

What is mitochondrial donation? And how might it help people have a healthy baby one day?

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Credit: [Jonathan Borba/Pexels](#)

Mitochondria are tiny structures in cells that convert the food we eat into

the energy our cells need to function.

Mitochondrial disease (or mito for short) is a group of conditions that affect this ability to generate the energy organs require to work properly. There are many different forms of mito and depending on the form, it can disrupt one or more organs and can cause organ failure.

There is no cure for mito. But an IVF procedure called mitochondrial donation now offers hope to families affected by some forms of mito that they can have genetically related children free from mito.

After a law to allow mitochondrial donation in Australia was passed in 2022, scientists are now preparing for a clinical trial to see if mitochondrial donation is safe and works.

What is mitochondrial disease?

There are two types of [mitochondrial disease](#).

One is caused by [faulty genes](#) in the nuclear DNA, the DNA we inherit from both our parents and which makes us who we are.

The other is caused by faulty genes in the mitochondria's own DNA. Mito caused by faulty mitochondrial DNA is passed down through the mother. But the risk of disease is unpredictable, so a mother who is only mildly affected can have a child who develops serious disease symptoms.

Mitochondrial disease is the most common inherited metabolic condition affecting [one in 5,000 people](#).

Some people have mild symptoms that progress slowly, while others have [severe symptoms](#) that progress rapidly. Mito can affect any organ, but organs that need a lot of energy such as brain, muscle and heart are

more often affected than other organs.

Mito that manifests in childhood often involves multiple organs, progresses rapidly, and has [poor outcomes](#). Of all babies born each year in Australia, around 60 will develop [life-threatening mitochondrial disease](#).

What is mitochondrial donation?

[Mitochondrial donation](#) is an experimental IVF-based technique that offers people who carry faulty mitochondrial DNA the potential to have [genetically related children](#) without passing on the faulty DNA.

It involves removing the nuclear DNA from the egg of someone who carries faulty mitochondrial DNA and inserting it into a healthy egg donated by someone not affected by mito, which has had its nuclear DNA removed.

The resulting egg has the nuclear DNA of the intending parent and [functioning mitochondria](#) from the donor. Sperm is then added and this allows the transmission of both intending parents' nuclear DNA to the child.

A child born after mitochondrial donation will have genetic material from [the three parties involved](#): nuclear DNA from the intending parents and mitochondrial DNA from the egg donor. As a result the child will likely have a reduced risk of mito, or no risk at all.

This highly technical procedure requires specially trained scientists and sophisticated equipment. It also requires both the person with mito and the egg donor to have [hormone injections](#) to stimulate the ovaries to produce multiple eggs. The eggs are then retrieved in an ultrasound-guided surgical procedure.

Mitochondrial donation has been pioneered in the United Kingdom where a [handful of babies have been born](#) as a result. To date there have been no reports about whether they are free of mito.

Maeve's Law

After three years of [public consultation](#) The Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021 was passed in the [Australian Senate](#) in 2022, making mitochondrial donation legal in a research and clinical trial setting.

Maeve's law stipulates [strict conditions](#) including that clinics need a special license to perform mitochondrial donation.

To make sure mitochondrial donation works and is safe before it's introduced into Australian clinical practice, the law also specifies that initial licenses will be issued for pre-clinical and clinical trial research and training.

We're expecting one such license to be issued for the [mitoHOPE](#) (Healthy Outcomes Pilot and Evaluation) program, which we are part of, to perfect the technique and conduct a clinical trial to make sure mitochondrial donation is safe and effective.

Before starting the trial, a preclinical research and [training program](#) will ensure embryologists are trained in "real-life" clinical conditions and existing mitochondrial donation techniques are refined and improved. To do this, many human eggs are needed.

The need for donor eggs

One of the challenges with mitochondrial donation is sourcing eggs. For

the preclinical research and training program, frozen eggs can be used, but for the clinical trial "fresh" eggs will be needed.

One possible source of frozen eggs is from people who have stored eggs they don't intend to use.

[A recent study](#) looked at data on the outcomes of eggs stored at a Melbourne clinic from 2012 to 2021. Over the 10-year period, 1,132 eggs from 128 patients were discarded. No eggs were donated to research because the clinics where the eggs were stored did not conduct research requiring donor eggs.

However, research shows that among people with stored eggs, the number one choice for what to do with eggs they don't need is to [donate them to research](#).

This offers hope that, given the opportunity, those who have eggs stored that they don't intend to use might be willing to donate them to mitochondrial donation preclinical research.

As for the "fresh" eggs needed in the future clinical trial, this will require individuals to volunteer to have their ovaries stimulated and eggs retrieved to give those people impacted by mito a chance to have a healthy baby. Egg donors may be people who are friends or relatives of those who enter the trial, or it might be people who don't know someone affected by mito but would like to help them conceive.

At this stage, the aim is to begin enrolling participants in the clinical trial in the next 12 to 18 months. However, this may change depending on when the required licenses and ethics approvals are granted.

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