Next Generation Sequencing for Myeloid Malignancies

Background

The detection of genetic variants identified in the DNA from patients with myeloid malignancies contribute to the diagnosis, prognosis and treatment of patients with AML, MDS, MPN and CMML. The use of Next Generation Sequencing (NGS) enables the sequencing of multiple genes and therefore the detection of clinically relevant variants in a single test in a cost-efficient manner.

The All Wales Genetics Laboratory (AWGL) has validated the Illumina TruSight Myeloid Sequencing panel which uses NGS technology to sequence key genomic regions in 27 genes (gene panel content is detailed below).

The panel has a target read depth of 500x and can therefore detect DNA variants (single nucleotide variants and indels) at a variant allele frequency of 5%. The clinical significance of detected variants is assessed and reported based upon the Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer (Li et al., JMD 2017, 19(1):4-23).

Gene Panel Content

The panel is composed of 27 genes: ASXL1, BCOR, CALR, CEBPA, CBL, DNMT3A, EZH2, ETV6, FLT3, GATA2, IDH1, IDH2, JAK2, KRAS, KIT, MPL, NRAS, NPM1, NOTCH1, PDGFRA, RUNX1, SF3B1, SRSF2, SETBP1, TET2, TP53 and U2AF1.

The whole coding region is sequenced for DNMT3A, EZH2, ETV6, TP53 and RUNX1. For the remaining genes, only oncogenic hotspots within the gene are sequenced.

Referral Criteria

The panel is designed for use in patients with a **confirmed** diagnosis of:

- Acute myeloid leukaemia
- Myelodysplasia
- Myeloproliferative neoplasm without a mutation in JAK2, CALR or MPL:
  - Chronic neutrophillic leukaemia
  - Chronic myeloproliferative disease unclassifiable
  - Polycythaemia vera

Essential thrombocythaemia
Chronic idiopathic myelofibrosis
- Chronic myelomonocytic leukaemia
- Juvenile chronic myelomonocytic leukaemia

This test is not suitable for referrals for mastocytosis

Referrals which do not meet this clinical criteria will not be processed. **This test is not suitable for minimal residual disease monitoring and testing will not be initiated.** DNA will be extracted and banked.

Please note this test is not designed for the investigation of suspected germline disorders. If germline testing is required please contact the laboratory. Due to the gene content of this panel, however, findings may be detected that are potentially germline in origin. Further analysis or subsequent testing, in order to confirm the nature of any finding, may be required to fully interpret such a result in the context of the referral.

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Prices* & Turnaround Times (TAT)
*Please contact the laboratory for pricing information

Sample Requirements
Please send 2ml of bone marrow or peripheral blood in an EDTA tube.
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request is received

TAT (Calendar days)
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Contact Details

All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW

Tel: 029 2074 2641
Fax: 029 2074 4043

Haematology.Genetics.cav@wales.nhs.uk.
lab.genetics@wales.nhs.uk
www.wales.nhs.uk/AWMGS/
Accredited to ISO 15189:2012 (8988)