









Technology



Next Generation Sequencing & Multiplex Ligation Dependent Probe Amplification

Specimen



FFPE Block & Whole Blood

Test Code







4 weeks



Detection and classification of sequence variants (SNVs & Indels) in 28 HRR pathway genes & BRCA1/2 Deletion-Duplication analysis by MLPA





Comprehensive assessment of Loss of heterozygosity (LOH), Telomeric allelic imbalance (TAI) and Large-scale transition (LST) across the entire genome to evaluate genomic instability



Determines the eligibility for PARP Inhibitors



Provides Genomic Scar Score

HRR pathway genes covered in Panel:

АТМ	BARD1	BRCA1	BRCA2	BRIP1	CDK12	CHEK2
FANCD2	MRE11	NBN	PALB2	PPP2R2A	RAD51B	RAD54L
TP53	CHEK1	FANCL	RAD50	RAD51	RAD51C	RAD51D
RAD52	XRCC2	KRAS	PIK3CA	POLD1	POLE	PTEN





Homologous Recombination Deficiency

Your Test Results

Case Number:

Patient Name:

Age/Sex:

Patient Location:

Hospital Name:

Physician Name:

Date & Time of Accessioning:

Date & Time of Reporting:

Test Performed

Homologous Recombination Deficiency

Specimen Information

Received 7 paraffin blocks

Clinical Indication

K/C/O ca ovary

Results

Genomic Instability (LOH+NtAI+ST)	Positive (HRD Score:61)		
HRR Pathway gene Mutations: (Germline+Somatic Study)	Positive		
BRCA1 & BRCA2 Deletion Duplication study by MLPA (Somatic+germine Study)	Negative		

Interpretation:

- GenomeSignature panel analysis of the provided FFPE tissue block (A0091648A-G) revealed HRD score
 of homologous re-combination deficient signature. HRD score 61 was calculated based on three
 biomarkers (loss of heterozygosity, telomericallelic imbalance and large-scale state transitions).
- Somatic HRR gene panel analysis revealed pathogenic variant in TP53 gene. (Details attached).

*Homologous Recombination Deficiency is calculated by proprietary algorithm developed at CORE Diagnostics based on threebiomarkers (loss of heterozygosity, telomeric allelic imbalance and large-scale state transitions)

Genomic Scar Score: > 50

HRD Status: Positive