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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 |

**Clinical Indication** CLINICAL\_INDICATION\_IN

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| **CEBPA GERMLINE VARIANT ANALYSIS REPORT** |

**Test Description** Germlinevariantanalysis of all coding regions of the CEBPA gene.

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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 v1) 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) and described according to HGVS nomenclature version 19.01 (http://varnomen.hgvs.org/) with minor differences in accordance with Peter MacCallum Cancer Centre Molecular Pathology departmental policy. **Germline variant analysis** – All rare germline variants in CEBPA are classified according to ACMG guidelines for the interpretation of sequence variants[1](#_ENREF_1) with class 3 (uncertain significance), class 4 (likely pathogenic) and class 5 (pathogenic) variants reported only.

**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 10-Jan-2023**