Requesting genomic test

THE GENOMIC INSTITUTE

This checklist highlights the essential steps for requesting clinical diagnostic genomic testing for patients with a suspected genetic condition.

Genomic testing includes targeted gene panel sequencing, Whole Exome Sequencing (WES), and Whole Genome Sequencing (WGS).
It is applicable to monogenic (single gene) diseases which are genetically heterogeneous.

Pre-test

1 Check for previous genetic testing

- ☐ Review all patient URNs via Auscare/Auslab to clarify any previous or concurrent genetic testing.
- ☐ Clarify if any family member has had genetic testing, and the outcome.

2 Choose genomic test and gene panel(s)

- ☐ Select a preferred platform for genomic testing (i.e. targeted panel, WES or WGS).
- ☐ Review gene panels at https://bit.ly/genepanels.
- ☐ Select the correct gene panel(s) for analysis based on the patient's phenotype.
- ☐ Determine the desired turnaround time based on clinical urgency.
- ☐ Determine the cost of test based on test type, panel size, and desired turnaround time.

Urgency	Turnaround time	Cost
Routine	2-6 months	$
Rapid/semi-urgent	2-3 weeks	$$
Ultra-rapid	<5 days	$$$

3 Choose testing laboratory

- ☐ Identify an accredited laboratory which can deliver the required test.
- ☐ Clarify any mandatory requirements for the test from the laboratory. Requirements may include consent form, test requisition form, and specimen type.

4 Identify test funding source

- ☐ Clarify how the test will be funded.
  - For publicly funded, get approval from consultant or clinical director.
  - For Medicare funded, review eligibility criteria for testing at https://bit.ly/mbsOnline.
  - For patient self-funded, ensure patient is aware of the costs.

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5 Obtain patient consent

- ☐ Complete a clinical consent form for genomic testing. Download the form from https://bit.ly/geneticTesting.
- ☐ Give the patient a genomic testing fact sheet. Download the fact sheet from https://bit.ly/geneticTesting.
- ☐ Scan or file the signed consent form in the patient's medical records.

6 Complete pathology form

- ☐ Download the form from https://bit.ly/geneticTesting.
  - For publicly funded, use the Public pathology form.
  - For Medicare funded, use the Private pathology form.
- ☐ For accurate test processing and correct billing, provide the following information on the pathology form:
  - Clinical phenotype and relevant family history
  - Name of testing laboratory
  - Type of genomic test
  - Name of gene panel(s)
  - Primary consultant's name, department, and contact details

7 Complete laboratory test requisition form (if required)

- ☐ Complete your chosen laboratory's test requisition form, which may be paper or online.
- ☐ For accurate test processing and correct billing, provide the following information on the test requisition form:
  - Clinical phenotype
  - Relevant family history
  - Type of genomic test
  - Name of gene panel(s)
  - Primary consultant's name, department, and contact details
- ☐ For interstate/international testing, email Pathology Queensland at Genetic-Referrals@health.qld.gov.au with the following information:
  - Patient details
  - Name of testing laboratory
  - Type of genomic test and name of gene panel(s)
  - Laboratory test requisition form
  - Patient consent form for genomic testing

Post test

8 Prepare for test results

Disclosing the genomic test result is the responsibility of the requesting clinician/consultant.

- ☐ Ensure there is an agreed plan to disclose the test results with the patient.
- ☐ Ensure you can interpret the outcomes of genomic testing and the implications. Go to 'Interpreting results' for guidance.
- ☐ Consider a referral to Genetic Health Queensland to discuss and interpret genomic test results.

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For further specialist advice, email Renal Genetics MDT at Renal_Genetics_MDT@health.qld.gov.au