

Patient Name
Age/Gender
MaxID/Lab ID
Ref By

Centre
OP/IP No/UHID
Collection Date/Time
Reporting Date/Time



TEST REQUESTED: Max Cancer 50 Gene Hotspot Panel

METHOD USED

Next Generation Sequencing

CLINICAL INFORMATION/ SAMPLE INFORMATION

As per pathology report, features are consistent with clear cell renal cell carcinoma in right kidney, adrenal sparing radical nephrectomy.

FFPE Block: (Block No.: BB-2545/24, Tumor Content: ~60%-65%)

TARGETED GENES

| | | | | | | |
|----------------------|----------------------|---------------------|--------------------|----------------------|-----------------------|--------------------|
| ABL1 (0) | CSF1R (0) | FGFR2 (0) | IDH1 (0) | MLH1 (0) | PTPN11 (0) | TP53 (0) |
| AKT1 (0) | CTNNB1 (0) | FGFR3 (0) | IDH2 (0) | MPL (0) | RB1 (0) | VHL (0) |
| ALK (0) | EGFR (0) | FLT3 (0) | JAK2 (0) | NOTCH1 (0) | RET (0) | |
| APC (0) | ERBB2 (0) | GNA11 (0) | JAK3 (0) | NPM1 (0) | SMAD4 (0) | |
| ATM (0) | ERBB4 (0) | GNAS (0) | KDR (0) | NRAS (0) | SMARCB1 (0) | |
| BRAF (0) | EZH2 (0) | GNAQ (0) | KIT (0) | PDGFRA (0) | SMO (0) | |
| CDH1 (0) | FBXW7 (0) | HNF1A (0) | KRAS (0) | PIK3CA (0) | SRC (0) | |
| CDKN2A (0) | FGFR1 (0) | HRAS (0) | MET (0) | PTEN (0) | STK11 (0) | |

Primary Findings

| Gene | CDS Variant | Amino Acid Change | Exon | Allele Frequency | Coverage | dbSNP ID | Pathogenicity (Clinvar/Varsome) |
|------------------------------|-------------|-------------------|------|------------------|----------|----------|---------------------------------|
| No Significant variant found | | | | | | | |

SAMPLE STATISTICS

| | |
|----------|---------|
| Coverage | 100% |
| Depth | 22,333X |

ADDITIONAL FINDINGS

No other variant that warrants to be reported was detected



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TEST METHODOLOGY

Background

Multi gene analysis through next generation sequencing allows the identification of variants to understand their prognostic and therapeutic implications in different cancer types, if any. Targeted application of next-generation sequencing (NGS) technology allows detection of specific mutations that can provide treatment opportunities to the patients. This panel with improved primer design and as little as 10 ng of DNA enable researchers to sequence challenging samples such as Formalin fixed, paraffin embedded (FFPE) tissue which exhibit variable quality. Additionally, even degraded samples can be used to generate reliable data using this panel as the primers are designed to produce, on average, 154 bp amplicons.

Method

The Ion AmpliSeq™ Cancer Hotspot Panel v2 was used to carry out next generation sequencing. After sequencing, automated analysis was performed with Torrent Suite™ Software. Variant annotations were then done using Ion Reporter™ Software. Clinically relevant mutations were also checked using published literature and databases.

Limitations

The accuracy and completeness may vary due to variable information available in different databases. The classification of variants of unknown significance can change over time. Synonymous mutations were not considered while preparing this report. The mutations have not been confirmed using Sanger sequencing and/or alternate technologies. To rule out germ line mutations i.e. variant with variant allele frequency at nearly 50% or 100%, whole blood sample is recommended to process along with tissue sample.

DISCLAIMER

A Negative result implying non-detection of mutation/deletion indicates a Benign/likely Benign polymorphism. A negative test result may also be due to the inherent technical limitations of the assay. Results obtained should be interpreted with consideration of the overall picture obtained from clinical, laboratory, and pathological findings. Rare polymorphisms may lead to false negative or positive results. False negative results may be due to sampling error/errors in sample handling as well as clonal density below the limit of detection. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Identification of a mutation in one or more of these genes does not guarantee activity of the drug in a given indication due to the presence of contraindicated mutation in the gene not covered by the panel.

The accuracy and completeness may vary due to variable information available in different databases. Classification of the variant may change overtime. An updated variant classification may be obtained on request. Insertions and deletions greater than 20bp in size may not be detected by this assay. The scope of this assay limits to SNVs, MNVs and short deletions/duplications. Due to poor quality of FFPE DNA, indeterminate result due to low gene coverage or low variant depth cannot be ruled out. The sensitivity of the assays depends on the quality of the block, and tumor content.

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 sound clinical judgment in arriving at any decision for patient care or treatment.

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