

Figure S1. Microsatellite analysis in OC tissues.

A: In GECO 23, where MLPA had detected BRCA2 duplication, the analysis of D13S153 marker is consistent with the duplication, but also shows LOH, which extends to the entire chromosome, according to the status of the other markers analysed (not shown).

B: In GECO 30, also found to harbor BRCA2 duplication, microsatellite marker D13S153 analysis rules out LOH.

C: In GECO 34, the analysis of microsatellite marker D17S938 demonstrates LOH at BRCA1 region, despite no CNV detected by MLPA (“Copy-neutral” LOH: CN-LOH).

PB = Peripheral blood; TT = SF tumor tissue; LOH= Loss Of Heterozygosity

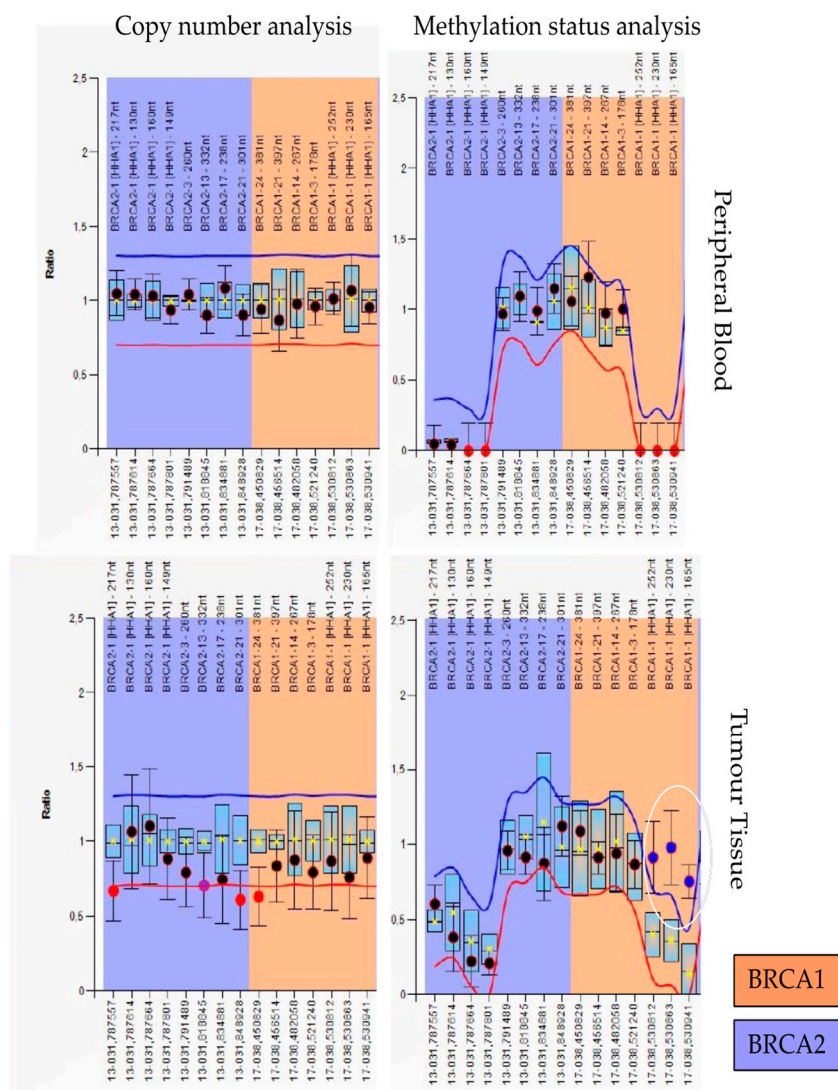


Figure S2. Promoter methylation plots in GECO 34.

Table S1. Comprehensive results of the molecular analyses performed on OC tissues in this study.

Patient	BRCA status	TP53 variant (classification ^a)	TP53 variant load (%)	Other gene variants (classification ^a)	Other gene variant load (%)	Number of CNVs detected by SNP-CGH	Germline variants ^b (gene)
GECO 1	BU	c.584T>C;p.Ile195Thr (P)	72.1	ATM: c.1272T>C;p.Pro424= (CI)	42.7	n.a.	/
GECO 2	BU	N	/	ATM: c.2978A>G;p.His993Arg (US)	47.3	n.a.	/
GECO 3	BD	n.a.	/	n.a.	/	n.a.	/
GECO 5	BD	n.a.	/	n.a.	/	n.a.	/
GECO 6	BU	c.395A>G;p.Lys132Arg (US)	59.7	N	/	99	/
GECO 7	BD	n.a.	/	n.a.	/	205	/
GECO 8	BD	n.a.	/	n.a.	/	78	BRCA1: c.5123C>A; p.Ala1708Glu
GECO 9	BU	N	/	ATM: c.4060C>A;p.Pro1354Thr (CI)	58.9	48	/
GECO 12	BU	c.990_991insT;p.Gln331SerfsTer6 (U)	76.2	N	/	n.a.	/
GECO 13	BD	n.a.	/	n.a.	/	89	BRCA2: c.7558C>T; p.Arg2520Ter
GECO 14	BU	c.818G>A;p.Arg273His (P/LP)	63.8	RAD51C: c.904+5G>T;p.? (LP)	79.4	n.a.	RAD51C: c.904+5G>T;p.?
GECO 15	BU	c.380C>T;p.Ser127Phe (CI)	54.5	PALB2: c.1001A>G;p.Tyr334Cys (US)	60.9	n.a.	/
GECO 16	BU	c.730G>A;p.Gly244Ser (P/LP)	59.2	ATM: c.2932T>C;p.Ser978Pro (CI)	89.9	49	/

GECO 17	BU	c.649delG;p.Val217TrpfsTer30 (P)	80.0	ATM: c.7475T>G;p.Leu2492Arg (US)	76.7	n.a.	/
GECO 18	BU	N	/	N	/	15	/
GECO 20	BD	n.a.	/	n.a.	/	n.a.	/
GECO 21	BU	c.469delG;p.Val157SerfsTer13 (U)	66.1	N	/	n.a.	/
GECO 22	BU	c.661G>T;p.Glu221Ter (U)	75.6	PTEN: c.388C>T;p.Arg130Ter (P)	56.5	n.a.	/
GECO 23	BU	c.818G>A;p.Arg273His (P/LP)	93.9	No	/	n.a.	/
GECO 24	BD	n.a.	/	n.a.	/	n.a.	BRCA2: c.1813delA; p.Gly602=fs*11
GECO 25	BU	N	/	N	/	107	/
GECO 26	BD	n.a.	/	n.a.	/	196	/
GECO 27	BD	n.a.	/	n.a.	/	n.a.	BRCA2: c.9118-1G>A;p.?
GECO 28	BU	N	/	N	/	n.a.	/
GECO 29	BD	n.a.	/	n.a.	/	72	BRCA1: c.4065_4068delTCAA; p.Asn1355Lysfs
GECO 30	BU	c.818G>T;p.Arg273Leu (P)	65.6	No	/	37	/
GECO 31	BD	n.a.	/	n.a.	/	90	BRCA1: c.547+2T>A;p.?
GECO 33	BU	c.742C>T;p.Arg248Trp (P)	50.0	N	/	n.a.	/
GECO 34	BD	n.a.	/	n.a.	/	n.a.	/
GECO 35	BU	c.824G>A;p.Cys275Tyr (P/LP)	85.6	APC: c.607C>G;p.Gln203Glu (CI)	91.0	60	/

^aClinVar (P = Pathogenic; US = Uncertain Significance; LP = Likely Pathogenic; CI = Conflicting Interpretations; U = Unreported)

^bPathogenic or Likely Pathogenic

N = No; n.a. = not applicable (not performed)