



June 2024 Update

The PreGen study provides funded genomic testing at three national diagnostic genomic laboratories - NSW Health Pathology Randwick, VCGS and SA Pathology.

Families who have fetal structural abnormalities identified through ultrasound in pregnancy may be offered participation in PreGen.

Information is being collected through family questionnaires to plan for future prenatal genomic testing resources for Australian families.

PreGen milestones to date

228 exome trios (target of 266 exome trios)

82 diagnoses - diagnostic rate 36%

240 initial interviews with an uptake rate of 75%

138 follow up interviews at 6 months with an uptake rate of 87%

There is funding for a further 38 Exome trios before PreGen moves to prenatal Whole Genome Sequencing. The move to WGS is expected by September/October 2024. Full details will be communicated ahead of time by email.

PreGen Staff Updates

The project has four staff conducting psychosocial interviews. Senior genetic counsellor, Sarah Long, is based in Perth. Reanu Gopal and Jayamala Parmar are genetics counsellors based in Sydney and research assistant, Natalie Hart, is also based in Sydney.

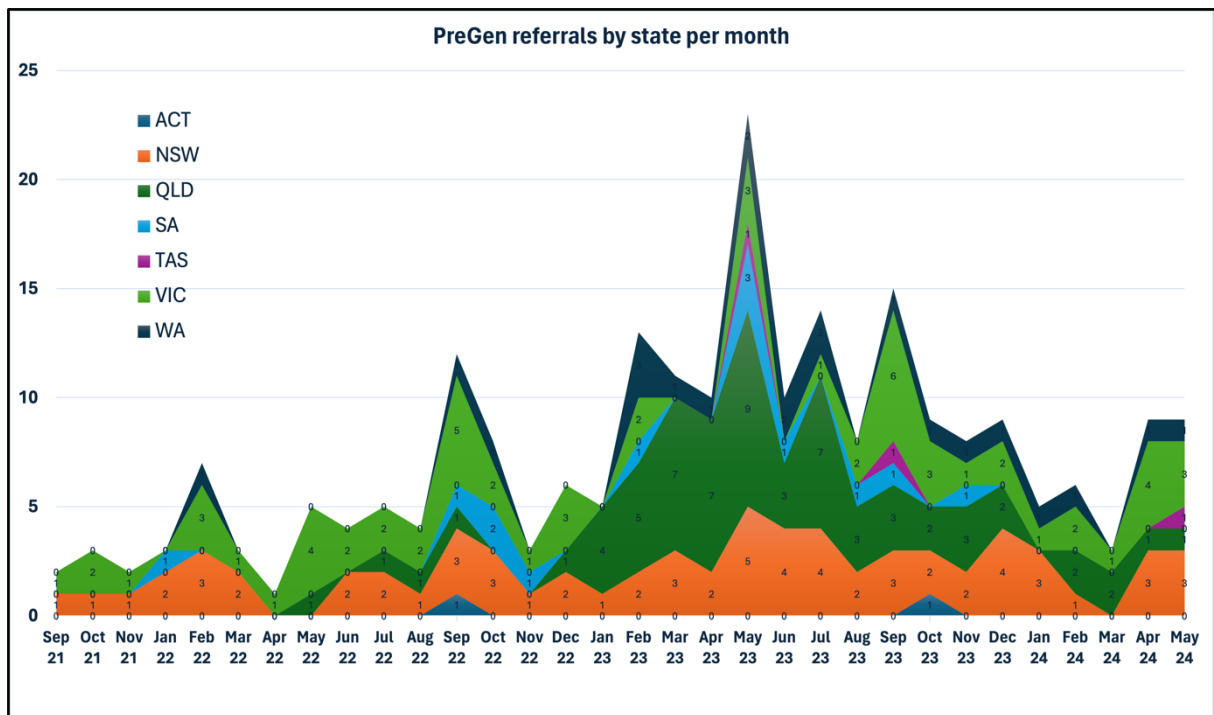
We also have a new program manager, Gemma Fernihough, to take us through to the end of the project in June 2025. We welcome all our new staff to the project and are confident they will make a great contribution to the essential counselling and management components of PreGen.

PreGen Website

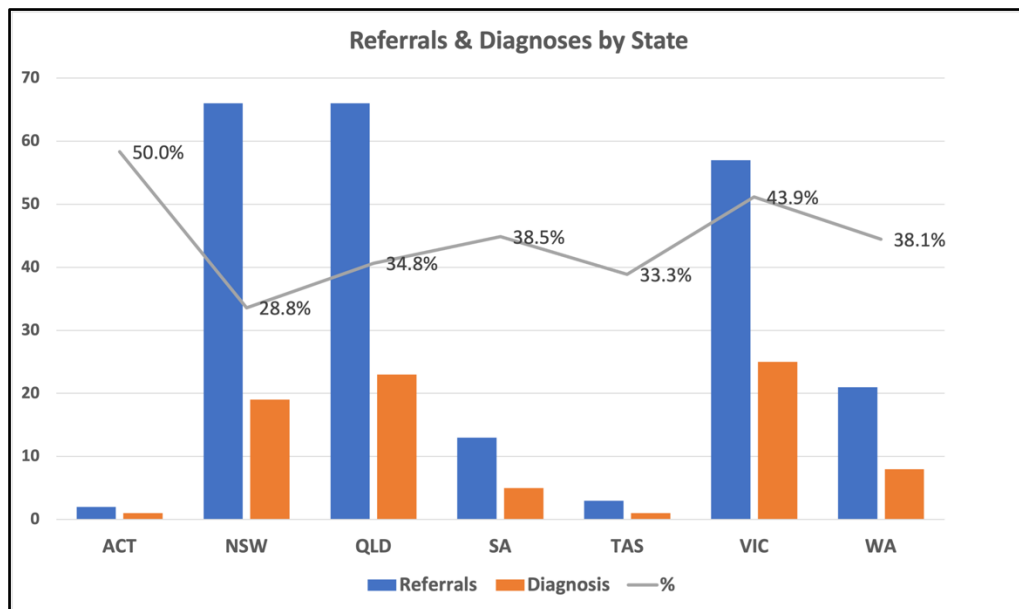
Please see the PreGen website (www.pregen.neura.edu.au) for the latest PreGen news as well as information for clinicians and families, clinical referral and consent documents. The most up to date PreGen inclusion and exclusion criteria can also be found there under 'useful documents'.

<https://pregen.neura.edu.au/useful-documents/>

Diagnoses and Referrals to PreGen from different Australian states

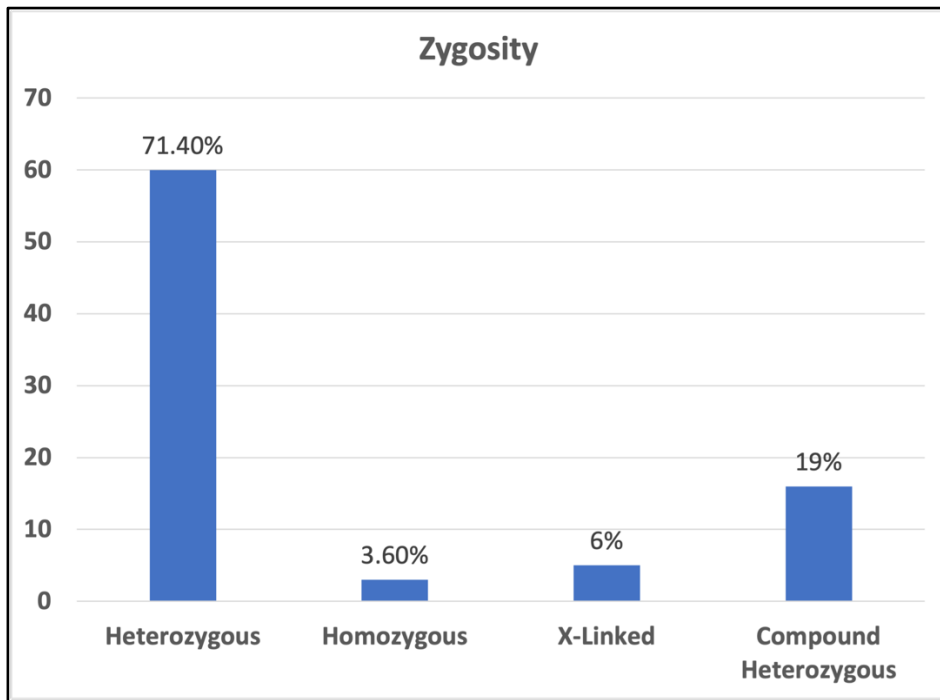


The average gestation at time of referral is 24 weeks. The diagnostic rate varies by state.



Zygosity & Inheritance

Of the 36% of families with diagnoses, 54.2% of variants were de novo and would be expected to have a low recurrence rate.



The most common referral HPO terms

The graph below shows the eight most common HPO from a total of 159 terms used in PreGen referrals.

