



**PreGen Antenatal Genomics  
and Newborn Care Study**  
Website: <https://pregen.neura.edu.au/>

**Information about PreGen for referrers located in VIC / TAS / NT**

- PreGen is a national multicentre MRFF funded research program.
- PreGen will assess the implementation and clinical utility of prenatal genomic testing for fetal anomalies. It will also assess clinical outcomes for fetuses and newborns who were tested, the psychosocial impact of testing on prospective parents and whether prenatal testing has health-economic benefits.
- When prospective parents consent to PreGen enrolment, the pregnancy is currently ongoing and the clinical inclusion criteria are met, PreGen funding will cover the cost of prenatal genomic testing via whole exome sequencing (WES) or whole genome sequencing (WGS) trios performed in diagnostic genomic laboratories (NSW Health Pathology Randwick, VCGS, SA Pathology).
- Families can be considered for enrolment in PreGen by the clinical genetic and maternal fetal medicine teams **after they have consented to clinical prenatal genomic testing**. Clinical teams will need to assess and consent the family for clinical testing using their usual counselling and consent processes. If a fetus / family meet the criteria for inclusion but the parents decline the option of enrolment in PreGen, clinical funding will need to be available to cover the cost of testing.
- To qualify for PreGen enrolment and funded testing, families will consent to complete two psychosocial questionnaires with a PreGen Genetic Counsellor (via phone/zoom interview, one before genomic results return and one six months later) and for researchers to access costing data related to pregnancy care via MBS/PBS/hospital data linkage.
- Information regarding clinical inclusion criteria can be found on the website: <https://pregen.neura.edu.au/health-professionals/>

**Referral Process** [For requests to VCGS Laboratory]

**1. Once a decision has been made to offer a couple clinical testing, please:**

- a. Complete clinical consent and clinical test pathology request and documentation (e.g. payment authorisation) for fetal rapid trio genomic sequencing and sample storage through VCGS in the usual way for your service.
- b. Genomic testing will proceed if other routine testing (eg. microarray, CMV PCR) does not explain the fetal findings.
- c. **Discuss the option of PreGen enrolment with your patient and their partner and obtain permission for our PreGen Genetic Counsellor, Rachael Stenhouse, to contact them to discuss the project further.**
- d. If your patient is interested in PreGen, **please write “For PreGen”** on clinical request form.

**2. Email a clinical summary to the PreGen Genetic Counsellor - [pregen@neura.edu.au](mailto:pregen@neura.edu.au)**

- Patient details: name, DOB and contact
- Clinical details: Pregnancy gestation / EDD, relevant medical or family history (include pedigree if relevant), fetal phenotype, date of CVS/amniocentesis, array result
- Referrer details: Doctor name, clinical genetic counsellor name, site and contact
- A referral template is available [here](#)

**Contacts (Victoria)**

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