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| **Patient** **PATIENT\_IN**  **URN** URN\_IN  **DOB** DOB\_IN  **Sex** SEX\_IN | **Lab No** LAB\_NO\_IN  **Ext Ref** EXT\_REF\_IN  **Collected** COLLECTED\_IN  **Received** RECEIVED\_IN  **Specimen** SPECIMEN\_IN | **Requester** REQUESTER\_IN  **Referral Lab** REFERRAL\_LAB\_IN |

**COMMENT\_IN**

**Clinical Indication** CLINICAL\_INDICATION\_IN

**Specimen Details** SPECIMEN\_DETAILS\_IN

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| **HAEMATOLOGICAL MALIGNANCY GENE PANEL REPORT** |

**Test Description** Somatic variant analysis of 79 genes with clinical significance in haematological malignancy. Refer to Panel Summary for gene list.

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| **Result Summary Failed assay due to suboptimal DNA quantity/quality.** RESULTS\_SUMMARY\_IN See comment below regarding the test limitations of cell free DNA analysis. |

**Test Methodology**

DNA is analysed by targeted gene sequencing of coding regions and flanking splice sites (within 2 bp) of the genes listed below. Libraries are prepared using a custom Twist Bioscience target enrichment panel (Peter MacCallum Cancer Centre AllHaem DNA Twist v1, design ID TE-98899881) and sequenced on an Illumina NovaSeq 6000 with 150 bp paired end reads. A custom Seqliner/Nextflow-based analysis pipeline is used to generate aligned reads and call variants (single nucleotide variants and short insertions or deletions) against the hg19 human reference genome. Variants are analysed using PathOS software (Peter Mac).

**Test Limitations**

Please note the quantity of tumour derived cell free DNA within this sample is unknown. In addition, the profile of mutations present within the cell free DNA compartment may differ from that in any given individually assessed tumour sample. Therefore a negative result, or the absence of detection of a particular mutation does not imply its absence from all tumour sites in the patient (Blombery et al. 2018, PMID: 28880377). In addition, the sensitivity of this assay for variant detection is not appropriate for monitoring or minimal residual detection but rather for determining mutation profile at diagnosis/relapse.

**Panel Summary**

\* Please note FLT3-ITDs are not reliably detected with this assay

Please contact the laboratory on 03 8559 7284 if you wish to discuss this report further.

**Reported by REPORTED\_BY\_IN**

**Authorised by AUTHORISED\_BY\_IN**

**Reported 16-Sep-2024**