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Hope for mitochondrial disease

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MITOCHONDRIAL DISEASE & MITOCHONDRIAL DONATION IN BRIEF

Mitochondrial disease is a debilitating genetic disorder that robs the body's cells of energy, causing multiple organ dysfunction or failure and sometimes death.

There is no cure. Current treatments aim to decrease the effect of the symptoms but do not change the course of the disease.

1 in 5,000 babies are born with a severely disabling and likely terminal form of mitochondrial disease. That is 56 per year in Australia, or more than one each week.

Mitochondria are small structures in our cells which generate the energy that powers every part of our body. When mitochondria are faulty, the body doesn't get the correct level of energy it needs to function.

Since the **high energy organs** require so many mitochondria within their cells, they are usually the first to be affected by mitochondrial disease. These include the brain, which uses 20% of our total energy, nerves, muscles, eyes, ears, heart, bowels, liver, kidney and pancreas. **Symptoms** can include childhood dementia, loss of motor control, strokes, seizures, visual or hearing problems, cardiac and/or liver disease, developmental delay and intellectual disability.

Many people with mitochondrial disease have repeated and/or prolonged visits to hospital. Many need to stop working and rely on full time care, therefore **impacting family and friends, and relying heavily on healthcare and social services systems**.

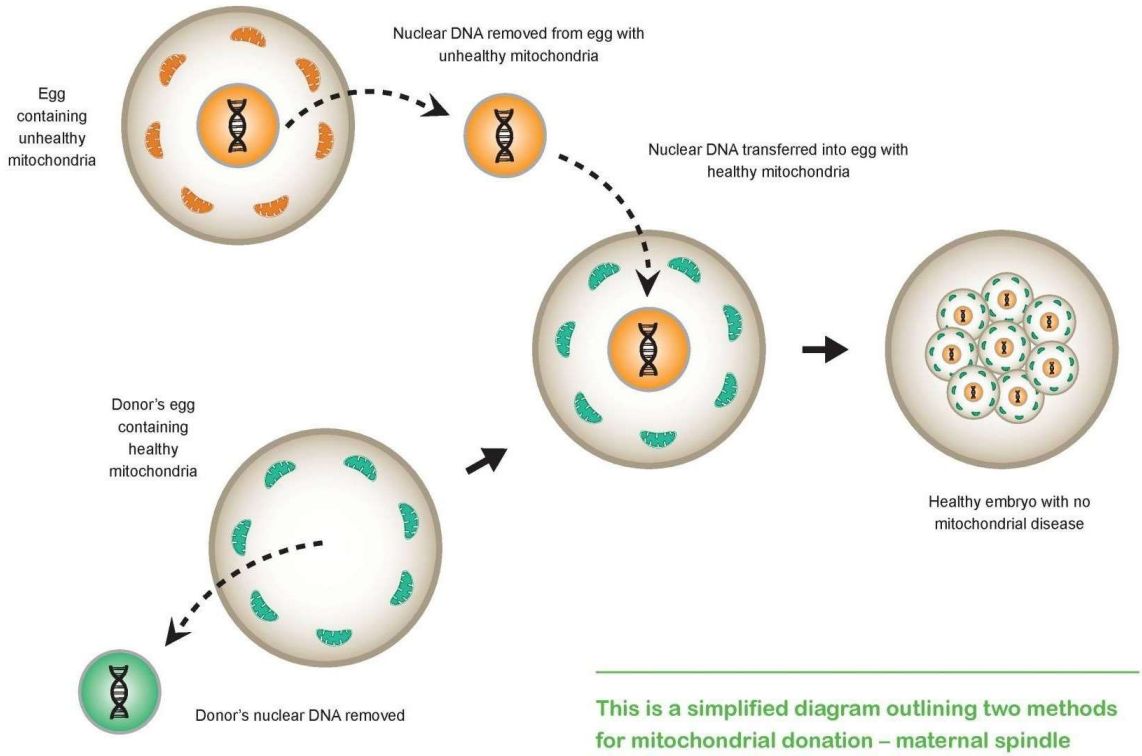
In about **half of all known cases**, mitochondrial diseases are caused by mutations in the separate mitochondrial DNA (mtDNA) that we **inherit only from our mother**. This is known as mitochondrial DNA (mtDNA) disease. About 1 in 200 people, or around 120,000 Australians, carry a mutation in their mitochondrial DNA that could potentially cause disease. It is likely that mtDNA disease is much more common in the community than previously thought.

Mitochondrial donation involves removing the nuclear DNA (the unique genetic information that makes us who we are and determines what we look like) from a patient's egg containing faulty mitochondria and inserting it into a healthy donor egg, which has had its nuclear DNA removed. This prevents mitochondrial DNA defects from being inherited by a genetically related offspring. There are two techniques:

Maternal spindle transfer (pre fertilisation): The nuclear DNA, which amounts to 99.9% of the total cell DNA, is removed from the donor egg, leaving the part of the cell containing the healthy mitochondria. The nuclear DNA from the mother's egg is then inserted into this cell. The healthy egg is fertilised and is then implanted into the mother's uterus in the same way IVF is carried out already.

Pronuclear transfer (post fertilisation): As above, but the nuclear DNA is removed from the mother's egg after the mother's egg is fertilised with the father's sperm (but prior to the development of the embryo), and then transferred to the donor egg containing healthy mitochondria, which has had its nuclear DNA removed. The healthy fertilised egg is then implanted into the mother's uterus in the same way as in maternal spindle transfer.

Approved by the UK Parliament in 2015, mitochondrial donation is not yet legal in Australia. Following a Senate Inquiry in 2018-19 and public consultation and work on scientific and other matters undertaken by the NHMRC and an Expert Working Committee since, on 24 March this year, Minister for Health Hon Greg Hunt MP introduced the ***Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021*** into the House of Representatives. This Bill, if passed, will give women who carry these genetic mutations the choice to eliminate the risk of their children inheriting this devastating and life-threatening disease by legalising mitochondrial donation.



Mitochondrial Donation

This is a simplified diagram outlining two methods for mitochondrial donation – maternal spindle transfer (prior to fertilisation) and pronuclear transfer (after fertilisation).