Australian Government
Department of Health and Ageing

SUBMISSION BY THE DEPARTMENT OF HEALTH AND AGEING

TO THE SENATE COMMUNITY AFFAIRS COMMITTEE

INQUIRY INTO GENE PATENTS

Introduction

The Department of Health and Ageing and portfolio agencies welcome the opportunity to make a submission to the Senate Community Affairs Committee Inquiry into Gene Patents.

Health practitioners, consumers, funders and managers are increasingly reliant on medical and scientific knowledge of the human genome and genetic factors for informed decision making, with genetic factors inherent in disease predisposition, manifestation, treatment and prognosis.

Genetics is a rapidly advancing scientific field with significant impact on individuals and the health care system. Genetic technologies are involved in the development of new diagnostic and therapeutic services. Increasingly, pharmaceuticals, vaccines and other biological therapies (often referred to as 'biologics') are being used for the targeted treatment of diseases such as cancer, rheumatoid arthritis, psoriasis, hepatitis and multiple sclerosis with other developments underway.

Numerous and complex issues surround genetic testing and services. The rate of discovery and the potential application of new knowledge calls for careful consideration of whether current ways of managing intellectual property claims, and associated diagnostic and treatment services, are congruent with community needs and values. Careful and prudent assessment of the issues and consequences are needed, within a framework that rigorously assesses the costs and benefits for individuals and society.

To date, the focus has been on diseases associated with single genes, but emerging trends suggest that many diseases are associated with changes in the expression of a number of genes, and thus complex genetic analyses are becoming increasingly required, providing added challenges to the regulatory frameworks underpinning these technologies. Genetic testing and related clinical services can have significant economic, psychological, social and family impacts that need to be appropriately managed with oversight by skilled clinicians and counsellors, supported by multi-disciplinary health care teams.

Genetic tests are commonly used for:

- *Preventative testing* - testing for conditions or risk factors for disease in asymptomatic individuals, that is, before there is clinical evidence of disease. This type of testing, or 'screening', can be aimed at either the individual, selected groups in the population or at a whole of population level. In Australia, only newborns are screened at the whole of population level for phenylketonuria, congenital hypothyroidism and cystic fibrosis.
- *Diagnostic testing* – used to establish a diagnosis in individuals displaying clinical symptoms or disease. For example, genetic testing is used to confirm Down's syndrome (the presence of Trisomy 21) or Fragile X syndrome in an individual who displays characteristics of the genetic disorder.
- *Testing to target treatment decisions (pharmacogenetics testing)* – used to enable informed decisions about effective clinical therapy. For example, women with breast cancer who test positive for the HER2 gene amplification can be expected to benefit from Herceptin.

This submission complements those provided by other government portfolios. It provides a brief overview of the key thematic health care policy issues related to the use of genetic technologies.

These policy themes relate to:

- affordable access to health care;
- research and development;
- safety and quality; and
- ethics and privacy.

Affordable access to health care

The Australian Law Reform Commission (ALRC) 2004 Report *Genes and Ingenuity: Gene Patenting and Human Health* found little evidence that gene patents and licensing practices with respect to genetic testing and clinical research and development have had any significant impact on the cost of health care in Australia. Since this Report, neither the Australian Health Ministers' Advisory Group on Human Gene Patents and Genetic Testing nor the National Health and Medical Research Council's (NHMRC) Human Genetics Advisory Committee has been advised of any systemic concerns about the impact of gene patents on the cost of health care.

Responses to genetic testing issues in Australia have been partly informed by specific events concerning affordable access to tests for the BRCA 1 & BRCA 2 genes, which are linked to breast and ovarian cancer. After initially seeking in 2002 to enforce its patent, Genetic Technologies Ltd (GTL) subsequently advised the Australian Stock Exchange in 2003 that it would not enforce its rights over the BRCA1 and BRCA2 patents. In June 2008, GTL again sought to enforce its rights by requiring others it considered to be using its patents for genetic testing to cease that testing. On 2 December 2008, GTL notified the Stock Exchange that it would not further seek to enforce its rights. The Department of Health and Ageing was involved in measures to address these issues.

The various current Australian, State and Territory government funding arrangements play a significant role in ensuring affordable access to health products such as pharmaceuticals, treatments, and genetic tests and services. States and Territories fund and provide the bulk of genetic testing and related clinical services. The Australian Government contributes to the funding of these genetic tests and services indirectly through the National Healthcare Agreements.

Affordable access to appropriately assessed health services is also directly supported through the Australian Government's Medicare Benefits Scheme (MBS), Pharmaceutical Benefits Scheme (PBS) and the Private Health Insurance rebate. Australia has a long established history and leads the world with a robust system that considers the evidence for funding of any new health-related tests, devices, treatments and medicines. MBS funding is provided for some genetic tests and the PBS funds those pharmaceuticals, vaccines and other treatments developed from genes, proteins and other related biological materials, assessed to be both effective and value for money.

On 18 December 2008, the Minister for Health and Ageing, the Hon Nicola Roxon MP, and the Minister for Finance and Deregulation, the Hon Lindsay Tanner MP, announced the Review of Health Technology Assessment (HTA Review) as a *Better Regulation Ministerial Partnership.* This Review will identify options for improving efficiency and reducing regulatory burden for Commonwealth HTA processes. It will consider ways to reduce impediments to medical innovation without compromising affordable patient access to health care that delivers improved health outcomes and value for money. One of the Terms of Reference of the Review - enhanced arrangements for assessment of co-dependent and hybrid technologies - will focus on issues around the assessment of pharmacogenetics testing, where a genetic test can usefully inform decisions about pharmaceutical treatments. The HTA Review is also expected to consider whether the current assessment framework is appropriately applied to other new and emerging genetic technologies.

Research and development

Australia has a longstanding and substantial commitment to genetics research, including investment in The International Cancer Genome Consortium. The NHMRC funded over $100 million for human genetics research in 2008. The NHMRC and the Innovation, Industry, Science and Research portfolio are both providing separate submissions to this Inquiry.

Access to genetic knowledge is critical for ongoing research and development of health applications. The patent system can provide useful incentives for investment in genetic research and development, by conferring a commercially valuable right on inventors of genetic technologies, which essentially provides a monopoly right for a period of years. The patents legislation also provides some mechanisms to help overcome the monopoly power which a patent confers on its owner, for instance by enabling a person to apply to a Court for a compulsory license to enable that person to use a particular technology which is patented, without fear of infringing the patent (s.133 *Patents Act 1990).*

As part of the Government's response to the Australian Law Reform Commission and Australian Health Ethics Committee's report *Essentially Yours – The Protection of Human Genetic Information,* the Commonwealth Government established an independent expert advisory body on human genetics as a principal committee of the NHMRC. The Human Genetics Advisory Committee provides advice on high level technical and strategic issues in human genetics, and on the social, ethical and legal implications of human genetics and related technologies.

A key policy issue is to ensure that patent protection, which is intended to encourage and disseminate research, does not impede access to basic health research, or render treatments founded on genetic technologies inaccessible by reason of cost to those who might require it. The June 2004 ALRC *Genes and Ingenuity* Report and the November 2005 Australian Government's Advisory Council Intellectual Property Report on *Patents and Experimental Use* recommended codifying the research use exemption. The Government is considering these recommendations.

Safety and quality

Ensuring the safety and quality of health services is of fundamental importance to governments and the community. This is achieved in multiple ways, including by workforce controls, rigorous evaluation and accreditation protocols, and a robust regulatory framework.

Health professionals most closely involved in genetic testing and services are clinical geneticists (specialist medical practitioners with a background usually in internal or paediatric medicine); genetic pathologists; geneticists (specialist medical laboratory scientists); and genetic counsellors.

Training and accreditation for the health care professions is a responsibility shared between the university sector and a range of professional bodies, such as the Australian Medical Council Ltd, specialist medical colleges, the nursing registration boards, and the Australian Psychology Accreditation Council Ltd. Specialist medical education is delivered by specialist colleges, faculties and chapters and is regularly reviewed. The Division of Paediatrics and Child Health in the Royal Australasian College of Physicians and the Royal College of Pathologists of Australasia are particularly involved in genetic testing and services.

The safety and quality of genetic testing is underpinned by the evaluation and validation of tests and treatments, accreditation of laboratories providing genetic tests and provision of appropriate clinical support and supervision in the clinical environments where testing is carried out.

The Therapeutic Goods Administration is currently developing a new classification and regulatory framework to ensure the quality of all therapeutic devices, including in vitro diagnostic kits used for genetic testing, and to reduce the risk of test kits producing unreliable results. The framework is being introduced to address concerns that many of these technologies are available on the Australian market with no regulatory oversight and no certainty that they perform as intended. Of key concern is genetic self-testing whereby people may order tests via the internet or direct from a provider, without essential information, counselling and support needed to deal with the results.

Laboratories accredited to conduct genetic tests that are funded through the MBS must meet the quality assurance standards of the Commonwealth/State National Pathology Accreditation Advisory Council. These include *Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection and Analysis, Classification of Human Genetic Testing* and *Requirements for the Development and Use of In-House In Vitro Diagnostic Devices*.

Ethics and privacy

Ethics and privacy policies are important concerns in the delivery of health services. The issues here are multiple, but include informed consent for research involving humans, privacy of patients, access to health information by insurance companies and possible discrimination on the basis of genetic status.

The guiding principle of the 2007 NHMRC *National Statement on Ethical Conduct in Human Research* is that a person's participation must be voluntary, and based on sufficient information and adequate understanding of the proposed research, including potential commercialisation. The *Statement* contains detailed requirements relating to human genetics covering research merit and integrity; justice in the use and disclosure of genetic information; beneficence; family and community involvement and confidentiality.

Traditional approaches to the management of privacy in health care have been challenged by the implications arising from genetic technologies, such as the right of a person to know about genetic information that may impact on his or her health. Since 2006, the *Privacy Act 1988* provides that medical practitioners and other health professionals can disclose genetic information to a patient's genetic relative without the patient's permission where necessary to lessen or prevent serious threat to life, health or safety of that person. Health practitioners are not however obliged to disclose information. Draft guidelines setting out a framework for such disclosures are currently being developed by the NHMRC.

The interaction of information made available by genetic technology with the operation of insurance law and practice also gives rise to some difficult issues of public policy. Currently, the position is that an insured person's duty of disclosure to his or her insurer includes an obligation to disclose knowledge which that person has acquired through genetic testing Moreover, insurers are not prevented from requesting family history and genetic testing results, from which they can make decisions about whether to insure individuals or not, and if so, upon what terms. However, IFSA Standard No. 11.00 provides that insurers are not to ask applicants for insurance to undertake genetic testing, nor are insurers to indirectly coerce applicants by using genetic tests as the basis for 'preferred risk underwriting' The area of insurance and disclosure is likely to yield further policy issues, and receive more public attention as medical science advances knowledge of the human genome and the prevalence of genetic testing increases.