

Supplementary Table 3. Clinical features of the tumors associated with DNA damage response gene variants

A. All *BRCA1* and *BRCA2* ^a

Category	Comparisons	N=21 <i>BRCA1/2</i> var	% of valid	N=2046 Wild type	% of valid	OR	95% CI	p
Tumor								
1		11	55%	1200	64%			
2		8	40%	584	31%			
3		1	5%	68	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	1.46	0.60–3.53	0.484
Nodus								
Neg		13	62%	1302	64%			
Pos	posit vs. neg	8	38%	725	36%	1.11	0.46–2.68	1.000
Metastasis								
Neg		20	95%	1993	98%			
Pos	posit vs. neg	1	5%	45	2%	2.21	0.29–16.86	0.379
ER								
Neg		6	30%	269	14%			
Pos	neg vs. posit	14	70%	1666	86%	2.65	1.01–6.97	0.051
PR								
Neg		9	45%	431	23%			
Pos	neg vs. posit	11	55%	1486	78%	2.82	1.16–6.85	0.028
HER2								
Neg		18	95%	1583	85%			
Pos	posit vs. neg	1	5%	281	15%	0.31	0.04–2.35	0.340
Grade								
1		1	5%	373	19%			
2		7	37%	914	47%			
3	3 vs. 1 and 2	11	58%	672	34%	2.63	1.05–6.58	0.049
Tumor histology								
Ductal		16	76%	1587	78%			
Lobular		4	19%	309	15%			
Other	Duct vs. others	1	5%	139	7%	0.90	0.33–2.48	0.794
Type								
LumA		13	68%	1414	76%			
LumB		0	0%	199	11%			
HER2 pos		1	5%	77	4%			
Triple-neg	TN vs. others	5	26%	160	9%	3.76	1.34–10.58	0.021
Ki67								
Neg		0	0%	145	8%			
1+		4	21%	763	41%			
2+	high vs. low	3	16%	501	27%			
3+	3 vs. others	12	63%	455	24%	5.31	2.08–13.56	0.0004
Multifocal								
Yes		3	14%	325	16%			
No	yes vs. no	18	86%	1718	84%	0.88	0.26–3.01	1.000
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	21	100%	1996	98%	NA	NA	1.000
Mean age at diag.		51 y		59 y				0.002

B. All *BRCA1*^a

		N=7		N=2062				
Category	Comparisons	BRCA1 var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		2	29%	1211	64%			
2		4	57%	588	31%			
3		1	14%	68	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	4.46	0.86–23.04	0.106
Nodus								
Neg		7	100%	1310	64%			
Pos	posit vs. neg	0	0%	733	36%	NA	NA	0.055
Metastasis								
Neg		7	100%	2008	98%			
Pos	posit vs. neg	0	0%	46	2%	NA	NA	1.000
ER								
Neg		4	57%	271	14%			
Pos	neg vs. posit	3	43%	1679	86%	8.26	1.84–37.11	0.009
PR								
Neg		5	71%	435	23%			
Pos	neg vs. posit	2	29%	1497	77%	8.60	1.66–44.50	0.008
HER2								
Neg		7	100%	1596	85%			
Pos	posit vs. neg	0	0%	282	15%	NA	NA	0.603
Grade								
1		0	0%	374	19%			
2		2	33%	920	47%			
3	3 vs. 1 and 2	4	67%	680	34%	3.81	0.70–20.83	0.191
Tumor histology								
Ductal		5	71%	1598	78%			
Lobular		1	14%	313	15%			
Other	Duct vs. others	1	14%	140	7%	0.71	0.14–3.67	0.654
Type								
LumA		3	43%	1426	77%			
LumB		0	0%	199	11%			
HER2 pos		0	0%	78	4%			
Triple-neg	TN vs. others	4	57%	161	9%	14.10	3.13–63.57	0.002
Ki67								
Neg		0	0%	145	8%			
1+		0	0%	769	41%			
2+	high vs. low	2	29%	502	27%			
3+	3 vs. others	5	71%	462	25%	7.66	1.48–39.63	0.012
Multifocal								
Yes		0	0%	328	16%			
No	yes vs. no	7	100%	1730	84%	NA	NA	0.606
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	7	100%	2012	98%	NA	NA	1.000
Mean age at diag.		47 y		59 y				0.015

C. All *BRCA2*^a

		N=14		N=2056				
Category	Comparisons	BRCA2 var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		9	69%	1204	64%			
2		4	31%	589	31%			
3		0	0%	69	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	0.79	0.24–2.56	0.78
Nodus								
Neg		6	43%	1310	64%			
Pos	posit vs. neg	8	57%	727	36%	2.40	0.83–6.95	0.159
Metastasis								
Neg		13	93%	2003	98%			
Pos	posit vs. neg	1	7%	45	2%	3.42	0.44–26.74	0.272
ER								
Neg		2	15%	274	14%			
Pos	neg vs. posit	11	85%	1671	86%	1.11	0.24–5.03	0.704
PR								
Neg		4	31%	437	23%			
Pos	neg vs. posit	9	69%	1490	77%	1.52	0.46–4.95	0.508
HER2								
Neg		11	92%	1593	85%			
Pos	posit vs. neg	1	8%	281	15%	0.52	0.07–4.01	1.000
Grade								
1		1	8%	373	19%			
2		5	38%	917	47%			
3	3 vs. 1 and 2	7	54%	678	34%	2.22	0.74–6.63	0.153
Tumor histology								
Ductal		11	79%	1595	78%			
Lobular		3	21%	310	15%			
Other	Duct vs. others	0	0%	140	7%	1.03	0.29–3.72	1.000
Type								
LumA		10	83%	1419	76%			
LumB		0	0%	199	11%			
HER2 pos		1	8%	77	4%			
Triple-neg	TN vs. others	1	8%	165	9%	0.93	0.12–7.28	1.000
Ki67								
Neg		0	0%	145	8%			
1+		4	33%	764	41%			
2+	high vs. low	1	8%	503	27%			
3+	3 vs. others	7	58%	462	25%	4.28	1.35–13.55	0.014
Multifocal								
Yes		3	21%	326	16%			
No	yes vs. no	11	79%	1727	84%	1.45	0.40–5.21	0.477
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	14	100%	2006	98%	NA	NA	1.000
Mean age at diag.		52 y		59 y		0.036		

D. *PALB2* c.1592delT (p.Leu531fs), rs180177102, 16-23634953-CA-C

		N=16		N=2054				
Category	Comparisons	PALB2 var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		7	47%	1208	64%			
2		7	47%	585	31%			
3		1	7%	68	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	2.04	0.74–5.66	0.182
Nodus								
Neg		6	34%	1311	64%			
Pos	posit vs. neg	10	63%	725	36%	3.01	1.09–8.33	0.035
Metastasis								
Neg		15	94%	2002	98%			
Pos	posit vs. neg	1	6%	45	2%	2.97	0.38–22.94	0.304
ER								
Neg		3	20%	273	14%			
Pos	neg vs. posit	12	80%	1671	86%	1.53	0.43–5.46	0.457
PR								
Neg		7	47%	433	23%			
Pos	neg vs. posit	8	53%	1493	78%	3.02	1.09–8.37	0.055
HER2								
Neg		13	87%	1593	85%			
Pos	posit vs. neg	2	13%	279	15%	0.88	0.20–3.91	1.000
Grade								
1		0	0%	374	19%			
2		4	27%	919	47%			
3	3 vs. 1 and 2	11	73%	674	34%	5.28	1.67–16.63	0.002
Tumor histology								
Ductal		15	94%	1589	78%			
Lobular		1	6%	313	15%			
Other	Duct vs. others	0	0%	141	7%	4.28	0.57–32.53	0.221
Type								
LumA		10	67%	1421	76%			
LumB		2	13%	196	11%			
HER2 pos		0	0%	78	4%			
Triple-neg	TN vs. others	3	20%	163	9%	2.6	0.73–9.31	0.141
Ki67								
Neg		0	0%	145	8%			
1+		3	20%	767	41%			
2+	high vs. low	3	20%	501	27%			
3+	3 vs. others	9	60%	459	25%	4.62	1.64–13.04	0.004
Multifocal								
Yes		2	13%	327	16%			
No	yes vs. no	14	88%	1724	84%	0.75	0.17–3.33	1.000
Bilateral								
Yes		1	6%	49	2%			
No	yes vs. no	15	94%	2005	98%	2.73	0.35–21.06	0.325
Mean age at diag.		60 y		58 y		0.587		

E. ATM c.7570G>C (p.Ala2524Pro), rs769142993, 11-108331498-G-C

		N=13		N=2057				
Category	Comparisons	ATM var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		10	77%	1204	64%			
2		3	23%	590	31%			
3		0	0%	69	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	0.53	0.15–1.93	0.398
Nodus								
Neg		9	69%	1309	64%			
Pos	posit vs. neg	4	31%	730	36%	0.80	0.25–2.60	0.781
Metastasis								
Neg		13	100%	2004	98%			
Pos	posit vs. neg	0	0%	46	2%	NA	NA	1.000
ER								
Neg		0	0%	276	14%			
Pos	neg vs. posit	13	100%	1669	86%	NA	NA	0.236
PR								
Neg		0	0%	440	23%			
Pos	neg vs. posit	13	100%	1487	77%	NA	NA	0.049
HER2								
Neg		11	85%	1594	85%			
Pos	posit vs. neg	2	15%	279	15%	1.04	0.23–4.71	1.000
Grade								
1		1	8%	373	19%			
2		8	62%	915	47%			
3	3 vs. 1 and 2	4	31%	681	35%	0.84	0.26–2.74	1.000
Tumor histology								
Ductal		10	77%	1594	78%			
Lobular		3	23%	311	15%			
Other	Duct vs. others	0	0%	141	7%	0.95	0.26–3.45	1.000
Type								
LumA		11	85%	1419	76%			
LumB		2	15%	196	11%			
HER2 pos		0	0%	78	4%			
Triple-neg	LumA vs. others	0	0%	166	9%	1.71	0.38–7.72	0.744
Ki67								
Neg		1	8%	144	8%			
1+		3	23%	767	41%			
2+	high vs. low	7	54%	497	27%			
3+	3 vs. others	2	15%	465	25%	0.55	0.12–2.49	0.747
Multifocal								
Yes		2	15%	327	16%			
No	yes vs. no	11	85%	1727	84%	0.96	0.21–4.35	1.000
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	13	100%	2007	98%	NA	NA	1.000
Mean age at diag.		60 y		58 y		0.711		

F. *CHEK2* c.1100delC (p.Thr367fs), rs555607708, 22-28695868-AG-A

		N=59		N=2012				
Category	Comparisons	CHEK2 var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		37	65%	1177	64%			
2		16	28%	577	31%			
3		1	2%	68	4%			
4	2, 3 and 4 vs. 1	3	5%	20	1%	0.96	0.55–1.66	0.890
Nodus								
Neg		35	60%	1283	64%			
Pos	posit vs. neg	23	40%	711	36%	1.19	0.70–2.02	0.579
Metastasis								
Neg		56	97%	1961	98%			
Pos	posit vs. neg	2	3%	44	2%	1.59	0.38–6.73	0.373
ER								
Neg		3	5%	273	14%			
Pos	neg vs. posit	53	95%	1630	86%	0.34	0.11–1.09	0.076
PR								
Neg		5	9%	436	23%			
Pos	neg vs. posit	51	91%	1449	77%	0.33	0.13–0.82	0.014
HER2								
Neg		44	82%	1561	85%			
Pos	posit vs. neg	10	19%	272	15%	1.30	0.65–2.62	0.561
Grade								
1		7	13%	367	19%			
2		29	53%	894	46%			
3	3 vs. 1 and 2	19	35%	666	35%	1.00	0.57–1.76	1.000
Tumor histology								
Ductal		47	80%	1558	78%			
Lobular		11	19%	303	15%			
Other	Duct vs. others	1	2%	140	7%	1.11	0.59–2.12	0.756
Type								
LumA		44	81%	1386	76%			
LumB		8	15%	191	11%			
HER2 pos		2	4%	76	4%			
Triple-neg	LumA vs. others	0	0%	166	9%	1.38	0.69–2.75	0.420
Ki67								
Neg		1	2%	144	8%			
1+		20	36%	750	41%			
2+	high vs. low	19	35%	485	26%			
3+	3 vs. others	15	27%	453	25%	1.14	0.63–2.09	0.751
Multifocal								
Yes		11	19%	318	16%			
No	yes vs. no	48	81%	1690	84%	1.22	0.63–2.37	0.587
Bilateral								
Yes		2	3%	48	2%			
No	yes vs. no	57	97%	1964	98%	1.44	0.34–6.05	0.652
Mean age at diag.		56 y		58 y		0.160		

G. *FANCM* c.5101C>T and c.5791C>T combined

		N=46*	N=2024					
Category	Comparisons	FANCM var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		22	56%	1193	64%			
2		15	38%	576	31%			
3		1	3%	68	4%			
4	2, 3 and 4 vs. 1	1	3%	22	1%	1.38	0.73–2.63	0.399
Nodus								
Neg		32	73%	1286	64%			
Pos	posit vs. neg	12	27%	721	36%	0.67	0.34–1.31	0.268
Metastasis								
Neg		43	98%	1973	98%			
Pos	posit vs. neg	1	2%	45	2%	1.02	0.14–7.57	1.000
ER								
Neg		8	19%	267	14%			
Pos	neg vs. posit	34	81%	1649	86%	1.45	0.67–3.17	0.366
PR								
Neg		12	29%	428	23%			
Pos	neg vs. posit	29	71%	1471	77%	1.42	0.72–2.81	0.345
HER2								
Neg		36	88%	1568	85%			
Pos	posit vs. neg	5	12%	277	15%	0.79	0.31–2.02	0.670
Grade								
1		12	29%	362	19%			
2		14	33%	908	47%			
3	3 vs. 1 and 2	16	38%	669	35%	1.17	0.62–2.19	0.743
Tumor histology								
Ductal		39	85%	1565	78%			
Lobular		4	9%	310	15%			
Other	Duct vs. others	3	7%	138	7%	1.60	0.71–3.59	0.287
Type								
LumA		30	73%	1400	76%			
LumB		3	7%	196	11%			
HER2 pos		2	5%	76	4%			
Triple-neg	TN vs. others	6	15%	159	9%	1.80	0.75–4.35	0.168
Ki67								
Neg		1	2%	144	8%			
1+		18	43%	752	41%			
2+	high vs. low	7	17%	496	27%			
3+	3 vs. others	16	38%	452	25%	1.90	1.01–3.57	0.048
Multifocal								
Yes		5	11%	323	16%			
No	yes vs. no	40	89%	1699	84%	0.66	0.26–1.68	0.422
Bilateral								
Yes		1	2%	49	2%			
No	yes vs. no	45	98%	1975	98%	0.90	0.12–6.63	1.000
Mean age at diag.		59 y		58 y				0.592

*one compound heterozygote

H. *FANCM* c.5101C>T (p.Gln1701Ter), rs147021911, 14-45189123-C-T

		N=34		N=2036				
Category	Comparisons	FANCM var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		19	63%	1196	64%			
2		9	30%	583	31%			
3		1	3%	66	4%			
4	2, 3 and 4 vs. 1	1	3%	22	1%	1.03	0.49–2.18	1.000
Nodus								
Neg		26	79%	1292	64%			
Pos	posit vs. neg	7	21%	727	36%	0.48	0.21–1.11	0.099
Metastasis								
Neg		33	100%	1984	98%			
Pos	posit vs. neg	0	0%	46	2%	NA	NA	1.000
ER								
Neg		5	16%	270	14%			
Pos	neg vs. posit	26	84%	1657	86%	1.18	0.45–3.10	0.793
PR								
Neg		7	23%	432	23%			
Pos	neg vs. posit	23	77%	1478	77%	1.04	0.44–2.44	1.000
HER2								
Neg		29	94%	1576	85%			
Pos	posit vs. neg	2	7%	279	15%	0.39	0.09–1.64	0.304
Grade								
1		9	28%	365	19%			
2		11	34%	911	47%			
3	3 vs. 1 and 2	12	38%	674	35%	1.14	0.55–2.34	0.712
Tumor histology								
Ductal		27	79%	1577	78%			
Lobular		4	12%	310	15%			
Other	Duct vs. others	3	9%	138	7%	1.10	0.47–2.53	0.842
Type								
LumA		24	77%	1407	76%			
LumB		2	7%	196	11%			
HER2 pos		0	0%	78	4%			
Triple-neg	TN vs. others	5	16%	160	9%	2.02	0.77–5.33	0.187
Ki67								
Neg		1	3%	144	8%			
1+		12	39%	758	41%			
2+	high vs. low	5	16%	498	27%			
3+	3 vs. others	13	42%	455	25%	2.22	1.08–4.57	0.035
Multifocal								
Yes		4	12%	324	16%			
No	yes vs. no	30	88%	1709	84%	0.7	0.25–2.01	0.640
Bilateral								
Yes		1	3%	49	2%			
No	yes vs. no	33	97%	1987	98%	1.23	0.17–9.17	0.567
Mean age at diag.		59 y		58 y				0.734

I. *FANCM* c.5791C>T (p.Arg1931Ter), rs144567652, 14-45198718-C-T

		N=13		N=2058				
Category	Comparisons	FANCM var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		4	40%	1211	64%			
2		6	60%	586	31%			
3		0	0%	69	4%			
4	2, 3 and 4 vs. 1	0	0%	23	1%	2.68	0.75–9.53	0.183
Nodus								
Neg		7	58%	1311	64%			
Pos	posit vs. neg	5	42%	729	36%	1.29	0.41–4.06	0.765
Metastasis								
Neg		11	92%	2006	98%			
Pos	posit vs. neg	1	8%	45	2%	4.05	0.51–32.06	0.238
ER								
Neg		4	33%	272	14%			
Pos	neg vs. posit	8	67%	1675	86%	3.08	0.92–10.30	0.076
PR								
Neg		6	50%	435	23%			
Pos	neg vs. posit	6	50%	1494	77%	3.43	1.10–10.70	0.035
HER2								
Neg		8	73%	1597	85%			
Pos	posit vs. neg	3	27%	279	15%	2.15	0.57–8.14	0.219
Grade								
1		3	27%	371	19%			
2		3	27%	920	47%			
3	3 vs. 1 and 2	5	46%	680	35%	1.58	0.48–5.20	0.528
Tumor histology								
Ductal		13	100%	1593	78%			
Lobular		0	0%	314	15%			
Other	Duct vs. others	0	0%	141	7%	NA	NA	0.085
Type								
LumA		6	55%	1424	76%			
LumB		1	9%	198	11%			
HER2 pos		2	18%	76	4%			
Triple-neg	TN vs. others	2	18%	164	9%	2.30	0.49–10.74	0.254
Ki67								
Neg		0	0%	145	8%			
1+		6	50%	764	41%			
2+	high vs. low	2	17%	502	27%			
3+	3 vs. others	4	33%	464	25%	1.52	0.46–5.07	0.506
Multifocal								
Yes		1	8%	328	16%			
No	yes vs. no	11	92%	1727	84%	0.48	0.06–3.72	0.704
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	13	100%	2008	98%	NA	NA	1.000
Mean age at diag.		58 y		58 y		0.986		

		N=34		N=2037				
Category	Comparisons	MCPH1 var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		16	57%	1199	64%			
2		10	36%	583	31%			
3		1	4%	68	4%			
4	2, 3 and 4 vs. 1	1	4%	22	1%	1.34	0.63–2.84	0.552
Nodus								
Neg		20	59%	1298	64%			
Pos	posit vs. neg	14	41%	721	36%	1.26	0.63–2.51	0.589
Metastasis								
Neg		33	97%	1985	98%			
Pos	posit vs. neg	1	3%	45	2%	1.34	0.18–9.99	0.538
ER								
Neg		6	19%	270	14%			
Pos	neg vs. posit	26	81%	1657	86%	1.42	0.58–3.47	0.440
PR								
Neg		12	38%	428	22%			
Pos	neg vs. posit	20	63%	1481	78%	2.08	1.01–4.28	0.054
HER2								
Neg		26	87%	1580	85%			
Pos	posit vs. neg	4	13%	277	15%	0.88	0.30–2.53	1.000
Grade								
1		2	7%	372	19%			
2		16	55%	907	46%			
3	3 vs. 1 and 2	11	38%	675	35%	1.16	0.54–2.47	0.845
Tumor histology								
Ductal		23	68%	1582	78%			
Lobular		9	27%	305	15%			
Other	Lob vs. others	2	6%	139	7%	2.03	0.94–4.94	0.087
Type								
LumA		23	77%	1408	76%			
LumB		2	7%	196	11%			
HER2 pos		2	7%	76	4%			
Triple-neg	LumA vs. others	3	10%	163	9%	1.02	0.43–2.38	1.000
Ki67								
Neg		0	0%	145	8%			
1+		9	30%	761	41%			
2+	high vs. low	14	47%	490	26%			
3+	3 vs. others	7	23%	461	25%	0.92	0.39–2.16	1.000
Multifocal								
Yes		13	38%	316	16%			
No	yes vs. no	21	62%	1718	85%	3.37	1.67–6.79	0.001
Bilateral								
Yes		1	3%	49	2%			
No	yes vs. no	33	97%	1988	98%	1.23	0.17–9.17	0.567
Mean age at diag.		60 y		58 y				0.405

K. *RAD50* c.687delT (p.Ser229fs), rs760146707, 5-132579996-GT-G

		N=28	N=2042					
Category	Comparisons	<i>RAD50</i> var	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		15	60%	1199	64%			
2		5	20%	588	31%			
3		3	12%	66	4%			
4	2, 3 and 4 vs. 1	2	8%	21	1%	1.18	0.53–2.65	0.835
Nodus								
Neg		15	60%	1302	64%			
Pos	posit vs. neg	10	40%	725	36%	1.2	0.54–2.68	0.678
Metastasis								
Neg		25	96%	1992	98%			
Pos	posit vs. neg	1	4%	45	2%	1.77	0.24–13.36	0.446
ER								
Neg		3	12%	273	14%			
Pos	neg vs. posit	23	88%	1660	86%	0.79	0.24–2.66	1.000
PR								
Neg		5	19%	435	23%			
Pos	neg vs. posit	21	81%	1480	77%	0.81	0.30–2.16	0.816
HER2								
Neg		22	85%	1584	85%			
Pos	posit vs. neg	4	15%	277	15%	1.04	0.36–3.04	1.000
Grade								
1		6	23%	368	19%			
2		13	50%	909	46%			
3	3 vs. 1 and 2	7	27%	679	35%	0.69	0.29–1.66	0.535
Tumor histology								
Ductal		21	78%	1583	78%			
Lobular		5	19%	309	15%			
Other	Duct vs. others	1	4%	140	7%	1.00	0.40–2.47	1.000
Type								
LumA		21	81%	1410	76%			
LumB		2	8%	196	11%			
HER2 pos		2	8%	76	4%			
Triple-neg	LumA vs. others	1	4%	165	9%	1.30	0.49–3.47	0.654
Ki67								
Neg		3	12%	142	8%			
1+		14	54%	756	41%			
2+	high vs. low	6	23%	498	27%			
3+	3 vs. others	3	12%	465	25%	0.39	0.12–1.31	0.167
Multifocal								
Yes		3	11%	326	16%			
No	yes vs. no	24	89%	1714	84%	0.66	0.20–2.20	0.790
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	28	100%	1992	98%	NA	NA	1.000
Mean age at diag.		63 y		58 y		0.090		

L. Double carriers

		N=11		N=2061				
Category	Comparisons	Double carriers	% of valid	Wild type	% of valid	OR	95% CI	p
Tumor								
1		5	50%	1210	64%			
2		2	20%	591	31%			
3		1	20%	68	4%			
4	2, 3 and 4 vs. 1	2	10%	21	1%	1.78	0.51–6.17	0.510
Nodus								
Neg		7	64%	1311	64%			
Pos	posit vs. neg	4	36%	731	36%	1.03	0.30–3.51	1.000
Metastasis								
Neg		11	100%	2007	98%			
Pos	posit vs. neg	0	0%	46	2%	NA	NA	1.000
ER								
Neg		3	27%	273	14%			
Pos	neg vs. posit	8	73%	1676	86%	2.30	0.61–8.73	0.193
PR								
Neg		4	36%	437	23%			
Pos	neg vs. posit	7	64%	1494	77%	1.95	0.57–6.70	0.284
HER2								
Neg		9	82%	1597	85%			
Pos	posit vs. neg	2	18%	280	15%	1.27	0.27–5.90	0.674
Grade								
1		1	10%	373	19%			
2		5	45%	918	47%			
3	3 vs. 1 and 2	5	45%	681	35%	1.58	0.48–5.20	0.528
Tumor histology								
Ductal		10	91%	1596	78%			
Lobular		1	9%	313	15%			
Other	Duct vs. others	0	0%	141	7%	2.85	0.36–22.28	0.473
Type								
LumA		7	64%	1424	76%			
LumB		1	9%	198	11%			
HER2 pos		1	9%	77	4%			
Triple-neg	LumA vs. others	2	18%	164	9%	0.54	0.16–1.85	0.301
Ki67								
Neg		0	0%	145	8%			
1+		3	27%	767	41%			
2+	high vs. low	3	27%	501	27%			
3+	3 vs. others	5	45%	464	25%	2.54	0.77–8.35	0.154
Multifocal								
Yes		2	18%	327	16%			
No	yes vs. no	9	82%	1730	84%	1.18	0.25–5.47	
Bilateral								
Yes		0	0%	50	2%			
No	yes vs. no	11	100%	2011	98%	NA	NA	1.000
Mean age at diag.		57 y		58 y		0.536		

*BRCA1 c.3607C>T (p.Arg1203Ter) rs62625308 17-43091924-G-A, BRCA1 c.3626del (p.Leu1209Ter) rs80357571 17-43091904-TA-T, BRCA1 c.4097-2A>G rs80358019 17-43091034-T-C, BRCA1 c.5095C>T (p.Arg1699Trp) rs55770810 17-43063931-G-A, BRCA2 c.771_775del (p.Asn257fs) rs80359671 13-32331003-ACAAAT-A, BRCA2 c.3860dupA (p.Asn1287Lysfs) rs80359406 13-32338208-G-GA, BRCA2 c.6275_6276del (p.Leu2092fs) rs11571658 13-32340629-CTT-C, BRCA2 c.7480C>T (p.Arg2494Ter) rs80358972 13-32356472-C-T, BRCA2 c.9118-2A>G (p.Val3040Metfs*20) rs81002862 13-32380005-A-G. CI: confidence interval, Diag: diagnosis, ER: estrogen receptor, HER2: human epidermal growth factor receptor 2, Lum: luminal, Neg: negative, OR: odds ratio, Pos: positive, PR: progesterone receptor, var: variant carrier, y: years. LuminalA was defined as positive ER expression and no HER2 overexpression, luminalB had positive ER and HER2 overexpression, HER2 type as negative ER and PR but with HER2 overexpression, and triple-negative as negative for all three markers. Genomic locations are reported in GRCh38.

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