

**S3 Table.** Characteristics of patients with pathogenic or likely pathogenic variants in unknown-risk genes beyond *BRCA* (n=24)

Case	Sex	Age at first diagnosis of breast cancer	Site	Path	Subtype	Family history Kind (degree*n)	Second cancer	Affected genes	Nucleotide change	Amino acid change	effect	Mode of inheritance	Zygoty	dbSNP
P047	F	38	Lt	IDC	TNBC	Uterus (2*1), CRC (2*1), liver (2*1)	-	<i>ALK</i>	c.2467-1G>C	-	Splicing	AD	Hetero	-
P056	M	68	Lt	IDC	ER+/HER2-	None	-	<i>BLM</i>	c.320dupT	p.Leu107Phefs	FS	AD/AR	Hetero	rs781221411
P066	F	39	Rt	IDC	ER-/HER2+	Larynx (1*1)	-	<i>CDKN2A</i>	c.260G>C	p.Arg87Pro	MS	AD	Hetero	-
P045	F	63	Rt	IDC	TNBC	Ovary (1*1), bile duct- (1*1)	-	<i>EPCAM</i>	c.555+2T>C	-	Splicing	AD/AR	Hetero	-
P015	F	54	Lt	IDC	ER+/HER2-	Breast (1*1)	-	<i>SDHA</i> <i>EXO1</i>	c.2T>C p.Met1? c.2071A>T	- p.Lys691Ter	MS NS	AD/AR AD	Hetero	-
P043	F	40	Rt	IDC	ER+/HER2-	Lung (2*1)	Thyroid	<i>EXO1</i>	c.2071A>T	p.Lys691Ter	NS	AD	Hetero	-
P008	F	53	Lt	DCIS	ER+/HER2-	Breast (2*1), leukemia (1*1)	-	<i>FANCM</i>	c.2112delA	p.Leu705Cysfs	FS	AR	Hetero	-
P007	F	58	Lt	IDC	TNBC	Ovary (1*1)	-	<i>MRE11A</i>	c.659+1G>A	-	Splicing	AD/AR	Hetero	rs759130031
P068	F	38	Rt	DCIS	Unknown	Hematology (3*1)	-	<i>MRE11A</i>	c.659+1G>A	-	Splicing	AD/AR	Hetero	rs759130031
P001	F	61	Lt	Medullary	ER+/HER2-	CRC (1*2), cervix (1*1)	Thyroid, ovary, endometrium, rectum	<i>MSH2</i>	c.256G>T	p.Glu86Ter	NS	AD	Hetero	-
P018	F	37	Lt	IDC	ER-/HER2+	None	Cervix	<i>JAK2</i> <i>NTRK1</i>	c.2189T>G c.1696C>T	p.Leu730Ter p.Arg566Ter	NS NS	AD AR	Hetero	- rs763758904
P058	F	35	Lt	IDC	ER+/HER2-	None	-	<i>PMS2</i>	c.164-1G>C	-	Splicing	AD	Hetero	rs763308607
P061	F	35	Rt	IDC	ER+/HER2+	Breast (2*1), CRC (1*1), thyroid (1*1)	-	<i>PMS2</i>	c.164-1G>C	-	Splicing	AD	Hetero	rs763308607

P057	F	38	Rt	IDC	TNBC	None	-	<i>PMS2</i>	c.2404C>T	p.Arg802Ter	NS	AD	Hetero	rs63751466
								<i>RAD50</i>	Exon 4-16 duplication	-	Exon duplication	AD/AR	Hetero	-
P003	M	82	Lt	IDC	ER+/HER2-	CRC (1*1), lung (3*1)	Lung	<i>PPM1D</i>	c.1423G>T	p.Glu475Ter	NS	AD	Hetero	-
P044	F	45	Rt	IDC	ER+/HER2-	Breast (1*1), Thyroid (1*1,2*1), stomach (1*1)	-	<i>RAD50</i>	c.1722dup	p.Gln575Thrfs	FS	AD/AR	Hetero	rs587782543
P004 <sup>a)</sup>	F	36	Bil(met)	IDC	ER+/HER2-	Panc (2*1), LeukemiaCervix (2*1)	-	<i>RAD50</i>	c.2165dup	p.Glu723Glyfs	FS	AD/AR	Hetero	rs397507178
P019 <sup>a)</sup>	F	39	Lt	IDC	ER+/HER2-	Breast (1*3)	Thyroid	<i>RAD50</i>	c.2165dup	p.Glu723Glyfs	FS	AD/AR	Hetero	rs397507178
P022 <sup>a)</sup>	F	35	Rt	IDC+ILC	ER+/HER2+	Breast (2*3)	-	<i>RAD50</i>	c.2165dup	p.Glu723Glyfs	FS	AD/AR	Hetero	rs397507178
P013	F	64	Lt	IDC	TNBC	Breast (1*1,2*1)	-	<i>RAD50</i>	c.2178delT	p.Asp727Metfs	FS	AD/AR	Hetero	-
P026	F	48	Bil(met)	IDC	ER+/HER2-	Stomach (1*1,2*1)	-	<i>RAD50</i>	c.3883C>T	p.Gln1295Ter	NS	AD/AR	Hetero	-
P012	F	51	Rt	IDC	ER+/HER2+	Breast (1*1)	Thyroid	<i>RAD50</i>	Exon 4-16 duplication	-	Exon duplication	AD/AR	Hetero	-
P002	F	35	Rt	Mucinous	Unknown	None	Thyroid, colon	<i>SDHB</i>	c.757del	p.Cys253Valfs	FS	AD/AR	Hetero	-
P074	F	36	Rt	DCIS	Unknown	None	Paraganglioma	<i>VHL</i>	c.242C>T	p.Pro81Leu	MS	AD/AR	Hetero	rs193922608

AD, autosomal dominant; AR, autosomal recessive; Bil(met), bilateral breast cancer, metachronous; CRC, colorectal cancer; dbSNP, Single Nucleotide Polymorphism Database; DCIS, ductal carcinoma in situ; ER, estrogen receptor; F, female; FS, frameshift; HER2, human epidermal growth factor receptor 2; IDC, invasive ductal carcinoma; Lt, left; M, male; MS, missense; NS, nonsense; Panc, pancreas; Rt, right; TNBC, triple negative breast cancer. <sup>a)</sup>Three patients were the cousins.