# Solid Tumor Mutation Panel, Sequencing

**Patient:** [redacted]
DOB: [redacted] Age: [redacted]
**Patient Identifiers:** [redacted]
**Visit Number (FIN):** [redacted]

Client: [redacted]
Physician: [redacted]

ARUP Test Code: 3004294

Collection Date: 02/10/2025
Received in lab: 02/19/2025
Completion Date: 02/28/2025

## Comment:

Block ID: SPP25-2787 A3
Clinical History: Serous adenocarcinoma
Sample Source: Left ovary

## TIER 1: Variants of Strong Clinical Significance

Variants with therapeutic, diagnostic, or prognostic significance in the patient's specific tumor type

None Detected

## TIER 2: Variants of Potential Clinical Significance

Variants with potential therapeutic, diagnostic, or prognostic significance

| Gene | Transcript ID | DNA Variant | Protein Variant |
|---|---|---|---|
| TP53 | NM_000546.5 | c.626_627del | p.Arg209fs |

### Interpretation

**TP53 -** Somatic mutations in TP53 are found in 96% of patients with high grade serous ovarian cancer (1). This mutation is predicted to alter the normal function of the tumor suppressor p53.

## TIER 3: Variants of Unknown Clinical Significance (VUS)

| Gene | Transcript ID | DNA Variant | Protein Variant |
|---|---|---|---|
| ALK | NM_004304.4 | c.2801G>A | p.Gly934Asp |

### Interpretation

**ALK -** Insufficient clinically relevant information was found on this variant.

## Low Coverage Regions

Listed below are regions where the average sequencing depth (number of times a particular position is sequenced) for 20% or more of the region is below our stringent cutoff of 300. Sensitivity for detection of low allellic frequency mutations may be reduced in areas with low depth of coverage. If high quality variants are detected in these regions, they will be listed above.

None

This result has been reviewed and approved by [redacted]

## References

(1) , Integrated genomic analyses of ovarian carcinoma. Nature 2011. PMID:21720365



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(2) Li MM, Datto M, Duncavage EJ et al, Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer: A Joint Consensus Recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists. J Mol Diagn 2017. PMID:27993330

BACKGROUND INFORMATION: Solid Tumor Mutation Panel by Next Generation Sequencing

CHARACTERISTICS: Specific somatic variants have been discovered in multiple cancer-related genes and have diagnostic, therapeutic and/or prognostic utility in several cancer types. Targeted next generation sequencing is utilized in this test for the detection of hotspot variants within 44 cancer-related genes and includes extended RAS variant detection. The personalized variant profile may be useful for prediction of patient diagnosis, prognosis and/or response to targeted therapies in solid tumors including, melanoma, gastrointestinal stromal tumors (GIST), colorectal cancers, bladder cancers, and hepatocellular cancers.

GENES TESTED: AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CTNNB1, DDR2, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KDR, KIT, KRAS, MAP2K1, MET, MTOR, NOTCH1, NRAS, NTRK1, PDGFRA, PIK3CA, PTEN, RB1, RET, ROS1, SMAD4, SMO, STK11, TERT promoter, TP53, VHL. A full list of targeted regions within the above genes is available through this link: http://ltd.aruplab.com/Tests/Pdf/375.

METHODOLOGY: Genomic DNA is isolated from microscopically-guided dissection of tumor tissue and then enriched for the targeted regions of the tested genes. The variant status of the 44 targeted genes is determined by massively parallel sequencing (next generation sequencing). The hg19 (GRCh37) reference sequence is used as a reference for identifying genetic variants.

LIMITATIONS: This test will not detect variants in areas outside the targeted genomic regions or below the limit of detection. Copy number alterations, translocations, microsatellite instability, and tumor mutational burden will not be detected. If clinical indication is lung cancer, additional clinical evaluation may be considered for complete genetic analysis including, detection of translocations or gene rearrangements. This test is not intended to detect minimal residual disease. This test evaluates for variants in tumor tissue only and cannot distinguish between somatic and germline variants. Therefore, if a hereditary/familial cancer is of clinical concern, consider additional clinical evaluation and genetic counseling prior to additional testing. In some cases, variants may not be identified due to technical limitations in the presence of known pseudogenes, homologous regions and/or low mapability regions. This includes variants in PTEN exons 1, 2, 4, 5, 6, 7, 8 and 9; MAP2K1 exons 2, 7 and 11; CDKN2A exon 2, PIK3CA exons 10 and 14; GNAQ exon 5; EZH2 exon 18; and BRAF exon 11. It is also possible that some large insertion/deletion variants (especially those greater than 60bp) may not be detected. Tissue samples yielding at least 10ng are acceptable but may yield suboptimal results if yield is less than 50ng.

LIMIT OF DETECTION: 5 percent mutant allele for single nucleotide variants (SNV), small to medium sized multi-nucleotide variants (MNV) (less than 60bp).



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ANALYTICAL SENSITIVITY (PPA): Analytical sensitivity for all variant classes available through this link: http://ltd.aruplab.com/Tests/Pdf/375.

CLINICAL DISCLAIMER: Results of this test must always be interpreted within the clinical context and other relevant data and should not be used alone for a diagnosis of malignancy. This test is not intended to detect minimal residual disease.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.



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