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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** ?Germline vs somatic origin of previously detected GENE\_IN variant.

**Correlative Morphology** CORRELATIVE\_MORPHOLOGY\_IN

**Specimen Details** SPECIMEN\_DETAILS\_IN

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| **GERMLINE VARIANT ANALYSIS REPORT** |

**Test Description** Germline variant analysis of GENE\_IN. Refer to Panel Summary for targeted region.

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| **Result Summary Failed assay due to suboptimal DNA quantity/quality** |

**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).

**Panel Summary**

Gene coverage in this sample is as follows

Please note variants may not be optimally detected in genes with less than 100% coverage. The gene coverage above is considered acceptable given the available information about the clinical context, however please contact the laboratory for further advice should specific genes covered at less than 100% require full coverage. A list of regions with suboptimal coverage is available upon request.

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**