

Title	Functional classification of DNA variants by hybrid minigenes: identification of 30 spliceogenic variants of BRCA2 exons 17 and 18
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S3 Table. Relative quantification of transcripts induced by DNA variants and microdeletions of *BRCA2* exons 17 and 18 in MCF-7 cells.

DNA variants	Canonical Transcript ¹	Exon 17 skipping	Ex17-del20	Ex17-del69	Exon17-insAG	lvs16-ins8	Ex17-del1	Other aberrant transcripts ³
EXON 17								
c.7806-9T>G		41.5% ±0.3		22.2% ±0.2		36.3% ±0.5		
c.7806-2A>G		20.1% ±0.3	51.8% ±0.3	28.1% ±0.1				
c.7806-1G>A							100%	
c.7806-1G>T			100%					
c.7806insAG		5.1% ±0.5		2.3% ±0.1	92.6% ±0.6			
c.7875A>G	95.3% ±1.1			4.7% ±1.1				
c.7975A>G	73.8% ±0.1	26.2% ±0.1						
c.7976G>C		100%)						
c.7976G>A		100%						
c.7976+1G>A		100%						
Microdeletions								
c.7944_7973del	61.6% ±0.2	38.4 ±0.2						
c.7944_7953del	100%							
c.7954_7963del	97.8%±0.5	1.0±0.1						1.2%±0.5
c.7964_7973del	95.4%±0.2	4.6%±0.2						
EXON 18								
DNA variants	Canonical Transcript	Exon 18 skipping	Ex18-ins6	Ex18-del309	Ex18-del298	Ex18-del164	Ex18-del191	Other aberrant transcripts ³
Wild type Minigene ²	99.3 ±0.8	0.7% ±0.8						
c.7977-7C>G		21.6% ±0.5	78.4% ±0.5					
c.7977-6T>G	66.7% ±0.8	31% ±0.8					2.3%	
c.7977-3_7978del		90% ±1.1					10% ±1.1	
c.7977-2A>T		93.3% ±0.1					6.7% ±0.1	
c.7977-1G>C		89.8% ±1.8					10.2% ±1.8	-
c.7977-1G>T		91.5% ±0.1					7%±0.1	ex18-del236=1.5%
c.7985C>G		90.2% ±4.4					5.0% ±0.1	4.7% ±4.5
c.7985C>T	96.7 ±1.3	3.3% ±1.3						
c.7988A>T	84.2% ±1.5	8.6% ±1.7						7.2% ± 0.4
c.7992T>A	68.6% ±1.0	31.4% ±1.0						
c.8007A>G	84.8% ±0.6	15.2%±0.6						
c.8009C>A	4.0 ±3.4	91.2% ±4.4					4.8% ±3.9	-
c.8009C>T	76.6% ±6.1	23.4% ±6.1						-

c.8009C>G	79.9 ±1.2	20.1% ±1.2						
c.8023A>G		0.7% ±0.1		93% ±0.3				1.0% ±0.1/ 4.2±0.2
c.8035G>T	2.4% ± 0.5				93.6% ± 0.4			4.0±0.2
c.8042C>G	97.7% ±0.1	2.3% ±0.1						
c.8072C>T	94.9% ±1.2	5.1% ±1.2						
c.8168A>G	69.6% ±0.9	4.5% ±0.4				25.9% ±0.5		
c.8249_50del	93.0 ±1.2	7.0 ±1.2						
c.8331G>A	40.7% ±0.6	52.0% ±0.4						2.1% ±0.1 / 1.7 ±0.1 / 3.5%±0.2
c.8331+1G>T		81.0 ±0.3						6.1%±0.2% / 3.7% ±0.3 / 6.4% ±0.4
c.8331+2T>C		87.1%±0.3						[ex17-del151 +ex18 skipping] (12.9%)
Microdeletions								
c.7979_8008del	47.2% ±0.4	49.6% ±0.4					3.2% ±0.1	
<i>c.7979_7988del</i>	79.8% ±0.7	20.2%±0.7						
<i>c.7989_7998del</i>	100%							
<i>c.7999_8008del</i>	48.3% ±0.4	51.7% ±0.4						
c.8004_8033del	30.6% ±0.2	64.7% ±0.0					4.7% ±0.1	
<i>c.8004_8013del</i>	16.3% ±0.1	78.1% ±0.1					5.6%	
<i>c.8014_8023del</i>	100%							
<i>c.8024_8033del</i>	100%							

Relative proportions (%) of each transcript with their standard deviations (±%) are indicated. Principal transcripts are shown in bold type.

¹ Size of the canonical transcript 1012 nt after cDNA amplification with primers RTBR2_ex16-FW and RTpSAD-RV.

² The wild type minigenes showed a minor transcript with exon 18 skipping ranging from 0% to 2.6%.

³ Other aberrant transcripts: [Ex17del151+Ex18 skipping], [Ex18 sk + ivs17ins58], [Ex18del156], [~710 nt],[775 nt], [822 nt], [867 nt], [878 nt], [1021 nt]