

Sample: 16K1040 Name: Mylonas,Joan DOB: 21-Aug-1944 URN: 13/11/02410

LUNG CANCER MUTATION ANALYSIS DRAFT

SPECIMEN

extref

PATHOLOGY

Histological typing:

The sample was reviewed by a pathologist and was considered to have% tumour cells within the area selected for analysis. Please note: This is not a formal pathology review and is based solely on an H&E of the tissue provided and not on ancillary clinical or pathology information that may be available elsewhere.

RESULT

FAILED SAMPLE

TEST DESCRIPTION

Tumour DNA was tested in duplicate for mutations in exons 19 to 21 of the EGFR gene, exons 2 to 4 of the KRAS gene, and exon 15 of the BRAF gene using massively parallel sequencing. This test detects single nucleotide variants and indels in the target exons only. At 1000x coverage, the limit of detection of this assay has been determined to be X%. At 500x coverage the limit of detection has been determined to be X%. The sample was sequenced to an average 2017 aligned reads per amplicon with 66.67% uniformity. Regions with less than 100x coverage have not been analysed. These are listed below.

INTERPRETATION

This sample failed to meet predetermined measures of quality/quantity for this assay. DNA has been scheduled for re-analysis by an alternative method. These results will follow.

COMMENTS

Mutations in the kinase domain of the epidermal growth factor receptor (EGFR) gene result in constitutive signalling leading to tumour development. Kinase domain mutations occur in approximately 10% of non-South East Asian and 35% of South East Asian NSCLC patients, the majority of which display a dramatic response to EGFR kinase domain inhibitors (1). Confirmation of EGFR mutation status is required before administering kinase domain inhibitors such as gefitinib (Iressa) and erlotinib (Tarceva).

Activating KRAS mutations occur in up to 40% of NSCLC and occur most frequently at codons 12, 13 and 61. KRAS mutations cause constitutive activation resulting in a continual proliferative signal downstream of EGFR. KRAS mutant NSCLC is insensitive to targeted EGFR inhibitors (2). In a retrospective series of 1,046 NSCLC patients, the BRAF V600E mutation was associated with shorter disease free survival (3).

Note: Testing of tissue treated with chemo and/or radiotherapy reduces the cellularity of the neoplastic element and reduces the sensitivity of the assay. Where possible tissue derived from untreated tumour should be tested.

REFERENCES

1. Mok, T.S., et al., N Engl J Med, 2009. 361(10): p. 947-57.
2. Sun, J.M., et al., PLoS One, 2013. 8(5): p. e64816.
3. Marchetti, A., et al., Journal of Clinical Oncology, 2011. 29(26): p. 3574-3579.

Low coverage amplicons:

There were 39 low read amplicons with <100 aligned reads:
not listed

Evaluation Only. Created with Aspose.Words. Copyright 2003-2011 Aspose Pty Ltd.

To: PETER MAC CANCER CENTRE
ST ANDREWS PL
EAST MELBOURNE
VIC 3002

Patient: Mylonas,Joan
URN: 13/11/02410
DOB: 21-Aug-1944
SEX: F
Location: X

Sample: 16K1040
Ext Ref:
Collected: 23-Apr-2013
Received: 15-Mar-2016
Specimen:
Block ID:

Requester: JENNENS,ROSS RALPH

ACTIONABLE CANCER PANEL DRAFT

Clinical Details

Histological typing:

The sample was reviewed by a pathologist and was considered to have% tumour cells within the area selected for analysis. Please note: This is not a formal pathology review and is based solely on an H&E of the tissue provided and not on ancillary clinical or pathology information that may be available elsewhere.

Results

FAILED SAMPLE

Interpretation

This sample failed to meet predetermined measures of quality/quantity for this assay. DNA has been scheduled for re-analysis by an alternative method. These results will follow.

Methods

Tumour DNA was tested in duplicate for mutations in targeted exons of the following genes using massively parallel sequencing: AKT1, ALK, BRAF, CDKN2A, EGFR, FGFR1, FGFR2, FGFR3, ERBB2, KIT, KRAS, NRAS, PDGFRA, PIK3CA, PTEN and TP53. This test detects single nucleotide variants and indels in the target exons only. At 1000x coverage, the limit of detection of this assay has been determined to be X%. At 500x coverage the limit of detection has been determined to be X%. The sample was sequenced to an average 2017 aligned reads per amplicon with 66.67 % uniformity. Regions with less than 100x coverage have not been analysed. These are listed below.

Comments

Processed DNA is assayed for concentration, purity and fragmentation status prior to massively parallel sequencing. Samples determined to be not capable of generating a representative amplicon library may be analysed by alternate methods.

Testing of tissue treated with chemo and/or radiotherapy reduces the cellularity of the neoplastic element and reduces the sensitivity of the assay. Where possible tissue derived from untreated tumour should be tested.

Please contact the laboratory on 03 9656 3595 if you wish to discuss this report further.

Reported by: Dr A Fellowes, Scientist in Charge Molecular Pathology Diagnostic Development
Authorised by: Prof Stephen Fox, Director of Pathology
Reported: 31-Aug-2015 9:54 AM

Low quality amplicons:

There were 39 low read amplicons with <100 aligned reads:
not listed

References: