

Guideline

Subject: Requesting Medical Human Genetic Testing
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Review by: Joint Statement: RCPA Genetics Advisory Committee (GAC) and Human Genetics Society of Australasia (HGSA)
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This Guideline has been jointly prepared and endorsed by the RCPA Genetics Advisory Committee (GAC) and the Human Genetics Society of Australasia (HGSA).

For the purpose of this document, the term “medical practitioner” refers to an individual who has completed an approved medical degree and is registered with AHPRA as a medical practitioner.

Background:

Genetics is a rapidly evolving field characterised by significant complexity. Genetic test selection, result interpretation and clinical follow-up require specialised knowledge. Results have potentially significant implications for diagnosis, treatment, reproductive decision-making, long-term patient management and the health of family members. Careful consideration is required when determining appropriate requestors for specific categories of genetic testing.

Following the legislative changes allowing nurse practitioners and midwives to access certain Medicare Benefits Schedule services and prescribe Pharmaceutical Benefits Scheme medications[1], NPAAC has released an update to the Requirements for Medical Testing for Human Genetic Variation (Fourth Edition, 2026) [2]. The document was updated in response to “*new and specific MBS item numbers of a single genomic test (Non-invasive prenatal testing of blood from an RhD negative pregnant patient)*” [2], specifically Items 73420 and 73421, which can be requested by nurse practitioners and midwives.

Both the RCPA and HGSA support efforts to ensure equitable access to appropriate genetic testing services and welcome increasing awareness around the importance of genetic testing amongst a range of health professionals.

The RCPA and the HGSA emphasise the need for the highest standards of clinical governance and patient safety, to maintain best practice in the provision of genetic testing as a clinical service. NPAAC guidelines do not provide guidance related to requests from requesting practitioners or specify the scope of practice for practitioners when ordering genetic tests. It is the responsibility of laboratories to assess whether a requesting practitioner has appropriate scope of practice for a given test. Below, we issue guidance to pathology laboratories on accepting requests for genetic tests to ensure clinically safe and appropriate patient care.

Appropriate requesting

Genetic tests are generally requested by a medical practitioner, who is accountable for and can guide medical decision-making. Some pathology tests may be suitable for referral by a broader range of practitioners, including midwives and nurse practitioners (See RCPA Guideline “*Pathology test requesting by Nurse Practitioners and Midwives*” [3]). While the NPAAC standards outline the minimum requirements for medical genetic testing laboratories [2], we recommend that in almost all cases, genetic testing should be requested by a medical practitioner, or an appropriately qualified practitioner in conjunction with a medical practitioner. In complex testing scenarios, consultation with a health professional with specialist training in human genetics is advised. This approach ensures that patients receive appropriate pre-test counselling, informed consent and post-test clinical follow-up.

As a general guide, it is recommended that requests for genetic testing consider the following:

1) General referral requirements for genetic testing

To ensure appropriate clinical oversight, genetic testing requests should be made by:

- a medical practitioner, or
- an appropriately skilled healthcare practitioner who has been granted scope of practice to request specific MBS items, or
- an appropriately skilled healthcare professional (e.g. genetic counsellor, nurse practitioner, or midwife) with relevant scope of practice in consultation with a medical practitioner. In practice, this could be easily achieved by ensuring the medical practitioner is copied in on the request form and receives a copy of the report.

2) Genetic testing requiring medical specialist oversight

Due to the clinical implications and potential impact, genetic tests with a high likelihood of complex results, such as predictive (presymptomatic), prenatal and most diagnostic testing, should only be requested by:

- a medical specialist with relevant expertise (e.g. clinical geneticist, neurologist, oncologist, metabolic specialist, maternal fetal medicine specialist), or
- a medical specialist working with a genetic counsellor.

Commitment to safe and responsible genetic testing

The RCPA and HGSA remain committed to ensuring equitable access to high-quality genetic testing, while ensuring that testing is clinically safe and appropriate. Non-medical healthcare professionals play an essential role in patient care, particularly in primary care and antenatal settings, enabling access to health care.

Individual laboratories should consider developing their own internal policies regarding appropriate referrers for each test that is offered, considering the characteristics of the requested test, including associated counselling and clinical follow-up requirements. In drafting these policies, laboratories should bear in mind that many genetic tests are characterised by a high level of complexity with associated downstream implications. Therefore, the RCPA and the HGSA call for the continued involvement of medical

practitioners with relevant expertise for such tests. This will ensure best practice and highest standards of patient safety in the provision and use of genetic testing.

References:

1. Medicare Benefits Schedule services and prescribe Pharmaceutical Benefits Scheme medications without the need for a collaborative arrangement with a medical practitioner <https://www.health.gov.au/our-work/collaborative-arrangements>
2. Australian Commission on Safety and Quality in Health Care. NPAAC Requirements for Medical Testing for Human Genetic Variation. Fourth edition. Sydney: ACSQHC; 2026. www.safetyandquality.gov.au/sites/default/files/2026-03/requirements-medical-testing-human-genetic-variation-fourth-edition-2026.pdf
3. RCPA Guideline “*Pathology test requesting by Nurse Practitioners and Midwives*”, issued November 2025