Senate Enquiry into Mitochondrial Donation:

Thank you for the opportunity for me to share my story and hopefully make a difference in many lives to come. It has been an incredible journey over the last 2 years, one that I would never wish on anyone. I had never heard of mitochondrial disease before this. So many things make sense now and there is so much I wish we could change. In our family of 4, my mother, brother and myself all experienced bilateral sensorineural hearing loss at varying ages, and have all been relatively thin in body frame. However, mitochondrial disease only became evident after my brother had a suspected heart attack in June 2015, with a muscle biopsy indicating mitochondrial myopathy. 2 months later my mother suddenly began to lose all the muscle in her body, struggled to breathe and passed away aged 63 in early February 2016. Months later I would receive my diagnosis of MELAS (mitochondrial encephelomyopathy, lactic acidosis and stroke-like episodes). June 2016 saw my brother experience myoclonus seizures, followed by episodes of cyclic vomiting and headaches. December 2016, he suddenly lost his vision and began to experience stroke-like episodes with numbness and psychotic episodes (hallucinations). Episodes became more severe and he passed away aged 34 in June 2017. The trauma of watching both my mum and brother deteriorate so rapidly and so devastatingly, will stay with me forever. To watch my mum lose all the strength in her body and being unable to communicate, eat, sit up, use or hands or get herself off the toilet, was extremely heartbreaking. Watching the nurses restrain my 36kg mum in the hospital because she wanted to get her tubes out to try to talk to me (because she had lost all muscle in her hands) will remain with me forever. To have my brother desperately search for Siri on his iphone to make a call to my father when he has lost all vision following a grand-mal seizure, is heartbreaking.

Knowing all this, and losing half my family in 16 months is difficult. What makes it even more life changing is finding out that I too have this disease and will possibly face the same devastating journey. But worse than this, I would almost certainly pass this disease on to my children.

It is strange just how strong the desire to have children became after my mother passed away. I suddenly yearned for those moments of comfort and her knowing exactly what to say to me. A mother-child relationship is something truly special. My husband and I had started trying to conceive about 6 months prior to my mother passing away. In hindsight we are grateful in a sense that we were not successful as we are now aware of the risk of having an unhealthy baby. It was after my diagnosis that we heard of Preimplantation Genetic Diagnosis (PGD). Through extensive consultation and designing a test for us, we began our journey with PGD IVF in June 2016. Since then we have undertaken 5 cycles (roughly 50 eggs retrieved), of which only 2 embryos have had the lowest mutation loading of 32%. While scientists and geneticists have indicated an embryo of less than 15% to be suitable, we find this a difficult benchmark to meet. We have faced many physical, financial and emotional challenges throughout the process. Financially, it places a great strain on us to pay roughly $7000 out of pocket per cycle, with previously having to travel from Hobart to Melbourne for the first 4 cycles. Physically, having mitochondrial disease has resulted in slower recovery and greater lack of energy and increased pain. Emotionally, the rollercoaster of having to desperately rely on success at each stage – retrieving as many eggs as possible, having them fertilise, seeing how many survive and grow to day 5, then biopsy the surviving embryos to

establish their mutation loading. Going through each stage of a cycle generally means going from 14 eggs down to 3 biopsied embryos only to find out that they do not meet the loading threshold. It is a difficult and draining process that seems unfair and frustrating. With any couple trying to have a child, time is a factor, as well as health. For us, PGD seems to show little success.

We have considered all the other avenues of egg donor, adoption or not having children. We have a strong desire to have our own child who inherits our genes, our personalities, ourselves. If something were to happen to either of us, we would still have a part of each other. When we heard about mitochondrial donation, we felt that this would provide us with the opportunity to have a child who would be genetically (99%) ours, but most importantly healthy. We reached out to the UK but unfortunately they are not currently taking international patients. It is unfortunate that we feel compelled to travel outside of Australia but every parent wants to have a healthy baby who will grow and develop into a happy child and learn and experience life to the fullest. For those who may believe that we are modifying babies or playing with nature, we don't want to play God or design our own child, we just want a healthy child that we ourselves have produced.

Mitochondrial disease is not a single disease. It is a multi-organ disease, an umbrella of diseases/disorders that greatly impact on the person and those close to them. Mitochondrial donation would have a massive impact on the health system and potentially reduce the prevalence of many diseases such as dementia, epilepsy, hearing loss, vision loss, intellectual disabilities, Parkinson's, Alzheimer's, atypical autism etc. Mitochondrial disease is the 2nd most commonly diagnosed, serious genetic disease after cystic fibrosis (AMDF).

Having worked in the disability industry for many years, I have witnessed firsthand the challenges people with disabilities face. The daily struggles and high care needs are often unsupported or provided by many who don't truly understand the person. The financial pressures that people with mitochondrial disease experience include constant visits to endocrinologists, cardiologists, neurologists, physiotherapists, psychologists and other professionals, as well as medication and transport costs etc. While the introduction of the NDIS may assist to coordinate services for people, this disease is still misunderstood and often takes years to diagnose. This results in a strain on the medical system for many more years than necessary, with contraindicative treatment being provided due to misdiagnosis.

Having witnessed the struggles, pain and suffering that my mother and brother went through, really hit home for me and made this awful disease far more real. I fear the future and my path ahead, hoping that I don't suffer the way they did. No one should have to say goodbye to their own child, nor should a person be faced with so many physical and emotional challenges that see their lives fade before them. This procedure can reduce much of that heartache and suffering. This procedure could make people's dreams of having a child come true. It appears that much of the groundwork has already been in the UK. This is not modifying a child for any other benefit than for the child to be healthy. Why would that be frowned upon? This disease is not a choice, the sometimes deadly consequences of this disease are not chosen. Sufferers see the devastation in the loss of relationships and social engagement. The inability to work, get out of bed each day, inject with insulin 4 times a day, wearing hearing aids, using a wheelchair, not being able to speak properly or suffering from

stroke effects, inability to get yourself off the toilet etc. These are all very real and daily occurrences for many people as they progress through mitochondrial disease.
My experience of this disease has opened my eyes to many aspects of the health care system, the impact on the person, the carer, the family, the repercussions in terms of not having a will, insurance challenges, grieving death, life and the future. Being able to prevent the multiple impacts on people's lives would be amazing.

Naturally, mitochondrial donation in Australia would require very careful planning and preparation. Safeguards would potentially involve ensuring that the patient is well enough to undergo the procedure of mitochondrial donation and pregnancy. This is standard for PGD IVF and my referral involved a cardiologist and endocrinologist being satisfied that this would be safe. Naturally counselling would be beneficial to both donor and couple. The procedures in place for current IVF would guide these safeguards. Selection criteria would be required to determine who would be a suitable candidate. I believe in the UK it is preferable for couples to have tried PGD beforehand. Screening of egg donors in terms of medical history would also be required. As with any procedures, mitochondrial donation would be suitable for particular cases, with the intention to prevent severe forms of this disease from being inherited.

Personally, my partner and I would jump at the opportunity to undergo a procedure that allowed us to have a healthy child. Having this procedure available in Australia would allow us to complete our family and have peace of mind that we are giving our child/ren the best opportunity of life possible. We thank you for your time and consideration of this procedure in Australia. We appreciate the complexity of this case and acknowledge the level of work involved in conducting this enquiry. Many thanks for your consideration.

Kind regards,