Bristol Genetics Laboratory is a UKAS accredited medical laboratory No.9307.

# Genomic testing in Acute Myeloid Leukaemia (AML)

North Bristol NHS Trust

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**Sample Required:**
**See Sample requirements page at www.nbt.nhs.uk/genetics for full details**

Bone marrow – in EDTA, Li Hep or heparinised bone marrow culture media (available from lab)

Blood 2-10 mls in EDTA or Li Hep

Samples should be accompanied by a FULLY completed request form (available as download at www.nbt.nhs.uk/genetics or from the laboratory).

Please include details of the test required, family history, address and POSTCODE, NHS number, referring clinician and centre.

**Consent and Storage:**
All genetic testing requires consent. **It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.**

DNA is stored from **ALL** patients undergoing DNA testing, unless consent for this is specifically denied.

Stored material from all referrals may be retained for quality assurance purposes and may be used anonymously for the development of new tests for the disorder in question.

## Clinical Background and Genetics

- Acute myeloid leukaemia (AML) is the most common acute leukaemia in adults and is an aggressive disease, although outcomes are more favourable in younger patients or within certain subtypes of AML
- Genomic testing has long played a role in the diagnosis of acute leukaemia and indeed recently recurrent genomic findings have begun to define the classification of AML[1]
- In addition to these characteristic, diagnostic entities a range of structural genomic changes have been used to develop prognostic as well as diagnostic frameworks in AML[2]
- As technologies develop recurrent molecular biomarkers have been added to the suite of tests that can provide diagnostic, prognostic and increasingly therapeutic opportunities to help with the management of AML[3]
- Moving forwards AML will be one of the initial cancers to access whole genome analysis as part of the NHS England Genomic Medicine Services

## Service Offered

- Genomic testing is delivered in line with the National Genomic Test Directory (NGTD) for Cancer
- The Bristol Genetics Laboratory (BGL) is part of the Bristol Haemato-oncology Diagnostic Service (BHODS) Specialist Integrated Haematological Malignancy Diagnostic Service (SIHMDS) and has access to a full range of complementary pathology services.

| NGTD code | Test | Turnaround time (days) |
|---|---|---|
| M80.7 | *PML-RARA* FISH/PCR | 1 |
| M80.5 | Core Binding Factor FISH/PCR | 3 |
| M80.18 | *NPM1* | 3 |
| M80.21 | *FLT3* tyrosine kinase domain (TKD) | 3 |
| M80.22 | *FLT3* internal tandem duplication (ITD) | 3 |
| M80.3 | AML karyotype | 7 |
| M80.2 | AML NGS panel | 14 |
| M80.1 | Whole Genome Analysis | 42 |

## Referrals

- Referrals are accepted from Consultant Haematologists and/or as part of agreed SIHMDS pathways

## Quality

- BGL participates in all UK NEQAS LI and GenQA external quality assurance programmes for AML

## Reference:

1. Swerdlow S.H. et al. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC; Lyon 2017
2. Grimwade D et al . Refinement of cytogenetic classification in acute myeloid leukemia: determination of prognostic significance of rare recurring chromosomal abnormalities amongst 5,876 younger adult patients treated in the UK Medical Research Council trials. Blood 2010.116:354-65
3. Döhner et al., Diagnosis and management of AML in adults: 2017 ELN recommendations from an international expert panel. Blood; 2017 129: 424-4474

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