Table S1a

Experiments performed by:		INT, Milano	INT, Milano	HVH, Barcelone	HVH, Barcelone	HVH, Barcelone	HCSC, Madrid
Samples provided by:		INT, Milano	INT, Milano and QIMR, Brisbane	HVH, Barcelone	QIMR, Brisbane	HVH, Barcelone	HCSC, Madrid
Samples (detection method):		**PBLs (capillary EP)**	**LCLs (capillary EP)**	**LEUs (agarose)**	**LCLs (agarose)**	**PBMCs (agarose)**	**LEUs (capillary EP)**
Splicing event	**mRNA position**						
Δ2	r.38_67del106	0/1	0/6	not tested	not tested	not tested	0/13
Δ3	r.68_316del249	1/1	6/6	**18/18**	**10/10**	**8/8**	13/13
Δ3,4	r.68_c.425del359	0/1	5/6	**0/18**	**0/10**	**0/8**	13/13
Δ3–7	r.68_631del564	0/1	5/6	not tested	not tested	not tested	not tested
Δ4	r.317_425del109	0/1	0/6	**0/18**	**0/10**	**0/8**	0/13
Δ4–7	r.317_631del315	0/1	6/6	not tested	not tested	not tested	5/20
Δ5	r.426_475del50	0/1	0/6	**0/19**	**0/10**	**0/8**	2/20
Δ5,6	r.426_516del91	0/1	1/6	**0/19**	**0/10**	**0/8**	2/13
Δ5–7	r.426_631del206	0/1	5/6	**0/19**	**0/10**	**0/8**	3/20
Δ6	r.476_516del41	0/1	0/6	**0/19**	**0/10**	**0/8**	17/20
Δ6q,7	r.478-631del154	1/1	6/6	**19/19**	**10/10**	**8/8**	20/20
Δ12	r.6842_6937del96	1/1	4/4	not tested	not tested	not tested	5/8
Δ12,13	r.6842_7007del166	0/1	0/4	not tested	not tested	not tested	1/13
Δ17	r.7806_7976del171	not tested	not tested	**0/21**	**0/10**	**0/8**	0/13
Δ17,18	r.7806_8331del526	2/2	3/3	**21/21**	**10/10**	**8/8**	23/23
Δ18	r.7977_8331del355	2/2	3/3	**21/21**	**10/10**	**8/8**	19/28

Δ19	r.8332_8487del156	not tested	9/9	**0/21**	**0/10**	**0/8**	1/13
Δ20	r.8488_8632del145	1/2	1/3	**0/16**	**0/10**	**0/8**	2/3
▼20A	c.8633-1327_c.8633-1264ins64	2/2	3/3	**0/16**	**10/10**	**0/8**	4/5
Δ22	c.8755_8953del199	0/2	0/3	**23/23**	**0/10**	**8/8**	5/10
Δ22,23p	c.8755_9004del250	not tested	not tested	**23/23**	**0/10**	**8/8**	4/10
Δ23p	c.8954_9004del51	0/2	0/3	**0/23**	**0/10**	**0/8**	0/13

Table S1b

HCSC, Madrid	HCSC, Madrid	HCSC, Madrid	HCSC, Madrid	HCSC, Madrid	MU, Maastrich	QIMR, Brisbane	Cologne University	Southampton General Hospital
HVH, Barcelone	USC, Santiago de Compostela	MU, Maastrich	QIMR, Brisbane	HCSC, Madrid	MU, Maastrich	QIMR, Brisbane	Cologne University	Southampton General Hospital
PBMCs (capillary EP)	**PBLs (capillary EP)**	**PBLs (capillary EP)**	**LCLs (capillary EP)**	**One healthy breast tissue (capillary EP)**	**PBLs (agarose)**	**LCLs (agarose)**	**LCLs (agarose)**	**LEU detected/tested (agarose)**

0/8	9/16	not tested	6/10	0/1	2/2	0/4	not tested	not tested
8/8	14/16	not tested	10/10	1/1	2/2	3/4	2/2	3/3
8/8	7/16	not tested	**10/10**	0/1	not tested	0/4	not tested	not tested
5/8	2/6	not tested	7/9	not tested	not tested	not tested	not tested	not tested
3/8	2/16	not tested	10/10	0/1	not tested	0/4	not tested	not tested
7/8	6/6	not tested	9/10	0/1	not tested	not tested	not tested	not tested
4/8	4/6	not tested	10/10	1/1	not tested	not tested	not tested	not tested
0/8	0/6	not tested	0/10	0/1	not tested	0/15	not tested	not tested
8/8	5/6	not tested	10/10	1/1	not tested	0/15	not tested	not tested
0/8	0/6	not tested	0/10	1/1	not tested	0/15	not tested	not tested
8/8	6/6	not tested	**10/10**	1/1	4/4	15/15	not tested	not tested
5/8	6/6	not tested	not tested	1/1	3/3	not tested	2/2	3/3
0/8	0/6	not tested	not tested	0/1	not tested	not tested	not tested	not tested
0/8	0/12	0/12	7/10	1/1	not tested	not tested	not tested	not tested
8/8	**12/12**	**12/12**	**10/10**	1/1	not tested	15/15	not tested	not tested
8/8	12/12	**11/12**	**10/10**	1/1	not tested	15/15	not tested	3/3
5/8	3/10	6/12	9/10	not tested	not tested	not tested	not tested	not tested
2/8	2/6	0/12	7/10	1/1	not tested	not tested	not tested	not tested
7/8	5/6	3/12	10/10	0/1	1/1	not tested	2/2	not tested
8/8	3/6	8/12	**9/9**	0/1	not tested	not tested	not tested	not tested
7/8	1/6	2/12	9/9	not tested	not tested	not tested	not tested	not tested
5/8	5/6	0/6	7/8	not tested	not tested	not tested	not tested	not tested

Table S2

Alternate splicing event to be detected	Forward or reverse	Primer name and sequence
Δ2	F (full length)	BR2ex1-2F: AGAACTGCACCTCTGGAGCGGACTTATTT
	F (alternate)	BR2ex1-3F: AGAACTGCACCTCTGGAGCGGATTTAGGA
	R	BR2CA2 Ex 7R: CTAAAGAACTTGACCAAGAC
Δ3	F (full length)	BRCA2 EX2-3F(a): ACACGCTGCAACAAAGCAGATT
	F (alternate)	BRCA2 EX2-4F: ACACGCTGCAACAAAGCAGGAA
	R	BRCA2 Ex 7R: CTAAAGAACTTGACCAAGAC
Δ3,4	F (full length)	BRCA2ex2-3F(b): TTTTAAGACACGCTGCAACAAAGCAGATTTA
	F (alternate)	BRCA2ex2-5F: TTTTAAGACACGCTGCAACAAAGCAGTCCTG
	R	BRCA2 Ex8-7R: CTTCATTTCTGACTATGAGC
Δ3-7	F (full length)	BR2ex2-3F(c): AAGACACGCTGCAACAAAGCAGATT
	F (alternate)	BR2ex2-8F: AAGACACGCTGCAACAAAGCAGTCA
	R	BRCA2 Ex 10R: GATCAGTATCATTTGGTTCC
Δ4	F (full length)	BRCA2ex3-4F(a): ATAAATTCAAATTAGACTTAGGAAG
	F (alternate)	BRCA2ex3-5F: ATAAATTCAAATTAGACTTAGTCCT
	R	BRCA2 Ex8-7R: CTTCATTTCTGACTATGAGC
Δ4-7	F (full length)	BR2ex3-4F(b): GATAAATTCAAATTAGACTTAGGAAGG
	F (alternate)	BR2ex3-8F: GATAAATTCAAATTAGACTTAGTCAGA
	R	BRCA2 Ex 10R: GATCAGTATCATTTGGTTCC
Δ5	F (full length)	BRCA2 Ex4-5F(a): CTAAATTCTTGTCTTAGTGAAAGTCCTG
	F (alternate)	BRCA2 Ex4-6F: CTAAATTCTTGTCTTAGTGAAAGTGGTA
	R	BRCA2 Ex8-7R: CTTCATTTCTGACTATGAGC
Δ5,6	F (full length)	BR2ex4-5F(b): CCACTTCTAAATTCTTGTCTTAGTGAAAGTCCT
	F (alternate)	BR2ex4-7F: CCACTTCTAAATTCTTGTCTTAGTGAAAGGGTC
	R	BRCA2 Ex 10R: GATCAGTATCATTTGGTTCC
Δ5-7	F (full length)	BRCA2ex4-5F(c): AAATTCTTGTCTTAGTGAAAGTCCTGT
	F (alternate)	BRCA2ex4-8F: AAATTCTTGTCTTAGTGAAAGTCAGAA
	R	BRCA2 Ex 10R: GATCAGTATCATTTGGTTCC

▼5p	F (full length)	BR2ex4-5F(b): CCACTTCTAAATTCTTGTCTTAGTGAAAGTCCT
	F (alternate)	BR2ex5+23F: CTAAATTCTTGTCTTAGTGAAAGGGA
	R	BRCA2 Ex8-7R: CTTCATTTCTGACTATGAGC
Δ6	F (full length)	BRCA2ex5-6F(a): CACCACAAAGAGATAAGTCAGTGGTA
	F (alternate)	BRCA2ex5-7F: CACCACAAAGAGATAAGTCAGGGTCG
	R	BRCA2 Ex10R: GATCAGTATCATTTGGTTCC
Δ6q7	F (full length)	BRCA2 EX 5-6F(b): ACAAAGAGATAAGTCAGTGGTAT
	F (alternate)	BRCA2 EX 5-8+2: ACAAAGAGATAAGTCAGTGTCAG
	R	BRCA2 Ex10R: GATCAGTATCATTTGGTTCC
Δ9-11	F (full length)	BR2ex8-9F: GTATTTCCTCATGATACTACTGCTAATG*
	F (alternate)	BR2ex8-12F: GTATTTCCTCATGATACTACTGCTGAGA
	R	BRCA2 Ex 14R: TGCTAAATTGCTTGAAGATT
Δ12	F (full length)	BR2EX11-12F(a): AGCCCCTTATCTTAGTGGGAGA
	F (alternate)	BR2EX11-13F: AGCCCCTTATCTTAGTGGGCAC
	R	BRCA2 Ex 14R: TGCTAAATTGCTTGAAGATT
Δ12,13	F (full length)	BR2ex11-12F(b): AAGAAGAGGAGAGCCCCTTATCTTAGTGGGAG
	F (alternate)	BR2ex11-14F: AAGAAGAGGAGAGCCCCTTATCTTAGTGGCAC
	R	BRCA2 Ex14R: TGCTAAATTGCTTGAAGATT
Δ17	F (full length)	BR2ex 16-17F(a): GGAAAAGAAGAATTTTATAGGGC
	F (alternate)	BR2ex 16-18F: GGAAAAGAAGAATTTTATAGATA
	R	BRCA2 Ex18R: TTTCAGATATATTTGCGCTC
Δ17-18	F (full length)	BR2ex16-17F(b): GGAAAGGCTGGAAAAGAAGAATTTTATAGGGC
	F (alternate)	BR2ex16-19F: GGAAAGGCTGGAAAAGAAGAATTTTATAGATT
	R	BRCA2ex22R: AACATTTGCCTGTGATTATT
Δ18	F (full length)	BR2ex17-18F: GCTTCTTCAACTAAAATACAGATATGA
	F (alternate)	BR2ex17-19F: GCTTCTTCAACTAAAATACAGATTTCT
	R	BRCA2 Ex23R: TGTATGTTAGCTCTTTCAGA
Δ 19	F (full length)	BR2ex 18-19F: GAATCTCTTATGTTAAAGATT
	F (alternate)	BR2ex 18-20F: GAATCTCTTATGTTAAAGTGG
	R	BRCA2 Ex23R: TGTATGTTAGCTCTTTCAGA
Δ20	F (full length)	BR2ex19-20F: ATTATTCAAAGAGCATACCCTATACAGTGG
	F (alternate)	BR2ex19-21F: ATTATTCAAAGAGCATACCCTATACAGAAA
	R	BRCA2 Ex23R:TGTATGTTAGCTCTTTCAGA

▼20A	F (full length)	BRCA2ex20-21F: GGAATTTGAAGAACATGAAGAAAAC
	F (alternate)	BRCA2ex20-i20F: GGAATTTGAAGAACATGAAGTTACT
	R	BRCA2 EX22R: AACATTTGCCTGTGATTATT
Δ22	F (full length)	BRCA2ex21-22F(a): CAGACCCAGCTTACCTTGAGGGT
	F (alternate)	BRCA2ex21-23F: CAGACCCAGCTTACCTTGAGTTATA
	R	BRCA2 Ex23R: TGTATGTTAGCTCTTTCAGA
Δ22,23p	F (full length)	BR2ex21-22(b): GACCCAGCTTACCTTGAGGGT
	F (alternate)	BR2ex21-delex23p: GACCCAGCTTACCTTGAGAAG
	R	BRCA2 Ex23R: TGTATGTTAGCTCTTTCAGA
Δ23p	F (full length)	BR2ex 22-23F: GCTATTCAAAAAAAGAAAAAGATTCAGTTA
	F (alternate)	BR2ex22-delex23pF: GCTATTCAAAAAAAGAAAAAGATTCAGAAG
	R	BRCA2 Ex23R: TGTATGTTAGCTCTTTCAGA

*Note: this may not always be a practical positive control as the predicted RT-PCR product is 6.5 kb.