

Response to: **Green Paper: Delivering Safe and Sustainable Clinical Services**

From: **Genetic Services Planning Group**

Date: **February 2015**

As a result of the mapping of the human genome, and the recent advent of Massively Parallel DNA Sequencing, there has been an explosion in knowledge about the genetics of human disease. This has resulted in an unprecedented increase in the number of genetic tests available for diagnosing and predicting human diseases and this has been matched by unprecedented demand for genetic tests from both the public and from medical professionals. Over the last two decades, Clinical Genetics has grown from a discipline primarily concerned with birth abnormalities and paediatrics to become an important component of clinical care in disciplines such as cardiology, neurology, gastroenterology, and oncology, just to name a few. With the increased demand for genetic testing comes an increased demand for genetic expertise to interpret and communicate the test results.

The cost of sequencing *an entire human genome* using massively parallel sequencing (MPS) has rapidly reduced, now nearing \$1,000 per genome, down from an estimated cost of US\$2.7 billion in 2003 when the Human Genome Project published the first human genome sequence. Whole genome sequencing is poised to become a routine part of pathology testing, preparing the way for individually targeted, precise clinical patient management. In May 2013, the McKinsey Global Institute (Manyika, et al 2013) identified MPS as one of 12 technologies likely to transform the global economy. It estimated that, by 2025, the economic impact would be between \$US700 billion to \$US1.6 trillion per year, 80 per cent of which would be realised by extending and enhancing lives through faster disease detection, more precise diagnoses, and new drugs and treatments customised to both the patient and the disease.

In this context it is disappointing to see that the discipline of Clinical Genetics is mentioned only twice in the Government's draft document entitled "Tasmanian Role Delineation Framework" – and only as a service to which a Level 4 Maternity Service or a Level 6 Integrated Cancer Service should have access to.

Clinical Genetics Services in Tasmania are currently supplied by the Tasmanian Clinical Genetics Service (TCGS). This service is based at the Royal Hobart Hospital and is staffed by genetic counsellors (Allied Health Professionals). The services of Clinical Geneticists are contracted from inter-state and provided by a combination of fly-in clinics and telehealth clinics, totalling 57 days per annum. In responding to the steady increase in demand for Clinical Genetics services over the last two decades, the TCGS has already implemented many of the strategies highlighted in the Government's supplementary papers to the Green Paper (*Sustainability and the Tasmanian Health System*, and *Tasmania's Health Workforce*) such as:

- maintaining a single Statewide referral service based at the RHH, and providing monthly outreach services to the LGH and MCH rather than spreading the expertise thinly over multiple regions.

- contracting consultant Clinical Geneticists from interstate rather than employing a locally-based specialist who would have insufficient workload to maintain clinical skills in a setting of professional isolation.
- expanding the scope of practice of allied health professionals – for example in 1998/1999 100% of all patients seen by the TCGS were seen by a visiting consultant geneticist from interstate. With the recruitment of trained genetic counsellors, from interstate and training of local allied health professionals in genetic counselling, this figure had dropped to 33% by 2013/2014, with the majority of consultations now provided by locally based genetic counsellors.
- remaining generalist – in contrast to the practice of the larger mainland states where genetic counsellors sub-specialise into discreet areas of clinical genetics (e.g. paediatrics, or familial cancer) the genetic counsellors at the TCGS have remained generalists and can work across all sub-specialties of clinical genetics.
- the use of telehealth for Familial Cancer Clinics in particular to reduce the costs of services contracted from interstate.

By streamlining clinical practice using these and other strategies, the TCGS has managed to increase its annual caseload 9 fold in the 20 year period from 1994 to 2014, (from 100 outpatient consultations per year in 1994 to 900 outpatient consultations per year in 2014) with only a 3 fold increase in staffing over the same period. However, the recent staggering advances in genetic technologies and the consequent unprecedented increase in demand for Clinical Genetics Services means that the service as it stands is not sustainable into the future. The following three areas are flagged for urgent attention:

- **Workforce:** Staffing levels at TCGS are no longer sufficient to cope with current demand, let alone the inevitable increase. The ratio of genetic counsellors per head of population in Tasmania is lower than in other jurisdictions (See attachment 1). An increase in staffing is needed immediately otherwise waiting times, which already exceed 200 days, will inevitably exceed one year.
- **Pathology:** Genetic tests are expensive – they range in cost from \$200 to \$3,000. However the vast majority of genetic tests are not funded by Medicare, but are paid for from the State Health budgets – in Tasmania through the TCGS budget. Although the cost of genetic testing is slowly coming down due to improved technology, the number of genetic tests available, and the demand for those tests, is steadily rising. Currently, in Tasmania, doctors in private practice who wish to organise genetic testing for their patients routinely refer those patients to the TCGS, even in situations where they do not require the diagnostic expertise or genetic counselling offered by the TCGS, because the patient does not wish to or cannot afford to pay for the genetic test. Likewise, doctors working in the four major public hospitals in Tasmania who wish to organise a genetic test for a patient also routinely refer to the TCGS, even though they may have the diagnostic expertise to correctly order the test and interpret the result themselves, simply to shift the cost of the genetic test onto a service which has a budget for genetic testing. This cost-shifting from both public and private services is contributing to an unprecedented increase in

referrals received by the TCGS, increased waiting times, and a budget blow-out. What is needed is a state-wide approach to the triaging and funding of genetic tests.

- **Cytogenetics:** There is a single public cytogenetics laboratory based at the RHH with expertise and capacity to service the entire state (there are no private cytogenetics laboratories in Tasmania). This cytogenetics laboratory should be the central referral centre for all cytogenetic testing in Tasmania to keep costs down and to maximise the caseload of this laboratory. However current practice at the other three major public hospitals in Tasmania, including the LGH, is to send cytogenetic testing to mainland laboratories, despite the expense and time delays associated with sample shipment. This is a waste of local resources.

Whilst addressing the issues flagged above may require the commitment of additional funding, it should be noted that increased expenditure on clinical genetics can be expected to result in long term savings to other areas within the health service, and indeed for other government services. This is because:

- Genetics testing enable couples to clarify their risk of transmitting an inherited disease to their offspring and also to *avoid transmitting that disease*.
- Genetics testing for hereditary cancer syndromes enables individuals at high risk of particular cancers to take preventative action to avoid developing those cancers.
- Genetic testing for hereditary cancer syndromes identifies individuals from high risk families who have *not* inherited the cancer-causing gene, who can be spared unnecessary cancer screening procedures. 50% of all patients who have predictive testing for hereditary cancer syndromes at the TCGS fall into this category, so this constitutes a considerable cost saving for the health services that provide the screening (in particular endoscopy services).
- Whole genome sequencing promises to deliver new drugs and treatments customised to both the patient and the disease, resulting in vastly more effective treatment for common diseases.

With Tasmania currently lagging behind other Australian states in so many measures of healthcare, it is critical that we maximise the health outcomes of these new genetic technologies by ensuring that Tasmanians can access a well funded Clinical Genetics Service in a timely manner.

References:

Manyika, J., Chui, M. et al (2013). Disruptive technologies: Advances that will transform life, business, and the global economy. [Internet]. McKinsey Global Institute. Available from:

http://www.mckinsey.com/insights/business_technology/disruptive_technologies

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Attachment I: Comparison of staffing levels at TCGS compared with interstate Genetic services.

It is instructive to compare staffing levels at the TCGS with staffing levels at interstate Genetics Services. The only Australian jurisdiction with an identical service structure to Tasmania (i.e. a single state-wide service staffed by genetic counsellors, with clinical geneticists contracted in from a larger service interstate) is the Australian Capital Territory Genetics Service. All other states employ clinical geneticists, but the service models used differ. SA and WA are the most similar to Tasmania in that they each have a single, centralised, state-wide Genetics service. The larger states have multiple separate Genetics Services serving separate regions of the state, many of which are specialised to specific areas of genetics such as familial cancer or pregnancy. As a result staffing levels for these states are more difficult to obtain and compare).

	TCGS	ACT	SA	WA
FTE - Administration	0.6	1.0	8.2	7.5
FTE – Genetic Counsellors	2.7	4.2	11.5	12
FTE - Geneticists	-	-	7.5	5.5
FTE - total	3.3	5.2	27.2	25
Population served*	513,400	382,900	1,674,700	2,535,700
Total FTE per million population	6.4	13.5	16.2	9.85

* ABS data: population at end Sept quarter 2013

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