AUSTRALIAN MITOCHONDRIAL DISEASE FOUNDATION

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Disease-free babies with donor DNA on the way

UK experts granted world-first licence for mitochondrial donation IVF

Eminent UK reproductive and genetics experts have become the first in the world licensed to provide a revolutionary IVF procedure using donor DNA – which they spent decades developing – enabling women carrying potentially fatal mitochondrial disease to have healthy, genetically-related babies.

Mitochondrial donation to prevent mitochondrial disease has been permitted in the UK since pioneering legislative changes in 2015 and endorsement in 2016 by the Human Fertilisation and Embryology Authority (HFEA), establishing the world's first regulated system to provide mitochondrial donation.

Today 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.

Australian Mitochondrial Disease Foundation (AMDF) CEO Sean Murray applauded the licensing as a major step forward for the mitochondrial disease community worldwide, and congratulated Mary Herbert, Professor of Reproductive Biology at the Institute of Genetic Medicine, and her team.

"Professor Herbert's work developing and refining mitochondrial donation has been widely published and is internationally-renowned, along with colleague Professor Sir Doug Turnbull, director of the Wellcome Trust Centre for Mitochondrial Research at the Institute of Neuroscience," Mr Murray said.

"It's very exciting that women can now access mitochondrial donation through licensed, reputable clinicians in the UK who are world experts in mitochondrial disease and reproductive technologies.

"The UK Government and regulators have undertaken a rigorous and comprehensive global scientific and ethical review of the treatment over a ten-year period.

"Each painstaking step has added to the evidence the AMDF expects the Australian Government to consider in changing our laws to make mitochondrial donation available to Australian women," he said.

Mitochondrial donation in the UK is restricted to women at risk of having a baby suffering severe mitochondrial disease, a debilitating genetic disorder that starves the body's cells of energy, impairing major organs like the brain, heart, liver, muscles, ears and eyes.

The disease has few treatments and no cure and can cause any symptom in any organ at any age.

The procedure is subject to numerous safeguards – overseen by the HFEA – such as carefully selecting women to undergo the procedure as a clinical risk reduction treatment, providing full information about potential limitations and risk, undertaking genetic testing when the embryo is at 15-weeks' gestation, and closely monitoring the outcomes over time.

"Mitochondrial donation could prevent at least 60 Australian babies each year from suffering a severely disabling and life-threatening form of mitochondrial disease," Mr Murray said.

"The Australian Mitochondrial Disease Foundation – and local families affected by mitochondrial disease – looks forward to the Australian Government following the UK's lead and amending our laws to give families here the choice to access mitochondrial donation to have healthy biological children."

Meanwhile, the first live birth of an apparently disease-free baby to a woman at risk of passing mitochondrial disease to her child was announced in September 2016 by US scientists who carried out the procedure in Mexico, where laws do not preclude it.

However, although the AMDF cautiously welcomed the news, Mr Murray said there is concern about the lack of published information, regulation or independent monitoring.

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"While this appears to be a promising development demonstrating a successful outcome for mitochondrial donation to prevent mitochondrial disease, the AMDF's Scientific and Medical Advisory Panel looks forward to details being published in a peer-reviewed journal so we can closely assess the matter," Mr Murray said.

Ukrainian scientists also announced the birth in January 2017 of a baby with donor mitochondrial DNA to a woman with unexplained infertility, but without mitochondrial disease, who had been unable to conceive with conventional IVF. Details have not been published to date.

"The AMDF supports making mitochondrial donation techniques available to women at risk for having children with severe forms of mitochondrial disease that could lead to a child's early death or substantial impairment," he said.

"We also support regulation and strict oversight of clinics offering the procedure and recognise it will be important to monitor outcomes closely, as it would be with any new IVF technique.

"The AMDF does not currently support mitochondrial donation to treat infertility. As far as we are aware, its use for purposes other than preventing inheritable disease has not been subject to rigorous scientific and ethical review or research, as has been the case for preventing mitochondrial disease."

Mitochondrial donation 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 mother's and father's nuclear DNA contributes more than 20,000 genes or 99.9 per cent of the baby's genetic make-up and determines its appearance, intelligence, behaviour and other personal characteristics.

The 0.1 per cent contribution (37 genes) from the donor egg means the resulting baby's cells can effectively convert food and oxygen into the energy needed to power its organs.

Mitochondria are the powerhouses of our cells that generate 90 per cent of the energy fuelling our bodies, particularly muscles and major organs. Depending on which parts of their bodies are most affected and to what extent, people with mito can lose their sight or hearing, be unable to walk, eat or talk normally, have strokes or seizures, develop liver disease or diabetes, suffer cardiac, respiratory or digestive problems, or experience developmental delays or intellectual disability.

More than 1 in 200 Australians – at least 120,000 people – have genetic mutations that predispose their mitochondria to fail early, and may develop mitochondrial disease sometime in their lives. Many people are symptomatic but undiagnosed or misdiagnosed, some are not yet symptomatic, and others are unknowingly at risk of passing the disease to their unborn children.

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- Prof David Thorburn, Mitochondrial Research Group, Murdoch Childrens Research Institute (MCRI)
- Prof Carolyn Sue, Kolling Institute of Medical Research
- Prof John Christodoulou, Neurodevelopmental Genomics Research Group, MCRI
- Assoc Prof Ainsley Newson, Centre for Values, Ethics and the Law in Medicine, University of Sydney
- Australians affected by mitochondrial disease

Resources:

- AMDF Position Statement on Mitochondrial Donation: www.amdf.org.au/mitochondrial-donation/
- Profile of Professor Mary Herbert: www.ncl.ac.uk/igm/staff/profile/mary.herbert#tab_research
- HFEA statement on mitochondrial donation licence: www.hfea.gov.uk/10635.html
- A video primer on mitochondrial donation: www.nature.com/nrdp/animations/mito-dis-16
- **AMDF Media Resources Dropbox** (releases, graphics, background): http://tinyurl.com/n9w963c