

## CLINICAL GENETICS SERVICES

### CHAPTER 7

Genetics is of increasing importance in clinical medicine as an understanding of genetics can underpin many areas of health care and medical practice. Genetics health professionals are involved in the translation and application process of genetics knowledge, which is growing exponentially from Human Genome Project and other research, while addressing the immediate needs of patients.

A full range of reproductive and general paediatric genetic services are available including diagnosis, counselling and organisation of genetic testing. Cancer genetics is currently not provided. Opportunities exist for SWSAHS to develop a Familial Cancer Clinic with appropriate funding for gene testing.

The service saw 1,106 patients in 2001, and 1,082 patients in 2002. The proportion of new patients in new families, to new family members in known families, to returned patients has remained the same. SWSAHS currently has high outflows for genetic services.

There is a growing demand for adult genetics especially neurogenetics with patients referred from all disciplines of medicine. In addition to genetic diagnosis and testing, genetic counselling for adult family members is offered which can include risk assessment, predictive testing for late onset disorders using agreed protocols, provision of health surveillance information and maintenance of genetic family pedigrees. Each year the genetic aetiology is identified for more medical conditions and therefore more families could utilise genetic services to obtain risk information of developing a genetic condition to improve their health surveillance and be informed of options for prenatal diagnosis.

An area that is increasing rapidly relates to cancer genetics where the genes for commonly inherited breast/ovarian and bowel cancers have been identified. There is growing health professional and public awareness of the importance of family history, risk assessment, counselling regarding surveillance and availability of certain cancer gene testing. This is the service provided by a Familial Cancer Clinic. In 2000, cancer was the second most common diagnosis for a new referral to NSW genetics units (after late maternal age) and the most common reason considering all referrals, new or follow-up. Research and development worldwide in this field continues rapidly and more gene tests are becoming available.

### Current Service Provision

The Department of Clinical Genetics (DCG) is based at Liverpool Hospital and provides a comprehensive reproductive and paediatric Area Genetic Service. It includes genetic diagnosis, counselling and investigation with the organisation of pre and post-natal chromosomal and molecular tests. Referrals are also accepted for general adult genetics including neurogenetics. Metabolic genetics services for children with inborn errors of metabolism are not provided at Liverpool as these conditions are best treated by specialised teams at the children's hospitals. The provision of educational services to hospital staff is an important component of the service. The service runs with 1.5 staff specialists and 1.6 genetic counsellors.

In addition, the Department of Clinical Genetics has a mandate to provide outreach genetics services to Southern Area Health Service (SAHS) as per Genetics Services Advisory Committee (GSAC) NSW Health, however this service ceased in 2002. Since then the genetic counsellor position based in SAHS has been vacant and the long term outlook for this service is unknown.

The Department of Clinical Genetics has a close working relationship with the Feto-Maternal Unit at Liverpool Hospital and with the antenatal clinics within SWSAHS.

Outreach services are currently provided to Campbelltown and Bankstown sectors with clinics in these hospital outpatients.

Specialised multidisciplinary clinics in conjunction with dermatology or ophthalmology are components of an enhanced service delivery model that need to be explored for the Area.

The majority of DNA/molecular tests, which are increasing in number, availability and demand, have no Medicare rebate and the laboratories in both Australia and overseas now charge for these tests. Testing where appropriate is an integral part of a genetics service and requires a targeted pathology budget to meet the growing demand.

### **Research and Teaching**

Clinical geneticists are resource persons for health care professionals and play an active role in teaching in hospitals. The DCG provides educational services for hospital medical, undergraduate and post-graduate health professionals, midwifery and antenatal staff. The department of clinical Genetics is also involved with Divisions of General Practice particularly with the Obstetric Shared Care Program and is a resource for community groups including genetic disease support groups and paediatric support organisations.

#### **RECOMMENDATIONS**

- Genetics services be progressively developed as per NSW Health Genetics Services Advisory Committee planning standards.
- The main service for Clinical Genetics continue to be delivered from Liverpool Hospital.
- Enhance outreach services at Campbelltown Hospital by the appointment of a genetic counsellor.
- An outreach service with weekly clinics at Bankstown and Fairfield be developed.
- A dedicated pathology budget be provided to support genetic testing.
- Develop comprehensive cancer genetics services for SWS.