

An overview of the **BRCA1**-specific amplicons and the extent of their upstream and downstream exon flanking regions

A BRCA1 exon-targeting amplicons (n=101)

[Amplicon resides within the exon]

BRCA1 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream [U] and Downstream [D] exon flanking regions covered (bp)
Exon2	99	2	94.5	[U]:+9 [D]:+71
Exon3	54	2	93.0	[U]:-68 [D]:+50
Exon4	78	2	98.5	[U]:-73 [D]:+32
Exon5	89	3	100.3	[U]:-97 [D]:+62
Exon6	140	3	94.7	[U]:-24 [D]:+90
Exon7	106	3	86.3	[U]:-49 [D]:+79
Exon8	46	3	105.7	[U]:-168 [D]:+104
Exon9	77	2	78.0	[U]:-31 [D]:+36
Exon10	3426	45	90.4	[U]:-57 [D]:-15
Exon11	89	2	79.0	[U]:-83 [D]:-33
Exon12	172	3	99.0	[U]:-24 [D]:+74

BRCA1 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream [U] and Downstream [D] exon flanking regions covered (bp)
Exon13	127	3	99.0	[U]:-34 [D]:+100
Exon14	191	3	100.7	[U]:-33 [D]:+41
Exon15	311	5	97.6	[U]:-27 [D]:+42
Exon16	88	2	105.0	[U]:-20 [D]:+31
Exon17	78	3	108.7	[U]:-83 [D]:+93
Exon18	41	2	120.5	[U]:-28 [D]:+102
Exon19	84	3	105.0	[U]:-79 [D]:+113
Exon20	55	2	115.5	[U]:-100 [D]:+33
Exon21	74	3	100.5	[U]:-66 [D]:+129
Exon22	61	2	114.0	[U]:-44 [D]:+34
Exon23	1508	3	110.7	[U]:-51 [D]:-1289

B BRCA1 intron-targeting amplicons (n=12)

BRCA1 Intron	Intron size (bp)	Number of amplicons	Mean size of amplicons (bp)
Intron3	9192	2	73.5
Intron8	1321	1	91.0
Intron10	402	1	93.0
Intron11	8368	1	93.0
Intron12	5789	3	117.0

BRCA1 Intron	Intron size (bp)	Number of amplicons	Size of amplicons (bp)
Intron15	3232	1	91.0
Intron17	500	1	96.0
Intron20	1868	1	79.0
Intron22	1840	1	114.0

Total number of BRCA1 amplicons=113

An overview of the **BRCA2**-specific amplicons and the extent of their upstream and downstream exon flanking regions

A BRCA2 exon-targeting amplicons (n=149)

[Amplicon resides within the exon]

BRCA2 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream and downstream exon flanking regions covered (bp)
Exon2	106	3	95.3	[U]:-69 [D]:+61
Exon3	249	3	116.7	[U]:-50 [D]:+28
Exon4	109	3	108.0	[U]:-101 [D]:+75
Exon5	50	2	95.5	[U]:-27 [D]:+107
Exon6	41	1	98.0	[U]:+9 [D]:+67
Exon7	115	3	104.0	[U]:-106 [D]:+30
Exon8	50	2	114.0	[U]:-63 [D]:+104
Exon9	112	3	87.7	[U]:-34 [D]:+78
Exon10	1116	14	102.9	[U]:-73 [D]:+32
Exon11	4932	56	103.4	[U]:-19 [D]:+45
Exon12	96	2	111.0	[U]:-42 [D]:+21
Exon13	70	2	95.5	[U]:-85 [D]:+22
Exon14	428	7	95.3	[U]:-76 [D]:+85

BRCA2 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream and downstream exon flanking regions covered (bp)
Exon15	182	3	105.7	[U]:-21 [D]:+88
Exon16	188	3	106.3	[U]:-67 [D]:+28
Exon17	171	3	88.0	[U]:-41 [D]:+29
Exon18	355	5	101.2	[U]:-29 [D]:+67
Exon19	156	3	99.3	[U]:-68 [D]:+49
Exon20	145	3	78.0	[U]:-35 [D]:+16
Exon21	122	3	88.7	[U]:-64 [D]:+55
Exon22	199	4	107.8	[U]:-67 [D]:+96
Exon23	164	3	103.7	[U]:-103 [D]:+15
Exon24	139	3	108.3	[U]:-90 [D]:+51
Exon25	245	4	96.0	[U]:-45 [D]:+47
Exon26	147	3	94.7	[U]:-31 [D]:+60
Exon27	1049	8	106.0	[U]:-92 [D]:-414

B BRCA2 intron-targeting amplicons (n=3)

BRCA2 Intron	Intron size (bp)	Number of amplicons	Size of amplicons (bp)
Intron6	216	1	85.0
Intron8	1426	1	117.0
Intron12	2173	1	82.0

Total number of BRCA2 amplicons=152

Estimation of the average and median upstream and downstream exon flanking regions of the BRCA1-specific amplicons

BRCA1 exon-targeting amplicons (n=101)

[Amplicon resides within the exon]

BRCA1 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream [U] and Downstream [D] exon flanking regions covered (bp)
Exon2	99	2	94.5	[U]:+9 [D]:+71
Exon3	54	2	93.0	[U]:-68 [D]:+50
Exon4	78	2	98.5	[U]:-73 [D]:+32
Exon5	89	3	100.3	[U]:-97 [D]:+62
Exon6	140	3	94.7	[U]:-24 [D]:+90
Exon7	106	3	86.3	[U]:-49 [D]:+79
Exon8	46	3	105.7	[U]:-168 [D]:+104
Exon9	77	2	78.0	[U]:-31 [D]:+36
Exon10	3426	45	90.4	[U]:-57 [D]:-15
Exon11	89	2	79.0	[U]:-83 [D]:-33
Exon12	172	3	99.0	[U]:-24 [D]:+74

BRCA1 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream [U] and Downstream [D] exon flanking regions covered (bp)
Exon13	127	3	99.0	[U]:-34 [D]:+100
Exon14	191	3	100.7	[U]:-33 [D]:+41
Exon15	311	5	97.6	[U]:-27 [D]:+42
Exon16	88	2	105.0	[U]:-20 [D]:+31
Exon17	78	3	108.7	[U]:-83 [D]:+93
Exon18	41	2	120.5	[U]:-28 [D]:+102
Exon19	84	3	105.0	[U]:-79 [D]:+113
Exon20	55	2	115.5	[U]:-100 [D]:+33
Exon21	74	3	100.5	[U]:-66 [D]:+129
Exon22	61	2	114.0	[U]:-44 [D]:+34
Exon23	1508	3	110.7	[U]:-51 [D]:-1289

Upstream [U] exon flanking region

for BRCA1-specific amplicons:

Average length=59bp

Median length=51bp

Downstream [D] exon flanking region

for BRCA1-specific amplicons:

Average length=70.3bp

Median length= 71bp

Estimation of the average and median upstream and downstream exon flanking regions of the BRCA2-specific amplicons

BRCA2 exon-targeting amplicons (n=149)

[Amplicon resides within the exon]

BRCA2 exon	Exon size (bp)	Number of amplicons	Mean size of amplicons (bp)	Upstream and downstream exon flanking regions covered (bp)
Exon2	106	3	95.3	[U]:-69 [D]:+61
Exon3	249	3	116.7	[U]:-50 [D]:+28
Exon4	109	3	108.0	[U]:-101 [D]:+75
Exon5	50	2	95.5	[U]:-27 [D]:+107
Exon6	41	1	98.0	[U]:+9 [D]:+67
Exon7	115	3	104.0	[U]:-106 [D]:+30
Exon8	50	2	114.0	[U]:-63 [D]:+104
Exon9	112	3	87.7	[U]:-34 [D]:+78
Exon10	1116	14	102.9	[U]:-73 [D]:+32
Exon11	4932	56	103.4	[U]:-19 [D]:+45
Exon12	96	2	111.0	[U]:-42 [D]:+21
Exon13	70	2	95.5	[U]:-85 [D]:+22
Exon14	428	7	95.3	[U]:-76 [D]:+85

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Exon15	182	3	105.7	[U]:-21 [D]:+88
Exon16	188	3	106.3	[U]:-67 [D]:+28
Exon17	171	3	88.0	[U]:-41 [D]:+29
Exon18	355	5	101.2	[U]:-29 [D]:+67
Exon19	156	3	99.3	[U]:-68 [D]:+49
Exon20	145	3	78.0	[U]:-35 [D]:+16
Exon21	122	3	88.7	[U]:-64 [D]:+55
Exon22	199	4	107.8	[U]:-67 [D]:+96
Exon23	164	3	103.7	[U]:-103 [D]:+15
Exon24	139	3	108.3	[U]:-90 [D]:+51
Exon25	245	4	96.0	[U]:-45 [D]:+47
Exon26	147	3	94.7	[U]:-31 [D]:+60
Exon27	1049	8	106.0	[U]:-92 [D]:-414

Upstream [U] exon flanking region

for BRCA2-specific amplicons:

Average length=59.9bp

Median length=64bp

Downstream [D] exon flanking region

for BRCA2-specific amplicons:

Average length=54.2bp

Median length=51bp