The Productivity Commission into Disability Care and Support to establish a new national disability care and support scheme is welcomed.

This submission highlights issues that are relevant for people living with disabilities due to genetic conditions, as well as their carers, with a particular focus on access to genetic testing through Preimplantation Genetic Diagnosis (PGD) as a means of prevention of the condition in future generations.

Genetic Testing and Technologies

Genetics is a rapidly evolving discipline, with emerging technologies that enable the development of screening and diagnostic tests which can be used in new and innovative ways. These provide innovative and often cost effective alternatives to current practices, but also raise legal, ethical and social questions.

Genetic technologies and testing is complex. Once a genetic test has been developed for a particular genetic condition, it can be used for multiple purposes: diagnosis, screening, genetic carrier testing and risk assessment. Each of these purposes requires different levels of guidance by medical practitioners and genetic counsellors. The medical, ethical, social or psychological risks to the patient and or families need to be considered by the practitioner and counsellor. With increasing availability of testing off shore, where quality cannot be guaranteed, issues such as misinformation and misdiagnosis can have significant negative impact on health outcomes and decision-making. Thus, consideration of access and funding for genetic testing and technologies is important, especially:

- Access to new, safe and effective, tests and technologies
- Equity and ease of access to genetic testing and technologies across Australia
- Education for health professionals as well as the public, about new technologies and tests, to ensure appropriate usage of testing and technologies.

Medical Genetic Testing, Information for health professionals, April 2010 published by the NHMRC, provides an invaluable insight into the legal, ethical and social issues as well as the regulatory framework that governs genetic testing in Australia. A brief background of genetic testing in Australia is provided below, with links to appropriate websites.

Reproductive choices for Families with a Genetic Disability

Couples who have a high risk of having a child with a serious genetic condition that would lead to lifelong disability have the following options when wanting to have a child:

- Natural conception, which could lead to
    - miscarriage
    - undertaking prenatal diagnosis (where the gene or chromosomal variation can be tested for) and facing the possible decision to terminate an affected pregnancy
    - the birth of a healthy child
    - the birth of an affected child
- Remain childless
- Adopt a child. However, where the gene or chromosomal variation causes infertility, limitations on the ability for infertile couples to adopt have been documented (Billings P.R., et al 1992)
- Having in-vitro fertilization (IVF) using donor eggs or sperm
- Having IVF with Preimplantation Genetic Diagnosis (PGD) where the embryo is tested for the gene or chromosomal variation for which it is at risk and then implanted only if free from the condition

It is important to note that all these options have consequences that impact on the couple and society, both socially and financially. This document doesn't allow detailed discussion of these items however the key points are:

- Life with disability and its diversity is valued in society; however the impact of the disability on the individual and their carers can be significant emotionally, psychologically, physically and financially.
- For couples at high genetic risk , there can be repeated cycles of pregnancy > prenatal diagnosis > termination of pregnancy, before a child without the genetic condition is conceived.
- The effect of termination of pregnancy (TOP) because of a genetic variant has a significant psychological impact on the couple. (Savulescu L.J., 2008) (Royal Australian & New Zealand College of Obstretricians and Gynaecologists, 2005).
- Adoption in Australia is complex and lengthy. In 2004-5, there were only 585 adoptions in Australia (Adoptions Australia 2004-05).

PGD is defined as: *screening of cells from preimplantation embryos for the detection of genetic and/or chromosomal conditions before embryo transfer*. This technique is an addition to the IVF process, to detect whether an embryo created in vitro has a specific genetic variant that will give rise to a serious condition. Embryos without the specific variant will be transferred to the patient while those excess to immediate needs can be cryofrozen for transfer at a later time. Success rates have improved significantly over the years, with Sydney IVF having 24.4 live births per 100 PGD cycles. In 2008 there were 971 PGD cycles completed by IVF clinics in Australia and New Zealand. (Wang Y.A. et al, 2008)

PGD can provide couples at risk of having a child with a genetic or chromosomal condition with the opportunity of having an unaffected child or avoiding multiple miscarriages. In many cases the couple already has a child or a family member with the genetic condition. The strongest drivers for choosing PGD are if the couple have an affected child or have had a previous termination for the genetic condition.

PGD therefore increases the couple's options and is a safe and well recognized method of conception; however, it is costly. The costs can play a significant role in the couple's decision-making and psychological wellbeing throughout the procedure. (Karatas J.C. et al, 2010). An additional barrier for couples in accessing this technology appropriately is the lack of referral by medical practitioners. This highlights the need for education in this area. (Karatas J.C. et al, 2010)

In Australia, PGD is available only in the private setting. It costs approximately $12,000 to $15,000 for the initial IVF with a PGD cycle. Subsequent cycle costs are less because the test is already developed. For families already facing increased financial strain in caring for a child with a profound or severe core function disability, this cost is significant. MBS does not currently provide rebates for PGD or the IVF costs associated with it. Funding PGD is complex, given the broad range of rare genetic conditions covered by PGD and the combination of IVF, genetic testing and technology involved. There are over 150 different conditions that PGD has been used for in Australia and structuring a system, which could support funding for technologies such as PGD, would be financially beneficial for the Australian health system.

Conclusion

We would appreciate it if the Productivity Commission would consider that education and equity of access are essential for appropriate and optimal use technologies such as PGD, which have a small but potentially valuable role to play in:

- enabling families, with a history of genetic conditions leading to lifelong disability, to have healthy children;
- the prevention of long-term disabling conditions which have high financial impact on the individual families and carers, and the Australian health system.

Funding technologies like PGD for the prevention of rare lifelong disabling genetic conditions would be financially beneficial to Australia as well as providing significant support to families coping with genetic abnormalities.

This submission is supported by:

Centre for Genetics Education

NSW HEALTH

Professor Kristine Barlow-Stewart
Director
Centre for Genetics Education, Royal North Shore Hospital

Date

SYDNEY IVF

Dr Kylie De Boer
Group General Manager
Sydney IVF Ltd

Date

MUSCULAR DYSTROPHY FOUNDATION • AUSTRALIA

JERRY LEWIS
International Patron

David Jack
Chief Executive Officer
Muscular Dystrophy Foundation

Date

The Fragile X Association of Australia

Doug Rodgers
Treasurer
Fragile X Association of Australia

Date

Thalassaemia Society of NSW

Nancy Lucich
Co-ordinator of Thalassaemia Society of NSW Centre
Thalassaemia Society of NSW

Date

BACKGROUND

Medical, Ethical, Social and Legal

The **National Health and Medical Research Council (NHMRC**) is Australia's peak body for providing guidance for health and medical research. The **Human Genetics Advisory Committee (HGAC)** is part of the NHMRC, and provides advice on medical, ethical, social, and legal aspects of human genetics and related genetic technologies. The HGAC provides national guidance in response to new technologies and advances in genetic health practices. ***Medical Genetic Testing, Information for Health Professionals, April 2010*** published by the NHMRC provides guidance on genetic testing from a technical, ethical, legal and social perspective. This resource is written for the health professional to help provide patients and families with appropriate guidance. http://www.nhmrc.gov.au/publications/synopses/e99syn.htm
Human Genetics Advisory Committee (HGAC) | National Health and Medical Research Council

Pathology

Pathology laboratories that do tests which are covered by Medicare are required to be accredited by the National Association of Testing Authorities (NATA) in Australia according to AS ISO 15189:2009 guidelines. National Pathology Accreditation Advisory Council (NPAAC) which is managed by the Australian Government Department of Health and Ageing published *Requirements for Pathology Laboratories (Revised document) 2007*. NPAAC sets standards for medical testing laboratories in Australia. According to the requirement there are two levels of genetic testing:

1) *Level 1 genetic tests carry similar risks (and require similar consent) as other medical tests. These are typically diagnostic tests, medical screening tests, or tests of cancer tissue (i.e. somatic cell testing) in which the genetic test is just one component of an accepted medical testing process.*
2) *Level 2 genetic tests are those associated with interpretive, ethical, or consent issues that are peculiar to the identification of a heritable mutation in an apparently unaffected person. These are typically predictive genetic tests. According to the NPAAC Standard, a Level 2 test requires specialist knowledge for the test to be requested and professional genetic counselling to precede and accompany the test.*

http://www.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-pathlabs.htm
Currently there is no requirement for pathology type laboratories to have additional accreditation if they do not perform testing which attracts a Medicare rebate. Except for Fragile X syndrome there is no Medicare rebate for testing of a specific genetic condition, thus laboratories performing these tests do not have to be NATA accredited. The laboratories are therefore self-regulating.

IVF

PGD is governed by Fertility Society of Australia's, Code of practice for Assisted Reproductive Technology Units. (Fertility Society of Australia, 2010)

> **Assisted reproductive Technology (ART**) *involves clinical treatments and laboratory procedures that include the handling of human oocytes, sperm or embryos. This includes IVF; gamete intrafallopian transfer; zygote intrafallopian transfer; intracytoplasmic sperm injection; embryo or gamete cryopreservation; oocyte, semen or embryo donation; blastomere biopsy for Preimplantation Genetic Diagnosis; gestational surrogacy and intrauterine insemination (IUI).*

The code also specifies that the ART facility must maintain compliance with the NHMRC Ethical Guidelines on the use of ART in clinical practice and research (2007) or New Zealand equivalent, except where specific alternate policies have been directed by a registered HREC affiliated to the Unit. http://www.fertilitysociety.com.au/

Funding

Medical Genetic Testing, Information for health professionals, April 2010 states that "The jurisdiction-based approach to funding genetic tests has led to inconsistent practices regarding who can order genetic tests. In some jurisdictions, certain genetic tests can only be ordered by specialist clinical geneticists employed in the public sector." Very few genetic tests are funded through the MBS and others are available through the Genetic departments in public hospitals, however the States and territories all have separate policies.
http://www9.health.gov.au/mbs/search.cfm?cat1=147&cat2=156&cat3=&adv=
http://www.health.nsw.gov.au/policies/PD/2005/pdf/PD2005_335.pdf

Education

There are many **Australian Education resources** available, here are two key resources:

- The Centre of Genetics education which provide a range of publications and fact sheets for health professionals and the public. Fact Sheets - Centre for Genetics Education
- Genetics in Family Medicine: The Australian Handbook for General Practitioners. http://www.nhmrc.gov.au/your_health/egenetics/practitioners/gems.htm

BIBLIOGRAPHY

Australian Institute of Health and Welfare (AIHW) 2005.
Adoptions Australia 2004–05.
AIHW cat. no. CSW 25. Canberra: AIHW.

Assisted Reproductive Technologies Review Committee. (2006).
Report of the Independent Review of the Assisted Reproductive Technologies.
Canberra: Assisted Reproductive Technologies Review Committee.

Australian Institute of Health and Welfare National Perinatal Statistics Unit. (2007).
Assisted Reproductive Technologies in Australia and New Zealand.
AIHW, Australian Institute of Health and Welfare National Perinatal Statistics Unit. Sydney: AIHW.

P.R. Billings, M.A. Kohn, M. de Cuevas, J. Beckwith, J.S. Alper, and M.R. Natowicz
Discrimination as a Consequence of Genetic Testing
Am. J. Hum. Genet. 50:476-482, 1992

L.J. de Crespigny and J. Savulescu
Pregnant women with fetal abnormalities: the forgotten people in the abortion debate
MJA Volume 188 Number 2 21 January 2008

Genetics Education in Medicine (GEM) Consortium. (2007).
Genetics in Family Medicine:The Australian Handbook for General Practitioners.
Sydney: Commonwealth of Australia 2007.

J. C. Karatas, K. Barlow-Stewart, K. A. Strong, B. Meiser, C. McMahon and C. Roberts
Women's experience of pre-implantation genetic diagnosis: a qualitative study
Prenat Diagn (2010) Published online in Wiley InterScience (www.interscience.wiley.com) DOI: 10.1002/pd.2542

J.C. Karatas, K. Barlow-Stewart, B. Meiser, C. McMahon, K.A. Strong, W. Hill, C. Roberts, and P. Kelly
Psychological adjustment, knowledge and unmet information needs in women undergoing PGD
Advanced Access publication on April 10, 2010 doi:10.1093/humrep/deq086
Human Reproduction, Vol.25, No.6 pp. 1481–1489, 2010

NHMRC (2007).
Ethical guidelines on the use of assisted reproductive technology in clinical practice and research.

Reproductive Technology Accreditation Committee. (2008).
Code of Practice for Assisted Reproductive Technology Units.
Melbourne: Fertility Society Of Australia.

Royal Australian & New Zealand College of Obstretricians and Gynaecologists. (2005).
Termination of Pregnancy; A resource for medical professionals.

L. J Savulescu (2008)
Pregnant women with fetal abnormalities:the forgotten people in the abortion debate.
Medical Journal of Australia , Volume 188 Number 2.

Sydney IVF Ltd Internal data. (2007). Sydney IVF.

Wang YA, Chambers GM, & Sullivan EA 2010.
Assisted reproductive technology in Australia and New Zealand 2008.
Assisted reproduction technology series no. 14. Cat. no. PER 49. Canberra: AIHW.

ABBREVIATIONS

AHEC	Australian Health Ethics Committee
AHTAC	Australian Health Technology Advisory Committee
AIHW	Australian Institute of Health and Welfare
ANZARD	Australian and New Zealand Assisted Reproduction Database
ART	Assisted Reproductive Technology
HGAC	Human Genetics Advisory Committee
HREC	Human Research Ethic Committee
IVF	*In Vitro* Fertilisation
IUI	Intrauterine Insemination
MBS	Medicare Benefits Schedule
MSAC	Medical Service Advisory Committee
NATA	National Association of Testing Authorities
NHMRC	National Health and Medical Research Council
PBS	Pharmaceutical Benefits Scheme
PGD	Preimplantation Genetic Diagnosis
RTAC	Reproductive Technology Accreditation Committee (Fertility Society of Australia)