

Supplementary Table 1: Target regions for *BARD1*, *BRIP1*, *NBN* and *PALB2* sequencing.

exon	# amplicons	Target region start	Target region stop	Amplicon size
<i>BARD1</i> , chromosome 2				
11	3	215593354	215593765	411
10	1	215595092	215595278	186
9	1	215609757	215609929	172
8	1	215610423	215610622	199
7	1	215617142	215617328	186
6	2	215632095	215632418	323
5	1	215633927	215634126	199
4	13	215645150	215646293	1143
3	1	215657067	215657218	151
2	1	215661801	215661902	101
1	1	215674121	215674320	199
<i>BRIP1</i> , chromosome 17				
20	9	59760555	59761557	1002
19	4	59763152	59763560	408
18	2	59770713	59770942	229
17	2	59793292	59793548	256
16	1	59820350	59820533	183
15	2	59821760	59822044	284
14	2	59853694	59853963	269
13	2	59857611	59857817	206
12	2	59858152	59858416	264
11	2	59861584	59861836	252
10	1	59870923	59871122	199
9	2	59876415	59876721	306
8	2	59878566	59878879	313
7	4	59885725	59886147	422
6	1	59924424	59924617	193
5	2	59926404	59926667	263
4	2	59934388	59934660	272
3	1	59937113	59937296	183
2	2	59938723	59938979	256
1	0	N/A	N/A	non coding
<i>NBN</i> , chromosome 8				
16	1	90947748	90947873	125
15	1	90949219	90949342	123
14	1	90955456	90955651	195
13	2	90958309	90958598	289
12	1	90960004	90960171	167
11	4	90965415	90965969	554
10	3	90967421	90967823	402
9	1	90970923	90971121	198
8	2	90976533	90976820	287
7	2	90982539	90982808	269
6	3	90983302	90990550	7248
5	1	90990451	90990650	199
4	2	90992902	90993175	273
3	2	90993553	90993809	256
2	2	90994896	90995147	251
1	1	90996705	90996847	142
<i>PALB2</i> , chromosome 16				
13	3	23614654	23615081	427
12	2	23619085	23619361	276
11	1	23625297	23625476	179
10	1	23632650	23632848	198
9	2	23634179	23634489	310
8	1	23635297	23635483	186
7	2	23637455	23637758	303
6	1	23640491	23640672	181
5	8	23640861	23641834	973
4	14	23646130	23647668	1538
3	1	23649139	23649301	162
2	1	23649327	23649505	178
1	1	23652356	23652513	157

Supplementary Table 2: Predicted deleterious mutations identified in ovarian cancer cases and controls

Index	Gene	Case/ control	Mutation details			Patient details			
			Nucl. Change*	Type	Predicted truncation	Age Dx	Tumor histology	OC FH	BC FH
4415	BARD1	Case	c.623dupA	FS	p.K208fs	55	HGS	0	0
4220	BARD1	Case	c.627_628delAA	FS	p.K209fs	52	HGS	0	0
3959	BARD1	Case	c.1690C>T	NS	p.Q564*	54	epithelial	0	0
2001	BARD1	Control	c.1996C>T	NS	p.Q666*	48	NA	0	1
1913	BARD1	Case	c.2300_2301delTG	FS	p.V767fs	39	HGS	0	0
5644	BRIP1	Case	c.55dupT	FS	p.Y19fs	60	HGS	0	0
4416	BRIP1	Case	c.78dupT	FS	p.A27fs	65	HGS	0	0
3050	BRIP1	Case	c.93+1G>T (1.07)	Sp	No start codon	48	HGS	0	0
6822	BRIP1	Case	c.133G>T	NS	p.E45*	66	HGS	0	0
2735	BRIP1	Case	c.168_171delACTT	FS	p.L56fs	76	HGS	0	0
1500	BRIP1	Case	c.379+1G>T (0.96)	Sp	In frame 58aa del	78	HGS	0	0
2250	BRIP1	Case	c.394dupA	FS	p.T132fs	75	HGS	0	0
6554	BRIP1	Case	c.394dupA	FS	p.T132fs	54	HGS	0	0
6033	BRIP1	Case	c.918+1G>A (0.86)	Sp	In frame 97aa del	68	LGS	0	0
5670	BRIP1	Case	c.919-2A>G (0.88)	Sp	In frame 74aa del	55	HGS	0	0
3380	BRIP1	Case	c.1236delA	FS	p.E412fs	53	HGS	0	0
1380	BRIP1	Case	c.1348G>T	NS	p.E450*	66	HGS	0	0
4606	BRIP1	Case	c.1510delA	FS	p.I504fs	57	HGS	0	0
1055	BRIP1	Case	c.1845delT	FS	p.G615fs	47	HGS	0	0
1382	BRIP1	Case	c.1845delT	FS	p.G615fs	58	serous	0	0
4513	BRIP1	Case	c.1871C>A	NS	p.S624*	71	HGS	0	0
6459	BRIP1	Case	c.1871C>A	NS	p.S624*	58	LGS	0	1
5801	BRIP1	Case	c.2010dupT	FS	p.E671fs	69	HGS	0	0
1010	BRIP1	Case	c.2010dupT	FS	p.E671fs	73	HGS	0	0
2930	BRIP1	Case	c.2038_2039dupTT	FS	p.L680fs	55	HGS	0	0
2753	BRIP1	Case	c.2108_2109insCC	FS	p.K703fs	74	HGS	0	0
1458	BRIP1	Case	c.2108_2109insCC	FS	p.K703fs	65	HGS	0	0
1534	BRIP1	Case	c.2108_2109insCC	FS	p.K703fs	74	HGS	0	0
908	BRIP1	Case	c.2255_2256delAA	FS	p.K752fs	65	serous	1	1
6500	BRIP1	Case	c.2255_2256delAA	FS	p.K752fs	50	LGS	0	1
4472	BRIP1	Case	c.2392C>T	NS	p.R798*	82	HGS	0	0
2577	BRIP1	Case	c.2392C>T	NS	p.R798*	67	HGS	0	0
1629	BRIP1	Case	c.2493-1G>C (0.80)	Sp	Stop at aa 834	56	HGS	0	0
5645	BRIP1	Case	c.2507_2508delGA	FS	p.R836fs	65	HGS	1	0
5201	BRIP1	Control	c.2575+1G>C (0.88)	Sp	Stop at aa 834	68	NA	0	0
11	BRIP1	Control	c.2765T>G	NS	p.L922*	52	NA	NA	NA
6975	BRIP1	Control	c.3401delC	FS	p.P1134fs	53	NA	0	0
6453	BRIP1	Case	c.3607G>T	NS	p.E1203*	68	LGS	0	0
3401	NBN	Case	c.38-10T>A (0.43)	Sp	Stop at aa 17	59	HGS	0	0
4303	NBN	Case	c.38-10T>A (0.43)	Sp	Stop at aa 17	55	HGS	0	0
6626	NBN	Control	c.38-10T>A (0.43)	Sp	Stop at aa 17	53	NA	0	0
1548	NBN	Case	c.38-3C>G (1.50)	Sp	Stop at aa 17	75	serous	0	0
4013	NBN	Case	c.657_661delACAAA	FS	p.K219fs	41	serous	0	0
4517	NBN	Case	c.657_661delACAAA	FS	p.K219fs	66	HGS	0	1
2433	NBN	Control	c.657_661delACAAA	FS	p.K219fs	70	NA	0	1
3919	NBN	Control	c.657_661delACAAA	FS	p.K219fs	48	NA	0	0
1189	NBN	Control	c.657_661delACAAA	FS	p.K219fs	54	NA	0	0
1262	NBN	Control	c.657_661delACAAA	FS	p.K219fs	50	NA	0	0
6895	NBN	Case	c.698_701delAACA	FS	p.K233fs	48	HGS	0	0
5947	NBN	Control	c.808_809delGT	FS	p.V270fs	48	NA	0	0
3685	NBN	Control	c.994+2T>C (0.91)	Sp	Stop at aa 316	26	NA	0	0
4166	NBN	Case	c.995-2A>G	Sp	Stop at aa 334	62	HGS	0	0
6191	NBN	Case	c.1125-2A>G (0.73)	Sp	In frame 91aa del	56	HGS	0	0
7096	NBN	Case	c.1142delC	FS	p.P381fs	64	LGS	0	0
3539	NBN	Control	c.2071-1G>C (0.91)	Sp	In frame 38aa del	33	NA	0	0
3240	PALB2	Case	c.509_510delGA	FS	p.R170fs	46	unknown	0	0
5235	PALB2	Case	c.1227_1231delTGTTA	FS	p.Y409fs	67	HGS	0	0
5822	PALB2	Case	c.3113G>A	NS	p.W1038*	40	LGS	0	0
6165	PALB2	Case	c.3113G>A	NS	p.W1038*	63	HGS	0	0
2391	PALB2	Control	c.3113G>A	NS	p.W1038*	54	NA	0	1
1199	PALB2	Control	c.3113G>A	NS	p.W1038*	56	NA	0	0
1626	PALB2	Control	c.3113+5G>C (0.60)	Sp	In frame 39aa del	52	NA	0	0
5778	PALB2	Case	c.3256C>T	NS	p.R1086*	50	HGS	0	0
3151	PALB2	Case	c.3507_3508delTC	FS	p.S1169fs	70	HGS	0	0
958	PALB2	Case	c.3549C>A	NS	p.Y1183*	47	CCC	1	1
4465	PALB2	Case	c.3549C>A	NS	p.Y1183*	33	HGS	1	0
1066	PALB2	Case	c.3549C>A	NS	p.Y1183*	67	HGS	0	0

Supplementary 3: Deleterious mutations identified in *BARD1*, *BRIP1*, *NBN*, *PALB2* in UKFOCSS subjects

ID	Gene	Nucl. Change*	Mutation details		Proband Characteristics		Ovarian- breast cancer family history			
			Type	Predicted truncation	Ref Age	Breast cancer (Age)	OvCa 1 st deg.	BrCa 1 st deg.	OvCa 1 st /2 nd deg.	BrCa 1 st /2 nd deg.
1	<i>BARD1</i>	c.1212C>G	NS	p.Y404X	37	No	1	0	2	0
2	<i>BARD1</i>	c.1921C>T	NS	p.R641X	53	No	0	0	0	0
3	<i>BARD1</i>	c.2291_2294delTAGA	FS	p.I764fs	42	No	1	1	1	1
All							2	1	3	1
4	<i>BRIP1</i>	c.66C>A	NS	p.Y22X	43	39	1	1	1	3
5	<i>BRIP1</i>	c.128_131delTGTT	FS	p.L43fs	40	No	2	0	2	0
6	<i>BRIP1</i>	c.133G>T	NS	p.E45X	0	Yes (34)	0	1	1	0
7	<i>BRIP1</i>	c.778insT	FS	p.T260fs	43	No	1	0	1	2
8	<i>BRIP1</i>	c.890delA	FS	p.K297fs	54	No	1	0	2	0
9	<i>BRIP1</i>	c.1058insA	FS	p.Y353*	44	No	1	1	1	1
10	<i>BRIP1</i>	c.1871C>A	NS	p.S624X	51	Yes (39)	1	0	1	0
11	<i>BRIP1</i>	c.1871C>A	NS	p.S624X	46	No	1	0	2	0
12	<i>BRIP1</i>	c.2096insCC	FS	p.K703fs	55	No	1	0	2	1
13	<i>BRIP1</i>	c.2255_2256delAA	FS	p.K752fs	48	No	1	0	2	1
14	<i>BRIP1</i>	c.2400C>G	NS	p.Y800X	64	No	3	1	3	1
15	<i>BRIP1</i>	c.2990_2993delCAAA	FS	p.T997fs	51	No	1	0	1	1
All							14	4	19	10
16	<i>NBN</i>	c.481-1G>A	SP	Stop 166	35	No	0	1	0	3
17	<i>NBN</i>	c.657_661delCAAA	FS	p.K219fs	30	No	0	1	0	3
18	<i>NBN</i>	c.1142delC	FS	p.P381fs	65	Yes (47)	0	1	1	1
All							0	3	1	7
19	<i>PALB2</i>	c.172_175delTTGTC	FS	p.Q60fs	42	Yes (39)	1	2	1	3
20	<i>PALB2</i>	c.509_510delGA	FS	p.R170fs	51	No	0	1	1	1
21	<i>PALB2</i>	c.2167_2168delAT	FS	p.M723fs	68	No	1	0	2	0
22	<i>PALB2</i>	c.2488delG	FS	p.E830fs	45	No	1	0	1	0
23	<i>PALB2</i>	c.3113G>A	NS	p.W1038X	46	Yes (44)	0	1	1	4
24	<i>PALB2</i>	c.3113G>A	NS	p.W1038X	44	No	0	1	1	1
25	<i>PALB2</i>	c.3549C>G	NS	p.Y1183X	35	No	0	1	1	1
All							3	6	8	10

Supplementary table 4: Non synonymous variants identified in *BARD1*, *BRIP1*, *NBN* and *PALB2*. Variants are those in which at least two of three measures of pathogenicity (Polyphen, Sift and PROVEAN) indicate they are possibly deleterious.

Variant	Cases	Controls	Total
<i>BARD1</i>			
p.A40V	1		1
p.D710V	1		1
p.E652G	1		1
p.G517R		1	1
p.G681V		1	1
p.H433P		1	1
p.H471Y		1	1
p.H466R		1	1
p.H483R	1		1
p.I525M		1	1
p.K205N	1		1
p.K205R	1		1
p.L211S	1		1
p.L220S	2	2	4
p.L432F		1	1
p.L447V	1		1
p.L772W		1	1
p.M26L	1		1
p.P167L		1	1
p.P464S	1		1
p.P747R	1		1
p.P89L		1	1
p.R38H	1		1
p.S363Y	1		1
p.S519Y	1	1	2
p.S586I	1		1
p.S760L	1		1
p.T463I		1	1
p.T562I		1	1
p.T605A	1		1
p.T714I	1		1
p.W462S	2		2
p.W629R	1		1
p.Y745D	1		1
Total	24	15	39
<i>BRIP1</i>			
p.A21V		1	1
p.A778T	1		1
p.C175R	1		1
p.C350R	1		1
p.C832Y		1	1
p.D1189E	1		1
p.D184Y	2	2	4
p.D791V		1	1
p.D846G	1		1
p.E1188V	4	2	6
p.E45K	1		1
p.E726K	1		1
p.G330E		1	1
p.G35R	1		1
p.G649S		1	1
p.G712D	1		1
p.G813D	1		1
p.H314R		1	1
p.H396R	2		2
p.I552T		1	1
p.I633M	1	1	2
p.I691L	1		1
p.K703I	3		3
p.K797N	1		1
p.L347P	1		1
p.N429T	1		1
p.P244A	1		1
p.P47A	4	2	6
p.Q740H	3	6	9
p.R264W	12	12	24
p.R419P	1		1
p.R419W	2	5	7

Variant	Cases	Controls	Total
p.R658W	2		2
p.R814C	1		1
p.R855C	2		2
p.R855H	1		1
p.S1239C		1	1
p.S206L	2		2
p.S31P		1	1
p.S624L	1		1
p.T1216I		1	1
p.T354I		1	1
p.T48I		1	1
p.T48K	1		1
p.Y22H	1		1
p.Y22N	1		1
p.Y352H	1		1
p.Y817C		1	1
Total	62	43	105
<i>NBN</i>			
p.D211E		1	1
p.D284A	1	1	2
p.D295V	5	1	6
p.D469Y	1		1
p.D95G	1		1
p.D95N	16	13	29
p.E179V		1	1
p.E254G	1		1
p.E552G	1		1
p.E564K		1	1
p.E658G		1	1
p.F106I		1	1
p.G224E	1		1
p.G246D		1	1
p.G246S		1	1
p.I171V	19	16	35
p.I290F	5	1	6
p.I293K	4	1	5
p.K237E		1	1
p.L297F		1	1
p.L297P		1	1
p.M152I	1	1	2
p.M294R	5	1	6
p.M296R		1	1
p.P170L		1	1
p.P266L		2	2
p.P357L		1	1
p.Q730R		1	1
p.R215W	22	20	42
p.S118T	1		1
p.S292P	5	1	6
p.S488Y	1		1
p.S93L	2		2
p.T226I	1		1
p.T268M		5	5
p.T76I		1	1
p.V151F	1	1	2
p.V210F	4	3	7
p.V691G		1	1
p.W289G	5	1	6
p.W722S		1	1
Total	102	85	187
<i>PALB2</i>			
p.A22V		1	1
p.D1053G	1		1
p.E1120V	1		1
p.E892K		1	1
p.G1043A		1	1
p.G514V		1	1
p.H1170Y	2	1	3

Variant	Cases	Controls	Total
p.I1051T		1	1
p.K18R		1	1
p.L1092P	1		1
p.L1143H	1	1	2
p.L362R	1		1
p.L583W	1		1
p.L939W	8	10	18
p.M1049T		1	1
p.P210R	1		1
p.P615H		1	1
p.P758L		1	1
p.P864S	34	31	65
p.R37C	1		1
p.R37H	1		1
p.S1054P	1		1
p.S382I		1	1
p.S417Y	2		2
p.S47Y	1		1
p.S804F		1	1
p.S869C		1	1
p.T1030A		1	1
p.T1099R		1	1
p.T247R	1		1
p.T275I		2	2
p.T734S	1	2	3
p.W1164C		1	1
Total^c	59	62	121