

**S2 Table.** *BRCA1/2* mutations in PIKO study (n=46)

Case	Age (yr)	Stage/ Histology	Site	Mutation <sup>a)</sup>	Type	FHBOC <sup>b)</sup>	FH of other cancer	Citation No. in BIC (N/R)
<b><i>BRCA1</i> (n=35)</b>								
PIKO-161	69	2/Serous	Exon 2	c.1A>G	MS	Breast (patient, mother)	-	8
PIKO-225	53	2/Serous	Exon 7	c.390C>A	NS	-	-	1 (R)
PIKO-031	50	4/Serous	Exon 7	c.390C>A	NS	Breast (patient)	Endometrium (aunt)	1 (R)
PIKO-082	46	3/Serous	Exon 11	c.922_923AGCi nsT	NS	-	-	0 (-)
PIKO-003	60	1/Serous	Exon 11	c.981_982delAT	NS	Ovary (sister)	-	13 (-)
PIKO-016	64	3/Serous	Exon 11	c.1179_1180dup AG	FS	-	-	0 (N, R)
PIKO-233	62	3/Clear cell	Exon 11	c.1179_1180dup AG	FS	-	-	0 (N, R)
PIKO-267	49	3/Serous	Exon 11	c.1716delA	FS	Breast (patient, mother)	-	0 (-)
PIKO-198	45	3/Serous	Exon 11	c.1831delC	NS	-	-	0 (R)
PIKO-159	49	3/Clear cell	Exon 11	c.1831delC	NS	-	Endometrium (aunt), Stomach (sister)	0 (R)
PIKO-147	60	2/Serous	Exon 11	c.1953delG	FS	Ovary (mother)	Cervix (patient)	0 (N)
PIKO-254	49	3/Serous	Exon 11	c.2433delC	FS	Breast (aunt)	-	11 (-)
PIKO-232	63	3/Serous	Exon 11	c.3442delG	FS	Ovary (sister)	-	2 (R)
PIKO-079	52	3/Serous	Exon 11	c.3442delG	FS	Breast (mother) Ovary (sister)	Pancreas (father)	2 (R)
PIKO-067	55	3/Serous	Exon 11	c.3627insA	FS	Ovary (mother, grandmother)	Colon (mother), Thyroid (sister)	9 (R)
PIKO-101	38	3/Serous	Exon 11	c.3627insA	FS	Ovary (mother)	Lung (father)	9 (R)
PIKO-148	46	3/Serous	Exon 11	c.3627insA	FS	-	-	9 (R)
PIKO-006	52	3/Serous	Exon 11	c.3627insA	FS	-	-	9 (R)
PIKO-061	44	3/Serous	Exon 11	c.3627insA	FS	Ovary (mother)	-	9 (R)

PIKO-181	56	3/Serous	Exon 11	c.3991C>T	NS	-	-	0 (-)
PIKO-186	53	3/Serous	Exon 16	c.4981G>T	NS	Ovary (sister)	-	1 (-)
PIKO-258	45	3/Serous	Exon 17	c.5030_5033del CTAA	FS	Breast (sister), ovary (sister)	Leukemia (sister)	17 (-)
PIKO-023	65	3/Serous	Exon 18	c.5080G>T	NS	Ovary (mother)	Melanoma (sister)	12 (-)
PIKO-236	48	3/Serous	Exon 22	c.5339T>C <sup>e)</sup>	MS	-	Thyroid (patient)	1 (R)
PIKO-41	52	3/Serous	Exon 22	c.5339T>C	MS	-	-	1 (R)
PIKO-267	48	3/Serous	Exon 23	c.5444G>A	NS	Ovary (mother)	Liver (mother)	4 (-)
PIKO-022	33	3/Serous	Exon 23	c.5445G>A	NS	Breast (sister, cousin)	Thyroid (sister)	0 (R)
PIKO-078	45	4/Serous	Exon 23	c.5445G>A	NS	-	-	0 (R)
PIKO-052	53	3/Serous	Exon 23	c.5445G>A	NS	-	Stomach (father)	0 (R)
PIKO-010	56	4/Serous	Exon 24	c.5470_5477del ATTGGGCA	FS	Breast (patient)	Colon (grandfather)	2 (-)
PIKO-071	46	4/Serous	Exon 24	c.5496_5506del GGTGACCCG AGinsA	FS	Breast (patient, sister)	-	3 (R)
PIKO-080	47	2/Serous	Exon 24	c.5496_5506del GGTGACCCG AGinsA	FS	-	Thyroid (patient) Stomach (brother)	3 (R)
PIKO-110	77	3/Clear cell	Exon 24	c.5496_5506del GGTGACCCG AGinsA	FS	-	-	3 (R)
PIKO-188	52	3/Serous	Exon 24	c.5496_5506del GGTGACCCG AGinsA	FS	Ovary (mother)	Endometrium (sister)	3 (R)
PIKO-142	42	3/Serous	IVS 6	c.302-2A>C	SP	-	-	1 (-)
<b>BRCA2 (n=11)</b>								
PIKO-056	75	4/Serous	Exon 3	c.276insA	FS	-	-	1 (-)
PIKO-179	56	3/Serous	Exon 10	c.1111A>T	NS	-	-	0 (N)
PIKO-272	67	2/Endomet rioid	Exon 10	c.1399A>T	NS	Breast (sister)	-	2 (-)
PIKO-219	42	3/Serous	Exon 11	c.2259delT	FS	-	Stomach (mother)	0 (-)

PIKO-095	46	4/Serous	Exon 11	c.2798_2799del CA	FS	-	-	1 (-)
PIKO-063	54	4/Serous	Exon 11	c.4207dupA	FS	-	-	0 (N)
PIKO-058	54	4/Serous	Exon 11	c.5576_5579del TTAA	FS	Breast (sister)	-	29 (-)
PIKO-244	55	3/Serous	Exon 11	c.5656C>T	NS	-	Pancreas (brother)	2 (-)
PIKO-170	72	3/Serous	Exon 13	c.6952C>T	NS	-	-	5 (-)
PIKO-131	67	3/Serous	Exon 20	c.8572delC	FS	-	Brain (sister)	0 (N)
PIKO-141	63	3/Serous	Exon 25	c.9431delC	FS	-	-	0 (-)

PIKO, Pusan National University, Inje University, and Kosin University; FHBOC, family history of breast/ovarian cancer; FH, family history; BIC, Breast Cancer Information Core; N, novel; R, reported; MS, missense; NS, nonsense; FS, frameshift; SP, splicing. <sup>a)</sup>Nomenclature system of the Human Genome Variation Society (<http://www.hgvs.org/mutnomen>), <sup>b)</sup>Patients with primary breast cancer or first- or second-degree relatives with breast or ovarian cancer, <sup>c)</sup>Recently implicated as a deleterious mutation rather than a variant of unknown significance.