## **Information about PreGen for referrers located in NSW / ACT / QLD**

* PreGen is a national multicentre MRFF funded translational 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 ACT and QLD will generally be sent to NSW Health Pathology Laboratory.
* Queries regarding clinical eligibility for PreGen may be directed to chief investigators, Tony Roscioli on (02) 9382 9169 / tony.roscioli@health.nsw.gov.au, George McGillivray / [george.mcgillivray@vcgs.org.au](mailto:george.mcgillivray@vcgs.org.au) and/or the PreGen genetic counsellors at pregen@neura.edu.au

**Referral Process** [For requests to NSWHP Genomic Laboratory Randwick]

1. Once a decision has been made to offer a family clinical testing, please:
   1. Complete clinical consent and clinical test pathology request for fetal rapid trio genomic sequencing in the usual way for your service.
   2. Send test request form to: NSWPATH-RandwickGenomics@health.nsw.gov.au Please note where consent is stored.
   3. Genomic testing will proceed in parallel to microarray testing but may be halted if an aetiology is identified by microarray testing.
   4. Discuss the option of PreGen enrolment with your patient and their partner and obtain permission for a PreGen Genetic Counsellor to contact them to discuss the project further.
   5. If your patient is interested in PreGen, please write “For PreGen” on clinical request form.
2. Email a clinical summary to the PreGen Genetic Counsellors - [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 if available
   * Referrer details: Doctor name, clinical genetic counsellor name, site and contact

**Samples**

* DNA from amniocentesis/CVS - 35-50ul at 30-60ng/ul. If you have a lower amount of DNA, please discuss with NSW Health Pathology prior to submission.
* Submit samples in labelled 1.5mL tubes. Please label tube and lid if possible.
* Ensure the DNA contains less than 1mM EDTA and is free from phenol, ethanol and other contaminants.
* DNA 500ng or EDTA from both parents

**Contacts**

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