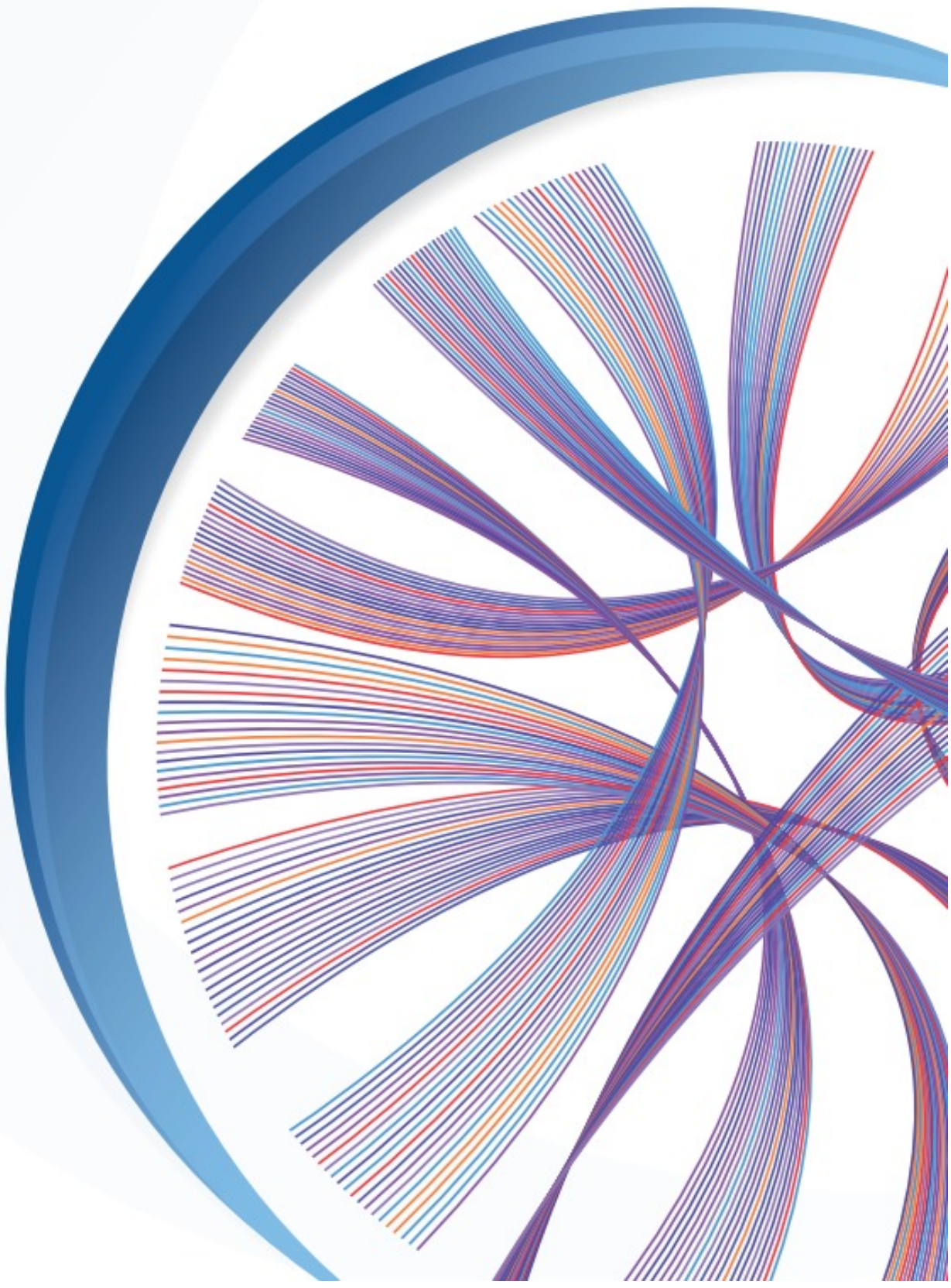
4baseCare
Together, We Beat Cancer

# TARGT First

## TEST REPORT



| PATIENT ID | CANCER SITE | SAMPLE ID | REGISTRATION DATE |
|---|---|---|---|
| IN-423-WBWA | UNKNOWN PRIMARY | IN-423-WBWA-F | 23/02/2023 |

## SCOPE OF THE TEST

SNVs, InDels, CNAs, Gene Fusions status

## CLINICAL INDICATION

Malignant effusion adenocarcinoma of unknown primary

## REPORT DETAILS

Name : KRISHNA

Gender : Female

Age/DOB : 56 Years

Reporting Date : 14/03/2023

Sample Source : B/3325/23

Tumor content : 30%

Consulting Clinician : Dr. Amit Jain

Hospital : Valentis Cancer Hospital, Mussoorie Mawana Road

## RESULTS

### GENOMIC FINDINGS FROM TUMOR PROFILING

| Genomic Alteration | Relevant Therapies (in Same Cancer Type) | Relevant Therapies (in Different Cancer) |
|---|---|---|

Clinically relevant genomic alterations associated with therapeutic significance were not detected.

**4baseCare Oncosolutions Pvt. Ltd**

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**Please note that this is not a medical report*

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## TEST DESCRIPTION

TAR**GT** First is a Next Generation Sequencing based test which identifies genetic alterations in a comprehensive panel of well curated 53 tumor genes which can impact response to approved therapy for a particular cancer type. Some of the alterations detected may have bearing on prognosis and/or therapeutic options and may provide relevant information that allows oncologists/clinicians to consider various lines of targeted treatment for the patient.

## GENES EVALUATED

TAR**GT** First detects mutations (SNVs and Short Indels), Copy Number Variations (CNVs), gene fusions and splice variants in the 53 genes :

### **SNVs/InDels** Covered in TAR**GT** First

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| *ABL1* | *ALK* | *AR* | *ATM* | *BARD1* | *BRAF* | *BRCA1* | *BRCA2* | *BRIP1* | *CDK12* |
| *CDK4* | *CDK6* | *CDKN2A* | *CHEK1* | *CHEK2* | *EGFR* | *EPCAM* | *ERBB2* | *ERBB3* | *EZH2* |
| *FANCL* | *FGFR3* | *GAPDH* | *IDH1* | *IDH2* | *JAK2* | *KIT* | *KRAS* | *MAP2K1* | *MAP2K2* |
| *MDM2* | *MET* | *MLH1* | *MLH3* | *MSH2* | *MSH6* | *NRAS* | *PALB2* | *PDGFRA* | *PDGFRB* |
| *PIK3CA* | *PMS1* | *PMS2* | *PPP2R2A* | *RAD51B* | *RAD51C* | *RAD51D* | *RAD54L* | *RET* | *ROS1* |
| *STK11* | *TSC1* | *TSC2* | | | | | | | |

### **CNAs** Covered in TAR**GT** First

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| *ABL1* | *AR* | *ATM* | *BARD1* | *BRCA1* | *BRCA2* | *BRIP1* | *CDK12* | *CDK4* | *CDK6* |
| *CDKN2A* | *CHEK1* | *CHEK2* | *EGFR* | *EPCAM* | *ERBB2* | *ERBB3* | *EZH2* | *FANCL* | *FGFR3* |
| *GAPDH* | *IDH2* | *JAK2* | *KIT* | *MDM2* | *NRAS* | *PALB2* | *PDGFRA* | *PDGFRB* | *PPP2R2A* |
| *RAD51B* | *RAD51C* | *RAD51D* | *RAD54L* | *STK11* | *TSC1* | *TSC2* | | | |

### **Gene Fusions** Covered in TAR**GT** First

| | | | |
|---|---|---|---|
| *ALK* | *MET* | *RET* | *ROS1* |

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## TEST METHODOLOGY

### Sample preparation and Library preparation :

DNA isolated from FFPE, or any other fresh tumor tissue source was used to perform targeted gene capture using a custom capture kit. The libraries were sequenced to mean >250X coverage on Illumina sequencing platform.

### Bioinformatics Analysis and Reporting :

The sequences obtained are aligned to human reference genome (GRCh37/hg19) and variant analysis was performed using set of Bioinformatics Pipeline. Only non-synonymous and splice site variants found in the panel consisting of specific set of genes were used for clinical interpretation. Silent variations that do not result in any change in amino acid in the coding region are not reported. Clinically relevant mutations were annotated using published variants in literature and a set of databases – ClinVar, COSMIC and dbSNP. Common variants are filtered based on allele frequency in 1000 Genome Phase 3, ExAC, dbSNP, gnomAD, etc. In the absence of a clinically significant reported known variation(s), pathogenicity will be predicted based on in-silico gene prioritization tools: CADD, SIFT, PolyPhen-2, Condel and Mutation taster and prioritized for clinical correlation. The identified pathogenic variant will be correlated with observed phenotypic features of the patient and interpreted according to American College of Medical Genetics (ACMG) guidelines.
Somatic variants are classified into two tiers based on their level of clinical significance in cancer diagnosis, prognosis, and/or therapeutics as per international guidelines: ACMG, ASCO, AMP, CAP, NCCN and ESMO

## LIMITATIONS AND DISCLAIMER

- DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of the many aspects used by the healthcare provider to help with a diagnosis and treatment plan.
- We are using the canonical transcript for clinical reporting which is usually the longest coding transcript with strong/multiple supporting evidence. However, in rare cases, clinically relevant variants annotated in alternate complete coding transcripts could also be reported.
- The contents of this test should be carefully assessed by the treating physician and further interpreted along with clinical, histopathological findings, contraindications and guidelines before deciding the course of therapy.
- The chromosomal aberrations like copy number variations and rearrangements may not be reliably detected with this assay and have to be confirmed by alternate method.
- The sensitivity of this assay to detect large deletions/duplications of more than 10 bp or copy number variations (CNV) is 70-75%. The CNVs detected have to be confirmed by alternate method.
- Most recent block is recommended for testing as the mutation profile may change in response to treatment and hence differ at different sampling points.
- TARGT FIRST test has been developed, validated and performed by 4baseCare Genomics Pvt. Ltd and has not been cleared or approved by the FDA.
- The identified pathogenic variant will be correlated with observed phenotypic features of the patient and interpreted according to (ASCO) guidelines.
- Certain genes may not be covered completely, and few mutations could be missed. A negative result cannot rule out the possibility that the tested tumor sample carries mutations not previously associated with cancer and hence not included in the panel.