

## **Genetics of Learning Disability (GOLD) Service**

This is a NSW state wide service based at Hunter Genetics in Newcastle and the Chatswood Assessment Centre in Sydney

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

### **What is X-Linked Intellectual Disability?**

Intellectual disability is common in the general population and affects males more frequently than females. If the intellectual disability is caused by alterations on the X chromosome it is referred to (in the medical literature) as X-Linked Mental Retardation (XLMR). XLMR affects 1 in 650 males. There are over 200 genes on the X chromosome that control brain function. To date, over 80 genes have been found to cause XLMR. The most common XLMR is fragile X syndrome and genetic testing for fragile X syndrome is routinely available. Genetic testing for other causes of XLMR is often only available through research studies.

### **How is X-Linked Intellectual Disability Inherited?**

Males have one X and one Y chromosome in their cells. The X chromosome is inherited from the mother and the Y chromosome is inherited from the father. If there is an alteration within a gene on the X chromosome, the male will have intellectual disability.

Females have two X chromosomes in their cells, one inherited from the mother and one inherited from the father. If there is an alteration within a gene on one of the X chromosomes, females usually will be unaffected because they have a working "backup" gene on the other X chromosome. These females are called carriers. Occasionally, some carrier females have mild learning problems.

### **What does the GOLD service offer?**

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

### **How to contact the service**

- Family members can contact the phone numbers listed below and speak with a genetic counsellor
- For a genetic consultation, a referral from your GP or medical specialist is required

### **What happens at the genetic consultation?**

- Prior to the genetic consultation, a genetic counsellor will call you by phone and obtain a detailed family tree and history
- Copies of previous medical investigations or assessments are obtained
- A clinical geneticist (a doctor trained in genetics) will examine the affected males, either at

- home or at a genetics clinic, and photographs are usually taken to be kept in the family file.
- Depending on the findings of the genetic examination, specific tests may be ordered to try and identify the cause of the intellectual disability within the family

### **How is the testing done?**

- Genetic testing can be performed on DNA which is extracted from blood or saliva samples
- Results can take months or even years and may not be successful in finding the gene alteration
- A DNA sample can be stored at the HAPS laboratory at the John Hunter Hospital in Newcastle, for future genetic testing as new genes are identified and genetic technology improves
- A transformed cell line (long lasting cells from which DNA can be extracted) can also be stored at Prof. Jozef Gecz's laboratory at the Molecular Genetics Laboratory at the Women and Children's Hospital, in Adelaide

### **Finding genes that cause X-Linked intellectual disability**

The development of new technology has led to the identification of many genes that cause X-linked intellectual disability. The GOLD service has strong links with researchers in Australia and overseas. Enrolment into research studies may be offered to families where the cause of intellectual disability is unknown.

### **Costs**

There will be no direct cost to patients and their families. This service is funded by the NSW Department of Health. The cost of the genetic consultation is covered by Medicare.

### **GOLD TEAM**

#### **Contact Details:**

**GOLD Service**

**Hunter Genetics**

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