ONCOLOGY genpath an OPKO Health Company

Final Report

PHYSICIAN

SAMPLE, PHYSICIAN
PATHOLOGY HOSPITAL
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C:

PATIENT

SAMPLE, PATIENT
DOB: 1/01/1966 **Age:** 54 Y **Sex:** M
Surgical #:
Patient ID:

SAMPLE

Specimen ID: XXXXXXXXX
Date of Report: 09/01/2020 11:51 AM
Date Collected: 08/07/2020 Time Unknown
Date Received: 08/18/2020 7:57 AM
Source: CYST, ORAL/DENTAL
Clinical Information:
mammary analog secretory carcinoma

ONKOSIGHT NEXT GENERATION SEQUENCING GENE FUSION PANEL

INTERPRETATION

POSITIVE: ETV6-NTRK3 gene fusion DETECTED

No pathogenic gene fusions detected involving AKT1, ALK, AXL, BRAF, CCND1, EGFR, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, PPARG, RAF1, RET, ROS1 or THADA.

ETV6-NTRK3 fusions are recurrent in mammary analog secretory carcinomas, and less frequently, among a wide variety of other solid tumor types. Among salivary glandular neoplasms, ETV6-NTRK3 fusions are relatively specific for mammary analogue secretory carcinoma (PubMed ID: 26492182; Mammary Analogue Secretory Carcinoma of Salivary Glands: Molecular Analysis of 25 ETV6 Gene Rearranged Tumors With Lack of Detection of Classical ETV6-NTRK3 Fusion Transcript by Standard RT-PCR: Report of 4 Cases Harboring ETV6-X Gene Fusion; Am J Surg Pathol. 2016 Jan;40(1):3-13.).

NTRK gene fusions are therapeutic indicators for Larotrectinib or Entrectinib under certain circumstances (NCCN Guidelines, Head and Neck Cancers, Version 2.2020; and, PubMed ID: 31803506. How I treat NTRK gene fusion-positive cancers. ESMO Open . 2019 Nov 25;4(Suppl 2):e000612).

RESULTS

Tumor Cellularity: >10%

5'GENE FUSION TRANSCRIPT	3'GENE FUSION TRANSCRIPT
NTRK3; exon:15; NM_002530.3	**ETV6; exon:5; NM_001987.4**

METHOD

Tissue sections are reviewed by a pathologist; specimens with minimal tumor cells may be rejected. RNA is isolated from the selected area of the sample. Anchored multiplex PCR for targeted next-generation sequencing is performed. The sequenced sample is a reverse transcription PCR-amplified fragment library in which each sample is uniquely identified by ligation of a short oligonucleotide barcode. The panel targets mutltiple rearrangements and the resultant sequence identifies the exons of the fusion transcript arising from that target and the partner gene. Each sample is monitored for quality to ensure reliable fusion detection. Variants are identified by an automated process that takes into account statistical confidence of base calling and alignment and mapping quality [Archer Analysis vs 3.3]. The software requires a single read spanning two separate genes of at least 23 bp each to be considered a valid fusion candidate and each read that spans the same breakpoint is grouped together. A final consensus sequence is constructed and used to annotate the two (or more) fusion partners by comparing to the human genome with BLAST and annotations from the RefSeq database cross-referenced with the manufacturers database of known fusions published in the literature [Archer Quiver Database]. The assay can detect RNA fusions in samples containing 5% or more cells with the chromosomal translocation.

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This report was electronically signed.

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