# SUBMISSION TO THE SENATE COMMUNITY AFFAIRS LEGISLATION COMMITTEE

Re: Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021

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16 JULY 2021

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The Mito Foundation welcomes the opportunity to provide this submission to the Senate Community Affairs Legislation Committee regarding the *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021.*

The Mito Foundation notes the Inquiry undertaken by the Senate Community Affairs References Committee into the *Science of mitochondrial donation and related matters* in 2018 and the Mito Foundation and the community that we support thank Senators for the time and effort they have given to this issue.

The Mito Foundation supports Australians impacted by mitochondrial disease, funds essential research into the prevention, diagnosis, treatment and cures of mitochondrial disorders, and increases awareness and education about these devastating diseases.

This submission provides information regarding the Mito Foundation's support for the *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021* and highlights some particular areas of the legislation that should be noted.

Maeve's Law is named after five year old Maeve Hood who has a life threatening form of mitochondrial disease. The Mito Foundation acknowledges Maeve and her family, as well as the thousands of Australians whose lives have been impacted by the devastating impacts of mitochondrial disease, in making this submission. The Australian mitochondrial disease community is the inspiration behind the Mito Foundation's wholehearted support of Maeve's law.


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## INTRODUCTION

The *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021* will enable parents to access mitochondrial donation – an IVF based technique – and mean that around 56 children born annually in Australia with mitochondrial disease could potentially avoid inheriting this deadly disease.

Maeve's Law makes mitochondrial donation legal in Australia by amending the *Prohibition of Human Cloning for Reproduction Act 2002* and the *Research Involving Human Embryos Act 2002*.

Maeve's Law will allow at risk families to have a biological child using a technique that minimises the risk of transmission of mitochondrial disease. This represents a significant step forward and offers hope to families who have often endured generations of suffering due to mitochondrial disease, a disease which has no known cure and very few treatments.

## MITOCHONDRIAL DISEASE

Mitochondrial disease is a debilitating genetic disorder that robs the body's cells of energy, causing multiple organ dysfunction or failure and sometimes death. When mitochondria are faulty, the body does not get the correct level of energy it needs to function.

There is no one age group affected by mitochondrial disease and people can develop it in infancy, in early childhood, in their teenage years or as adults.

1 in 5,000 Australian babies are born that will develop a severely disabling form of mitochondrial disease that can cause death in infancy, childhood or adulthood. That is, more than one baby born every week in Australia.

In some cases, mitochondrial disease is caused by genetic mutations in the nuclear DNA inherited equally from our biological parents. Mitochondrial disease can also arise as a spontaneous (i.e. not inherited) genetic mutation at conception.

However, in about half of all known cases, mitochondrial diseases are caused by mutations in the separate mitochondrial DNA (mtDNA) that are inherited only from our biological mother. About 1 in 200 people, or around 120,000 Australians, carry a mutation in their mitochondrial DNA that could potentially cause disease. Mitochondrial donation will enable impacted families the opportunity to minimise the risk of passing this type of mitochondrial disease from mother to child.

### THE IMPACTS OF MITOCHONDRIAL DISEASE

The impacts of mitochondrial disease can be devastating and virtually all forms of it have significant impact on patients. Babies and young children die from some types of mitochondrial disease, whilst other types of mitochondrial diseases can impact sufferers in different ways.

Depending on the person and the form of their mitochondrial disease, they may suffer symptoms ranging from loss of motor control, dementia (including childhood dementia), blindness, deafness, strokes, seizures, cardiac and/or liver disease, developmental delay and intellectual disability. Problems with balance and digestive or eating difficulties are also common and all require significant


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treatment and care with the result that many people with mitochondrial disease need repeated and/or prolonged visits to hospital.

Repeated seizures and loss of motor control can result in people of all ages having to give up school or work and can result in them needing full time care. This has significant impact on them, their families and friends and means they rely heavily on healthcare and social services systems.

With few effective treatments and no known cure, the impact of mitochondrial disease is devastating.

## SCIENCE INFORMING MITOCHONDRIAL DONATION

It is possible to significantly reduce the risk of mitochondrial disease being passed on. Mitochondrial disease caused by mutations in the nuclear genes involved in mitochondrial function can be prevented through prenatal testing or an IVF based procedure called preimplantation genetic testing.

These approaches are generally not as reliable when the mutation is in mitochondrial DNA inherited from the mother. Mitochondrial donation is an alternate approach.

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.

### INTERNATIONAL EXPERIENCE

Mitochondrial donation has been legal in the United Kingdom since 2015.

The process undertaken in the UK prior to the legalisation of mitochondrial donation firmly established the scientific evidence base for mitochondrial donation and its role in preventing transmission of mitochondrial donation between mother and child. The scientific review undertaken in the UK was in fact recognised as the most rigorous look at any scientific endeavour being introduced into humans.[1] It was based on expert international advice and information.

In addition, an open letter to The Guardian[2] on this issue co-signed by 40 international experts in the field, recognised the process that the UK went through, acknowledging the admiration it attracted internationally. This letter also noted the consideration given to the various benefits, risks, ethical and other issues that had occurred.

The science informing mitochondrial donation has been extensively reviewed and there is no suggestion or evidence of those international experts changing their stance since the introduction of mitochondrial donation in the UK.

The UK Human Fertilisation and Embryology Authority has issued the licenses to enable parents to use mitochondrial donation. Due to patient confidentiality, no announcement has or will be made

[1] The full quote of this, from Sir Jeremy Farrer, Director of Wellcome, is "'*I don't think there's been any more rigorous look at any scientific endeavour coming into humans*".
[2] The Guardian (UK), *Parliament should approve regulations for mitochondrial donation*, https://www.theguardian.com/science/2015/jan/30/parliament-should-approve-regulations-for-mitochondrial-donation Accessed July 2021


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about babies' births as a result of mitochondrial donation but children are likely to have been born using this technique. It is not new nor untested.

## EXTENSIVE AUSTRALIAN PUBLIC, SCIENTIFIC AND PARLIAMENTARY ENGAGEMENT

Australia has undertaken significant public, scientific and parliamentary engagement regarding the introduction of mitochondrial donation over a number of years.

### SENATE INQUIRY INTO SCIENCE OF MITOCHONDRIAL DONATION AND RELATED MATTERS: 2018

In 2018, the Senate Community Affairs Committee undertook an inquiry into the *Science of mitochondrial donation and related matters.* The inquiry examined the impacts of mitochondrial disease and the legal and ethical considerations associated with mitochondrial donation.

The Committee received 60 submissions from individuals including patients, scientists, clinicians and family members of patients as well as from medical, patient and other organisations. Submissions were overwhelming positive although, given the nature of the issue under consideration, there were some that voiced concerns regarding issues relating to the creation, use and destruction of embryos, donor rights and others. These were largely and legitimately informed by their authors' belief systems.

Reporting in June 2018, the Senate Community Affairs Committee made a series of recommendations including that a public consultation be undertaken about the introduction of mitochondrial donation and that 'the Australian Government prepare a consultation paper, including options for legislative change that would be required'.

### PUBLIC AND SCIENTIFIC CONSULTATION: 2019-2020

Following these recommendations, the Government tasked the NHMRC to bring together an Expert Working Committee to work on scientific and other matters raised by the Inquiry. Between March 2019 and March 2020, the Expert Working Committee engaged in robust discussion informed by the range of expertise on the Committee and its diversity of views. The Committee delivered their final report and advice to the Australian Government in 2020.

In addition, a significant program of public consultation was undertaken over some months in late 2019 to explore the Australian community's opinions and views regarding the social and ethical issues raised by mitochondrial donation.

A Citizens' Panel was convened and met over two weekends to consider and reach a position on mitochondrial donation. The Panel identified a number of reasons why mitochondrial donation might be desirable as an option for people at risk of passing on mitochondrial disease to their children. These included helping prevent suffering and death whilst improving quality of life; the opportunity to have healthy genetically related children; breaking the cycle of mitochondrial disease in families, reducing emotional trauma and improving mental health; and reducing the costs to the community of mitochondrial disease.


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The Panel concluded that "We are aware of the unknowns related to mitochondrial donation, but the majority have the view that mitochondrial donation should be permitted".[3]

In addition to the Panel, hundreds of people contributed to the NHMRC's consultation through public forums, webinars, online submissions, and other activities.[4]

Themes raised included outcomes from introducing mitochondrial donation; the wellbeing and rights of children; factors relating to egg donation and the donor; factors relating to the embryo in mitochondrial donation; and implementation considerations.[5]

A common theme among many of the responses opposed to the introduction of mitochondrial donation was the use of embryos, with the destruction of embryos in the process considered to be unethical.

A number of responses suggested that mitochondrial donation should initially be introduced as part of a clinical research study and mitochondrial donation should only be used in certain circumstances, such as where significant risk of mitochondrial disease existed. Other respondents also indicated that appropriate oversight mechanisms would need to be introduced, including limiting mitochondrial donation only to clinics which had appropriate expertise.

A number of responses that were supportive of mitochondrial donation indicated that one of the reasons for this support was the option for families to have healthy children free from mitochondrial disease thus avoiding the impact of the disease on the child and their family. A number of respondents acknowledged that risks exist with the procedure but those supportive of introducing mitochondrial donation tended to regard the benefits as outweighing the risks.

## PUBLIC AND SCIENTIFIC CONSULTATION: 2021

In February 2021, a public discussion paper was released by the Department of Health requesting input from the Australian community regarding a two-stage implementation proposal for introducing mitochondrial donation.

This public discussion paper sought feedback about legalising mitochondrial donation initially for research and training and for a pilot program to enable affected families to access mitochondrial donation; a further roll-out of this to clinical practice depending on the outcomes of the trial; and a clear regulatory framework to guide this work under the oversight of the Embryo Research Licensing Committee of the NHMRC. Ethical matters, such as pre-treatment counselling, privacy, monitoring and reporting, were also proposed as were issues relating to the identification of donors and their legal status.

[3] Mitochondrial Donation Community Consultation Citizens' Panel Statement, https://www.nhmrc.gov.au/file/15363/download?token=1Drxz1-1 Accessed July 2021.
[4] NHMRC, 'CEO Statement: should Australia introduce mitochondrial donation?", 5 June 2020. https://www.nhmrc.gov.au/file/15368/download?token=2im6Rys3 Accessed July 2021.
[5] NHMRC, *Report on NHMRC's public consultation on the social and ethical issues raised by mitochondrial donation,* June 2020. https://www.nhmrc.gov.au/file/15362/download?token=s-VFszfn Accessed July 2021.


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This public consultation identified support for the proposed implementation approach and Maeve's Law is designed to give effect to the publicly support approach to introducing mitochondrial donation in Australia.[6]

## MAEVE'S LAW

The *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021* recognises the various issues and challenges in relation to introducing mitochondrial donation, including those raised during both the 2018 Senate Inquiry and subsequent consultation process, and addresses them in a practical, considered and proportionate manner.

Maeve's Law as currently drafted allows for the introduction of mitochondrial donation into Australia in a carefully staged and managed manner with a variety of safeguards and protections incorporated so as to support Australian families, children and the scientific, medical and general community.

### CAREFULLY STAGED INTRODUCTION

Maeve's Law allows for a very carefully staged introduction of mitochondrial donation into Australia and establishes governance arrangements that ensures it will be closely monitored and appropriately overseen by experts within the NHMRC.

The first stage of the two-stage implementation process outlined in Maeve's Law sees mitochondrial donation first legalised for certain research and training purposes, including a clinical trial of the use of mitochondrial donation.

This first stage will ensure that there is an Australian-specific evidence base in relation to mitochondrial donation whilst at the same time allowing the establishment of Australian expertise in mitochondrial donation techniques. At the same time, some affected Australian families will have the opportunity to access mitochondrial donation in a timely manner, avoiding the risk of passing on mitochondrial disease to their children whilst ensuring they do not miss the window available to them for having genetically related children.

It is anticipated that this first stage will be undertaken over a number of years, enabling the collation of trial data and the appropriate establishment of the necessary training and other skills needed in this area.

The second stage will commence some years later following the introduction of regulations that would permit mitochondrial donation to be used in clinical practice. This will enable appropriate review of data related to mitochondrial donation and consideration of any new and/or additional information regarding mitochondrial donation and the science regarding it to be considered at that time.

[6] The Parliament of the Commonwealth of Australia, *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021: Explanatory Memorandum.* https://www.aph.gov.au/Parliamentary_Business/Bills_Legislation/Bills_Search_Results/Result?bId=r6697 Accessed June 2021.


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## A CLEARLY DEFINED REGULATORY AND LICENSING SYSTEM

Maeve's Law outlines a clearly defined licensing process whereby two types of mitochondrial donation techniques will be permits under either a clinical trial research and training license or a clinical trial license. This recognises the current state of the science in relation to mitochondrial donation and also reflects the UK experience where only these two techniques are identified as being suitable for use for human reproductive purposes.

Maeve's Law also will also ensure that clinic/s are appropriately licensed before being permitted to undertake research and training or a clinical trial in mitochondrial donation. The establishment of five types of licenses under which different activities can occur are clearly thought out and defined and inform the licensing system throughout the proposed two-stage implementation process.

It should be noted that an additional advantage of the need to review and potentially introduce additional regulations prior to mitochondrial donation being introduced into clinical practices is the opportunity to again review this science at an appropriate time in the future. By then, data may support the adoption of additional mitochondrial donation techniques whilst the built-in reviews may ensure that regulations do not lag significantly behind science if that is the case

## LICENSING PROCESS ENSURES EXPERT OVERSIGHT

The licensing and approval processes outlined in Maeve's Law deliver appropriately expert oversight under the supervision of the NHMRC's Embryo Research Licensing Committee (ERLC). This will ensure that the clinic/s undertaking these techniques will need to be licensed by the ERLC and the use of mitochondrial donation techniques will only be undertaken for the appropriate at-risk candidates.

This is an appropriate means of ensuring that mitochondrial donation is performed only in accordance with the legislation and only by people and clinics who have the appropriate qualifications and knowledge. It reflects closely the system introduced in the UK whilst, as outlined below, tailoring this for Australia's own environment, community expectations and other governance arrangements.

***RECOMMENDATION**: The Mito Foundation urges that the licensing and approval processes must be undertaken in a timely and appropriate manner so as to avoid unnecessary delays and the associated impacts for patients.*


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## ACCESS ONLY FOR THOSE AT SIGNIFICANT RISK

Maeve's Law provides that mitochondrial donation will only be available to those families who have a significant risk of passing mitochondrial disease to their children.

***RECOMMENDATION**: To ensure that the appropriate families can access mitochondrial donation, the Mito Foundation strongly recommends that the ERLC has access to and utilises one or more **clinical** experts in mitochondrial disease. This will ensure that all appropriate information and matters are considered.*

The Mito Foundation is aware of concerns regarding the so-called 'slippery slope' regarding the introduction of mitochondrial donation whereby it is argued that allowing this technique to become legal may open the way to further amendments to laws governing the treatment of human embryos.

Given the nature of mitochondria and their DNA as distinct from nuclear DNA and the contents of Maeve's Law, this concern is unfounded. Maeve's Law clearly specifies that the only use of mitochondrial donation is to minimise the possibility of this devastating disease from parent to child and does not expand beyond this. This is clearly defined and cannot be misinterpreted to enable the mitochondrial donation techniques to be used for any other purpose.

## CHANGES TO MITOCHONDRIAL OR NUCLEAR DNA ARE NOT PERMITTED

The above points are further emphasised by the fact that Maeve's Law clearly and expressly excludes the intentional modification of either nuclear or mitochondrial DNA during mitochondrial donation. As such, the donor mitochondria cannot be modified nor can the nuclear DNA of the biological parents.

This clear position within Maeve's Law ensures that there is no risk of changes to either type of DNA that could be considered to be of a 'choice' of the parents nor lead to the creation of so-called 'designer babies'. Mitochondrial donation, as outlined in Maeve's Law, can only be used to minimise the risk of Australian parents passing on mitochondrial disease to their children.

## MANAGEMENT OF ETHICAL ISSUES

In drafting Maeve's Law, a number of key ethical issues have been considered and outlined to ensure that families, donors and the community have access to appropriate support and a clear management process.

Some of these matters will be managed in a manner that mirrors that utilised in the UK whilst others build on the knowledge gleaned there and tailored for the Australian context.


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## PRE-TREATMENT COUNSELLING

As in the UK, parents will be required to undergo counselling prior to accessing mitochondrial donation. This is an appropriate and proportionate measure and will ensure that the potential risks involved with mitochondrial donation as well as any potential alternatives available to specific individuals are fully explained and understood. This approach will allow families to make informed decisions about mitochondrial donation and permit them appropriate reproductive choice.

## PRIVACY OF FAMILIES AND CHILDREN

The privacy of families and children has long been recognised as an important issue in relation to mitochondrial donation as it should be for all people undergoing health or medical treatment.

Maeve's Law prioritises the privacy of families and children while at the same time ensuring that health monitoring of children will occur as will reporting of any adverse events. This monitoring is intended to take place wherever possible within the mainstream healthcare system providing additional privacy for families and their children. Maeve's Law also ensures that children will not be subject to unnecessary or routinely invasive monitoring, providing a strong framework that again is appropriate and proportionate.

## LEGAL STATUS AND IDENTIFICATION OF DONORS

As with the UK, mitochondrial donors will not be considered parents under Maeve's Law. This mirrors other Australian donor laws and delivers upon community expectation.

Likewise, although in contrast to the UK, children born from mitochondrial donation will be able to receive identifying information about their donor when they turn 18. This again mirrors other laws that govern donation in Australia and meets the expectations of our community.

## OTHER SAFEGUARDS AND PROTECTIONS

The manner in which Maeve's Law has been drafted ensures that a number of safeguards and protections are established within it as well as the opportunity for additional parliamentary review. This is a sophisticated and appropriate way to manage this issue and the Mito Foundation congratulates the Government and the team involved in drafting Maeve's Law for their foresightedness and proportionate management of these issues.

## BUILT-IN REVIEWS

The two-stage implementation process proposed by Maeve's Law ensures that a number of reviews of the scientific and regulatory frameworks governing mitochondrial donation will be undertaken prior to mitochondrial donation becoming introduced into clinical practice in Australia.

Maeve's Law is currently being reviewed by the Senate Committee and will then be considered by both the House of Representatives and the Senate before the first stage can be introduced. This is a significant task and one which builds on the work already done through the engagement process outlined above.


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Once the first stage is completed, regulations will then need to be made before mitochondrial donation can be introduced into clinical practice. This essentially ensures that, at that time, a further review will be undertaken and, should concerns arise, these can be addressed at that time. This two-stage implementation process provides multiple opportunities for mitochondrial donation to be considered and any necessary changes to its governance systems made.

### CONSIDERATION BY BOTH COMMONWEALTH AND STATE AND TERRITORIES

Further, given the various responsibilities of the different levels of Australian Governments, before mitochondrial donation can be introduced into clinical practice in Australia, consideration will be required by both the Commonwealth *and* State and Territory Governments.

Whilst changes to Commonwealth law are necessary to enable the proposed clinical trial to go ahead, the use of mitochondrial donation in clinical practice will require both changes to Commonwealth regulations *and* the introduction of State and/or Territory laws. This is because, before mitochondrial donation can be used in a particular State or Territory, that jurisdiction will need to enact their own laws authorising and regulating its use.

The Mito Foundation fully supports these measures.

## ADDITIONAL BENEFITS OF MAEVE'S LAW

In addition to the specific elements of Maeve's Law, it is important to consider the additional benefits that it grants to Australian families and the community more broadly.

Principal of these is undoubtedly the opportunity to have a healthy genetically related child, something identified as a key benefit of mitochondrial donation in all the public consultations and engagements regarding this issue. The opportunity also exists to leverage Australia's scientific expertise and leadership to the benefit of the Australian community and also to protect Australian families from potential exploitation in relation to mitochondrial donation.

### GIVING AUSTRALIAN FAMILIES CHOICE

As has been highlighted throughout the processes of engagement undertaken by the Senate Community Affairs Committee, the NHMRC and the Department of Health, offering parents at risk of passing mitochondrial disease onto their children the opportunity to minimise this risk is of major value.

In addition to the opportunity to have a healthy child, mitochondrial donation offers some families the only means to have a child genetically related to both parents who is healthy. While some people do not consider genetic relationship to be of significant importance, evidence indicates that this is not the case for a large number of people. This was upheld throughout the consultations mentioned above and has also been highlighted by members of the Australian mitochondrial disease community who are clear that they are not seeking to have a 'designer baby' but simply a child related to both parents and one who will not suffer from mitochondrial disease. A case study relating to this issue is incorporated at Appendix One – Case Study: Shelley's Story.


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Maeve's Law also allows for parents, following pre-treatment counselling and if safe and practicable, to choose to implant only male embryos. Given that mitochondrial DNA is passed only through the female line, this option would allay any potential risk that mitochondrial disease might reappear in future generations and, as such, is being offered to parents. The Mito Foundation does not hold significant views on this issue but allowing parents this option aligns with the broader philosophy underpinning Maeve's Law that families should be fully informed and fully empowered to make decisions whether or not to access mitochondrial donation.

## LEVERAGING AUSTRALIA'S SCIENTIFIC EXPERTISE AND LEADERSHIP

Australia is well-known for its scientific expertise and leadership and has long been pioneers in the use and development of safe IVF treatment. Further developing this expertise in the area of mitochondrial donation presents an opportunity for our scientists and researchers.

In addition to this, the introduction of a carefully designed regulatory and licensing system in Australia will ensure that Australian families have access to mitochondrial donation, and their children to appropriate health monitoring, which might otherwise not be available.

The Mito Foundation is aware of instances where mitochondrial donation has been utilised in unregulated health systems or for conditions not related to the avoidance of mitochondrial disease. The Mito Foundation has been approached on occasion by overseas organisations seeking to offer access to mitochondrial donation to Australian families and has long been concerned that, without an appropriate domestic regulatory framework, the risk exists that vulnerable families might be tempted to access these techniques in other jurisdictions.

## CONCLUSION

The Mito Foundation welcomes the *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021* and the clear regulatory and licensing systems it is seeking to establish. This is being done in the best interests of the families and children involved and will enable families the opportunity to choose to minimise the risk of passing this devastating disease onto their children and future generations.

The Mito Foundation wishes to acknowledge the significant work undertaken on this issue by the Australian Government, the Minister for Health and his office, the Senate Community Affairs Committee, a number of supportive Parliamentarians from all parties, the NHMRC and the Department of Health.

It is humbling for a small organisation and community such as ours to receive the benefit of the time and effort invested by so many people and, on behalf of the Australian mitochondrial disease community, we wish to record our thanks.


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## APPENDIX ONE – CASE STUDY: SHELLEY'S STORY

I had never heard of mitochondrial disease until 3 years ago.

My journey started when my mother experienced a rapid decline in health, losing muscle in most parts of her body and struggling to breathe. Her, my older brother and I all experienced hearing loss at a young age (32, 13, 21 respectively). After several doctors' appointments and hospital admissions, it was established that my mother had heart failure, muscle weakness and fluid on her lungs – she passed away shortly after symptoms progressed in Feb 2016.

Only a few months prior to her decline, my brother experienced what appeared to be a heart attack. It was only after my mother's passing that a report from a biopsy came back to indicate probable mitochondrial cytopathy in my brother. I was then genetically tested through a blood test at the genetic centre and was given the diagnosis of MELAS (Mitochondrial Enchephalomyopathy, Lactic Acidosis and Stroke-like symptoms) - a maternally inherited form of mitochondrial DNA disease.

A few months after my diagnosis, my brother started experiencing myoclonus seizures, his leg twitching resulting in severe falls. Following this, he had migraines, vomiting and hallucinations, then stroke-like episodes where he lost his eye sight for weeks on end. In June 2017 he got an infection and passed away, 3 weeks short of his 35th birthday.

Mitochondrial disease has not only taken away half of my family and my best friend (my mum), it has left me fearing for my own future and for my partner. There are so many adjustments required in terms of employment, relationships, responsibilities, finances, health access and starting our own family. When my mother passed away, my maternal instinct only increased, knowing what a truly special person she was and how incredibly important she has been in moulding me as a person.

***"Mitochondrial disease has not only taken away half of my family … it has left me fearing for my own future"***

There are so many times I reflect and give thanks that I possess some of her quirks and personality traits. I so want that for my own child, yet I fear passing on this awful disease to them. Throughout the last 3 years I have done a great deal of research on mitochondrial disease and at times have stopped myself from learning more as the prognosis is bleak and there are no treatments currently available.

My current symptoms of mitochondrial disease include bilateral sensorineural hearing loss (moderate to profound), daytime hypertension and thickened left side of heart (high risk of heart failure due to family history), mitochondrial diabetes insulin-dependent, elevated lactate CSF, muscle weakness and exercise intolerance. My energy levels fluctuate greatly from day to day and at times I am paranoid about getting sick or taking medications that may be contraindicated to the condition. Lifestyle changes have to be made to accommodate my diabetes and weakness to ensure that I don't overexert myself.

My month is most often filled with various doctors' appointments: cardiologist, neurologist, endocrinologist, gastroenterologist, audiologist, physiotherapist, psychologist and IVF treatments. Not only does this consume my time and energy but is naturally expensive and requires me to take a large amount of time off work resulting in financial loss. There is sadness, guilt, frustration, fear, paranoia, anxiety and anger about how incredibly devastating this disease is and the vast impact it has on every single aspect of our lives. Guilt surrounding my lack of understanding when my mum


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and brother were sick, guilt at having to place a great amount of responsibility on my husband at such a young age and stage in our marriage, fear of experiencing what my mum and brother did, fear that my husband may have to witness this and be left with the consequences. Yet there is still hope that we can progress with change and learn quickly from our international mitochondrial disease partners about how to save lives and prevent this disease from spreading.

While having to come to terms with my losses both of family and my own limitations, I am devastated that this disease could take away the opportunity to have my own child. While I am aware that there are alternative options such as fostering, adopting or egg donation, my partner and I have a strong desire to have our own biological child. This desire is not only because of the genetic components, but because I truly appreciate and value what I inherited from my mum and often look in the mirror to reflect what part of me belongs with her.

***"I have a strong desire to have our own biological child … because I truly appreciate and value what I inherited from my mum and often look in the mirror to reflect what part of me belongs with her"***

Additionally, if anything were to happen to me, my partner has a piece of me within our children. While this may not be everyone's choice, it should be an option for people with this disease to be able to have their own biological child, disease free. We have tried and exhausted all other available options within Australia and are considering accessing mitochondrial donation in the UK as we are running out of time to start our family. Naturally it would be the preferred option to access this procedure in Australia.

***"… it should be an option for people with this disease to be able to have their own biological child, disease free. We have tried and exhausted all other available options"***

In my many years of working professionally in the field of disability, mitochondrial disease can be utterly incomprehensible and has such a large spread of symptoms impacting several organs at a time. This is challenging both physically and mentally (at the same time!). As stated above, mitochondrial disease has already taken a great deal from me and my family and I am hopeful that we can bring about change, if not for me personally, for our future generations and society.

**Shelley Beverley**
2019


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