA

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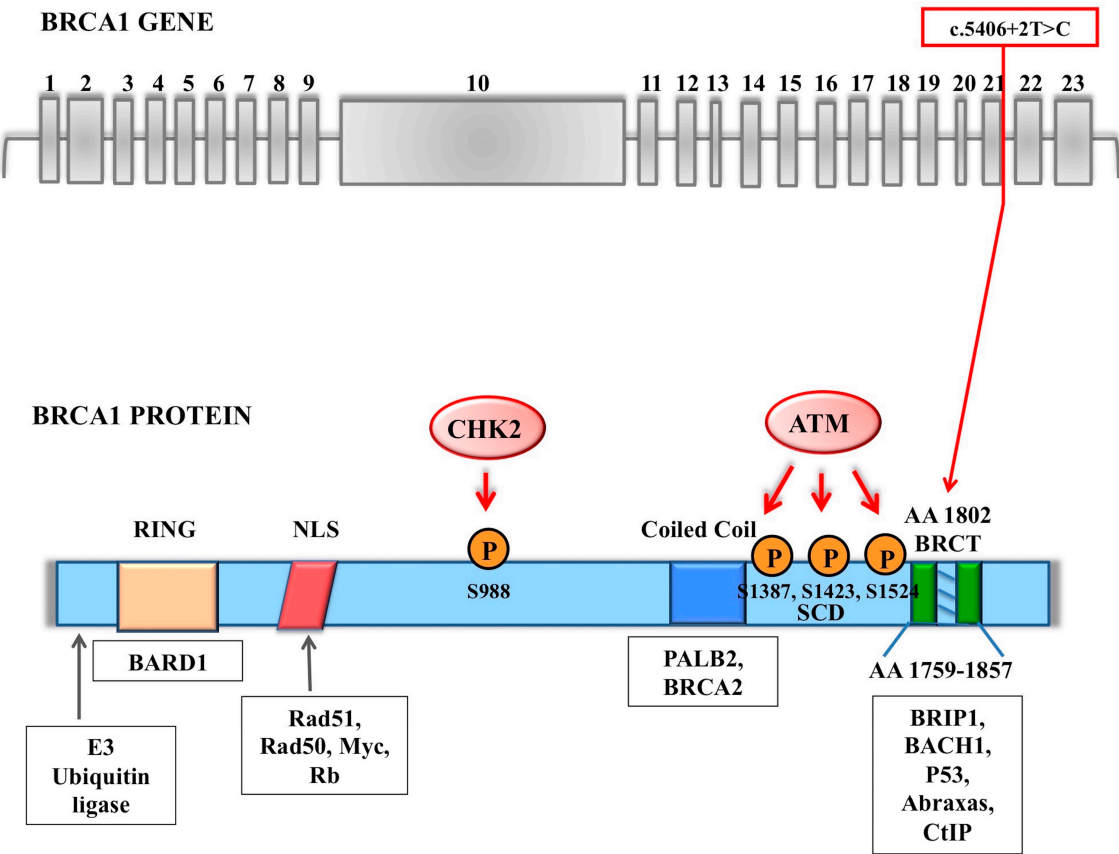
a

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b

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GTGTGAGGCACCTGTGGTGACCCGAGAGTGGGTGTTGGACAGTGTAGCACTCTACCAGTG
CCAGGAGCTGGACACCTACCTGATACCCCAGATCCCCCACAGCCACT
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**Figure S1.** (a) FASTA sequence of a *wt* cDNA spanning from exon 20 to 23: exon 20 in yellow, 21 in green, 22 in violet and 23 in blue; (b) FASTA sequence of the cDNA of the patient bearing the c.5406+2T>C splice variant (in lower case and in red) showing the 156 retained bases of intron 21: exon 20 in yellow, 21 in green, 22 in violet and 23 in blue; the retained intron sequence is reported in lower case and white.



**Figure S2.** Schematic view of the BRCA1 protein and its interacting proteins. The function of the 3'-end proximity BRCT domain could be impaired by the c.5406+2T>C splice variant consequent to the altered repair of the double-strand break. The cross-talk with other proteins that interfere with the function of BRCA1 are indicated in boxes and the activating kinases are indicated by pink ovals and arrows.

**Table S1.** Primers used to assess the sequence regions of the novel *BRCA1* splicing variant (red and blue arrows correspond to those in Figure 1c).

Primer Sequences on BRCA1 cDNA		Product Length (bp)	
		Wild Type	5406+2T>C
Blue arrows →	(a) BRCA1_cDNA_Fw: CAACATGCCCACAGATCAAC	825	981
	(b) BRCA1_cDNA_Rw: AATTTCCTCCCCAATGTTCC		
Red arrows →	(c) cDNA_ex20/21_Fw: CAACATGCCCACAGATCAACTGGAATGG	165	321
	(d) cDNA_ex22/23_Rw: CATCTGCCCAATTGCATGGAAGCCATTG		