GENEVOLVE
Genomics Division

LPL – PRODUCTION TEST COLLECTION CENTRE
SECTOR - 18, BLOCK-E
ROHINI
DELHI 110085

Name	: DUMMY		
Lab No.	: DUMMYN149 Age: 25 Years	Gender: Male	
A/c Status	: P	Ref by : DR. DUMMY DUMMY	

Collected : 01/06/2021 05:06:00
Received : 01/06/2021 17:06:16
Reported : 04/06/2021 15:19:34
Report Status : Final

TEST CONDUCTED	**Cancer Any three marker: C-KIT, PDGFRA, TP53*** Next generation sequencing

***Third marker not mentioned**

INDICATION & SAMPLE INFORMATION	40 year old female diagnosed with Gastrointestinal stromal tumor (GIST) from ? Mesentric cyst excision with no known treatment history. The patient has a clinical history of ? Mesentric cyst. Sample type - FFPE (Block Number-432892/19; Tumour content 60%)

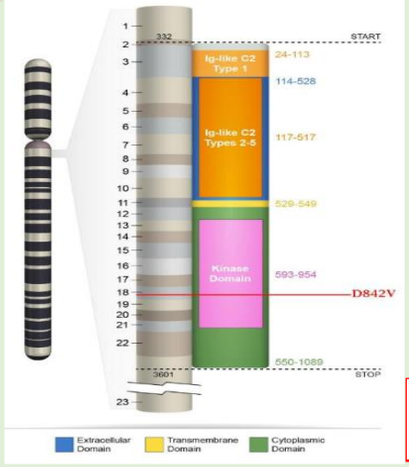
MUTATIONS DETECTED IN SAMPLE

PDGFRA	KIT	TP53*
Detected	**Not detected**	**Not detected**

INDIVIDUAL VARIANT INTERPRETATION FOR PATHOGENIC MUTATION

PDGFRA: NM_006206.5(PDGFRA):c.2525A>T (p.Asp842Val) - Pathogenic

The D842V mutation results in an amino acid substitution at position 842 in PDGFRA aspartic acid (D) to a Valine (V). This mutation occurs within the TK2 domain. 842V is located in the kinase domain (amino acids 593-954). It is the most common PDGFRA mutation associated with GIST (~5%) and has shown to increase growth factor-independent growth and basal activity (PMID: 22745105). PDGFRA D842V confers primary resistance to imatinib (PMID: 14645423), sorafenib (PMID: 18794084) and suntinib (PMID: 18955458). However preclinical studies provide evidence that tumors with D842V may respond to second generation PDGFR TKIs, such as crenolanib and midostaurin. In vitro, crenolanib is a 100 fold more potent than imatinib for inhibition of D842V kinase activity (PMID: 18955458). The PDGFRA alpha monoclonal antibody, olaratumab, was tested in a phase 2 clinical trial assessing previously treated patients with metastatic GIST (PMID: 28426120).



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METHODS

This panel targets 3 genes and uses methodologies of Next generation sequencing using ion ampliseq cancer hotspot panel V2.

The sensitivity of the assays depends on the quality of the block, and tumor content. In validation studies using control material and a variety of cell lines, the minimum analytic detection limit for each of the assays is:
Next Generation Sequencing – 5%
Genomic positions are given in reference to the GRCh37 (hg19) assembly of the human genome

LIMITATION

The accuracy and completeness of this information may vary due to variable information available in different databases Variants with variant allele frequency at nearly 50% or 100% should be considered Germline mutation. To rule out germ line mutations, whole blood sample is recommended to process along with tissue sample. Synonymous mutations were not considered while preparing this report. UDG treatment has not been done. The mutations have not been confirmed using Sanger sequencing and/or alternate technologies and additional testing might be required if clinically indicated. False

DISCLAIMER

This report provides information about the patient's mutations that may aid the physician's decision making process, but this test should not be the sole source of information for making decisions on patient care and treatment. These tests should be interpreted in the context of standard clinical, laboratory, and pathological findings. Identification of a mutation in one or more of these genes does not guarantee activity of the drug in a given indication. Insertions and deletions greater than 20bp in size may not be detected by this assay. Benign mutations in the intronic regions have not been included in this report.

The information provided in this report was collected from various sources that we believe to be reliable and quality control procedures have been put in place to ensure the information provided is as accurate, comprehensive, and current as possible. The information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician. Patient care and treatment decisions should only be made by the physician after taking into account all relevant information available including but not limited to the patient's condition, family history, findings upon examination, results of other diagnostic tests, and the current standards of care. This report should only be used as an aid and the physician should employ clinical judgment in arriving at any decision for patient care or treatment.

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DMC-28322

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PhD, Biotechnology
National Head - Molecular Diagnostics /R & D
NRL - Dr Lal PathLabs Ltd

Dr Lopamudra Deka
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Deputy HOD Histopath, Cytopath &
Chief of Academic Program – NRL

IMPORTANT INSTRUCTIONS

·Test results released pertain to the specimen submitted. ·All test results are dependent on the quality of the sample received by the Laboratory. ·Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician. ·Sample repeats are accepted on request of Referring Physician within 7 days post reporting. ·Report delivery may be delayed due to unforeseen circumstances. Inconvenience is regretted. ·Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours post reporting. ·Test results may show interlaboratory variations. ·The Courts/Forum at Delhi shall have exclusive jurisdiction in all disputes/claims concerning the test(s) & or results of test(s). ·Test results are not valid for medico legal purposes. *Contact customer care Tel No. +91-11-39885050 for all queries related to test results.
(#) Sample drawn from outside source. (*) Not in NABL scope.