|  |  |  |
| --- | --- | --- |
| **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

|  |
| --- |
| **HAEMATOLOGICAL MALIGNANCY RNA FUSION PANEL REPORT** |

**Test Description** Identification of clinically significant fusion transcripts in haematological malignancy. Refer to Panel Summary for gene list.

|  |
| --- |
| **Result Summary FUSION TRANSCRIPT DETECTED. See Test Results table below for details.** RESULTS\_SUMMARY\_IN  **Clinical Interpretation** CLINICAL\_INTERPRETATION1\_IN  CLINICAL\_INTERPRETATION2\_IN  CLINICAL\_INTERPRETATION3\_IN |

**Test Results**

**Test Methodology**

RNA is analysed by targeted RNA fusion sequencing of the genes listed below. Libraries are prepared using a custom QIAGEN QIAseq RNA Fusion XP single primer extension-based panel (Peter MacCallum Cancer Centre AllHaem RNA v1) and sequenced on an Illumina NextSeq500. A customised CLC bioinformatics pipeline including QIAGEN CLC enterprise solutions is used to identify candidate fusion genes against the hg19 human reference genome. Please note candidate fusion genes may involve gene partners that are not targeted by this assay. Candidate fusion genes are analysed using PathOS software (Peter Mac). The following databases are commonly used in addition to literature review to assist gene fusion interpretation: the Catalogue of Somatic Mutations in Cancer (COSMIC; cancer.sanger.ac.uk), Mitelman (https://mitelmandatabase.isb-cgc.org), Quiver (https://quiver.archerdx.com), StJude PeCan (https://pecan.stjude.cloud), FusionGDB2 (https://compbio.uth.edu/FusionGDB2) and ChimerDB (https://www.kobic.re.kr/chimerdb). Gene fusions considered clinically significant or previously documented in haematological malignancy are reported. Novel fusions of unknown clinical significance are generally not reported. In addition, where multiple fusion transcripts are detected for a single gene fusion, only the major transcript (i.e. the transcript with the greatest number of supporting reads) is reported. Fusion transcripts arising from reciprocal gene fusions on the partner derivative chromosome are generally not reported in addition to the primary (i.e. clinically significant) gene fusion. **Gene fusion categorisation –** fusions are curated and categorised according to the clinical context of the patient and categorised as **DIAGNOSTIC** (the variant either defines a diagnostic category or is sufficiently specific for the clinical context to contribute to diagnostic subcategorisation), **PROGNOSTIC** (the variant has been associated in large trials/series with inferior or superior outcomes in either the context of a specific therapy or independent of therapy.), **DRUG TARGET** (the variant or variant class is specifically targeted by a therapeutic agent, this category only includes therapeutic agents that are clinically advanced and generally available through either reimbursement or clinical trials [i.e. not early stage investigational agents]), **MRD MARKER** (the variant is an established biomarker for which assessment at MRD sensitivity after therapy is accepted practice). If the variant is not categorised into any of the above categories it is assigned **CLONAL MARKER** indicating its utility in defining the presence of a clonal haematopoietic process in the specimen. These categorisations are general in nature and may not be applicable to the specific clinicopathological context of the patient.

**Test Limitations**

The detection limit of this assay is approximately 5%-10% of ABL1 equivalent. This assay is qualitative and is not suitable for the monitoring of measurable residual disease (MRD). Complex gene fusions involving novel boundaries, intronic sequences, or gene rearrangements not resulting in the expression of an abnormal transcript may not be reliably detected by this assay or accurately described. Gene expression and structural variants such as tandem duplications are not assessed. Gene fusions are analysed using the reference transcripts listed below unless otherwise stated. The performance of sample types other than peripheral blood, bone marrow and FFPE tissue have not been validated for testing using this assay. Please note Peter Mac assumes sample identification and clinical diagnoses are as stated on the request. Our clinical recommendations may be based on evidence from third-party data sources and should be interpreted in the context of all other clinical and laboratory information for this patient.

**Panel Summary**

Selected regions of the following genes are targeted by this assay.

| Gene | Preferred Transcript | Gene | Preferred Transcript | Gene | Preferred Transcript | Gene | Preferred Transcript | Gene | Preferred Transcript | Gene | Preferred Transcript |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| ABL1 | NM\_005157.5 | CRLF2 | NM\_022148.3 | GLIS2 | NM\_032575.2 | MLLT3 | NM\_004529.3 | NUTM1 | NM\_175741.2 | RBM15 | NM\_022768.4 |
| ABL2 | NM\_007314.3 | CSF1R | NM\_005211.3 | HLF | NM\_002126.4 | MNX1 | NM\_005515.3 | PAX5 | NM\_016734.2 | RET | NM\_020975.4 |
| AFDN | NM\_001040000.2 | DEK | NM\_003472.3 | IL2RB | NM\_000878.4 | MRTFA | NM\_020831.4 | PBX1 | NM\_002585.3 | RUNX1 | NM\_001754.4 |
| AFF1 | NM\_001166693 | ELL | NM\_006532.3 | JAK2 | NM\_004972.3 | MYB | NM\_001130173.1 | PCM1 | NM\_006197.3 | RUNX1T1 | NM\_175635.2 |
| ALK | NM\_004304.4 | EPOR | NM\_000121.3 | KAT6A | NM\_006766.4 | MYC | NM\_002467.4 | PDCD1LG2 | NM\_025239.3 | TCF3 | NM\_003200.3 |
| BCL11B | NM\_138576.3 | ERG | NM\_001136154.1 | KMT2A | NM\_001197104.1 | MYH11 | NM\_001040113.1 | PDGFRA | NM\_006206.5 | TSLP | NM\_033035.4 |
| BCR | NM\_004327.3 | ETV6 | NM\_001987.4 | MECOM | NM\_004991.3 | NPM1 | NM\_002520.6 | PDGFRB | NM\_002609.3 | TYK2 | NM\_003331.4 |
| BRAF | NM\_004333.4 | FGFR1 | NM\_023110.2 | MEF2D | NM\_005920.3 | NTRK1 | NM\_002529.3 | PICALM | NM\_007166.3 | UBTF | NM\_014233.3 |
| CBFA2T3 | NM\_005187.5 | FGFR3 | NM\_000142.4 | MLF1 | NM\_022443.4 | NTRK2 | NM\_006180.4 | PML | NM\_033238.2 | USP2 | NM\_004205.4 |
| CBFB | NM\_022845.2 | FIP1L1 | NM\_030917.3 | MLLT1 | NM\_005934.3 | NTRK3 | NM\_001012338.2 | PTK2B | NM\_004103.4 | ZMYM2 | NM\_003453.4 |
| CPSF6 | NM\_007007.2 | FLT3 | NM\_004119.2 | MLLT10 | NM\_001195626.1 | NUP214 | NM\_005085.3 | RARA | NM\_000964.3 | ZNF384 | NM\_133476.4 |
| CREBBP | NM\_004380.2 | FUS | NM\_004960.3 | MLLT11 | NM\_006818.3 | NUP98 | NM\_016320.4 | RARG | NM\_000966.5 |  |  |

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 11-Jun-2024**

**CLINICAL\_CONTEXT\_IN**