

# HRD Report

Patient	Analysis
<b>First name:</b> <b>Last name:</b> <b>Patient ID:</b> S2144-00107 <b>DOB:</b> Unknown <b>Sex:</b> Unknown <b>Pathology:</b> Serous ovarian carcinoma	<b>Analysis ID:</b> 200036613 <b>MID:</b> S2 <b>Run date:</b> 15/03/2022 <b>Run name:</b> Homologous Recombination Deficiency Solution <b>Sequencer:</b> Illumina NextSeq

The present analysis was performed according to patient consent(for details see Gene Panel Section)

**SOPHiA DDM™ HRD status POSITIVE**

Supporting evidence

**Proposed SOPHiA DDM™ HRD status** Positive

**Genomic Integrity status** **POSITIVE**

Supporting evidence

**Proposed Status** Positive **GI index** 10.3 **QA status** High

**BRCA status** **POSITIVE**

Supporting evidence

**Proposed Status** Positive

Gene	Coding DNA alteration Protein alteration	Variant Fraction	BRCA pathogenicity	BRCA score
<i>BRCA1</i>	c.3748G>T p.(Glu1250*)	85.3 %	Pathogenic	Positive

Validated by:

Date:

Signature:

Analyzed by:

Date:

Signature:

## OTHER INFORMATION AND RESULTS

### Variant description

#### Genomic alterations associated to approved therapies in patient's disease

None reported

#### Other potentially actionable genomic alterations

None reported

#### Other genomic alterations

None reported

#### Diagnosis - Prognosis

None reported

**Application:** HRD\_v1 **Reference genome:** GRCh37/hg19 **SOPHiA DDM™:** 5.10.19--b375-e3e3d24  
**JAX-CKB™ version:** v20211210

**Sample type:** FFPE **Pipeline ID / Revision number / Splitting ID:** ILL1XG1S7\_FFPE\_CNV\_NextSeq\_1 / v5.5.57 / GEN1GN1FSQ2

## ANNEXES

### Gene Panel (28)

*Version:v1*

AKT1 , ATM , BARD1 , BRCA1 , BRCA2 , BRIP1 , CCNE1 , CDK12 , CHEK1 , CHEK2 , ESR1 , FANCA , FANCD2 , FANCL , FGFR1 , FGFR2 , FGFR3 , MRE11 , NBN , PALB2 , PIK3CA , PPP2R2A , PTEN , RAD51B , RAD51C , RAD51D , RAD54L , TP53

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).