

Test Description

The MolQ Liquid Precision Panel includes 50 genes, involving hotspot regions and 3159 unique variants, applicable to a wide range of tumor types for detection of SNV (single and multiple nucleotide variation), Insertion-Deletion, Copy Number Variation (CNV), and gene Fusions. Fusion and splice variants are detected in RNA.

Patient Demographic

Name: Mr. Dharam Prakash
Sex: Male
Date of Birth/Age: 80 years
Disease: Soft Tissue Sarcoma

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Dr AV Institute of Personalized Cancer Therapy and Research
Pathologist: Not Provided

Specimen

Booking ID: 012311230173
Sample Type: Blood
Tumor Content Percentage: NA
Date of Collection: 23-11-2023
Date of Booking: 23-11-2023

CLINICAL SYNOPSIS

Dharam Prakash, is a known case of soft tissue sarcoma. He has been evaluated for pathogenic variations in the genes listed in Appendix 2.

RESULT SUMMARY

Variants detected as per NCCN Guidelines:

No clinically relevant alteration detected.

Other variants detected:

GNA11 mutation (p.Gln209Lys, VAF= 2.7%), **GNA11** mutation (p.Gln209Arg, VAF= 0.4%), **FGFR3** mutation (p.Asp641Gly, VAF= 0.4%) and **AKT3** mutation (p.Glu17Lys, VAF= 0.3%) are present in the given sample in the filtered data.

Note: Our reporting cut off is 0.5% for SNVs, however as per request by clinician we have included the variants below 0.5% and also from filtered data.

The sequencing was performed on 19.8 ng of cfTNA in the given specimen. The average coverage of sequencing was 32460 in this sample.

RESULTS

No clinically relevant alteration was detected.

RELEVANT SOFT TISSUE SARCOMA FINDINGS


Gene	Findings	Gene	Findings
<i>ALK</i>	None detected	<i>NTRK2</i>	None detected
<i>CDK4</i>	None detected	<i>NTRK3</i>	None detected
<i>CDKN2A</i>	None detected	<i>RET</i>	None detected
<i>CTNNB1</i>	None detected	<i>ROS1</i>	None detected
<i>NTRK1</i>	None detected		

RELEVANT BIOMARKERS

No biomarkers associated with relevant evidence.

RECOMMENDATIONS

- Validation of the variant(s) by Sanger sequencing is recommended to rule out false positives.
- Genetic counselling is advised for interpretation on the consequences of the variant(s).
- If results obtained do not match the clinical findings, additional testing should be considered as per referring clinician's recommendations.



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APPENDIX 1: TEST METHODOLOGY

Method

Circulating cell-free total nucleic acid (cfTNA) were isolated from samples using the MagMAX Cell-Free Total Nucleic Acid Isolation Kit. Quantity and quality is checked by Qubit assay and Tape station, respectively. After quality check the isolated and purified sample was directly loaded on Ion Torrent Genexus Next Generation Sequencer and subjected to automated library preparation and template preparation followed by in-depth sequencing.

It utilizes unique molecular tags to enable high sensitivity detection of variants. Analysis is done using Ion Torrent Reporter Software, the data is visualized on Integrative Genomics Viewer (IGV) and analyzed. The final report is generated using OncoPrint curated knowledgebase reporter and includes clinical trials information continuously being updated for the best of the patient management as per clinical guidelines.

DISCLAIMER

- This report was generated using the materials and methods as recommended which required the use of quality reagents, protocols, instruments, software, databases and other items, some of which were provided or made accessible by third parties. A defect or malfunction in any such reagents, protocols, instruments, software, databases and/or other items may compromise the quality or accuracy of the report.
- The report has been created based on, or incorporated inferences to, various scientific manuscripts, references, and other sources of information, including without limitation manuscripts, references, and other sources of information that were prepared by third parties that describe correlations between certain genetic mutations and particular diseases (and/or certain therapeutics that may be useful in ameliorating the effects of such diseases). Such information and correlations are subject to change over time in response to future scientific and medical findings. MolQ Laboratory makes no representation or warranty of any kind, expressed or implied, regarding the accuracy of the information provided by or contained in such manuscripts, references, and other sources is later determined to be inaccurate, the accuracy and quality of the Report may be adversely impacted. MolQ Laboratory is not obligated to notify you of any of the impact that future scientific or medical findings may have on the report.
- The report must always be interpreted and considered within the clinical context, and a physician should always consider the report along with all other pertinent information and data that a physician would prudently consider prior to providing a diagnosis or developing and implementing a plan of care for the patient. The report should never be considered or relied upon alone in making any diagnosis or prognosis. The manifestations of many diseases are caused by more than one gene variant, a single gene variant may be relevant to more than one disease, and certain relevant gene variants may not have been considered in the report. In addition, many diseases are caused or influenced by modifier genes, epigenetic factors, environmental factors, and other variables that are not addressed by the report. This report is based on a Next Generation Assay which does not distinguish between a somatic and a germline variant. If germline variant is in question, further testing is recommended. The report provided by MolQ Laboratory is on a "as is" basis. MolQ Laboratory makes no representation or warranty of any kind, expressed or implied, regarding the report. In no event will MolQ Laboratory be liable for any actual damages, indirect damages, and/or special or consequential damages arising out of or in any way connected with the Report, your use of the report, your reliance on the report, or any defect or inaccurate information included within the report.
- Medical knowledge and annotation are constantly updated and reflects the current knowledge at the time.
- Due to inherent technology limitations of the assay, not all bases of the exome can be covered by this test. Accordingly, variants in regions of insufficient coverage may not be identified and/or interpreted. Therefore, it is possible that certain variants are present in one or more of the genes analyzed, but have not been detected. The variants not detected by the assay that was performed may/ may not impact the phenotype.
- It is also possible that a pathogenic variant is present in a gene that was not selected for analysis and/or interpretation in cases where insufficient phenotypic information is available.
- The report shall be generated within turnaround time (TAT), however, such TAT may vary depending upon the complexity of test(s) requested. MolQ Laboratory under no circumstances will be liable for any delay beyond afore mentioned TAT.
- It is hereby clarified that the report(s) generated from the test(s) do not provide any diagnosis or opinion or recommends any cure in any manner. MolQ Laboratory hereby recommends the patient and/or the guardians of the patients, as the case may be, to take assistance of the clinician or a certified physician or doctor, to interpret the report(s) thus generated. MolQ Laboratory hereby disclaims all liability arising in connection with the report(s).
- In a very few cases genetic test may not show the correct results, e.g. because of the quality of the material provided to MolQ Laboratory. In case where any test provided by MolQ Laboratory fails for unforeseeable or unknown reasons that cannot be influenced by MolQ Laboratory in advance, MolQ Laboratory shall not be responsible for the incomplete, potentially

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- misleading or even wrong result of any testing if such could not be recognized by MolQ Laboratory in advance.
- A negative value in liquid biopsy does not mean true absence of mutation. It may not be detectable in the blood sample but may still be positive in tissue biopsy.
 - This is a laboratory developed test and the development and the performance characteristics of this test was determined by reference laboratory as required by the CLIA 1988 regulations. The report, and the tests used to generate the Report have not been cleared or approved by the US Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. The test results have scientifically shown to be clinically useful.

APPENDIX 2: GENE LIST WITH COVERAGE

DNA Hotspots					
<i>AKT1</i>	<i>AKT2</i>	<i>AKT3</i>	<i>ALK</i>	<i>AR</i>	<i>ARAF</i>
<i>BRAF</i>	<i>CDK4</i>	<i>CDKN2A</i>	<i>CHEK2</i>	<i>CTNNB1</i>	<i>EGFR</i>
<i>ERBB2</i>	<i>ERBB3</i>	<i>ERBB4</i>	<i>ESR1</i>	<i>FGFR1</i>	<i>FGFR2</i>
<i>FGFR3</i>	<i>FGFR4</i>	<i>FLT3</i>	<i>GNA11</i>	<i>GNAQ</i>	<i>GNAS</i>
<i>HRAS</i>	<i>IDH1</i>	<i>IDH2</i>	<i>KIT</i>	<i>KRAS</i>	<i>MAPK1</i>
<i>MAPK2</i>	<i>MET</i>	<i>MTOR</i>	<i>NRAS</i>	<i>NTRK1</i>	<i>NTRK2</i>
<i>NTRK3</i>	<i>PDGFRA</i>	<i>PIK3CA</i>	<i>PTEN</i>	<i>RAF1</i>	<i>RET</i>
<i>ROS1</i>	<i>SMO</i>	<i>TP53</i>			
CNVs					
<i>ALK</i>	<i>AR</i>	<i>CD274</i>	<i>CDKN2A</i>	<i>EGFR</i>	<i>ERBB2</i>
<i>ERBB3</i>	<i>FGFR1</i>	<i>FGFR2</i>	<i>FGFR3</i>	<i>KRAS</i>	<i>MET</i>
<i>PIK3CA</i>	<i>PTEN</i>				
Inter-genetic Fusions					
<i>ALK</i>	<i>BRAF</i>	<i>ESR1</i>	<i>FGFR1</i>	<i>FGFR2</i>	<i>FGFR3</i>
<i>MET</i>	<i>NRG1</i>	<i>NTRK1</i>	<i>NTRK2</i>	<i>NTRK3</i>	<i>NUTM1</i>
<i>RET</i>	<i>ROS1</i>	<i>RSPO2</i>	<i>RSPO3</i>		
Intra-genetic Fusions					
<i>AR</i>	<i>EGFR</i>	<i>MET</i>			