PREGEN

March 2025 Update

PreGen milestones to date

280 exome trios and 38 genome trios (diagnostic rate 35.0% overall)

WGS diagnostic rate so far: 11/27 – 40.7%

296 initial interviews (uptake rate of 78%) / 169 follow up interviews at 6 months (uptake rate of 54%)

Our application for federal funding of prenatal genomic testing

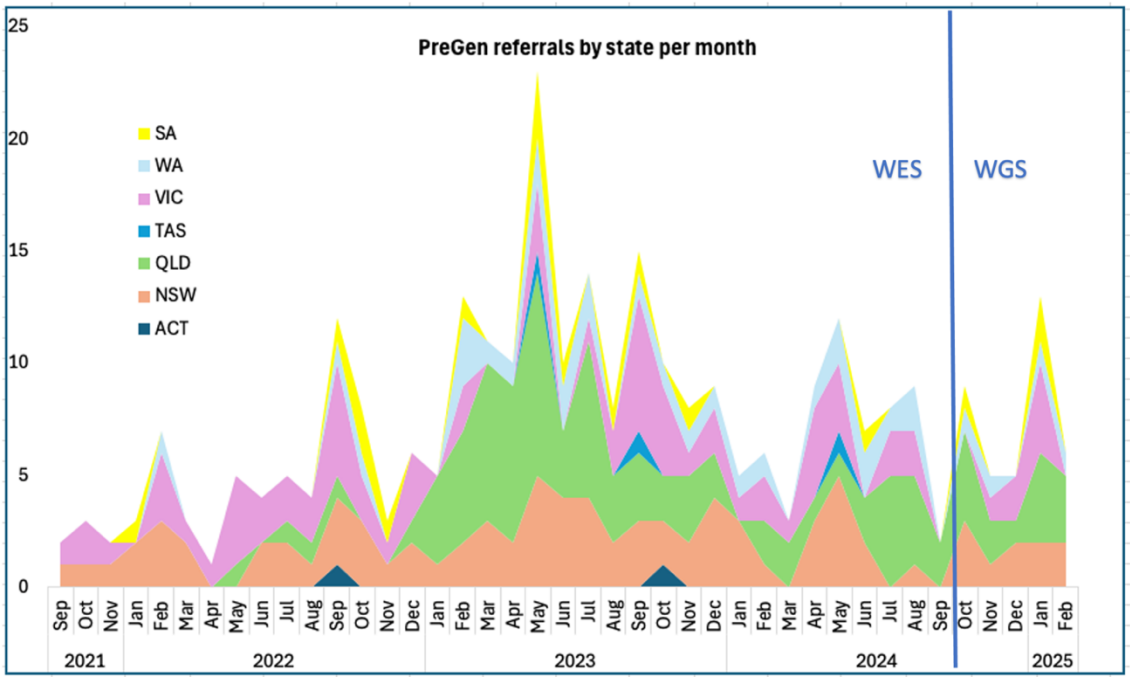
The PreGen team has been working hard on a submission to the medical Services Advisory Committee (MSAC) to secure an MBS item number for WES and WGS for fetal anomaly. This funding will enable equitable access for all Australian families. **Our initial suitability application has been successful**, and we are now developing a full assessment report due in November 2025. It is a long process with several stages likely to take longer than 12 months. We will keep you informed of the applications progress. **Thank you for your ongoing support of this project to provide equitable prenatal genomic testing to Australian families.**

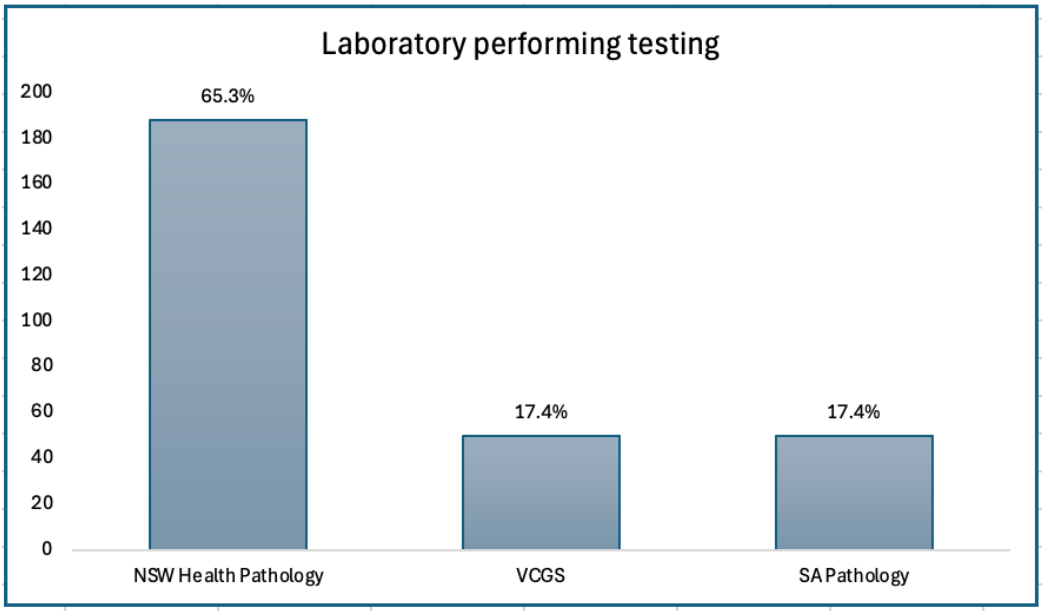
First of many PreGen papers is published

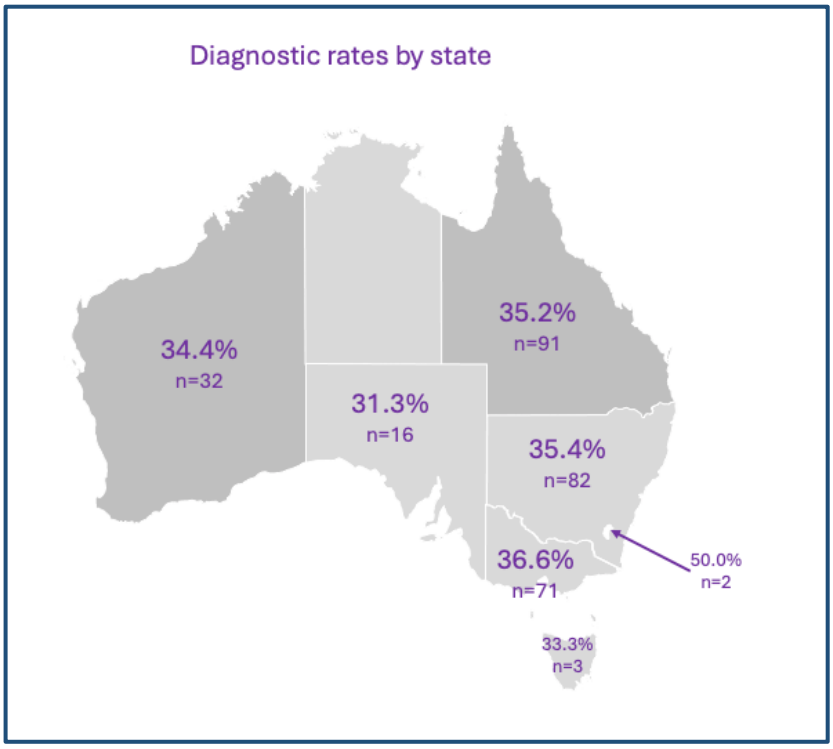
"The PreGen Research Program: Implementing Prenatal Genomic Testing in Australia—A Commentary" is published online in Australian and New Zealand Journal of Obstetrics and Gynaecology (ANZJOG). https://pubmed.ncbi.nlm.nih.gov/40123302/

DATA SUMMARY

Diagnoses and Referrals to PreGen from different Australian states

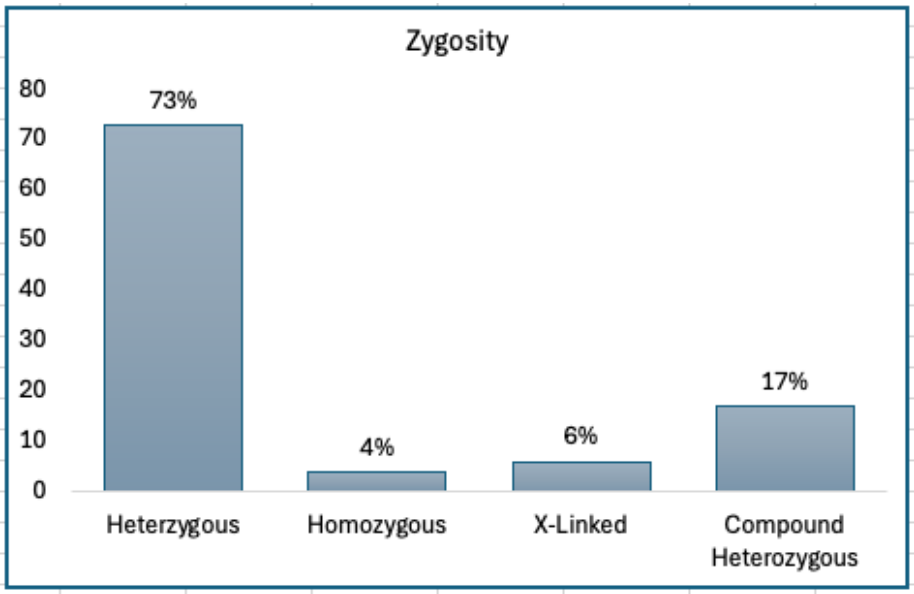


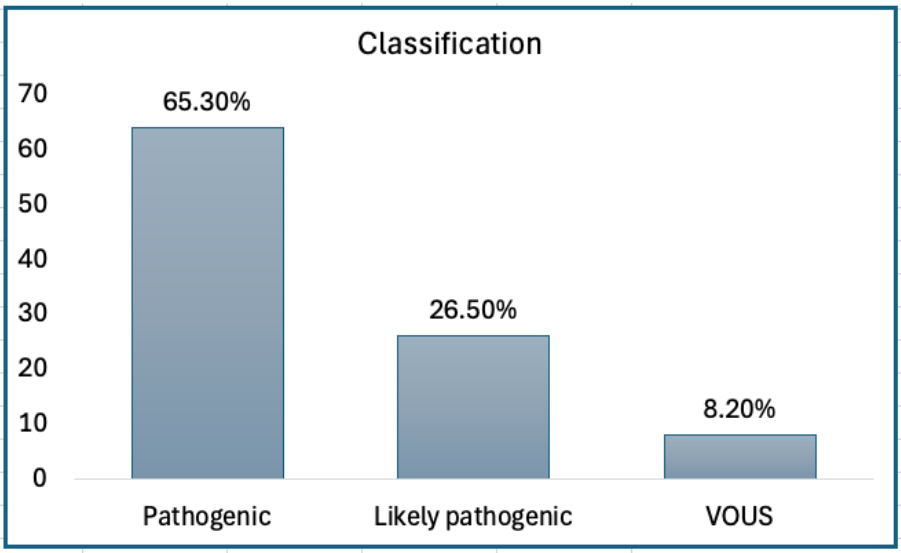

Laboratory performing testing
200
180
160
140
120
100
80
60
40
20
0
65.3%
17.4%
17.4%
NSW Health Pathology
VCGS
SA Pathology


Diagnostic rates by state
34.4%
n=32
31.3%
n=16
35.2%
n=91
35.4%
n=82
36.6%
n=71
50.0%
n=2
33.3%
n=3

Zygosity, Inheritance and Classification

Of the 35.3% of families with diagnoses, 58% of variants were *de novo* and would be expected to have a low recurrence rate.





Within the VOUS (n=8), seven were heterozygous and one was compound heterozygous. For the compound heterozygous VOUS, the classification of the variant *in trans* was pathogenic.