ABN 42 100 504 883

NATA & RCPA ACCREDITED LABORATORY Number 2465

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

Patient: Hurley,Kevin
URN: 16/11/65856
DOB: 24-Jul-1933
SEX: M
Location: MP

Requester: PRINCE,HENRY MILES

Sample: 16M1662
Ext Ref:
Collected: 07-Mar-2016
Received: 07-Mar-2016
Specimen:
Block ID:

NEXT GENERATION SEQUENCING REPORT DRAFT

Clinical indication –

Sample type –

Estimated tumour burden of sample –

ASSAY	26-GENE FULL MYELOID PANEL or MYELOPROLIFERATIVE DIAGNOSTIC PANEL
Genes	JAK2 (exon 12, 14(V617F)), MPL (exon 10), CALR (exon 9), c-KIT (exon 17), SF3B1 (exon 14,15,16), CSF3R (exon 14, 17), ASXL1 (exon 12 [*excluding c.1934 dupG; p.Gly646Trp fsX12*]) ASXL1 (exon 12 [*excluding c.1934 dupG; p.Gly646Trp fsX12*]), BRAF (exon 15), CALR (exon 9), CBL (exon 8, 9), CSF3R (exon 14, 17), DNMT3A (exon 23), EZH2 (exon 2-20), FLT3 (exon 14, 15, 20), GATA2 (exon 4, 5), IDH1 (exon 4), IDH2 (exon 4), JAK2 (exon 12, 14, 16), JAK3 (exon 13), KIT (exon 8, 10, 11, 17), KRAS (exon 2, 3, 4), MPL (exon 10), NPM1 (exon 11), NRAS (exon 2, 3, 4), RUNX1 (exon 4-9), SETBP1 (exon 4), SF3B1 (exon 14,15,16), SRSF2 (exon 1), TET2 (exon 2-11), TP53 (exon 2-11), U2AF1 (exon 2, 6), WT1 (exon 7, 8, 9)

MUTATIONS DETECTED	
Gene	**Mutation**
ASXL1	NM_015338.5, c.2077C>T, NP_056153.2:p.(Arg693*)
U2AF1	NM_006758.2, c.470A>C, NP_006749.1:p.(Gln157Pro)

SUMMARY

Individual Variant Analysis

ASXL1: CurVariant chr20:g.31022592C>T not yet curated.

U2AF1: A Gln157Pro missense mutation was detected in U2AF1. The Gln157Pro occurs in the zinc finger domain of the U2 small nuclear RNA auxiliary factor 1 (www.uniprot.org).

Method

DNA is analysed using a custom-designed myeloid amplicon gene panel (Myeloid v5.4). 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 an amplicon-optimised pipeline. Only plausible pathogenic variants passing multiple functional and quality filters and that are present in the "*Genes analysed*" list above are reported. Amplicons with less than 100 aligned reads are not analysed. 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%. For the variants JAK2 V617F and c-KIT D816V the sensitivity of the assay is approximately 1%. Please note, systemic mastocytosis and related disorders frequently have allelic burdens of <1% and therefore will not be reliably detected with this assay.

DNA extraction produced sufficient good quality material for myeloid amplicon gene panel testing. Sample processing passed all expected QC metrics and high quality sequence with high coverage (1568 mean aligned reads/amplicon) and uniformity (98.61 % amplicons >0.2 mean aligned reads) was obtained.

Please contact the laboratory on 03 9656 5297 if you wish to discuss this report further.

Reported by: **Dr. Piers Blombery (Consultant Haematologist)**
Authorised by: **Ms. Michelle McBean**

Reported: **19-Dec-2015 11:37 AM**

Low quality amplicons:

There were 2 low read amplicons with <100 aligned reads:
TET2_EX3_1_1 (reads 1)
JAK3_Ex13 (reads 81)