

Comprehensive Genomic Profiling (CGP) & HRD Status

Sample Report — Case A

CANCER TYPE High-Grade Serous Carcinoma	METHOD DNA Whole Exome Sequencing
QC STATUS PASS	CGP SPECIMEN FFPE tissue
HRD SPECIMEN Peripheral blood	CGP COVERAGE 89.03x average depth

Clinically Significant Gene Mutations

Genes tested: BRAF, BRCA1, BRCA2, BRIP1, CHEK1, CHEK2, PALB2, RAD51C, RAD51D, TP53 — all negative except:

GENE	MUTATION	VAF	SIGNIFICANCE
PIK3CA	c.G1798A (p.E600K)	15.5% (heterozygous)	Pathogenic

COSMIC ID COSV55891897 — observed in endometrium (3), cervix (1) and bladder (2) carcinoma samples.

Additional Likely / Non-Clinically Significant Findings (VUS)

GENE	MUTATION	VAF	SIGNIFICANCE
GNAS	c.G2531A (p.R844H)	24.8% (heterozygous)	Likely Pathogenic

COSMIC ID COSV55670349 — observed in ovarian carcinoma, adenoma and low malignant potential (borderline) tumor samples (9), plus thyroid, soft tissue, small intestine and pituitary cancers.

HRD Status

HRD FINAL STATUS

POSITIVE — GIS 95 / 100 (Threshold ≥ 42)

41

LOH

32

TAI

22

LST

Genomic Instability Score combines Loss of Heterozygosity (LOH) + Telomeric Allelic Imbalance (TAI) + Large-Scale state Transitions (LST). HRD-positive tumors combined with BRCA1/BRCA2 inactivation are susceptible to PARP inhibitors and platinum chemotherapy.

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