

Table 1: Characteristics of ovarian cancer case-control populations analyzed in this study

Study	Country	Controls		Cases					
		Number	Mean age (range)	Number	Mean age (range)	High-grade serous ^c	(%)	Stage 3/4	(%)
AOC	Australia	648	57 (20-80)	630	61 (23-80)	573	(91)	545	(87)
GRR ^a	USA	0	NA	126	48 (21-80)	70	(56)	NA	
HJO/HMO	Germany /Belarus	526	36 (18-68)	335	58 (18-88)	173	(52)	156	(47)
LAX	USA	210	61 (34-90)	199	62 (32-88)	199	(100)	183	(92)
MAY	USA	660	63 (26-93)	650	64 (23-91)	642	(99)	581	(89)
RMH ^b	UK	0	NA	64	53 (27-73)	64	(100)	NA	
SEA	UK	843	53 (28-66)	712	57 (24-74)	356	(50)	393	(55)
STA	USA	171	48 (20-66)	163	53 (23-64)	129	(79)	121	(74)
UKO	UK	373	65 (52-78)	357	61 (25-90)	329	(92)	244	(63)
Total		3,431	55 (18-93)	3,236	58 (18-91)	2535	(78)	2223	(69)

All studies are case-control studies except for: ^a familial ovarian cancer registry study and ^b case only hospital study; ^c includes 304 cases that were serous subtype but of unknown grade

Table 2: Truncating mutations in *BRIP1*, *BARD1*, *NBN* and *PALB2* identified in ovarian cancer cases and controls and in the UKFOCSS subjects

Subjects	Carrier status	<i>BARD1</i>		<i>BRIP1</i>		<i>NBN</i>		<i>PALB2</i>	
		N	%	N	%	N	%	N	%
Cases	Carrier	4	0.12	30	0.92	9	0.28	9	0.28
	Non carrier	3,257	99.88	3,227	99.08	3,248	99.72	3,248	99.72
Controls	Carrier	2	0.06	3	0.09	8	0.23	3	0.09
	Non carrier	3,447	99.94	3,444	99.91	3,439	99.77	3,444	99.91
P-value			0.39		0.0001		0.61		0.078
UKFOCSS	Carrier	3	0.15	12	0.6	3	0.15	7	0.35
	Non carrier	1997	99.85	1988	99.40	1997	99.85	1993	99.65
P-value ^a			0.15		0.0008		0.045		0.76

^a Based on a comparison of the frequency of deleterious mutations identified in UKFOCSS subjects compared to mutation frequency in controls from the other studies

Table 3: Mutation frequency by clinical characteristics in ovarian cancer cases

Characteristic	Subjects	Carrier status	BARD1		BRIP1		NBN		PALB2	
			N	%	N	%	N	%	N	%
Age at diagnosis	<40 yrs	Carrier	1	25	0	0	0	0	1	11
		Non carrier	148	4	149	5	149	5	14	5
	40-59 yrs	Carrier	0	0	2	7	2	22	3	33
		Non carrier	460	14	458	14	458	14	457	14
	50-59 yrs	Carrier	3	75	9	30	3	33	1	11
		Non carrier	977	30	971	30	977	30	979	30
	≥ 60yrs	Carrier	0	0	19	63	4	44	4	44
		Non carrier	1662	51	1643	51	1658	15	1658	51
Tumor Subtype	High-grade serous	Carrier	3	75	25	83	6	67	6	67
		Non carrier	2279	70	2257	70	2276	70	2276	70
	Other	Carrier	1	25	5	17	3	33	3	33
		Non carrier	974	30	970	30	972	30	972	30
	None	Carrier	4	100	26	87	8	89	7	78
		Non carrier	2,357	80	2,335	80	2353	79	2,354	79
Family history	OvCa family history	Carrier	0	0	2	7	0	0	2	22
		Non carrier	275	9	273	9	275	9	273	9
	BrCa family history	Carrier	0	0	3	10	1	11	1	11
		Non carrier	427	14	424	14	426	14	426	14
	Ov/Br Ca family history	Carrier	0	0	4	13	1	11	2	22
		Non carrier	612	21	608	21	611	21	610	21

Table 4: Mutation frequency by family history in UCKFOCSS subjects

Family history	Carrier status	<i>BARD1</i>		<i>BRIP1</i>		<i>NBN</i>		<i>PALB2</i>	
		N	(%)	N	(%)	N	(%)	N	(%)
1 ovarian cancer, ≤ 2 breast cancer case (under 60yrs)	Carrier	1	(33)	5	(42)	2	(66)	4	(57)
	Non carrier	1087	(54)	1083	(54)	1086	(54)	1084	(54)
2 ovarian cancer	Carrier	1	(33)	6	(50)	0	(0)	1	(14)
	Non carrier	558	(28)	553	(28)	559	(28)	559	(28)
Other related cancers in extended family	Carrier	1	(33)	1	(8)	1	(33)	2	(29)
	Non carrier	283	(14)	283	(14)	283	(14)	282	(14)

30 individuals with 1 OV & 3 colorectal cancers (1 <50y) in primary relatives and 39 individuals with ≥ 3 breast cancers had no carriers detected for any gene.