SUMMARY: [redacted] are parents of two children who died of mitochondrial disease, their oldest son [redacted] who died aged 7 and their youngest daughter [redacted] who died a week short of her 30th birthday.
This is their story.

SUBMISSION: to the Senate Community Affairs Committee inquiry into the science of mitochondrial donation.

May we thank the Senators of this Committee of Inquiry for hearing our story.

We had five children, two of whom died of mitochondrial disease. On each occasion their illness put our family through years of disruption, dashed hopes, heartbreak and utter despair.

Our oldest son, [redacted], presented in 1980 at the age of four with intermittent rhythmic convulsions of his right hand. These became progressively more frequent and more violent until he died three years later.

When a precious seven-year-old son, his frail body wracked by convulsions, asks if he's going to die what does one say? [redacted] knew, and told us what he wanted to do before he died - another visit to the Snake Hut at Taronga Zoo, a special cake for afternoon tea and much more.

Treatment for his convulsions at that time required rushing him to the Children's Hospital in Camperdown, Sydney, where the doctors would render him unconscious. Apparently it was the only reliable way of bringing the convulsions under control. The convulsions could start anytime without warning so [redacted] demanded our care and attention 24 hours a day. This completely disrupted our family life and resulted in ongoing neglect of our younger children. [redacted] gave up work completely and [redacted] curtailed his work-time considerably during these three years.

Owing to [redacted] medical background and [redacted] paediatrican Professor Robert Ouvrier, we had access to the best medical experts from around the world. Despite this however, at the time of his death, the cause of [redacted] convulsions remained unknown.

Twenty-five years later in 2007, our youngest daughter [redacted], who was 20 and at university, developed similar convulsions in her right shoulder. Professor Ouvrier was then able to tell us that in retrospect [redacted] had almost certainly died of mitochondrial disease and that [redacted] most likely had the same. It was our worst nightmare come true; and the first time we heard the term mitochondrial disease or mito.

Subsequently, [redacted] had a muscle biopsy which confirmed the diagnosis and our hell started in earnest all over again.

Gone were her hopes of a normal happy life, a career, marriage and having children. Instead [redacted] faced a life of overwhelming tiredness, progressive isolation from her friends and increasingly degrading disability. She was unable to work, drive a car or socialize. Eventually she lost all independence and could not cook or care for herself. One of us, usually her mother, had to be with her constantly.

[redacted] seldom managed a day without twitching or convulsions. She gradually lost strength in her limbs, walked clumsily and loathed the frequent comments from strangers suggesting she was a druggie or had been drinking. Her greatest wish was to go out with her friends and be normal again.

By 2010 much more was known about mitochondrial disease. [redacted] convulsions were initially reasonably well controlled and new drugs offering treatments were in the pipeline. What kept her going, and what really gave us hope, was the understanding that mitochondrial science was developing at an increasingly rapid rate, and there was a real chance some form of treatment, or even a cure, would be found. As an example, when [redacted] had her diagnostic biopsy in 2007, it took the Murdoch Children's Research Institute approximately six months to sequence one single gene, reportedly at a cost of about $15,000. Ten years later it's now possible to sequence all 22,000 nuclear genes and 37 mitochondrial genes more accurately in just a few days, and for just a few hundred dollars.

By attending to [redacted] every symptom and preventing irreversible damage, we hoped that gene therapy, protein therapy, stem cell therapy or some other form of a treatment would be found in time.

Unfortunately for us, time ran out. [redacted] died last year. Her 10-year ordeal is now over – only her memory and a deep empty sadness are left.

Our remaining children have had their DNA sequenced and they know they won't develop mitochondrial disease so in that sense the uncertainty for our family is over. But for other families with mito around Australia who want to have children, their nightmare is only beginning. The constant anxiety and sense of hopeless foreboding at knowing that their precious child could be affected is overwhelming.

Mitochondrial donation offers a real chance to end this nightmare. It can give others an opportunity to experience the joys and sorrows of a full life that [redacted] and [redacted] couldn’t.

We urge you to take the opportunity while it presents to make mitochondrial donation legal. We believe it offers a real opportunity for women with mtDNA mutations to have healthy babies and to not pass on the curse of this truly horrible disease.

Thank you,