Taking action

on the preventable burden of mito

Responding to the economic study on the preventable burden of mitochondrial disease (mito) in Australia

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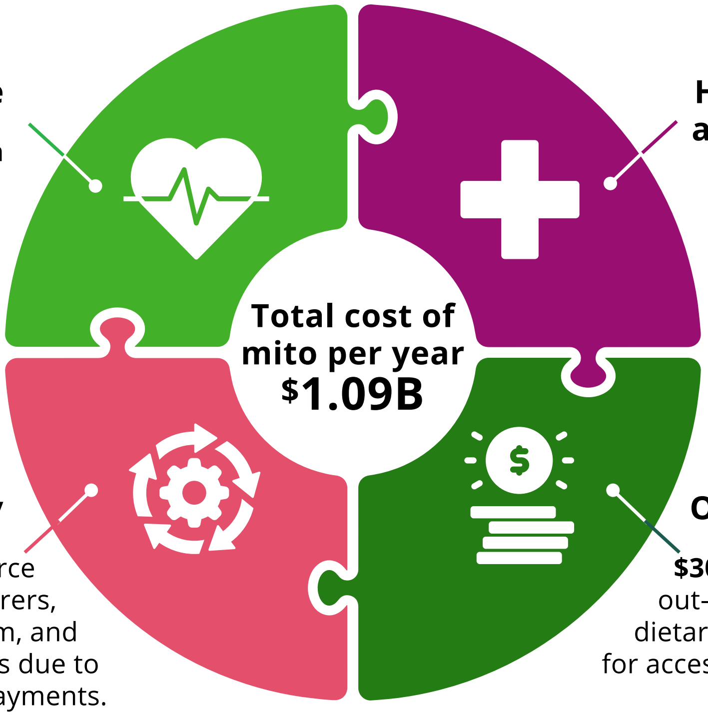
Highlights

Mitochondrial disease (mito) costs Australia over $1 billion each year*.



Total cost of mito per year
$1.09B

Quality of life

$860M from reduced quality of life and early death.

Health care and NDIS

$93M in health system expenditure, and **$40M** in NDIS costs.

Lost productivity

$34M in reduced workforce participation of carers, **$21M** in absenteeism, and **$7M** in economic costs due to income support payments.

Out-of-pocket costs

$30M per year including out-of-pocket health care, dietary supplements, travel for accessing health, etc.

*For more information on the calculation of these costs, please refer to the economic study on the *Preventable burden of mito* in Australia.[1]

We can reduce this

Reducing the cost of mito in Australia

Diagnose mito better

124,000 Australians have a genetic risk factor for mito. An unknown share of them may have experienced symptoms of mito and are at risk of deteriorating health. Diagnosing mito earlier could save $20 million each year: $17 million in health care costs and $3 million in productivity losses.

Support prevention options for mito

We could save $3 million for every healthy baby born without mito. We must continue support for prevention options for mito, including mitochondrial donation and reproductive carrier screening.

Improve health care and support

Many Australians with mito are missing out on the care they need. Improving health care and other support services can keep people with mito healthier for longer, improve their quality of life, reduce loneliness, and increase productivity.

Boost research in Australia to develop new treatments and cures for mito

Research is discovering new therapeutic options and can improve access and refine existing treatments for mito. Australia is the leader in diagnosing mito and is an ideal location for clinical trials.

Front cover image: **Bodhi and his mum Alysha**

The preventable burden of mito

Calculating a disease's burden and cost involves adding up how it affects people's lives and the community. This includes looking at factors like the number of people affected, the duration of their illness, its impact on their quality of life, expenses related to health care, and the impact on people's productivity.

Mito Foundation commissioned the Centre for International Economics (CIE) to calculate the total burden of mito in Australia for the first time. We also asked them to identify potential ways to reduce this burden.[1]

What is mito?

Mito is a group of rare genetic disorders that reduce the body's ability to produce energy. One in 4,300 Australians is affected by mito, and one in 200 carries genetic changes that may cause mito.[2-4] This progressive and potentially fatal disease can affect whole families. There are many forms of mito. It can cause any symptom in any organ at any age.[5] This makes mito a very challenging disease to diagnose and treat.

There is currently no known cure for mito. Depending on the type of mito and its severity, people can experience various symptoms. Fatigue and muscle weakness are the most common.[5] Other symptoms of mito can include:

hearing loss
vision loss
heart problems
diabetes
liver failure
anaemia
immunodeficiency
kidney disease
seizures
depression
anxiety
and more

What impact does mito have?

A framework for understanding the burden of mito in Australia

Individual impacts on people with mito

- Health and wellbeing
- Home and community life
- Education and learning
- Employment and financial security

Individual impacts on carers

- Reduced employment and financial security
- Caring responsibilities
- Household/parenting responsibilities

What this means for Australia

- Health system impacts including unnecessary health care costs
- Impact on government services including disability and income support
- Productivity losses due to reduced participation in workforce by people with mito and carers
- Psychosocial impacts including impacts on families

Mito affects every part of life, including work, study, and play. People with mito often have complex health care needs, which means they need care from many different specialists at the same time. They may also have non-health-related needs. These can include disability and financial support, access to housing and assistive technology, and so forth. Mito also impacts family members of both children and adults with mito, as they often take on the carer role.

Francesca's story

For Francesca, it started as some tingling in her arms. But when it became a throbbing, aching pain, the doctor told her she had repetitive strain injury (RSI). She didn't know, but this was one of her first symptoms of mito. It would take her 30 years to get a formal diagnosis of mito.

Over that time, she developed more symptoms: hearing loss, diabetes, pain in her legs, fatigue, and muscle weakness. This slow decline in health chipped away at the life Francesca had built. She recalls the day she finally had to retire from her job and the toll it took on her mental health:

"It was really devastating. I had a lot of counselling through that period."

"I also became quite agoraphobic and withdrawn because I didn't want to see anybody I knew. I just didn't want people to see me as I felt I had become. And using the walking frame...the way people perceive me and treat me is so very different now. I basically just dropped off the grid."

Francesca thought she might have mito after a relative passed away from it. But initially, she didn't get checked because she worried it might affect her children's insurance.

Today, Francesca mainly relies on her disability pension and National Disability Insurance Scheme (NDIS) plan. Accessing and using them hasn't been easy for her:

"So, I applied for a disability pension and jumped through all the hoops of fire that you have to, to do that. For the NDIS, it took me 4 months just to pull together all the reports I needed for the application. It was a very confusing process, and I needed a lot of support applying."

Despite all this, Francesca maintains a positive mindset. She stresses the importance of community and family and counts herself very fortunate for the assistance she has received over the years.

"I am lucky that I am well educated. I feel for people that are not. This system is hard enough for me, it must be impossible for someone who cannot navigate the system or speak English."

Key findings

Over 4,600 Australians live with symptoms of mito; almost 8% are children.

Population and burden of mito in Australia

345 children and **4,283 adults** living with symptoms of mito

2,613 carers of people with mito

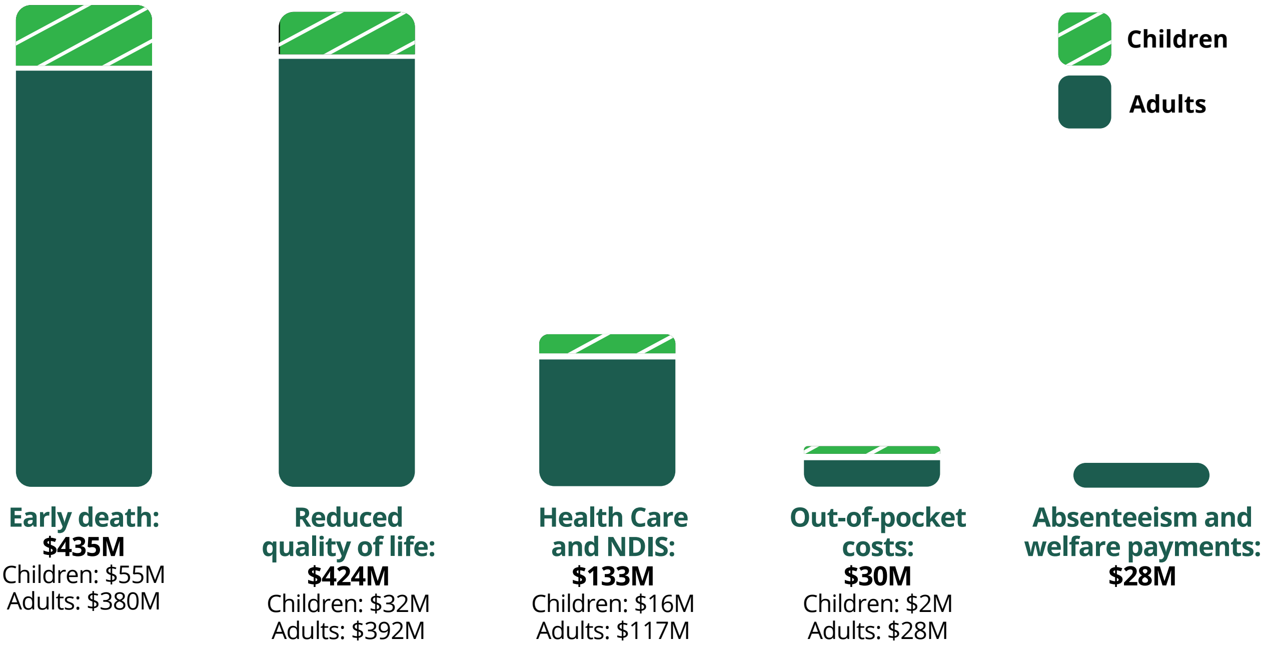
81 deaths and **1,805 years** of healthy life lost each year

Unknown share of **124,000 people** at risk of developing symptoms of mito

Another 124,000 Australians are estimated to have a genetic risk factor for mito.[1] An unknown share of them may have experienced symptoms of mito and are at risk of deteriorating health and wellbeing. There are approximately 700 Australians with symptoms of mito today who remain undiagnosed.[1]

There is approximately one carer for every 2 adults with mito. However, children with mito usually have more than one carer (often both parents).[1]

Mito costs Australia over $1 billion each year: $107 million for children and $948 million for adults. 80% of these costs are due to reduced quality of life and early death.



Priority areas for action

Action is required in 4 areas:

Diagnosis

Prevention

Treatment

Support

Progress requires collaboration between the mito community, health professionals, researchers, industry, philanthropy, and government. Mito Foundation is committed to working with these stakeholders to achieve positive outcomes for people impacted by mito.

Shelley and James
Picture courtesy Peter Mathew/Good Weekend

Australia is well-placed to continue making progress on reducing its burden of mito. Action in these areas can help governments to deliver on the commitments in the *National Strategic Action Plan for Rare Diseases.*[6] Many of the steps proposed will also progress the recommendations made by the House of Representatives Standing Committee on Health, Aged Care, and Sport in *The New Frontier: Inquiry into approval processes for new drugs and novel medical technologies in Australia.*[7]

Diagnosis

The study found: **Diagnosing mito earlier could save $20 million each year**

Over 120,000 Australians have a genetic risk factor for mito. We must accelerate progress on making diagnosing mito faster and less traumatic.

Diagnosing mito is a long and draining process, taking 6.4 years for adults and 1.8 years for children on average. Diagnosing mito earlier could save $17 million in unnecessary health costs and $3 million in productivity losses per year. Genomic testing for children saves up to $10,000 per child and improves the health care they receive.

Faster and less traumatic diagnoses of mito are possible. Adding genomic testing for mito to Medicare in 2023 recognised this. Further improvements are needed in detecting mito symptoms in primary care and connecting to specialist mito tertiary care. This can improve access to surveillance and proactive management — keeping people with mito healthier for longer. The National Health and Medical Research Council's support of a technology-driven project in this area is welcome.

The government's commitment to newborn screening, including research into genomic screening, has the potential to shorten diagnosis time in children. More research is needed to identify the health risks for the 124,000 Australians estimated to have a genetic risk factor for mito. This includes researching which sub-groups may benefit most from early detection and intervention.

Next steps:

- Reduce barriers to genetic testing, including a total ban on the use of genetic testing results by life insurance companies.
- Remain committed to the expansion of newborn screening, including working towards genomic screening.
- Collaboration with medical education organisations to improve the health workforce's ability to recognise symptoms of mito. This includes increasing the number of GPs completing the *Maybe It's Mito* training.[8]

How these actions deliver on existing policy

The first 2 pillars of the *National Strategic Action Plan for Rare Diseases* emphasise raising awareness and education about rare diseases across Australia and ensuring better diagnosis of rare diseases.[6]

The New Frontier report recommends the establishment of a jointly funded national genomic testing and counselling program that would allow all Australians to get tested.[7] It also stresses the potential of newborn screening to improve diagnosis and access to early intervention.

Prevention

The study found: **We can save $3 million for each healthy baby born without mito**

We can prevent mito. We must continue supporting prevention options for mito, including mitochondrial donation and reproductive carrier screening.

Australia is well-placed to use genomic and reproductive technologies to help families reduce their risk of having a child with mito. The CIE has calculated that each case of mito prevented can save approximately $3 million.

Mitochondrial donation offers new hope to families impacted by mitochondrial DNA changes. The Australian Government has acknowledged its importance by passing Maeve's Law (*Mitochondrial Donation Law Reform Act 2022*). This allows for the use of reproductive techniques for mitochondrial donation. The Australian Government has also funded the mitoHOPE Program (through the Medical Research Future Fund). This program will pilot the introduction of mitochondrial donation in Australia. The program will start recruiting in 2024.

Reproductive genetic carrier screening can identify families at risk of having a child with mito due to nuclear DNA changes.[9] Currently, Medicare funding is only available for Fragile X Syndrome, Spinal Muscular Atrophy, and Cystic Fibrosis — not mito.

Next steps:

- Australia must continue its support for the mitochondrial donation pilot program. This program is currently funded until 2028.
- Improve access to expanded reproductive genetic carrier screening through equitably funded access to testing, counselling, and reproductive options.

How these actions deliver on existing policy

The National Strategic Action Plan for Rare Diseases emphasises facilitating reproductive confidence in families at risk of passing down a rare disease by providing them equitable access to peri-conception genetic testing and counselling.[6]

Support

The study found: **Improving support, within and beyond health services, can improve quality of life and reduce costs of mito.**

Many Australians with mito are missing out on the care they need. We must continue to improve health care and other support services for mito and ensure these are sustainably funded.

Australians impacted by mito use many health services, including primary care, specialist care, and allied health. Often, these services are fragmented and need ongoing negotiation to access.[10] Many Australians living with mito are not accessing the health care they need. This includes support available post-diagnosis, particularly in emergency management.[11,12] Investment in centres of expertise for mito that provide multidisciplinary care is required.

Many people with mito also have significant disability and psychosocial support needs. This forces many to navigate Australia's complex support systems themselves. Over 50% lack helpful support in key areas of their life, including transport, education, employment, personal care, finances, and housing.[13] Improvements across these key areas would have many far-reaching benefits. For example, improved supports could enable people with mito and their carers to continue to work. The CIE found that if people with mito and carers could reduce their work hours by 50%, instead of leaving the workforce, productivity would be boosted by $52 million each year.[1] Australians living with mito would also benefit from recommended improvements to services such as the NDIS. This includes improved access to supports that can delay the progression of symptoms.

A significant proportion of the mito community have low mito-specific knowledge and confidence in managing their symptoms.[14] Non-government organisations like Mito Foundation play a key supporting role. Their services improve access to health care and provide the direct support people with mito need. For instance, accessing Mito Foundation's Pathways Telehealth Nurse reduces anxiety and improves confidence in managing mito for 89% and 70% of people, respectively.[14] Since 2018, over $2 million of these services' funding has come from private philanthropy, and only $90,000 has come from government contributions.

Next steps:

- A whole of government approach is needed to implement the *National Recommendation for Rare Disease Health Care* for people with mito, including a network of Centres of Expertise.[15]
- Explore and secure sustainable funding for Mito Foundation's support services.

How these actions deliver on existing policy

The *National Strategic Action Plan for Rare Diseases* calls for further development and investment into multidisciplinary care teams, which includes the establishment of Centres of Expertise.[6] *The New Frontier* report additionally recommends the formation of a Centre for Precision Medicine and Rare Disease within the Department of Health.[7]

Research

The study found: **New therapeutic options can lead to new ways to reduce the burden of mito**

The dream of mito treatments is within reach. We must increase research activity in Australia, particularly clinical trial activity, to accelerate progress on new therapies.

Clinical trial activity for mito is increasing worldwide. However, Australians need more opportunities to participate in these. Mito Foundation has identified 59 new mito treatment options in development. Of these, 21 therapies have had phase II/III clinical trials. **Australians only had access to clinical trials for 3 of these.**

Australia is a world leader in diagnosing mito. This makes it an ideal location for mito clinical trials. Creating the *National One Stop Shop* can contribute to overcoming some of the current barriers.[16]

We need more research to refine and improve access to existing treatments and active management approaches for mito. These can include structured exercise programs, diet interventions, and specific dietary supplements. This means funding through support systems such as the NDIS can then focus on evidence-based interventions.

Other rare diseases, such as cystic fibrosis, demonstrate what is possible when targeted therapies are supported. Similar progress could transform both life expectancy and symptoms for people with mito. This will ultimately reduce the burden of mito in Australia.

Next steps:

- Work with stakeholders to develop a plan to increase mito-related clinical trial activity in Australia. This work should include investments in natural history studies, patient registries, patient-reported outcome measures, and clinical trial networks.
- Allocate resources for targeted investments in mito clinical research, co-funded by philanthropy and the government.

How these actions deliver on existing policy

Increasing research funding and clinical trial activity in Australia is a priority across all existing frameworks and policies. The *National Strategic Action Plan for Rare Diseases* and *The New Frontier* report both highlight the importance of stakeholder collaboration in planning for this.[6,7]

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ISBN 978-1-7635228-0-0

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