Response by the
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The Health Workforce

Productivity Commission Issues Paper
May 2005

Response by the
Human Genetics Society of Australasia
Special Interest Groups

Australasian Association of Clinical Geneticists (AACG)
Australasian Society of Cytogeneticists (ASoC)
Australasian Society of Genetic Counsellors (ASGC)
Australasian Society for Inborn Errors of Metabolism (ASIEM)

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The Human Genetics Society of Australasia.

The Human Genetics Society of Australasia (HGSA) is a professional society of those working in Human Genetics. It provides a forum for the study, investigation and practice of Human Genetics with the following objectives:

- High ethical standards among those working in Human Genetics.
- Communication between those working in Human Genetics.
- Training and professional recognition for those involved in Human Genetics.
- Professional and lay education about Human Genetics.
- Promotion of public awareness of Human Genetics.
- Consideration and comment upon matters relevant to Human Genetics or the interests of the Society.
- Representation of the interests of Human Genetics and those working in the field, and of the Society and its members, in public, professional, governmental and other forums.
- Promotion and support of research in Human Genetics.
- Performance or support of any activity which may achieve all or any of the foregoing.

There has been exponential growth in the information on the human genome, providing ever increasing knowledge on the causes of inherited diseases and health problems. The delivery of genetics services will require expanded access to both genetic diagnostic tests and support services. In the consideration of the Australian health workforce, it is essential that the needs for growth of genetic health provision are contemplated. Both the number of tests and the range of genetic diseases for which tests can be offered will increase and require an increase in the number of laboratory scientists for processing. This must be combined with expert advice and information in the delivery of genetic counselling and support to patients with equity of access across Australia.

The issues and needs for the future workforce have been separately considered by the Special Interest groups of the HGSA. These groups comprise professionals in particular areas of Human Genetics research and clinical service delivery:

- Australasian Association of Clinical Geneticists (AACG)
  The AACG represents the subspecialty of Clinical Genetics. It is a Special Interest Group of the HGSA, and provides expert advice about this field of health care to other bodies including the Royal Australasian College of Physicians, to which most Australasian members of the AACG belong.

- Australasian Society of Genetic Counsellors (ASGC)
  Counsellors specifically trained in the delivery of information and support regarding genetic diseases. They usually will have a background in science, nursing or psychological counselling.

- Australasian Society of Cytogeneticists (ASoC)
  Scientists with specific (usually on the job training) in human clinical cytogenetics.

- Australasian Society for Inborn Errors of Metabolism (ASIEM)
  ASIEM is a multidisciplinary special interest group of HGSA with membership drawn from metabolic clinical geneticists, metabolic nurses, dietitians and laboratory scientists

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Submission by the Australasian Association of Clinical Geneticists

What is clinical genetics?

The majority of Australians have a family history or personal medical history of genetic disease, and could potentially benefit from genetic counselling. Although most (not all) genetic disease is uncommon, the number of different diseases is huge and knowledge of these is beyond the capacity of health professionals outside clinical genetics. Undergraduate, graduate and postgraduate training programs are becoming shorter and busier, and teaching time and curriculum topics are jealously guarded so that most medical students, general practitioners and specialists have been taught very little genetics. Not all have the time or training to collect and interpret a family history from their clients. In the next few years, all medical practitioners will need to have, or have access to, some knowledge of genetics to interpret and explain genetic screening tests that will identify predisposition to common multifactorial diseases, and predict desirable or adverse responses to different treatments or preventions. This knowledge will improve if adequate basic and clinically relevant genetics is taught in undergraduate or postgraduate programs. Until then a major function of Australasian Clinical Genetics services will be to advertise their service to the public and the profession, interpret genetic concepts to clients and their caregivers, and offer to train students at all levels.

Consideration of future productivity requires an understanding of the function of clinical genetics services. Members of the public, media and especially the health professions have misconceptions about genetics and health. The following vignettes illustrate typical Australasian clinical genetics services provide.

- ***A couple was referred with their child for diagnosis. The child had severe global developmental delay and a number of malformations, including congenital heart disease that required surgery. The parents felt guilty and wanted to know what caused their child's problem. They were not prepared to have another child unless they could be assured that future children would be unaffected. Previous assessments and tests had not reached a diagnosis, and the parents were beginning to lose hope and faith in the medical system. The service provided by the clinical genetics service included supportive genetic counselling, validating the parents' hopes, fears and frustrations, and helping them to deal with the uncertainty about their child's diagnosis and prognosis. Although a genetic diagnosis was not immediately apparent, after 2 years a new test became available. Luckily, the parents were referred back to Genetics for review. They agreed to do the test, which showed that their child had a tiny previously undetectable chromosome deletion. Further testing showed neither parent carried this. They were reassured that they could not have done anything to cause or avoid their child's problem, and that their future children will be unaffected.***

- ***A young woman, whose father died with Huntington disease (HD) when she was 15, requested presymptomatic DNA testing for HD so that she could plan her career and family. Preliminary counselling informed her of the need to finalise plans for insurance, the need for confidentiality about the test and the availability of supportive counselling and assessment by a psychiatrist and neurologist. Subsequently when the test was done, the HD Social Worker based at the genetics service supported her and her family during the stressful period of waiting for results. The DNA test showed she had the gene, which was extremely disappointing. The Social Worker continued to see her and accompanied her on subsequent visits to a psychiatrist who managed her depression and a neurologist who reassured her each year that no signs of HD were present. She decided to have her children early in life, to maximise the time she would be well while they were dependent on her. Five years later, although still very sad that she had the gene, she stated that she appreciated knowing the result, would not do things differently if given the time over***

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again and hoped that a treatment might be available in time for her and her children to benefit.

- ***An ultrasound scan showed that the baby of a couple seen at the Prenatal Diagnosis Service had a lethal condition which was probably genetic. The geneticist at the clinic talked to the couple about a range of diagnostic possibilities and they saw the value of performing several tests on the baby. The baby was stillborn and was examined by the geneticist who made a clinical diagnosis, recommended tests that identified the condition, and explained how the condition could be detected in a future child or embryo. The couple would not terminate pregnancy, but chose to have IVF and transfer an unaffected embryo by preimplantation genetic diagnosis.***

- ***A healthy man whose father, grandmother and aunt had bowel cancer saw a cancer Genetic Counsellor, who also offered genetic counselling to the surviving affected relatives and with their consent obtained documentation of their diagnoses. Testing of a sample of the father's tumour suggested an inherited form of bowel cancer. Subsequent DNA testing of the father's blood identified a mutation in a hereditary non-polyposis colorectal cancer gene. This allowed testing of our client who found he did not carry the mutation. He was greatly relieved, and now knew he and his children had no increased risk to develop cancer. He, his descendants and the health system were saved the time, expense and hazard of screening colonoscopies. His brother also had the DNA test which showed he carried the mutation, and he had a colonoscopy which detected a small cancer early enough for definitive surgery.***

- ***A woman whose mother, grandmother and aunt died in their 30s with breast cancer, and whose sister had just been diagnosed with ovarian cancer, was referred by her GP for genetic counselling and a BRCA1 mutation was found in her sister's blood sample. The client then had a blood test and found she had the BRCA1 mutation. The cancer genetic counsellor gave her the latest information about research on effects on hereditary cancer of oral contraceptives, prophylactic mastectomy or oophorectomy and screening. She started having regular screening for breast cancer and planned to have a prophylactic oophorectomy when her family was complete.***

- ***A grandmother whose two adult sons had severe intellectual disability and still lived with her, and whose daughter also had an affected son, had stayed in touch with the genetics service for 15 years. Her gran- daughters were unwilling to have children for fear of having an affected son. This year a laboratory collaborating with the genetics service found the gene change that caused the intellectual disability, providing women in the family with a free blood test that will tell them if they are carriers, and providing a test for prenatal diagnosis or preimplantation genetic diagnosis for carriers. Genetic counsellors contacted all members of the family who previously asked for help. Those who had moved out of town could be referred to their closest service. All unaffected relatives who were still of childbearing age had decided not to have children unless a test became available, and were very relieved that they could be assured of an unaffected baby.***

Therefore, a primary function of clinical genetics services, in addition to being a reasonable definition of the art and science of Genetic Counselling, is to provide clients with the knowledge they need to plan their future and their family. Knowledge is a family's greatest weapon, along with the courage and optimism shown by most, against the uncertainty, fear and suffering engendered by diseases that cannot be cured or easily prevented.

What is a clinical genetics service?

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This is a multidisciplinary team consisting of clinical geneticists, genetic counsellors, specialist social workers, administrative officers and data managers or data entry officers.

Most clinical geneticists are consultant physicians or paediatricians trained by the Royal Australasian College of Physicians. Others trained equivalently overseas or with other Colleges such as the RACOG.

Genetic counsellors come from a wide range of backgrounds including nursing, social work, laboratory science, psychology and teaching. There are several postgraduate university courses in genetic counselling, successful completion of which confers part 1 of professional accreditation by the Genetic Counselling Board of Censors of the HGSA. Complete accreditation requires at least 2 years of full time equivalent supervised training while employed as a genetic counsellor.

Most genetic social workers work with patients and their families affected by Huntington disease or other neurogenetic diseases. Their work differs from most other social work because their affected clients are usually young and are poorly served by nursing institutions designed for aged patients, and files can rarely be closed when a patient dies. In contrast, this event usually leads to several relatives from the next generation coming to ask for advice and support, and the enrolment of new nuclei in each family as they get older.

Australasian Genetics Services are located in state or regional capitals with close affiliations, in both research and clinical service, to tertiary teaching hospitals, medical schools and specialised laboratory services including cytogenetics, molecular genetics, biochemical genetics and newborn screening. Most patients are seen as outpatients and are referred by general practitioners or organ specialists. The services usually have several subspecialty interests and conduct clinics or research in these areas, often with a state-wide or national referral base. The majority conduct outreach clinics. In NSW and Queensland, outreach services in regional cities are staffed by genetic counsellors who reside in the outreach area.

Clinical Governance

The HGSA and the Clinical Governance Committee of the British NHS (CGS) have published guidelines for the structure and function of clinical genetics services, and their practice of genetic counselling. The HGSA guidelines for Australasian Services were published in 1999, and are to be revised this year (HGSA 1999; HGSA 1999):

Genetics units - Levels of service

Level 1

- No on-site genetics counsellor.
- Staff able to arrange on site clinics with clinical geneticists/genetic counsellors.
- Phone consultation with clinical geneticist/genetic counsellor is available.
- Access to general genetics educational information.
- Access to interpreters.

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Level 2

- Services provided by a genetics counsellor.
- Provide information and counselling for individuals and family members.
- Counselling and diagnostic services provided by visiting clinical geneticists.
- Phone consultation with clinical geneticist/genetic counsellor is available.
- Access to interpreters.
- Access to pathology services for diagnostic purposes.
- Collection of relevant service data and submission to reporting authority.

Level 3

Services as level 2

plus

- Clinical genetics services provided by a less than full-time clinical geneticist.
- Clinical geneticist on call service usually available.

Level 4

Services as at level 3

plus

- Service operates as a separate unit providing outreach referral services.
- Full-time clinical geneticist with after hours availability.
- Access to relevant specialists eg oncology, neurology, gastroenterology and cardiology as required by disease diagnosis.

Level 5

Services as at level 4

plus

- May provide state-wide or national expertise in a specific disorder or disorders
- Usually co-located with genetics laboratory services.

Each Genetics Unit with on-site staff, level 2 and above should have:

- A clear definition of the range of services provided.
- Procedure manuals outlining the mission, vision and goals of the genetics unit.
- A confidential genetics record system, accessible by authorised staff of the unit.
- Appropriate and dedicated funding.
- A commitment to ethical provision of services.

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Staffing

The recommended staffing per population served is one clinical geneticist, one genetic counsellor and one support staff member per 300,000 population. [1,2]
The minimum composition for a level 4/5 service is:

- 2.0 Full Time Employed (FTE) accredited clinical geneticists.
- 2.0 FTE accredited genetic counsellors.
- 2.0 FTE administrative support staff (clinic coordination/secretarial duties/data management).

Additionally a level 4/5 service may provide positions for:

- Trainee clinical geneticists, trainee genetic counsellors and trainee medical specialists.
- Staff engaged in research.
- Staff involved in professional or community education about genetics.
- Social workers

The staff of a clinical genetics unit may be based in one institution or spread over multiple sites within the region served. For example, genetic counsellors working in country centres may be supervised by a clinical geneticist based in the central unit of the region.

The minimum staffing for an outreach genetic counselling service, level 2/3 serving a population of 150,000 or less, should be:

- 1.0 FTE accredited genetic counsellor
- 0.5 FTE administrative support staff (clinic coordination/secretarial duties/data management)

The CGS recommends that 90% of general appointments should be within 13 weeks of referral. The HGSA recommends no longer than 6 weeks for general appointments, 7 days from referral for urgent appointments such as those that involve a current pregnancy and 2 months, or the next available clinic, for outreach clients. Most NSW clinical genetics services have a waiting time exceeding 3 months, some are closer to 6 months, and some outreach centres have a waiting list of a year.

Genetics services hold weekly intake meetings where new referrals are presented, and collection of further relevant information is organised so that diagnoses can be confirmed at the clinic appointment. A genetic counsellor or social worker contacts the family to collect a family history, find what questions the family wants answered and arrange for documentation of diagnoses made earlier. At the clinic interview which usually lasts an hour, a team or individual sees the family, depending on complexity and need for a diagnostic assessment by the geneticist. Most of the work is done by counsellors and trainees where diagnoses like cystic fibrosis, Huntington disease or familial cancer have already been made elsewhere. The geneticists supervise counsellors and trainees, provide diagnoses or confirm them where there is uncertainty, order and interpret tests. A letter is usually written to the referrer, the family, and other agencies if the family desires. Follow-up arranged with the family might include a phone call from the counsellor or seeing the family to review test results, attempt another diagnostic assessment or see relatives. Outreach clinics are run differently in different states. Some outreach centres are visited by a geneticist and counsellor who are based in the main service. Outreach counsellors who live in the outreach region arrange clinic appointments, send pedigrees and documentation to the visiting geneticist before the clinics and arrange follow-up

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locally. Telemedicine or "telegenetics" is being introduced for outreach cases that are urgent or do not need examination.

Most genetics services hold clinical review meetings, where clients seen in the previous week are presented. This peer review process helps to confirm diagnosis, ration pathology testing and ensure continuity of follow up for large families that might be seen by several staff or different services. Staff also attend grand rounds, journal clubs, meetings with affiliated laboratories, ward consultations and multidisciplinary or subspecialty clinics. Genetic counsellors who live in outreach areas in Queensland and NSW visit the main service for continuing education and business planning.

Current workforce

Table 1: Number of staff (FTE) in each state, July 2005.

State	*Geneticists[a] (FTE per million population[b])*	*Metabolic genetics specialists[c]*	*Clinical genetics trainees*	*Counsellors*	*Social Workers[e]*	*Administrative staff[f]*
Qld	*5 (1.28)*	*1.5*	*1*	*13.6*		*7.2*
NSW	*15.9 (2.38)*	*2.5*	*4*	*27.1*	*2*	*16.5*
Vic	*7 (1.41)*	*2*	*3*	*13.5*	*2.3*	*10.3*
SA	*3.6 (2.36)*	*1*	*0*	*6*		*5.1*
WA	*4.5 (2.27)*		*1*	*8*		*6*
Tas	*0.4 (0.8)*		*0*	*2*		*0.2*
Total	*36.4 (1.78)*	*7*	*9*	*70.2*	*4.3*	*45.3*

Notes:

[a] General and cancer geneticists. Excludes metabolic genetics, and clinical geneticists working solely in public or private fertility or prenatal diagnosis clinics.

[b] Based on census data 2003-4

[c] All except one trained as clinical geneticists, all except 0.5 FTE work entirely in metabolic genetics. Excludes biochemical genetic laboratory personnel.

[d] Genetic Counsellors, nurses, includes cancer genetic counsellors. Includes outreach genetic counsellors: 11 FTE in NSW, 3 FTE in Qld and 1 FTE in Victoria.

[e] Specialist social workers eg for Huntington disease

[f] Clerical, business management and data entry/management staff. Some services only have clerical staff. Includes staff working in clinical cancer genetics units. Does not include the few staff working in genetic metabolic specialist units (less than 5 FTE nationally) as these are shared with academic and laboratory services.

Changing benchmarks:

The submission of the specialty of clinical genetics to the Australian Medical Workforce Advisory Committee (AMWAC) in 2003 forecast that Australia would need one clinical geneticist per 200,000-250,000 people, or 4-5 per million. The best-served states have less than 50% of the optimum number of geneticists. A minimum of 2.0 FTE clinical geneticists is required per urban service that serves 500,000 people. 2.0 FTE genetic counsellors and 1.5 FTE administrative staff are required per geneticist. For a resident rural practice the recommendation to AMWAC was 1.0 FTE clinical geneticist, 1.0 FTE genetic counsellor and 1.0 FTE administrative officer for a population of 150,000. It is difficult to predict future trends in genetics as demand for services will depend largely on the availability and costs of tests. At least as many clinical geneticists working in cancer as general clinical geneticists will possibly be required. The ratio of genetic counsellors or social workers per geneticist is not standardised and optima are not known. The AMWAC submission recommended an optimum ratio of 2:1

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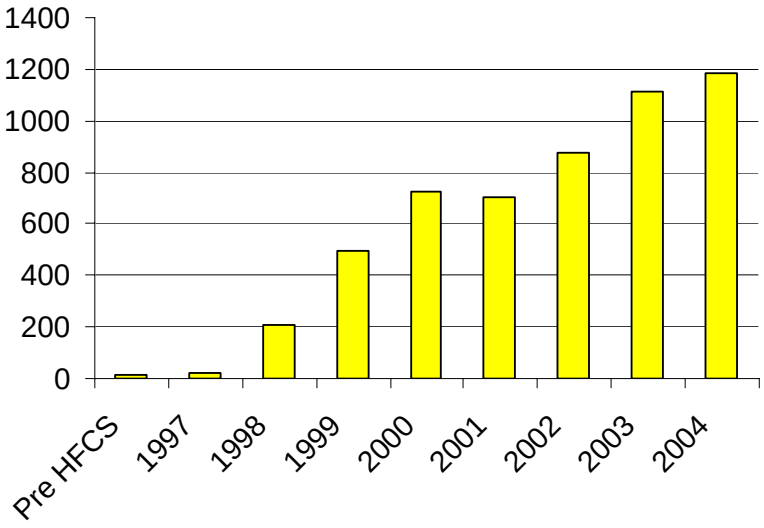
counsellors to geneticists. The ratio could be 4:1 for conditions diagnosed by other specialties, for which counselling and DNA testing protocols have been well documented, eg adult onset conditions like familial breast/ovarian or colorectal cancer and neurogenetic conditions like Huntington disease. AMWAC has reported that an urban or rural population of at least 80,000 is required for a clinical genetics service (Anonymous 2005) but it does not specify the structure of the service. The UK Clinical Genetics Society set a benchmark of 3 clinical or cancer geneticists per million population in 2000 (Hughes, Donnai et al. 2000).

Administrative/clerical/data management staff are often the first to go when funding is tight and the last to be added to services. Planning for the next 10 years needs to include the administrative staff including those who maintain databases. As clinical genetics involves the care of families, follow-up is essential so that relatives can be given appropriate information as they get older. A Canadian geneticist has been sued for providing advice that was the standard of care for patients referred in 1985, but not having the resources to call all patients back, or prompt referring doctors to do this, when new tests became available (Hunter, Sharpe et al. 2001). Ideally, genetics services should act like cancer registers for every disease they see, so that they can contact consenting clients when new assessments or treatments are indicated. It will be essential for all genetics services to have data managers or at least data entry staff, but not every service has one.

After the available benchmarks were set, the demand for genetic counselling and the number of conditions for which DNA tests are available have increased. Figure 1 shows the increase in cancer services since DNA testing became available, although this trend represents increased in all areas of service, not just the cancer data shown.

Figure 1: Yearly occasions of service of a familial cancer service serving a local population of 450,000.



All services report increases in their activity. Budgets have usually not taken into account the increasing seniority of staff, increase of salaries with CPI, a predominance of women in clinical genetics services, who plan to start families now that they have finished training and have a job, changed attitudes of newer graduates to 80 hour weeks, and OHS requirements and common sense dictating that geneticists no longer drive 5 hours to and from 9 hour clinics. All these changes have meant that increases in activity are more likely to be achieved by use of multidisciplinary teams rather than by increasing the activity or number of geneticists. The cost of testing borne by clinical genetics services has also increased, reflected by colorectal cancer testing in a typical service (figure 2). Tests are only informative for a proportion of

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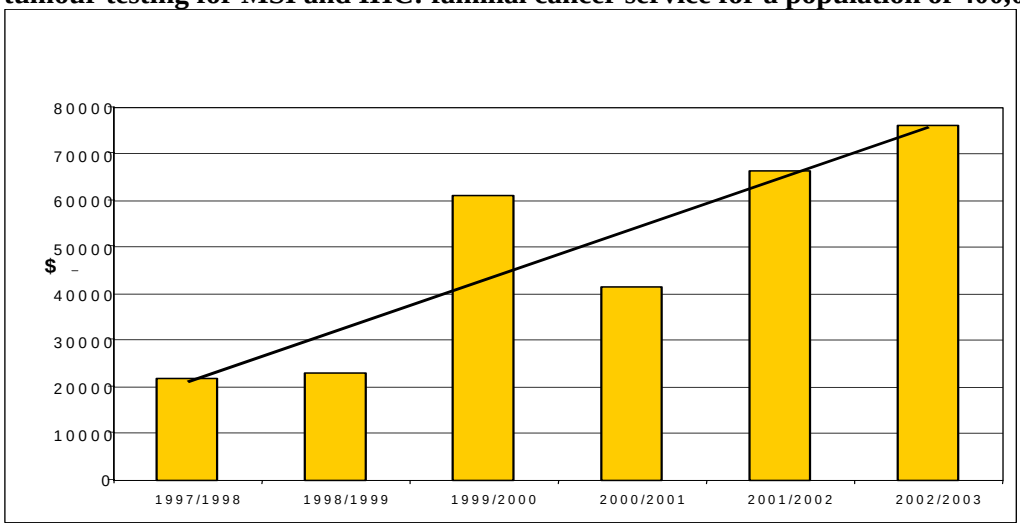
cancer families and those with intellectual handicap. As new tests, and especially therapies are developed, the rate of increase in demand is likely become exponential, while the activity of services will be constrained to an inadequate linear increase, unless there is a huge leap forward in foresight and funding (see UK White Paper, following).

Figure 2: Total costs of breast and colorectal cancer genetic testing including bowel tumour testing for MSI and IHC: familial cancer service for a population of 400,000.



United Kingdom Genetics White Paper

The UK Department of Health has published a white paper that sees past the next election or change of minister for health, and provides for a service designed for the next 10 years (Anonymous 2003). The UK government invested £30 million into genetics services in 2001 and planned for an additional £50 million in 2003, aiming to: (a) Strengthen specialist genetics services (b) Build genetics into mainstream services (c) Spread genetics knowledge across the NHS (d) Generate new knowledge and applications and (e) Ensure public confidence. Additional funds will enable the immediate training or appointment of 30 new clinical geneticists, 50 new genetic counsellors and 90 new laboratory staff, plus training staff and resources, for a population of 59.6 million. The UK will have trouble filling all the new positions made available by this funding, and some migration of Australasian geneticists and genetic counsellors to the UK is possible.

A genetic counsellor recently moved to a position in the UK where she now has a full time position with a good salary and her own secretary. She previously had to work two jobs as the health service for 1.3 million people could only afford to employ her for 3 days per week, gave her no administrative support and had no clinical geneticist. After years of requests for funds to establish a genetics service in this area, and hours of business planning workshops for the future of genetics in NSW, the NSW government has recently funded a half-time clinical geneticist position for this 1.3 million people. This equates to 4% of what the UK has given an equivalent population in enhancement funding over the last 4 years, assuming £1 buys the same in the UK as $1 buys here. A brilliant cancer geneticist with FRACP and an Australian PhD is moving to the UK with her husband because she cannot find a job in her field.

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System Constraints, and Recommendations

Genetics services do not make a short-term profit for health services and many of the savings do not go to the local health service. Budgets are not indexed against cost-effectiveness, including the provision of laboratory tests. As laboratories recover costs and bill to cover service tests and the costs of accreditation, clinical genetics services that ordered the tests will have to pay the bills for public patients. Every diagnosis made by a clinical genetics service avoids unnecessary scanning, pathology testing or subspecialty diagnostic assessment for each relative at risk in a family. For example, the detection of a connexin 26 mutation ($150) in a blood spot of a baby found to have hearing loss on newborn hearing screening obviates CT or MRI scanning for inner ear malformation under general anaesthetic (Smith 2004). Although it is professionally satisfying to the genetics staff, the financial benefit of each diagnosis goes to the family and to health or community service providers other than clinical genetics. Screening procedures such as colonoscopy and mammography are demonstrated to be unnecessary for relatives shown by the genetics service not to have a high risk of inherited cancer. Medical and socioeconomic costs are also reduced for those shown to have a high genetic risk because their cancers are detected earlier. **Recommendation:** Funding plans for clinical genetics services should take account of their long term national benefit to society, not just their short term cost to one part of a health service.

More studies need to be done on the cost-benefit of general clinical genetics, although screening programs for individual diseases have been analysed. Screening of relatives in families with hereditary haemochromatosis allows presymptomatic detection and the prevention of disease by becoming a blood donor at a cost of $US3665 per year of life saved. Screening of the young male population might be cost-effective (Asberg, Tretli et al. 2002; Provenzale 2002) but the cost-benefit of screening the general population for a single gene has been confirmed. The cost-effectiveness of screening within families for genetic colorectal cancer has been demonstrated (Rozen and Ron 1989; Goldberg, Madden et al. 1998; Debniak, Kurzawski et al. 2000; Ramsey, Clarke et al. 2001). The cost of screening within families at high risk for breast and ovarian cancer was 753 Euros per year of life gained in Norway (Heimdal, Maehle et al. 1999) and 4294 Euros in Spain (Balmana, Sanz et al. 2004). On average, investigations performed to reach a new genetic diagnosis cost a NSW community genetics service $A350, varying from $240 per diagnosis for general genetics consultations, and $600 per cancer family gene identified to $680 per syndrome diagnosis (Hackett and Field 2004). Most of the children diagnosed to have a syndrome had much more than $680 worth of alternative specialist assessments, imaging or pathology tests, muck of which could be avoided by genetic assessment. One FTE genetic social worker managing 100 families with Huntington disease in a clinical genetics service with a total wages budget less than $1.5 million per year, saves the federal government $1.4 million per year by keeping 100 patients at home with their family carers for one extra year before being admitted for residential care, based on estimates of the cost of home care versus residential care (Anonymous 1999; Henwood 1999; Anonymous 2002). **Recommendation:** Budgets for clinical genetics services should be indexed to financial benefits they confer on a range of hospital, health, community and social services. A national database for the collection and analysis of cost-effectiveness data would address inequities.

Clinical genetics services improve efficiency by the use of multidisciplinary teams. Some revenue is generated by bulk-billing Medicare for privately referred patients seen by consultants, although each case is time-consuming as diagnoses have to be documented before reliable genetic counselling can be given, patients usually need several expert explanations of the options available to them as the concepts are usually not understood easily even by their referring doctors. Tests are usually not covered by Medicare. General clinical genetics or familial cancer services cannot be run effectively and equitably as private companies, they need the infrastructure and salaried multidisciplinary staff provided by public health services, which also have the most long term benefit to gain from their diagnoses and counselling. Medicare

items 110 and 116 and the multidisciplinary care items do not adequately reimburse services for the time spent on each family or the type of multidisciplinary care required. They are only valid for the patient referred to the service, whereas clinical genetics services have to manage whole families. Clinical genetics services are essential for this type of family screening in the current health system because private physicians can only bill Medicare for the relative referred to them. To provide equitable service, clinical genetics services usually have to see families near their homes because the families are often too poor to be able to travel to a capital city, and because more than one branch of a family often needs to be seen. **Recommendations:** Medicare item numbers or other funding appropriate to the intensive nature of assessment, large geographic areas covered, and the level of multidisciplinary care provided by clinical genetics services.

Telemedicine is a cost-effective way to see patients whose diagnoses are already made, especially those who have a family history of cancer. State government telemedicine grants have enabled outreach genetic counsellors to run clinics without a geneticist having to drive or fly to the clinic or stay overnight. Until a Medicare item is allocated for telegenetics consultation, clinical genetics services will lose potential revenue each time a patient is not seen face to face. Once the state grants expire, clinical genetics services or the areas they serve will have to pay for telemedicine. Costs of telegenetics will be reduced when communication channels can be transferred from telephone lines to secure broad band. **Recommendation:** permanent or long term Medicare or other funding for telemedicine.

Laboratories need to be staffed and funded so that they can offer a wider variety of tests. The only tests covered by Medicare are routine karyotyping, factor V Leiden, fragile X and haemochromatosis. Other tests such as FISH, CGH microarray and DNA mutation analysis cost clinical genetics services between $400 and $3000 each. Although most cancer DNA tests can be done in Australia, more than 50% of general genetics DNA testing costs are paid to overseas laboratories, and these could be done more cheaply in Australia if Medicare or other specific funding were allocated to different categories of laboratory investigation. These should be classified according to complexity, type of analysis (i.e. sequencing vs. PCR or MLPA) and need for expensive consumables rather than awarding a different Medicare item for each disease or gene, such as haemochromatosis, fragile X, etc. Funding for some complex tests should depend on their being ordered by qualified specialists who can give the appropriate counselling before and after the test is done. Clinical genetics services need to have laboratory testing budgets adequate for equitable access to crucial DNA tests for families who wish to avoid developing cancer or having severely intellectually or physically disabled children. The present rate of spending for laboratory testing, even though it is steadily increasing, reflects individual service budgets rather than demand and need. Some services have no budget for certain tests, so that testing is limited by socioeconomic status and ability of families to pay. At least one family had to sell their car to pay several hundred dollars for a karyotype requested by a private practitioner from a private laboratory. Special genetic tests are expensive in relation to common pathology items covered by Medicare, but they are now an indispensable part of preventive medicine. Their cost needs to be placed in perspective, most clinical genetics service testing budgets are dwarfed by the essential amounts spent on acute medicine and the care and investigation of chronic disease and ageing. **Recommendation:** appropriate Medicare or other funding scales for different classes of specialised genetic testing, and a goal that Australia should be self sufficient for common DNA tests.

Clinical genetics services have to obtain funding from their local health service to employ trainees. Several genetics services cannot afford to train clinical geneticists, or teach relevant clinical genetics to paediatric or adult medicine trainees. Unique opportunities for excellent training are being wasted, often in rural or regionally based outreach centres. Once a medical postgraduate trainee has been orientated to a genetics service they can do a lot of routine preliminary work in the genetics clinics, freeing the consultant for more complicated tasks and

costing about the same as a genetic counsellor. Directors of paediatric or adult medicine training programs are reluctant to release trainees to genetics services without salary compensation. Area health services are under pressure to hire trainees for acute services rather than genetics. Genetics services with trainee positions are tempted to use the trainee budget to keep their trainees on as staff specialists. Dedicated protected training positions need to be funded for all clinical genetics services, not only to train more clinical geneticists but also for advanced trainees in paediatrics, medicine, surgery and obstetrics and gynaecology who are interested in genetics. There are insufficient clinical genetics trainees being trained to succeed geneticists who are close to retirement age. **Recommendations**: funds for each clinical genetics service to have at least one extra clinical genetics trainee and one extra trainee visiting from another specialty eg medicine or paediatrics. Genetics services will need the resources to teach relevant clinical genetics topics to medical and other undergraduate health students.

There is no national or uniform state award for genetic counsellors. Current awards for allied health workers in many areas do not recognise the need for genetic counsellors to attend national conferences and travel for regular clinical genetics and counselling supervision. Some genetic counsellors have to share intake rosters with non-genetic services. Although some allied health staff are served well by generic continuing professional education programs offered by community health services, genetic counsellors are different to other allied health staff, and need regular contact with their counterparts in other regions or states. They need access to the internet in order to stay in touch with new tests, new syndromes and their patients' questions: many families know more about the genetics of their condition than their specialist or general practitioner. The low rates of pay and lack of opportunity for career advancement in some areas cause a high rate of burnout and turnover. **Recommendation:** national or state position descriptions and awards for genetic counsellors that provide for continuing education and research into genetic counselling.

Many Clinical Genetics Services are funded by their local area health service, but have historical obligations to provide services across regional or state boundaries. There is no satisfactory state-wide arrangement allowing funding to be coordinated across different regions of NSW. The state health department has devolved responsibility for this type of planning to individual area services without any specific funding. Some services have developed service level agreements but client services have insufficient funds to pay for professional services that were historically provided without charge. **Recommendation**: supra-regional funding for all clinical genetics services that provide services across regional or state or territory boundaries, state government support for the introduction of service level agreements.

Genetics services need sufficient budget to allow equity of access to genetic counselling and testing for the whole population. Genetics services have developed guidelines or protocols to ration limited funds for genetic testing as fairly as possible with priority for urgent circumstances or where most benefit will be gained from a test. There will never be Medicare item numbers for some of the rarer tests, and clinical genetics services will need to fill a gatekeeper role for testing. It is difficult to predict new diagnostic tests or their costs, but plans for the next 10 years will need to accommodate this uncertainty. Flexibility and a rapid response from laboratories and those who fund them will become more urgent when new treatments are developed for previously untreatable diseases and increase demand for genetic testing. The funds for these tests must be accompanied by appropriate funding for the infrastructure and staff of molecular genetics, cytogenetics, metabolic testing and newborn screening laboratories. Clinical and laboratory genetics are generally too labour-intensive and expensive to make a profit by billing Medicare and Medicare does not cover most laboratory tests. Billing patients is an unattractive concept in Australia, exacerbated by the high costs of most of the tests, and the tendency for genetic conditions such as intellectual disability or Huntington disease to make families poor. Some public hospital laboratory genetics services are managed by hospital pathology service business units, but these laboratories should more

appropriately be considered to be services rather than profit-making businesses. Although the assessments and tests can be expensive in the short term, they save resources for families and other areas of the health and social services, and go some way towards providing families with confidence in the future. **Recommendations:** government directive to all financial managers of specialist genetic clinical and laboratory services that financial plans should be appropriate for a service rather than a business unit. Financial planning for of laboratory services will most appropriately be linked to their contributions to "social capital" in addition to financial benefits to the community, although methods will need to be developed to assess "social capital".

There will not be enough clinical genetics service staff in Australia to meet demands of the next 10 years. The ACT, Northern Territory and Tasmania have no clinical geneticist although ACT and Tasmania are visited by geneticists. States with genetics services have very different staff to population ratios, and are mostly concentrated within teaching hospitals: see *Changing Benchmarks*, page 5. **Recommendations**: Although available data do not allow accurate prediction, it seems safe to predict that Australia will need at least one clinical geneticist per 200,000 population, 2-4 genetic counsellors per geneticist and 1.5 administrative staff per geneticist and appropriate testing budgets. This would need to increase if new treatments or tests became available for common genetic conditions. To achieve this over the next 10 years, budgets for employing new trainees and new trained staff will need to aim for this target as soon as possible.

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Asberg, A., S. Tretli, et al. (2002). "Benefit of population-based screening for phenotypic hemochromatosis in young men." Scand J Gastroenterol **37**(10): 1212-9.

BACKGROUND: Hereditary hemochromatosis (HH) is a common genetic disease leading to accumulation of iron in the body, most notably in the liver. More men than women become clinically ill. The prognosis is excellent if phlebotomy treatment is started before liver cirrhosis develops. Screening has been recommended, but the benefit of population-based screening has never been shown in a randomized clinical trial. In this article, we estimate the benefit of screening young men, using a theoretical model. METHODS: A phenotypic screening scenario was modelled using a decision tree. Gain of quality-adjusted life-years was used as a measure of benefit, and estimated using Markov processes. Data on the accuracy of the screening tests, the prevalence of HH and the risk of liver cirrhosis were mainly derived from a cross-sectional study on the prevalence and morbidity of HH in 30509 men. Data on the excess mortality of cirrhosis were taken from relevant literature. Sensitivity analysis was done for important variables. RESULTS: Assuming basal case values for variables, screening a cohort of

1000 men aged 30 years for phenotypic HH would gain about 8 quality-adjusted life-years, compared to awaiting symptomatic disease. Based on actual costs of our cross-sectional study, the screening cost was US$250 per quality-adjusted life-year gained. The prevalence of phenotypic HH, the excess mortality of liver cirrhosis, the quality of life in non-cirrhotic HH patients, and the fractions of patients compliant with treatment were the most important variables in the sensitivity analysis. CONCLUSION: Incorporating screening for phenotypic HH in health survey programmes for young men may be worthwhile.

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There is a consensus among medical geneticists that it is desirable to recontact patients as new information becomes available. Furthermore, some have suggested that there are legal arguments to support an obligation, creating a duty to recontact. Thus far much of the discussion among medical geneticists has focused on the practical concerns of implementing such a policy. However, we think that any such policy raises a number of important ethical concerns that must first be considered. Furthermore, there has not been a careful evaluation of the legal precedents that may reflect on a hypothetical duty to recontact. In this paper we first present an analysis of the scope of approaches and issues to be addressed in the development of ethical policy on this question. Secondly, we examine whether there is a legal obligation to recontact former patients about advances in genetics, as well as the legal implications if such a policy were to be adopted. Finally, we consider some of the functional and resource implications of adopting a policy of recontact. Our goal is to provide a framework for further discussion of this question and to stimulate further debate and research.

Provenzale, D. (2002). "Cost-effectiveness of screening the average-risk population for colorectal cancer." Gastrointest Endosc Clin N Am **12**(1): 93-109.

This article reviews several of the recent models addressing the cost-effectiveness of colorectal cancer screening in the average-risk individual (Table 1). How can clinicians and policy makers use this information for decision making regarding colorectal cancer screening? The cost-effectiveness ratios reported by themselves do not identify cost-effective practices. They must be placed in a decision context that is expressed in one of two forms. In the first form, an explicit threshold or maximum amount that a policy maker is willing to spend is stated (e.g., $40,000 per LY gained, as has been quoted as an acceptable amount for a prevention program). In the second form of decision context, a list of medical practices and their associated cost-effectiveness ratios, also known as a league table (Table 2) is used as a basis for comparison with the practice

under evaluation (e.g., colorectal cancer screening). The practice with the lowest cost-effectiveness ratio is the most cost-effective practice on the list. Practices with lower cost-effectiveness ratios are considered cost-effective compared with those with higher ratios. Table 2 lists incremental cost-effectiveness ratios for common medical practices. The models discussed in this article suggested that colorectal cancer screening using annual FOBT, flexible sigmoidoscopy at 3 or 5 years, the combination of FOBT and flexible sigmoidoscopy, barium enema, colonoscopy, and even virtual colonoscopy had incremental cost-effectiveness ratios ranging from $6300 to $92,900 per LY saved with most of the cost-effectiveness ratio ranging from $10,000 to $40,000 per LY saved. These ratios are similar to the cost of another widely accepted practice, breast cancer screening with annual mammography in women age 50 and older ($22,000 per LY gained). Colorectal cancer screening with any of the modalities discussed is considered less cost-effective than screening for hemochromatosis, which has an incremental cost-effectiveness ratio of $3665 per LY saved. Based on these ratios, however, screening for colorectal cancer is considered cost-effective compared with cervical cancer screening in women age 20 and older with Pap smear every 3 years, which has an incremental cost-effectiveness ratio of $250,000 per LY gained. The clinician can use these incremental cost-effectiveness ratios to evaluate the risks and benefits of alternative practices for the individual, and the policy maker with a limited health care budget can use these ratios to set priorities for funding based on the costs and the expected gains in life expectancy for colorectal cancer screening and for alternative health care programs.

Smith, R. J. (2004). "Clinical application of genetic testing for deafness." Am J Med Genet A **130**(1): 8-12.

Advances in the molecular biology of hearing and deafness have identified many genes essential for normal auditory function. Allele variants of these genes cause nonsyndromic deafness, making mutation screening a valuable test to unequivocally diagnose many different forms of inherited deafness. In this study, genetic testing of GJB2, SLC26A4 and WFS1 is reviewed.

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Submission by the Australasian Society of Genetic Counsellors (ASGC)

A. Background

Genetic counselling has been practiced in Australia since the 1970's but only more recently, progress has been made toward the professionalisation of non-medical "genetic counsellors" (GCs). In 1989 the Human Genetics Society of Australasia (HGSA) adopted a recommendation that a training and certification program be initiated, and the first HGSA Board of Censors (BOC), which is responsible for professional certification, was established [(1)]. Postgraduate courses in genetic counselling emerged in 1995 (University of Newcastle), followed by Griffith University, Melbourne University, and Charles Sturt University in 1996.

In 1995 the Australasian Society of Genetic Counsellors (ASGC) formed as a special interest group (SIG) of the HGSA, with the aim of representing the interests of the profession both within and externally to the society proper, and sharing the common interests in the training and education of GC's. The BOC recruits volunteer senior GC members from the SIG, as well as two clinical geneticist HGSA members who serve for a defined term. The ASGC currently has a membership of approximately 245, including student members, associate GCs (trainees) and fully certified GCs.

Genetic counsellors are, and their practice is, by definition "allied health" (i.e.. non-medical, non-nursing), although the (undergraduate) professional background of many genetic counsellors was previously nursing until the advent of postgraduate programs in the mid 1990's. An undergraduate science background is likely to (or already has) taken over from nursing as the commonest educational starting point of GCs. Genetic counsellors employed prior to 1995 and those employed in familial cancer genetics units however, may be more likely to have a nursing qualification.

The ASGC is currently in the planning stages of conducting a SIG funded professional survey. This survey (planned for 2006) will hopefully provide an insight into the educational backgrounds and employment of, and the opportunities, obstacles, and future directions for GCs in Australasia.

B. Terms of Reference

1. Regulatory and institutional factors

The genetic counselling profession is not currently regulated by statute. The certification process is overseen by the professional body (HGSA). There is, however, general interest within the SIG for pursuing statutory registration for the profession along the lines of the profession in the USA and strong support for statutory registration was indicated in the recent Australian Law Reform Commission (ALRC) of inquiry into the protection of human genetic information[(2)]. The USA has so far only achieved registration of the profession in a small number of states. The Association of Genetic Nurses and Counsellors (AGNC) in the UK is also pursuing statutory registration. A national registration system for GCs in Australia may be a more cost effective approach due to the small numbers of the profession. Statutory registration may provide GCs with enhanced autonomy and accountability whilst protecting the public from potential harm from unsuitably qualified individuals.

Within Australia, the majority of GCs are employed by state health authorities in centralized genetics units or area health services. Major genetics units are based in tertiary health facilities which may conduct "outreach" geneticist clinics in regional areas and may have locally based GC in a few regions. NSW is the only state that employs a significant number of GCs in primary health facilities such as community health centres or child and family health centres.

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These GCs are employed by the regional area health service to provide a genetic counselling service, which is augmented by visiting clinical geneticists from larger genetics units.

In all states of Australia, except NSW, GCs are employed under a single award, although these are not specific for GCs eg. Queensland employs GCs under the Professional Officer's award. In NSW, GCs are aligned to an award relating to their educational background. For example, the State nursing award for those GCs with a nursing qualification. In more recent years, GCs in NSW have been employed under the Health Education Officer's Award. Consequently, there has been great disparity between level of training and remuneration for GCs employed in NSW and potential disadvantage when moving between differing area health services. The differing state awards may potentially impinge upon mobility of GCs between the states due to lack of recognition of prior experience and lack of comparability.

There are only a small number of GCs employed by private health facilities (such as specialist obstetric ultrasonology services), and lack of statutory registration and access to clinical supervision may be a significant obstacle to employment in the private health sector.

Although there are postgraduate courses in genetic counselling at both a Master of Science and Graduate Diploma level, these qualifications serve only to qualify graduates for the first phase of professional GC training, as defined by the HGSA. The clinical practice required during training is a limiting factor to the number of applicants able to gain entry into the postgraduate courses due to the small number of suitable supervisors (fully certified GCs) available and the workload restraints inherent in their workplaces. In addition, for full HGSA certification as a genetic counsellor, substantial experiential learning and clinical supervision is required, which necessitates employment within a clinical genetics unit. The net outcome of this system is that around 10 to 40 individuals graduate each year with a postgraduate qualification in genetic counselling but they are unable to achieve full certification as GCs due to lack of employment opportunities and alternative mechanisms for supervision. A review of the tertiary genetic counselling courses and their relationship to certification may be needed to investigate this shortfall. Alternative supervision requirements and settings may provide opportunities for greater diversification of the profession. It is acknowledged that in comparison to our overseas counterparts, Australasian GC graduates are essentially less clinically qualified [(1)] and thus further away from achieving professional certification.

Although it is likely that some dissatisfaction with pay scales will be reported, the genetic counselling profession is considered appealing as evidenced by the high level of competition for employment positions and the numbers of applicants applying to the postgraduate courses. New permanent GC positions in the public sector are relatively uncommon, with most advertisements for locum or temporary employment, indicating a low attrition rate. The majority of GCs are women and conditions of employment generally support flexible working arrangements conducive to family life such as part-time employment and no shift or weekend work. Levels of GCs satisfaction with their employment will hopefully be gauged from the planned professional survey in 2006. In the USA, the average working life for a GC is only five years [(3)]. This does not appear to be the case in Australia as a significant number of GCs have been in employment for ten or more years.

Productivity of GCs may be influenced by employment environment. No Australian state currently employs GCs to the level recommended by the HGSA[(4)] (i.e.. one GC for population less than 150000). The majority of GCs work with medically trained clinical geneticists and there is a degree of role redundancy or overlap. Opportunities exist for GCs to become specialized in certain subspecialities of clinical genetics such as cancer genetics and prenatal genetic counselling. Autonomy of genetic counsellors may be inhibited by lack of training, lack

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of statutory registration, and unwillingness of clinical geneticists to delegate roles although the profession developed due to an acknowledged shortage of suitably qualified medical geneticists. In the UK, a comprehensive study of the role and practice of the genetic nurse was undertaken in response to perceived or actual opposition to their growing autonomy [5]. Workplace practices may also impact upon GCs such as the "drive" to enhance public service funding through the Medicare rebate scheme. As there is no rebate for consultation with a GC, patients who might otherwise more appropriately be seen by a GC only, may be preferentially assigned to the care of a medical geneticist.

2. Structure and distribution

The majority of GCs are located within tertiary or secondary health facilities. There great potential for integration of GCs within primary care facilities (as in the northern half of NSW) to improve access to services and enhance partnerships with general practitioners. There is opportunity for the existing workforce (nursing and allied health) in remote and rural health settings to gain genetics education and skills through distance tertiary education and telehealth supervision. This would enable remote health workers to conduct primary genetic health assessments and genetic counselling, and facilitate referral to a specialist genetics service only if necessary.
Although rare in practice, there is great potential for GCs to be employed in the private sector. Genetic counsellors could be employed by groups of general practitioners to conduct high risk assessment clinics, or by obstetric practices to conduct prenatal genetic counselling. These opportunities would be enhanced by the provision of provider numbers for GCs. In addition, the private pathology sector may benefit from employing a GC for liaison with consumers and medical practitioners regarding genetic testing. These areas of employment are currently in evidence in the USA although in small numbers only.

3. Demand for services

With the unravelling of the human genome there is great scope for increased demand for genetic counselling for conditions that previously had no identified genetic component and multifactorial conditions such as complex heart disease [5,7]. Emerging technologies in genetic testing and increased access to testing will also place an increased burden on an already stretched genetic counselling workforce [2]. It has been postulated that primary care providers will need to contribute to the delivery of genetics services in the future as it is recognized that clinical geneticists will be unable to meet the demands of the population alone, particularly in the wake of new genetic discoveries [8,9,10].

New models of care have been proposed to assist the delivery of genetics services at the primary care level and to support PCP's such as the introduction of the "genetics liaison nurse" or other "genetic associates" [8,11]. Trent *et al*., suggest that a genetic counsellor could provide this supporting role, but conclude that the financial infrastructure in Australia could not support such an initiative. However, no detailed industry assessment has been conducted in Australia to support this view.

In recent times the ASGC has advertised positions on behalf of employers and some interest has been derived from overseas trained GCs. Less than half of the ASGC memberships have full certification, resulting in limited applicant pools for senior genetic counselling vacancies. Exchange programs between Australian and international universities have been developed for GC university students and, in addition, a reciprocity agreement has been developed between the UK and Australia [1]. Reciprocity between Australian and the USA is more difficult to achieve due to the disparity between the respective styles of graduate programs, although the

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American Board of Genetic Counselling has established the International Counselor Eligibility Program [(1)].

Conclusions

Genetic counselling is an emerging allied health profession in Australia. Detailed analyses of future workforce requirements should be conducted including an assessment of the current training and certification program overseen by the HGSA and its relationship to the tertiary educational preparation of future GCs. The benefits and disadvantages of statutory regulation of the profession should be investigated. New models of delivering genetics services, such as genetic counsellors based in primary care, should be assessed with the aim of improving equitable access. In addition, barriers to increasing the number of GCs in the private health sector, such as lack of appropriate regulation and improved opportunity for reimbursement of services should be fully considered.

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Submission by the Australasian Society of Cytogeneticists

Although the field of molecular biology continues to expand within clinical genetics, laboratory geneticists who have an understanding of and diagnostic skills in both cytogenetics and molecular cytogenetics will be needed within the 10 year time frame being considered by the Commission. Cytogenetic referrals for prenatal diagnosis, constitutional analysis and cancer testing (in particular bone marrow karyotyping for leukaemia) have increased on average 11% since 1994 (Source HIC data See Table 2)

Table 2 Total referrals for cytogenetic analysis by state

	State									
	NSW	VIC	QLD	SA	WA	TAS	ACT	NT	Total	% Growth
Calendar Year										
1994	9,262	5,971	4,053	1,382	1,442	362	360	252	23,084	
1995	9,316	6,647	4,598	2,598	1,510	459	381	268	25,777	12%
1996	9,215	7,018	4,972	2,127	3,199	416	374	302	27,623	7%
1997	10,990	7,859	5,792	2,213	3,544	398	247	257	31,300	21%
1998	10,807	8,646	6,068	2,270	4,214	492	263	287	33,047	20%
1999	12,238	9,207	6,656	2,459	4,142	547	298	249	35,796	14%
2000	11,177	8,413	6,704	1,957	4,449	610	315	276	33,901	3%
2001	12,695	8,629	7,542	2,410	4,628	668	661	294	37,527	5%
2002	12,343	8,571	7,030	2,274	4,531	695	558	277	36,279	7%
2003	13,827	8,625	9,028	2,567	4,870	709	590	286	40,502	8%
2004	14,137	8,406	10,209	2,279	5,225	608	731	315	41,910	16%
									Average increase	11%

The NPAAC guidelines recommend a staffing ratio of 250 specimens per FTE. There is a nationwide shortage of scientists trained in cytogenetics, which partly reflects the poor status currently of science as a career path for young people. In recent years, heads of laboratories have frequently had to resort to recruiting overseas-trained scientists to fill staff vacancies.

There are a considerable number of senior cytogeneticists in charge of clinical cytogenetics laboratories, who will face retirement over the next few years and replacing them with adequately trained and experienced people will be difficult.

Factors affecting the available resource pool of scientists trained in cytogenetics/ molecular cytogenetics are as follows:

- Nearly all Cytogenetics training is performed within clinical laboratories: only a small number of clinical cytogenetics courses being offered within Australian universities. The training process within clinical cytogenetics laboratories is laborious, taking a minimum of 12 months to complete rudimentary training. Cytogenetics is a labour-intensive process, with most processes defying substantial automation.

- Qualifications in cytogenetics are gained through a certification process supervised by the Human Genetics Society of Australasia (HGSA) and are extremely rigorous. There are two levels of certification in cytogenetics.

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Eligibility to sit *Membership of the HGSA* (*MHGSA*) examinations requires a minimum of 3 years' working experience in cytogenetics laboratories. Candidates are required to submit extensively researched case studies of a publishable quality and subsequently to sit for an extremely challenging examination. There is no specific training scheme for examination candidates in Australia (as exists in the UK and USA), so commitment to this qualification procedure requires considerable self-motivation and dedication to self-education. There is currently no uniform recognition of these qualifications within State salary awards, and without this young scientists are lacking the incentive to commit to the examination process.

Eligibility to sit *Fellowship of the HGSA* (*FHGSA)* examinations requires a mandatory minimum of 10 years' working experience in cytogenetics laboratories. The process is similar to the Membership process, but a much higher level of expertise is required. Case reports of a publishable standard are submitted on a wide range of tissues. Examinations take place over two days, and currently the success rate is very low, requiring candidates to re-sit examinations the following year. If unsuccessful the second time, the process needs to be re-started with another series of case studies and examinations. This qualification requires an enormous dedication over a protracted period of time to the process by candidates. Again, as stated above, there is no specific training scheme for examination candidates in Australia and there is no currently no provision within the State Award for rewarding successful candidates, so many senior scientists are also opting not to sit for these qualifications. However, NPAAC has recently recognised Fellowship of HGSA as an eligible qualification for a senior scientist in charge of a lab, thereby providing at a national level some parity with a PhD and the basis for its recognition and appropriate remuneration in State awards.

Recommendations: improve availability of qualified Cytogenetics personnel by increasing funding for the expansion of courses in clinical cytogenetics within Australian universities and through promotion of uniform recognition of HGSA qualifications within State Salary Awards.

Cytogenetics is an area experiencing rapid technological expansion. Laboratories are expected to perform a wide range of molecular techniques to rapidly diagnose cytogenetic abnormalities in a wide range of situations: prenatally, in the newborn, adolescents, sub- fertile couples and in leukaemia and cancer. With rapidly increasing workloads and demands for faster turnaround times, cytogeneticists are experiencing reduced opportunities for continuing education and research. These factors have resulted in loss of job-satisfaction and the departure of scientists to other areas of genetics, such as genetic counseling and bioinformatics. An increasing emphasis on turnaround times of tests, particularly in prenatal testing, has also had a detrimental impact on the employment of trainee scientists as new employees are expected to be 'instantly useful' to carry out complex cytogenetic analysis.

Recommendations: recognise the rapidly evolving technologies within this field of laboratory genetics by increasing the funding for implementation of these technologies and the training of staff in these technologies. Provide funding for both traineeships and trainers within laboratory genetics.

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Submission by the Australasian Society for Inborn Errors of Metabolism

There is a clear and demonstrable shortage of consultant genetic metabolic physicians in all States. This is also a worldwide problem so Australia will not be able to address this by migration of overseas trained specialists. This is particularly a problem in the expanding area of adult metabolic patients as the success of treatment during childhood leads to more and more patients reaching adulthood but still requiring lifelong management. There is, to our knowledge, only one funded training place in Australia for clinical geneticists wishing to specialise in inherited metabolic disease. Unless this shortage is addressed we see the advances in diagnosis and treatment made over the past 20 years being compromised by the lack of suitably trained and funded medical staff.

Alongside the requirement for metabolic clinicians is a need for metabolic nurses. The roles performed by nurses specially trained in metabolic disease are vital to ensure optimum care. As expanded newborn screening continues to identify patients who are treated from birth to prevent the development of pathologies resulting from their condition, much of the task of providing an easily accessible source of advice for primary care givers falls to the metabolic nurse. There is also likely to be a steady growth in the number of patients receiving enzyme replacement therapy for lysosomal storage diseases, treatment which requires dedicated nursing staff familiar with the conditions and complications of the treatments.

The dietitian's own professional body will be making a submission separately but of particular concern for ASIEM is the fact that there are not enough dietitians to cover all the jobs with the metabolic patients being often very difficult to control and requiring a great depth of understanding in interpreting laboratory results. Although this is well understood in paediatric centres there is still competition for time dedicated to metabolic patients from the growing problems of juvenile diabetes and obesity. In adult centres the dietitians lack experience and exposure to metabolic disease and the long overdue development of adult services must include provision for and training of adult metabolic dietitians.

The laboratory scientists working in this field are also very highly specialised. There is less of a problem in recruiting junior scientists (more people with degrees in biomedical science than scientific officer positions in pathology in general) but retention of quality staff requires a recognition that availability of senior positions should not be determined by bed or test numbers but by the complexity of testing and interpretative skill required. All the centres in Australasia rely heavily on the experience and expertise of one or two individuals. Without the expansion in senior staff positions the very comprehensive training (under the HGSA board of censors) will be wasted as the lack of career progression forces many capable people to leave the profession.

Response by the
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Workforce for delivery of molecular genetics services

As a consequence of the ever expanding knowledge on the human genome, it is the area of molecular genetics service which will experience the greatest demand in growth. The delivery of molecular genetics testing is via university, public hospitals and a private pathology laboratories. The funding for genetics testing is limited to funding grants to hospitals and Medicare payment for only a single genetic syndrome (Fragile X syndrome) and 2 genetic based health problems (Factor V Leiden and haemochromatosis). The increasing importance of molecular genetic diagnosis for single gene traits and multifactorial disease will gorw the demand for trained molecular genetic scientists.

Two years ago the HGSA began its accreditation program for molecular geneticists. There are now accredited scientists at both the Fellow and Member level. A training program with assessment by examination is in preparation.