AUSTRALIAN
MITOCHONDRIAL
DISEASE FOUNDATION

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UK report adds weight to calls for Australian government to overhaul laws so babies won't inherit severe genetic disorder

Women at risk of passing a life-threatening genetic disorder to their babies are a step closer to having healthy biological children following a UK report recommending IVF using donor DNA to prevent severe mitochondrial disease, which affects at least 60 Australian babies born each year.

The Australian Mitochondrial Disease Foundation (AMDF), which advocates for patients and funds research, has welcomed the 30 November report on mitochondrial donation by an independent expert panel convened by the UK Human Fertilisation and Embryology Authority (HFEA).

Mitochondrial donation – where an affected woman's faulty mitochondrial DNA is replaced by healthy mitochondrial DNA from a donor egg – has been permitted in the United Kingdom since legislative changes in October 2015 enabled the HFEA to offer licences to IVF clinics.

The report recommends mitochondrial donation be approved for "cautious" use in "specific circumstances", based on a comprehensive global review of the latest scientific developments and input from international experts including Australian researchers.

AMDF CEO Sean Murray welcomed the expert report as an important step towards mitochondrial donation being available to affected women who would otherwise have no options to have a healthy biological child who won't suffer the consequences of severe mitochondrial disease.

"It's exciting that mitochondrial donation could be available to women in the UK in 2017, and the first babies born later next year, if the HFEA board agrees to the panel's recommendations at its 15 December meeting," Mr Murray said.

"This development adds weight to the Australian Mitochondrial Disease Foundation's call for the federal government to amend legislation so that affected Australian women can also have the chance to have healthy babies.

"Based on the extensive evidence available, the Australian Mitochondrial Disease Foundation believes the potential benefits of mitochondrial replacement outweigh the risks for unborn children who would otherwise almost certainly develop potentially deadly mitochondrial disease.

"The Australian Mitochondrial Disease Foundation supports making this ground-breaking treatment available for in-clinic use under certain specific conditions and strict regulation," he said.

At least one Australian child born each week – or 62 children every year – will develop a severe or life-threatening form of mitochondrial disease, and half will die in childhood. A further 30 Australian children born each week – or 1540 every year – are at risk for developing a mild to moderately disabling form of mitochondrial disease during their lifetime.

Professor David Thorburn is Head of Mitochondrial Research at Murdoch Childrens Research Institute and a member of the AMDF Scientific and Medical Advisory Panel and the AMDF Mitochondrial Donation Working Group, and made a submission to the independent panel in favour of in-clinic mitochondrial donation.

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"Research evidence indicates mitochondrial donation techniques are sufficiently developed and safe for in-clinic use, subject to specific conditions defined by the UK legislation and the expert report recommendations," Professor Thorburn said.

"The report to the HFEA concludes that recent scientific advances have sufficiently addressed the potential carry-over of faulty mitochondrial DNA.

"It also recommends numerous safeguards such as carefully selecting women to undergo the procedure as a clinical risk reduction treatment, providing full information about potential limitations and risk, and undertaking genetic testing when the embryo is at 15-weeks' gestation.

"It is important to note that the donor mitochondrial DNA only replaces 37 mtDNA genes – contributing about 0.1 per cent of the baby's genetic make-up – compared with approximately 20,000 genes in the nucleus, which are not replaced.

"The mitochondrial DNA contribution is important for converting food into energy but appears to make no significant contribution to appearance, behaviour or other features, which are overwhelmingly determined by the nuclear genes and environment," he said.

Mitochondrial disease (mito) is a debilitating and potentially fatal genetic disorder that starves the body's cells of energy, depriving our major organs of the power they need to function properly. The ability to walk, run or even just stand up unaided can be a daily struggle for people with mito, which has few treatments and no cure and can cause any symptom in any organ at any age.

Mitochondria are the powerhouses of our cells that generate 90 per cent of the energy fuelling our bodies, particularly muscles and major organs like our brain, heart, liver, ears and eyes. 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 with these genetic mutations 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.

The Australian Mitochondrial Disease Foundation was set up in 2009 by family members, friends and doctors of sufferers to fund research into the diagnosis, treatment and cure of mitochondrial disorders, support affected individuals and their families, and educate the general public and the medical profession about mitochondrial disease. It does not receive government funding and relies solely on donations.

The AMDF funds numerous research projects, an Australia-wide mitochondrial patient database and priority access to a new Next-Generation DNA Sequencing Facility to enable faster, less expensive and more accurate diagnoses of mitochondrial disease. It operates a telephone helpline (1300 977 180), runs support groups including teleconferences and a Facebook group, and holds information days and symposia for patients, GPs, specialists and the public.

Australian Mitochondrial Disease Foundation: 1300 977 180, www.amdf.org.au

- The AMDF Position Statement on Mitochondrial Donation (including an illustration of the process) and further information is available at www.amdf.org.au/mitochondrial-donation/
- The HFEA media release is available at www.hfea.gov.uk/10559.html?platform=hootsuite
- The independent expert panel report to the HFEA - *Scientific review of the safety and efficacy of methods to avoid mitochondrial disease through assisted conception: 2016 update* - is available at www.hfea.gov.uk/10557.html
- A video primer on mitochondrial donation is available at www.nature.com/nrdp/animations/mito-dis-16

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