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Department of Molecular Oncology

Patient information		Ref. Doctor/ Hospital/ Specimen	
Patient Name:	Mr. Om Prakash	Ref Hospital:	Sage Path Labs Pvt Ltd
Barcode:	A0320829	Sample type:	Biopsy from right lung (FFPE Block)
Age/Gender:	67/Male	Received date:	01-03-24
Test name:	Lung Cancer Panel	Reported date:	18-03-24

Indication: Non-small cell lung cancer

Sample type: FFPE Block

Tumor Percentage: Adequate tumor cells (50 %) were noted in the submitted tissue block.

Result:

1. Clinically relevant pathogenic *EGFR* gene mutation Identified.
2. Negative for fusion in the tested genes.

Variant Identified:

Gene	Locus	Variant	Depth/VAF	Impact	Variant Classification
EGFR NM_005228.5		c.2235_2251delGGAATTAAGAGAAGCAAin			

Relevant Biomarkers

Tier	Genomic Alteration	Relevant Therapies (In this cancer type)	Relevant Therapies (In other cancer type)	Clinical Trials
IA	*EGFR exon 19 deletion* epidermal growth factor receptor Allele Frequency: 24.95% Transcript: NM_005228.5	**afatinib** [1, 2] **bevacizumab* + erlotinib** [2] **dacomitinib** [1, 2] **erlotinib** [1, 2] **erlotinib + ramucirumab** [1, 2] **gefitinib** [1, 2] **osimertinib** [1, 2] **atezolizumab + bevacizumab + chemotherapy** **gefitinib + chemotherapy**	None	262

Public data sources included in relevant therapies: FDA[1], NCCN, **EMA**[2], ESMO
*** Includes biosimilars/generics**

Alerts informed by public data sources: Contraindicated, Resistance, Breakthrough, Fast Track

EGFR exon 19 deletion	**patritumab deruxtecan** [1] **CPO-301** [1], **osimertinib + quaratusugene ozeplasmid** [1]

Public data sources included in alerts: FDA[1], NCCN, **EMA**[2], ESMO

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Recommendations: These findings should be correlated with other clinical and laboratory tests for a definite conclusive interpretation.

Variant Description

EGFR: c.2235_2251delGGAATTAAGAGAAGCAAinsAATTC: p.Glu746_Thr751delinsIlePro: Pathogenic:

The p.Glu746_Thr751delinsIlePro variant (also known as c.2235_2251delGGAATTAAGAGAAGCAAinsAATTC), pathogenic non-frame shift deletion was detected in exon 19 of EGFR gene at position 55242465 with variant allele frequency of **24.95% at a total depth of 1928X**. It belongs to Oncomine Variant class of EGFR Exon19 Deletion which leads to Gain-of-Function. It is represented as rs727504332 in dbSNP and COSM6225 in Cosmic database. It is interpreted as pathogenic according to ClinVar database [VCV000177808]. It is predicted as pathogenic by MutatonTatster2. It is absent in the population frequency database like gnomAD, ExAC and 1000G database.

Biomarker Descriptions

EGFR (epidermal growth factor receptor)

Background: The EGFR gene encodes the epidermal growth factor receptor (EGFR) tyrosine kinase, a member of the ERBB/human epidermal growth factor receptor (HER) family. In addition to EGFR/ERBB1/HER1, other members of the ERBB/HER family include ERBB2/HER2, ERBB3/HER3, and ERBB4/HER41. EGFR ligand induced dimerization results in kinase activation and leads to stimulation of oncogenic signaling pathways including the PI3K/AKT/MTOR and RAS/RAF/MEK/ERK pathways. Activation of these pathways promote cell proliferation, differentiation, and survival [2,3].

Alterations and prevalence: Recurrent somatic mutations in the tyrosine kinase domain (TKD) of EGFR are observed in approximately 10-20% of lung adenocarcinoma, and at higher frequencies in never-smoker, female, and Asian populations4,5,6,7. The most common mutations occur near the ATP-binding pocket of the TKD and include short in-frame deletions in exon 19 (EGFR exon 19 deletion) and the L858R amino acid substitution in exon 218. These mutations constitutively activate EGFR resulting in downstream signaling, and represent 80% of the EGFR mutations observed in lung cancer. A second group of less prevalent activating mutations include E709K, G719X, S768I, L861Q, and short in-frame insertion mutations in exon 20 [9,10,11,12]. EGFR activating mutations in lung cancer tend to be mutually exclusive to KRAS activating mutations13. In contrast, a different set of recurrent activating EGFR mutations in the extracellular domain include R108K, A289V and G598V and are primarily observed in glioblastoma8,14. Amplification of EGFR is observed in several cancer types including 30% of glioblastoma, 12% of esophageal cancer, 10% of head and neck cancer, 5% of bladder cancer, and 5% of lung squamous cell carcinoma [5,6,7,14,15]. Deletion of exons 2-7, encoding the extracellular domain of EGFR (EGFRvIII), results in overexpression of a ligand-independent constitutively active protein and is observed in approximately 30% of glioblastoma [16,17,18].

Relevant Therapy Summary

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
afatinib	●	●	●	●	● (IV)
osimertinib	●	●	●	●	● (IV)
dacomitinib	●	●	●	●	● (II)
erlotinib	●	●	●	●	● (II)
erlotinib + ramucirumab	●	●	●	●	✖
gefitinib	●	●	●	●	✖
bevacizumab + erlotinib	✖	●	●	●	✖
bevacizumab (Allergan) + erlotinib	✖	✖	●	✖	✖
bevacizumab (Celltrion) + erlotinib	✖	✖	●	✖	✖
bevacizumab (Mabxience) + erlotinib	✖	✖	●	✖	✖
bevacizumab (Pfizer) + erlotinib	✖	✖	●	✖	✖
bevacizumab (Samsung Bioepis) + erlotinib	✖	✖	●	✖	✖
bevacizumab (Stada) + erlotinib	✖	✖	●	✖	✖
atezolizumab + bevacizumab + carboplatin + paclitaxel	✖	✖	✖	●	✖
gefitinib + carboplatin + pemetrexed	✖	✖	✖	●	✖
afatinib, bevacizumab, chemotherapy	✖	✖	✖	✖	● (IV)
almonertinib	✖	✖	✖	✖	● (IV)
apatinib + EGFR tyrosine kinase inhibitor	✖	✖	✖	✖	● (IV)
apatinib, camrelizumab	✖	✖	✖	✖	● (IV)
apatinib, gefitinib	✖	✖	✖	✖	● (IV)
bevacizumab + osimertinib, osimertinib	✖	✖	✖	✖	● (IV)
bevacizumab, almonertinib, chemotherapy	✖	✖	✖	✖	● (IV)
catequentinib, toripalimab	✖	✖	✖	✖	● (IV)
EGFR tyrosine kinase inhibitor	✖	✖	✖	✖	● (IV)
gefitinib, chemotherapy	✖	✖	✖	✖	(IV)
gefitinib, endostatin	✖	✖	✖	✖	(IV)
gefitinib, radiation therapy	✖	✖	✖	✖	(IV)
icotinib hydrochloride, chemotherapy	✖	✖	✖	✖	(IV)

natural product, gefitinib, erlotinib, icotinib hydrochloride, osimertinib, almonertinib, furmonertinib	✖	✖	✖	✖	● (IV)
almonertinib, apatinib	✖	✖	✖	✖	● (III)
almonertinib, chemotherapy	✖	✖	✖	✖	● (III)
almonertinib, radiation therapy	✖	✖	✖	✖	● (III)
almonertinib, radiation therapy, chemotherapy	✖	✖	✖	✖	● (III)
ASK120067, gefitinib	✖	✖	✖	✖	● (III)
befotertinib, icotinib hydrochloride	✖	✖	✖	✖	● (III)
bevacizumab, osimertinib	✖	✖	✖	✖	● (III)
CK-101, gefitinib	✖	✖	✖	✖	● (III)
erlotinib, chemotherapy	✖	✖	✖	✖	● (III)

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.

Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
FHND9041, afatinib	✖	✖	✖	✖	● (III)
furmonertinib	✖	✖	✖	✖	● (III)
gefitinib, afatinib, erlotinib, metformin hydrochloride	✖	✖	✖	✖	● (III)
gefitinib, icotinib hydrochloride, erlotinib, radiation therapy	✖	✖	✖	✖	● (III)
icotinib hydrochloride, catequentinib	✖	✖	✖	✖	● (III)
icotinib hydrochloride, radiation therapy	✖	✖	✖	✖	● (III)
ivonescimab, chemotherapy	✖	✖	✖	✖	● (III)
osimeítinib, bevacizumab	✖	✖	✖	✖	● (III)
osimertinib, chemotherapy	✖	✖	✖	✖	● (III)
patíitumab deíuxtecan	✖	✖	✖	✖	● (III)
savolitinib, osimertinib	✖	✖	✖	✖	● (III)
SH-1028, gefitinib	✖	✖	✖	✖	● (III)
SHR-1701, bevacizumab, chemotherapy	✖	✖	✖	✖	● (III)
SKB264	✖	✖	✖	✖	● (III)
targeted therapy	✖	✖	✖	✖	● (III)
IY-9591, osimeítinib	✖	✖	✖	✖	● (III)
zipalertinib, chemotherapy	✖	✖	✖	✖	● (III)
PM-8002, chemotheíapy	✖	✖	✖	✖	● (II/III)
afatinib, chemotherapy	✖	✖	✖	✖	● (II)
almoneítinib, bevacizumab	✖	✖	✖	✖	● (II)
almonertinib, chemoradiation therapy	✖	✖	✖	✖	● (II)
almoneítinib, dacomitinib	✖	✖	✖	✖	● (II)
amivantamab, lazertinib	✖	✖	✖	✖	● (II)
amivantamab, lazeítinib, chemotheíapy	✖	✖	✖	✖	● (II)
atezolizumab, bevacizumab, tiragolumab	✖	✖	✖	✖	● (II)
bevacizumab, afatinib	✖	✖	✖	✖	● (II)
bevacizumab, furmonertinib	✖	✖	✖	✖	● (II)
BL-B01D1, osimeítinib	✖	✖	✖	✖	● (II)

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.

Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
camrelizumab, apatinib	✖	✖	✖	✖	● (II)
capmatinib, osimertinib, ramucirumab	✖	✖	✖	✖	● (II)
catequentinib	✖	✖	✖	✖	● (II)
catequentinib, almonertinib	✖	✖	✖	✖	● (II)
catequentinib, gefitinib	✖	✖	✖	✖	● (II)
catequentinib, gefitinib, eílotinib, icotinib hydíochloíide	✖	✖	✖	✖	● (II)
catequentinib, icotinib hydíochloíide, gefitinib	✖	✖	✖	✖	● (II)
chemotheíapy, atezolizumab, bevacizumab	✖	✖	✖	✖	● (II)
dacomitinib, osimeítinib	✖	✖	✖	✖	● (II)
EGFR tyíosine kinase inhibitoí + chemotheíapy, EGFR tyíosine kinase inhibitoí	✖	✖	✖	✖	● (II)
EGFR tyíosine kinase inhibitoí, íadiation theíapy	✖	✖	✖	✖	● (II)
eílotinib, OBI-833	✖	✖	✖	✖	● (II)
eílotinib, íamuciíumab	✖	✖	✖	✖	● (II)
famitinib, almoneítinib	✖	✖	✖	✖	● (II)
fuímoneítinib, bevacizumab	✖	✖	✖	✖	● (II)
fuímoneítinib, catequentinib	✖	✖	✖	✖	● (II)
fuímoneítinib, chemotheíapy	✖	✖	✖	✖	● (II)
fuímoneítinib, chemotheíapy, bevacizumab	✖	✖	✖	✖	● (II)
fuímoneítinib, icotinib hydíochloíide	✖	✖	✖	✖	● (II)
gefitinib, bevacizumab, chemotheíapy	✖	✖	✖	✖	● (II)
gefitinib, eílotinib, afatinib	✖	✖	✖	✖	● (II)
gefitinib, thalidomide	✖	✖	✖	✖	● (II)
icotinib hydíochloíide	✖	✖	✖	✖	● (II)
icotinib hydíochloíide, autologous RAK cell	✖	✖	✖	✖	● (II)
icotinib hydíochloíide, osimeítinib	✖	✖	✖	✖	● (II)
keynatinib	✖	✖	✖	✖	● (II)
lazeítinib	✖	✖	✖	✖	● (II)
lazeítinib, chemotheíapy	✖	✖	✖	✖	● (II)

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.

Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
olaparib, durvalumab	✖	✖	✖	✖	(II)
osimertinib, amivantamab	✖	✖	✖	✖	(II)
osimertinib, datopotamab deruxtecan, savolitinib, gefitinib, necitumumab, chemotherapy, durvalumab, alectinib, selpercatinib, selumetinib	✖	✖	✖	✖	● (II)
osimertinib, radiation therapy	✖	✖	✖	✖	● (II)
osimertinib, savolitinib	✖	✖	✖	✖	● (II)
PD-1 Inhibitor, chemotherapy	✖	✖	✖	✖	● (II)
ramucirumab, erlotinib	✖	✖	✖	✖	● (II)
serplulimab, chemotherapy	✖	✖	✖	✖	● (II)
SH-1028	✖	✖	✖	✖	● (II)
sintilimab, chemotherapy	✖	✖	✖	✖	● (II)
SKB264, chemotherapy	✖	✖	✖	✖	● (II)
SKB264, osimertinib	✖	✖	✖	✖	● (II)
sunvozertinib	✖	✖	✖	✖	● (II)
sunvozertinib, golidocitinib	✖	✖	✖	✖	● (II)
sutetinib	✖	✖	✖	✖	● (II)
tislelizumab, chemotherapy	✖	✖	✖	✖	● (II)
tislelizumab, chemotherapy, bevacizumab	✖	✖	✖	✖	● (II)
tocilizumab, ipilimumab, nivolumab	✖	✖	✖	✖	● (II)
toripalimab, bevacizumab, Clostridium butyricum, chemotherapy	✖	✖	✖	✖	● (II)
TY-9591	✖	✖	✖	✖	● (II)
tyrosine kinase inhibitors, radiation therapy	✖	✖	✖	✖	● (II)
zipalertinib	✖	✖	✖	✖	● (II)
zorifertinib, pirotinib	✖	✖	✖	✖	● (II)
AFM-24_I, atezolizumab	✖	✖	✖	✖	● (I/II)
almonertinib, icotinib hydrochloride	✖	✖	✖	✖	(I/II)
AP-L1898	✖	✖	✖	✖	● (I/II)
BBT-207	✖	✖	✖	✖	(I/II)

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Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
BEBT-908, BEBT-109	✖	✖	✖	✖	● (I/II)
BLU-451, chemotherapy	✖	✖	✖	✖	● (I/II)
BLU-945, osimertinib	✖	✖	✖	✖	● (I/II)
bozitinib, osimertinib	✖	✖	✖	✖	● (I/II)
BPI-361175	✖	✖	✖	✖	● (I/II)
CBT-502, catequentinib	✖	✖	✖	✖	● (I/II)
chemotherapy, cetuximab, natural killer cell therapy	✖	✖	✖	✖	● (I/II)
dacomitinib, catequentinib	✖	✖	✖	✖	● (I/II)
dositinib	✖	✖	✖	✖	● (I/II)
dresbuxelimab, ivonescimab, chemotherapy	✖	✖	✖	✖	● (I/II)
E01001	✖	✖	✖	✖	● (I/II)
EMB01	✖	✖	✖	✖	● (I/II)
erlotinib, chemotherapy, bevacizumab	✖	✖	✖	✖	● (I/II)
FWD-1509	✖	✖	✖	✖	● (I/II)
GB263T	✖	✖	✖	✖	● (I/II)
H-002	✖	✖	✖	✖	● (I/II)
IN10018, furmonertinib	✖	✖	✖	✖	● (I/II)
JFAN-1001	✖	✖	✖	✖	● (I/II)
JIN-A-02	✖	✖	✖	✖	● (I/II)
MCLA-129	✖	✖	✖	✖	● (I/II)
MRG-003, HX008	✖	✖	✖	✖	● (I/II)
MRTX0902	✖	✖	✖	✖	● (I/II)
necitumumab, osimertinib	✖	✖	✖	✖	● (I/II)
necitumumab, trastuzumab, osimertinib	✖	✖	✖	✖	● (I/II)
nivolumab, pembrolizumab, atezolizumab, ipilimumab, radiation therapy	✖	✖	✖	✖	● (I/II)
ONC-392, osimertinib	✖	✖	✖	✖	● (I/II)
PLB-1004, baricitinib	✖	✖	✖	✖	● (I/II)
quaratusugene ozeplasmid, osimertinib	✖	✖	✖	✖	● (I/II)

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Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

EGFR exon 19 deletion (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
TAS-3351	✖	✖	✖	✖	● (I/II)
telaglenastat, osimertinib	✖	✖	✖	✖	● (I/II)
TQ-B3525, osimertinib	✖	✖	✖	✖	● (I/II)
ABBV 400	✖	✖	✖	✖	● (I)
AZD-9592	✖	✖	✖	✖	● (I)
BAY-2927088	✖	✖	✖	✖	● (I)
BBP-398	✖	✖	✖	✖	● (I)
BBP-398, osimertinib	✖	✖	✖	✖	● (I)
BCA101, pembrolizumab	✖	✖	✖	✖	● (I)
BDTX 1535	✖	✖	✖	✖	● (I)
catequentinib, gefitinib, metformin hydrochloride	✖	✖	✖	✖	● (I)
cemiplimab, sarilumab	✖	✖	✖	✖	● (I)
CPO-301	✖	✖	✖	✖	● (I)
EGFR tyrosine kinase inhibitor, catequentinib	✖	✖	✖	✖	● (I)
genolimzumab, fruquintinib	✖	✖	✖	✖	● (I)
HBI 2376	✖	✖	✖	✖	● (I)
HS-10241, almonertinib	✖	✖	✖	✖	● (I)
HSK-40118	✖	✖	✖	✖	● (I)
IBI-318, lenvatinib	✖	✖	✖	✖	● (I)
MRX-2843, osimertinib	✖	✖	✖	✖	● (I)
nivolumab, ipilimumab, radiation therapy	✖	✖	✖	✖	● (I)
osimertinib, carotuximab	✖	✖	✖	✖	● (I)
osimertinib, ipilimumab	✖	✖	✖	✖	● (I)
osimertinib, Minnelide	✖	✖	✖	✖	● (I)
osimertinib, tegatrabetan	✖	✖	✖	✖	● (I)
patritumab deruxtecan, osimertinib	✖	✖	✖	✖	(I)
QLH-11811	✖	✖	✖	✖	(I)
repotrectinib, osimertinib	✖	✖	✖	✖	(I)
RLY-1971, osimertinib	✖	✖	✖	✖	(I)

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Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
sintilimab, catequentinib	✖	✖	✖	✖	● (I)
sunvozertinib, midazolam, digoxin, rosuvastatin calcium	✖	✖	✖	✖	● (I)
TAS 2940	✖	✖	✖	✖	● (I)
TAVO-412	✖	✖	✖	✖	● (I)
TNO-155, nazartinib	✖	✖	✖	✖	● (I)
VIC-1911, osimertinib	✖	✖	✖	✖	● (I)
WJ13404	✖	✖	✖	✖	● (I)
WSD-0922	✖	✖	✖	✖	● (I)
WTS-004	✖	✖	✖	✖	● (I)
YL-202	✖	✖	✖	✖	● (I)
ZZ06	✖	✖	✖	✖	● (I)

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EGFR exon 19 deletion (continued)

Methodology

DNA/RNA was extracted from FFPE tissue samples using standard Qiagen kits. Briefly,10ng of extracted DNA/RNA was amplified with a custom 12-gene panel following the kit's instructions. Ion S5 platform sequencing was performed according to its user manual. Sequencing reads underwent quality control and mapping to the hg19 human reference genome using Ion Reporter™ Software 5.18.2.0. This software also called variants (SNVs, small InDels, CNVs, Fusions) and annotated them. Various databases were employed to identify and characterize genes associated with these variants, with OMIM and ClinVar specifically focusing on disease-related annotations. Common variants/polymorphisms were eliminated based on population frequency data from 1000 genomes, ExAC, GnomAD, and ESP. Finally, PolyPhen-2 and SIFT scores predicted the potential impact of coding non-synonymous SNVs on protein structure and function, while Oncomine Reporter software annotated variants with relevant labels, guidelines, and information from a curated list of global clinical trials.

Run QC statistics

Sample is sequenced at Average base coverage depth of 5,969. The Target base coverage at 500X is 99.99%.

Variant classification

Pathogenic — A **pathogenic variant** is a specific type of change in a gene sequence that is known to **increase the risk of developing a particular disease**. These changes are classified as pathogenic based on strong evidence demonstrating a causal link between the variant and the disease.

Likely Pathogenic — A strong candidate variant exhibiting functional and/or genetic evidence highly **suggestive of pathogenicity**, but lacking definitive proof of causality for the presenting symptoms. Requires further investigation for conclusive classification.

Variant of uncertain significance — A variation in a genetic sequence for which the association with disease risk is unclear and there is insufficient evidence to prove a connection between the variant and disease. They are not used as a basis for clinical decisions. A variant may be reclassified over time as more information becomes available.

Variant Categorization

Tier I	Variants with strong clinical significance	Level A evidence	FDA -approved therapy guiding treatment decisions with the highest confidence.
		Level B evidence	Findings supported by observational studies or expert consensus, providing valuable but slightly less robust evidence for clinical decision-making.
Tier II	Variants with potential clinical significance	Level C evidence	FDA -approved therapies for various tumor types rely on multiple small-scale studies with varying degrees of consensus among researchers.
		Level D evidence	Preclinical trials or few case reports without consensus
Tier III	Variants of unknown clinical significance		Genetic alterations with limited clinical evidence,and lacking validation through clinical studies or functional assays. Not observed at significant allele frequency in the subpopulation or pan-cancer or tumor specific databases thus uncertain in guiding treatment decisions.

Genes Assayed

Genes Analyzed for the Detection of DNA Sequence Variants

BRAF, EGFR, ERBB2, KRAS, MET, NRAS, PIK3CA, RET, ROS1, ALK

Genes Analyzed for the Detection of Copy Number Variations

ALK, BRAF, EGFR, ERBB2, FGFR2, MET, PIK3CA

Genes Analyzed for the Detection of Fusions

ALK, MET, BRAF, EGFR, ERBB2, ROS1, NTRK1, NTRK2, NTRK3, RET

Limitations and Disclaimer:

1. This test was developed and its performance characteristics determined by YODA Laboratory, it has not been cleared or approved by the US Food and Drug Administration and NABL.
2. This NGS test used does not allow definitive differentiation between germline and somatic variants. However, variants with variant allele frequency at nearly 50% or 100% should be considered Germline mutation. To rule out germ line mutations, repeat analysis using peripheral blood/saliva sample is recommended.
3. The test was performed in a third-party lab
4. Certain genes may not be covered completely, and few mutations may not be detected in the presence of pseudogenes or in repetitive or homologous regions.
5. False negative results may be due to sampling issues, errors in sample handling, mislabeling, transportation issues, technical limitations of the assay and mutations frequency below the limit of detection of the assay, i.e., 5% for SNVs and 10% for short indels. It is also possible some complex insertion/deletion variants may not be identified.
6. Sanger confirmation of reported mutations is available on request with additional charges.
7. This test is not intended to detect minimal residual disease.
8. Results of this test need be interpreted within the context of clinical findings and other relevant clinical and laboratory data and should not be used alone.

Disclaimer: The data presented here is from a curated knowledgebase of publicly available information, but may not be exhaustive. The data version is 2023.11(007). The content of this report has not been evaluated or approved by FDA, EMA or other regulatory agencies.

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