

Table S11. Relative expression analysis of *BRCA1* mRNA junctions using qPCR, with comparison to RNA-seq data.

Sample Name	Sample #	Target	Reference	Ct Target	Ct Control	Target/Ref qPCR	Target/Ref RNA-seq†
<i>BRCA1</i> c.594-2 A>C	5	Δ 10	FL2-3	28.7	27.37	0.398	0.578
Control	19	Δ 10	FL2-3	35	28.884	0.014	0.077
Control	24	Δ 10	FL2-3	35	27.615	0.006	0.029
Control	20	Δ 10	FL2-3	35	29.439	0.021	0
Carrier:controls						28.688	16.262
<i>BRCA1</i> c.594-2 A>C	5	Δ 9-10	FL2-3	27.82	27.248	0.671	1.241
Control	19	Δ 9-10	FL2-3	28.61	27.893	0.607	0.852
Control	24	Δ 9-10	FL2-3	27.92	27.037	0.542	2.38
Control	20	Δ 9-10	FL2-3	29.33	28.595	0.601	1.055
Carrier:controls						1.15	0.869
<i>BRCA1</i> c.594-2 A>C	5	FL10-11	FL2-3	28.2	27.435	0.587	2.222
Control	19	FL10-11	FL2-3	27.97	27.84	0.915	7.66
Control	24	FL10-11	FL2-3	27.37	26.724	0.64	4.335
Control	20	FL10-11	FL2-3	28.32	28.158	0.895	3.615
Carrier:controls						0.718	0.427

†Number of raw reads mapped to the full length exon 2-3 junction compared to the target junction in targeted RNA-seq data.