

NextGen geneCORE Somatic

Homologous Recombination Deficiency

Patient Selection



Patients with
Ovarian Cancer/
Fallopian Tube
Cancer/Primary
Peritoneal Cancer

Technology



Next Generation
Sequencing

Specimen



Formalin Fixed
Paraffin Embedded
(FFPE) Tissue Block

Test Code



N8274

TAT



28 days



Provides HRD status which facilitates successful selection and identification of patients most likely to benefit from PARP inhibitors



Calculates HRD score by analyzing genome-wide SNPs and comprehensive assessment of loss of heterozygosity (LOH), telomeric allelic imbalance (TAI) and large-scale state transitions (LST) across the entire genome



Qualitative detection and classification of single nucleotide variants, insertions and deletions, and copy number variants in protein coding regions and intron/exon boundaries of the 29 HRR pathway genes including BRCA1 and BRCA2



Sensitivity: >99% for single nucleotide variants (5% VAF) and indels (10% VAF)
Specificity: > 99%

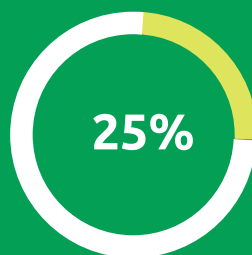
HRR pathway genes:

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

Understanding the Molecular Components of HRD



1 in 2 patients with ovarian cancer are HRD+



1 in 4 HRD+ patients have a BRCA1/2 mutation

Knowing HRD status is essential

HRD is defined as the inability of a cell to accurately repair DNA double-strand breaks



DNA double-strand breaks

When the HR pathway is disrupted by gene mutations, promoter methylation or undetermined causes, the HR pathway stops working leading to Homologous Recombination Deficiency (HRD).

Homologous Recombination Deficiency

HRR pathway genes

29 genes

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

BRCA1/2 Status
SNVs, Indels,
Structural rearrangements

+

Genome-wide SNPs

Heterozygous dbSNP variants

Loss of heterozygosity (LOH),
Telomeric allelic imbalance (TAI),
Large-scale state transitions (LST)



Are you aware of definition

"HRD Status Positivity"?

(Homologous recombination Deficiency Criteria)

01

Detection of
BRCA1 and BRCA2
mutations (+)

02

Positive Genomic
Instability Score (+)

03

Detection of
HRR related genes
mutations (+)

NextGen geneCORE Somatic Homologous Recombination Deficiency Panel Report

Case Number:
 Patient Name:
 Age/Sex:
 Patient Location:
 Hospital Name:
 Physician Name:
 Date & Time of Accessioning:
 Date & Time of Reporting:

Test Information

The NextGen geneCORE Somatic Homologous Recombination Deficiency (HRD) panel is a targeted next-generation sequencing (NGS) assay that provides an assessment of Homologous Recombination Deficiency (HRD) score and enables the detection of clinically relevant genomic alterations from DNA, within the 29 unique Homologous Recombination Repair (HRR) pathway genes, from formalin-fixed paraffin-embedded (FFPE) tumor block samples. This assay is designed to facilitate successful selection and identification of samples most likely to derive responses from PARP inhibitors.

Specimen Information

Received 01 paraffin block labelled as (block no).

Clinical History

k/c/o Carcinoma ovary.

Results



Hrd Status: Positive

Multi-Gene Biomarker Findings

Assay Biomarker	Result
HRD Score	51
Clinically relevant somatic variants in HRR* genes, including BRCA1 & BRCA2 genes	ATM, p.R2832H & TP53, p.R110Lfs*

*HRR: Homologous Recombination DNA Repair

