





To: PETER MAC CANCER CENTRE

ST ANDREWS PL EAST MELBOURNE

VIC 3002

Patient: SPENCER, STUART

URN: T-30442

DOB: 29-Jul-1982 SEX: M

SEX: M Location: X

Requester: SAMBROOK, JOE

Sample: 15K3182

Fxt Ref:

Collected: 14-Aug-2015 Received: 14-Aug-2015

Specimen:

Block ID:

TRUSEQ AMPLICON CANCER PANEL REPORT DRAFT

Clinical Details

XXX, approximately XX% tumour cell content, ? mutation profile for personalized medicine

Results

FAILED SAMPLE.

Interpretation

TruSeq Amplicon testing of a repeat sample is required. Avoid blocks >12 months post-fixation, containing <5mm³ tissue, and with low cellularity.

Method

Tumour DNA is analysed using the Illumina TruSeq Custom Amplicon Cancer Panel, which targets the mutation hotspots of 48 cancer genes in 212 targeted amplicons. Samples are uniquely indexed, pooled and sequenced on the Illumina MiSeq using MiSeq v2 chemistry at 2x151bp reads. Alignment, variant calling and annotation are performed using Peter Mac's amplicon-optimised pipeline v1.0. Only plausible pathogenic variants passing multiple functional and quality filters are reported above. Amplicons with less than 100 aligned reads are not analysed. These are listed below.

This mutation panel is designed to detect single nucleotide variants and indels in the target regions only. Mutations in the 48 cancer genes that lie outside the target regions will not be detected. The technology employed here is not suitable for detecting loss of heterozygosity, copy number variations, gross structural rearrangements, or aneuploidies. At 1000x coverage, this assay has a detection limit of approximately 5%. The variants detected by this assay should be confirmed by a second method before being used to guide clinical decisions.

Comments

DNA extraction of this tissue sample produced sufficient good quality material for TruSeq Amplicon testing. Sample processing passed all expected QC metrics and high quality sequence with high coverage (1667 mean aligned reads/amplicon) and uniformity (65.67 % amplicons >0.2 mean aligned reads) was obtained.

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

This test has not yet been fully validated to the current NPAAC requirements for an in-house IVD and results should be interpreted accordingly. All findings should be confirmed by an independent clinical assay. For further information, please contact the laboratory.

Reported by: Dr A Fellowes, Scientist in Charge Molecular Pathology Diagnostic Development

Authorised by: Prof Stephen Fox, Director of Pathology

Reported: 31-Aug-2015 9:49 AM

Low quality amplicons:

There were 176 low read amplicons with <100 aligned reads:

not listed