pulmo CÖRE 20 Gene Panel

Primary Tumor Site	Specimen Site	Specimen Type
N/A	Right side supraclavicular lymph node biopsy	FFPE
Specimen Collection Date & Time		Date & Time of Accessioning
15/07/2024 00:00 Hrs		17/07/2024 13:07 Hrs

Case Number:	**24010010573**
Patient Name:	**S. J. S. L. Samarasinghe**
Age/Sex:	**28 Yrs/Male**
Patient Location:	**Colombo**
Hospital Name:	**Aegle Omics Pvt Ltd**
Physician Name:	**Dr. Mahendra Perera**
Date & Time of Reporting:	**30/07/2024 12:38 Hrs**

TEST INFORMATION

***pulmo* CORE 20 gene panel** is a **Next Generation Sequencing (NGS)** based assay that identifies clinically relevant genomic alterations within the 20 genes, that are most frequently altered in Non Small Cell Lung Cancer (NSCLC). (Detailed information has been provided in additional information section).

SPECIMEN INFORMATION

Received 1 paraffin block labelled as 2511/1/23. Tumor content: 30%.

CLINICAL HISTORY

Metastatic deposits of carcinoma.

RESULTS

MUTATION TYPE	GENES TESTED	RESULTS
SNVs, short indels, Fusions & CNV	**ALK**	Not Detected
SNVs, short indels, Fusions & CNV	**BRAF**	Not Detected
SNVs, short indels, Fusions & CNV	**EGFR**	Not Detected
SNVs, short indels, Fusions & CNV	**ERBB2**	Not Detected
SNVs, short indels, Fusions & CNV	**FGFR2 & FGFR3**	Not Detected
SNVs and short indels	**HRAS**	Not Detected
SNVs, short indels & CNV	**KIT**	Not Detected
SNVs, short indels & CNV	**KRAS**	Not Detected
SNVs and short indels	**MAP2K1 & MAP2K2**	Not Detected
SNVs, short indels, Fusions & CNV	**MET**	Not Detected
SNVs and short indels	**MTOR**	Not Detected
SNVs and short indels	**NRAS**	Not Detected
SNVs, short indels, Fusions & CNV	**PDGFRA**	Not Detected
SNVs, short indels & CNV	**PIK3CA**	Not Detected
SNVs, short indels & Fusions	**RET**	Not Detected
SNVs, short indels & Fusions	**ROS1**	Not Detected
Fusions	**METex14 skipping mutation**	Not Detected
Fusions	**NTRK (1,2,3)**	Not Detected
Copy number variation (CNV)	**MYC**	Not Detected

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AU-ID-2010258

Aditi Aggarwal, Molecular Scientist

Dr. Shivani Sharma
DCP, DNB, DipRCPath. Reg. No. 1906

Dr. Rahul Katara, Ph.D.

CÖRE DIAGNOSTICS™

pulmo CÖRE 20 Gene Panel

Case Number: **24010010573** Patient Name: **S. J. S. L. Samarasinghe** Ordering Physician Name: **Dr. Mahendra Perera**

METHODOLOGY

pulmoCORE 20 gene panel is a multibiomarker NGS assay that enables detection of variants in 20 genes relevant to Non Small Cell Lung Cancer (NSCLC). The test utilizes AmpliSeq technology based NGS assay. DNA and mRNA from FPPE samples were extracted and subjected to Next Generation Sequencing (NGS) using the Ion S5 System. High quality nucleic acids that passed QC checks were subjected to library preparation and analysed for relevant genomic alterations in both the DNA and RNA to simultaneously detect multiple types of variants, including hotspots, single-nucleotide variants (SNVs), indels, copy number variants (CNVs), and gene fusions as mentioned below:

Hotspot genes (SNVs and short indels): ALK, BRAF, EGFR, ERBB2, FGFR2, FGFR3, HRAS, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, PDGFRA, PIK3CA, RET and ROS1.

Gene Fusions: ALK, BRAF, EGFR, ERBB2, FGFR2, FGFR3, MET, NTRK (1,2,3), PDGFRA, RET and ROS1

Copy number variation (CNV) genes: ALK, BRAF, EGFR, ERBB2, FGFR2, FGFR3, KIT, KRAS, MET, PDGFRA, PIK3CA and MYC

Sequencing was performed to achieve a minimum 500x depth of coverage. The output sequences were aligned to the human reference genome hg19 (GRCh37). Raw data analysis, alignments, and variant calling was done using the Torrent Variant Caller. High quality sequencing data was then analysed using the optimized ION Torrent Suite and the ION Reporter software to accurately detect rare somatic variants. The hotspots, indels and fusions were analyzed with the help of the ION Reporter Software and variants were annotated according to the American College of Medical Genetics (ACMG) and AMP guidelines. NCBI dbSNP, Catalogue of Somatic Mutations in Cancer (COSMIC), The Exome Aggregation Consortium and ClinVar as well as protein function prediction and classification algorithms FATTHM, SIFT, PolyPhen-2 and Grantham score were used for variant annotation.

For the index patient, on target was 97.79% with a 98.20% (76.79%) target base coverage at 100X (500X).

VARIANTS OF UNKNOWN FUNCTIONAL/THERAPEUTIC SIGNIFICANCE (VUS)

Gene (Exon) [Transcript]	Variant	VAF
-	-	-

LIMITATIONS AND DISCLAIMER

Despite all precautions taken, the error (administrative and technical) associated with these types of molecular diagnostic tests can be as high as 1% to 2%. Rare polymorphisms may be present that could lead to false negative or false positive results. The quality of sequencing and coverage varies between regions; many factors such as homopolymers, GC-rich regions etc. influence the quality of sequencing and coverage. This may result in an occasional error in sequence reads or lack of detection of a particular genetic variant. Furthermore, A negative (wild type) result does not rule out the presence of a mutation or rearrangement resulting in targeted fusion, that may be present but below the limits of detection of this assay. Variants that have not been confirmed by an independent analysis could represent technical artifacts. Not all variants detected may be listed in the report. Inclusion of variants is dependent upon our assessment of their clinical significance. Additionally, the presence of a mutation may not be predictive of response to therapy in all patients. The selection of any potential treatment/course of action based on this report rests solely within the decision and judgment of the treating physician and patient. Decisions on patient care should be based on the independent medical judgment of the treating physician based upon all available clinical information, according to the applicable standard of care and should not be based solely on the tests and information contained in this report. Accurate interpretation of this report is dependent on provided detailed clinical history of the patient. In the event of unavailability of detailed clinical history, the lab cannot guarantee the accuracy of the interpretation. The results and interpretation are based on current knowledge and might change in the future. Some findings listed in this report may be based on pre-clinical studies or studies not in the given patient's tumor type.

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Dr. Rahul Katara, Ph.D.

Scan to Connect

If you have any questions about this report or would like to have a conversation about the test results, please feel free to reach out to us at

+91 88828 99999 or **info@corediagnostics.in**

CONDITIONS OF REPORTING

1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient named or identified in the bill/test request form.
2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory.
3. The reported results are for information and are subject to confirmation and interpretation by the referring doctor.
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