Belfast Health and Social Care Trust
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Scan this QR code to link to the genetic testing section of our website

Northern Ireland Regional Genetics Service

Genetic Testing for Non-Geneticists – Information and Tips Sheet

1. Is my patient suitable for genetic testing?

Use the National Genomic Test Directory to check (PDF document):

- If the patient meets testing criteria.
- If your specialty is authorised to request the test.

Refer to Clinical Genetics (REF1021) if:

- The patient meets criteria for multiple tests. A more extensive test may be appropriate (e.g. exome sequencing).
- You are unsure which test is most appropriate.
- A patient requires more careful counselling, whatever the reason.
- Predictive (asymptomatic) testing is typically requested by Clinical Genetics only.

Before requesting a test, consider:

- Will the result impact clinical management or family planning? Therapies for genetic disorders are rare.
- Does the patient want testing, and understand the implications?
- Is this the right time in the patient's care journey?
- Does the patient have any mental health concerns for which a genetic test result would impact?
- Will the result have implications for other family members, and how would this be communicated?

2. How do I request a genetic test?

- Search for the test on Epic using the R number (from the National Genomic Test Directory).
- You can follow the step-by-step guide on our website (or scan the QR code above).
- Include the National Genomic Test directory testing criteria. Requests without this will be rejected by the lab.
- Most tests use EDTA (purple top) sample.
- If unsure, request R346 DNA storage and activate testing later by emailing: GeneticsLabs@belfasttrust.hscni.net

3. Possible test outcomes

- **Normal result:** No findings. No genetic test can rule out an underlying genetic diagnosis.
- **Diagnostic result:** Confirms a genetic condition; may require referral to other specialties and/or Genetics.
- **Variant of Uncertain Significance (VUS):** A genetic change is found, but its significance is unclear. The vast majority of VUSs which are not diagnostic. If your patient has a clinically plausible VUS, please refer.
- **Incidental findings:** Unrelated but potentially important health information may be discovered. Some tests may reveal unexpected family relationships (e.g. non-paternity, consanguinity).
- **Need help interpreting results?** Feel free to refer to Clinical Genetics (REF1021).

Results are sent to the requesting physician and are available on Epic (media/lab folder).
To follow-up on outstanding or historical results, you can contact GeneticsLabs@belfasttrust.hscni.net.
Turnaround time of test varies (typically months). Information on in-house genetic test TATs can be found here.
Clinical Genetics team do not routinely review all reports. If a Clinical Genetics appointment is needed, please refer.

4. How to consent a patient for genetic testing.

Use SmartPhrase .GENETICSCONSENT to record the consent discussion in your Epic notes.
Consent discussion should cover:

- **Purpose and limitations:** Genetic testing can help explain medical conditions but may not detect all disorders.
- **Uncertainty:** Some results may be unclear, and classification of genetic variants may change over time as scientific understanding evolves.
- **Unexpected findings:** Testing may reveal unrelated health risks or unexpected family relationships (e.g. non-paternity, consanguinity).
- **Family implications:** Results may affect relatives and could inform their healthcare.
- **DNA storage:** DNA will be stored for future testing or quality assurance unless otherwise requested.
- **Data storage:** Genetic data will be securely stored and may be reanalysed to support ongoing care.
- **Data sharing:** Genomic data may be securely shared between labs under strict data protection protocols.
- **Health records:** Test results will be included in the patient's NHS medical record.
- **Partial consent:** Any limitations should be clearly documented in the Comments box.

If you prefer signed paper consent, use the genetics test request form or NHS England Record of Discussion.
Genetic Alliance UK provides information on Insurance and Genetic Testing.

Laboratory contact: GeneticsLabs@belfasttrust.hscni.net / 028 9504 7353
Clinical contact: genetic.medicine@belfasttrust.hscni.net / 028 9504 8022 / REF1021