

CLINICAL GENETIC SERVICE



Dr Rani Sachdev
Dr Mary-Louise Freckmann

Phone: 61 2 9113 3635
Fax : 61 2 9113 3694

The St George Hospital and
Community Health Service
Level 1, Prichard Wing
Gray Street, Kogarah 2217
Australia
61 2 9113 1111

Genetic factors make a substantial contribution to physical and intellectual disability, chronic ill-health, psychiatric illness and familial cancer

50% - people will have a disease with a genetic component during their lifetime

5% - people experience gene related illness, impairment or disability by age 25

Hospital admissions ~50% of paediatric
~12% of adult

GENETIC COUNSELLING - REFERRALS

- AMA / LMA : ≥ 35 yrs at delivery
 - Chromosomal abnormalities eg., Down syndrome
 - Chromosomal rearrangement
 - PHx of abnormal sib or child / stillborn / NND
 - Known AD, AR, XL or multifactorial disorders
 - ♦ Thalassaemia
 - ♦ cystic fibrosis (CF)
 - ♦ neural tube defect
 - ♦ Huntington disease (HD)
 - ♦ cardiomyopathy
 - ♦ FRAXA
 - Consanguinity
 - Abnormalities on ultrasound scan
 - Multiple miscarriage - ≥ 3
 - Teratogens
 - Paediatric eg., dysmorphism, FTT
 - Adult referrals eg., peripheral neuropathies
-

GENETICIST VS GENETIC COUNSELLOR

A **Clinical Geneticist** is a physician trained in the application of the principles of human genetics including the interpretation of laboratory genetic findings to the diagnosis and management of genetic disorders and the counselling of patients and families. Clinical geneticists have had widely based training combining knowledge and skills in genetic counselling and in formal genetics.

Genetic Counsellors come from varied backgrounds. In the past many Nurses or Social Workers moved from their traditional roles into supporting families affected by a genetic condition. Today Scientists, Teachers, Psychologists and university graduates are swelling the ranks of the Genetic Counselling fraternity.

Training to become a certified Genetic Counsellor is a two part programme established by the Human Genetics Society for Australasia (HGSA) and overseen by the Genetic Counselling Board of Censors. Part I may be completed through any of four established post graduate university courses throughout the country. Part I has two aspects, one covering genetic knowledge and the other counselling skills. Associate Genetic Counsellors have Part I training with most jobs today requiring Part I as a pre-requisite for employment.

Part II training leading to certification as a Genetic Counsellor [FHGSA (Genetic Counselling)] requires a minimum of two years full time employment with a Clinical Genetics Unit. The certification process includes extensive written case reports, genetic and counselling supervision along with detailed continuing education activities. There is also a Maintenance of Professional Standards (MOPS) programme designed to enhance and maintain existing Counsellor's skills as well as the development of new learning directions to meet growing and changing knowledge in genetics and counselling.
