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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

**Correlative Morphology** CORRELATIVE\_MORPHOLOGY\_IN

**Specimen Details** SPECIMEN\_DETAILS\_IN

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| **UBA1 EXON 3 VARIANT ANALYSIS REPORT** |

**Test Description** Somatic variant analysis of UBA1 exon 3.

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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 QIAGEN QIAseq single primer extension-based panel (Peter MacCallum Cancer Centre AllHaem v4) and sequenced on an Illumina NextSeq500 with 150 bp paired end reads. A customised CLC bioinformatics pipeline including QIAGEN CLC enterprise solutions 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**

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

**Reported by REPORTED\_BY1\_IN**

**REPORTED\_BY2\_IN**

**Authorised by AUTHORISED\_BY\_IN**

**Reported 25-Oct-2023**