Where will my appointment be?

We are a NSW state-wide service based at two locations:

Hunter Genetics, Corner Turton & Tinonee Rds WARATAH NSW 2298

&

Royal North Shore Hospital, Reserve Rd ST LEONARDS NSW 2065

We see individuals and families at either of these locations. Alternatively, we can arrange to see you either at home or a local community health centre in surrounding areas of Sydney and Newcastle.

We also regularly see families at regional outreach locations across NSW and the ACT.

Please inform the genetic counsellor involved with your family if you are unable to travel to a clinic appointment and require a home visit.

Version 1, 17/08/2016. Pending CPGAG approval

How to contact our team:

Hunter Genetics (Newcastle and any administrative questions):

Phone: (02) 4985-3100
Fax: (02) 4985-3105

RNSH (Sydney-based enquiries):

Phone: (02) 9463-1555 or
(02) 9463-1558
Fax: (02) 9463-1057

Postal Address:
PO Box 84,
WARATAH NSW 2298

Please direct referrals to:

Dr Mike Field
Dr Anna Hackett
Dr Tracy Dudding-Byth
Dr Emma Palmer

Please include with your referral:

- Previous clinic letters
- All genetic and any relevant non-genetic investigation reports
- Name(s) and contact details for parent/guardian/consenting adult

Kaleidoscope
THE CHILDREN'S HEALTH NETWORK

The NSW Genetics of Learning Disability (GOLD) Service

Incorporating the Fragile X Service

Rafa, 6 years old

NSW State-wide clinical genetics service

NSW GOVERNMENT Health
Hunter New England Local Health District

The NSW Genetics of Learning Disability (GOLD) Service

The GOLD Service provides genetic services to NSW families with fragile X syndrome and families who have two or more relatives with intellectual disability (ID).
The aim is to identify the cause of ID within these families and provide reliable genetic counselling for all family members.

Who are we?

Our team is made up of:

- Clinical geneticists
- Clinical nurse consultants / Genetic counsellors
- Administrative support staff
- A Bioinformatician

What families do we see?

- Families with two or more individuals with ID (at least one with moderate ID). At a minimum, affected individuals should be 1st or 2nd degree relatives.
- Any X-linked families with ID (2 or more affected males through related females)
- Individuals and/or families with ID who have an array abnormality on the X chromosome that is likely to be related to the ID.
- Individuals and /or families with an expanded Fragile X (*FMR1*) gene.

What does the GOLD service offer?

- A comprehensive clinical genetic service to those families within NSW with fragile X syndrome
- A comprehensive clinical genetic service to those families within NSW with inherited intellectual disability
- Co-ordination of genetic testing for affected individuals
- Testing other family members if the specific gene alteration responsible for intellectual disability within the family is known
- Provision of current information on inherited forms of ID to families, health professionals and the community.
- Counselling regarding reproductive choices, and community resources and support groups
- Participation in research studies to identify the causes of inherited conditions resulting in ID.

How much will it cost?

There will be no direct cost to you for the appointment or any recommended genetic testing. This service is funded by the NSW ministry of health. The cost of the consultation is covered by Medicare.

Referral process

To arrange an appointment, a referral from a medical specialist (such as a paediatrician or neurologist) to one of our doctors is required. For families with a Fragile X genetic change, either a specialist or GP referral is acceptable.

What happens after the referral?

- A genetic counsellor will call you to discuss the referral and obtain more information about your family history. An appointment will then be arranged with one of our geneticists.
- At your appointment, the clinical geneticist will examine the referred individuals. Frequently, this will include a basic neurological examination like checking reflexes. Sometimes photos of you and your family will also be taken to keep in your family's file.
- Depending on the examination, genetic testing may be offered and a blood sample (or other samples such as saliva/cheek swab) may be collected.
- The geneticist may also recommend and refer you or your family member to be seen by other specialists.

What should I bring to my appointment?

The genetic counsellor will advise you on what to bring with you, but typically this includes for each referred family member:

- The 'Blue Book' for children,
- Allied health, developmental clinic and school reports,
- Clinic letters and results from other doctors
- Sometimes family pictures can also be useful to bring along.