UKAS MEDICAL 9059

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NGS BASED SOMATIC GENE FUSION ANALYSIS RESULTS

Patient Details		Source Information		Sample Information	
Lab Number:	MP20-XXXX	Requester Ref:		Date Received:	01/06/2020
Surname:	Atient	Surgical No.:	20S0001 A1	Primary Tumour Site:	Lung
Forename:	Patricia	Sample Format:	FFPE Block	Tumour Subtype:	Adenocarcinoma
D.O.B. (D/M/Y):	01/01/2001	Consultant:	Consultant	Tissue Sample Site:	Lung
Gender:	Female	Hospital:	Random DGH	% Tumour (Whole):	51-75%

Fusions assessed: MGP-4 (RNA)

IVD: ALK, BRAF, EGFR (incl. exon 2-7 skipping), MET (incl. exon 14 skipping), NTRK1, NTRK3, RET, & ROS1

RUO: For additional RUO fusions see sarahcannon-md.co.uk.

IVD – Validated and/or certified for clinical diagnostic use, **RUO** – Assessed but not clinically validated, for Research Use Only.
Fusion – An intra- or inter-genic rearrangement (caused by mutations, translocations, interstitial deletions, or chromosomal inversions)
A full description of the analytical scope and methodology utilised is detailed in the footer of this document.

Results

Fusion Detected

Gene 1	Exon 1		Gene 2	Exon 2
MET	13	⇝	MET	15

Summary Comments

A **MET** exon 14 skipping transcript sequence was detected in this sample. This has been associated with increased sensitivity to selected MET inhibitors (e.g. capmatinib) in non-small cell lung cancer.

Report Signed by / Date:

Dr F. Irst
Clinical Scientist
08/06/2020

Report Checked by / Date:

Dr S. Econd
Clinical Scientist
08/06/2020

Sample QC:

Expression Control Gene Performance (4 out of 5 passes required for valid result):

HMBS Pass **ITGB7** Pass **LRP1** Pass **MYC** Pass **TBP** Pass

Sample Performance (>1,200 Expression Control Reads AND >10,000 Total Reads required for valid result):

Expression Control Reads 16,765 (Pass) **Total Reads** 81,246 (Pass)

Analytical Scope & Methodology:

Assay Details: Selected regions from a commercially available multi-gene RNA panel (Life Technologies Oncomine Focus Panel: see below) are reverse transcribed and amplified using a highly multiplex Polymerase Chain Reaction approach. These are labelled using 'DNA barcodes' unique to each specimen and then collectively sequenced on a Life Technologies PGM or S5 instrument using Ion Torrent Hi-Q™ View chemistry and a 318v2 or Ion 520 chip respectively. Data is analysed using Torrent suite v5.10.2 and an in-house developed pipeline detects fusions by identification of sequences corresponding to transcripts with specific fusion partners, results are then reviewed manually. **Limitations**: Whilst every precaution has been taken to ensure this assay is as sensitive as possible, it has been optimised with total RNA input of not less than 10ng. Fusions with unlisted* gene partners cannot be detected. Furthermore, at least 5% of nucleated cells in the sample must comprise tumour. Below either of these cut off levels, fusions may not be consistently identified. **RUO/IVD status**: This assay has been validated for in-vitro diagnostic use (IVD) for the detection of gene rearrangement events (gene fusions) involving ALK, BRAF, EGFR, MET, NTRK1, NTRK3, RET and ROS1 only. Due to their rarity, and hence the lack of extensive material suitable for comprehensive validation and ongoing IQC, IQA & EQA, all other gene fusions are currently designated as clinical research use only (RUO). Any findings relating to these RUO fusions should not be used (without independent confirmation) to aid decisions regarding therapeutic patient management. **Interpretation**: This report primarily details laboratory test findings. Only limited interpretive comments are provided. Any fusions of unknown or unclear significance should be discussed in an appropriate MDT or other suitable forum. **Risk of false negative findings:** For validated fusions the likelihood of false negative findings arising from assessment/analytical errors is estimated at 1-2%, for novel fusions the risk of false negative findings is not readily assessable.
*A full list of the fusion partners assessed by this assay is available at sarahcannon-md.co.uk