

**Submission
No 10**

**IMPROVING ACCESS TO EARLY CHILDHOOD HEALTH AND DEVELOPMENT
CHECKS**

Organisation: Human Genetics Society of Australasia

Date Received: 21 February 2024



Human Genetics Society of Australasia

ARBN 076 130 937 (Incorporated Under the Associations Incorporation Act)
The liability of members is limited.

Submission to NSW Parliament Legislative Assembly Committee on Community Services- Improving access to early childhood health and development checks.

About HGSA

The Human Genetics Society of Australasia (HGSA) is the leading professional organisation in human genetics, working collaboratively and inclusively to advance the provision of quality genetic health for all, through research education and advocacy. We have over 1400 members working across all disciplines of human genetics, including clinicians, genetic counsellors, academics and diagnostic and research laboratory professionals.

Submission

The early identification of children who are not meeting developmental milestones is key to identification of any underlying causes and initiating appropriate intervention. Some of those children identified will have a genetic condition with the need for timely referral to clinical genetics services offering specialist diagnostic testing, treatment and counselling on potential implications for future pregnancies.

The HGSA believes limited access to clinical genetics services are part of the block to specialty assessment services and wishes to highlight the following issues:

- These services are almost exclusively located in metropolitan hospitals and are facing long wait times for non-urgent referrals. This is a particular barrier for rural and remote families with a wait of up to 2 years to be seen by a clinical geneticist.
- This extended wait time means families may be unable to access appropriate intervention without specialist testing leading to a specific diagnosis.
- There is a risk that without formal diagnosis and intervention there could be a family recurrence in some cases.
- There are a growing number of conditions where new therapies, including gene therapy, have the potential to significantly improve outcomes if started early enough.

The HGSA believes there is the opportunity to bridge gaps in services with targeted investment to assist mainstreaming support for paediatricians with access to online genetic counselling to initiate consent and testing as soon as possible.

The HGSA would be happy to answer any detailed questions arising from this submission.