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

Myeloproliferative neoplasia

North Bristol NHS Trust

Contact details

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Sample Required

3-5ml blood in EDTA

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

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

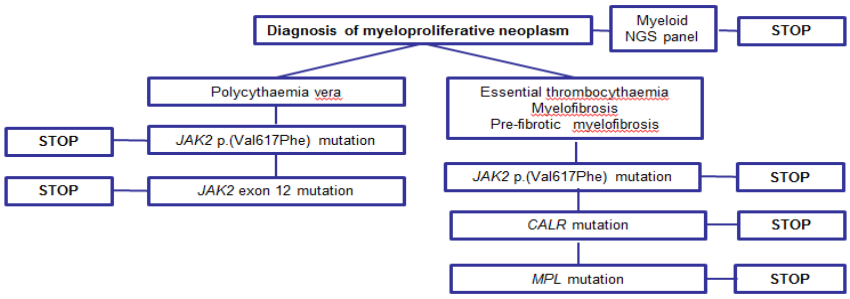
All genetic testing requires consent. **It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.**

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

Clinical Background and Genetics

- A significant proportion of cases of Myeloproliferative Neoplasia (MPN), (excluding CML), have no visible chromosome abnormality. The presence of acquired molecular mutations is now a well-established finding, providing additional diagnostic and prognostic information to patients with MPN.
- *JAK2* p.(Val617Phe) mutations (formerly V617F) have been described in approximately 98% of patients with Polycythaemia Vera (PV) and approximately 50% of patients with either Essential Thrombocythaemia (ET) or Idiopathic Myelofibrosis (IMF). Therefore, the detection of a mutation can distinguish between true MPN and a reactive disorder.
- Patients with PV that do not have a *JAK2* p.(Val617Phe) mutation may possess a mutation within exon 12 of the *JAK2* gene.
- 67% patients with *JAK2* p.(Val617Phe)-negative ET and 88% patients with *JAK2* p.(Val617Phe)-negative PMF have been found to possess mutations in exon 9 of the *CALR* gene.
- Approximately 5% *JAK2* p.(Val617Phe)-negative patients with ET and PMF possess an acquired mutation within exon 10 of the *MPL* gene.
- A wider range of genes have also been identified as having clinical significance in MPNs, which can be tested for by NGS
- The Bristol Genetics Laboratory (BGL) offers testing for the above mutations.



Service Offered

- The BGL is part of the Bristol Haemato-oncology Diagnostic Service (BHODS) and has access to a full range of complementary pathology services
- Testing for the *JAK2* p.(Val617Phe) mutation is carried out on genomic DNA by digital droplet PCR (ddPCR) which is routinely capable of detecting a mutant allele when present at levels of 1% or above.
- Testing for *JAK2* exon 12 and *CALR* exon 9 mutations is carried out on genomic DNA using multiplex and simplex PCRs followed by fragment analysis on the CEQ8000 with a sensitivity of approximately 4%
- Testing for mutations in exon 10 of the *MPL* gene is carried out on genomic DNA using a simplex PCR and bi-directional Sanger sequencing with a sensitivity of approximately 20%.
- BGL also offers targeted Myeloid Next Generation Sequencing (NGS) analysis for *JAK2, CALR* and *MPL* and an extended MPN NGS panel using an Illumina custom capture 54 gene panel.

Referrals

Referrals are accepted from Consultant Haematologists or Healthcare professionals that have taken advice from a Consultant Haematologist

Target reporting Times

14 calendar days

Quality

BGL participates in the GenQA programme for *JAK2* p.(Val617Phe) status and UK NEQAs LI Myeloproliferative Neoplasms Gene Panels (not accredited).

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