LABORATORY REPORT

Reg. No	:	**30100200094**	Reg. Date	:	18-Jan-2023 09:47
Name	:	**KUNJLAATA SAHU**	Collected on	:	18-Jan-2023 15:21
Sex/Age	:	**Female / 53 Years**	Approved Date	:	09-Feb-2023 18:48
Ref. By	:		Tele. No	:	
Location	:	LILAC INSIGHTS PVT. LTD. @ MUMBAI	Dispatch At	:	

Please find detailed report of NGS Oncomine cfTNA Pan Cancer assay (DNA+RNA) in the following pages.

---------- End Of Report ----------

Neeraj Arora

Dr. Neeraj Arora
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22396

Patient Details

Patient Name	**KUNJLAATA SAHU**	**Sample Id/LabID**	**30100200094**
Gender	Female	**Sample Type**	Peripheral Blood in Streck tube:10ml
DOB/AGE	53 yrs	**Date of Sample Collection**	18/01/2023
Ref.By	LILAC INSIGHTS PVT. LTD.	**Date of Receipt**	18/01/2023
Sample QC Criteria	Pass(Yield->20ng)	**Date of Report**	09/02/2023

NGS Oncomine cfTNA Pan Cancer assay (DNA+RNA)

Clinical Details:

Known case Carcinoma Pancreas(On treatment)

RESULT

NEGATIVE:

No Clinically Relevant Pathogenic Mutations Identified.

No fusion Identified.

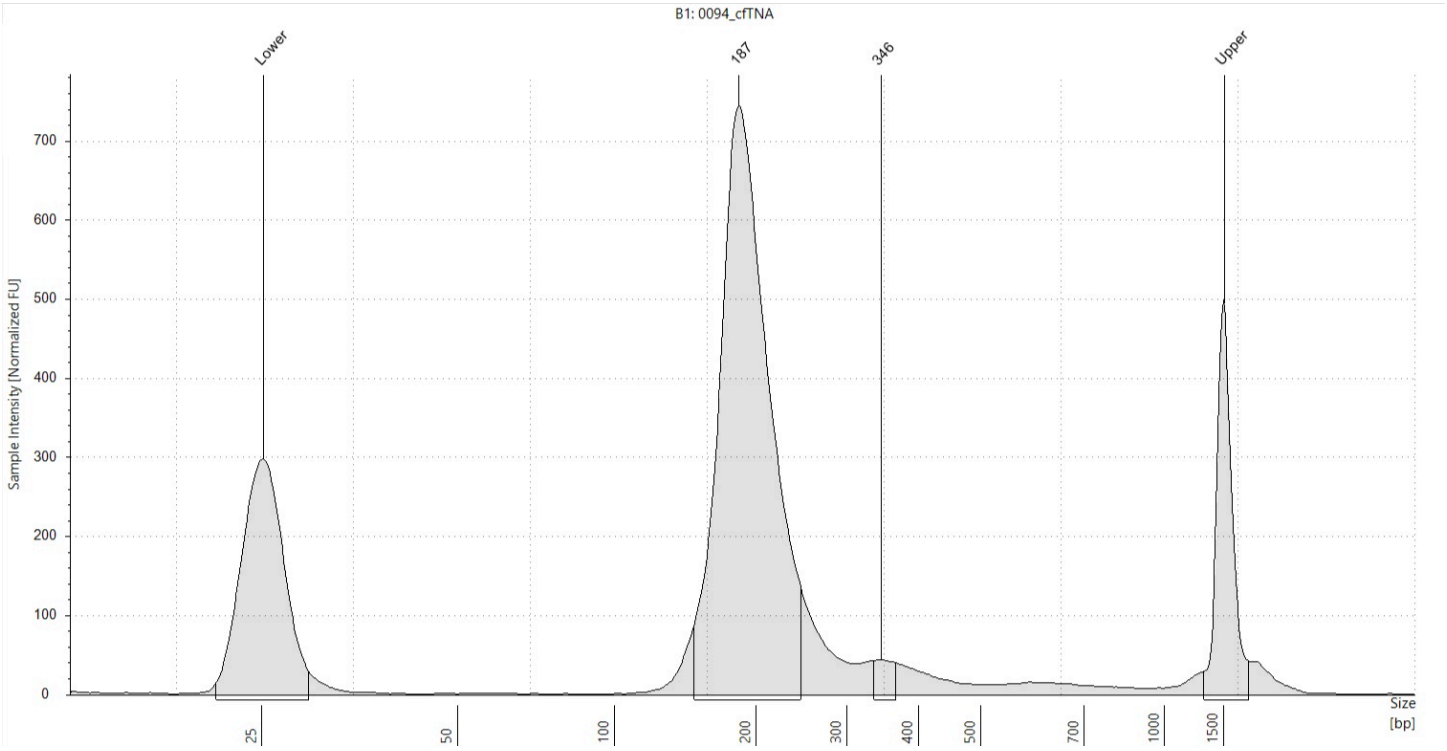
Comments:

These findings should be corelated with other clinical and laboratory tests for a definite conclusive interpretation.

Methodology

Total Nucleic acid (cfTNA) was extracted from provided peripheral blood in streck tube, using standard Total Nucleic Acid Isolation Kit. Briefly, 10ng of cfTNA was amplified using Oncomine Pan-Cancer Cell free Assay as per the instruction manual and sequencing was performed using the Ion S5 platform as per user manual. The sequencing reads QC, mapping on hg19 human reference genome, variant calling (SNVs, small InDels, CNVs, Fusions), and annotation was carried out with IonReporter™ (IR) Software 5.18.2.0. Latter uses different databases for the identification and characterization of genes-associated variants. The annotation for variants was derived using various diseases databases like OMIM and ClinVar. The population frequency information from 1000 genomes, ExAC, GnomAD, and ESP was used for the elimination of common variants/polymorphism. For the prediction of the possible impact of coding non-synonymous SNVs on the structure and function of a protein, PolyPhen-2 and SIFT score was used. Further Oncomine Reporter software was used for annotating variants with a curated list of relevant labels, guidelines, and global clinical trials. The Oncomine Pan-Cancer Cell-Free Assay is part of a complete solution to detect multiple targets in tumor-derived DNA and RNA isolated from the plasma fraction of whole blood.The 52-gene panel includes:Hotspot genes (SNVs) and short indels,Gene fusions,MET exon 14 skipping,Copy number genes (CNVs) and Tumor suppressor genes.The assay has a 0.1% limit of detection (LOD).

CfTNA Profile on Tapestation using DNA1000 screen



Run QC statistics
Sample is sequenced at Average base coverage depth of 6,356. The Target base coverage at 500X is 100.0%.

Variant Classification

- **Pathogenic**: The pathogenic variant are the one which is believed to account for the symptoms. It increases an individual's susceptibility or predisposition to a certain disease or disorder. This mutation is always included in results section of report.
- **Likely Pathogenic**: The likely pathogenic variant are the one which most likely have harmful effect but, there is insufficient evidence that a variant is the definite cause for symptoms. This mutation is always included in results section of report
- **Variant of Uncertain Significance**: The Variant of Uncertain Significance (VUS) are the one which have limited and/or conflicting evidence regarding pathogenicity. Its exact effect on gene function is not known. With more information available over time, a VUS may be reclassified as likely pathogenic or likely benign. This mutation is always included in results section of report.
- **Likely benign**: The likely benign variants are the one which are most likely not associated with disease risk. However, additional evidence is needed to confirm this assertion. This mutation is not included in report
- **Benign**: The benign variants are the one which are represented by alteration in gene compare to wild-type allele but it is not associated with disease risk. This mutation is not included in report.

Evidence Based Variant Classification

Tier I	Variants with strong clinical significance	Level A evidence	FDA-approved therapy included in professional guidelines
		Level B evidence	Well-powered studies with consensus from leaders in the field
Tier II	Variants with potential clinical significance	Level C evidence	FDA-approved therapies for different tumor types or investigational therapies. Multiple small published studies with some consensus
		Level D evidence	Preclinical trials or few case reports without consensus.
Tier III	Variants of unknown clinical significance		Not observed at significant allele frequency in the general or specific subpopulation databases, or pan-cancer or tumor-specific variant databases. No convincing published evidence of cancer association

Genes Assayed

Genes Assayed for the Detection of DNA Sequence Variants

AKT1, ALK, AR, ARAF, BRAF, CHEK2, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, NTRK1, NTRK3, PDGFRA, PIK3CA, RAF1, RET, ROS1, SF3B1, SMAD4, SMO, APC, FBXW7, PTEN

Genes Assayed for the Detection of Copy Number Variations

CCND1, CCND2, CCND3, CDK4, CDK6, EGFR, ERBB2, FGFR1, FGFR2, FGFR3, MET, MYC

Genes Assayed for the Detection of Fusions

ALK, BRAF, ERG, ETV1, FGFR1, FGFR2, FGFR3, MET, NTRK1, NTRK3, RET, ROS1

Genes Assayed With Full Exon Coverage

TP53

Limitations and Disclaimer

1. This test was developed and its performance characteristics determined by Unipath Specialty Laboratory Ltd, Ahmedabad. It has not been cleared or approved by the US Food and Drug Administration and NABL.
2. This test is a screening test and enables researchers to develop tests that may impact treatment selection, treatment monitoring, and recurrence monitoring in the future
3. False negative results may be due to sampling issues, errors in sample handling, mislabelling, transportation issues, and technical limitations of the assay and mutations frequency below the limit of detection of the assay, i.e., 0.1% for somatic variants. It is also possible some complex insertion/deletion variants may not be identified.
4. Extreme low circulating mutant cell free DNA leads to false negative result. Therefore in all negative cases, it is recommended to perform retesting on fresh tissue biopsy/tissue block.
5. This test is not intended to detect minimal residual disease.
6. Results of this test need be interpreted within the context of clinical findings and other relevant clinical and laboratory data and should not be used alone.

Report Signed by:

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(Sr. Scientist, NGS Division)

DR. EKTA JAJODIA, MD (Path.),
PDF (Mol. Hemat),Consultant Pathologist

DR. NEERAJ ARORA, MD ,
PDF (Mol.Hemat), PDF (Haematopath)
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