PreGen Updates
May 2022

The PreGen study is now providing 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.

There is funding for a further 245 Exome trios before PreGen moves to prenatal WGS.

Please note that **site specific approvals are not required to refer** to PreGen.

Please contact the PreGen team (pregen@neura.edu.au) if you have questions or if you have a family you would like to refer.

PreGen Staff Updates

Alongside our current PreGen Genetic Counsellors – Rebecca Vink (NSW) and Rachael Stenhouse (VIC) – we have our third Genetic Counsellor – Kate Ross – commencing work in SA.

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.

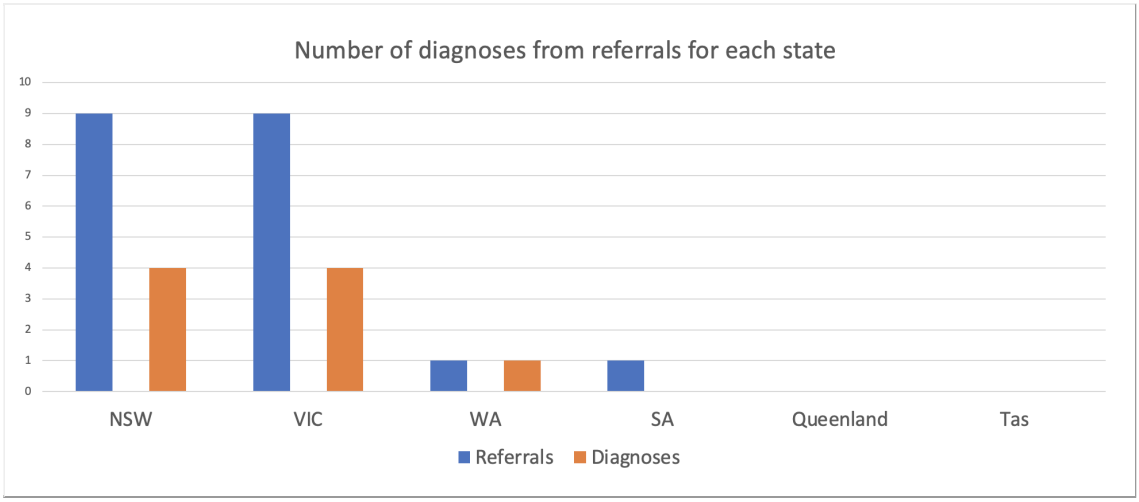
We have also **added a page on diagnostic rates** from recently published articles to the website which will be updated regularly.

You can find this under the ***Health Professionals Tab:***

https://pregen.neura.edu.au/diagnostic-rates/

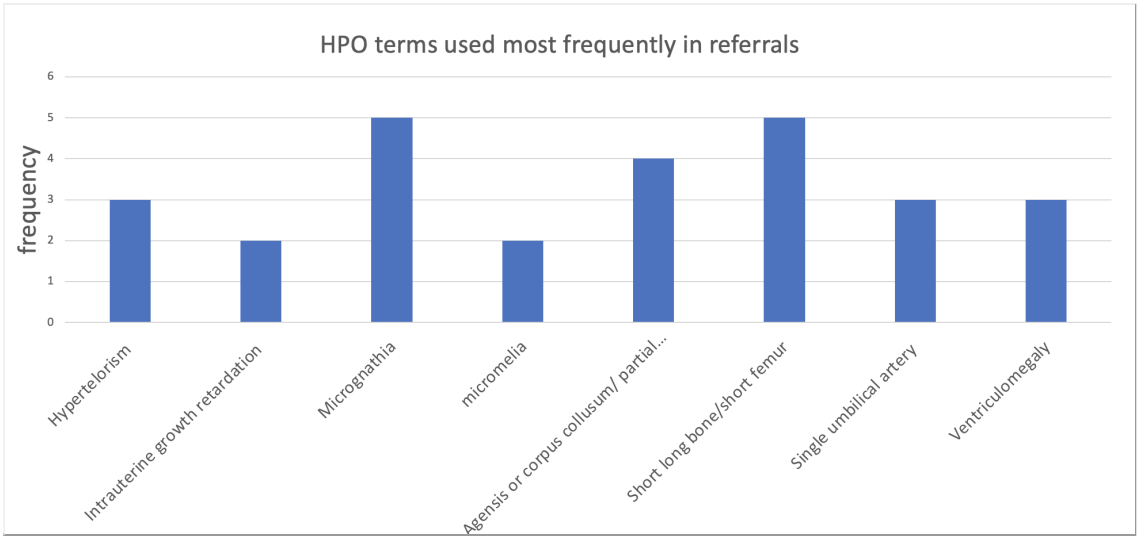
Diagnoses and Referrals to PreGen from different Australian states

The first 20 referrals to the PreGen Study have a **diagnostic rate of 45%** with average gestation at referral being 25 weeks. All families have completed their initial psychosocial questionnaire and 6 monthly follow up questionnaires have commenced.



PreGen's most common referral phenotype terms

Micrognathia
Short long bones / short femurs
Agenesis / partial agenesis of corpus collosum



Upcoming Events

PreGen meeting updates are now being provided across sites in Australia covering progress on the project so far and answering questions on family suitability and referrals to the project.

Please contact PreGen Program Manager, Alyssa Wilson, if you would like to organise an update for your site.

PreGen Contacts

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