

Plan for NSW Familial Cancer Services 2010 - 2015

September 2010

DISCUSSION PAPER



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Introduction

It is estimated that between 5-10% of all cancer is inherited¹, and that for some cancers family members have between 40% (ovarian) to 80% (breast and colorectal) lifetime chance of developing cancer if a specific gene is present². It is estimated that in one year- 2007-, some 1798-3595 individuals inherited cancer (table 12). It is also estimated that in 2011, almost 6,000 people should be referred to familial cancer services (Table 14). This number rises to approximately 6,700 in 2016.

1 Executive Summary

The Cancer Institute NSW engaged a consultant to undertake the development of a statewide cancer genetics plan. The following is a de-identified version of the report provided under this consultancy and is based on cancer genetics services in NSW in 2009.

1.1 Project Objective

The main objective of this project is to provide options for a structure for statewide familial cancer genetics services, both clinical and laboratory, which will meet current and future demand and be accessible to all potential clients across NSW.

1.2 Project Liaison and Direction

A Steering Committee was established to provide advice to the consultant on the scope of the project and issues that may arise; and provide and facilitate access to information as required. The Committee met on three occasions during the project.

1.3 Methodology

The project consisted of the following components:

- Consultations with key stakeholders;
- Review of the provisions of familial cancer services (FCS) in other States of Australia and internationally;
- A Data analysis; and
- Future services demand projections.

1.4 Current Service Arrangements

The current service configuration of FCS can be seen in the Figure 1.

Figure 1: Current Service Arrangements of Familial Cancer Services in NSW.

Familial Cancer Clinic Hospital 1

1.6 FTE staff specialist
 2.6 FTE counsellor
 1.0 FTE research nurse
 2.0 FTE data/admin

Laboratory services are provided primarily by SEALS, HAPS.

Outreach services from Hospital 1 are provided by the Director/CMO to:

Hospital 2 Hospital 1 1 visit per mth (approx. 10 times per year) by Director. There is 1.0 FTE cancer genetic counsellor based at Hospital 2 hospital.

Hospital 3 Hospital - 1 visit per mth by CMO. There is 0.8 FTE cancer genetic counsellor based at Hospital 3 Hospital

Hospital 25 - 1 /mth via telehealth by Director. There are 1.25 FTE cancer genetic counsellors based at Hospital 25

Laboratory services are provided by SEALS for breast testing, HAPS for bowel testing

Familial Cancer Service Hospital 4

1.6 FTE staff specialist
 2.0 FTE counsellor
 0.6 FTE nurse
 1.2 FTE data/admin

Laboratory services provided primarily by molecular laboratory within the FCS and HAPS

Outreach services are provided by the Director or staff specialist of the Familial Cancer Service to:

Hospital 24 2 visits per month by Director and counsellor

Hospital 17 - 1 /mth via telehealth (not currently being provided). At Hospital 17 there is 1.0 FTE cancer and general genetics counsellor

Hospital 19 - 1/mth via telehealth by staff specialist. There is 1.0 FTE cancer and general genetic counsellor.

Laboratory services provided by the molecular laboratory within the Familial Cancer Service for breast testing, HAPS for bowel testing,

Hospital 6 Familial Cancer Service

0.2 FTE HMO
 1.6 FTE counsellor
 1.0 FTE admin
 0.5 FTE data manager

Laboratory services provided primarily by HAPS

Outreach services are provided by the HMO at the Hospital 6 Familial Cancer Service to:

Hospital 8 1 visit every 2 months ie 6 per year

Hospital 9 1 visit every 2 months ie 6 per year

Hospital 11 1 visit every 3 months ie 4 per year

Hospital 10 1 visit every 3 months ie 4 per year

Hospital 20 and surrounding towns of Hospital 13, Hospital 29, Hospital 30 and Hospital 31 on demand via telehealth

Hospital 12 and Hospital 32 on demand via telehealth

Hospital 14 1 visit per month

Hospital 15 on demand via telehealth

Hospital 16 none at present because there is no counsellor at Hospital 16

Hospital 18 8 visits are planned for 2010 with 2 clinics each in Hospital 18, Hospital 33, Hospital 34 and Hospital 35.

The genetic counsellors based in each of these towns provide both cancer and general genetic counselling (see FTE numbers in Table 5.) Both general geneticists and cancer geneticists visit to provide clinics. The visits described above are those provided by the HMO from the Hospital 6 FCS.

Laboratory services are provided by HAPS and GTG.

Familial Cancer Clinic Hospital 5

0.05 FTE clinical academic
 1.0 FTE genetic counsellor

Laboratory services are provided primarily by HAPS and GTG

Health Service 4 Familial Cancer Clinic

0.05 FTE VMO
 1.0 FTE genetic counsellor

Laboratory services are provided by GTG.

Familial Cancer Service Hospital 7

0.4 FTE staff specialist
 0.6 FTE genetic counsellor

Laboratory services are provided by HAPS

1.5 Challenges facing the current services

- Unequal access by the population to FCS- see Table 6 which shows the FCS staff to population ratios for each AHS.
- Outreach services have developed on an ad hoc basis by the three main FCS.
- Use of telehealth for outreach is adversely influenced by the fact that it cannot be reimbursed through Medicare despite the fact the FCS medical consultations are suited to this medium.
- There is variation between FCS in the way services are provided- the counsellor role differs between services as indicated by the very different doctor counsellor ratios by FCS outlined in Table 7.
- The three largest services (1, 4 and 6) depend on temporary funding- between 14% and 33% of their total budget- see Table 8.
- There is not a clear training path for medical specialists so it is difficult to attract trainee clinical cancer geneticists. Training for genetic counsellors is currently being revised with genetic counsellors required to do an undergraduate and masters degree (minimum 5 years) at considerable cost which may make it difficult to attract people to the profession, especially if their future role in FCS is not clear and can vary from service to service.
- Clinical governance of FCS differs between services. Some services are located within the Cancer Stream, some are in Primary and Community Health and others in the Chronic and Complex Care Stream. There are no formal service agreements between the AHS providing the medical outreach services and the AHS receiving the medical outreach services. If there is a vacant genetic counsellor position in outreach, no medical outreach services are provided until the position is filled.
- Statewide planning and coordination is currently being undertaken by both the Cancer Institute NSW and NSW Genetic Service Advisory Committee (GSAC) with no clear definition of the roles and responsibilities of the two organisations in relation to planning for FCS.
- There will be increasing numbers of patients who should be referred to the FCS. In 2008 it is estimated that there were around 3500 new referrals to FCS in NSW. This falls far short of the estimated number of people who would need the service in 2011 (around 6,000 people) and in 2016 (approximately 6,800 people). The demands on the FCSs are set to increase exponentially as new genes linked to inherited cancer are discovered and as there is a greater need for the FCS to become more acute with testing done at the time of patient diagnosis because treatment will depend on the results.
- There is variation in the waiting time for tests by the laboratories from 2-3 weeks for urgent cases to 12 months. As the clinical FCS begin to operate more acutely, it is not clear whether the current laboratories will be able to handle the workload.
- There is variation between services in terms of data collection systems. The three main FCS use Genesis but they have all modified it to suit their own purposes and there are no common data definitions. The outreach services do not use Genesis- some do not have a data system at all, others have to enter data twice- into CRS, a genetics database, and the community health database.

1.6 Issues Affecting Future Services Delivery

- Access to services varies in terms of both geographic factors and waiting times.
- There is no common model of service with variation between services in terms of staffing, funding, protocols and guidelines, follow up management, role of doctor vis a vis genetic counsellor, nature and provision of outreach, attendance at multidisciplinary team meetings.
- The governance of FCS varies between Area Health Services. Each Area is clinically and operationally responsible for the services within their boundaries but

within the Areas they report in different ways (see section 3.2.4). Both the FCS at Hospital 1 and Hospital 4 are within cancer services in their Areas, the Hospital 6 FCS is part of a stand-alone genetics unit which has been recently incorporated into Pathology North, the Area Health Service 4 service is part of the general genetics service which in turn is part of chronic and complex care, the FCS at Hospital 7 has also been incorporated into Pathology North and most of the outreach counsellors are in community health.

- Responsibility for the statewide planning and coordination of FCS resides with two bodies, GSAC and the Cancer Institute NSW, so direction is unclear.
- There are no formal agreements between the AHS which provide the outreach services and the AHSs who receive the outreach services. As a result there are problems of isolation for genetic counsellors in rural areas because they are sole practitioners who are employed by the AHS where they are located but who are clinically responsible to the service providing the outreach service- which in their case is both the general genetics service and the cancer genetics service because they provide both general and cancer genetic counselling. These services have no formal responsibility for the outreach counsellors in terms of mentoring, clinical supervision or continuing education.
- Potentially there is a shortage of medical staff for familial cancer services as there has not been a clear training path for cancer geneticists.
- Genetic counsellors are about to have their education requirements changed which may have an impact on the number of people undertaking genetic counselling courses.
- Differential funding of services has inevitably contributed to the variation in the way services are provided.
- There is variation between the laboratories in terms of the time it takes to complete the genetic testing.

1.7 Comparison of NSW with other states in Australia and two Provinces in Canada.

The main points of comparison of six services (four states of Australia and two provinces of Canada) have highlighted the following::

- Four of the six services are coordinated at the statewide or equivalent level- NSW and Ontario are the two jurisdictions that are not.
- Most FCS are decentralised in nature with the exception of South Australia (SA), which has the smallest population (1.5 million people in total). In SA there is one service for the state and staff provide outreach to some regional towns.
- The model of service is similar across all jurisdictions. There are mostly metropolitan services which provide outreach services to regional areas. In some cases (SA, Victoria) the staff travel from the metropolitan area to provide services whereas in others the genetic counsellors are based in the regional areas and medical staff travel to the regional areas from the metropolitan services (NSW, Queensland). Others have a mix of both approaches with medical and counselling staff travelling to some regional locations and genetic counsellors based in other regional locations (BC and Ontario).
- The extent to which protocols are used varies between jurisdictions.

- There is variation between jurisdictions in the clinical governance of the FCS. Some are located organisationally within cancer services, some are in general genetics and some are in pathology. In NSW, two of the largest services are in cancer services (Hospital 1 and Hospital 4) and one is in general genetics services (Hospital 6); of the smaller services, one is in general genetics (Health Service 4), one is in cancer (Hospital 5) and one is in pathology services (Hospital 7). All the outreach staff based in regional locations are in Community Health.
- The nature and extent of the follow up management varies by jurisdiction- two provide high risk clinics although this varies by FCS (NSW, Vic) , two do not provide any follow up management (SA, BC), and for two jurisdictions it was not known (Qld, Ontario).
- Outreach responsibilities are not equally divided between the three largest services in NSW. In the other jurisdictions (with the exception of Ontario) there was a statewide decision about how outreach services should be provided.
- Administration support in NSW was the least adequate.
- Two jurisdictions (SA, QLD) had a statewide database. In NSW a statewide database is about to be rolled out. Victoria, BC and Ontario do not have a statewide database.
- Laboratory waiting times were longest in NSW compared with other states in Australia. In both BC and Ontario laboratory waiting times were substantially worse than for any state in Australia. In BC the laboratory waiting times were seen as a real bottleneck in the system. In Ontario there are 13 laboratories involved and, as well long waiting times, there is variation in the quality of testing between laboratories.

1.7 Next Steps

It is recommended, given the issues highlighted above that;

1. CINSW facilitate a statewide planning exercise to develop a service model for cancer genetics services in NSW;
2. CINSW, in partnership with key stakeholders, produce a strategic plan for cancer genetics services in NSW for the next 5 years, 2011-2016; and,
3. CINSW and NSW Health identify respective roles and responsibilities in regards to familial cancer genetics services.

2. Introduction

2.1 Background to this Project

A key program of the Cancer Services and Education Division of the Cancer Institute NSW is the better coordination of specialised cancer care. In support of this program the Cancer Institute NSW has undertaken in 2007 a Review of Cancer Genetics Services in NSW in 2007 (The Review). The Review provides information and baseline data about the current cancer genetics services that people at risk of familial cancer are able to access in NSW. These services provide medical, counselling and testing services to individuals and families with or at risk of inherited cancer to help such individuals make informed choices about such risk.

The Review maps current Cancer Genetics Services in NSW, presents quantitative data on the numbers and types of patients using these services, current staffing levels, and the number and types of cancer genetics tests provided through laboratories. Qualitative data provides information about key trends, strengths and gaps, suggestions for improvements to services and the implications for current service provision.

The Review provided valuable information that will assist the Cancer Institute NSW to identify key trends, prioritise needs and assist in informing the development of an effective and coordinated strategic plan for cancer genetic services in NSW. A strategy for cancer genetics services would set out the principles and directions for the planning, development and delivery of services for the next 5 years and beyond.

Stage 2 of the project will offer an amylase the scope and data collected in the Review, although providing valuable baseline information is limited. Additional evidence to complete the picture of provision of cancer genetics services and to inform the development of a comprehensive Cancer Genetics Statewide Services Plan. The aim of this project is to obtain additional information and develop a statewide plan for the provision of cancer genetics services.

2.2 Project Scope

The main objective of this project was:

To provide options for a structure for statewide familial cancer genetics services (also referred to in this report as familial cancer services (FCS)), both clinical and laboratory, that will meet current and future demand and be accessible to all potential clients across NSW.

More specifically the plan will:

- Assess future services' demand taking account of developments in genetics knowledge;
- Examine state, national and international models of cancer genetics or familial cancer services delivery to inform development of options for NSW;
- Examine the Cancer Institute NSW's Review of Cancer Genetic Services in NSW and undertake an analysis of the strengths, gaps or weaknesses, opportunities and threats of current familial cancer genetics delivery;
- Undertake costing of current cancer genetics services and identify the source of funding;

- Propose options for current cancer genetics services' organisation, structure, governance and configuration, taking account of future demand, access across metropolitan and rural areas, coordination, integration, funding, and workforce sustainability;
- Model future workforce requirements, including how the workforce should be distributed;
- Articulate the relationship between cancer genetics services and broader cancer services, general genetics services, NSW Health and The Cancer Institute NSW;
- Comment on the role of a suitable information technology/patient management system;
- Undertake a cost/benefit analysis of proposed options including funding mechanisms;
- Assess current laboratory based genetic testing and screening capabilities;
- Propose options for laboratory cancer genetics service organisation and configuration;
- Identify laboratory workforce requirements;
- Identify appropriate mechanisms for funding laboratory services and purchasing testing;
- Propose statewide options to ensure the independent review of new tests for familial disease and to prioritise their introduction based on an assessment of need, benefit and cost; and
- Propose an implementation plan for an improved structure for Familial Cancer Genetics Statewide Services.

Inadequate data (both cost and utilisation), lack of access to information and data (laboratories), limited budget and project timeframe have made it not possible to complete the following tasks:

- A cost/benefit analysis of proposed options including funding mechanisms;
- The assessment of current laboratory workforce requirements;
- The identification of laboratory workforce requirements;
- The identification of appropriate mechanisms for funding laboratory services and purchasing testing; and
- The development of statewide options to ensure the independent review of new tests for familial disease and to prioritise their introduction based on an assessment of need, benefit and cost.

2.3 Project Liaison and Direction

A Steering Committee was established to provide advice to the consultant on the scope of the project and issues that may arise, and provide and facilitate access to information required.

2.4 Methodology

2.4.1 Consultations with key stakeholders.

Over 65 people were interviewed either in person or by phone as part of this project. They included the Medical Directors of all the FCS, most of the Genetic Counsellors in metropolitan FCS and outreach FCS (all were extended an invitation), the Directors of Molecular Laboratories at South Eastern Area Laboratory Services (SEALS), Hospital 6 Area Pathology

Service (HAPS) and Hospital 4, most of the Directors of Area Cancer Services (all were extended an invitation), most of the Cancer Services Development Managers (all were extended an invitation), one NSWOG Chair and the GMCT Gynaecology Chair (all NSWOG chairs were extended an invitation), staff of NSW Health's Statewide Services Development Branch (SSDB) and others who were recommended.

The Cancer Institute NSW provided an initial list of 35 people to consult. The additional 30+ people interviewed included another 16 Genetic Counsellors, other staff of the FCS (administrators or data managers) the chairs of GSAC, other laboratory staff including some from interstate, Directors and other staff of FCS in other states and internationally, medical and gynaecological oncologists who were referred by Directors of FCS, DACS or genetic counsellors.

The Cancer Institute NSW sent out a letter to the Chief Executives (CEs) of Area Health Services advising them of the project.

2.4.2 Review of the provision of FCS in other States and Internationally

Information on FCS in Victoria, South Australia and Queensland was obtained from interviews with staff of those services and from information on relevant websites. Discussions were also held with cancer genetics counsellors in British Columbia (BC) and Ontario in Canada regarding the form on which FCS are provided in those provinces. Information about those services was also obtained from relevant websites. The service provision model of FCS in the United Kingdom was gleaned from key reports and articles.

2.4.3 Data Analysis

Data on the utilisation of FCS services were obtained from the FCSs database, Genesis. As a result there are four years of data for the three largest services – 2005, 2006, 2007 and 2008. It was decided at the first Steering Committee meeting that it would not be practicable for the smaller services nor the outreach services to provide data in this format. These other services provided what data were available. In some of the outreach services, there were no data readily available and estimates were made of the number of familial cancer patients who were seen by those services.

Data on the number and type of laboratory tests performed were sought from the three molecular laboratories that do most of the breast and bowel testing for the FCS- SEALS, HAPS and the laboratory which is part of the FCS at Hospital 4. Only the laboratory at Hospital 4 provided the data that were requested.

To estimate the projected demand for FCS, projections on the incidence of cancer by type was obtained from a report by the Cancer Institute NSW (Tracey et al. 2009) and the methodology for undertaking the projections was based on an article by Featherstone et al. (2007).

The brief also sought a cost-benefit analysis. However basic cost information on services was not available nor were data on outcomes – both pre-requisites of a cost-benefit analysis. An estimate was made of the cost of services based on standard salary rates.

3. Current Familial Cancer Services in NSW

3.1 History of service provision

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 Statewide Services Development Branch (SSDB).

Increasing awareness of developments in cancer genetics created a demand from cancer families for access to cancer genetics services. A Cancer Genetics Working Party was established as part of GSAC to examine health and service implications. A report was produced in April 1996 which recommended, among other things:

- Four teams each consisting of a clinical geneticist, genetic counsellor, secretary and laboratory staff be established at four hospitals in Sydney where there is existing expertise in cancer genetics at the clinical and laboratory level together with existing oncology services.
- Services to be provided through integrated cancer genetics and oncology clinics to give maximum benefit to individuals attending.
- Cancer genetics centres to be used as a base from which to provide decentralised services to all Areas through outreach genetic counsellors and GPs.

In recognition of the newly developing field of cancer genetics, an allocation of \$720,000 in Statewide Service Improvement funding was made in 1997/98 to four AHSs to assist in setting up clinical and laboratory services.

Familial cancer services were subsequently established in Health Service 1, Area Health Service 2, Health Service 4 and Hospital 6. In Hospital 7 a cancer genetics service was offered between 1997/98 and 2000 but there was no cancer genetics service between 2001 and 2007. A cancer genetic counsellor was appointed in 2007. The genetics counsellors provides a cancer genetics service to both Hospital 21 and Hospital 22 and Hospital 6 cancer geneticist provides medical support to clinics at both Hospital 21 and Hospital 22 once a month. The cancer geneticist is on a VMO renewable contract while AHS 4 attempts to recruit a staff specialist in cancer genetics. These familial cancer services also provide outreach services to a number of rural centres where genetic counsellors are based.

NSW Health has produced two reports in 2002 and 2005 on genetics services in NSW. Both reports provide a description of services and some data on their utilisation. In 2004 a draft business plan was developed for the period 2003- 2006 which provided a plan for the development of services. It has 5 key result areas (KRA) with intended outcomes and performance indicators for each KRA. KRA 3 requires a strategic increase in capacity with a intended outcome 3.5 of 'Cancer genetics services are available state-wide'. Strategies include:

- Build on existing Familial Cancer Clinics and establish links to all Comprehensive Cancer Centres;
- Negotiate for a cancer genetic counsellor, or trained oncology nurse, in all Comprehensive Cancer Centres ie 5 additional counsellors at Hospital 21, Hospital 7, Hospital 22, Hospital 24, Hospital 3;
- Develop agreed uniform policies to be implemented in all familial cancer clinics;
- Develop a rational approach to the provision of cancer genetic testing in NSW;
- Determine funding needs for cancer genetic testing, including tumour testing
- Develop a workable system for cost recovery for tests;
- Negotiate for outreach services using telehealth links between regional genetics counsellors and major familial cancer clinics;
- Provide data to NSW Health on state-wide genetics services to support a state-wide cancer genetics data collection system to achieve a networked service;
- Explore the role of the NSW Familial Cancer Register within a networked service;
- Develop a cancer familial history questionnaire to triage average/above average risk families;
- Provide standardised written surveillance advice to all cancer genetics families seen by genetics services; and
- Implement on-going professional education for the cancer genetics workforce, particularly the currently employed genetics counsellors.

It appears that only some of these KRAs have been achieved – a statewide cancer genetics data collection system (KinTrack) is currently being rolled out and there has been a review of the NSW Familial Cancer Registry.

3.2 Current service arrangements

3.2.1 Overview

The current service configuration of familiar cancer services (FCS) in NSW can be summarised as follows:

- Three main services – The Familial Cancer Clinic at Hospital 1; The Hospital 4 Familial Cancer Service; and the Hospital 6 Familial Cancer Service within Hospital 6 Genetics Service.
- Smaller services – Familial Cancer Clinic at Hospital 5, Area Health Service 4 Clinic at Hospital 21; and Hospital and the Familial Cancer Service at Hospital 7.
- Outreach services provided by the FCS at Hospital 1 to Hospital 2, Hospital 3 Hospital 26.
- Outreach services provided by the FCS at Hospital 4 to Hospital 24, Hospital 17 and Hospital 19.
- Outreach services provided by the FCS in Hospital 6 to Hospital 15, Hospital 16, Hospital 8, Hospital 9, Hospital 10, Hospital 11, Hospital 12, Hospital 20, Hospital 14 and Hospital 18

3.2.2 Staffing

Staffing of the three main familial cancer services are provided in Table 1. The Area Director of Cancer Services for Area Health Service 1 provides a familial cancer service for half a day a week at Hospital 1 (0.1 FTE) and the Professor of Surgery at Hospital 5 provides the medical services at the FCS in the Hospital 6 for one day a week (0.2 FTE) as an Honorary Medical Officer (HMO).

Table 2. Staffing numbers (in FTE) of the main familial cancer services, 2009

Staff category	Hospital 1	Hospital 4	Hospital 6
Honorary Medical Officer (HMO)			0.2
Clinical academic	0.1		
Senior staff specialist	0.8	1.0	
Staff specialist		0.6	
Career Medical Officer (CMO)	0.7		
Genetic counsellor	2.6	2.0	1.6
Nurse - Risk Management Clinic		0.4	
Research nurse	1.0	0.2	
Admin officer	1.3	1.0	1.0
Data manager	0.7		0.5
Clerical assistant		0.2	
Total	7.2	5.4	3.3

Staffing numbers of the smaller services are provided in Table 2. The service at Area Health Service 4 has only one full time staff member at present- the genetic counsellor. The administration position is currently vacant and a VMO is providing the medical care one day a month (in lieu of the appointment of a staff specialist).

Table 2. Staffing numbers (in FTE) of the smaller familial cancer services, 2009.

Category	Hospital 5	AHS 4	Hospital 7
VMO		0.5	
Clinical Academic	0.05		
Staff Specialist			0.4
Genetic Counsellor	1.0	1.0	0.6
Admin Officer		0.5	
Total	1.05	1.55	1.0

Outreach services are provided by the Director of Familial Cancer Clinic at Hospital 4, Hospital 1 to Hospital 2, Hospital 3 and Hospital 25. There are genetic counsellors based at each of these hospitals (see Table 3). At Hospital 2 and Hospital 3 the counsellors provide only cancer genetic counselling. However at Hospital 25, the genetic counsellors do general and cancer genetics counselling. The Director of the Hospital 1 service visits patients once a month at Hospital 2 and provides telehealth consultations (one clinic per month) with patients at Hospital 25. Previously this provided telehealth consultations (one clinic per month) with patients at Hospital 3 but in 2010 the VMO is providing monthly visits.

Table 3. Staff numbers (in FTE) at the outreach services of the Hospital 1- Familial Cancer Clinic

Category	Hospital 2	Hospital 3	Hospital 26
Genetic Counsellor	1.0	0.8	1.25

The Familial Cancer Service at Hospital 4 provides outreach services to Hospital 17 and Hospital 19. Staff, (doctor and counsellor) from the FCS at Hospital 4 also travel every fortnight to Hospital 24 and see patients there. The medical clinics at Hospital 17 are not operating at the present time. The doctor used to fly from Hospital 4 but most recently medical consultations have been via telehealth. Hospital 19 was without a counsellor for some time however a genetic counsellor has been recently appointed and the clinics have recommenced. Telehealth is mostly used. The genetic counsellors at Hospital 17 and Hospital 19 (Table 4) see patients with cancer and their relatives as well as patients with general genetics conditions. General geneticists visit in addition to the cancer geneticists and the counsellors divide their time between the two groups of patients. The counsellor at Hospital 19 estimates that 60% of time is spent on cancer patients and their relatives. The Hospital 17 counsellor estimated that she previously spent 40% of time was previously spent with cancer patients when the medical telehealth clinics were operational but the amount of time has decreased to about 20% now that the medical clinics have ceased.

Table 4. Staff numbers in FTE at the outreach services of the Familial Cancer Service at Hospital 4 Hospital

Category	Hospital 17	Hospital 19
Genetic Counsellor	1.0	1.0

The HMO at the Familial Cancer Service within Hospital 6 provides outreach to all the hospitals in Table 5. In each town there is a full or part time counsellor who works with the HMO from the Familial Cancer Service of Hospital 6 to provide services to people with inherited cancer or to their relatives. The counsellors also work with general geneticists to provide general genetics services. On average they mostly estimate that they spend about half of their time on cancer genetics and half on general genetics. The HMO from the Familial Cancer Service of Hospital 6 does telehealth consultations to patients in Hospital 20, Hospital 12 and Hospital 15 but visits all the other towns. The counsellor position in Hospital 16 is currently vacant so there are no outreach services to Hospital 16 at the present time.

Table 5. Staff numbers in FTE at the outreach services of the Familial Cancer Service of Hospital 6

	Hospital 12	Hospital 20	Hospital 14	Hospital 11	Hospital 10	Hospital 9	Hospital 8	Hospital 15	Hospital 16	Hospital 18
Genetic Counsellor	1.0	1.0	0.6	1.0	1.0	0.5	0.5	1.0	0.2	1.0

Table 6 shows the number of staff by staff category in FTEs by AHS. The staff in each Area includes the outreach staff for that service. For example, the staff in Area Health Service 1 includes all the staff in the FCS at Hospital 1, Hospital 2, Hospital 3, Hospital 5, and Hospital 25. The staff in AHS 2 includes the FCS staff at Hospital 4, Hospital 17 and Hospital 19. The FCS staff for Area Health Service 3 includes the outreach counsellors where the Hospital 6 Familial Cancer Service provides an outreach service – at Hospital 8, Hospital 9, Hospital 10, Hospital 11, Hospital 12, Hospital 20, Hospital 18 and Hospital 15. As the counsellors in most of the outreach locations provide both cancer and general genetic counseling, only half of their positions are included here to provide an estimate of the cancer genetic counselling component.

Table 6 also provides the 2007 population of the AHS where the service is located and the population of the AHS where outreach is provided to give an indication of the catchment population. A staff to population ratio is calculated to give some indication of the variation in the distribution of services. The staff/population ratio is lowest in AHS 1 at 1:134,000 and highest at in AHS 4 and AHS 5 at 1:866,000 and 1:850,000 respectively.

Table 6. Staff in FTEs (including outreach staff) providing cancer genetics services by AHS

	AHS 1	AHS 2 & Outreach	AHS 3 & outreach	AHS 4	AHS 5
HMO			0.4		
VIVIO				0.05	
Clinical Academic	0.15				
Senior Staff Specialist	0.8	1.0			
Staff Specialist		0.6			0.4
CIVIO	0.7				
Genetic Counsellor	6.65	3.0	5.3	1.0	0.9
Nurse – RIVIC		0.4			
Research Nurse	1.0	0.2			
Admin Officer	1.3	1.0	1.0	0.5	
Data Manager	0.7		0.5		
Clerical Assistant		0.2			
Total	11.30	6.4	7.20	1.55	1.3
2007 Population of Catchment	1,515,667	1,484,192	1,711,362	1,342,316	1,104,624
Staff/Population Ratio	1:134,130	1:231,905	1:237,689	1:866,010	1:849,711

Some indication of the variation in the way that services are provided is given by the doctor/counsellor ratio (Table 7). These vary significantly between services. These have been calculated on the basis of all staff (including outreach staff) providing the service. The Hospital 6 Familial Cancer Service and its outreach have the equivalent of 1 doctor to 13.5 counsellors compared to 1 doctor to 1.9 counsellors at the Hospital 4 Familial Cancer Service.

Table 7. Doctor/Counsellor Ratio

Service	Actual Doctor/ Counsellor Ratio	Standardised Doctor/ Counsellor Ratio
AHS 1	1.65:6.65	1.0:4.0
AHS 2	1.6:3.0	1.0:11.9
Hospital 6	0.45:4	1.0:13.5
AHS 4	0.05:1.0	1.0:20.0
AHS 5	0.4:0.9	1.0:2.25

2.3.3 Workforce Training

Doctors

The medical staff at the three main FCS at Hospital 1, Hospital 4 and Hospital 6 have come to the subspecialty of cancer genetics from different backgrounds. The director of the FCS at Hospital 1 is a clinical geneticist; the director of the FCS at Hospital 4 is a paediatric oncologist and the HMO at the FCS in Hospital 6 is a surgeon. Certification as a Clinical Cancer Geneticist is granted by the Special Society (Human Genetics Society of Australasia (HGSA)).

Counsellors

Genetic counsellors come from a variety of backgrounds – nursing, psychology, social work, science. Most current genetic counsellors have an undergraduate degree in one of the disciplines listed and have a 1- year post graduate diploma in genetic counselling – mostly from the University of Newcastle. To become fully accredited as a genetic counsellor, the genetic counsellor has to do certification training (also granted by the HGSA) while working, which takes a minimum of one year. The postgraduate diploma course at the University of Newcastle has recently been abolished and the requirement for genetic counselling will be a 2 year masters degree. A new degree is currently being developed and current estimates are that it will cost the prospective student in the order of \$50,000. Certification requirements of the HGSA following the masters also have to be finalised. Initial indications, however, suggest that a genetic counsellor working in a FCS cannot achieve certification while working solely in FCS and that they have to spend some time working in a general genetics service.

3.2.4 Governance

The Area Health Service Executive consists of the following positions:

- Chief Executive
- Director of Clinical Operations
- Director of Finance and Corporate Services
- Director of Clinical Governance
- Director of Nursing and Midwifery services
- Director of Population Health, Planning and Performance
- Director of Workforce Development

Each Area has a matrix organisational structure with Directors of Operations of Acute hospitals/networks/clusters and Directors of Primary and Community Care services/networks/clusters who are operationally responsible for the provision of hospital and community services.

There are also Area Directors of a range of services – Cancer, Cardiovascular and Respiratory, Surgery and Anaesthetics, Chronic, Complex and Aged Care, Women, Children and Youth, Critical Care and Emergency, Mental Health and Drug and Alcohol, Allied Health, Pathology, Imaging, Pharmacy, etc. These Directors do not have direct line responsibility for staff but work strategically in their stream across the Area. Both the operational managers of acute and community services and the Area Directors of clinical streams mostly report to the Director of Clinical Operations on the Area Executive.

Whilst this is a basic description of a complex organisational structure and there are some differences between Areas however it describes what occurs in most AHSs.

The Director of the Familial Cancer Clinic at Hospital 1 reports to the General Managers of the Northern Hospital network of AHS 1 (comprising Hospital 1, Hospital 26 and Hospital 27) who in turn reports to the Director of Clinical Operations. The Familial Cancer Clinic at Plan for NSW Familial Cancer Services

Cancer Institute NSW

Hospital 1 is part of the Clinical Program 2 which includes emergency medicine and Cancer services which also has a dotted line relationship (strategic not operational) to the Director of Area Cancer Services.

At Hospital 2 the cancer genetic counsellor reports operationally through the Surgical, Preoperative and Cancer stream to the General Manager of the Central Hospital Network which included Hospital 2 and Area Health Service 1. The General Manager of the Central Hospital network reports to the Area Director of Clinical Operations. The manager of the clinical stream of Surgery, Perioperative and Cancer services has a dotted line relationship (strategic not operational) to the Director of Area Cancer Services. The genetic counsellor is clinically responsible to the Director of the Familial Cancer Clinic at Hospital 1. However there is no line responsibility between the Director of the Familial Cancer Clinic at Hospital 1 and the cancer genetic counsellor at Hospital 2 even though they are in the same AHS.

At Hospital 3 the cancer genetic counsellor reports to the Nursing Co-Director of Critical Care and Cancer who reports to the Director of Clinical Services at Hospital 3 who in turn reports of the Southern Hospital Network (SHN) Manger who reports to the Area Director of Clinical Operations. The Clinical Division of Cancer services of the Southern Hospital Network also has a dotted line relationship (strategic not operational) to the Director of Area Cancer Services. The genetic counsellor is clinically responsible to the Director of the Familial Cancer Clinic at Hospital 1 and they meet every two weeks. However there is no line responsibility between the Director of the Familial Cancer Clinic at Hospital 1 and the cancer genetic counsellor at Hospital 3 even though they are in the same AHS.

At Hospital 4 the Director of the Familial Cancer Services reports to the General Manager of Hospital 4 and Eastern Hospitals through the Cancer Program. The Director also reports strategically to the Director of the Cancer Network (also known as the Director of Area Cancer Services).

The Hospital 6 Familial Cancer Service is part of Hospital 6 Genetics which is a stand-alone service comprising general genetics, cancer genetics and the Genetics of Learning Disability (GOLD) Service. An HMO provides the medical services for the Familial Cancer Service. He reports both to the Clinical Director and the Service Manager of Hospital 6 Genetics. The Clinical Director and Service Manager of Hospital 6 Genetics report to the Director of Operations, Primary and Community Networks who in turn reports to the Area Director of Clinical Operations. Unlike the FCS at Hospital 1 and Hospital 4, the Hospital 6 Familial Cancer Services is not in an acute hospital therefore does not report to the General Manager of an Acute Hospital/Network. The cancer services stream also does not have a strategic relationship with the Area Director of Cancer services. Hospital 6 Genetics is about to be incorporated into Pathology North, an amalgamation of all pathology services in AHS 5, Hospital 6 and Area Health Service 6. Presumably this new entity will report to the Director of Clinical Operations. In this new structure, the Hospital 6 Familial Cancer Service will be part of Pathology services not Cancer services.

The Familial Cancer Service at Hospital 7 will also be incorporated into Pathology North and it is not yet clear how that service will relate to the Familial Cancer Service in Hospital 6 Genetics.

The Area Health Service 4 Familial Cancer Clinic is currently part of the Area Chronic and Complex Care stream not the cancer stream. However when a staff specialist cancer geneticist is appointed the service will be located within the Cancer stream. At the present time medical services are provided on a temporary VMO basis by the Professor of Surgery and Director of Cancer at Hospital 5, who is also the HMO at the Hospital 6 Familial Cancer Service.

At Hospital 5 the Familial Cancer Service is part of Cancer services. As Hospital 5 is a third schedule hospital it is not part of the hospital network of AHS 1.

The outreach counsellors are employed and funded by the AHS where they are located and are usually based in community health centres. They report to the general manager of the community health service where they are based who reports to the network/cluster general manager who reports to the Director of Primary and Community Health who in turn reports to the Area Director of Clinical Operations. In Area Health Service 6 both the hospital and community health managers in the four networks report to one of the four network coordinators who report directly to the Director of Clinical Operations. Mostly the outreach counsellors provide both cancer and general genetics counselling. As described, they are operationally responsible to their local network/cluster/hub managers and the Director of Primary and Community Health and the Director of Clinical Operations yet they are clinically responsible to the general geneticists and cancer geneticists who visit to provide clinics. However there are no formal agreements between the AHS providing the outreach services and the AHS receiving those outreach services.

Statewide planning and coordination is the responsibility of the NSW Genetic Service Advisory Committee (GSAC) according to the Draft Business Plan of the NSW Genetics Service. GSAC is supported by the Statewide Services Development Branch of the Strategic Development Division, NSW Department of Health.

In August 2005, the NSW Oncology Group (NSWOG) was established by the Cancer Institute NSW to bring together cancer specialist doctors and nurses, consumers, patients and planners. NSW Oncology Groups (NSWOGs) were established with 12 cancer specific groups and 8 discipline groups, (one of which is cancer genetics). The roles and responsibilities of NSWOGs, as articulated on the Cancer Institute NSW website are:

- To advise on standard treatments and the treatment pathways and protocols for site specific cancers;
- To encourage an increased participation in clinical trials and other research;
- To establish the data requirements for the specific cancer in addition to the minimum data set;
- To develop educational programs to encourage sub-specialisation and exchange of knowledge for the specific cancers; and
- To advise on other issues to promote best practice in the specific types of cancer.

It appears that with the creation of NSWOG Cancer Genetics, the Cancer Genetics Working Party of GSAC was disbanded and the members of that committee joined the NSWOG Cancer Genetics. However it is not clear who has the overall planning and coordination responsibility for familial cancer services because in addition to this project CINSW commissioned GSAC to develop a framework for Genetics Services which includes FCS.

3.2.5 Funding

Familial Cancer Services are funded from Area Health Service budgets. The FCS at Hospital 1 and Hospital 4 employ staff specialist who have certain rights to private practice and they bill Medicare for some of their consultations. A proportion of the revenue they generate goes to the hospital and a proportion goes back to the service. The service at Hospital 6 is provided by an Honorary Medical Officer who receives payment directly from Medicare for the consultations provided.

The FCS at Hospital 1, Hospital 3, Hospital 4, Hospital 6 and Hospital 7 are all dependent on some non recurrent funding from the Cancer Institute NSW which was due to expire in June

2010 but has been extended until the end of June 2011. Table 8 shows the number of FTEs by staff category who are dependent on temporary funds by service, the total number of FTEs per service who are dependent on temporary funds and FTEs dependent on temporary funding as proportion of total FTEs. A significant proportion of the three largest services – 14% and 15% in the case of Hospital 1 and Hospital 6 and a third of the total FTE at Hospital 4 – are funded on a temporary basis.

Table 8. FTE staff numbers dependent on temporary funding by service, 2009

	Hospital 1	Hospital 3	Hospital 4	Hospital 6	Hospital 7
Senior Staff Specialist			0.2		
Staff Specialist			0.6		0.2
Genetic Counsellor	1.0	0.8	0.4		
Nurse – RMC			0.4		
Research Nurse			0.2		
Data Manager				0.5	
Total Temporary FTEs	1.0	0.8	1.8	0.5	0.2
Total FTEs	7.2	0.8	5.4	3.3	1.0
Temporary FTEs as a percentage of Total FTEs	13.9%	100%	33.3%	15.2%	20%

3.2.6 Costing

In order to obtain information about the cost of NSW familial cancer services, budgets were requested from services. However budgets were not always available and those that were available were not necessarily comparable. Some included superannuation, while others did not. Some had a budget for pathology, while others did not.

To get a standardised view of the cost of the clinical familial cancer services, the staff of services were costed using median award rates for each staff category. Allowances and on-costs were not included, so the costs may be an under estimation but they are at least comparable. Table 9 shows the estimated cost of staff at each of the services including the outreach services.

Table 9. Estimate of the cost of staff providing clinical familial cancer services in NSW

	AHS 1	AHS 2 and outreach	AHS 3 and outreach	AHS 4	AHS 5	Total
Total	\$889,000	\$674,000	\$563,000	\$135,000	\$115,000	\$2,376,000

Table 10 estimates the cost of the molecular genetics tests ordered by the familial cancer services. It is not clear whether the tests ordered for patients seen in outreach are included in the numbers of the services who provide the outreach. In other words, it is not clear whether the tests ordered for patients seen at Hospital 3 and Hospital 2 are included in the Hospital 1 numbers. If they are, then the tests for Hospital 3 are double counted. There are a number of services for which testing data are not available (n/a). Based on the data available, testing costs are estimated to be in excess of \$2m in 2008.

Table 10. Number of molecular genetics tests requested by each service in 2008 and the estimated cost of these tests.

Familial Cancer Service	Number of Mutation Screens	Number of Predictive Tests
Hospital 1	212	160
Hospital 3	41	22
Hospital 2	n/a	n/a
Hospital 4	153	163
Hospital 17	n/a	n/a
Hospital 19	n/a	n/a
Hospital 6	293	145
Hospital 12	n/a	n/a
Hospital 20	n/a	n/a
Hospital 14	n/a	n/a
Hospital 11	n/a	n/a
Hospital 10	n/a	n/a
Hospital 9	n/a	n/a
Hospital 8	n/a	n/a
Hospital 18	n/a	n/a
Hospital 15	n/a	n/a
Hospital 16	n/a	n/a
Hospital 28	n/a	n/a
AHS 4	146	53
Hospital 7	92	39
Total	937	582
Cost Per Test	\$2000	\$300
Total Estimated Cost	\$1,874,000	174,600

If it is assumed that the salaries and wages in Table 9 are 80% of total costs (excluding pathology) then the total clinical costs would be \$2,970,000. If it is assumed that pathology costs are an additional \$2m, then it would seem that the total cost (clinical and pathology) of providing the current services would be in the order of \$5m.

3.2.7 Utilisation of FCS

Data on the utilisation of all FCS was not readily available. The FCS at Hospital 1, Hospital 4 and Hospital 6 all use the Genesis database which each service has modified for its own purposes. There are no standard data definitions so it is not clear how comparable the information is. The outreach counsellors do not have Genesis but some of them use a statewide genetics database, known as CRS. Others record patient numbers and occasions of service in CHIME, the community health database.

It was decided at the first Steering Committee meeting of this project that data would only be obtained from the three main services at Hospital 1, Hospital 4 and Hospital 6 and in the format that was provided to the previous review. Other services would provide whatever they could. In some of the other rural Areas only estimates of total numbers of patients could be made as there was no electronic data system.

- The FCS at Hospital 1, Hospital 4 and Hospital 6 each saw around 600 new families in 2008;
- The total number of patients (new and returning) in 2008 was 1440 for the Familial Cancer Clinic at Hospital 1, 1304 for the Familial Cancer Service at Hospital 4 and 1261 for the Familial Cancer Service at Hospital 6. ;
- The estimated total number of new referrals seen by all FCS for NSW in 2008 was 3464;
- Where these new referrals were seen is estimated as follows:
 - 18 % at Hospital 1
 - 17% at Hospital 4
 - 17% at Hospital 6
 - 10% at AHS 4
 - 6% at Hospital 7
 - 6% at Hospital 3
 - 6% at Hospital 2
 - 3% at Hospital 5
 - 17% in rural outreach
- For the three FCS at Hospital 1, Hospital 4 and Hospital 6, approximately 60% of new referrals live in the AHS where the service is located; and
- At the FCS at Hospital 1, Area Health Service 4 and Hospital 7 around 80% of referrals are from other specialists; at the FCS at Hospital 4 less than 50% of referrals are from other specialists and about 50% are from GPs; at the FCS, Hospital 6 57% of referrals are from other specialists and 38% are from GPs.

3.2.8 Services for Children

There is a higher proportion of children with inherited cancer than adults. However the numbers of new cancer cases are in NSW about 250 children get cancer per year compared with 40,000 adults.

There are three types of cancer referrals among children to genetics services:

- Children with pediatric cancer who required genetic testing;
- Children who may have a syndrome (eg. as Wilms syndrome) that puts them at increased risk of cancer; and
- Well patients who may be a risk of cancer in later life.

There is only a small group of inherited pediatric onset tumor. Services for these children are provided by the medical oncologists and clinical geneticists at the three children's hospitals in NSW. However these tumours have ongoing implications for other family members and for the children themselves. Some of the adult family members may need to see the adult FCS. GPs need to manage the ongoing surveillance of the adults and children.

In planning for FCS, consideration needs to be given to the provision of paediatric services because of the long term implications for both the children and their families.

3.2.9 Laboratory Services

Three main public laboratories provide genetic testing for inherited cancer in NSW:

- Hospital 6 Molecular Genetics laboratory within Hospital 6 Area Pathology Service (HAPS);
- Genetics Laboratory Services within South Eastern Area Laboratory Services (SEALS); and
- A small molecular genetics laboratory within the Familial Cancer Service at Hospital 4. This laboratory is not part of the Institute of Clinical Pathology Medical Research (ICPMR) at Hospital 4.

The HAPS laboratory does all the bowel cancer testing for NSW and the breast cancer testing for the Hospital 6 Familial Cancer Service and some of its outreach. The SEALS laboratory does the breast cancer testing for the Familial Cancer clinic at Hospital 1 and some of the testing of rare disorders. Bowel cancer testing for the Hospital 1 clinic is sent to HAPS. The laboratory at Hospital 4 does the breast cancer testing for the Familial Cancer Service at Hospital 4. Hospital 4 sends bowel cancer testing to Hospital 6. Testing for rare disorders for all services goes to a number of laboratories in NSW, interstate or overseas depending on the required test.

There is also a private genetics laboratory in Melbourne – Genetics Technologies Group (GTG) – which holds the license for breast cancer testing in Australia from Myriad Genetics in the US, the company which holds the patent on BRCA1 and 2 testing. The GTG has been used by the familial cancer service at AHS 4 and some of the outreach services use GTG in Melbourne for breast cancer testing and the familial cancer services at Hospital 1, Hospital 4 and Hospital 6 at times when their own laboratory services' waiting times have blown out.

There is variation in waiting times between all laboratories and at each laboratory these times may fluctuate from time to time. Mutation screens take longer than predictive tests, with mutation screens taking from between 2-3 weeks for urgent cases to 8 months or more. The average waiting time is 3-4 months. Predictive tests are quicker, usually taking between 4 to 6 weeks.

It was not possible to get utilisation data for the laboratories themselves, with the exception of the molecular laboratory at Hospital 4. Data on tests requested by the FCS was obtained from the FCS themselves but the actual workload of each of the laboratories was not provided. Each laboratory is quite different. Most of the testing (90-95%) in the HAPS molecular laboratory is for inherited cancer. This is not the situation at SEALS where inherited cancer testing is a smaller proportion of the total workload that includes both germline and somatic testing. At Hospital 4, the laboratory was established as part of the Familial Cancer Service and it does all the breast cancer testing for patients of the service.

3.3 Demand projections for FCS in NSW

3.3.1 Population by Area Health Service

Familial cancer services are required for the population of NSW. The population distribution and projected growth by Area Health Service (AHS) is given in Table 11. Sydney West and Sydney South West are expected to show the greatest growth and the North Coast is the fastest growing AHS- all three AHS are expected to have greater growth to 2016 than the average of 11.6% for NSW.

Table 11 Population distribution and projected growth by AHS

Area Health Service	2006	2011	2016	Per Cent Change Between 2006 & 2016
Greater Southern Area Health Service	473,578	492,985	512,259	8.2
Greater Western Area Health Service	300,528	301,560	302,331	0.6
New England Area Health Service	844,765	880,812	915,938	8.4
North Coast Area Health Service	479,544	511,146	542,696	13.2
Northern Sydney / Central Coast Area Health Service	1,104,624	1,158,068	1,203,417	8.9
South Eastern Sydney / Illawarra Area Health Service	1,173,593	1,237,286	1,282,555	9.3
Sydney South West Area Health Service	1,342,316	1,447,390	1,559,596	16.2
Sydney West Area Health Service	1,097,139	1,178,394	1,284,710	17.1
Grand Total	6,816,087	7,207,642	7,603,502	11.6

3.3.2 Epidemiology of inherited cancer

It is estimated that between 5% and 10%¹ of all cancer is inherited. Genetic testing of these families means that family members who are at high risk of developing cancer can be advised of appropriate surveillance and risk-reduction strategies, whereas members of their family who have not inherited the mutated gene are at no higher risk of cancer than the general population and can be thus reassured.

It is estimated that 5-10% of breast cancer cases, 10% of ovarian cancer cases and 10-15% of colorectal cancer cases are due to inherited genetic mutations. Women with a BRCA1 mutation have a lifetime risk of up to 80-90% of developing breast cancer, 40-60% chance of developing ovarian cancer and possibly an increased risk of developing colorectal cancer. Mutations in the familial genes nonpolyposis colorectal cancer (HNPCC) and familial adenomatous polyposis (FAP) predispose carriers particularly to colorectal cancer. A HNPCC germline mutation confers up to an 80-90% lifetime risk of developing colorectal cancer during their lifetime. Familial genes nonpolyposis colorectal cancer and FAP also convey risks of developing cancers in the duodenum, ureteric tract and other organs.²

Table 12 shows that incidence of all cancers in 2007 by AHS. If we assume that between 5% and 10% of cancer is inherited, there are between 1798 and 3595 individuals who inherited their cancer in 2007. Table 12 also shows the estimated number of people with inherited cancer in 2007 by AHS. It is these patients and their families who should be referred to cancer genetics services.

Table 12 Cancer incidence by AHS in 2007

Area Health Service	Cancer Incidence in 2007	5% of Cancer Incidence in 2007	10% of Cancer Incidence in 2007
Sydney South West	5636	282	564
South East and Sydney/Illawarra	6298	315	630
Sydney West	4593	230	459
North Sydney and Central Coast	6446	322	645
Hunter New England	5263	263	526
North Coast	3369	168	337
Greater Southern	2686	134	269
Greater Western	1659	83	166
Total	35950	1798	3595

3.3.3 Projected Need for Familial Cancer Services

Featherstone³ estimated the optimal proportion of new patients diagnosed with cancer who require assessment and evaluation for familial cancer genetic risk. They argue that ‘an optimal service should include referral of patients who are at high and moderate probability of having a genetic susceptibility, where confirmation of the familial history and more detailed assessment of their probability of genetic predisposition can be determined by genetic counsellors and specialists’. They assess the proportion of cases of each cancer type that should be referred and calculate ‘the optimal proportion of patients diagnosed with cancer who should be referred for genetic assessment for high or moderate risk was calculated to be 0.07. That is, 6 % of all patients diagnosed with cancer in Australia should be referred for genetic assessment, based on the best available evidence. The optimal proportion of ‘high probability’ patients is 1%.

On the basis of the above calculation, 6% of patients diagnosed with cancer in 2007 and 6% of the projected numbers of cancer patients in 2011 and 2016 were calculated (Table 13). These numbers represent the indexed cases or the number of people affected with cancer. Inevitably relatives of these people should also be referred for assessment, counselling and testing. The number of relatives requiring referral will range from none to several.

Table 13 Cancer incidence in 2007 and cancer projection in 2011 and 2016 by AHS

Area Health Service	2007		2011		2016	
	Actual cancer cases	6% of cancer cases	Projected cancer cases	6% of projected cancer cases	Projected cancer cases	6% of projected cancer cases
Area Health Service 4	5636	338	6422	385	7408	444
Area Health Service 1	6298	378	7076	425	7853	471
Area Health Service 2	4593	276	5109	307	5986	359
Area Health Service 5	6446	387	6881	413	7559	454
Area Health Service 3	5263	316	5599	336	6353	381
Area Health Service 6	3369	202	3948	237	4581	275
Area Health Service 8	2686	161	2990	179	3426	206
Area Health Service 7	1659	100	1847	111	2043	123
Total	35950	2157	39872	2392	45209	2713

For the purpose of estimating the potential demand of cancer genetics services, it is assumed that for half of the projected indexed cases, a definitive causative gene fault would be found, so testing could be offered to an average of another five family members. Table 14, column 4, shows the estimated number of people (affected and unaffected by cancer) by AHS who could benefit from referral to cancer genetics services in 2007. Column 7 and Table 14 shows the estimated number of people (affected and unaffected by cancer) by AHS who could benefit from referral to cancer genetics services in 2007. Column 7 shows the estimated number of people (affected and unaffected by cancer) by AHS who could benefit from referral to cancer genetics services in 2011 and column 10 shows the estimated number of people (affected and unaffected by cancer) by AHS who could benefit from referral to cancer genetics services in 2016 – ie 5981 and 6781 respectively.

Table 14. Actual (2007) and projected cancer cases (2011 and 2016) by AHS, estimated numbers with inherited cancer by AHS and estimated numbers of affected and unaffected cases who should be referred to a cancer genetics service.

	2007			2011			2016		
AHS	Actual number of cancer cases	Estimated numbers with inherited cancer	Estimated numbers for Referral for cancer genetic services	Projected Numbers of cancer cases	Estimated number with inherited cancer	Estimated numbers for referral to cancer genetics services	Projected numbers of cancer cases	Estimate numbers with inherited cancer	Estimated numbers for Referral to cancer genetics services
4	5636	338	845	6422	385	963	7408	444	1111
1	6298	378	945	7076	425	1061	7853	471	1178
2	4593	276	689	5109	307	766	5986	359	898
5	6446	387	967	6881	413	1032	7559	454	1134
3	5263	316	789	5599	336	840	6353	381	953
6	3369	202	505	3948	237	592	4581	275	687
8	2686	161	403	2990	179	449	3426	206	514
7	1659	100	249	1847	111	277	2043	123	306
Total	35950	2157	5393	39872	2392	5981	45209	2713	6781

3.4 Challenges facing the current Familial Cancer Services

- Unequal access by the population to FCS;
- Outreach services have developed on an ad hoc basis by the three main FCS;
- Use of telehealth for outreach is adversely influenced by the fact that it cannot be reimbursed through Medicare despite the fact that FCS medical consultations are suited to this medium;
- There is variation between FCS in the way services are provided – the counsellor role differs between services as indicated by the very different doctor/counsellor ratios by FCS;
- The three largest services depend on temporary funding – between 14% and 33% of their total budget;
- There is not a clear training path for medical specialists so it is difficult to attract trainee clinical geneticists;
- Training for genetic counsellors is currently being revised with genetic counsellors required to do an undergraduate and masters degree (minimum of 5 years) at considerable cost which may make it difficult to attract people to the profession, especially if their future role in FCS is not clear and can vary from service to service;
- Clinical governance of FCS differs between services. Some services are located within the Cancer stream, some are in Primary and Community Health and one is

currently in the Chronic and Complex Care stream but will move to Cancer when a staff specialist clinical cancer geneticist is appointed. There are no formal service agreements between the AHS providing the medical outreach services. If there is a vacant genetic counsellor position in outreach, no medical outreach services are provided until the position is filled;

- Statewide planning and coordination is currently being undertaken by both the Cancer Institute NSW and GSAC with no clear definition of the roles and responsibilities of the two organisations in relation to planning for FCS;
- There will be increasing numbers of patients who should be referred to the FCS. In 2008 it is estimated that there were around 3500 new referrals to FCS in NSW. This falls far short of the estimated number of people who would need the service in 2011 (around 6000 people) and in 2016 (approximately 6800 people). The demands on the FCSs are set to increase exponentially as new genes linked to inherited cancer are discovered and as there is a greater need for the FCS to become more acute with testing done at the time of patient diagnosis because treatment will depend on the results;
- There is variation in the waiting time for tests by the laboratories from 2-3 weeks for urgent cases to 12 months. As the clinical FCS begin to operate more acutely, it is not clear whether the current laboratories will be able to handle the workload; and
- There is variation between services in terms of data collection. The three main FCS use Genesis but they all modified it to suit their own purposes and there are no common data definitions. The outreach services do not use Genesis – some do not have a data system at all, others have to enter data twice – into CRS, a genetics database, and the community health database.

4. Review of Models of Familial Cancer Service Delivery in Other Australian States and Internationally

Table 15, summarises and compares NSW FCS against those in other Australian States and the UK and Canada. A more detailed description of these FCSs is given in Appendices M and N.

The most relevant comparison for NSW is with other States in Australia and the provinces in Canada because these jurisdictions are most similar in terms of population size, geography and the federated nature of their health services. The UK is a national health service, the population is much larger and the funding arrangements are substantially different.

4.1 Comparison of NSW with other states in Australia and two provinces in Canada

The main points of comparison of the six services (four states of Australia and two provinces of Canada) are:

- Four of the six services are coordinated at a statewide or equivalent level – NSW and Ontario are the two jurisdictions that are not.
- Most FCS are decentralised in nature with exception of SA which has the smallest population (1.5 million people in total). In SA there is one service for the state and staff provide outreach to some regional towns.
- The model of service is similar across all jurisdictions. There are mostly metropolitan services which provide outreach services to regional areas. In some cases (SA, Victoria) the staff travel from the metropolitan area to provide services whereas in others the genetic counsellors are based in the regional areas and medical staff travel to the regional areas from the metropolitan services (NSW and Queensland). Or they can have a mix of both approaches with medical and counselling staff travelling to some regional locations and genetic counsellors based in other regional locations (BC and Ontario).
- The extent to which consistent protocols are used varies between jurisdictions.
- There is variation between jurisdictions in the clinical governance of the FCS. Some are located organisationally within cancer services, some are in general genetics and some are in pathology. In NSW itself two of the largest services are in cancer services and one is in general genetics of the smaller services, one is in general genetics, one is in cancer and one is in pathology. All the outreach staff based in regional locations are in community health.
- The nature and extent of follow up management varies by jurisdiction - two provide high risk clinics although this varies by FCS (NSW, Vic), two do not provide any follow up management (SA, BC) and for two jurisdictions it was not known (Qld, Ontario).
- Outreach responsibilities are not equally divided between the three largest services in NSW. In other jurisdictions (with the expectation of Ontario) there was a statewide decision about how outreach services should be provided.
- Administration support in NSW was by far the least adequate (table 6).
- Two jurisdictions (SA, Qld) had a statewide database. In NSW a statewide database is about to be rolled out. Victoria, BC, and Ontario do not have a statewide database.
- Laboratory waiting times were longest in NSW, (up to 8 months) compared with the other States in Australia. In both BC and Ontario laboratory waiting were waiting times were substantially worse than for any State in Australia. In BC the laboratory waiting times were seen as a real bottleneck in the system. In Ontario there are 12 laboratories involved and, as well as long waiting times, there is variation in the quality of testing between laboratories.

4.2 United Kingdom

In the UK a model was defined for the provision of FCS in 1996 in the Harper report.⁴ It recommended the organisation of cancer genetics services for England and Wales in a 3-tier structure:

- Primary Care
- Cancer Units
- Specialist genetics services

Despite the existence of this model, a national study⁵ in 2001 showed ongoing great diversity in all aspects of cancer genetics care provision.

In 2003 a genetics white paper⁶ recognised the potential impact of genetics on lives and on healthcare. One of its initiatives was to co-fund with Macmillan Cancer Relief pilots in six cancer network areas of a model of service delivery for people at risk of familial cancer. Issue 2 of the 2007 edition of the Journal of Familial Cancer outlines the progress of the pilots to date, gives an indication of the different approaches used and highlights the issues that have arisen.

Two important outcomes of these projects should be noted:

- All projects required non genetics practitioners to undertake risk assessment activities within agreed protocols and with close supervision, mostly in mainstream services. 'For all projects, the most important element was the support of the genetics department, including on-going direction and mentorship, clinical supervision and observation and continuing opportunities to discuss cases with colleagues and with experts at the genetics centre...[Furthermore] it was important that the relationship of practitioners with the specialist genetics department was formalised; and; Issues relating to cancer genetics straddle two specialties (genetics and oncology). This made it difficult to engage managers, establish service sustainability and secure on-going funding.

More detail of FCS in the UK is given in Appendix M and N.

Table 15 Summary of key characteristics of FCS in NSW and other States in Australia, the UK and Canada.

Characteristic	NSW	Vic	SA	Qld	UK	British Columbia, Canada	Ontario, Canada
Estimated 2007 Population ('000)	6,927	5,246	1,575	4,131	60,975	4,480	13,119
Integrated Statewide service	no	yes	yes	yes	no	yes	no
Centralised/Decentralised Service	decentralised	decentralised	centralised	decentralised	decentralised	partially decentralised	decentralised
Model of Service	3 metropolitan FCS where the medical staff provide outreach to 15 regional centres (and the ACT) where genetic counsellors are based; 3 smaller metro FCS that do not provide outreach	4 metropolitan FCS which provide outreach to a quadrant of the State. It is not known whether the genetic counsellors are based in the regional areas or whether they go out with the medical staff	1 metropolitan FCS that provides outreach to a number of regional centres. Both the genetic counsellors and the doctors go out from the metro area.	1 metropolitan FCS that provides outreach to 8 regional locations. Genetic counsellors are based in 6 of the 8 locations.	?	1 metropolitan service based at the Cancer Centre in Vancouver. Medical and counselling staff provide outreach to regional centres. Recently genetic counsellors have been based in 2 regional Cancer Centres.	13 regional FCS and 9 Northern or satellite clinics. A Northern or satellite clinic has a limited complement of staff on-site, chiefly genetic nurses/ counsellors , augmented by a formal relationship with a clinical geneticist in a regional centre.
Services Offered:							
Counselling	✓	✓	✓	✓	✓	✓	✓
Genetic Testing	✓	✓	✓	✓	✓	✓	✓
Medical	✓	✓	✓	✓	✓	✓	✓

Characteristic	NSW	Vic	SA	Qld	UK	British Columbia, Canada	Ontario, Canada
Consistent protocols for eligibility, referral, assessment, testing	X	X	✓	Unknown	Some FCS would have consistent protocols	✓	There are standard criteria for counselling, testing and clinical management but they are not used consistently.
Clinical governance	FCS report to one of the following Cancer, General Genetics, Genetics Community Health, Pathology Staff are employees of the AHS where services are located.	General Genetics	Pathology	General Genetics All Staff (including those based in regional towns are employees of Genetics Health Queensland.	?	Cancer	Cancer and General Genetics.
Follow up management	Some high risk clinics provided by some FCS.	Some high risk clinics are provided by the FCS; the Familial Cancer Register maintained by the Cancer Council Victoria sends out reminders for	FCS does not provide surveillance reminders to patients; it does provide an annual mailout of latest advice to patient.	Unknown	Varies by service	No ongoing screening or management by the Familial Cancer Program. The BC Cancer Agency provides some	?

Characteristic	NSW	Vic	SA	Qld	UK	British Columbia, Canada	Ontario, Canada
		screening and testing				high risk screening for BRCA1 and 2 mutations. The GIP is responsible for coordinating other screening.	
Nature of outreach services	There are 12 genetic counsellors based in regional and rural areas and medical specialists visit or provide consultations via telehealth	Genetic counsellors and medical staff from the 4 hospitals where FCS are based, visit rural areas	Genetic counsellors and medical staff from the centralised FCS based at the Women's and Children's Hospital in Adelaide, visit rural areas.	There are 6 genetic counsellors based in regional towns and medical staff based at GHQ in Brisbane visit 8 regional locations on a regular basis.	Varies by FCS	There are genetic counsellors based in 2 regional locations. Most outreach services are provided by the genetic counsellor and the clinical geneticist based at the Vancouver Cancer Centre who visit regional areas.	Varies by FCS
Adequacy of administration support	Not adequate	Adequate	Adequate	All counsellors Based in regional towns have admin support 1 day/week	?	Adequate 1.0 genetic counsellor to 0.5 clerk	Varies by FCS

Characteristic	NSW	Vic	SA	Qld	UK	British Columbia, Canada	Ontario, Canada
Standardised S/W database	X A statewide standardised database (KinTrack) is currently being rolled out	X	✓ KinTrack	✓ KinTrack	?	X	X
Laboratories • No. of laboratories • Urgent tests • Mutation Screens • Predictive tests	3 public, 1 Private 2-3 weeks 3-8 months 4-6 weeks	3 public 2-3 weeks 3-4 months 4 weeks	3 public ? 8 weeks 2 weeks	1 public ? ? ?	? Genetics White 3 days 8 weeks 2 weeks	1 public 8-12 weeks 12 months ?	13 public ? 4 months – 2 years ?

5. Issues affecting future service delivery

5.1 Access to services

Access to FCS is highly variable in terms of geographical availability (see staff of FCS/population ratio in Table 6), the waiting times for clinical appointments and laboratory tests and access by specific population such as Aboriginal peoples and people from a culturally and linguistically diverse background.

Table 6 shows that the best access in terms of FCS staff per head of population is in AHS 1 (1:134,000) followed by AHS 2 (1:232,000) and AHS 3 (1:238,000). AHS 4 and AHS 5 have substantially different staff/population ratios with 1:866,000 and 1:850,000 respectively. In AHS 4 there is a large non English speaking population as well as lower socio-economic status population.

Although access appears best in AHS 1, only high risk patients are seen. Despite the recommendation that all people at high or moderate risk should be seen³ this is not occurring even in the most resourced service.

Waiting times for clinical appointments and testing also vary between services. The waiting time for an appointment is 8 months in the Hospital 25, 2 months in AHS 4 and 2 weeks in Hospital 6. At Hospital 8 there was no genetic counsellor for some time and the waiting list grew so long that at the beginning of 2006, when the current counsellor took up the position it took 3 years to get through the backlog.

There is also a variable waiting time for genetic testing results – from a few weeks to 8- 12 months. There are differing views about the importance of this variability with some people saying it does not matter and others who say it does because it affects the screening regimen of the proband and has implications for testing of the relatives who are potentially at risk. In the UK as a result of the 2003 report⁸ there were funding enhancements of the laboratories to reduce the waiting time for testing.

Only about half of the expected number of referrals to the familial cancer services is made. In Section 4.3 it was estimated that 5400 patients should have been seen by the familial cancer services in NSW in 2007, 6000 in 2011 and 6800 in 2016. In section 5.5.1 it was estimated that 3500 patients were seen in 2007. The difference between the number seen and the number expected to be seen varies by AHS with AHS 4 and AHS 5 showing the greatest discrepancy.

Although data are not available, anecdotal evidence is the Aboriginal population and people from culturally and linguistically backgrounds are not well represented in the utilisation of the familial cancer services.

5.2 Model of Service

There is no common model of service across all of the FCS in NSW. Variation occurs in a number of areas:

- There are no standard protocols across all FCS for assessment, testing and referral of patients;
- Some FCS provide follow up clinics for breast and ovarian cancer whereas others provide no follow up within the FCS but there may be follow up clinics outside the FCS eg in oncology or gynaecological oncology or colo-rectal surgery;
- Staff of some FCS attend relevant cancer multidisciplinary team meetings but others do not;
- There is no formalised relationship between the molecular laboratories who provide the germline testing for the FCS;
- The provision of outreach from the FCS – ie whose responsibility it is and the appropriate mode (doctor visits or telehealth) – is ad hoc; and
- The role of the generic counsellor vis a vis the cancer geneticist varies by FCS.

Protocols and guidelines

There are no common protocols used by all FCS and different guidelines are in use to assess risk and decide whether testing should occur. Some of this variation is being addressed by the development of guideline and protocol development in eviQ at the Cancer Institute NSW. There are also different requirements in relation to referrals with some FCS insistent on a doctor's referral while others are not.

Follow up management

There are differences between the FCS in terms of the extent to which they provide ongoing risk management of patients. For example, at Hospital 4 there is a high risk management clinic as part of the familial cancer service for people with mutation identified breast genes. At Hospital 6 there is a review process but no high risk clinic. There is a gynaecological oncology risk clinic held monthly at the Familial Cancer Service at Hospital 6. At Hospital 21 there is a high risk clinic for people with BRCA mutations which is held outside the familial cancer service.

In SA the Familial Cancer Service does provide an annual mailout of the latest advice to the patient and a letter to the referring doctor with a request for them to report back the outcome of surveillance of the patient. It does not provide any surveillance itself. In the UK, at least at the Teeside service, the familial Cancer service provides testing and surveillance.

There are differing views on how ongoing management should be provided:

- A familial cancer registry (not just for bowel cancer) which would send out relevant reminders to patients regarding their surveillance;
- The familial cancer services would not provide the high risk clinic but they would be aware of the relevant ones for their patients and refer them. The familial cancer service would provide case management; and
- Another suggestion was that the familial cancer services would not necessarily provide high risk management but they should provide standardised guidelines for how they should be provided.

Multidisciplinary Team Meetings:

The involvement of familial cancer service staff in multidisciplinary team meetings is recognised as important by all services but the extent to which this is possible depends on the availability of staff to attend.

Relationship of FCS to molecular laboratories

The provision of laboratory services is described in detail in section 3.2.9. Despite the clear relationship between the laboratories and the FCS, there is no formal contact between the FCS and the laboratories that do the testing.

Outreach Provision

The provision of outreach is described in detail in section 3.2.2. It seems to be ad hoc in nature with the FCS at the Hospital 6 providing most of the outreach services in NSW. The HMO at the FCS in Hospital 6 provides outreach to the regional areas of AHS 3, the Area Health Service 6 (Hospital 8, Hospital 9, Hospital 10, Hospital 11), the Area Health Service 7 (Hospital 18). At some places – Hospital 20, Hospital 12, Hospital 15 and Hospital 16 – outreach is provided by telehealth. At other locations, the doctor visits. Outreach is not provided if the genetic counsellor position is not filled. As there is no genetic counsellor in Hospital 16 at present, no outreach services are provided there.

By comparison, the FCS at Hospital 1 provides outreach to Hospital 2, Hospital 3 and Area 1. Medical services to the Hospital 25 are via telehealth and they used to be via telehealth to Hospital 3 too but in 2010 the CMO will be visiting Hospital 3. The FCS at Hospital 4 provides outreach services to Hospital 24, Hospital 17 (not provided at the present time) and Hospital 19 (via telehealth).

There are a number of inconsistencies in the provision of outreach FCS:

- In most rural outreach the genetic counsellor is based in community health and provides both general and cancer genetics counselling; however at Hospital 2 and Hospital 3 the genetic counsellor only does genetic counselling and is part of cancer services;
- Some outreach is provided by telehealth and some by doctor visits. Telehealth consultations do not attract a Medicare item number so this may influence the extent to which it is used. The Hospital 1 FCS has shown that telehealth can be used very successfully in this environment where the patient does not need to be examined by the doctor. The Hospital 1 FCS has done over 1000 telehealth consultations with no report adverse events. A study in Canada⁹ showed that telemedicine is useful alternative by which to provide cancer genetics services when geographical distance is an issue;
- Cancer genetic counsellors are not available to all major cancer centres in NSW; and
- The HMO at the FCS in Hospital 6 is only available on a 0.5 FTE basis to provide the FCS for Hospital 6 and all the outreach compared to 1.6 FTE staff specialists at the FCS at Hospital 1 and 1.6 FTE staff specialists at the FCS at Hospital 4 who, combined, provide less outreach than does the FCS at Hospital 6.

5.3 Role of genetic counsellors

There is variation between FCS in the role of the genetic counsellor. This variation is reflected in the very different doctors/counsellor ratios shown in Table 7 and the variation that exists in the views of doctors and counsellors about what they consider to be optimal doctor/counsellor ratios. These views vary from 1:2 to 1:10.

The genetic counsellors indicated that there was variation in the role of genetic counsellors that was not related to their level of experience and that were differences between services in what they could do. For example there are differing views on whether counsellors can order predictive tests and whether they can/should give screening advice. There are differing views on how results should be given – on the phone or in person – and who should give the results – the doctor or the genetic counsellor.

The role of the doctor vis a vis the genetic counsellor may be partially driven by the need for the doctor to see the patients and to bill Medicare. Both Hospital 1 and Hospital 4 FCSs are dependent to a certain extent on revenue from Medicare. However this is not relevant in Hospital 6 where the doctor providing the medical services is an HMO and bills Medicare privately.

In an attempt to standardise the role and to illustrate the career pathway of the genetic counsellor, two of the current genetic counsellors developed guidelines and competencies for the genetic counsellor's role in familial cancer. The purpose of these guidelines was to indicate what can be expected from a counsellor with each year of experience. The guidelines have not really been used to date despite the general agreement among doctors and counsellors that they are a good idea.

There was a problem with the pay of genetic counsellors given the view that they were recompensed adequately for their training which consists of an undergraduate degree and a post graduate counselling course (soon to become a Master's degree). However that issue has been resolved to a certain extent with the inclusion of genetic counsellors in the Allied Health Award. However there are still some discrepancies in the pay of genetic counsellors depending upon whether they are nurses and on the Nursing Award or

counsellors on the Allied Health Award. The idea of creating the equivalent of a genetic counsellors practitioner, similar to a nurse practitioner, was also raised as a way of genetic counsellors achieving autonomy once they have reached the required level of competency.

5.4 Lack of administration support

Lack of administration support is an issue for most of the services – the exceptions are Hospital 6 and Hospital 1. Administration support is needed for assistance in making patient bookings, for patient reception, to type letters, for photocopying, filing, faxing, to track down pathology reports and to assist with data entry. One of the outreach genetic counsellors recorded time useage over the period of a month and she found that 40% of her time was spent on administration tasks. None of the outreach services has any administration support (although the Hospital 6 service does provide them with some) and this affect s their avaiability to attend multidisciplinary team meetings, to provide community health centres, sometimes available to the outreach genetic counsellors, is not always appropriate because of the confidential nature of their work. They need dedicated administration support.

In Queensland, the Genetics service is a statewide service so the outreach counsellors, although based in regional centres, are employed by the Statewide service. They are provided with administration support for one day per week. In BC, Canada, the Familial Cancer Program has a formula which advocates that there be 0.05 FTE administration staff for every 1.0 FTE genetic counsellor position.

5.5 Governance

The governance of FCS varies between Area Health Services. Each area is clinically and operationally responsible for the services within their boundaries but within the Areas they report in different ways (see section 3.2.4). Both the FCS at Hospital 1 and Hospital 4 are within Cancer services in their Areas, the Hospital 6 FCS is part of the a standalone Genetics Unit which has been recently incorporated into Pathology North, the AHS 4 service is part of the general genetics service which in turn is part of Chronic and Complex Care, the FCS at Hospital 7 has also been incorporated into Pathology North and most of the outreach counsellors are in Community Health.

Responsibility for the statewide planning and coordination of FCS resides with two bodies – GSAC and the Cancer Institute NSW, so direction is unclear.

There are no formal agreements between the AHS with the FCS which provide the outreach services and the AHS who receive the outreach services. As a result there are problems of isolation for genetic counsellors in rural areas because they are sole practitioners who are employed by the AHS where they are located but who are clinically responsible to the service providing the outreach service – which in their case is both general and cancer genetics counselling. These services have no formal responsibility for the outreach counsellors in terms of mentoring, clinical supervision or continuing education. This lack of supervision, apart from the obvious shortfall, has implications as it affects the ability of the genetic counsellor to complete Part II of their counselling course. In 2009 the genetic counsellors from Hospital 9, Hospital 8, Hospital 11, Hospital 10, Hospital 20, Hospital 12 had to organise their own education day in Hospital 6. These genetic counsellors have also independently initiated monthly peer support sessions via videoconferencing.

The consensus view is that the outreach genetic counsellors need good telecommunications with the doctors providing the outreach services; mentoring and supervision; and they need to go to the hub service for at least

one week per year to update skills and receive education. Managers may not be aware of the genetic counselors requirements - their need for clinical supervision and continuing education. Moreover it is often difficult obtaining funding to attend training and conferences.

5.6 Workforce

A number of significant issues that will affect the future sustainability of the FCS workforce are as follows:

- Potentially there is a shortage of medical staff for familial cancer services as there has not been a clear training path for cancer genetics. A speciality training path for cancer geneticists has been proposed but not yet approved and after it is approved it will then take 3-5 years for doctors to be available for recruitment. Many of the doctors currently occupying the cancer geneticists' positions are of a similar age so that when they retire there will be a loss of most of the cancer genetics medical workforce.
- Genetic counsellors are about to have their education requirements changed which may have an impact on the number of people undertaking genetic counselling courses for a number of reasons:
 - The masters degree that they are being required to undertake is both longer and more expensive
 - The accreditation requirements have not been finalised and it may not be possible to meet these requirements by working solely in a FCS.
 - The role of the genetic counsellor is not clearly defined and they are not graded accordingly to their competency and experience.
- Genomic medicine¹⁰ is expected to be a significant development in future health care so undergraduate medical and nursing curricula need to reflect this.
- Genetics is predicted to become a significant part of mainstream medicine and its accommodation within mainstream medicine needs planning ie. What category of staff will provide these mainstream services, what services will they provide and how will they relate to specialist FCS.

5.7 Funding of Services

There is no obvious pattern to the funding of FCS and the differential funding of services has inevitably contributed to the variation in the way services are provided. Table 9 shows the estimated cost of staff by service which represents 80-90% of the budget.

The three main FCS at Hospital 1, Hospital 4 and Hospital 6 are all dependent on some temporary funding – 1.8 FTE at Hospital 4 (one third of their funding), 1.0 FTE at Hospital 1 (14% of their funding) and 0.5 FTE at the Hospital 6 (15% of their funding). The genetic counsellor position at Hospital 3 was created entirely with temporary funding. The funding for all these temporary positions is from the Cancer Institute NSW and was due to expire June 2010 but has been extended to June 2011.

The services at Hospital 1 and Hospital 4 are also dependent on revenue from Medicare billing because they are outpatient services. The staff specialists employed by those services can bill Medicare for outpatient consultations and the revenue goes to the hospital and the familial cancer services. The Hospital 6 service does not generate any Medicare revenue for the service itself because the specialist there is not a staff specialist, but a HMO who bills Medicare for the patient consultations he made.

5.8 Data

The previous report indicated the need for a Statewide database and this remains an important issue. Approval has been given to purchase KinTrack, the genetics database used in South Australia, for NSW. Roll out is due to commence shortly. Some of the requirements of KinTrack for the familial cancer services include:

- Statewide data sharing so that it is possible to access details of a patient's familial if they are being seen by another FCS;
- The ability to satisfy the requirements of rural AHS community health databases, where outreach counsellors are part of community health, without having to enter the data twice; and
- Training, support and maintenance of the database so that optimum use can be made of the functionality of the system.

5.9 Role of the Familial Cancer Registry

A review of the Familial Cancer Registry (HCR) has just been completed for the Cancer Institute NSW and is in draft form. It makes a number of recommendations including the implementation of a single database across all familial cancer services and the expansion of the scope of the register to collect all inherited cancers.

These recommendations agree with suggestions made by some of the people interviewed for this project.

5.10 Laboratories

The main laboratory issues are;

- testing turnaround time;
- public versus private laboratories;
- centralisation versus decentralisation of laboratories;
- gene patenting; and
- future developments

5.10.1 Testing turnaround time

As noted in section 3.2.9 there is variation between the laboratories in the time it takes to do the genetic tests. There is a difference of opinion about what is an appropriate waiting time. Some people argue that it is important to know quickly if the cancer is inherited because treatment decisions for breast cancer, in particular, are becoming more dependent on knowing the inherited status of the cancer. Timing affects decisions about the nature of surgery (partial mastectomy or double mastectomy and reconstruction) and clinical trials are currently underway for different chemotherapy drugs. Others believe that the results revealing an inherited cancer need to be mulled over and decisions should be made after initial treatment. Some of the genetic counsellors say that regardless of the result, there is a problem if there is a variation between services in the waiting times for tests. For example, if someone is tested at one service with one waiting time but their relative is tested at another service with a different waiting time, it is difficult to explain why this difference exists.

In 2003 in the UK⁶, the government invested up to £18 million capital on upgrading NHS genetics laboratory facilities in England with the aim of reducing genetic testing times to within two weeks where the potential genetic mutation is already known (predictive tests) and within eight weeks for unknown mutations in large genes.

5.10.2 Public versus private laboratories

Section 3.2.9 identified that there were three public laboratories and one private laboratory where most of the genetic testing for inherited cancer is performed. There is a difference of opinion over the importance of whether the testing should be done in the public sector or the private sector.

The arguments, expressed to the consultant as part of this project, against using the private sector laboratories include the following:

- there is not the same level of interpretation of results provided by private laboratories;
- if a result is inconclusive public laboratories will often do other studies to try and find an answer. However it is not profitable for private laboratories to do this extra work;
- private laboratories may keep samples but they are not likely to do extra testing without further payment;
- private laboratories are unlikely to transfer samples to other laboratories for further testing to explore questions of pathology within families;
- private laboratories are less likely to be interested in research because it is not profitable; and
- if a private laboratory had a monopoly over a particular form of testing, they could charge any price and they would have no incentive to develop new tests.

In South Australia there is no outsourcing of testing to private laboratories even if testing is more expensive in the public laboratory. Similarly in Victoria all genetic testing for inherited cancer is done in public laboratories. The only instance where samples may be sent to a private laboratory is where the patient is not eligible for testing in the public system and they pay for the test to be done. In NSW there is no policy about the use of private molecular laboratories for inherited genetic testing.

5.10.3 Centralisation versus decentralisation of laboratories

There is debate over whether testing should be centralised or decentralised and in close proximity to the clinical services. The main argument in favour of centralisation is that there could be cost efficiencies if all the testing was performed in the one laboratory. As testing becomes more automated these savings could be significant. Some people argue that laboratory robotics are not too far away and that they will also reduce costs.

However there are disadvantages that were expressed to the consultant to the centralisation of laboratories:

- there would not be the close proximity and collaboration of the laboratory staff with the clinical service and the benefits that arise from this proximity and collaboration;
- there would not be the range of expertise that currently exists in a number of different laboratories; and
- centralisation may create the conditions for a monopoly to develop and stifle healthy competition.

The comparison of NSW with other jurisdictions in section 4.1.1 shows that the number of molecular laboratories used by the FCS in NSW is very similar to other jurisdictions when population size is taken into account.

5.10.4 Gene patenting

Currently GTG, the private laboratory in Melbourne, holds the licence in Australia for breast cancer testing which means that in principle it is the only laboratory in Australia which can do BRCA testing. Currently GTG has allowed public laboratories to do BRCA testing, but this arrangement needs to be formalised.

5.10.5 Future developments in laboratories

A couple of future developments that may influence the rate of growth of the molecular laboratories:

- Epigenetics - which refers to changes in appearance or gene expression caused by mechanisms other than changes in the underlying DNA sequence. Epigenetic theory is an emergent theory of development that includes both the genetic origins of behaviour and the direct systematic influence that environmental forces have, over time, on the expression of those genes. It is anticipated that testing for epigenetics is imminent (in the next three years); and
- Somatic profiling of tumours - i.e. genetic testing of tumours - will be the big growth area in the next few years. As the technology for tumour testing is similar to genetic testing for inherited disease, the current molecular laboratories could be set to grow substantially.

5. 11 Future predicted developments

There are a number of predicted future developments that will impact on the way familial cancer services are provided:

- It is known that there is a familial component to prostate, kidney and endocrine cancer but the genetic tests are not yet available. When these tests are discovered (and it is not known when), there will be a greater demand for familial cancer services.
- It is predicted that the next disease for which testing will become available is prostate cancer. (Prostate cancer had the highest incidence of all cancers for men and women in 2007 according to the Cancer Institute NSW report¹¹);
- Genetic counselling and testing is increasingly likely to be offered to women with breast cancer shortly after diagnosis while treatment options are being considered. This is occurring for a number of reasons: technical advances in genetic testing mean that genetic test results can be available within a few weeks (staffing permitting) so the results can assist with decisions about the nature of surgery - breast conserving or mastectomy. In the not too distant future it may also direct the selection of adjuvant chemotherapy (clinical trials are currently underway). Meiser at al. (2008) describe the impact that the linking of the results of genetic counselling and testing to subsequent patient management will have on familial cancer services:
 - an increase in the number of patients and their unaffected relatives referred for genetic counselling and testing;
 - the need for patients to be seen soon after referral in already under resourced services
 - the subsequent increase in the number of mutation-positive families and the need to provide additional psycho-social support to these women and their families will impact even further on services already struggling under personnel and funding constraints
 - current management protocols and established work practices will change in breast oncology units
- These changes are not only occurring for breast cancer patients. If someone has Lynch Syndrome, they receive different chemotherapy than someone who does not. Similarly someone with P53 should not have radiation treatment.
- Treatment focused genetic counselling and testing implies that counselling and testing may become part of mainstream medicine - i.e. within oncology units, and the familial cancer services will be needed more to see the relatives of the person with cancer and for complex cancer syndromes and for difficult and rare cases.
- At the present time a high proportion of people who are tested get an inconclusive result but they are still regarded as high risk and need regular screening. However as the genes of major and minor effect become better known, there will be less inconclusive cases and only the people who are proven to be at high risk will need to be managed on an ongoing basis.
- Increased knowledge about the lower impact penetrance genes and the interaction of genes will mean increased referrals to familial cancer services.
- Over the next five years there will be dramatic changes in testing for non familial genetic errors and those errors will guide treatment and prognosis but the signature of those somatic errors will also be important for

familial cancer. For example, at the present time if a woman has breast cancer and a strong familial history, she will be tested for a BRCA mutation. However if there is no familial history available (perhaps she has been adopted), she will not be tested. However in future the molecular profiling of the breast cancer tumour may show similarities to the somatic profile of a BRCA mutation.

- Testing guidelines may change as testing becomes cheaper and quicker e.g. there may be testing of all women with ovarian cancer except those with borderline/non invasive cancer; women who develop breast cancer under 50 years of age, with a triple negative tumour, or are Ashkenazi Jewish or have a familial history may be offered testing. If testing does become low cost, there may even be a time when all women are offered a BRCA test.
- At some time in the future - probably more than five years away - all young adults (who are over the age of consent) may be able to get a blood test to identify if they have inherited a set of genes that puts them at risk of cancer later in life. Their risk profile will convert into lifestyle advice such as never smoke, protect oneself in the sun, have an MRI or ultrasound rather than an x-ray.

6. Options for Future Service Delivery of Familial Cancer Services

This section looks at options for the future delivery of FCS in terms of factors, namely:

- Service configuration
- Organisation and Governance
- Outreach
- Funding
- Data
- Workforce
- Laboratories

Each section will identify options and indicate the preferred option based on the following key planning principles:

- Accessibility
- Sustainability
- Evidence based practice
- Quality, reliability and timeliness
- Efficiency and effectiveness

The preferred options below incorporate the issues made in Section 5.

6.1 Service configuration

6.1.1 Option 1- Maintain services as is (status quo)

The features of this option are as follows:

- Relatively large familial cancer services (FCS) at Prince of Wales Hospital, Westmead Hospital and the Hunter;
- Smaller FCS at St Vincents, Sydney South West Area Health Service and Royal North Shore Hospital;
- Outreach FCS from each of the main services - by telehealth to the outreach services provided by Prince of Wales Hospital and Westmead Hospital and a mixture of telehealth and visits to the outreach services provided by Hunter; and
- Rural outreach counsellors provide general and cancer genetics counseling.

6.1.2 Option 2: Three hubs based on current services at Prince of Wales Hospital, Westmead Hospital and the Hunter.

- Three main FCS hubs at Prince of Wales Hospital, Westmead Hospital and the Hunter, each with responsibility for the provision of FCS to a designated third of NSW
- The service hub would be responsible for the provision of services within their AHS as well as for outreach services in their designated third of the State.

Outreach genetic counsellors would be based at appropriate regional and rural locations. The 3 segments would be as follows:

- The Prince of Wales Hospital hub service would be responsible for SESIAHS, half the population of SSWAHS and the eastern half of GSAHS including the ACT - the projected population in 2016 of SESIAHS, half the population of AHS 4 and half the population of GSAHS is 2,318,483 (see Table 11). (The ACT population in 2016 is projected to be 377,400.
- The Westmead hub service would be responsible for SWAHS, half the population of SSWAHS, half the population of GSAHS and GWAHS - the projected population in 2016 is 2,622,969.
- The Hospital 6 hub service would be responsible for the population of three AHS - HNE, NSCCAHS and NCAHS.

6.1.3 Option 3 : Four main service hubs for NSW at Prince of Wales Hospital, Westmead Hospital, the Hunter and Sydney South West Area Health Service (SSWAHS).

- Four main hubs, at Prince of Wales Hospital, Westmead Hospital, the Hunter and SSWAHS, each with responsibility for the provision of FCS to a designated quadrant NSW.
- The service hub would be responsible for the provision of services within their AHS as well as outreach services in their designated quadrant of the State. Outreach genetic counsellors would be based at appropriate regional and rural locations. The 4 quadrants would be as follows:
- The Prince of Wales Hospital hub service would be responsible for SESIAHS and the eastern half of GSAHS including the ACT - the projected population in 2016 of SESIAHS and half of GSAHS is 1,538,685.
- The Westmead Hospital hub service would be responsible for SWAHS and GWAHS the projected population in 2016 is 1,587,041.
- The Hunter Hospital hub service would be responsible for three AHS - HNE, NSCCAHS and NCAHS - the projected population in 2016 is 2,662,135.
- The SSWAHS hub service would be responsible for the SSWAHS and western half of GSAHS - the projected population in 2016 is 1,815,726.

The rationale for this preferred option is that each of the metropolitan AHS provides a hub service compared to the previous option where SSWAHS is not a hub service. This option seems appropriate especially as SSWAHS is the largest AHS in the State with a large culturally and linguistically diverse population and a large low socio-economic population whose access to services may be less than average. SSWAHS also has major cancer centres at both RPA and Liverpool Hospitals.

6.2 Organisation and Governance

6.2.1 Option 1 Status quo

The features of the status quo are as follows:

- Overall planning and coordination by GSAC and the Cancer Institute NSW
- AHS responsible for provision of FCS within their AHS
- Different reporting mechanisms in each AHS (see sections 3.2.4 and section 5.5)
- No common model of service (see section 5.2)
- Variation between services in the role of genetic counsellors (see section 5.3)
- Inadequate administration support (see section 5.4)
- No formal agreements between the FCS providing medical outreach and the AHS receiving that medical outreach (see section 5.5)
- Isolated genetic counsellors in rural AHS who have no formal relationship with the FCS providing medical outreach so no formal mentoring, clinical supervision and continuing education (see section 5.5)

6.2.2 Option 2: Statewide coordination of FCS

The most appropriate governance structure to achieve the development of a single organisational structure for familial cancer services across NSW will be decided by the Cancer Institute NSW and NSW Health, and in particular the resolution of the respective roles of NSW Health, GSAC and the Cancer Institute NSW in the planning and coordination of FCS. However Statewide coordination of the FCS could take various forms:

- A Statewide Coordination Committee; and
- A Statewide manager/coordinator and a Statewide Coordination Committee; or
- Managers/coordinators at each of the hubs with an overarching governance structure.

Statewide coordination of the planning and provision of FCS offers the prospect of:

- Designated formal responsibility for the provision of FCS to the population of NSW, including the specific needs of Aboriginal people and people from a culturally and linguistically diverse background.
- A statewide model of service and equitable resourcing ie. staff numbers (doctors, genetic counsellors and administration support) per hub would be based on population numbers and cancer incidence projections, the role of the counsellor in relation to the doctor would be defined, there would be standard guidelines and protocols for the assessment, testing and referral of patients, decisions about appropriate outreach destinations would be determined and the role of telehealth in the provision of outreach would be defined, the nature of ongoing patient management and surveillance by the FCS would be specified, links to molecular laboratories would be seen as part of the provision of FCS so waiting times across laboratories could be monitored centrally.
- The development of similar lines of accountability even if reporting structures within AHSs differ. Hub FCS should be fully accountable to Cancer Services in their own AHS and in the AHS where they provide outreach regardless of their position within the organisation structure. Staff of the FCS should attend relevant cancer multidisciplinary team meetings. Outreach staff who provide general and cancer genetic counselling should stay in Community Health where they are located currently but would be accountable to the hubs for the provision of FCS. Service agreements would be drawn up between the hubs and the relevant AHS to formalise these arrangements.
- Hub FCS to take clinical responsibility for outreach staff even if they remained administratively responsible to the AHS where they are located. This means that the hub would provide mentoring, clinical supervision, continuing education, guidelines and protocols for service provision. Hub staff would be cross credentialed in their own AHS and in the AHS where they provide outreach.
- Pathways for referral to the FCS to be developed by the hub services for every cancer centre in their quadrant of the State with a genetic counsellor attached to all the major cancer centres.
- The nature of the relationship of the FCS to general Genetics to be defined and each FCS would nominate a clinical geneticist who would be available for consultation by staff of the FCS in each quadrant.

The rationale for Statewide coordination is that it would enable:

- The equitable allocation of budgets to the hub services based on an agreed service model;
- The fostering of common protocols and practice standards;
- Agreement to be reached on and promotion of the use of standardised genetic counsellor competency levels;
- Facilitation of the use of a common database;
- Facilitation of the training of cancer geneticists, genetic counselors;
- Planning of professional development of FCS staff;
- Facilitation of Statewide research;
- Liaison with the Genetics Education Service to prepare education materials;
- The monitoring of service quality;
- Decisions to be made about new developments/issues such as. if testing becomes cheaper and is done by the oncologists; how should the FCS support the oncologist; whether genetic counsellors should be employed by the oncology unit; whether there is a role for a new staff category such as the GRAPS in the UK. As the public becomes more aware of genetics, work with the Centre for Genetics Education to determine how best to handle this increased demand for information; and
- Advocacy and management of changes e.g. the inclusion of genetic testing and counselling as Medicare rebateable items, the changing role of counselors.

6.3 Outreach

6.3.1 Option 1 Centralised

Centralised outreach means that the doctors and genetic counsellors are based at the hub FCS and for them both to visit regional and rural locations to provide outreach services. This is the model for outreach services used in SA where the population for the state is only 1.5 million people.

There are advantages to this approach - all the staff are based together at the hub service so there are not the problems of isolation of sole genetic counsellors discussed in section 5.5 above. However from a patient's point of view this model is disadvantageous. It is more preferable for the counsellor to be based locally. They can see the patient before the medical consultation, do an initial assessment, and obtain the necessary paperwork. The basing of the genetic counsellor in the rural location also means that telehealth consultations with the medical specialist are possible because the genetic counsellor is present with the patient when the medical consultation occurs via videoconferencing. The counsellor is available to talk with the patient before and after the medical consultation.

6.3.2 Option 2: Decentralised

Under this option the genetic counsellors are based in the regional locations and the medical specialists provide consultations via telehealth or visits. Currently this is the way medical outreach for FCS is provided in NSW. It may be the best model for patients because it provides better access than if counsellors visited and it is the most appropriate model for the provision of telehealth. The main disadvantage with the current approach is the isolation of the genetic counsellor in the regional location (see section 5.5).

However if there was statewide coordination of FCS and the genetic counsellors were clinically responsible to the hub service isolation issues may be resolved. There needs to be service agreements between the hub FCS and the AHS receiving the medical outreach to formalise the arrangement. In Queensland the genetic counsellors, although based in regional locations, are employees of Genetics Health Queensland. All the outreach counsellors in Queensland participate in weekly staff meetings via teleconference.

The rationale for decentralised outreach is better access to FCS for people who live in regional and remote areas.

6.4 Funding

6.4.1 Option 1: Status quo

Current clinical costs is estimated to be in the order of \$3m (section 3.2.6) and a substantial proportion of this funding on a temporary basis. The number of patients seen per annum is 3500 (section 3.2.7) which is only 65% of patients that should have been seen in 2007 from the estimates (see Table 14) and slightly over half the number of new referrals who should be seen in 2016 (Table 14 estimates that nearly 6800 patients should be referred to FCS in 2016).

If the funding for FCS remains the same in real terms, there will be a substantial shortfall in the number of patients seen by FCS. Moreover the number not seen will be cumulative over time, so if the funding remains the same, less than half the number of new patients who should be seen in 2016, will be seen.

6.4.2 Option 2: Substantially enhanced funding

In order to meet estimated future demand, funding for FCS needs to be substantially enhanced. The cost of enhancing the services to meet the estimated service demand is difficult to quantify because it depends on the model of service and staff employed but based on discussions with key stakeholders during this project it is estimated that each hub should have 2-3 doctors and genetic counsellor numbers should be increased by at least 10 FTE. Appropriate administrative support is also needed. On this basis it is estimated that the current staffing budget should be doubled from \$2.4m (Table 9) to nearly \$5m. More detailed costing of services is needed to enable a more accurate estimation of the additional funding needed.

Statewide coordination is essential so that resourcing is based on a standard model of service and takes the current inequitable distribution of resources into account. The FCS in both SSWAHS and NSCCAHS have relatively less resources than do the other metropolitan AHS. The development of these services should be seen as a priority. Furthermore the funding of FCS should be fully recurrent and there should be no temporary funding of FCS.

The rationale for enhanced budgets for FCS is based on the estimated number of new referrals to FCS - nearly double the current number by 2016.

6.5 Data

6.5.1 Option 1: Individual FCS databases

At the present time the metropolitan FCS all use a database called Genesis. This database is unsupported and each service has modified it to meet their own needs, so there are no standard data definitions. The outreach counsellors do not have Genesis but some of them use a statewide genetics database, known as CRS. Others record patient numbers and occasions of service in CHIME, the community health database.

Most importantly, the individual databases for each FCS are not networked so it is not possible to know whether some members of a family are being seen by another FCS, which is essential to efficient and effective service delivery.

6.5.2 Option 2: Preferred option: Statewide database

NSW Health has approved the development of a Statewide database - KinTrack - and roll out is due to commence shortly. Some of the requirements of KinTrack for the familial cancer services include: statewide data sharing so that it is possible to access details of a patient's family if they are being seen by another FCS; the ability to satisfy the requirements of rural AHS community health databases, where outreach counsellors are part of community health, without having to enter the data twice training, support and maintenance of the database so that optimum use can be made of the functionality of the system. The rationale for a statewide database is the ability of all FCS to share information about a patient and their familial so that data collection is not duplicated and so that the FCS are as efficient and effective as possible.

6.6 Workforce

Genetic counsellors are the key to the future provision of FCS for at least two reasons:

- The predicted shortfall of cancer geneticists because of the lack of a formal training path and the fact that many of the current cancer geneticists will be retiring at around the same time
- The estimated increase in demand for FCS due to:
 - unmet existing demand for services;
 - the discovery of new genes that are responsible for an familial component to cancer;
 - the need for an inherited cancer diagnosis to be more acute because cancer treatment will depend on this knowledge; and
 - the inevitable mainstreaming of some inherited cancer services.

There are two options:

6.6.1 Option 1: Status quo

If the status quo is maintained there will be a future shortage of both cancer geneticists and genetic counsellors.

6.6.2 Option 2: Take significant action to ensure a sustainable workforce

Significant action is needed in the following areas:

- The training path for cancer geneticists needs to be finalised so that potential new recruits can begin training as soon as possible;
- The new requirements for genetic counsellor education and accreditation need to be addressed to ensure that they will not impact adversely on the cancer genetic counsellor workforce;
- Competencies for cancer genetic counsellors need to be introduced;
- The new genetic counsellor award should be reviewed in the light of changes to the education requirements for genetic counsellors and in relation to the scale of competencies developed;
- Undergraduate medical and nursing curricula should reflect the significant impact that Genomic medicine will have on future health care delivery; and
- Ways of incorporating inherited cancer genetics services into mainstream medicine need to be identified in order to meet future demand for FCS and to cope with the more acute nature of the service.

The rationale for action in relation to the workforce is that FCSs are operating in an unsustainable way.

A separate project, Building an Appropriate Workforce, is planned which will focus on workforce issues.

All services would need to be enhanced to cope with the projected demand identified in section 4 (only about half of the estimated optimal number of patients are referred to FCS - compare projected demand in section 3 with utilisation in section 5 - and some services are clearly under resourced e.g. Sydney South West Area Health Service, RNS and many of the outreach services).

6.7 Laboratory services

The discussion of issues in section 5.10 indicates that there are many unknown factors facing the future of molecular laboratories. Consequently it is difficult to provide prescriptive advice about how laboratories doing testing for familial cancer should be organised without viewing molecular laboratories in the broader context of pathology services generally. It is also premature because the Australian government is only just developing a national framework for genetic testing.

However a number of known factors need to be considered in decisions about the future of molecular laboratories:

- Genetic testing is a relatively new area of testing and somatic testing is predicted to be a significant growth area in the next few years. As the technology for germline and somatic testing is similar, the future of the laboratories that currently provide germline testing is linked to their interest/willingness in providing somatic testing. It also means that links have to be formed with anatomical pathology departments.
- This project has shown that there will be an increase in the demand for inherited cancer testing in the future.
- The public laboratories around Australia have valuable expertise in the area of familial cancer testing and that expertise should be nurtured and consulted in any decisions about the future of molecular laboratories.
- Even though the activities of the current laboratories may appear ad hoc, there are very real reasons why they have developed in the way they have. Area Health Service 3 started to do bowel cancer testing because it did not exist elsewhere in NSW. Breast cancer testing constituted the bulk of the testing and laboratories close to the three main clinical services developed breast cancer testing for their local service. This arrangement enabled close collaboration between the scientists and the clinicians especially for the interpretation of testing results.

At Hospital 4 the breast cancer testing laboratory was developed within the FCS because breast cancer testing was still evolving from the research phase, it was expensive and with a slow turnaround time. It was not suited to being within the main pathology service at Hospital 4 because it was not a routine test that is highly automated and able to generate large revenues. Hospital 4 laboratory has led the change in testing for large genomic rearrangements, to the enormous benefit of patients and their clinical care and the also to the large pathology service providers who then became adopters of the new testing protocols based on the Hospital 4 (and the emerging international) evidence base. The Hospital 4 laboratory has also played a collaborative role in the interpretation of variants of uncertain significance with major research groups, to the benefit of other testing laboratories. Because it is outside the main pathology department the laboratory that is part of the FCS at Hospital 4 has been able to extend testing beyond the routine sequence analysis that is generally the primary activity of a routine laboratory within a large pathology service.

The Hospital 6 laboratory is also a research laboratory and has become a major bowel cancer testing laboratory not just within NSW but within Australia and internationally. Because of its research base it is also able to do additional testing and is not focused on generating revenue from routine testing. Area Health Service 1 laboratory service molecular laboratory, although part of a larger pathology department, has developed expertise in testing some of the rare inherited disorders.

The main issue is the variation that exists between laboratories in their turnaround time but this variation also occurs within laboratories. Each laboratory has experienced a period when their turnaround time has been a year or more. Turnaround time is set to become an issue again if treatment becomes dependent upon testing. It will have an impact on the staff needed in the laboratories.

GTG is a private laboratory with a license to do all the breast cancer testing in Australia. Formal arrangements between FCSs and GTG will need to be made in order to ensure appropriate and timely access to breast cancer genetic testing.

A number of options were proposed in the consultations undertaken as part of this project:

1. Retain the status quo
2. Centralise all breast and bowel testing to one laboratory for the State
3. Estimate the workload of the all FCS in NSW and tender for the testing to be done in any private or public laboratory in Australia.

There are advantages and disadvantages to each of these options.

6.7.1 Option 1: Status quo

Advantages

- Close collaboration between scientists and clinicians in the interpreting of test results
- Expertise residing in a number of laboratories.
- The number of scientists and the range of expertise and experience mean that there are more people available to consider and interpret difficult test results.
- Healthy competition between laboratories so no threat of a monopoly occurring that could artificially inflate the price of testing

Disadvantages

- Ad hoc development of services without strategic decisions about who does testing and where testing should be done
- The Hospital 6 service is small so appears vulnerable moreover being part of a larger pathology service does not make it less vulnerable

6.7.2 Option 2: Centralise all testing into one public laboratory

Advantages

- Appears more efficient and streamlined but there is no evidence that this is the case as there are no cost economies derived from doing more tests
- The expertise of the centralized service may be enhanced but there is not the cross fertilisation that exists from having other laboratories involved.

Disadvantages

- Risk of monopoly developing
- Reduced expertise available
- Transport costs

6.7.3 Option 3: Tender for all inherited cancer testing to be done by one public or private laboratory

Advantages

- Standard turnaround times for tests
- Standard price for all tests

Disadvantages

- Level of interpretation of all test results cannot be specified because it may not be possible to anticipate the number and/or nature of the variants
- Extra testing may not be available within the agreed price
- Loss of public sector expertise in a growth area
- Potential for a monopoly to develop as expertise is lost and fewer laboratories tender

The conclusion of this discussion is that there is not enough information to make definitive decisions. The preferred option, from the author of this report's point of view, is that the status quo should be maintained until a wider investigation specifically into laboratory services is undertaken.

It is proposed that a statewide coordination committee be established to take stock of the current laboratory situation, to make decisions about the future direction of laboratory services and to liaise with the statewide coordination of the clinical services.

The rationale for maintaining the status quo is that there is not enough information to make an informed decision about the future of the laboratory services because the laboratories need to be viewed in the broader context of pathology services generally. However, as the relationship between the clinical services and the laboratory services are vital, a statewide molecular laboratory coordination committee is needed to develop and maintain this link with the clinical services.

7. Conclusion

There is significant variation between FCS across NSW. This variation occurs in the following key areas:

- Governance and reporting arrangements;
- Waiting times;
- Referral pathways;
- Access to services;
- Service models;
- Staffing levels;
- Staffing mix;
- Roles and responsibilities;
- Outreach arrangements;
- Protocols and guidelines;
- Funding arrangements;
- Administration support;
- Data collection and reporting; and
- Laboratory services and waiting times.

The options provided in this paper are summaries in the table 16 below.

Table 16: Summary of Options

Issue	Option 1	Option 2	Option 3
Service Configuration	Status Quo	Three Hubs	Four Hubs
Governance	Status Quo	Statewide Coordination	-
Outreach	Centralised	Decentralised	-
Funding	Status Quo	Enhance Funding	-
Data	Individual Databases	Statewide Database	-
Workforce	Status Quo	Sustainable Workforce	-
Laboratory Services	Status Quo	Centralised Public Testing	Statewide Tender (public &/or private)

Given this variation and the importance of the FCS in preventing cancer incidence, it is recommended that:

1. CINSW facilitate a statewide planning exercise to develop a service model for cancer genetics services in NSW;
2. CINSW, in partnership with key stakeholders, produce a strategic plan for cancer genetics services in NSW for the next 5 years, 2011-2016; and,
3. CINSW and NSW Health identify respective roles and responsibilities in regards to familial cancer genetics services.

APPENDIX E

1) Discussion points for Cancer Genetics Service Directors:

Area of discussion	Specific detail required
1. Current services provided	Nature of services provided (including outreach services) Referral pathways Staff numbers and category Funding amount and source Relationship to other FCS Relationship to Cancer services and involvement in multidisciplinary teams Role of GPs Role of telehealth Data system
2. Clinical workload and projections	What data is readily available? Is it feasible to obtain data on utilisation - no of patients, type cancer, AHS of residence, referral source, number of patients with a positive result of no of patients with a negative result (to understand the extent of positive vs other patients), no. of relatives per patient Method for projecting future workload
3. Workforce issues	Medical training and qualifications Counsellor training and qualifications Counsellor career structure
4. Governance	Relationship of FCS to Cancer services Relationship of FCS to general Genetics services Where do children fit?
5. Issues affecting current service delivery (as defined in previous Review) - Do these issues still exist? Are there any additional issues?	Ad hoc development of services Inadequate staffing Expanding need for services Governance - should Cancer Genetics be separate from general Genetics? Variable geographical patient access Role of Familial Cancer Registry Different databases and privacy issues present difficulties for information sharing Isolation and lack of peer support for senior clinicians Training of medical staff Variation of roles and responsibilities of counsellors and lack of career structure Waiting/turn around times for lab results
6. Lab test projections	Method
7. Literature	Published articles and government reports for info on: Policies/plans for cancer genetics - State and national Best practice guidelines - evi-Q? Models of service in other jurisdictions - other States and internationally Access for rural and remote communities Developments in cancer genetics knowledge Role of GPs in cancer genetics Projection
8. Other people who need to be consulted both within the service and outside	

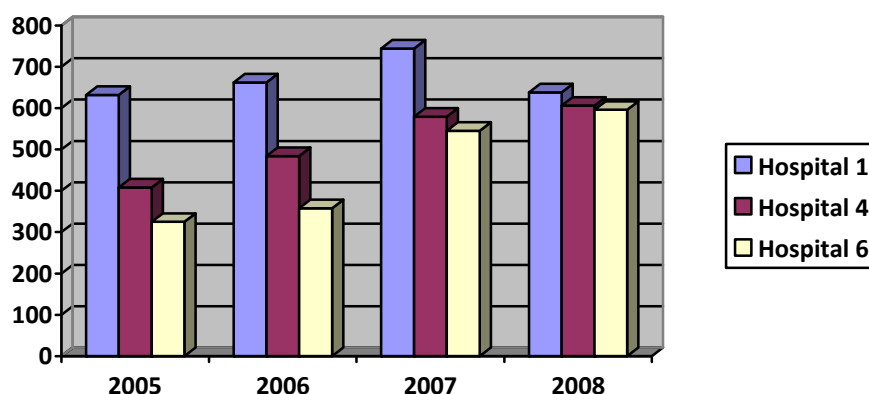
APPENDIX L

20.7 Utilisation of Services

20.1.1 Number of patients

Figure 1 shows the number of new referrals to the three largest cancer genetics services over the past four years. The Hospital 4 and Hospital 6 services have grown over this period to be comparable in size to the Hospital 1 service. In 2008 the three largest services saw around 600 families each.

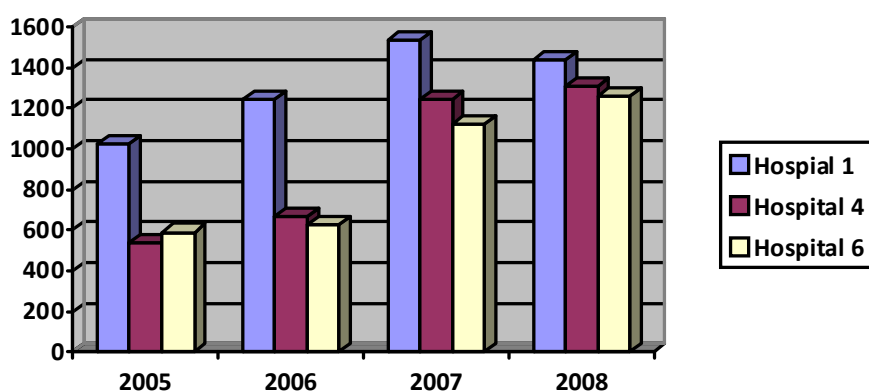
Figure 1 Number of new referrals to the three main cancer genetics services for the years 2005, 2006, 2007 and 2008



Source: Genesis database at Familial Cancer Clinic at Hospital 1, Familial Cancer Service at Hospital 4, Familial Cancer Service, Hospital 6 Genetics

Figure 2 shows the total number of patients (new and returning) seen by the three largest services over the past four years. Returning patients are those people who have already been seen by the service and are attending follow up appointments.

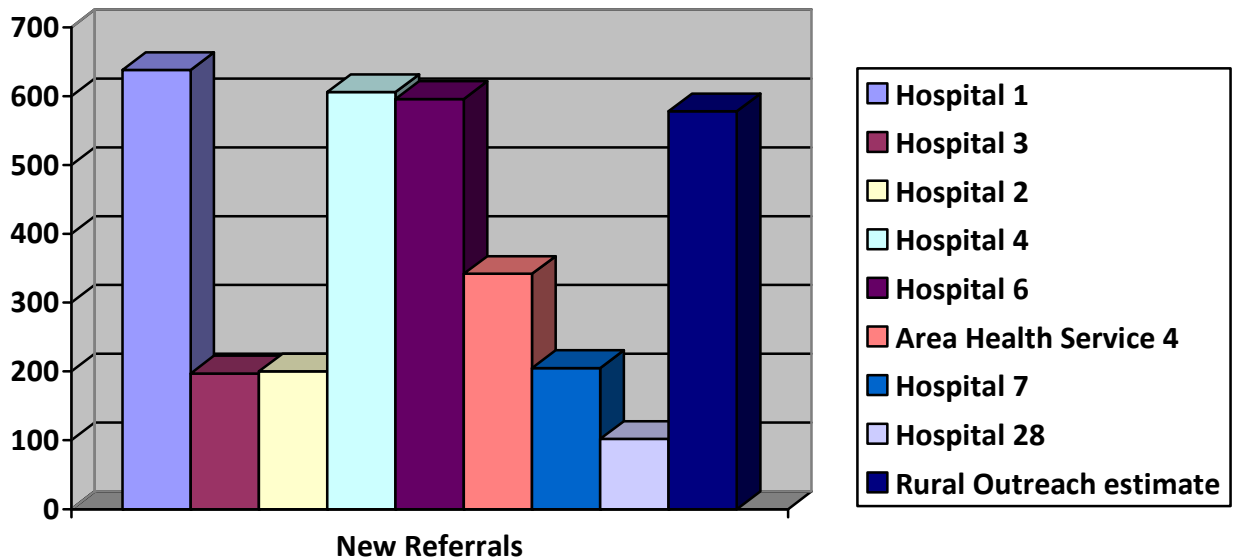
Figure 2 Total number of patients (new and returning) seen by the three main cancer genetics services for the years 2005, 2006, 2007 and 2008



Source: Genesis database at Familial Cancer Clinic at Hospital 1, Familial Cancer Service at Hospital 4, Familial Cancer Service, Hospital 6 Genetics

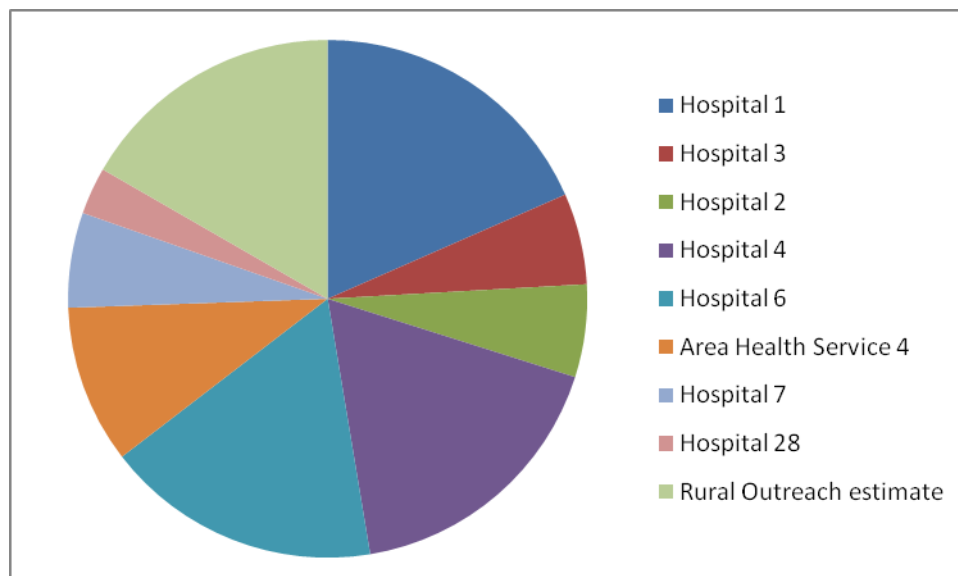
Figure 3 shows the number of new referrals to all the familial cancer services in NSW. There is some double counting in these figures as patients seen by counsellors in outreach centres may have also been counted as a new referral to the main centre if seen by the doctor in outreach. These numbers are expressed as a percentage in Figure 4 illustrating each service's share of the total State's workload.

Figure 3 Number of new referrals to cancer genetics services, 2008



Source: Databases at services or estimates based on information provided by services

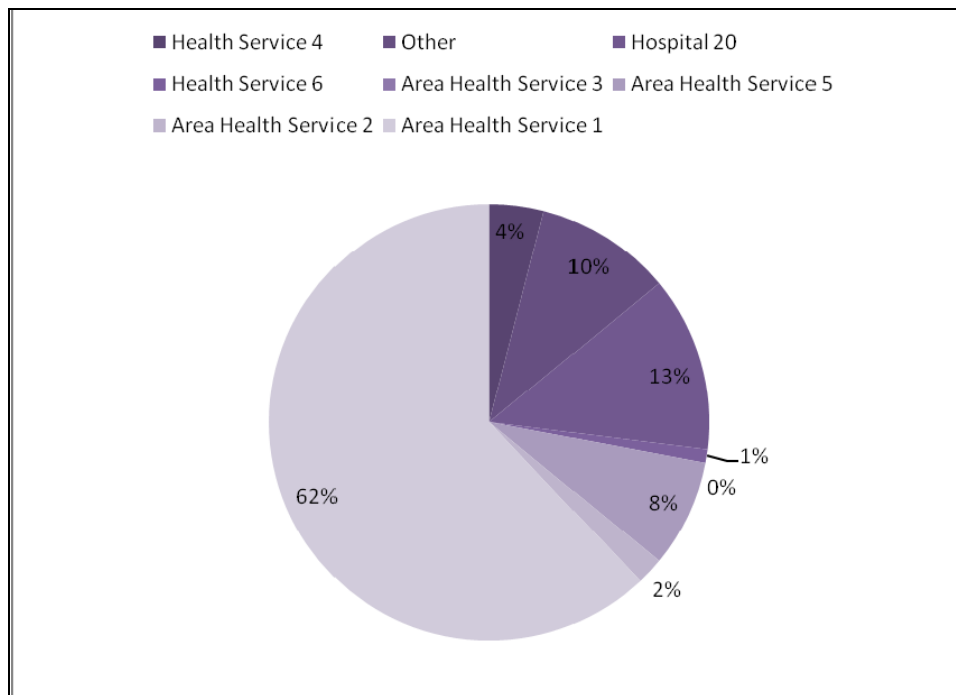
Figure 4 Proportion of new referrals seen by NSW cancer genetics services, 2008



20.1.2 Area of residence of patients

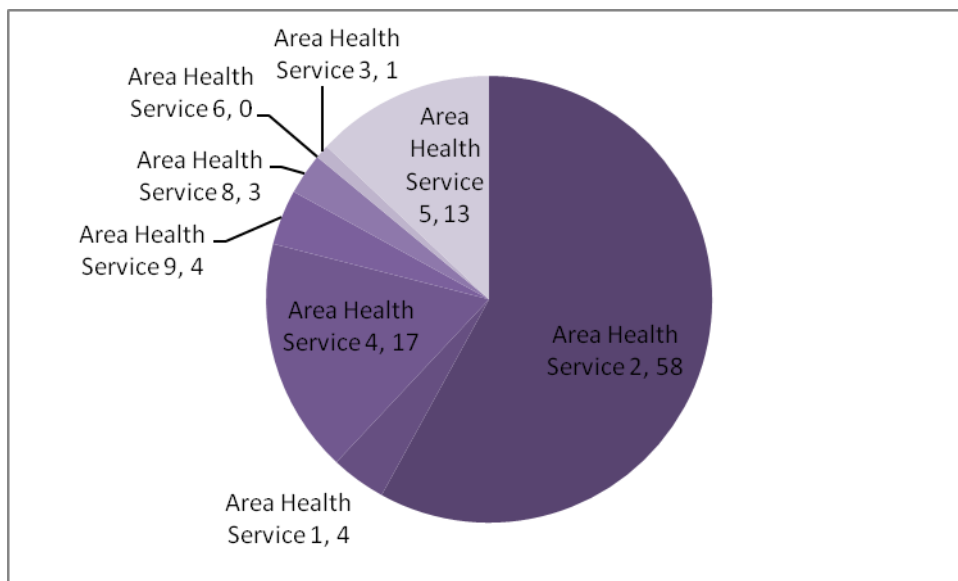
Figures 5, 6 and 7 show the AHS of residence of the new referrals to the three largest services. In each case around 60% of the new referrals live in the AHS where the service is located. The large number of cases for Hospital 1 from the ACT are patients seen in outreach via telehealth in Canberra. The counsellor estimates that 25% of these patients live in the Area Health Service 8 in NSW. In the absence of a large service at Area Health Service 4, a large number of Area Health Service 4 residents go to Hospital 4 and Hospital 1. Similarly in the absence of a large service at Hospital 7, it is inferred that the Area Health Service 5 residents who go to Hospital 4 (80) and Hospital 1 (52) are probably residents of Area Health Service 6 and Area Health Service 3 residents (90) who go to the Hospital 6 or are seen in outreach by the Hospital 6 service are Area Health Service 5 residents. At the Area Health Service 4 service, 81 % of patients live in Area Health Service 4, 6% live in Area Health Service 1 and 5% live in Area Health Service 5. At the Hospital 7 service, 81 % live in Area Health Service 5 and 16% live in Area Health Service 1.

Figure 5 Area of residence of new patients to the Familial Cancer Clinic, Hospital 1, 2008



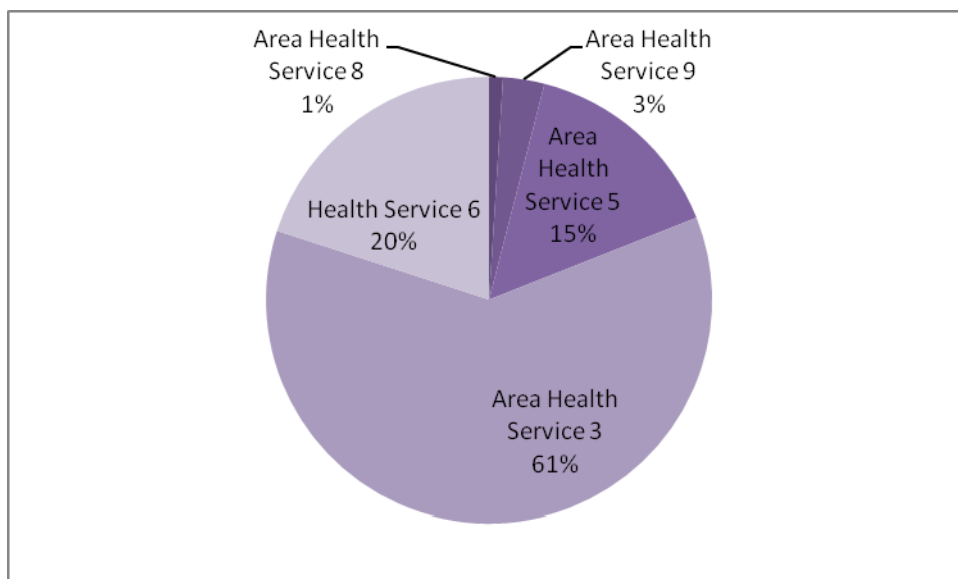
Source: Genesis database at Familial Cancer Clinic at Hospital 1

Figure 6 Area of residence of new patients to the Familial Cancer Service, Hospital 4, 2008



Source: Genesis database at Familial Cancer Service at Hospital 4

Figure 7 Area of residence of new patients to the Familial Cancer Service, Hospital 6, 2008



Source: Genesis database at Familial Cancer Service, Hospital 6 Genetics

20.1.3 Source of referral

Half of referrals to the Hospital 4 service and 38% of referrals to the Hospital 6 Service are from GPs. At Hospital 1, Hospital 7 and Area Health Service 4, GP referrals are much smaller - 24%, 27% and 12% respectively. At these services around 70% of all referrals are from other health professionals, primarily cancer specialists (see Table 16).

Table 17 Source of referral of new patients to the familial cancer services, 2008

Referral Source	Hospital 1		Hospital 4		Hospital 6		Area Health Service 4*		Hospital 7	
	No.	%	No.	%	No.	%	No.	%	No.	%
GP	105	23.7	204	50.9	158	38.2	58	12.0	53	27.0
Self	32	7.2	8	2.0	18	4.4	66	13.7	7	3.6
Other Health Professionals	306	69.1	189	47.1	234	57.4	358	74.3	136	69.4
Total	443	100.0	401	100	408	100.0	482	100	196	100

Source: Genesis database at each service

*Source of referral at Area Health Service 4 is for all patients not just new patients

APPENDIX M

2 1.7 Victoria

The Genetics Services Strategy for Victoria 2005-2009¹² provides an overview of Victoria's Familial Cancer Genetics Services.

The Victorian Familial Cancer Genetics Service (VFCGS) is an integrated statewide service for people concerned about their risk of cancer due to the families cancer history. It is delivered by five providers - Genetics Health Services Victoria (GHSV), Royal Melbourne Hospital, Monash Medical Centre, Peter MacCallum Cancer Institute and The Cancer Council Victoria. The service's goal is to reduce the morbidity and mortality associated with cancer by identifying individuals with an inherited pre-disposition and to address the concerns of people with a familial history of cancer.

The Department of Human Services (DHS) funding covers the provision of clinical genetics and laboratory services, together with the establishment and maintenance of a Familial Cancer Register and support for education of health professionals. Service agreements between DHS and each institution document the aims of the service, guidelines and standards, service arrangements, staff and consumables, data management and allocation of laboratory testing.

The VFCGS offers genetic counselling, genetic testing, medical advice, psychological support and the opportunity to participate in research, to people concerned about their risk of cancer due to the familial's cancer history. Services are provided through four familial cancer clinics with clinics also run in some other hospitals, regional and rural centres. Services are provided on the basis of a doctor's referral or self-referral. A person's family history is taken and an estimate of their inherited risk of cancer is given. Follow up and options for early detection and prevention are discussed. Depending on a person's familial history, genetic testing is offered, together with counselling so that limitations and potential benefits can be discussed.

Services are organised and delivered through a shared care model. Key characteristics are:

- Multidisciplinary service comprising medical oncologists, clinical geneticists, and other medical specialists (surgeons and gastroenterologists), genetic counsellors and laboratory staff;
- Encompasses breast, bowel, ovarian and other cancers and syndromes as required;
- Four familial cancer service providers - Monash Medical Centre (MMC), Peter MacCallum Cancer Institute (PMCI), Royal Melbourne Hospital (RMH) and GHSV;
- Clinical genetics support provided by GHSV though separate agreements negotiated by GHSV with the other three service providers;
- Testing concentrated in three laboratories - originally testing was performed at MMC; however, following a 2002 review of laboratory services (Healthwise Consulting 2002), testing was reduced to three laboratories;
- Reciprocal testing arrangements across service providers with laboratories specialising in tests to optimise quality and efficiency;
- Outreach services to some metropolitan and rural areas provided mainly by GHSV or in collaboration with other service providers (e.g. Peter MacCallum Cancer Institute);
- Catchment areas for referrals to centres that are defined loosely on historical grounds with no centre responsible for a specific region;
- Provision of feedback to a person's doctor including recommendations for follow up care;
- A Victorian Familial Cancer Register, a confidential database of families who have an inherited risk of developing cancer that is stored and maintained at The Cancer Council Victoria, that helps families by sending reminders when screening and tests

are due and updates familial histories so that other familial members are followed up with appropriate care; and

- An education program to improve GP's awareness and management of patients concerned about familial cancer delivered by The Cancer Council Victoria.

This strategy recommended that:

- Current arrangements for the provision of the Victorian familial cancer genetics services should be examined to ensure a unified, integrated and coordinated statewide service. The areas requiring attention include:
 - consistency of eligibility criteria and referral processes, service and testing protocols, risk assessment and reporting processes, and other policies and procedures; and
 - efficiency and cost effectiveness of the current distribution of familial cancer services (clinical, laboratory and outreach).
- Decisions need to be taken in the context of developments occurring in Victoria's cancer services and adult genetics services.

Victoria's Cancer Action Plan 2008-2011¹³ was released in December 2008. One of its priorities was the increase in capacity of the Familial Cancer Genetics Service. The Department of Health re-structured services so that all genetics (general adult and cancer) services were devolved from GHSV to adult genetics departments in four hospitals, with each taking on certain regional responsibilities. The four hospitals are:

1. Peter MacCallum (cancer genetics services only)
2. Royal Melbourne
3. Monash
4. Austin

As part of this re-structure and decentralisation of services, there was a funding enhancement to enable the achievement of one of the key numeric targets of Victoria's Cancer Action Plan namely: The provision of an additional 2,700 patient appointments in clinical cancer genetics across Victoria each year until 2011.

Each service received some enhancement funding for additional staff and the geneticists who had previously been based at GHSV and visited the hospital, were allocated to the hospitals.

2 7.2 Queensland

Genetic Health Queensland (GHQ) is a statewide service which provides all clinical genetics services funded by Queensland Health to urban, rural and remote Queenslanders- a population of nearly 4.5 million people. Services provided include diagnosis, genetic testing, genetic counselling, predictive and prenatal testing counselling.

Clinical genetics covers the full spectrum of patient age group:

- Prenatal genetics testing or diagnosis of foetal abnormality;
- Paediatric genetic conditions and, for example, multiple congenital anomaly syndromes; and
- Adult onset disorders including familial cancer conditions, neurological and cardiac conditions.

Current trends suggest the adult component of the workload is likely to increase. The current adult component is about 60%.

There are 5.4 FTE geneticists and 12 genetic counsellors and administration support. Genetic counsellors based in Cairns, Townsville, Bundaberg, Nambour, Toowoomba and Gold Coast. The genetic counsellors provide a local resource for clinicians and are aware of local services. They develop informal networks with these local services to enable effective referrals of patients. However they are employed by GHQ and participate in a weekly meeting via teleconference to discuss all new referrals. All the outreach counsellors have access to administration support for one day per week.

Medical staff undertake regular clinics in each of the outreach centres;

- Twice monthly in the Gold Coast;
- Monthly in Toowoomba, Nambour and Townsville; and
- Every 2 months in Cairns, Mackay, Bundaberg and Rockhampton.

GHQ uses KinTrack and all sites are networked so there is full access from anywhere in the State.

21.3 South Australia

The South Australian Clinical Genetics Service comprises two discrete units - the Clinical Genetics Unit and the Familial Cancer Unit - and is based at the Women's and Children's Hospital in Adelaide. The Directors of each of these services reports to the Director of the SA Clinical Genetics Service who reports to the Manager of the Genetics and Molecular Pathology Directorate in SA Pathology. The Familial Cancer Unit (FCU) provides genetic counselling and testing for individuals and families at increased genetic risk of developing cancer. The service aims to both prevent the incidence of cancer and to increase the early detection of cancer in the community. The staff of the FCU consists of:

- 1.6 FTE staff specialists;
- 4.0 FTE counsellors;
- 1.0 FTE secretary;
- 1.0 FTE data manager; and
- 0.8 FTE data officer for the cancer registry paid for by The Cancer Council.

There are no counsellors based outside Adelaide but the doctors and counsellors do visit a number of regional centres on a regular basis.

The counsellors are each assigned to a hospital in Adelaide where they provide services. They organise and manage the clinics in that hospital and attend multidisciplinary meetings.

The FCU does not provide follow-up clinics although the breast surgeons, for example, offer high risk management clinics. The FCU does not provide surveillance reminders to patients. However it does provide an annual mailout of the latest advice to the patient and a letter to the referring doctor with a request for the doctor to report back the outcome of their surveillance of the patient.

The FCU uses KinTrack and, according to the Director, it is the core of the service: 'good data management underpins good clinical services'. Doctors and counsellors enter data about each patient so they remain familiar with the system and are aware of the capacities of the system.

The SA FCU reviewed its activities in 2000 and 2006 and it found:

- a slight reduction in overall FTE (5%) - the decrease was mainly in administration staff;
- an increase in clinical staff;
- twice as many referrals;
- 74% increase in patients;
- 32% increase in clinics; and
- 160% increase in the number of families with mutations.

The Director of the service attributes these changes primarily to KinTrack because it enabled staff to operate more efficiently.

27.4 United Kingdom

In 1996, the UK Department of Health set up a working group on cancer genetics services chaired by Professor Peter Harper. The Harper Report¹⁴ recommended organisation of cancer genetics services for England and Wales in a 3-tier structure. First, the role of primary care would be to refer on individuals newly identified as being at potentially high genetic risk of cancer, to follow-up existing high-risk individuals and to reassure individuals at population-level risk. Second, cancer units (based at district general hospitals), staffed by oncologists and surgeons with an interest in genetics, would provide risk assessment and, if appropriate, screening for individuals at moderately increased risk. Third, specialist genetics services would be integrated into the specialist cancer centre, serving a population of 1-2 million. This specialist service would be led by a consultant with training in both oncology and genetics, supported by 2 clinical nurse specialists (or one nurse and one clinical assistant). The specialist service would deal with high-risk individuals and would provide referral and management guidelines for the primary care and cancer unit teams within its region.

While the Harper Report¹⁴ made recommendations on service provision for familial cancer a national study⁴² in 2001 showed great diversity in all aspects of cancer genetics care provision in the UK:

- There were only 11 dedicated cancer geneticists across the 22 centres
- Referral rates varied up to five-fold among the regions and they were 75% higher in centres with a dedicated cancer geneticist
- The proportion of attendees at different risk levels varied significantly
- Individuals at population level risk accounted for 27% of consultations (range 0%, 58%)
- GPs referred significantly more individuals at population risk than did hospital clinicians
- Only 7% of referrals were for men
- 3% were for males and females from ethnic minority groups
- Median waiting time for an appointment was 19 weeks ranging from 4 to 53 weeks

A Genetics White Paper, *Our Inheritance, Our Future: Realising the potential of genetics in the NHS*⁶, published in 2003, recognised the potential impact of genetics and the genome project on lives and on healthcare, and the importance of preparing the National Health Service (NHS) to be able to respond to this new knowledge. It focused on five areas: how to integrate genetics into health services; how to spread genetics knowledge across health services; how to strengthen existing specialised services; how to invest in research and research facilities to drive further discoveries; and how to increase public awareness and understanding of genetics. The investment that resulted from the White Paper enabled development of new genetics knowledge, skills and provision of services within the NHS.

One of the initiatives proposed in the White Paper was to co-fund with Macmillan Cancer Relief pilots in six cancer network areas of a model of service delivery for people at risk of familial cancer.

Issue 2 of the 2007 edition of the journal *Familial Cancer* outlines the progress of the pilots to date, gives an indication of the different approaches used and highlights the issues that have arisen.

Seven sites competitively bid to run the pilot services from 2004-2007: South West London (Royal Marsden NHS Foundation Trust/St George's Hospital); South East London (Guy's and St Thomas' Hospitals); Yorkshire Genetics Service (St James's University Hospital, Leeds and North Kirklees Primary Care Trust); Northern Genetics Service (James Cook University Hospital, Middlesbrough, Teesside); Oldham Primary Care Trust; Somerset Coast Primary Care Trust; and Poole Primary Care Trust. A description of the projects is provided in Appendix N.

Each project was based on the Kenilworth model of service delivery which has the following aims:

- Consistent, correct and appropriate information is available to patients
- Risk assessment is carried out to an agreed framework; referrals are streamlined in accordance with agreed pathways . individuals are consistently managed according to their level of risk.

The pilots explored a number of issues:

- Different approaches to triage of risk ie in terms of different information technology and the point at which risk triage occurs - not in terms of risk guidelines (all followed standard cancer genetics risk principles)
- How to increase the uptake of services by black and ethnic minorities using various strategies
- Method of delivering care - telephone, home visiting
- The skill base of cancer genetics' service staff and the need for core competencies for counsellors and medical staff
- The experience of the users of the service

The evaluation of these projects is detailed and complex and cannot be summarised easily here. Two important outcomes of these projects should be noted:

- All projects required non genetics practitioners to undertake risk assessment activities within agreed protocols and with close supervision, mostly in mainstream services. 'For all projects, the most important element was the support of the genetics department, including on-going direction and mentorship, clinical supervision and observation and continuing opportunities to discuss cases with colleagues and with experts at the genetics centre [Furthermore] it was important that the relationship of practitioners with the specialist genetics department was formalised.

- Issues related to cancer genetics straddle two specialities (genetics and oncology). This made it difficult to engage managers, establish service sustainability and secure on-going funding.

Central funding for the pilots was to come to an end in 2007, but at the time of these articles in Familial Cancer all are seeking ongoing NHS funding and ensuring that the learning from the evaluations is rolled out into routine NHS services.

As space is limited in this report two case studies from these pilots, reported in the Progress Review of the 2003 White Paper⁶, are reproduced here.

Case study: Teesside Cancer Familial History Service

This service was set up in 2004 with funding from the Department of Health's Macmillan Cancer Care Support Familial Cancer Programme. It aimed to provide a more efficient and accurate genetic risk assessment for families affected by cancer within the Cancer Care Alliance network, which covers a population of about one million.

In order to meet the needs of patients more effectively, a new role of genetic risk assessment practitioner (GRAP) was created to work with patients who have a moderate risk of developing cancer. Within the pilot area, patients assessed as high risk were referred to the genetics cancer specialist to discuss the possibility of genetic testing, while patients with a moderate risk were cared for by GRAPs, who organised additional cancer screening and provided extra monitoring and advice on prevention.

In the 30 months of the pilot study, the Cancer Familial History Service received over 2,600 referrals, two-thirds from cancer clinics. In most cases, referrals were much more accurate than previously, as the GRAPs worked with an improved risk assessment model and were able to collect more accurate familial history information.

A collaborative breast cancer audit showed that, before the new service was introduced, some 30% of women who were undergoing regular extra mammography screening were wrongly thought to be at increased risk. Cancer genetics specialist Dr Paul Brennan, who led the project, said: "We are proud that we have successfully managed change in a large clinical network. We have established a novel, efficient service and can demonstrate its benefit in a number of ways. We have broken the boundaries between primary, secondary and tertiary care."

In 2001, Poole Primary Care Trust set up a unique Community Cancer Nursing Service (CCNS) comprising five cancer specialist nurses experienced in working within the community. This team works with 28 GP practices, the hospital cancer services and palliative care teams to provide a comprehensive care service to the people of Poole.

Until now, risk from specific genes was assessed by specialist genetics services. Poole's nearest genetics service is based in Southampton. As more people became aware of the risk of cancer, many who were worried, but not at very high risk, were being referred. GPs did not know how to identify patients at highest risk. In addition, many families struggled to produce accurate cancer history details to help the clinic with risk assessment. These two factors meant that waiting times rose to unacceptable levels.

With funding from the Department of Health/Macmillan Familial Cancer Programme, the Poole CCNS worked with a genetics counsellor from Southampton General Hospital to develop the skills and knowledge required to:

- Provide risk assessment in cases referred by GPs
- Help patients gather information about familial illness history

- Explain the risk assessment process.

Patients greatly appreciated this new personal and sympathetic local service. In the first few months, a small number of people were found to be at high risk. However, many more patients found that their risk was no greater than that of the general population. This helped reassure many worried individuals and families and reduced pressure on the specialist genetics clinic.

2 7.5 Canada

21.5.1 British Columbia

The Familial Cancer Program (HCP) is a result of the BC Cancer Agency (BCCA) and the BC Provincial Medical Genetics Program working together to provide information and genetic counselling for individuals and families with a strong history of cancer. The service has been available for about 15 years and comprises of 6 genetic counsellors, a nurse educator, clerical staff and consulting medical geneticists. The service is based in Vancouver but recently genetic counsellors have been based in cancer centres at Abbotsford and on Vancouver Island. Outreach services are also provided from the Vancouver office. Whereas the counsellors are employed by BCCA, the medical geneticists are contracted from the BC Medical Genetics Program. The HCP uses the following staffing ratio: 1.0 genetic counsellor: 0.1 medical geneticist: 0.5 clerk.

When a referral is made, the patient is sent a familial history form and, based on the information provided, an appointment is made with a genetic counsellor or a medical geneticist. The genetic counsellors see all patients with common familial disorders and the doctor sees patients with rare cancers or patients who require a physical examination. At present the waiting time for an appointment is 3-4 months. This time can be expedited for medical reasons. If the patient is eligible for testing, blood is taken and sent to the laboratory in the BCCA in Vancouver. The waiting time for mutation screens is currently one year. Most test results are given over the phone at a scheduled time. All patients who receive their results by phone are offered the option of a follow-up appointment with a genetic counsellor.

The HCP does not provide any ongoing screening or management. Women with a BRCA1 or 2 mutation attend the high risk screening clinic at the Cancer Centre in Vancouver. For patients with other conditions, screening recommendations are sent back to the familial doctor to provide on-going management.

Currently the HCP sees 1200 new referrals per year. About 400 are index cases with suspected BRCA 1 or 2 mutations and 100 are index cases with suspected Lynch syndrome. The remaining 700 cases are unaffected relatives of the index patients.

At the present time there is no formal leadership of the HCP because the previous Director has retired. Currently the service is deciding the nature of its future leadership.

21.5.2 Ontario

A 2008 report describes the cancer genetics services in Ontario¹⁵. There are 13 regional centres and 9 northern or satellite clinics throughout Ontario offering cancer genetic services. A regional centre has a full complement of skilled clinical and laboratory staff and provides a wide range of services to the catchment area. A northern or satellite clinic has a limited complement of staff on-site, chiefly genetic nurses/counsellors, augmented by a formal relationship with a clinical geneticist in a regional centre who visits on some pre-determined basis. Such services may serve a large catchment area, usually with lower population density.

According to the report, there is variable consistency among all service sites in terms of using a standard service delivery model (i.e. types of referrals seen at each site, types of health care professionals providing services, and wait times). Although there are standard criteria for inherited cancer for counselling, testing and clinical management, the report notes that there is a need to ensure that these are up to date and used consistently, and for the services to have the ability to track utilisation of counselling services. This would facilitate the monitoring of access to services, ensure a quality standard for the services delivered across the province, and highlight gaps in services related to wait times and availability of services in various regions, as well as assist in resource planning (personnel, funding, service locations).

According to a survey of 22 of the centres/clinics conducted by the Task Force in June 2008, referrals for cancer-related genetic counselling have increased by 61 % since 2002 (3,966 referrals reported in 2002, compared with 6,378 referrals reported in 2007) with no evidence of a plateau to date. As it is estimated that 5% to 10% of cancers are currently believed to be related to an inherited predisposition, cancer incidence projections suggest that this would equate to an anticipated demand of 6,300 primary referrals for counselling, plus eligible familial members. According to the report, there was consensus within the Task Force that clinical services are not able to meet current need.

The survey revealed that familial breast and ovarian cancer referrals dominate current service use and account for about 80% of all referrals. In the opinion of both the survey respondents and representatives of the Ontario Familial Colon Cancer Registry, colorectal cancer patients are currently being under-referred.

There are 5.5 FTE geneticists, 3.85 FTE oncologists and 34.54 FTE genetic counsellors providing cancer genetics services for the province. Based on these figures, the ratio of doctors to counsellors is 1.0: 3.69. On average there is 0.9 FTE administration staff per centre with a range of administration staff per centre of 0.4 FTE to 1.8 FTE.

There are 13 molecular genetics laboratories across the province providing testing for inherited cancer. According to the report demand has increased between 2003 and 2007 but not to the same extent as demand for genetic testing of acquired disease. For higher volume molecular tests, there may be several labs offering the test. The decision about which tests to offer rests largely with individual lab directors and is often based on institutional demand. For rarer tests, there was an effort amongst the labs in the past to centralise testing in fewer labs for quality assurance and cost-effectiveness purposes. In recent years, there has been a trend toward more duplication of services, with less regard for volume and cost considerations.

The recommendations made by the Task Force in this report are:

- 1 Immediately establish a provincial oversight body for molecular oncology services to oversee system planning for these services in Ontario; and to advise key stakeholders on test approval, delisting of obsolete tests, funding, licensing, accreditation, credentialing, quality assurance, location of services, and to ensure alignment of oncology services with all other genetic services.
2. Implement a mandatory approval process for each genetic test performed by laboratories in Ontario and administered by the appropriate regulatory agencies to ensure that only appropriately accredited and licensed laboratories with credentialed personnel are reporting and interpreting laboratory results that impact patient care.
3. Ensure that each genetic test performed by a laboratory meets rigorous quality assurance criteria and is regularly subjected to proficiency testing (external validation).

4. Establish and fund an existing or new information-sharing mechanism to ensure that critical information is readily available to referring physicians and the public regarding availability of tests, how to refer, what data the test will yield, its limitations, patient eligibility criteria, specimen handling guidelines, and clinical management.

5. A comprehensive and competitive approach to funding should be established that addresses dynamic volumes and variation in complexity of testing and clinical care, as well as the costs associated with risk assessment and genetic counselling. Funding should capture province-wide utilisation of testing and clinical services and be linked to quality and utilisation data.

6. Promote translational research and establish an "advance notice" process to ensure that the system is prepared to implement new tests and technologies when evidence warrants.

The report warns that 'Not addressing these issues will result in insufficient infrastructure to meet growing demands and an inability to implement and fund new tests'. In the absence of a coordinated system with proper oversight and quality assurance mechanisms in place, the potential for a breakdown of the system that affects patient safety and treatment is a genuine risk.

APPENDIX N

22.7 Summary of information provided by UK pilot projects and main roles developed

Project	Focus of project	New role in non genetics service	New roles in specialist genetics services
Teeside Cancer Familial History Service – Cancer Care Alliance of South Durham, Teeside and North Yorkshire in Teeside Hospitals NHS Trust and Newcastle upon Tyne Hospital Foundation NHS Trust	To develop a model for risk assessment and counselling for families at risk of breast, bowel and ovarian cancers	3 Genetic Risk Assessment Practitioners (GRAPs)	
South East London Genetics Service – Guy's and St Thomas' Hospitals, London in conjunction with Lambeth and Southwark Primary Care Trusts.	Provide a 'whole systems' approach – promote awareness of cancer familial history amongst the local population and health care professionals. Targeting black and ethnic minority groups in deprived inner city London boroughs of South East London. A community based cancer familial history risk assessment service with a strong emphasis on self referral.	Macmillan Community Clinical Nurse Specialists	Maemillan Cancer Familial History Nurse Trainer
Oldham Primary Care Trust	Taking service to primary care and community setting	GP with a Special Interest (GPwSI); Clinical Nurse Specialist for patients with history	
Somerset Coast Primary Care Trust, Avon, Wiltshire and Somerset Cancer Network, Cancer Service Users	To develop a new primary care led cancer genetics service tailored for a rural population, which aims to reduce inequalities in breast, ovarian and colorectal cancer service provision. It explores a new service model that is patient focuses, provides service close to home and reduces hospital visits and pressure on genetics departments	Cancer Genetics Project Coordinator; Primary Care Genetics Nurse Specialist	
The South West Genetics Service – St George's	To test the feasibility, accessibility, and effectiveness of providing genetic nurse counsellor clinics in primary and secondary	Cancer Genetics Nurse/ Macmillan Project Coordinator	

	care in the SW Thames Region		
The South West Genetics Service – The Royal Marsden Hospital	To evaluate a teleconference counselling model for provision of risk assessment	Cancer Genetic Nurse Counsellor	
(Leeds) Yorkshire Regional Genetics Service, North Kirklees Primary Care Trust and Bradford Hospital NHS Trust	Improving access to cancer genetics services for people from ethnic minorities and lower socio-economic groups.	Practice nurse; GPwSI (reigned); Breast care nurse/genetic counsellor (50%/50%)	Genetic counsellor working to set up clinic in primary care.

References

1. *Human cancer syndrome: clues to the origin and nature of cancer.* **ER, Fearon.** Science 278: 1043 -1050, 1997.
2. *Cancer genetics services: a systematic review of the economic evidence and issues.* **GL Griffith, RT Edwards, and J Gray.** British Journal of Cancer, 2004, Vol. 90. 1697-1703.
3. *Estimating the referral rate for cancer genetic assessment from a systematic review for the evidence.* **C. Featherstone, A. Colley, K. Tucker, J. Kirk and M. Barton.** The British Journal of Cancer, 2007, Vol. 96. 391-398.
4. **Officer, Working group for the Chief Medical.** *Genetics and cancer Services.* London : Department of Health, 1996.
5. *A descriptive study of UK cancer genetics services: an emerging clinical response to the new genetics.* **D. Wonderling, P. Hopwood, A. Cull, F. Douglas, M.Watson, J.Burn, and K. McPherson.** The British Journal of Cancer, 2001, Vol. 85 (2): 166-170..
6. **Department of Health.** *Our Inheritance, Our Future: Realising the potential of genetics in the NHS.* London : Government White Paper, 2003.
7. *Competencies, education and support for new roles in cancer genetics services: outcomes from the cancer genetics pilot projects.* **C. Bennett, H. Burton, P. Farndon.** Familial Cancer , 2007, Vol. 6 (2): 171-180.
8. *National evaluation of NHS genetics service investments: emerging issues from the cancer genetics pilots.* **G.P. Martin, R. Finn, and G. Currie.** Familial Cancer, 2007, Vol. 6(2): 257-263.
9. *An assessment of the efficacy of cancer genetic counselling using real-time videoconferencing technology (telemedicine) compared to face-to-face consultations.* **J. Coelho, A. Arnold, J. Nayler et al.** European Journal of Cancer, 2005, Vol. 41: 2257-2261..
10. **Science and Technology Committee.** *Genomic Medicine.* House of Lords, 2009.
11. **Cancer Institute NSW.** *Review of Cancer Genetics Services in NSW.* Sydney, 2007.
12. **Australian Healthcare Associates.** *Genetics Services Strategy for Victoria 2005-2009.* Department of Human Services, 2005.
13. **Victorian Department of Human Services.** *Victoria's Cancer Action Plan 2008 - 2011.* Melbourne: 2008.
14. *A descriptive study of UK cancer genetics services: an emerging clinical response to the new genetics.* **D. Wonderling, P. Hopwood, A. Cull, F. Douglas et al.** The British Journal of Cancer, 2001, Vol. 85(2): 166-170.
15. **Molecular Oncology Task Force.** *Ensuring access to high quality molecular oncology laboratory testing and clinical cancer genetic services in Ontario.* Cancer Care Ontario, 2008.