



# Genetics Services in NSW

# 1996-2000



**NSW HEALTH DEPARTMENT**

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Produced by:

The NSW Genetics Service Advisory Committee

Statewide Services Development Branch

NSW Health Department

Locked Mail Bag 961

North Sydney NSW 2059

Tel. (02) 9391 9520

For further copies:

Better Health Centre

Locked Mail Bag 5003

Gladesville NSW 2111

Tel. (02) 9816 0452

Fax. (02) 9816 0492

A full copy of this report and others in this series can be downloaded from the NSW Health Web site:  
[www.health.nsw.gov.au](http://www.health.nsw.gov.au)

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# Highlights and achievements 1996-2000



The NSW Genetics Service Advisory Committee (GSAC) was established in 1987 to provide policy and planning advice to the Director-General of the NSW Health Department, through the NSW Genetics Service Executive Office located in the Statewide Services Development Branch.

The GSAC's membership represents all aspects of genetics services. The consensus approach of the Committee and Working Parties has fostered the rational planning and development of a coordinated network of services across the state. This process has been documented in several earlier publications listed at the end of this paper.

## **New clinical and counselling services**

Over the last few years the planning process has resulted in the establishment of:

- new clinical and counselling services at Royal Prince Alfred Hospital, Liverpool Health Service and Nepean Hospital.
- amalgamation of genetics services at Westmead Hospital and the Children's Hospital at Westmead.
- new genetic counselling services at St George Hospital.
- expansion of rural genetic counselling services with new services at Mudgee/Dubbo, Port Macquarie, Taree/Forster, Gosford and upgrading of services in other Areas.
- expansion of genetics services in Northern Sydney Area in conjunction with services in South Eastern Sydney Area.

## **New cancer genetic services**

Technological advances in genetics occur with such rapidity that the implications of cancer genetics were not considered within the scope of the *Five Year Plan 1993-98*. However, the identification of cancer predisposing mutations has propelled the development of specialised services. Five familial cancer services have been established at Prince of Wales, Westmead,

Liverpool and St Vincent's Hospitals and Hunter Genetics. Clinical services and laboratory testing have been made available with the support of Area Health Services and the Department of Health.

## **Biochemical genetics and newborn screening**

The introduction in 1998, of new technology using tandem mass spectrometry has improved laboratory efficiency by enabling testing for a number of extremely rare disorders in one analysis.

## **New DNA testing technology**

In the past five years the introduction of new equipment for high throughput mutation screening, such as denaturing HPLC and multicapillary DNA sequencers, has dramatically increased the number of patient samples analysed as well as significantly reducing turnaround times and costs associated with testing.

## **Database development**

Improved reporting processes developed and implemented by the NSW Genetic Service now enable the collection of non-identifying information from clinical and laboratory services across the State to produce a Statewide picture of genetics service activity. The data reproduced in this report will provide the basis for future service planning.

The clinical genetics database also serves as an efficient patient record management tool for all clinical and counselling services.

## **MotherSafe**

MotherSafe was established in January 2000 as the Statewide Medications in Pregnancy and Lactation Service at the Royal Hospital for Women and was officially opened by the Director General of Health, Mr Michael Reid, in May 2000.

### **Policies and guidelines**

The Committee has been responsible for producing policies and guidelines, including:

- *The Goal and Objectives of the NSW Genetic Service* (1995) which sets directions for genetics services.
- *The Ethical Code Governing the Provision of Genetics Services* (1998) considers the shared implications of genetic inheritance for family members and aspects of genetic testing and procedures which raise specific ethical issues. It acknowledges the need for, and importance of, meaningful ethical principles to guide health professionals and individuals through the difficult personal and collective decisions which are encountered in the application of genetic technologies.
- *Specialised Testing for Genetic Disorders* (2000) provides guidelines for testing based on this ethical approach, in a context which addresses full information, consent, privacy issues and impact on other family members. It makes recommendations for service provision and on cost recovery processes.
- A number of Information Bulletins, Circulars and other publications listed at the end of this document.

The NSW Genetics Service is an umbrella term used to unite all genetics service disciplines and service providers. It is coordinated by the NSW Genetics Service Advisory Committee and Statewide Services Development Branch in the NSW Health Department.

## Goals

Planning for genetics services follows the principles of *NSW Health Strategic Directions for Health (2000-2005)* and is structured around the following four goals.

### Healthier people

To reduce the impact of genetic disorders and birth defects on affected individuals, individuals at risk and their families.

### Fairer access

To enhance patient and family wellbeing through provision of conveniently located genetics services.

### Quality health care

To maximise genetic health outcomes through provision of information, services and support appropriate to the needs of the people of NSW.

### Better value

To support the development of clinical and laboratory services and implementation of beneficial new technologies.

## Services

A list of all services by Area Health Service is provided in Appendix 1. Full address details are provided in Appendix 3.

### Clinical and counselling services

Available at six teaching hospitals and staffed by clinical geneticists and genetic counsellors who provide information, clinical, counselling and diagnostic services on a cross area basis.

- **Clinical geneticists** are physicians with a specialist qualification in clinical genetics.
- **Genetic counsellors** are non-medical graduate health professionals with specialist training who work closely with clinical geneticists.

### Outreach genetic counselling services

Located in all Area Health Services in NSW. Locally employed genetic counsellors are supported by visiting clinical geneticists from major units, who conduct outreach clinics on a regular basis.

### Cancer genetics services

Specialised services are available at five teaching hospitals, provided by physicians trained in oncology/genetics and cancer genetic counsellors. Clinical and genetic counselling services also provide cancer genetics services or arrange referral to a specialised cancer genetics service.

### Prenatal diagnosis services

Located in seven teaching hospitals and providing expertise in the detection of fetal abnormalities.

### Cytogenetics services

Services are provided from four public hospital laboratories and one private laboratory, for the detection of cytogenetic abnormalities, infertility investigations and some cancer diagnoses.

### Molecular genetics services

Presymptomatic, predictive and carrier testing is provided for the diagnosis of specific genetic disorders. Each of the seven laboratories provides a Statewide service for a specific range of disorders.

### Newborn screening services

#### Biochemical genetics service

#### Genetic metabolic diseases service

Statewide programs based at the Children's Hospital at Westmead, for the diagnosis of a number of genetic conditions in newborn babies, inborn errors of metabolism and other rare biochemical genetic conditions.

### **Genetic education**

The Statewide NSW Genetic Education Program is based at Royal North Shore Hospital and produces genetic education information and resources for the community and health professionals.

### **The Association of Genetic Support of Australasia (AGSA)**

A community service providing peer support and information for families and individuals affected directly or indirectly by genetic conditions.

### **MotherSafe**

A Statewide advisory service for pregnant and lactating women whose pregnancies may be at risk as a result of exposure to medications or other teratogens. It is based at the Royal Hospital for Women.

# The NSW Genetics Service Advisory Committee

# 3

The NSW Genetics Service Advisory Committee (GSAC) reports to the Director-General of NSW Health through Statewide Services Development Branch.

The Committee's membership is drawn from all genetics service disciplines, as shown in the table below.

Genetics service discipline	Representative 2001/2002
Clinical Genetics SESAHS	Dr Anne Turner
Clinical Genetics SWSAHS	Dr Alison Colley
Clinical Genetics HAHS	Dr Matt Edwards
Clinical Genetics CHW	Dr Meredith Wilson
Clinical Genetics CSAHS	Dr Rani Sachdev
Clinical Genetics WAHS	Dr Mary-Louise Freckmann
Biochemical Genetics and Newborn Screening	A/Prof Bridget Wilcken, <b>Chair 1998 – present</b>
Molecular Genetics	A/Prof John Christodoulou
Cytogenetics Services	Ms Mary Suter
Genetic Education Services	Dr Kristine Barlow-Stewart, <b>Deputy Chair</b>
Genetic Counsellors	Ms Judith Elber
Prenatal Diagnosis Services	Dr John Smoleniec
Cancer Genetics Services	Professor Michael Friedlander
Community Representative (AGSA)	Ms Dianne Petrie
Genetics Service/NSW Health Department	Ms Jennifer Blackwell
	Prof David Sillence
	A/Prof Graeme Morgan, <b>Chair 1987-1998</b>

Rural services are represented by clinicians from major units who provide rural outreach services.

## Terms of reference

To advise and make recommendations on:

- relevant policy issues
- the development of guidelines for the delivery of high quality clinical, counselling, laboratory and educational genetics services
- current and projected service requirements consistent with NSW Health Department service delivery processes for equitable access
- evaluation of outcomes of genetics service provision.

## Current working parties

- Cancer Genetics
- Data Storage and Computer Systems
- DNA/Molecular Genetics Laboratory Services
- Cytogenetics
- Genetic Education
- Newborn Screening
- Predictive and Presymptomatic Testing
- Prenatal Diagnosis

# 4

## Genetic disorders

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### What are genetic disorders?

Genetic disorders result from a change in the hereditary (genetic) material contributed by the parents at the time of conception of an individual. Symptoms may be apparent at birth or may occur later in life. Genetic disorders can affect growth, development and all aspects of health and include for example:

- cystic fibrosis
- Down syndrome
- fragile x syndrome
- haemochromatosis
- haemophilia
- Huntington disease
- neural tube defects
- muscular dystrophies
- neurofibromatosis
- phenylketonuria
- polycystic kidney disease
- spinal muscular atrophies
- thalassaemias

And some forms of:

- asthma
- cancer
- diabetes
- epilepsy
- heart disease
- hearing disorders
- hypothyroidism
- intellectual disability
- visual disorders
- short stature syndromes

### How common are genetic disorders?

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

- Cystic fibrosis – incidence 1 in 2500<sup>1</sup>.
- Down syndrome – incidence 1 in 770 pregnancies<sup>2</sup>.
- Duchenne muscular dystrophy – incidence 1 in 3300 males<sup>2</sup>.
- Fragile X syndrome – incidence 1 in 4000 males<sup>3</sup>.
- Haemophilia – incidence 1 in 10,000<sup>2</sup>.
- Neural tube defects (spina bifida, anencephaly, encephalocoele) – incidence 1 in 500-1,000<sup>2</sup>.
- Phenylketonuria – incidence 1 in 10,000<sup>4</sup>.
- Approximately 5% of cancers are inherited.

- Moderate to severe intellectual disability – incidence 1 in 400, 80% genetically determined.
- 1 in 20 people experience gene related illness, impairment, or disability by age 25.
- During their lifetime 50% of people have a disease with a genetic component.
- About half of the admissions to paediatric hospitals and 12% of adult admissions to general hospitals are for genetic disorders.
- Many chronic diseases of middle and old age are due to a combination of genetic and non-genetic factors.
- Genetic factors are a major cause of psychiatric illness.

### What can be done about genetic disorders?

Genetics services provide information, education, clinical, counselling and diagnostic services and support to individuals and families who are concerned about a disorder with a hereditary or genetic basis.

**Genetic counselling** for people at risk assists them to make informed choices about their health, lifestyle and reproductive options. It is essential before undertaking testing, to ensure that tests are conducted on an informed basis.

**Prenatal testing** is the process of detecting and diagnosing fetal abnormalities before birth. It is available for women or couples at high risk for more common disorders. Tests include ultrasound, amniocentesis and chorionic villus sampling. Screening blood tests are also used.

**Genetic carrier testing** may be available for people who have a family history of an inherited condition to determine if they are carriers of the faulty gene involved. This information may be useful in planning pregnancies. Gene mutation carriers usually do not show any signs or symptoms of the disorder. However, in the reproductive context, if both parents are mutation carriers, their offspring may be at risk of being affected.

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**Presymptomatic and predictive testing** for individuals at high risk can enable prevention of occurrence of some conditions, or minimise their impact through close monitoring and early treatment intervention.

**Newborn screening** and early intervention achieves significantly improved outcomes for babies born with phenylketonuria, galactosaemia, hypothyroidism and cystic fibrosis.

# 5

## Genetic health outcomes

In 1995 Genetics Services health objectives<sup>5</sup> were published with the aim of monitoring data collected by genetics services units and other facilities, to determine the impact of service provision. Following is an indication of progress in achieving these objectives and outcomes.

### Pregnancy and genetic disorders

#### Down syndrome

##### Outcome

Reduced impact of Down syndrome through improved awareness of, and access to a range of services including education, counselling, diagnosis and options to enable couples to arrive at informed decisions about pregnancy and genetic disorders.

Down syndrome has an incidence of about 1 in 700 pregnancies. It results from the presence of an extra chromosome 21, usually in all cells. The extra chromosome results from an incorrect division which arises mostly from an egg cell and occasionally a sperm. It is usually a chance event but this chance increases as the mother grows older.

#### Risk of a live born affected with Down syndrome

Years of age	Risk
20	1:1530
25	1:1350
30	1:910
35	1:380
40	1:110
45	1:30
50	1:6

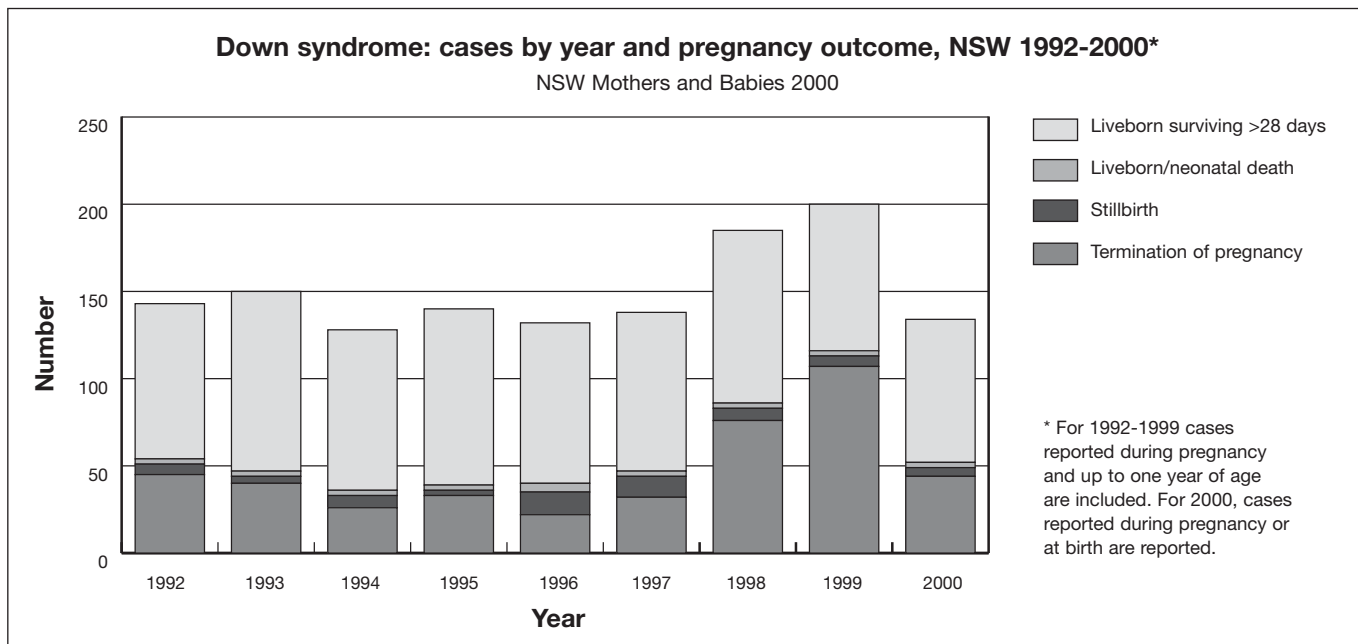
The number of women using prenatal diagnosis by cytogenetic testing between 1990 and 2000 has almost doubled from 3869 to 7070. 72% of women using cytogenetic testing are referred for reason of advanced maternal age (see Section 6 on Clinical genetics services and Section 11 Cytogenetics services). Women in this group tend to plan much wanted pregnancies and many use genetic counselling services to help them with their choices.

#### Confinements for women 35 years and over

Confinements	1990		2000	
	No	%	No	%
Maternal age 35+	8,974	10.4	15,334	17.7
Total all ages	86,499	100.0	86,460	100.0
No of women using prenatal diagnosis by amniocentesis or chorion villus sampling	3,869		7,070	

New South Wales Mothers and Babies 2000 and NSW Genetics Service

Reproductive patterns are changing so that the percentage of women confining at 35 years of age and older has increased from 10.4% in 1990 to 17.7% in 2000. This has the potential to increase the number of babies born with Down syndrome, highlighting the importance of genetic counselling, provision of information and prenatal testing. The following chart shows that the number of liveborn babies with Down syndrome does not appear to have increased during this time.



### Neural tube defects: Spina bifida, anencephaly and encephalocele

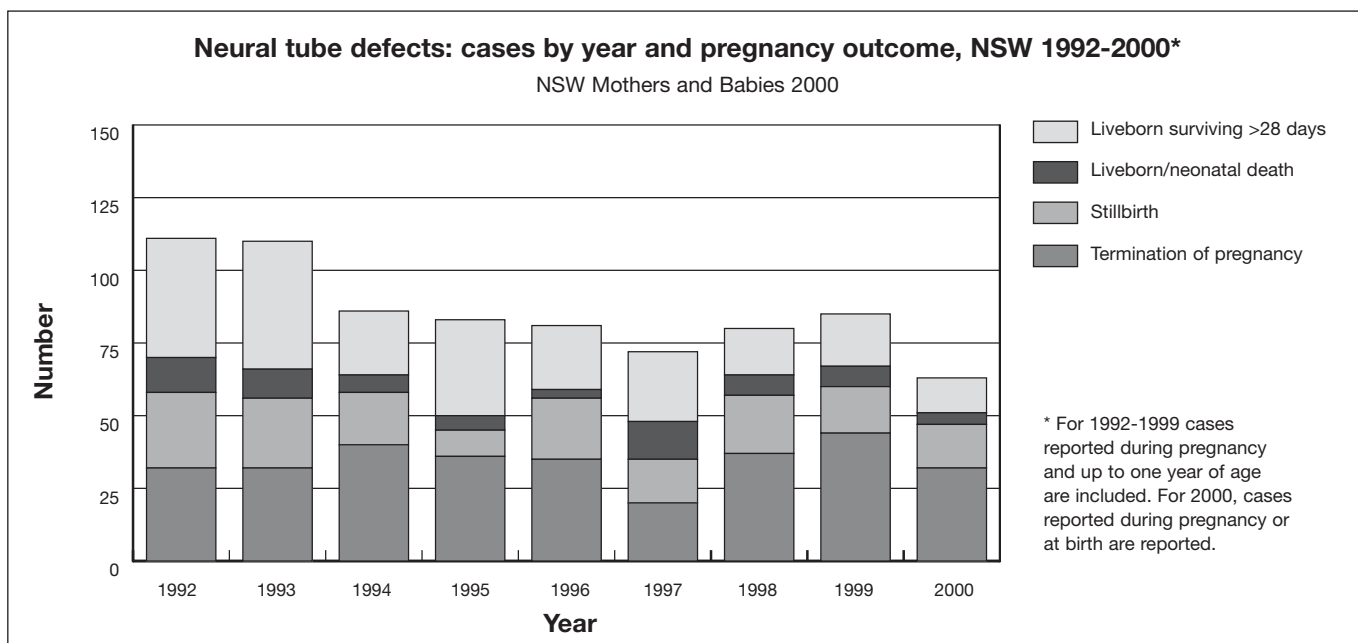
#### Outcomes

- Increased intake of folate among women of childbearing age.
- Reduced incidence of neural tube defects: spina bifida, anencephaly and encephalocele.

disability. At that time evidence from multicentre trials showed that increased intake of folic acid by women of childbearing age reduced the incidence of neural tube defects. A number of organisations including the NSW Health Department, the Commonwealth Department of Health and Aged Care and the food industry undertook a range of health promotional initiatives, including improving dietary intake of folic acid rich foods, promoting tablet supplementation and folate fortification of a number of breakfast cereals.

In the early 1990s, in NSW, about 110 children were born each year affected by neural tube defects. Most infants are stillborn or die early in life. The remainder usually have lifelong physical and often intellectual

Data from the NSW Birth Defects Register indicate a fall in reported cases from 111 in 1993 to 85 in 1999. Year 2000 data are incomplete.



## Newborn screening for genetic disorders

### Cystic fibrosis, phenylketonuria, galactosaemia, hypothyroidism

#### Outcome

Early diagnosis and immediate treatment reduces severe mental and physical impairment and mortality.

The NSW Newborn Screening Programme tests all babies born in NSW and ACT for several disorders, including phenylketonuria, cystic fibrosis, galactosaemia and hypothyroidism.

In 1999/2000 about 80 babies were diagnosed with one of these conditions and many more affected children were monitored. Early detection and intervention results in significantly improved outcomes.

As an example, phenylketonuria has a frequency of 1 in 10,000 births. Each year the Programme diagnoses about 8-10 infants with this disorder. Early treatment with a special milk formula and diet prevents severe mental retardation and results in normal growth and development.

#### New diagnoses April 1998-April 2001

Disorder	Number Diagnosed
Cystic Fibrosis	59
Galactosaemia	4
Hypothyroidism – primary congenital	60
Phenylketonuria	31
Other rare disorders	43
<b>Total</b>	<b>197</b>

NSW Newborn Screening Programme

## X-linked disorders

### Fragile X

#### Outcomes

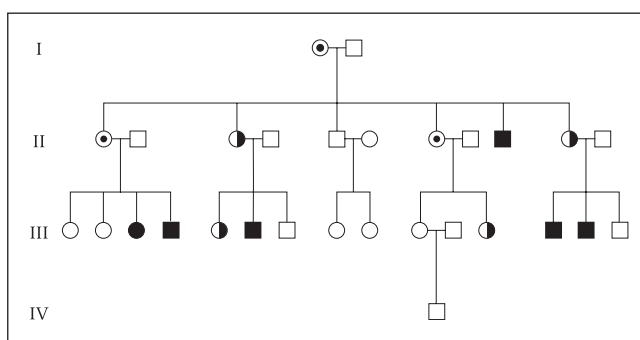
- Knowledge of carrier status allows informed choices enabling the possibility of having a child without fragile X.
- A 60% reduction in incidence of fragile X in NSW since 1986 from 1 in 4,000 to 1 in 10,000 through access to counselling and testing<sup>3</sup>.

The fragile X syndrome is a cause of moderate to severe intellectual disability and affected individuals might require lifelong extra educational help and supervision. The gene mutation is on the X chromosome. Both males and females can be unaffected but carriers.

The following pedigree illustrates four generations of a fragile X family. The first generation has a female unaffected carrier (circle with dot). This female carrier and her partner had six children – two unaffected carrier daughters, two daughters with some degree of learning disabilities (circles half black and half white), one son who is not affected (blank square) and one son with fragile X (black square). The third generation has a number of normal and affected individuals. With genetic counselling and testing it is possible for the third generation family members to have the opportunity to have fourth generation children who are not affected by fragile X intellectual disability.

The Fragile X Programme:

- runs a register of 245 extended families
- offers family members testing when they come of age
- helps arrange prenatal diagnosis
- arranges, on average, 12 prenatal tests per year
- studies and assists families with other forms of X-linked mental retardation



## Duchenne muscular dystrophy

### Outcomes

- Improved carrier detection.
- Reduction in unnecessary terminations of pregnancy from >50% to 2-5%.
- Reduced incidence of Duchenne muscular dystrophy in NSW between the mid 1960s and 1980s from 1 in 3,500 to 1 in 5,000<sup>7</sup>.

Duchenne muscular dystrophy is a progressive, profoundly disabling and terminal genetic disease of muscles. Affected males are usually diagnosed between 3 and 5 years of age and are wheelchair dependent by 13 years of age. Average survival is to the third decade of life.

At present there is no cure. It is inherited in such a way that it affects only males, with rare exceptions. It is usual that when a boy is diagnosed with Duchenne muscular dystrophy several of his female relatives are recognised to be possible carriers, meaning that although unaffected they may pass the disease on and have affected sons and grandsons.

The decreased incidence from 1 in 3,500 male births in the mid 1960s to 1 in 5,000 in the 1980s reflects the effectiveness of genetic counselling and DNA diagnostic testing since 1986.

Before the availability of prenatal DNA diagnostic testing, women who were known to be or likely to be carriers had very limited reproductive options. They usually remained childless or opted for fetal sex determination with termination of pregnancy if the fetus was male. This almost certainly meant that more than 50% of the pregnancies terminated were normal rather than affected. Today, with DNA testing, male fetuses can be tested to see if they are affected and the risk of terminating an unaffected pregnancy is very much diminished.

Advances in DNA technology mean that carrier mothers can be identified and prenatal diagnosis can be offered with a higher degree of accuracy. Of the 88 prenatal diagnostic tests performed between 1993 and 1998, 11 would not be necessary today as the mother has since been shown not to be a carrier.

## Single gene disorders

### Huntington disease predictive and prenatal testing

#### Outcome

- Knowledge of risk status in families with a history of Huntington disease allows informed choices to be made by individuals and couples regarding issues such as family, career and financial planning.

Huntington disease (Hd) is an autosomal dominant neurodegenerative condition with age of onset usually between 35 and 45 years (range 2–80 years) The incidence of Hd in the population is 6–7 per 100,000 and NSW has about 400 affected people. The ‘at risk’ estimate is 25.1/100,000. The Hd mutation, identified in 1993, is a trinucleotide repeat expansion on chromosome 4.

Predictive and prenatal testing for Hd is available for persons at risk for Hd aged 18 years and older. Because of the complexity of issues, Hd testing is best accessed through an expert team, including a clinical geneticist, genetic counsellor, neurologist and neuropsychologist/psychiatrist who can provide detailed information about the test; its limitations, possible outcomes and supportive pre and post result counselling. Ongoing assessment and management by a neurologist and psychiatrist is recommended.

Worldwide, the uptake of predictive testing is 15–20%, and uptake of prenatal testing is extremely low.

There is an established Huntington Disease Clinic at Westmead Hospital. Services are also available through genetic clinical and counselling units. The Huntington Disease Unit at Lottie Stewart Hospital in Sydney provides 18 beds and services. Residential and counselling services are also provided in the Hunter Area.

#### Predictive Testing Program participation 1993-2000

	Female	Male	Total
No. enrolled for predictive testing	383	405	788

**Results of Huntington disease testing 1993-2000**

	<b>Mutation +ve</b>	<b>Mutation -ve</b>	<b>Inter-mediate</b>	<b>Total</b>
No. of predictive test results	238	365	26	629
No. of prenatal test results	2	4		6

**Hereditary haemochromatosis****Outcome**

- Early diagnosis and treatment prevents organ damage and allows normal life expectancy.

Hereditary or genetic haemochromatosis is the commonest genetic disorder in Australia, affecting one in 200-300 people. One in eight to one in ten people is likely to be a carrier of the gene mutation. Carriers usually show no symptoms of the disease but may develop it if they have diabetes, are alcohol dependent or have some other triggering factor. Couples who are both carriers of the mutated gene have a 1 in 4 chance in every pregnancy of having a child who will develop haemochromatosis as an adult.

It is characterised by accumulation of iron in various organs which leads to conditions such as cirrhosis, cardiomyopathy and diabetes. Early diagnosis and treatment is important to prevent organ damage and allow normal life expectancy. However, it is under-diagnosed because of its late onset (average age of onset of symptoms is late 40s) and multiple non-specific clinical presentations.

Treatment consists of regular blood collection from the patient to reduce excess body iron stores. The collected blood can potentially be used for blood transfusion services.

Excess body iron is commonly due to haemochromatosis. Patients exhibiting symptoms would usually undergo fasting iron studies in the first instance. Where indicated by family history and other risk factors, gene mutation testing can be carried out. Mutation testing has only been available since 1997. The risk implications for other family members can then be ascertained and early diagnosis and monitoring can avoid development of more serious conditions.

**Genetically determined cancers**

About 5-10% of cancer is caused by inheritance of a genetic susceptibility<sup>8</sup>.

Genetic counselling and pedigree construction provides information to enable initial risk estimation. Decisions can then be made on the appropriateness of genetic testing for more accurate risk estimation on inheritance of mutations in cancer susceptibility genes.

People identified at high risk then have a basis for making informed choices about preventive and early detection strategies or prophylactic interventions.

**Breast cancer****Outcome**

- Reduced impact of genetically determined and partly genetically determined cancers.

Preliminary results from a survey show the effectiveness of genetic counselling in decreasing psychological distress in women at moderate or high risk of developing breast cancer. Genetic counselling was also shown to improve women's knowledge of breast cancer genetics and that this knowledge was retained in the long term.

**Colorectal cancer****Outcomes**

- Improved detection of families at risk through increased registration of families with familial adenomatous polyposis (FAP), hereditary non polyposis colorectal cancer (HNPCC) and other polyposis syndromes at the NSW and ACT Hereditary Bowel Cancer Registers and at family cancer clinics statewide.
- Genetic testing introduced for these syndromes.

There has been a 30% increase in registrations (families) since NSW & ACT Hereditary Bowel Cancer Registers were relaunched in 1998. Improved collaboration between the Registers and other state and international registers, as well as with family cancer clinics, has allowed multiple families, previously thought to be independent, to be linked. This can simplify the gene testing process, achieving savings in time with a concomitant reduction in anxiety, and also in costs.

## Genetic education

### Genetic education

#### Outcome

- Raised awareness of genetic disorders and genetic technologies to reduce impact of genetic disorders.

The NSW Genetic Education Program develops information, resources and educational programs for individuals affected by genetic disorders, the health professionals who serve them, community and school educators.

A 'Train the Trainer' concept was instituted as a means to maximise the use of limited resources in meeting the demand from teachers and students for information on the 'New Genetics'. In 1997 and 1998, six workshops were held for science teachers in NSW with 145 participants representing 68 government and independent schools.

Evaluation of workshops showed that over 95% of participants rated the workshops as very good. Overwhelmingly, participants considered the content of the workshops relevant to all teaching levels and 56% wanted the workshop to go on longer. The majority indicated improvement of their scientific knowledge and understanding of the implications of the genetic technology. Student responses to ethical scenarios ranged from interested to enthusiastic.

Importantly, 64% of teachers of Years 7-10 and 57% of teachers of Years 11 & 12 reported that they had changed the content of their teaching syllabus to incorporate human genetics material.

# 6

## Clinical genetics and genetic counselling services

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### **The genetic counselling process**

Genetic counselling is provided by a team which may include clinical geneticists, genetic counsellors and genetic social workers.

Genetic counselling provides an individual or family with current information and supportive counselling (advice or guidance) regarding problems in growth, development and health which may have a genetic basis. Counselling can assist families and individuals to understand and adjust to the diagnosis of a genetic disorder. A diagnosis of a genetic disorder may be made or confirmed in a pregnancy, after birth, in childhood or later in life. The diagnosis may be made on the basis of clinical features or laboratory analysis of tissue or blood, using cytogenetic, molecular genetic or biochemical genetic testing techniques.

A diagnosis may mean that other family members are at risk. During the consultation a family health history is taken. Where there is a genetic disorder in the family, genetic counselling may provide advice on the risks that other family members or future children will be affected.

### **Informed consent for genetic testing**

A key feature of genetic counselling is the provision of information to individuals, so that where genetic testing is an option, it can be undertaken on the basis of full information.

Because genetic disorders are family health problems, a diagnosis in one member has implications not only for that person but also for other family members. The shared implications of genetic inheritance raise specific ethical issues, which are different from those associated with other medical interventions. Further information is available in the *Ethical Code Governing the Provision of Genetics Services, 1998*.

Issues for consideration prior to consenting to testing include privacy, ownership of and access to information, responsibilities and obligations to family members and impact of a positive diagnosis on access to insurance and employment.

### **Data collection**

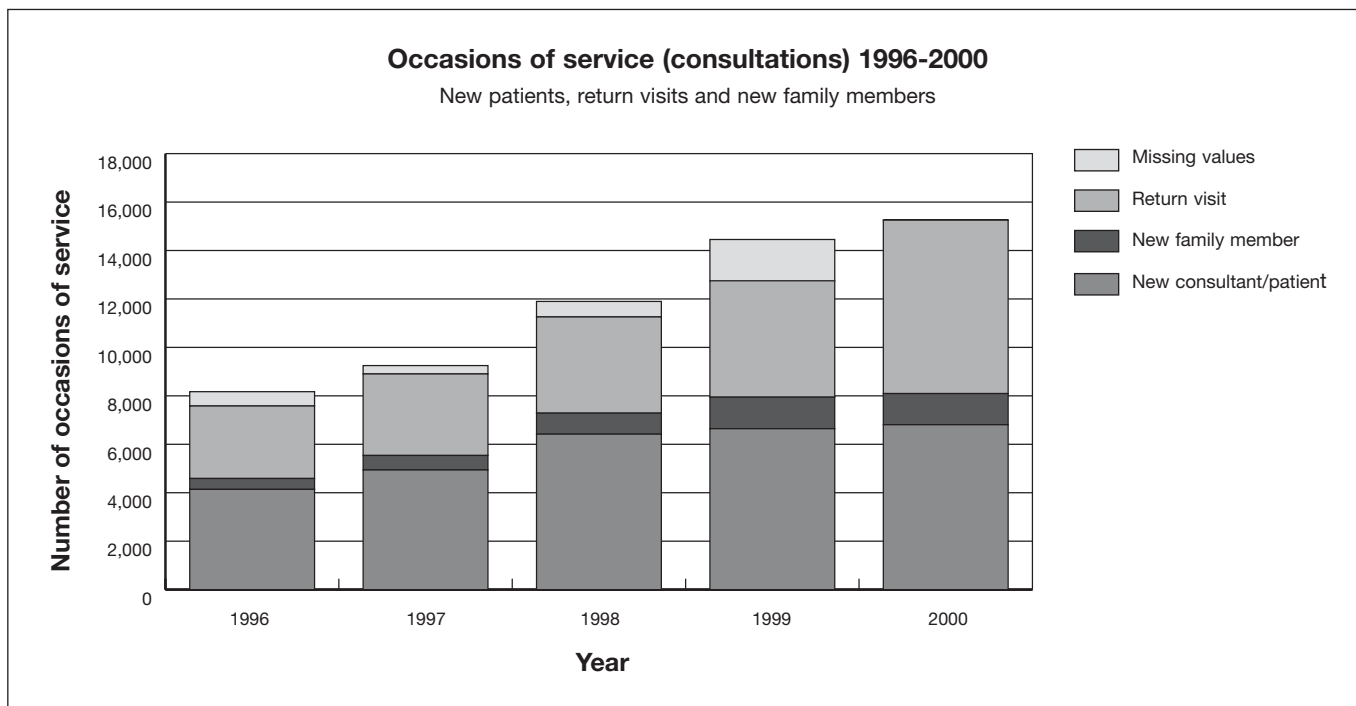
The development and installation of a genetics service database for all clinical and genetic counselling units has allowed the gathering of non-identifying data on service activity.

Data on service use for the 1996 to 2000 period are summarised in the charts on the following pages. The data indicate venue of service provision, most common reasons for referral, major diagnostic categories, referral sources and types of services.

The majority of cancer genetics services are provided by five specialised familial cancer services. Liverpool Hospital and Hunter Cancer Genetics Services gather service data using the clinical genetics database. Familial Cancer Services at Prince of Wales, Westmead, and St Vincent's Hospitals have collected data using different processes. Their data has been adjusted to fit existing data fields and should be considered as estimates. From the beginning of 2001, all cancer genetics services will record data in a uniform format consistent with the clinical genetics database.

## Service use

- The chart below shows growth in demand for genetics services. Reported consultations have almost doubled from 8,172 occasions of service in 1996 to 15,254 in 2000.
- In 2000, 45% of patients were new referrals, 47% return visits and 8% new family members.

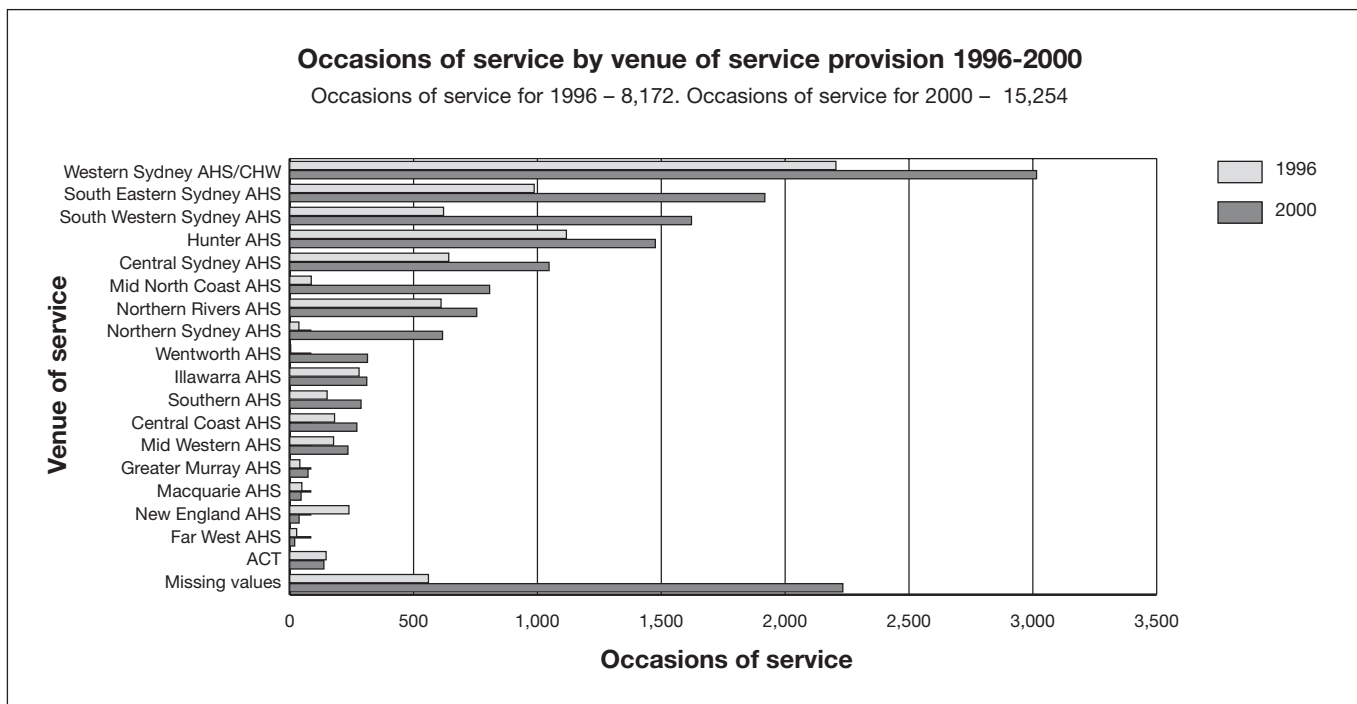


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**Note 2:** Two units (Central Sydney and Northern Rivers) were unable to provide 2000 data. Figures for this year have been estimated. For Central Sydney this is likely to be an over-estimate as staff changes in 2000 resulted in reduced services during the year.

## Venue of service provision

- As indicated in the chart below growth is evident in all venues of service provision, particularly in the larger and more well established units in Western Sydney/Children’s Hospital at Westmead (CHW), South Eastern Sydney and Hunter.
- New services commenced between 1996 and 1999 in Central Sydney, South Western Sydney, South Eastern Sydney at St George Hospital, Wentworth and the Mid North Coast.
- Data for the ACT represent patients seen by NSW Genetics Service staff. The ACT has now established its own Genetics Service. Its data are not included here. Flows in and out of NSW and ACT are approximately equal.



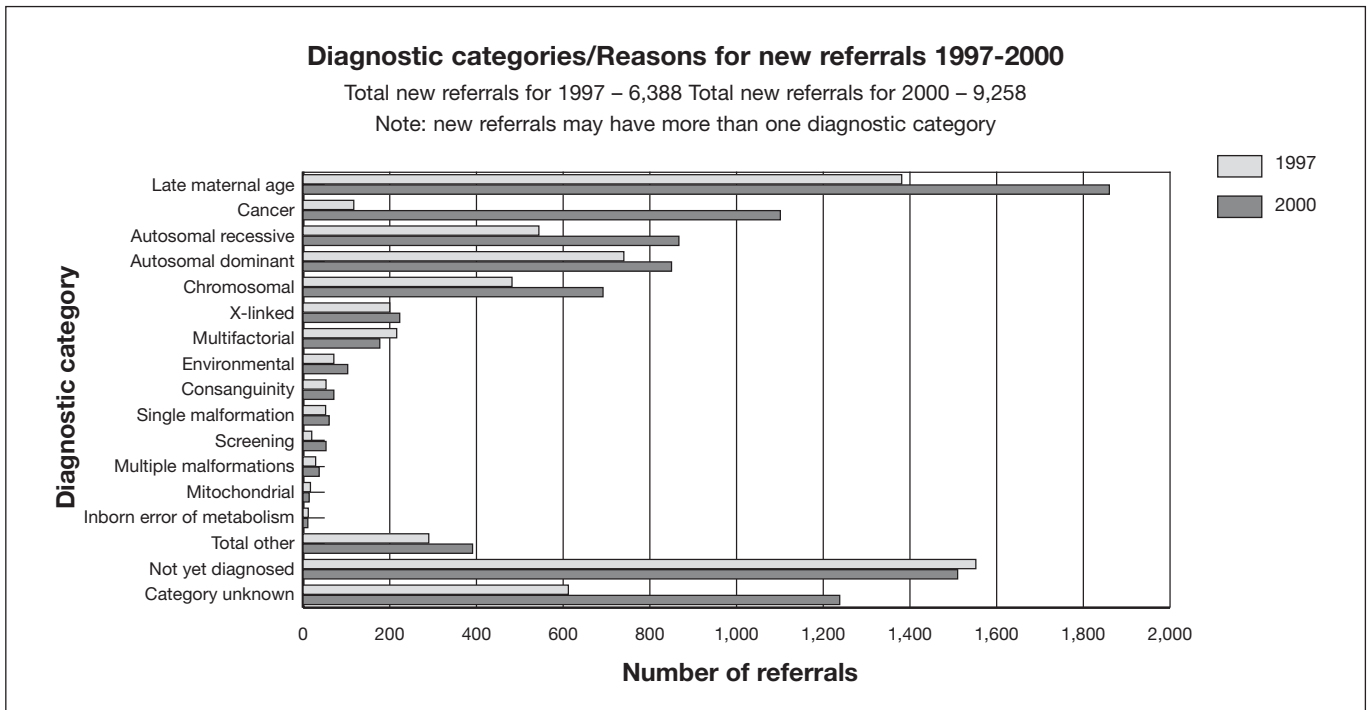
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**Note 3:** Cancer Genetics Services. Data on cancer genetics services provided by the Familial Cancer Services at Prince of Wales, Westmead, and St Vincent’s Hospitals have been collected using different processes and adjusted to fit existing formats. Most of the missing values (2,174) are cancer genetics services provided at Western Sydney AHS and South Eastern Sydney AHS. The venue of service provision was not recorded by these units.

## Most common reasons for referral

- Genetic conditions are classified according to standard genetic diagnostic categories listed below as well as by their syndrome or condition name.
- The following chart shows most common reasons for referral according to standard genetic diagnostic categories.
- Growth is apparent in most categories, particularly cancer and late maternal age.
- Cancer Genetics Services were introduced in the mid 1990s and new referrals have grown rapidly since that time.

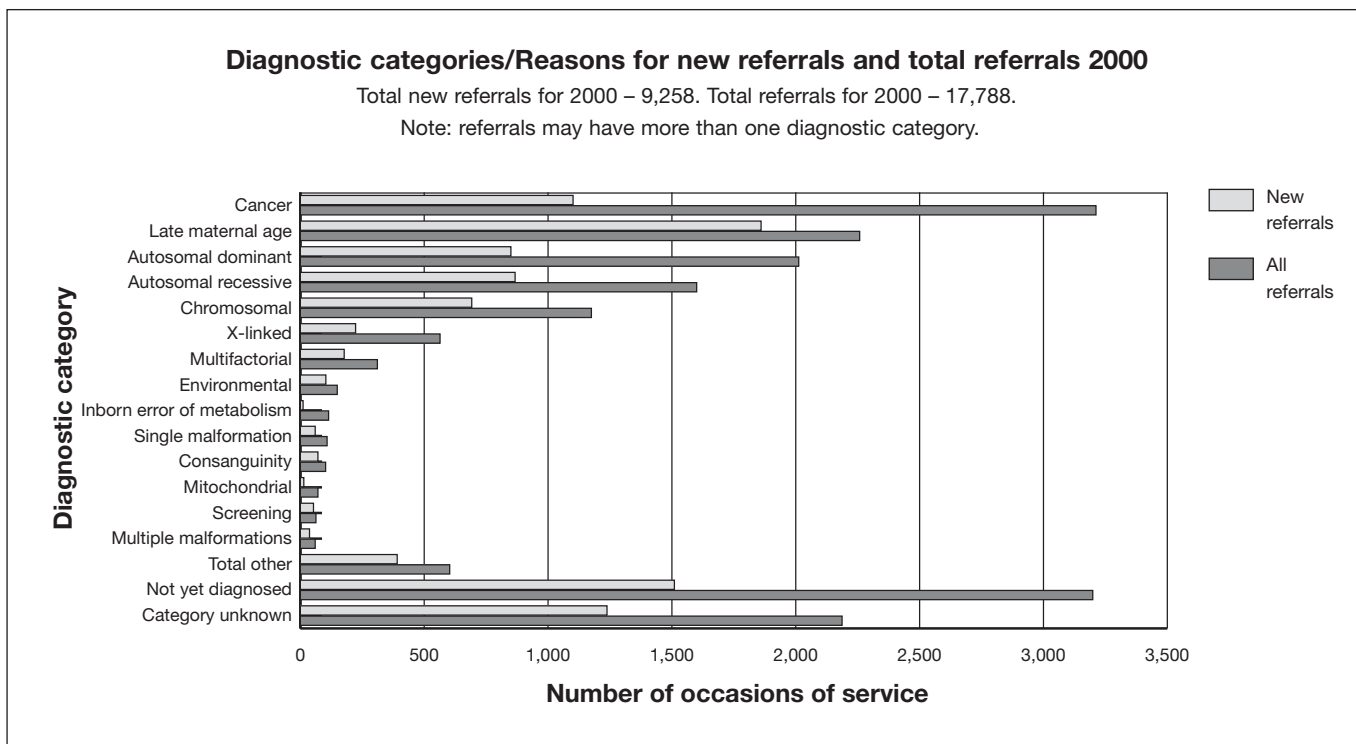


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**Note 3:** Cancer Genetics – Data on cancer genetics services provided by the Familial Cancer Services at Prince of Wales, Westmead and St Vincent's Hospitals have been collected using different processes and adjusted to fit existing formats.

- This chart indicates that cancer is the most common reason for service consultation. The complexities of cancer genetics mean that two to three consultations per patient are necessary
- Generally referrals for late maternal age require only one consultation.

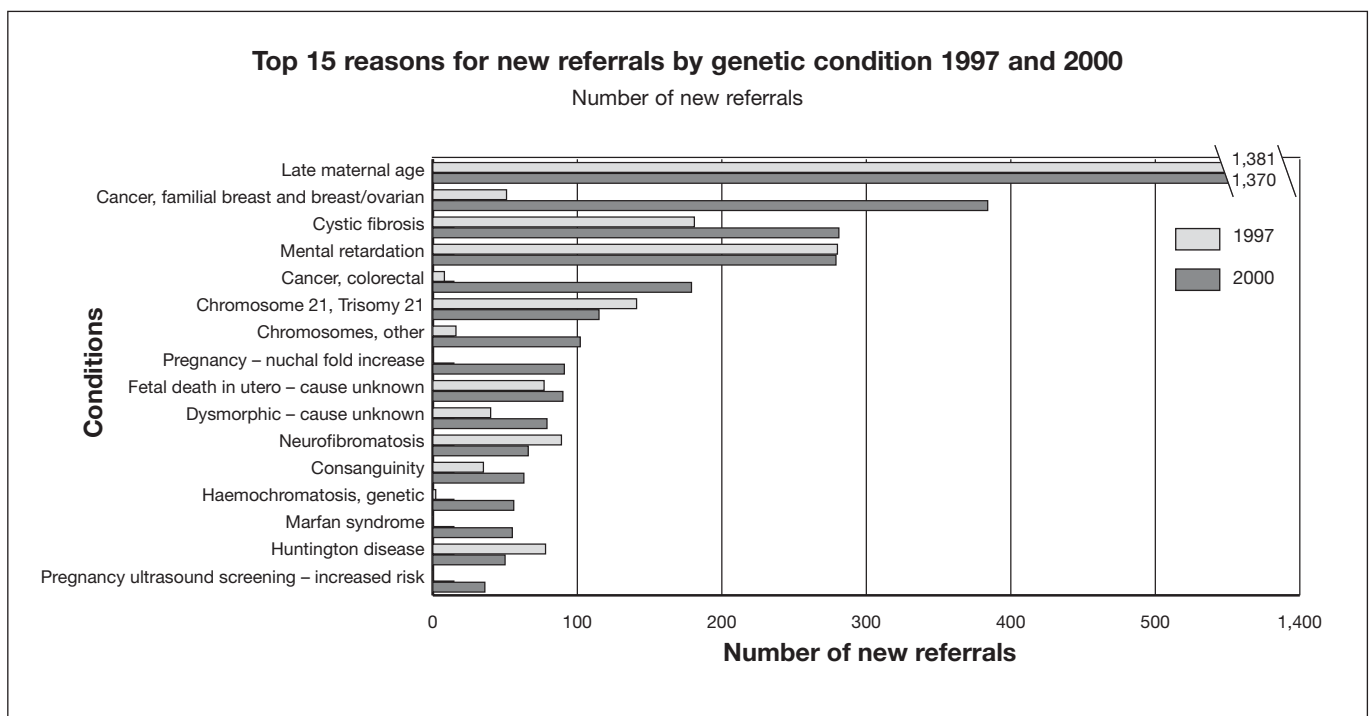


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**Note 3:** Cancer Genetics – Data on cancer genetics services provided by the Familial Cancer Services at Prince of Wales, Westmead and St Vincent’s Hospitals have been collected using different processes and adjusted to fit existing formats.

- As well as classifying genetic conditions to a limited number of diagnostic categories as in the previous two charts, conditions are also coded under their condition name or syndrome.
- Every unit reports its top ten reasons for referral by genetic condition which are amalgamated to produce the top 15 shown in the chart below. The data shown do not represent total referrals, as data which are not included in each unit's top ten reasons are not counted.
- Growth has occurred in most categories, particularly referrals for more newly diagnosable conditions such as familial breast, breast/ovarian cancer and colorectal cancer.
- Recent advances in antenatal ultrasound techniques, particularly the detection of increased nuchal translucency are reflected in the increasing number of referrals of women with increased risk pregnancies
- Genetic haemochromatosis is also attracting a growing number of referrals. Many more people with genetic haemochromatosis would be seen outside genetics units.



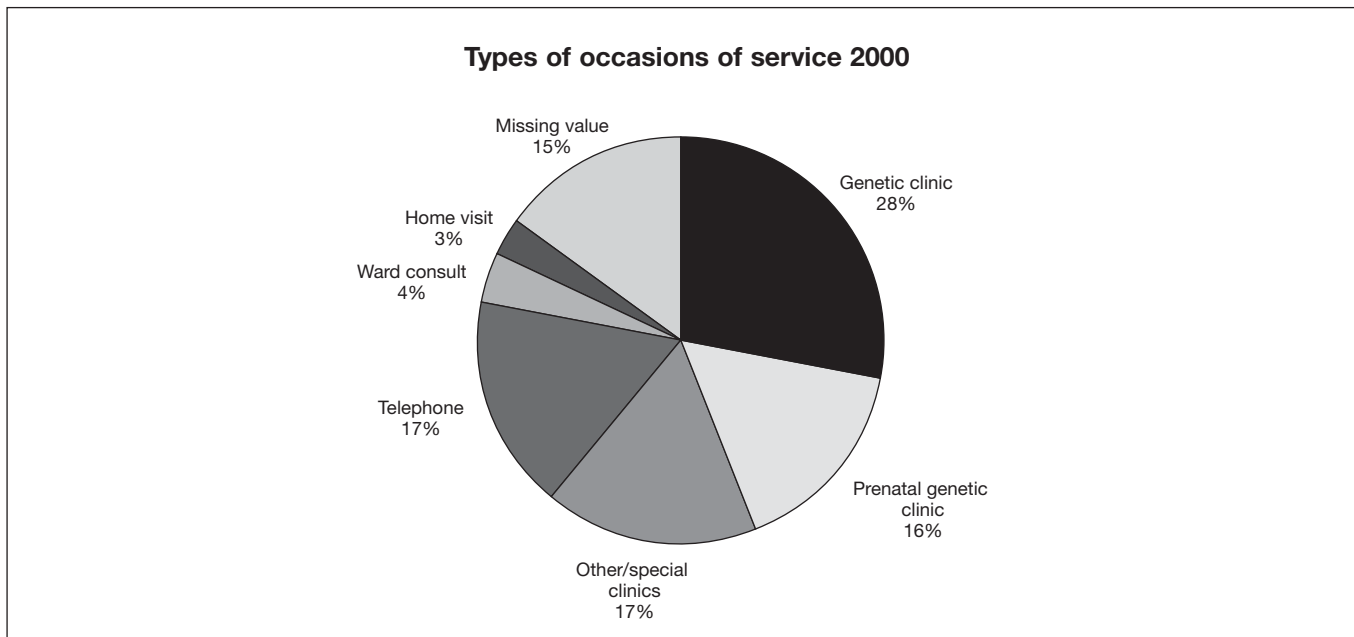
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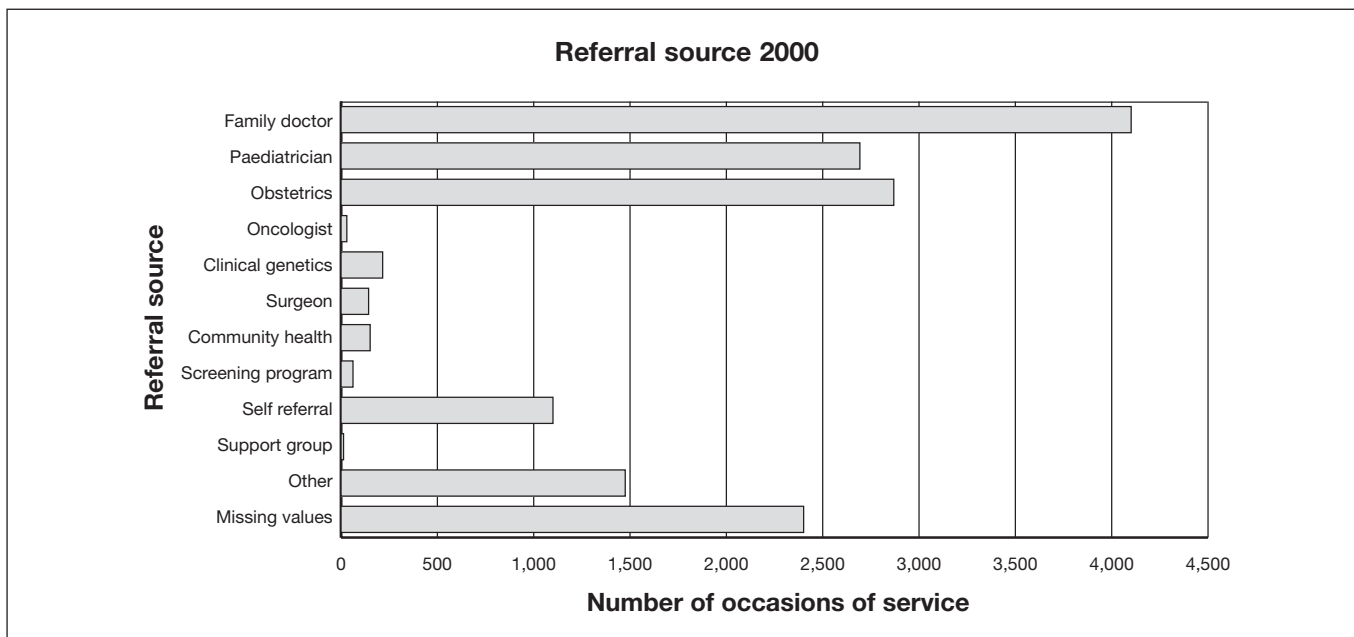
## Types of services

- 60% of consultations or occasions of service are held in clinics on a non-inpatient basis.
- 17% of consultations are by telephone. Telephone consultations of 15 minutes duration and of therapeutic value are counted as occasions of service.
- Telephone consultations have an important role in eliciting relevant information prior to the clinic visit and for consultant follow up concerning the understanding, assimilation and management of complex information.



**Note:** The majority of missing values are cancer genetics services provided by Western Sydney AHS and South Eastern Sydney AHS and would be cancer clinic visits and telephone consultations

## Referral sources



**Note:** The majority of missing values are cancer genetics services provided by Western Sydney AHS and South Eastern Sydney AHS. Referral sources are most commonly family doctors and oncologists.

## Length of service

- Typically a genetics consultation lasts between 30 and 70 minutes with an average of 65 minutes.

## Age and sex of consultands

- The greatest frequency of consultands is those of reproductive age – 25 to 44 years. The next most frequent patient age group is the 0 to 24 years group. Over 45 year olds are less frequent users of the service according to the data presented. This is an underestimate for this age group as age data for cancer genetics services are not included.
- Women are the most likely consultands to access services. In most instances they are consulting for themselves, although frequently they are the parents of a child who is affected with a genetic disorder.

## Interpreter use

- Data are incomplete but from data reported, interpreters are used most frequently in Central Sydney and South Western Sydney.
- Most commonly used languages excluding English, are Mandarin, Vietnamese, Cantonese and Arabic.

# 7

## Cancer genetics services

### Cancer genetic conditions

5–10% of cancers are considered to be caused by inheritance of genetic susceptibility. These include:

#### ***Inherited breast and/or ovarian cancer***

About 5% of all individuals with breast or ovarian cancer are thought to have inherited, from a parent, a mutation in one copy of the genes controlling cell division and growth in breast and/or ovarian tissue. There are several different genes which have recently been identified as important in the development of breast and/or ovarian cancer: the breast cancer 1 gene (BRCA1) and the breast cancer 2 gene (BRCA2).

#### ***Inherited bowel cancer***

Two types of bowel cancer FAP (familial adenomatous polyposis) and HNPCC (hereditary non-polyposis colorectal cancer) contribute to about 2–5% of all bowel cancer.

### Cancer genetic services

**Genetic counselling**, often in conjunction with specialist cancer services, is available to individuals concerned about their risk because of a strong family history of these cancers. For individuals assessed to be at high risk, counselling can help with strategies for reducing risk of developing the cancer or appropriate management and surveillance of the disorder.

**Molecular genetic testing** may be possible for some high risk individuals, in conjunction with genetic counselling, to identify those individuals who have a mutation in one of the genes known to be involved in the development of the cancer. Testing is only an option when the gene has been isolated or closely linked markers are known and the family structure is appropriate.

### Development of cancer genetics services

The benefits of new advances in cancer genetic technology have been made available through the establishment of specialised cancer genetics services or Familial Cancer Clinics in South Eastern Sydney, Western Sydney, South Western Sydney and Hunter Area Health Services. Generally services are provided jointly by Clinical Genetics and Oncology Departments.

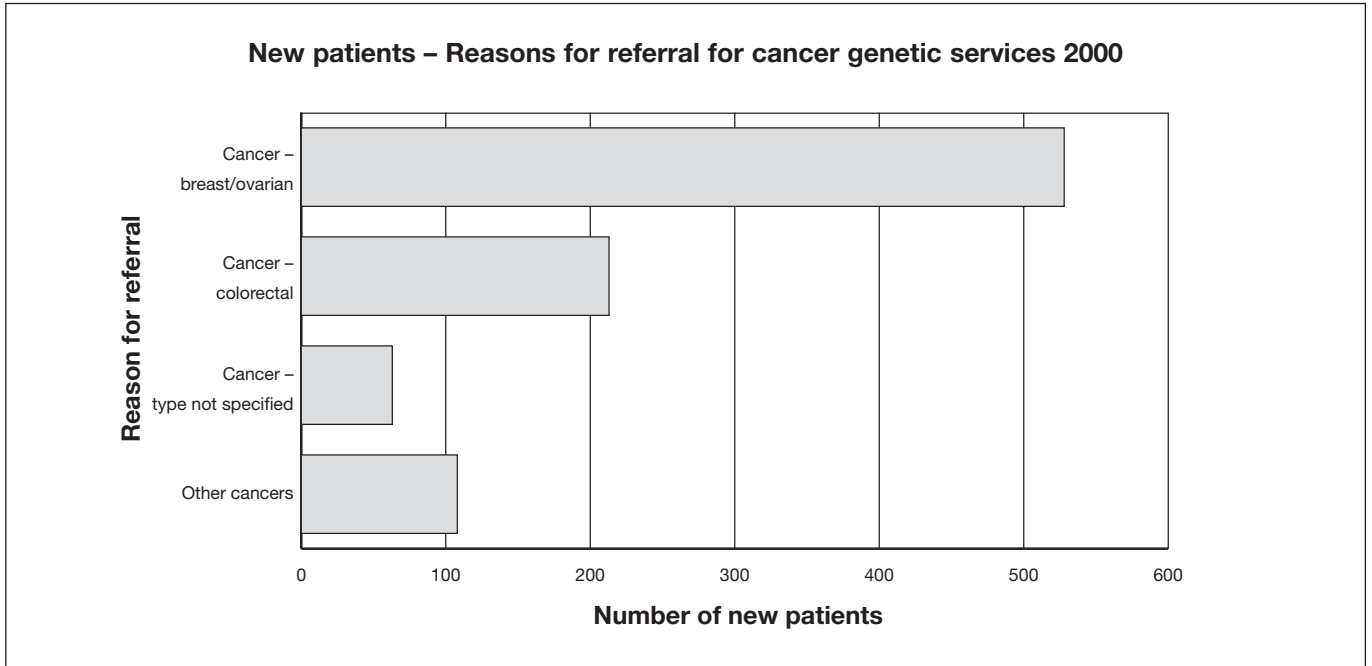
In addition to specialised Familial Cancer Clinics, cancer genetics services are available through all metropolitan and outreach genetics units.

### Statistics from cancer genetics clinics

In 2000 a total of 3,212 occasions of service for cancer were recorded, 1,101 of these were new referrals. The majority of services were provided through the specialised cancer genetics services (2,853 total services of which 931 were new services) and the remaining services were recorded by general Genetics Units in the State. There is likely to be a small underestimate of total cancer services provided as some units have coded cancer under autosomal dominant.

### Laboratory testing statistics

Type of cancer test	1999/2000
Ashkenazi screen	49
BRCA1&2 screen	215
BRCA1&2 predictive	27
Von Hippel Lindau	19
Familial Adenomatous Polyposis (FAP) screen	37
Familial Adenomatous Polyposis (FAP) predictive	49
Hereditary Non-Polyposis Colorectal Cancer (HNPCC) screen	94
Hereditary Non-Polyposis Colorectal Cancer (HNPCC) predictive	52
CDKN2A predictive	9
<b>Total</b>	<b>551</b>





# The NSW Newborn Screening Programme

A few diseases that are severe, without specific early symptoms and treatable, can be detected by tests carried out on dried blood samples obtained by heelprick. The NSW Newborn Screening Programme tests all babies born in NSW and in ACT for a number of such disorders. Around 94,000 tests are carried out each year.

Until 1998 the Programme tested for five conditions:

Disorder	Testing started	Analyte	Frequency
Phenylketonuria	1965	Phenylalanine	1:10,000
Hypothyroidism	1977	TSH	1:3,500
Cystic fibrosis	1981	IRT/DNA	1:2,500
Galactosaemia	1983	Gal metabolites	1:40,000
Homocystinuria	1994	Methionine	1:60,000

The screening laboratory offers a monitoring service for patients with PKU, who have blood tests monthly, or, in the case of patients who are pregnant, weekly. Over 300 patients are monitored in this way.

In 1998 new technology was introduced, using tandem mass spectrometry to test for groups of compounds, so that a number of extremely rare disorders can be tested for in a single operation.

## Diagnosis of rare disorders by Tandem Mass Spectrometry April 1998-April 2001

<b>Babies screened</b>		<b>275,000</b>
Resamples requested (0.13%)		357
Samples referred to Biochemical Genetics (from 105 babies). This represents 1.4% of the total samples received by Biochemical Genetics		226
<b>Detected</b>		<b>74</b>
PKU	31	
BH4 def	2	
Other amino acidurias	6	
Organic acidurias	8	
MCAD deficiency	12	
Other fatty acid oxidation	8 plus 2 affected siblings	
Secondary (B12 def, hepatitis)	7	
<b>Positive predictive value</b>		<b>21%</b>
<b>Undetected</b> (CbIC, Tyr 1, NKHG, GA1) (patients born during the period, whose screening test result was within the normal range)		4
<b>Other diagnoses</b>		<b>123</b>
Detected		
Cystic Fibrosis	59	
Galactosaemia	4	
Hypothyroidism – primary congenital	60	

# The NSW Biochemical Genetics Programme

# 9

Biochemical genetic testing can identify errors in the body's metabolism or its chemical processes. Testing can be done on the fetus during pregnancy, in the newborn period ie *newborn screening*, or later in life. The laboratory provides a statewide service and employs complex analytical techniques requiring in-depth knowledge of human biochemistry for interpretation of results.

A wide range of investigations is available including amino acid and organic acid profiles, quantitative assays of specific metabolites, diagnostic enzymology and molecular biology. The laboratory has had a long-standing interest in the disorders of fatty acid oxidation and carnitine transport and maintains an active program for developing assays in this area.

The clinical geneticist and senior scientific staff provide an advisory service to clinicians and other laboratories about the investigation of suspected inborn errors of metabolism in sick babies, children and adults. Comprehensive and up-to-date information is available concerning diagnosis, treatment, management and prenatal diagnosis of inborn errors.

## New diagnoses 1999-2000

	1999	2000
Urea cycle	1	8
Organic acid	12	10
Transport	9	5
Amino acid	13	6
Storage disorder	10	8
Fatty acid oxidation	8	12
Others	3	1
<b>Totals</b>	<b>56</b>	<b>50</b>

Many, though not all, fetal abnormalities can now be diagnosed prenatally. Prenatal diagnostic services are provided in conjunction with genetics services by fetal medicine units in South Eastern, Central, Western, Northern and South Western Sydney, Wentworth and Hunter, and some outreach areas.

### **Information and counselling**

Maternal serum testing, screening ultrasound, nuchal translucency testing alone, and nuchal translucency testing combined with first trimester serum screening are used during pregnancy to determine risk of abnormalities such as Down syndrome. These tests are risk estimates, not definitive tests. This concept requires careful explanation because of the possibility of false positive and false negative results. Ultrasound or maternal serum testing are not threatening to the pregnancy, but an increased risk result leads to the option of more invasive testing by amniocentesis or CVS which does carry a small procedure related risk.

Before amniocentesis and CVS are offered, women should be advised about the risks to the pregnancy associated with these procedures, and the time lag between testing and receiving results. The main reasons for prenatal diagnostic testing using amniocentesis or CVS are:

- where a woman is having a baby when she is in her mid thirties or older
- having a family history of a person with a serious disorder or of being a 'carrier' for a mutation
- where one of the partners in a couple has a serious disorder which may be passed on to a baby
- having a previous child affected by a serious problem in growth, development and/or health
- where the results of tests such as the maternal serum test have determined that the woman is at an increased risk for having a baby with a particular disorder.

### **Prenatal tests**

#### ***Nuchal translucency measured by ultrasound***

Nuchal translucency measurement is a targeted form of ultrasound. An increase in nuchal translucency between 11½ and 13½ weeks is known to be associated with babies at increased risk of a chromosomal abnormality such as trisomy 21 (Down syndrome), trisomy 13 and trisomy 18. This is a risk assessment rather than an absolute diagnosis. About 70–80% of babies with Down syndrome will be identified as being at 'increased risk' and about 20–30% of affected babies will be missed.

#### ***First trimester serum screening***

The nuchal translucency screen may be combined with a biochemical first trimester screening test which improves the detection rate to 80–90%.

#### ***Maternal serum testing***

The maternal serum test is available to women between 15 and 17 weeks of pregnancy to assist in the prenatal identification of several birth defects including Down syndrome and neural tube defects. This blood test, combined with age and other factors can provide an estimate of risk that the baby is affected. It is not a diagnostic test. Women identified at high risk usually follow up with more definitive diagnosis by amniocentesis or ultrasound. It is estimated that this process will lead to the detection of 60% of Down syndrome affected pregnancies. Results are not usually available until 17 to 19 weeks of pregnancy.

#### ***Ultrasound***

Ultrasound or fetal imaging can be conducted at any time during the pregnancy. The optimal time for dating the pregnancy or determining the number of babies is between 8 and 12 weeks. A fetal anomalies scan to check the baby's physical development is best done between 18 and 20 weeks.

***Chorionic Villus Sampling (CVS)***

A small amount of the placental tissue is sampled either vaginally or abdominally guided by ultrasound scanning. The procedure is undertaken during the 11th to 14th week of pregnancy and sometimes later. Approximately 1% of the procedures are followed up by an amniocentesis when tissue is inadequate or the result is equivocal. The result is usually available within 18 days. There is about a 1% risk of loss of the pregnancy due to the procedure.

***Amniocentesis***

A sample of amniotic fluid is withdrawn by a needle through the abdomen during the 14th to 18th week of pregnancy. The result is available within 18 days. There is a less than 1% risk of loss of the pregnancy due to the procedure.

**Prenatal testing results**

The results of prenatal cytogenetic and molecular genetic testing after amniocentesis or CVS are discussed on the following pages. Ultrasound and maternal serum testing are a routine part of obstetric care and service data are not collected for inclusion in this report.

## Prenatal cytogenetics services

Cytogenetic testing is a process used to identify any change in the usual number or structure of an individual's chromosomes and may aid in the diagnosis of a genetic disorder.

Prenatal cytogenetic diagnosis is available to women at increased risk of having a child with a chromosomal disorder. The majority of referrals are for women over the age of 35 years, who are at higher risk for Down syndrome and other chromosomal abnormalities for reason of advancing maternal age (see Section 5 page 8). Other major reasons for referral include women whose pregnancies are found to be at increased risk on ultrasound or maternal serum testing, and women with a previous child or family history of a chromosomal/genetic disorder.

### Chromosomal disorders

The most commonly occurring chromosomal disorders are:

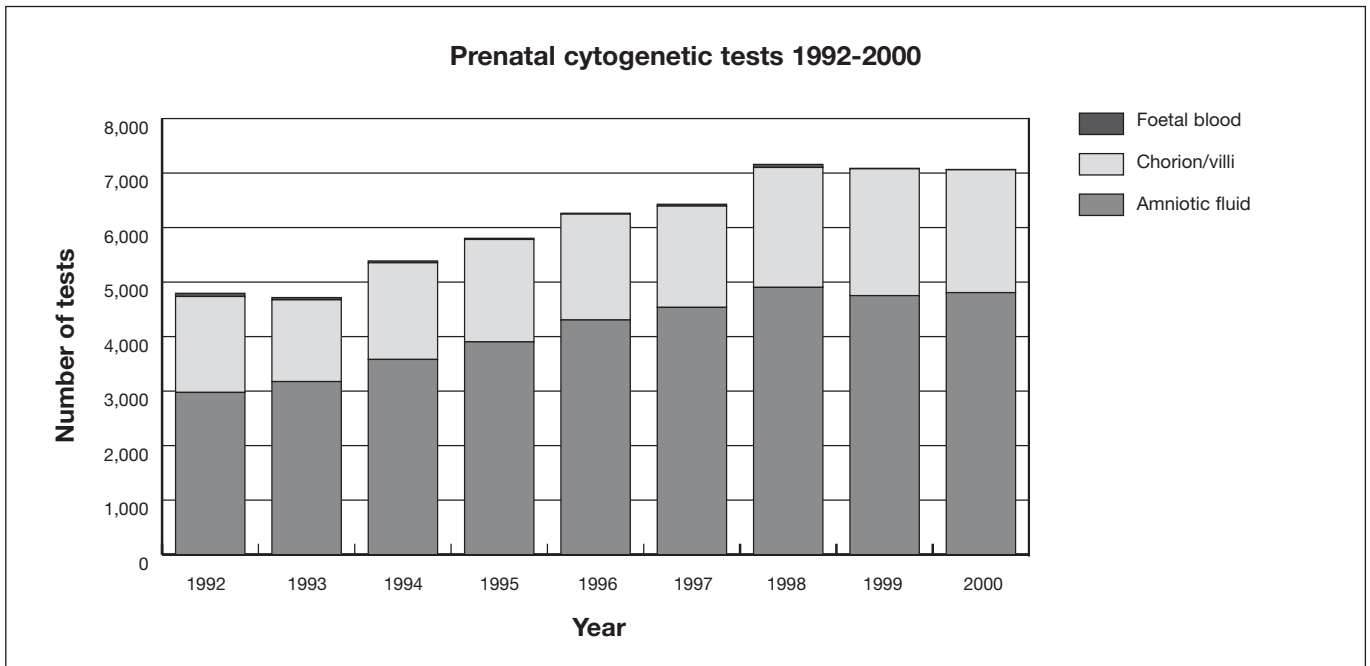
- **Trisomy 21/Down syndrome** – Incidence is about 1 in 770 births. Congenital heart disease is common and is a major cause of early death. Mild to moderate mental retardation is usual.
- **Trisomy 13** – Incidence is about 1 in 8,000 births. All patients are profoundly retarded. Half the babies born die by one month of age and others rarely survive more than 3 years.
- **Trisomy 18** – Incidence is about 1 in 6,600 births. All patients are profoundly retarded. 30% die by 1 month, 90% by 1 year and 99% by 10 years.
- **Klinefelter syndrome** – Incidence is about 1 in 500 males. Intelligence is usually normal. The majority of Klinefelter syndrome adults will be infertile and male hormone therapy may be required.
- **Turner syndrome** – Incidence is about 1 in 2,000 females. Intelligence is usually normal but there may be specific learning difficulties. It is characterised by short stature, possible infertility, and lack of sexual maturation at puberty. There are a number of potential physical problems such as heart defects.

### Prenatal cytogenetic testing data

Non-identifying data for the State have been collected since 1992 from laboratories located in South Eastern Sydney (SEALS), Northern Sydney (PaLMS), Western Sydney Genetics Program (Children's Hospital, Westmead), Hunter Area (HAPS); and one private laboratory (Sydney Genetics). Data have been evaluated to provide a comprehensive picture of service utilisation and outcomes of testing.

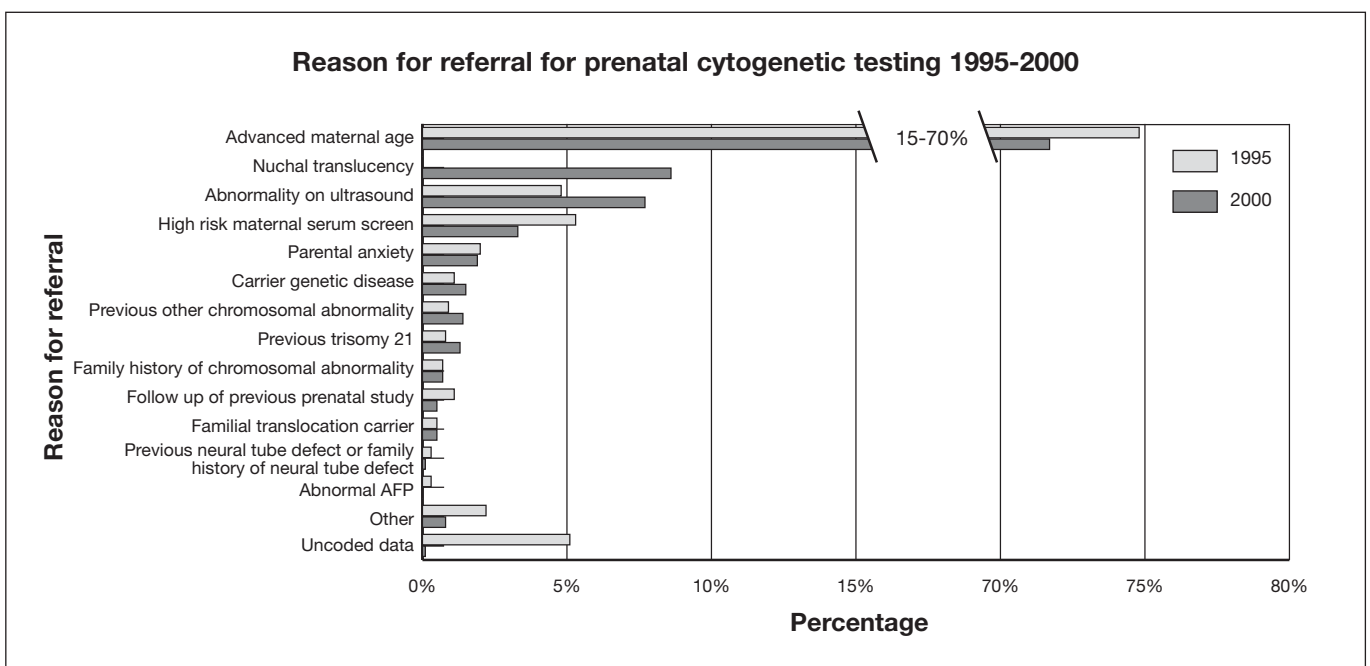
### Prenatal cytogenetics test use

- The chart opposite shows that referrals have grown at a rate of approximately 10% per annum, and have more than doubled since 1989 to 7,070 test requests in 2000.
- Amniocentesis is used twice as frequently as chorionic villus sampling (CVS).
- A levelling off in numbers of tests occurred between 1998 and 2000. This coincides with the introduction in 1997/98 of nuchal translucency measurement in ultrasound to detect increased risk of chromosomal abnormalities such as Down syndrome. Data on the total numbers of nuchal translucency measurements or ultrasounds are not available but their use is known to be growing significantly.



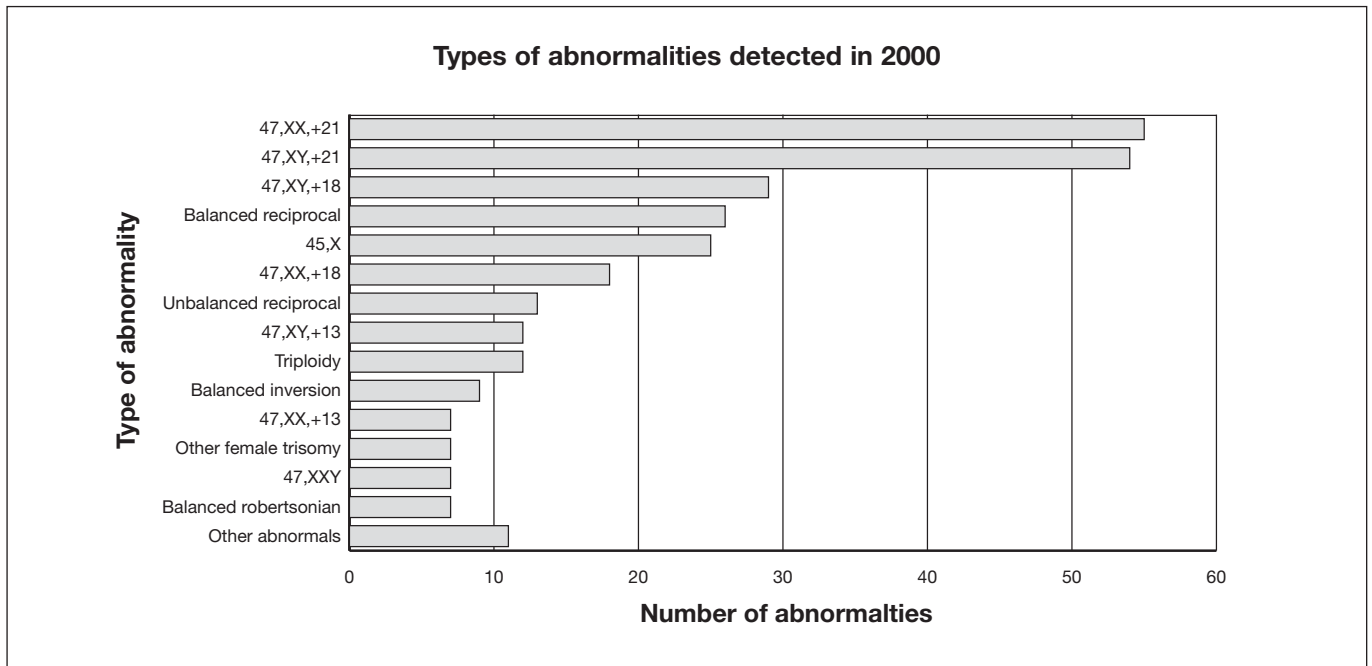
## Reason for referral for prenatal cytogenetic testing

- Cytogenetic testing through amniocentesis and CVS has been available since the 1970s to women of advanced maternal age who are at greater risk of having a child with Down syndrome or other cytogenetic abnormality (see Section 5, page 8). Advanced maternal age is the most common reason for referral for cytogenetic testing. In 1996 referrals for advanced maternal age reached a peak of 82%, compared with a 75% rate for 1995 and a decline to 72% in 2000.
- This decline has been offset by an increase in referrals for abnormality on ultrasound to 8% in 2000. A further 9% of referrals arose from increased nuchal translucency measurement introduced in 1997/98, making a total of 17% for both categories. These data may reflect women's preferences for ultrasound and nuchal translucency measurement over the more invasive procedures of amniocentesis and CVS.



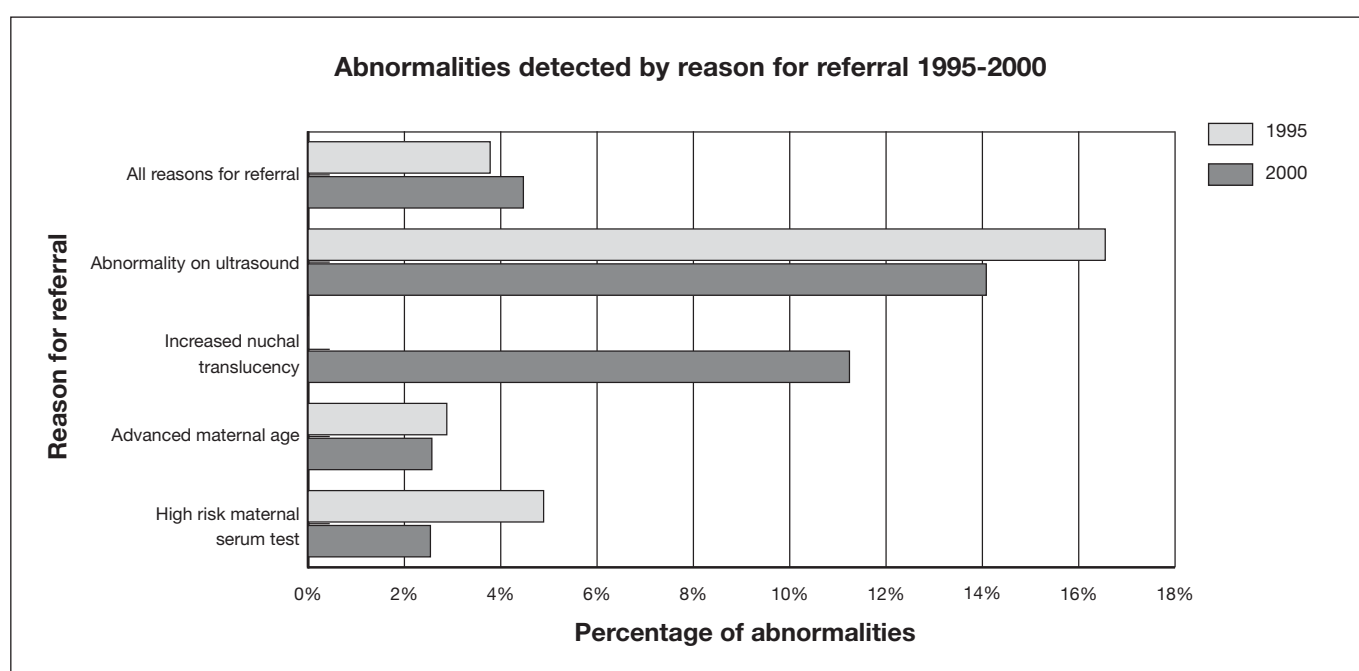
## Types of abnormalities detected by prenatal cytogenetic testing

- Trisomy 21 or Down syndrome comprised 109 (34%) abnormalities detected; 54 were male trisomy 21 (47,XY,+21) and 55 female trisomy 21 (47,XX,+21).
- Trisomies 21, 18, 13 and other made up 59% of the abnormalities detected.



## Abnormalities and reason for referral

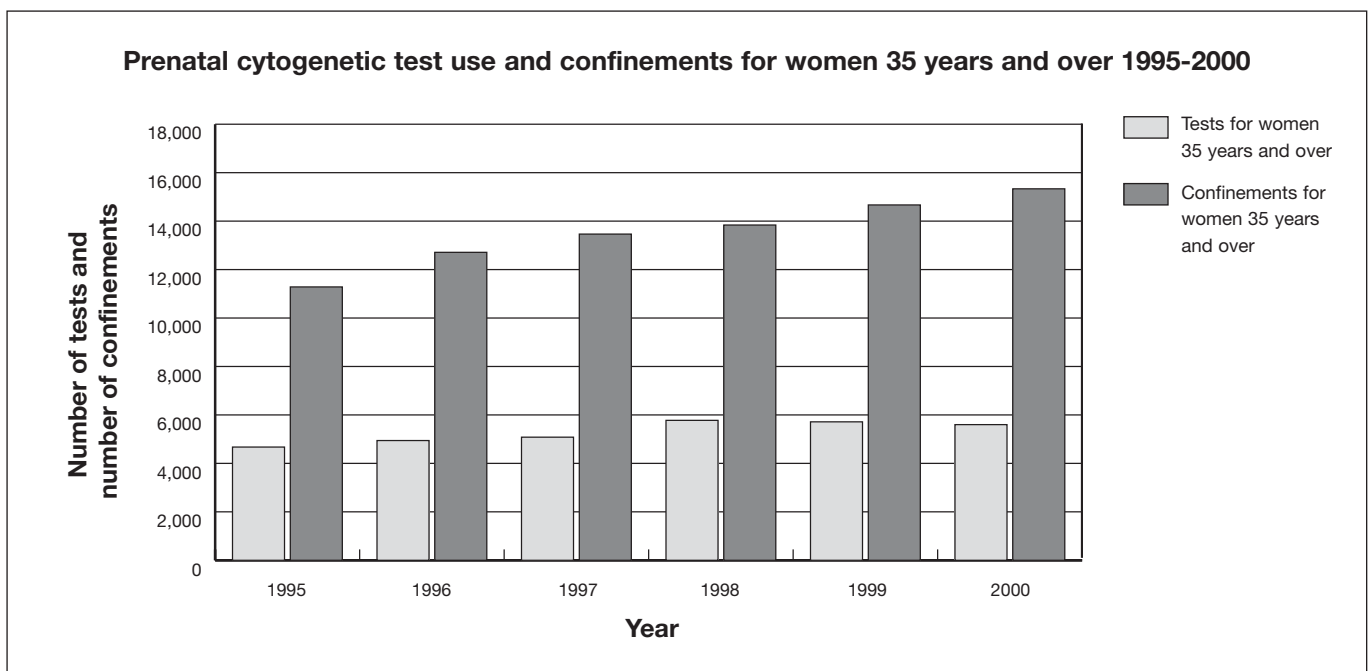
- The chart and table below show that the detection rate for abnormalities for *all reasons for referral* has increased from 3.78% in 1995 to 4.47% in 2000. This is likely to be the result of growth in referrals for abnormality on ultrasound and increased risk on nuchal translucency measurement.
- In 2000 the highest abnormality detection rate was 14% for those patients referred because of *fetal abnormality on ultrasound* (77 abnormalities among 547 referrals in 2000). During 1998, laboratories began to record referrals for increased nuchal translucency separately from other ultrasound abnormalities. The detection rate in 2000 for abnormalities for patients referred for increased nuchal translucency risk was 11% (68 abnormalities among 605 referrals).
- The most common reason for referral, ie *advanced maternal age* has the lowest detection rate.
- There has been a decline in referrals for *high risk maternal serum testing*.



	All reasons for referral		Abnormality on ultrasound		Increased nuchal translucency		Advanced maternal age		High risk maternal serum test	
	No.	%	No.	%	No.	%	No.	%	No.	%
<b>1995</b>										
Total normal	5,447	89.05%	240	81.08%			4,400	96.15%	308	94.19%
<b>Total abnormal</b>	<b>231</b>	<b>3.78%</b>	<b>49</b>	<b>16.55%</b>			<b>132</b>	<b>2.88%</b>	<b>16</b>	<b>4.89%</b>
Total other	439	7.18%	7	2.36%			44	0.96%	3	0.92%
<b>Total</b>	<b>6,117</b>	<b>100%</b>	<b>296</b>	<b>100%</b>			<b>4,576</b>	<b>100%</b>	<b>327</b>	<b>100%</b>
<b>2000</b>										
Total normal	6,678	94.46%	460	84.10%	532	87.93%	4,903	96.78%	228	96.61%
<b>Total abnormal</b>	<b>316</b>	<b>4.47%</b>	<b>77</b>	<b>14.08%</b>	<b>68</b>	<b>11.24%</b>	<b>130</b>	<b>2.57%</b>	<b>6</b>	<b>2.54%</b>
Total other	76	1.07%	10	1.83%	5	0.83%	33	0.65%	2	0.85%
<b>Total</b>	<b>7,070</b>	<b>100%</b>	<b>547</b>	<b>100%</b>	<b>605</b>	<b>100%</b>	<b>5,066</b>	<b>100%</b>	<b>236</b>	<b>100%</b>

## Prenatal cytogenetics test use and confinements for women 35 years and over

- The number of confinements has been relatively constant between 1990 and 2000 at around 85,000 to 86,500, but confinements to women aged 35 years and over have risen from 10.4% in 1990 to 13.1% in 1995 and to 17.7% in 2000.
- The chart below indicates that in 1995, 11,284 confinements were to women 35 years and over. By 2000, the number of confinements to women in this age group had risen to 15,334. The corresponding use of cytogenetic testing rose from 4,674 tests in 1995 to 5,591 tests in 2000. However, the growth in numbers of tests has not matched the growth in numbers of confinements to women over 35 years of age, so that the overall percentage of women using prenatal cytogenetic testing has declined from 41% in 1995 to 36.5% in 2000.
- As observed in Section 5, page 8 of this report the risk of having a child with Down syndrome increases with maternal age. The expected increase in numbers of liveborn children with Down syndrome has not occurred. This could be attributed to the impact of genetic counselling, provision of information about options available and increased use of nuchal translucency testing.



## Abnormalities and maternal age

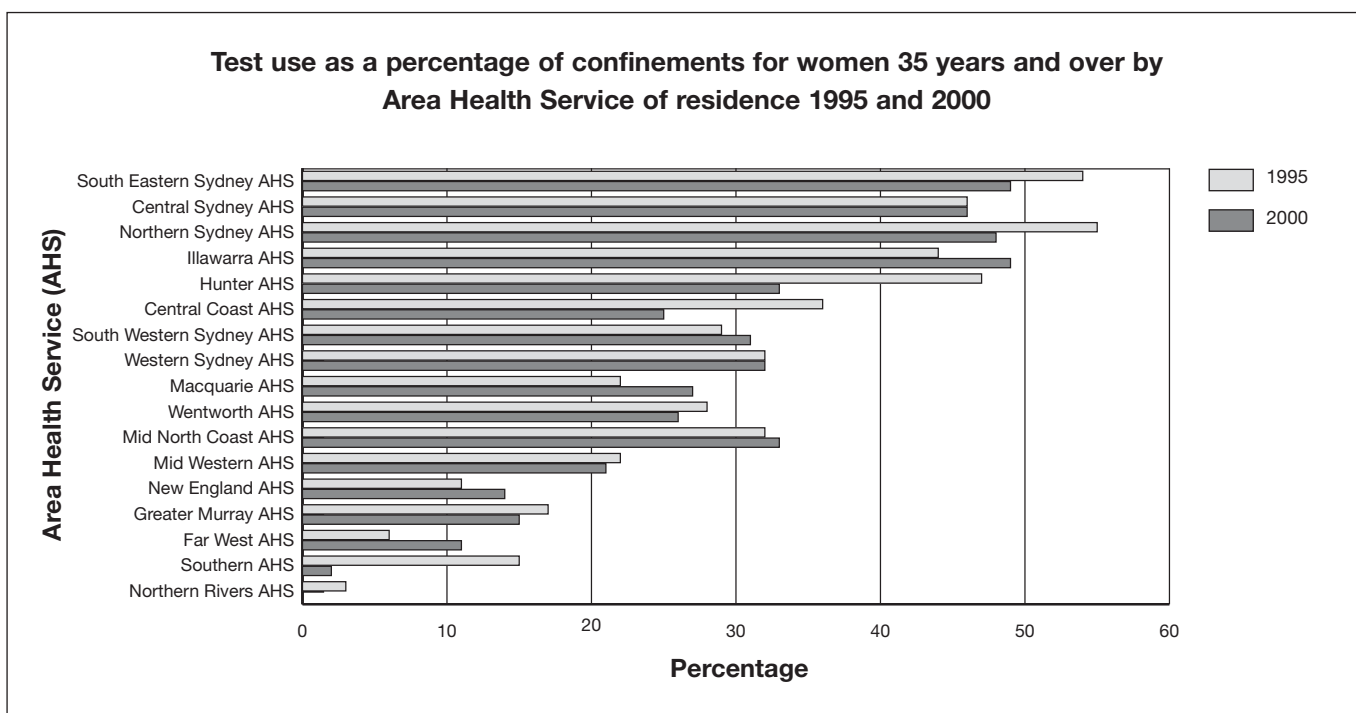
- In 2000, approximately 55% of prenatal cytogenetic referrals were for women aged between 35 and 39. This is a decline from 59% in 1995, although the number of women in this age group who are delivering babies has increased from 13.1% to 17.7% in the same period.
- The last five years have seen an increase in referrals in other age categories. Again this is likely to be related to greater use of ultrasound and nuchal translucency measurement.
- 24% of referrals in 2000 were for women aged between 40 and 44, ie an increase from 20% in 1995. Factors may include declining reproductive opportunities with advancing age, previous experience, availability of CVS earlier in the pregnancy or awareness of options. As women get older their preference for CVS over amniocentesis increases.
- Of the total 316 abnormalities detected in 2000, the 35-39 year age group had the highest number of abnormalities ie 111 or 35.13%.
- Although the 35-39 age groups had the highest number of tests and the highest number of abnormalities detected, it had the lowest detection rate of abnormalities at 2.87%.

### Tests and abnormalities by maternal age 1995 and 2000

Age	No of tests by age groups				No of abnormalities detected by age group				Abnormalities within age groups as a percentage of total abnormalities	
	1995		2000		1995		2000		1995	2000
	No	%	No	%	No	%	No	%	%	%
<20	16	0.26%	31	0.44%	2	0.87%	3	0.95%	12.50%	9.68%
20-24	89	1.45%	144	2.04%	5	2.16%	5	1.58%	5.62%	3.47%
25-29	215	3.51%	350	4.95%	15	6.49%	28	8.86%	6.98%	8.00%
30-34	568	9.29%	886	12.53%	42	18.18%	81	25.63%	7.39%	9.14%
<b>35-39</b>	<b>3,613</b>	<b>59.06%</b>	<b>3,874</b>	<b>54.79%</b>	<b>96</b>	<b>41.56%</b>	<b>111</b>	<b>35.13%</b>	<b>2.66%</b>	<b>2.87%</b>
40-44	1,211	19.80%	1,686	23.85%	63	27.27%	80	25.32%	5.20%	4.74%
>44	43	0.70%	88	1.24%	6	2.60%	8	2.53%	13.95%	9.09%
Unknown	362	5.92%	11	0.16%	2	0.87%	0	0.00%	0.55%	0.00%
<b>Total</b>	<b>6,117</b>	<b>100%</b>	<b>7070</b>	<b>100%</b>	<b>231</b>	<b>100%</b>	<b>316</b>	<b>100%</b>	<b>3.78%</b>	<b>4.47%</b>

## Prenatal cytogenetics tests and confinements by Area Health Service

- Cytogenetic testing as a proportion of confinements has traditionally been greater for women living in South Eastern Sydney, Central Sydney and Northern Sydney. The levelling off of invasive testing has occurred in most Area Health Services.
- **Note:** Adjustments have not been made for:
  - women who choose to terminate a pregnancy or miscarry after testing
  - women who change residence between time of testing and time of confinement.
  - testing carried out across State borders which occurs for New England, Greater Murray, Far West, Southern and Northern Rivers residents.



## Other cytogenetics services

Cytogenetics laboratories also conduct tests for:

- The diagnosis of congenital cytogenetic abnormalities, as well as cytogenetic aberrations which lead to developmental delay in childhood and adolescence. Cytogenetic investigation of some of these syndromes is carried out by peripheral blood sampling.
- The investigation of infertility or repeated miscarriages in couples. Testing is carried out on peripheral blood samples of affected couples.
- The diagnosis of patients with haematological disorders such as leukaemia, myelodysplasia and lymphoma, as well as for the characterisation of solid tumours. Cytogenetics is carried out on a bone marrow aspirate sample, peripheral blood, lymph node or on tumour biopsy material.

A diagnostic karyotype may provide confirmation of specific haematological disorders and crucial prognostic information. Follow-up karyotypes are crucial for the detection of minimal residual disease (MRD), following treatment regimens.

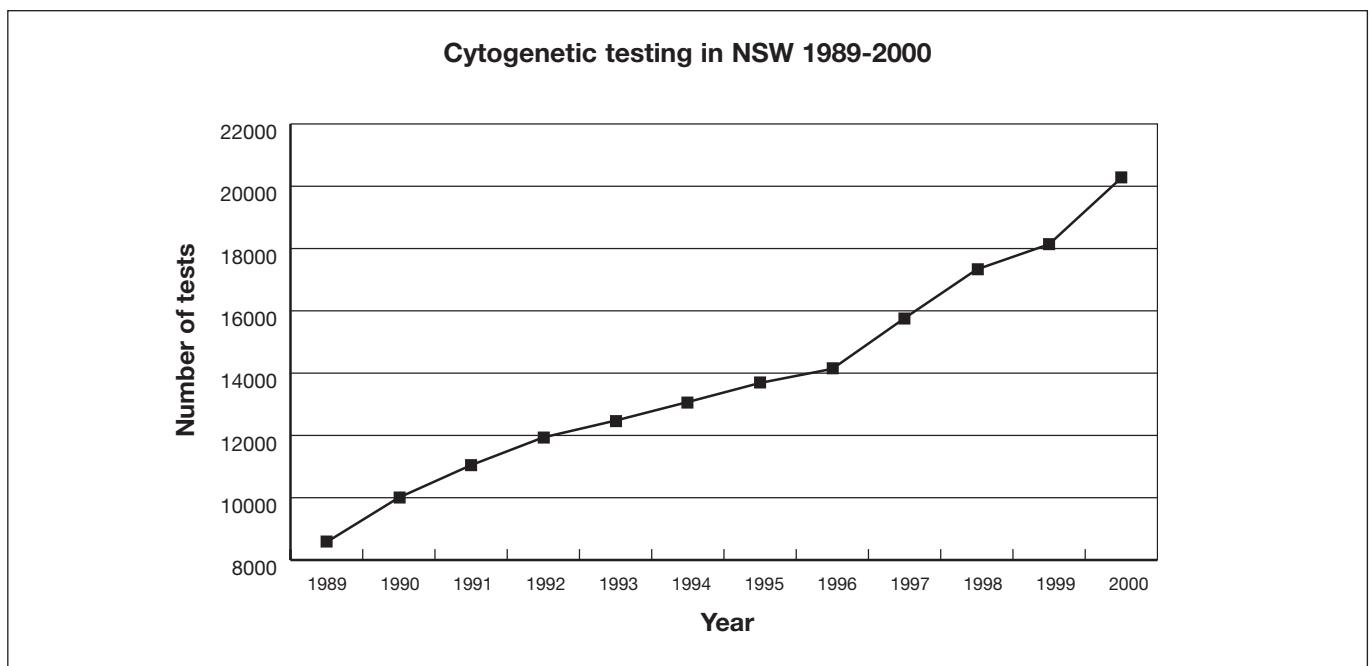
Currently, molecular cytogenetic techniques, such as Fluorescent in situ hybridisation (FISH) techniques are being utilised in increasing numbers for the purposes of prenatal screening for autosome and sex chromosome aneuploidy. These techniques are also used widely for the detection of microdeletion syndromes, such as Williams or diGeorge syndromes, and for the detection of MRD in haematological malignancies

### Cytogenetic testing data

Data presented below for all cytogenetic testing including prenatal tests have been collected since 1989 from the four public laboratories located in South Eastern Sydney, Northern Sydney, Royal Alexandra Hospital for Children and Hunter Area; and one private laboratory (Sydney Genetics).

### Test use

- The demand for cytogenetic testing has grown steadily over the past 10 years, from 8,576 studies in 1989 to 20,268 studies in 2000.
- A significant proportion of testing (7,070 tests in 2000) is for prenatal diagnosis.



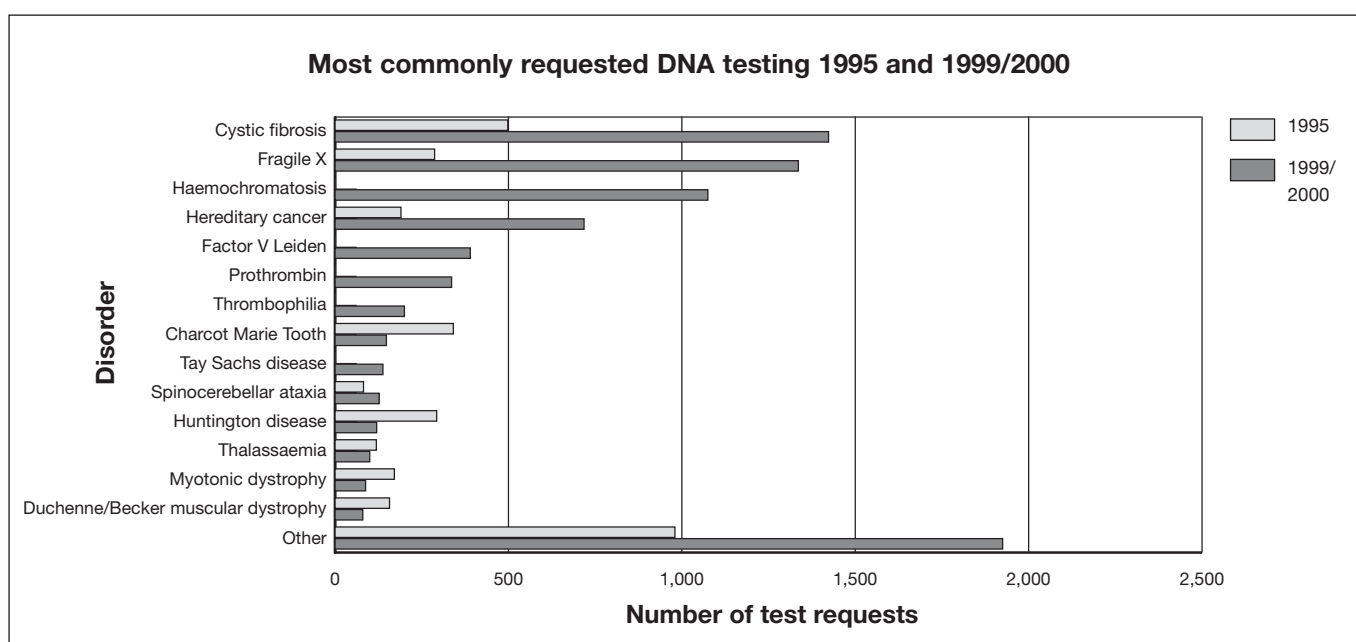
This type of testing involves examination of the DNA (deoxyribonucleic acid) to look for changes (mutations) in genes which may indicate a specific genetic disorder. DNA is the chemical compound which makes up genes within chromosomes and is the basic material of heredity.

DNA diagnostic testing is provided by eight molecular genetics laboratories in the State. In past years, each genetic disorder was tested in only one laboratory. This approach was adopted because the rarity of genetic disorders was such that individual laboratories would see too few cases to maintain proficiency of diagnosis and interpretation; and because low sample numbers together with high consumables and labour costs meant that costs could be minimised and duplication of services avoided through concentration of expertise. More recently some specific genetic disorder testing is being undertaken in more than one laboratory. Several factors have contributed to this change, including improved testing techniques, wider application of DNA testing than for genetic disorders alone, introduction of new tests and increased demand for genetic testing. Furthermore, the networking of pathology services is influencing the way DNA testing is provided.

The data which follow have been provided by molecular genetics laboratories in South Eastern Sydney, Central Sydney, Northern Sydney, Hunter and the Children's Hospital at Westmead. DNA cancer genetic testing at Westmead Hospital is included.

### Test use

- Technological developments have expanded the range of tests available and improved techniques. Since 1995, testing has been introduced for haemochromatosis, factor V leiden, prothrombin, and thrombophilia. Testing has increased for cystic fibrosis, fragile X, hereditary cancers and Tay Sachs disease. In other cases, testing has declined. This pattern is typical of DNA testing for many genetic disorders where there is an initial demand which tapers off after the identification of a significant number of people within the affected group.
- More detail on hereditary cancers can be found in the Cancer Genetics Services section on page 22.



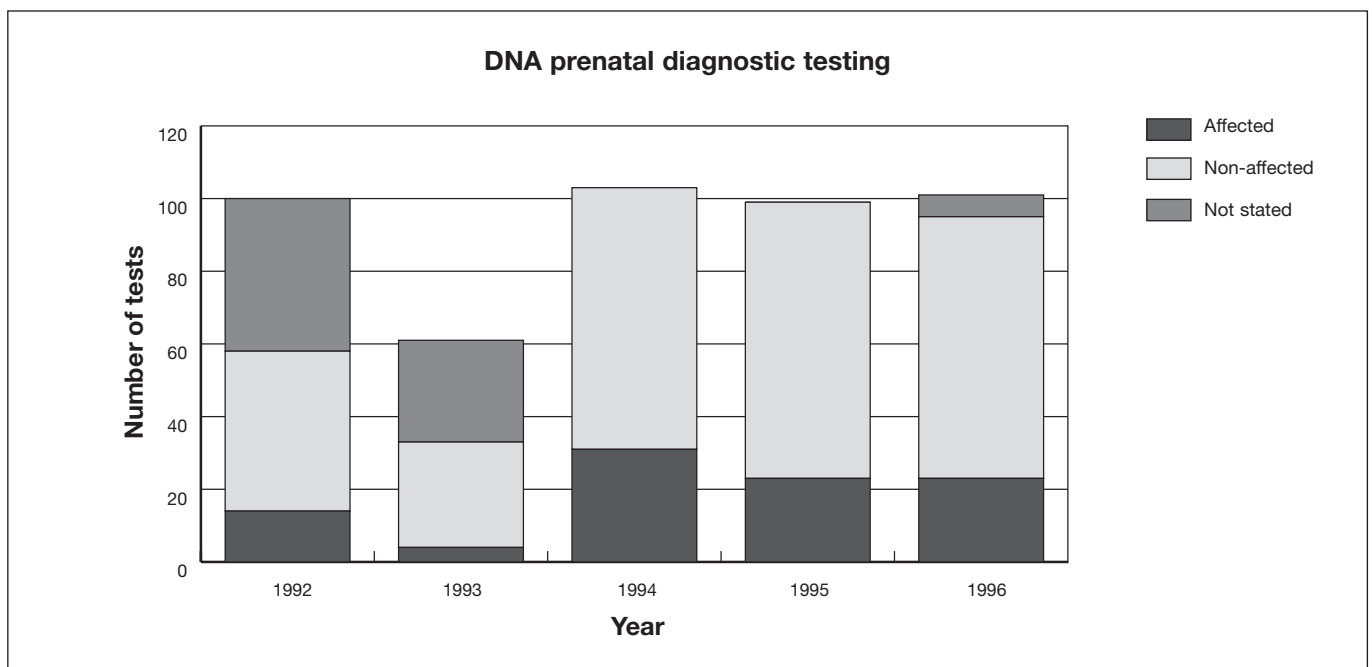
**Note 1:** The figures presented are best estimates for each 12 month period, 1995 and 1999. Participating laboratories provide data in differing forms, eg calendar year, financial year, or part year on which a full year has been estimated.

## Prenatal diagnostic testing outcomes

Outcomes of prenatal diagnosis are shown below for disorders listed opposite. *Affected* indicates that a specific mutation for the disorder being tested has been identified in the fetus. *Non-affected* means that the fetus does not have the specific mutation being tested for. Sometimes results are unclear and these are included in the *not stated* category.

Where a mutation has been identified in one or both parents, the risk for their offspring can be estimated. For example: a parent with the Huntington disease gene mutation has a 1 in 2 risk of having an affected offspring. In the case of cystic fibrosis, if both parents are mutation carriers, the offspring has a 1 in 4 risk of being affected. The results in the chart are consistent with these risk assumptions.

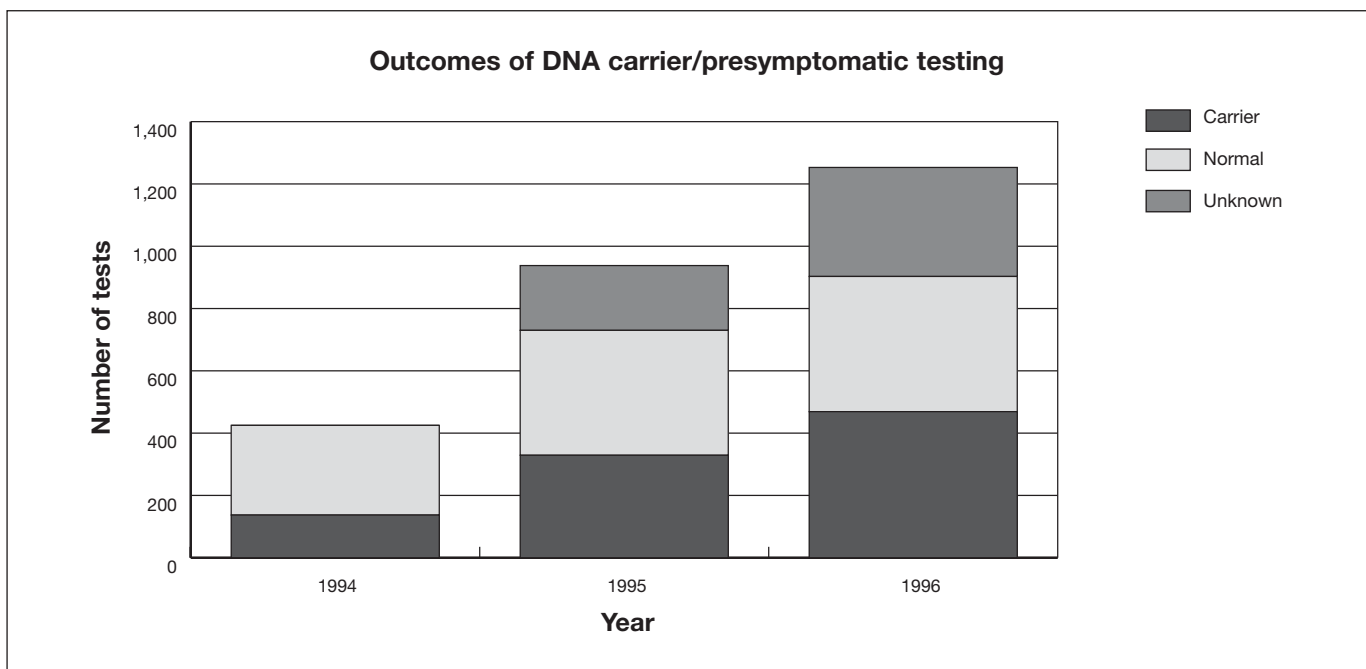
Disorder	Testing started
Cystic fibrosis	1988
Huntington disease	1990
Thalassaemia	1984
Haemophilia	1986
Duchenne & Becker muscular dystrophy	1989
Spinal muscular atrophy	1991
Fragile X cytogenetically	1981
Fragile X	1991
Charcot Marie Tooth	1992
Uniparental disomy	1992
Myotonic dystrophy	1992
Neurofibromatosis I	1994



## Carrier testing/ presymptomatic diagnosis

Outcomes of carrier testing are shown below for disorders listed opposite. *Normal* indicates that the person tested does not have the specific mutation/s being tested for. *Carrier* means that a mutation has been identified in the person being tested. Sometimes results are unclear and these are included in the *unknown* category.

Carrier testing has been conducted for the following disorders	
• Cystic fibrosis	• Breast/ovarian cancer
• Huntington disease	• Myotonic dystrophy
• Thalassaemia	• Fragile X
• Haemophilia	• Charcot Marie Tooth
• FHC	• Kennedy disease
• Familial adenomatous polyposis	• MMD
• Angelman/Prader Willi syndrome	• MJD
• Duchenne & Becker muscular dystrophy	• SCD
	• Other
	• MEN 2



# The NSW Genetics Education Program 13

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The Genetics Education Program of the NSW Genetics Service is based at the Royal North Shore Hospital, St Leonards, Sydney. The major focus of the program is the development and provision of information, resources and educational programs about:

- availability and access to genetics services (including clinical, counselling, specialist and laboratory services) for diagnosis, management, prenatal testing, and genetic counselling about problems in growth, development and health which may have an hereditary basis
- specific genetic disorders
- peer and family support groups for those affected by genetic conditions
- recent advances and technologies: their impact on individuals and families affected by genetic conditions and the community at large.

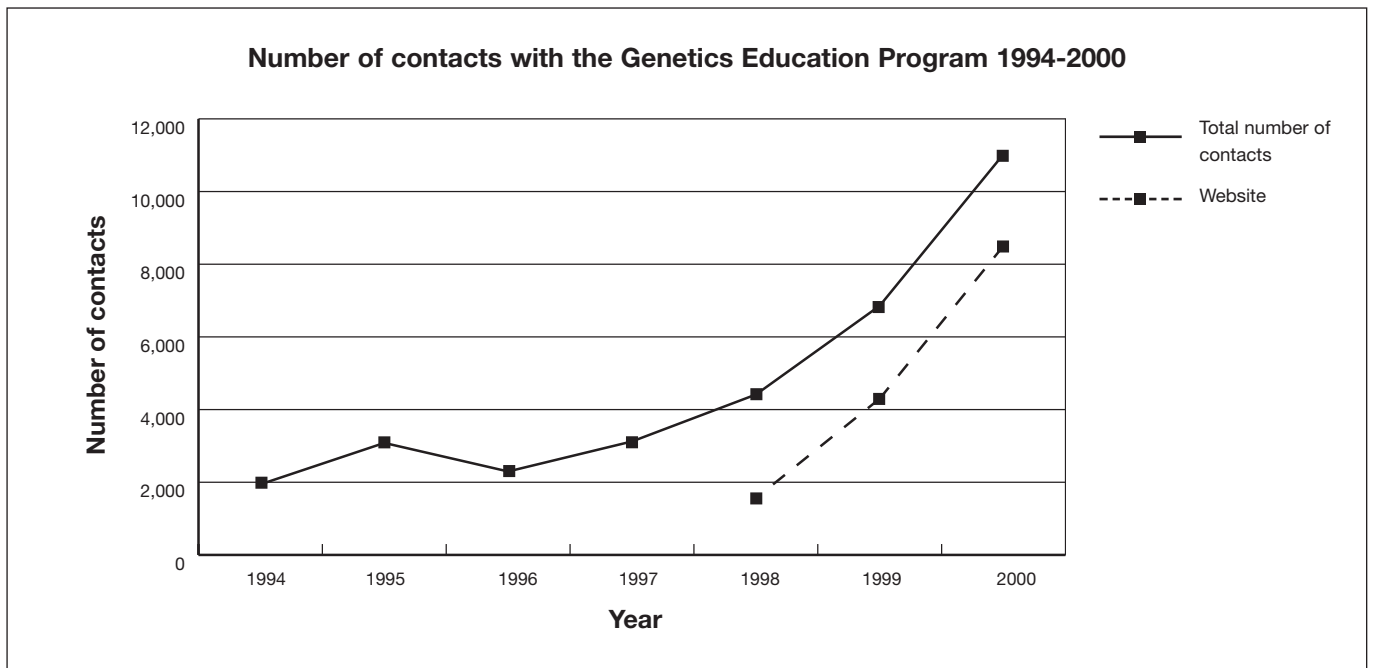
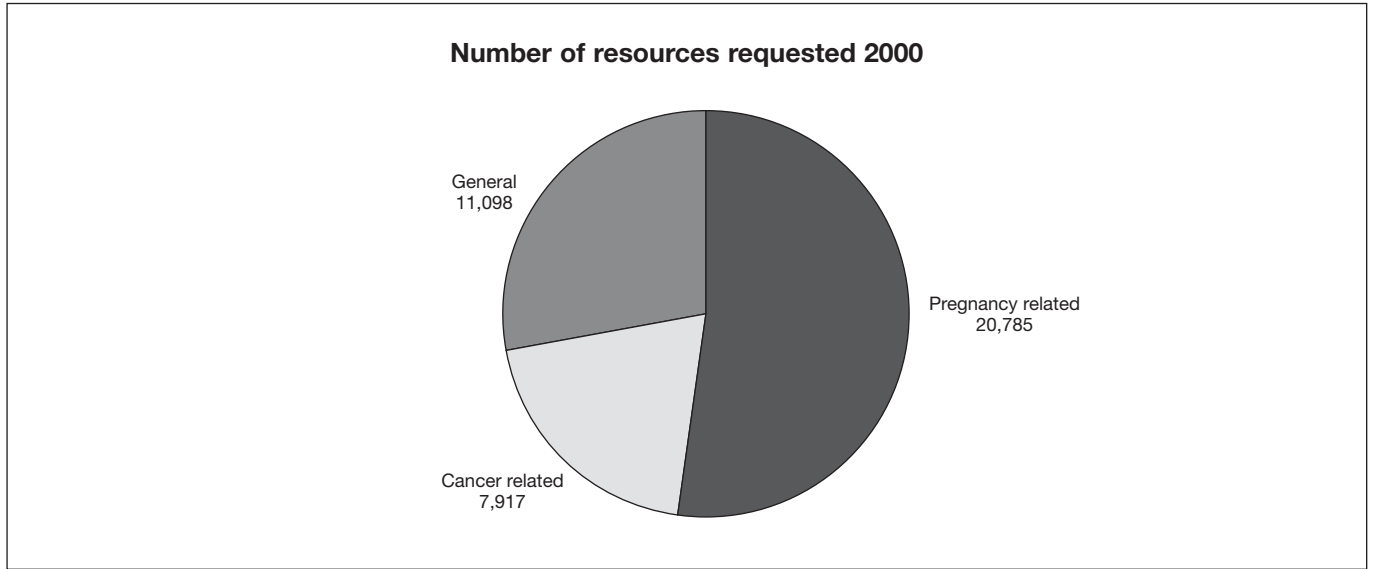
Activities of the program are targeted at all levels of the community: government, professionals, individuals and families affected by a genetic disorder and the general public. It is the central referral base for genetics services and information for NSW.

From 1986–1992, the program focussed on producing basic resources on genetic counselling, prenatal diagnosis, specific disorder information and support groups.

Since 1993 the program has targeted health professionals so that they can provide information to the families, using a ‘train the trainer’ concept by conducting workshops for science teachers and health professionals, as well as conducting health promotion campaigns to raise awareness of the importance of genetics in family health. Enhancement funding for a Statewide Cancer Genetic Education Project Officer has enabled the program to begin to address the need for information in this new and developing area. The success of these initiatives is reflected in the rapid increase in requests to the program since 1994.

*The Genetics Resource Book: a Directory of Support Groups, Services and Information for Australia and New Zealand* is now in its 5th edition. It has been evaluated highly and it is widely used by hospitals, libraries, schools and professionals. The program’s web site ([www.genetics.com.au](http://www.genetics.com.au)) has proven to be an effective tool in providing access to its resources.

All resources are produced in response to needs assessments or in response to requests for information from consumers. The program has surveyed support groups for their needs and experiences following genetic counselling, assessed public knowledge and attitudes to genetics technologies through questionnaires, utilised focus groups in the development and evaluation of resources and participated in research into educational programs for high school students to enable informed decision making regarding genetic screening.



MotherSafe provides telephone counselling for women and their healthcare providers throughout the State regarding agents of all types:

- prescription and over-the counter medications
- radiation
- chemicals
- infections
- occupational exposures.

The service aims to provide the most up-to-date information available to callers who are concerned about the potential risks of exposures during pregnancy and breastfeeding.

Apart from the telephone counselling service there is also a face-to-face counselling clinic for those women (around 5% of callers) who have been exposed to agents with increased risk or who wish to have further discussion about their concerns for other reasons.

In the first year of service since January 2000, MotherSafe has handled over 4,000 calls. Most calls have come from consumers (women and their partners) with around 40% of calls from health care professionals including general practitioners, obstetricians, clinical geneticists, genetic counsellors, pharmacists, midwives, lactation consultants and early childhood nurses.

Mothersafe is based in South Eastern Sydney Area Health Service but provides a service for women throughout the whole state of NSW. Approximately 70% of the calls have come from Sydney, with 27% from non-metropolitan NSW and the remainder from interstate, including the ACT, and overseas.

The majority of calls (62%) have been regarding exposures during pregnancy, while 25% have been questions about exposures during lactation. The remainder of the calls has been from women planning a pregnancy or questions about exposures during previous pregnancies.

MotherSafe has been involved in professional and public education as well as public health initiatives such as the promotion of pre-conceptual folic acid.

MotherSafe is also establishing follow-up studies to improve the information available to pregnant women and their health care providers on the outcomes of exposures to new or unusual medications during pregnancy and lactation.

AGSA provides support and information for individuals and families affected by a genetic condition. It has been supported under the NSW Department of Health's Non-Government Organisation program since 1993.

## How AGSA helps

The diagnosis of a genetic condition in a family member, particularly a child, places enormous stress on a family. Families may feel the need for personal support offering a specialised understanding of their particular condition. Whilst there are support groups established for a number of genetic disorders, AGSA may provide the only contact point for families affected by a rare condition. AGSA will endeavour to facilitate contact with another family/individual affected by the same, or similar conditions, and/or provide information about an overseas support group.

## AGSA also provides:

- a Peer Support and Information Officer who deals with inquiries and facilitates ongoing support for families, health professionals and other interested groups
- regular bi-monthly newsletter
- resources relating to education, medical services, other helpful organisations, allowances and respite care
- information seminars
- local and regional contacts.

## Achievements over the last five years

- Employment of Peer Support & Information Officer – 1994
- First Genetic Awareness Week – 1994
- Increase in membership to 400
- Treble number of incoming calls
- Establishment of twenty new support groups
- 30 seminars held on genetic conditions
- Organised disability seminars in rural areas
- Established a Contact Register of over 400 genetic conditions
- Undertook Pilot Survey 'What are the needs after the diagnosis of a genetic condition?'
- Poster presentation at HGSA Meeting in Melbourne 1998
- Development of the Health Information folder

## NSW Genetics Service

### Publications

- *Specialised Testing for Genetic Disorders*, May 2000
- *Ethical Code Governing the Provision of Genetics Services*, June 1998
- *Report on the Impact of Cancer Genetic Technology and Recommendations for Cancer Genetics Service Provision*, April 1996
- *The Goal and Objectives of the NSW Genetics Service*, September 1995
- *Genetics Services in NSW Five Year Plan 1993-1998*, July 1993
- *Genetics Services in NSW 1987-1991*, August 1991
- *Report of the Genetics Services and Birth Defects Sub-Committee on Super-Specialty Genetics Services in NSW and ACT: Review and Planning for Future Development of Delivery, Distribution and Organisation*, March 1987

### Reports and Circulars relevant to Genetics Services, released by NSW Health

- *Guidelines for diagnostic and predictive DNA testing for adult onset neurogenetic disorders with no definitive treatment such as Huntington disease*, Circular No. 2001/87
- *Guidelines for Newborn Screening*, Circular No. 2001/45
- *Haemochromatosis – Information for Health Care Providers on Diagnosis and Management*, Information Bulletin 2000/9
- *Guidelines for Diagnosis and Management of Haemoglobinopathies*, Information Bulletin 99/14
- *Guidelines for Testing for Genetic Disorders*, Circular No. 97/48
- *Folic Acid and Neural Tube Defects*, Circular No. 94/68
- *Genetics Services Policy Guidelines*, 1987
- *Guidelines for Investigation of a Stillbirth*, Circular No. 97/107
- *Reporting and Submission Requirements of Data for the NSW Birth Defects Register (BDR)*, Circular No. 2001/85

## NSW Genetics Education Program

### Publications

Following is a comprehensive list of resources available from:

The NSW Genetic Education Program  
PO Box 317  
ST LEONARDS NSW 2065  
Tel. (02) 9926 7324  
Fax. (02) 9906 7529  
Web. [www.genetics.com.au](http://www.genetics.com.au)

### General

- *Do You Know Your Genes? – A Do-It-Yourself Guide to Drawing Your Family Health Tree*
- *The Genetics Resource Book (2000/2001)*: an Australasian directory of genetics support groups, services and information – A comprehensive resource for Australia and New Zealand, published annually for the health, education and welfare professionals; and for the individuals and families affected by genetic conditions
- *Genetic Information Fact Sheets* on over 550 genetic disorders – information is prepared concerning a particular disorder utilising the most recent data available about the condition and existing support services. Each information sheet is dated and will not be distributed if more than 6 months has passed since its last update
- *Autopsy Information for parents about the postmortem examination* – pamphlet

### Genetic Counselling

- *What you should know about inherited disorders* – Pamphlet. Available in Arabic, Armenian, Chinese, Croatian, Greek, Italian, Khmer/Cambodian, Lao, Macedonian, Maltese, Polish, Portuguese, Serbian, Spanish, Turkish, Vietnamese

### Folate

- *Facts Sheet* for health professionals

### **Cystic Fibrosis**

- *Genes and Cystic Fibrosis – Basic Facts about Chromosomes, Genes and Cystic Fibrosis – Information kit*

### **Huntington Disease**

- *Predictive Testing for Huntington Disease Information Kit*
- *Predictive Testing – Information for Physicians – Pamphlet*

### **Screening In Pregnancy**

#### **Ultrasound**

- Pamphlet

#### **First Trimester Screening**

- *Screening tests for your baby in early pregnancy – brochure for consumers*
- *First trimester screening using nuchal translucency ultrasound and biochemical testing – information for health professionals*

#### **Maternal Serum Testing:**

- *A Blood Test to Determine the Risk of Certain Problems in your Pregnancy, The Maternal Serum Test – Pamphlet*
- *Some questions and answers when your test result shows an ‘increased risk’ of your baby having Down syndrome – Pamphlet*
- *Some questions and answers when your test result shows an ‘increased risk’ of your baby having a neural defect such as Spina Bifida – Pamphlet*

### **Prenatal Diagnosis**

- *Checking your baby’s health before birth – Pamphlet. Available in Arabic, Chinese, Croatian, Khmer/Cambodian, Korean, Lao, Macedonian, Polish, Portuguese, Spanish, Turkish, Vietnamese*
- *Special Tests for your Baby During Pregnancy, Chorionic Villus Sampling (CVS), Ultrasound and Amniocentesis, Prenatal Diagnosis – Booklet*
- *Prenatal Diagnosis – Towards an Informed Decision – an educational video produced for use in the community*

### **Support After Fetal Diagnosis Of Abnormality (SAFDA)**

- *Support after Fetal Diagnosis of Abnormality (Safda) – Pamphlet*
- *Diagnosis of Abnormality in an Unborn Baby, A Booklet for Parents*
- *When your unborn baby has a problem: How to manage the weeks ahead – booklet*

### **Cancer resources**

- *What if I have a family history of cancer – brochure for consumers*
- *Cancer: the significance of family history – brochure for health professionals*
- *Information for women considering preventive mastectomy because of a strong family history of breast cancer – booklet*
- *Is breast cancer inherited? – video for consumers*

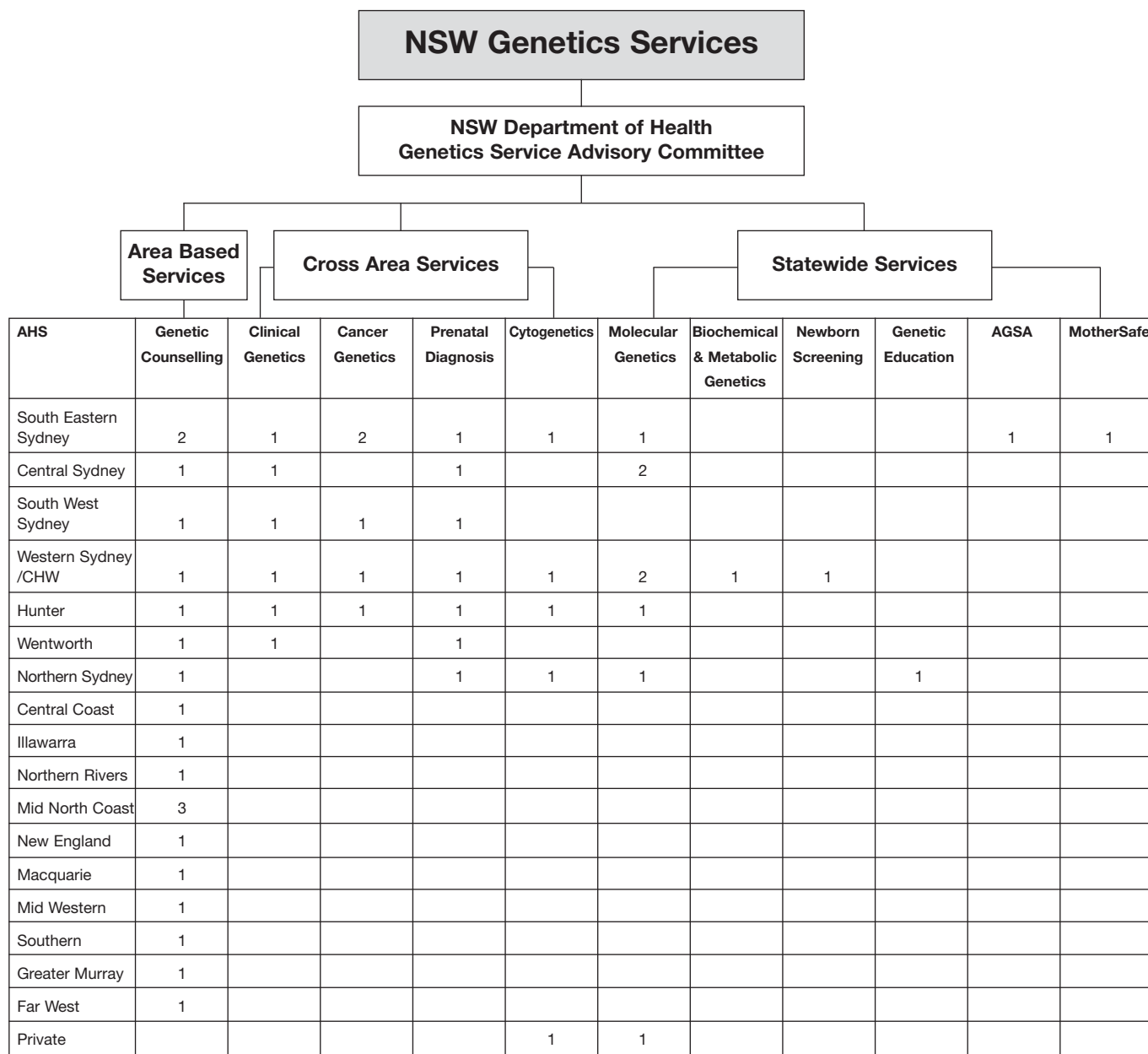
# References

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- 1 Wilcken B, Wiley V, Sherry G, Bayliss U. *Newborn Screening for Cystic Fibrosis: a comparison of two strategies for case detection in 1.2 million babies.* J.Pediatr 1995; 127:965-72
- 2 Buyse ML, Editor in Chief, *Birth Defects Encyclopedia*, 1990, USA
- 3 Turner G, Robinson H, Wake S, Laing S, Partington M, *Case finding for the Fragile X syndrome and its consequences*, BMJ 1997, 315:1223-1226
- 4 Mowat DR, Hayden M, Thompson S, Wilcken B. *Maternal phenylketonuria: a continuing problem* Med J Aust. 1999; 170: 592-595
- 5 *The Goal and Objectives of the NSW Genetics Service*, 1995
- 6 *New South Wales Mothers and Babies*, 2000, NSW Public Health Bulletin Supplement, Vol 12, Number S-3, November, 2001, NSW Health Department
- 7 Cowan J, Macdessi J, Stark A, Morgan G, *Incidence of Duchenne muscular dystrophy in NSW and the ACT*, J Med Genetics, Vol 17:245-249
- 8 Easton DF, *The Inherited component of cancer*, British Medical Bulletin (1994) Vol 50, No3, pp 527-535



# Appendix 1 – NSW Genetics Service Locations



All genetic counselling services are supported by visiting clinical geneticists

# Appendix 2 – Genetic services workforce

Services/Staff	1998 FTE	1999 FTE	2000/2001 FTE
<b>Clinical Genetics</b>			
Clinical Geneticist	10.00	10.70	11.20
Clinical Academic	0.60	0.60	0.60
Clinical Geneticist Administration	0.50	0.50	0.50
Genetic Fellows	4.00	4.00	4.00
Genetic Counsellor (Certified)		24.10	6.50
Associate Genetic Counsellor	27.35		18.30
Social Worker		2.25	2.25
Administration	12.10	12.90	13.90
<b>Total</b>	<b>54.55</b>	<b>55.05</b>	<b>57.25*</b>
*Approximately 6 Clinical Geneticists work in specialties such as neurology, maternal/fetal medicine with some time in clinical genetics			
<b>Cancer Genetics</b>			
Clinical Geneticists	1.00	1.00	1.80
Oncologist/Genetics	1.00	1.00	1.00
Genetic Counsellor (Certified)	1.00	1.00	1.50
Associate Genetic Counsellor	1.00	3.00	3.00
Cancer Genetic Education	1.00	1.00	1.00
Administration	2.50	3.50	3.50
Other	1.50	1.00	1.00
<b>Total</b>	<b>9.00</b>	<b>11.50</b>	<b>12.80</b>
<b>Cytogenetics Services (Public Hospital Labs)</b>			
Staff Specialists/Registrars	1.6	1.60	1.50
Scientific Staff	57.5	58.56	61.00
Administration	1.9	1.73	1.73
Other			
<b>Total</b>	<b>61</b>	<b>61.89</b>	<b>64.23</b>
<b>Molecular Genetics Services</b>			
Staff Specialists/Registrars	1.30	1.40	3.30
Scientific Staff	24.20	33.60	38.10
Associate Genetic Counsellor	0.40	0.40	0.00
Administration	2.10	2.00	2.50
Other			0.00
<b>Total</b>	<b>28.00</b>	<b>37.40</b>	<b>43.90</b>
<b>Biochemical Genetics and Newborn Screening</b>			
Staff Specialists/Registrars	0.50	0.50	0.50
Scientific Staff	21.50	20.00	20.50
Administration	3.00	2.50	2.50
Other	1.00	1.00	1.00
<b>Total</b>	<b>26.00</b>	<b>24.00</b>	<b>24.50</b>
<b>Metabolic Service</b>			
Metabolic Physician			1.30
Honorary Fellow			1.00
Dietitian			0.90
Social Worker			0.20
Nurse			1.00
<b>Total</b>		<b>4.10</b>	<b>4.40</b>
<b>MotherSafe</b>			
Clinical Geneticist			0.50
Associate Genetic Counsellor			0.50
<b>Total</b>			<b>1.00</b>
<b>Maternal Serum Testing</b>	<b>2.00</b>	<b>2.00</b>	<b>2.00</b>
<b>Genetic Education (including 1 Cancer Education in 1999/2000)</b>	<b>3.70</b>	<b>3.70</b>	<b>3.40</b>
<b>Executive Administration</b>	<b>2.00</b>	<b>2.00</b>	<b>2.00</b>
<b>Total Staff</b>	<b>186.25</b>	<b>201.64</b>	<b>215.48</b>

# Appendix 3 – Clinical and genetic counselling service locations and contact details

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## **Clinical and Genetic Counselling Service Locations**

### **Camperdown**

Department of Molecular and Clinical Genetics  
Royal Prince Alfred Hospital  
Missenden Road  
CAMPERDOWN NSW 2050  
Tel. 9515 5080  
Fax. 9515 7595

### **Liverpool**

Department of Clinical Genetics  
Health Services Building  
Cnr Campbell and Goulburn Streets  
LIVERPOOL NSW 2170  
Tel. 9828 4665  
Fax. 9828 4650

### **Penrith**

Nepean Hospital  
Summerset Street  
PENRITH NSW 2750  
Tel. 4734 3362  
Fax. 4734 2567

### **Randwick**

Department of Medical Genetics  
Sydney Children's Hospital  
High Street  
RANDWICK NSW 2031  
Tel. 9382 1708  
Fax. 9382 1711

### **Westmead**

Department of Clinical Genetics  
The New Children's Hospital  
Hawkesbury Road  
WESTMEAD NSW 2145  
Tel. 9845 3273  
Fax. 9845 3204

### **Newcastle**

Hunter Genetics  
Cnr Turton & Tinonee Streets  
WARATAH NSW 2298  
Tel. 4985 3100  
Fax. 4985 3105

## **Genetic Counselling**

**Services** in conjunction with visiting clinical genetics services.

### **Kogarah**

Women's and Children's Health  
2nd Floor Prichard Wing  
St George Hospital  
Gray Street  
KOGARAH NSW 2217  
Tel. 9350 2315  
Fax. 9350 3901

### **St Leonards**

Fetal Medicine Unit  
Royal North Shore Hospital  
Pacific Highway  
ST LEONARDS NSW 2065  
Tel. 9926 6478  
Fax. 9906 1872

### **Bathurst**

Community Health Centre  
158 William Street  
BATHURST NSW 2795  
Tel. 6331 5533  
Fax. 6332 2039

### **Broken Hill**

Community Health Centre  
BROKEN HILL NSW 2880  
Tel. (08) 8080 1556  
Fax. (08) 8080 1611

### **Canberra**

The Antenatal Clinic  
The Canberra Hospital  
Gilmore Crescent  
CANBERRA ACT 2605  
Tel. 6244 4042  
Fax. 6244 3422

### **Coffs Harbour**

Coffs Harbour Health Campus  
Pacific Highway  
COFFS HARBOUR 2450  
Tel. 6656 7806  
Fax. 6656 7817

### **Gosford**

Child Health Centre  
297 Henry Parry Drive  
WYOMING NSW 2250  
Tel. 4337 0207  
Fax. 4337 0217

### **Goulburn**

Child Development Unit  
Cnr Albert and Clifford Streets  
GOULBURN NSW 2580  
Tel. 4827 3951  
Fax. 4827 3958

### **Lismore**

Child and Family Health Centre  
37 Oliver Avenue  
GOONELLABAH NSW 2480  
Tel. 6625 0111  
Fax. 6625 0102

### **Mudgee/Dubbo**

Mudgee Community  
Health Centre  
MUDGEES NSW 2850  
Tel. 6372 6455  
Fax. 6372 7341

### **Muswellbrook**

Community Health Centre  
Brentwood Street  
MUSWELLBROOK NSW 2332  
Tel. 6542 2083  
Fax. 6542 2005

### **Port Macquarie**

Hastings Macleay  
Community Health  
Morton Street  
PORT MACQUARIE 2444  
Tel. 6588 2882  
Fax. 6588 2800

**Tamworth**

Community Health Centre  
Cnr Dean and Johnson Streets  
TAMWORTH NSW 2340  
Tel. 6766 2555  
Fax. 6766 3967

**Taree/Forster**

Community Health Centre  
64 Putney Street  
TAREE NSW 2430  
Tel. 6592 9315  
Fax. 6592 9607

**Wagga Wagga**

Wagga Base Hospital  
Edward Street  
WAGGA WAGGA NSW 2650  
Tel. 6938 6393  
Fax. 6921 5632

**Wollongong**

Maternal and Paediatric Services  
Wollongong Hospital  
Crown Street  
WOLLONGONG NSW 2500  
Tel. 4222 5216  
Fax. 4222 5477

**MotherSafe**

**Statewide Medications in  
Pregnancy and Lactation  
Advisory Service**

Royal Hospital for Women  
Barker Street  
RANDWICK NSW 2031  
Tel. 9382 6539 (Sydney calls)  
Tel. 1800 647 848 (Other calls)

**AGSA**

**Association of Genetic Support  
of Australasia Inc.**

66 Albion Street  
SURRY HILLS NSW 2010  
Tel. 9211 1462  
Fax. 9211 8077  
Email. [agsa@ozemail.com.au](mailto:agsa@ozemail.com.au)  
Web. [www.agsa-  
geneticsupport.org.au](http://www.agsa-geneticsupport.org.au)

**Prenatal Diagnosis &  
Counselling Services**

**Camperdown**

Fetal Medicine Unit  
King George V Hospital  
Missenden Road  
CAMPERDOWN NSW 2050  
Tel. 9515 8258  
Fax. 9515 6579

**Liverpool**

Fetal Medicine Unit  
Liverpool Hospital  
Elizabeth Drive  
LIVERPOOL NSW 2170  
Tel. 9828 4145  
Fax. 9828 4146

**Randwick**

Prenatal Diagnosis  
Royal Hospital for Women  
Barker Street  
RANDWICK NSW 2031  
Tel. 9382 6098  
Fax. 9382 6706

**Penrith**

Fetal Medicine Unit  
Nepean Hospital  
Summerset Street  
PENRITH NSW 2750  
Tel. 4734 3163  
Fax. 4734 3206

**St Leonards**

Fetal Medicine Unit  
Royal North Shore Hospital  
Pacific Highway  
ST LEONARDS NSW 2065  
Tel. 9926 7280  
Fax. 9906 1872

**Westmead**

Fetal Medicine Unit  
Westmead Centre  
Hawkesbury Road  
WESTMEAD NSW 2145  
Tel. 9845 6802  
Fax. 9845 7793

**Newcastle**

Prenatal Diagnosis Unit  
John Hunter Hospital  
NEWCASTLE NSW 2310  
Tel. 4921 4694  
Fax. 4921 3133

**Cancer Genetics  
Specialised Services**

**Darlinghurst**

Family Cancer Clinic  
Department of Medical Oncology  
St Vincent's Hospital  
Victoria Street  
DARLINGHURST NSW 2010  
Tel. 8382 3395  
Fax. 8382 3386

**Kogarah**

Cancer Care Centre  
St George Hospital  
Belgrave Street  
KOGARAH NSW 2217  
Tel. 9350 3815  
Fax. 9350 3958

**Liverpool**

Liverpool Hospital  
Elizabeth Drive  
LIVERPOOL NSW 2170  
Tel. 9828 4665  
Fax. 9828 4650

**Randwick**

Hereditary Cancer Clinic  
Prince of Wales Hospital  
High Street  
RANDWICK NSW 2031  
Tel. 9382 2587  
Fax. 9382 2588

**Westmead**

Familial Cancer Services  
Westmead Hospital  
Hawkesbury Road  
WESTMEAD NSW 2145  
Tel. 9845 5079  
Fax. 9687 2331

**Newcastle**

Hunter Genetics  
Cnr Turton & Tinonee Streets  
WARATAH NSW 2298  
Tel. 4985 3100  
Fax. 4985 3105

**Further Information**

On services in other areas and  
newly developed services:  
NSW Genetic Education Program  
PO Box 317  
ST LEONARDS NSW 2065  
Tel. 9926 7324  
Fax. 9906 7529  
Web. [www.genetics.com.au](http://www.genetics.com.au)

