australasian society of clinical immunology and allergy

10 March 2025

Health Technology Assessment Team (HTA Team)
Medical Services Advisory Committee
Email: hta@health.gov.au

To whom it may concern,

Re: RCPA application for genomic testing for the diagnosis of inborn errors of immunity (including primary immunodeficiency diseases)

On behalf of the Australasian Society of Clinical Immunology and Allergy (ASCIA) we are writing in support of the application by the Royal College of Pathologists of Australasia (RCPA) for generation of an MBS item number for genomic testing for the diagnosis of **inborn errors of immunity (IEI) which includes primary immunodeficiency diseases (PID).**

These are a group of more than 520 serious, chronic medical conditions, caused by defects in genes that control the immune system. They can lead to frequent or severe infections, other chronic immune system disorders, including autoimmune problems, and a predisposition to cancer.

ASCIA supports this application for a Medicare Benefits Schedule (MBS) item number for genomic testing to diagnose IEI including PID for the following reasons:

- One of the main goals of the **ASCIA Immunodeficiency Strategy for Australia and New Zealand** (the Strategy) is to improve access to expert genetic diagnosis by using genomic and immunological testing for patients with a suspected or recently diagnosed primary immunodeficiency (PID), or people at a high risk based upon their family history.
- As stated in the Strategy;
  - **Genomic testing is required to make a definitive genetic diagnosis** for an increasing number of IEI/PID.
  - **Genomic testing enables targeted therapies and counselling** about outcomes based on what is known about that gene, and informed reproductive/family planning decisions.
  - **Genomic testing for genetic diagnosis of IEI/PID is currently unfunded and expensive**, often sent to overseas laboratories at considerable cost, or performed in research laboratories, that may not be accredited by National Association of Testing Authority (NATA).
  - **Access to testing, funded or otherwise, varies considerably across regions**, centres and in private versus public systems. There are well established collaborations across Australia that are attempting to provide services to meet these complex needs, but without adequate or ongoing resourcing.
  - **Equitable access to funded and accredited genomic testing for IEI/PID requires funding of diagnostic genomic testing performed by accredited diagnostic laboratories**, including

the introduction of an MBS item number in Australia similar to those introduced in 2020 for identification of childhood dysmorphology syndromes and intellectual disability.

- As part of the Strategy, ASCIA has developed a **Clinical Care Standard for Inborn Errors of Immunity (Primary Immunodeficiencies)** which states;
  - People with IEI/PID should have equitable and timely access to appropriate, funded and accredited genomic and functional immunological testing.
  - This requires funded diagnostic genomic testing performed by accredited diagnostic laboratories.
  - Genomic testing should only be ordered by health professionals with appropriate knowledge and expertise, supported by genomic medicine and genetic counselling expertise.

Further information is available on the **ASCIA website**

Please email jill@allergy.org.au if you require additional information.

Yours sincerely,

Dr Michael O'Sullivan — ASCIA President

Jill Smith — ASCIA CEO