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The Program Manager
Reproductive Technology Unit
Patient Safety & Clinical Quality
Clinical Excellence Division
Department of Health
189 Royal Street
PERTH WA 6004

16 March 2018

Dear Sir/Madam,

I am writing to provide a submission into the Review of the Western Australian Human Reproductive Technology Act 1991. This submission has been written on behalf of the Australian Mitochondrial Disease Foundation (AMDF). The AMDF supports mitochondrial disease (mito) sufferers and their families, funds essential research into the prevention, diagnosis, treatment and cures of mitochondrial disorders, and increases awareness and education about these devastating diseases. As such, I am responding to this Review only in relation to the *Human Reproductive Technology Act 1991* Terms of Reference regarding new technologies, in particular the techniques known as mitochondrial donation, and provide information below as to the AMDF's support for the introduction of mitochondrial donation in Australia.

Mitochondrial disease

One in 200 people may develop mito during their lifetime. In about half of cases, the disease is caused by changes in mitochondrial genes (mtDNA), which contribute about 0.1 per cent of a child's genetic make-up and are inherited only from the mother.

Mito is a debilitating genetic disorder that starves the body's cells of energy, causing multiple organ dysfunction or failure and potentially death. It primarily affects the muscles and major organs such as the brain, heart, liver, inner ears and eyes, but can cause any symptom in any organ at any age.

Depending on which parts of their bodies are affected and to what degree, sufferers may: have strokes or seizures; be unable to walk, eat, swallow or talk normally; develop liver disease or diabetes; suffer heart, respiratory or digestive problems; lose their sight or hearing; suffer muscle weakness and pain; and/or experience developmental delays or intellectual disability. There are few effective treatments and no cures for mito.

Reproductive options currently available

Currently, prenatal diagnosis or in vitro fertilisation (IVF) using preimplantation genetic diagnosis (PGD) are the only reproductive options available to prospective Australian parents who are at risk of passing on mito and want to have a healthy, genetically-related child.

However, these techniques are not an option where the exact gene involved is unknown or where most of the woman's eggs may carry substantial amounts of an mtDNA change, such as in maternally inheritable mtDNA disease. Also, PGD can only reduce and does not eliminate the risk of mito in the resulting child.

Mitochondrial donation

Mitochondrial donation (also called mitochondrial replacement, transplant or transfer) involves transferring nuclear genetic material from the affected mother's egg into a donor egg that has had its nuclear DNA removed and retains only its healthy mitochondrial DNA. The resulting child therefore does not inherit mito.

It has been estimated the average number of births per year among women at risk for transmitting mtDNA disease is 152 in the United Kingdom and 778 in the United States. Assuming roughly equal age distribution and fertility, this conservatively equates to approximately 56 Australian babies who could be born each year free from maternally inheritable mito. These children could, instead of suffering from a dreadful and, ultimately, deadly disease, live 'normal' lives free from mito.

Support for mitochondrial donation

AMDF supports mitochondrial donation being available under strictly controlled, tightly regulated conditions to women at risk for having children with severe forms of mtDNA disease that could lead to a child's early death or substantial impairment. The Foundation has organised over 50 meetings between members of the mito community and federal and state MPs and stakeholders. We are also in discussions with the National Health and Medical Research Council (NHMRC) regarding emerging technologies in this area and consideration of the *Prohibition of Human Cloning for Reproduction Act 2002* and the *Research Involving Human Embryos Act 2002* in light of these.

AMDF is committed to expanding the reproductive options available to Australian women and couples affected by mito, as has been done in the United Kingdom.

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Overseas experience of introducing mitochondrial donation

The United Kingdom recently established the world's first regulated system to provide mitochondrial donation.

Following an extensive scientific and ethical review process involving ten years of public consultation and three expert reports, in October 2015 the UK Parliament approved regulations to allow mitochondrial donation to prevent maternally inheritable mito. In December 2016, the regulations were endorsed by the Human Fertilisation and Embryology Authority (HFEA).

UK clinics apply to the HFEA for a special licence to provide mitochondrial donation. Patients also apply individually to the HFEA to undergo mitochondrial donation treatment in a licensed clinic. In March 2017, the HFEA granted the first clinical mitochondrial donation licence to the Newcastle Fertility Centre at the International Centre for Life in Newcastle-upon-Tyne, United Kingdom, whose reproductive and genetics experts were instrumental in developing the procedure. On 6 February 2018, two UK women carrying mtDNA changes were granted permission to undergo mitochondrial donation, giving them the opportunity to have children free of mito.

Current legislative environment in Australia

In Australia, current legislation does not allow the use of mitochondrial donation techniques in the clinic, and research is significantly restricted. Research and clinical applications of mitochondrial donation are overseen by federal and state governments; state laws are, for the most part, consistent with federal law. In all states except Western Australia, research on a limited range of mitochondrial donation is permissible up to day 14 of embryo development, subject to a licence being granted.

No treatment can claim to be 100 per cent safe and effective and there are risks and benefits with any medical technique, including "traditional" IVF. Risks with mitochondrial donation procedures are expected to be low and comparable to the risk for any couple of having a child with a severe genetic condition; the latter is about three per cent in the general population.

Mitochondrial donation has been shown to be safe and effective in producing monkeys whose mtDNA has been almost completely replaced by donor mtDNA. Experiments in very early human embryos suggest the techniques allow normal embryo development. AMDF supports selecting a mitochondrial donor with the same ancestral mtDNA background, to increase the likelihood of a successful pregnancy and a healthy child.

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Offering parents choice

AMDF values people affected by mito. While AMDF's ultimate vision is to cure mito, its mission is to support sufferers and their families, fund research into mito, and educate the general public and the medical profession. In advocating for techniques to prevent children being born with the disease, AMDF supports the rights of prospective parents to choose to have healthy biological children who will not suffer the debilitating, disabling and potentially fatal consequences of severe forms of mito.

AMDF believes the choice to utilise mitochondrial donation should ultimately be made by the affected woman or couple. They should be supported to make informed reproductive choices based on clearly understanding the relevant issues.

AMDF calls upon the Western Australian Government to acknowledge the significant developments and advances in mitochondrial donation techniques, and to revise its legislation to allow affected women the choice to access mitochondrial donation in Western Australian clinics.

Yours faithfully,

Sean Murray
Chief Executive Officer

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