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

Information about PreGen for referrers located in WA

- 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/ Prenatal genomic testing for WA will generally be sent to SA Pathology.
- Queries regarding clinical eligibility for PreGen may be directed to chief investigator, Tony Roscioli on (02) 9382 9169 / tony.roscioli@health.nsw.gov.au, and/or PreGen genetic counsellor, Kate Ross on +61 403 217 360 / kate.ross@sa.gov.au

Referral Process

1. **Once a decision has been made to offer a couple clinical testing, please:**
    a. **Notify testing laboratory SA Pathology when samples are being sent from WA.** Please email the following: Prof Hamish Scott (Hamish.Scott@sa.gov.au), Kate Ross (genetic counsellor, Kate.Ross@sa.gov.au), Lucas De Jong (Genomic analyst SA Pathology, Lucas.DeJong@sa.gov.au).
    b. Complete clinical consent and clinical test pathology request for fetal rapid trio genomic sequencing in the usual way for your service.
    c. Obtain approval to offer couple prenatal trio exome testing via PreGen (please contact Tony Roscioli and/or Kate Ross with clinical summary for review and approval).
    d. **Once accepted for PreGen testing, discuss the option of PreGen enrolment with your patient and their partner and obtain permission for our PreGen Genetic Counsellors, to contact them to discuss the project further.**
    e. Send test request form to PathWest (Philip Asquith) to arrange DNA extraction and send away to SA Pathology. If your patient is interested in PreGen, **please write "For PreGen"** on clinical request form.
    f. Genomic testing will proceed if other routine testing (eg., microarray, CMV PCR) does not explain the fetal findings.

2. **Email a clinical summary to the PreGen Genetic Counsellors**
   Kate Ross: Kate.Ross@sa.gov.au **(Cc** pregen@neura.edu.au**) and please include:**

- 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

Samples

- DNA from amniocentesis is 30ul of 30ng/ul of DNA
  - **Please** discuss with SA Pathology laboratory if less DNA is available
- DNA from both parents extracted in Perth – 500ng
  - Alternatively, whole blood in EDTA tubes can be couriered

For any sample queries, please contact Genome Analyst, Lucas DeJong, at SA Pathology on (08) 8222 3438 or Lucas.Dejong@sa.gov.au.

Contacts (WA)

- **Ben Kamien** – Principal Investigator, WA - P: +61 8 6458 1242; E: benjamin.kamien@health.wa.gov.au
- **PreGen Genetic Counsellors**: Kate Ross (SA & WA) – **P:** +61 403 217 360 Kate.Ross@sa.gov.au
  - Interstate GCs (NSW & Vic) – **P:** (02) 9399 1817
    E: pregen@neura.edu.au
- **SA Pathology:** Laboratory enquiries on **(08) 8222 3438** (or free call 1800 188 077) may be directed to Genome Analyst, Lucas DeJong, Lucas.Dejong@sa.gov.au
  - SA lead investigator – Prof. Hamish Scott (Hamish.Scott@sa.gov.au)
- **Tony Roscioli**: Chief Investigator (02) 9382 9169 / tony.roscioli@health.nsw.gov.au
- **Pathwest Sendaways team**: Philip.Asquith@health.wa.gov.au and Sendaways.DiagnosticGenomicsQE.PathWest@health.wa.gov.au
  Philip's phone is 63834233 or he can be contacted via general DGS number 63834234