

Attention:

Committee Secretary

Senate Community Affairs Committee

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May 6th, 2018

Dear Committee Secretary,

On the morning of March 5th, 2007, I awoke to our gorgeous, 8yr old redheaded daughter, having a seemingless innocent bout of gastro. Sadly, 24hours later and still in the depths of seizures and a stroke, she was diagnosed with MELAS, one of the most common forms of mitochondrial disease. Two weeks later, the genetic diagnosis was confirmed (3243A>G mutation), and confirmed later in our other 2 children and myself as well. Our world had changed.

As a general practitioner, mitochondrial disease just was not taught, and nor did I ever realise that such a horrific multi-system progressive illness existed, and now it was our lives. A conservative incidence of 1 in 200 Australians was inconceivable at first, but after extensive volunteering with the AMDF and running their helpline 24/7 for 7years (2009-2016), the impact of this illness and the path of devastation it left in so many lives, and continues to do so, was simply stupefying. So many of the hundreds of patients and families I spoke to over those years, were like us, mortified, existing to survive now rather than live. Trying their utmost to create as many precious memorable moments, always looking for the smile in their child's or partner's face.

Nearly 10yrs later to the day, some 20+ strokes later, our daughter lost her battle with mito, resting now in the loving arms of her Lord. She had gone from the flourishing 8yr old, full of hope, dreams and motivation, to having dementia, deafness, partial blindness, tube feeds, incontinence, immobile, seizures, diabetes, multi-organ failure, dysphasia and extreme fatigue. The impact of her illness on her younger brother of one year, who was her best friend, can **NEVER** be measured. Now with **his** MELAS also progressing quite rapidly (in the last 2-3 years), he must now live knowing the path which lies ahead of him, with a likely prognosis not extending much past 10yrs as well, with **NO CURE** or even a valid treatment in sight. For a young man nearly 18, trying to finish school and plan his future, it is a lot to take on. Imagine if it was your child, what would you say.....

Also, not forgetting our youngest daughter, now 12yrs old, watching her big sister deteriorate is all she knew growing up, and the separations and restrictions that mitochondrial disease brought to the entire family. How many 12yr old daughters are already saying that they don't think they want children because they fear they will die like their sister, knowing she will pass on the same genetic mutation **100%** to every child she ever has?! She has little choice but to think that having her own healthy child is not achievable.

As for my husband and myself, we could NEVER have envisaged some 11yr ago, that our 3 "healthy" kids were to be so impacted by an illness that we'd never even heard of or even have dreamt up. An illness that is so painfully and slowly progressive, and is like dying slowly from 4+ illnesses at one time over years and years. An illness that cruelly makes you pay for every good day you have had by being followed up by a "crash" or even another stroke the next day, always on tender hooks waiting for the next downturn. When the news first came a few years ago of the UK changing their legislation to allow mitochondrial donation, I will never forget where I was, as it was a sudden light shining bright in the midst of riding a dark merry-go-round. A chance that this vicious cycle could be broken in other families, or even our youngest daughter's life was intensely uplifting.

Some years before that UK announcement, when I first heard of mitochondrial donation, I thought deep and hard about the personal, religious and medical implications of the technique and its impacts. Being a Christian, the line between playing God and using gifts from God can be blurred in most areas of medicine I have dealt with over the years. All the way from the most basic of treatments to end stage palliative care,

to future potential cures and genetic modification. After much investigation and searching, I personally would prefer the “maternal spindle” transfer technique over the “pronuclear” transfer as I would rather avoid the production of unwanted embryos. However, this aspect is an old argument in Australian legislation as the excess production of human embryos has been happening for many years in the IVF world, and is therefore a moot point.

As for the argument of genetic modification or manipulation, when the genetic transfer during an organ transplant is questioned, then scientists and ethicists might have the right question our mitochondrial “mini-organ” or organelle transplant. Surely the cystic fibrosis (CF) patient receiving a new lung with healthy cells carrying healthy genes from TWO other people, may never be accused of genetic manipulation. Sadly, any child I had under a mitochondrial donation technique will still have the same untouched 46 chromosomes, the same bad traits as me and my same ugly looks!! The only genes touched are the ones within the mitochondria and ONLY influence it, just like the CF lung transplant.

Will all this affect future generations or germ lines? I cannot answer that, **HOWEVER** nor can the organ transplant patient, consuming multiple highly toxic immunosuppressants daily, know how it affects their own eggs or sperm when they conceive. The same applies to the leukaemic child after massive doses of chemotherapy and possible bone marrow transplants, they know not the risks to their future germ lines as well. The same can be applied to all medicines and treatments in medicine, we may not grossly see a change now or next generation but we cannot see what lies 3 to 4 generations ahead. Our Western lifestyle has had many documented impacts on our health and well-being but still we practice it, despite rising diabetes, obesity and vascular disease to name a few. **WHY SHOULD** all others be offered their treatment options but not a generation of mitochondrial patients who cannot escape passing on the genetic mutation to their children?? What have we done to be disregarded? When our cultural, scientific and medical worlds can **ALL** be 100% confident that every treatment, medicine, diet, processed food, chemical spray, etc has **NO impact at ALL on ALL future generations** then they **MAY** have an argument against mitochondrial donation.

I have always believed that you can never fully understand a person till you have walked every step with them in every moment of their lives. As much as I cannot fully understand the journey of a dying cancer patient nor a person with Parkinson's, our journey with mito can never be fully comprehended either. But the numbers speak for themselves, 1 in 200 Australians is a large number of people affected or potentially affected or to have their future offspring affected, by a severely debilitating multi-system illness that drains the health dollar..... so doesn't it financially make sense to keep these future generations “disease-free” too?

The personal, emotional, social and financial impact of the illness within our family, the loss of a daughter at age 18 after a 10yr battle, the decline in our 17yr old son who is potentially repeating the same battle again, and to look into the eyes of our 12yr old daughter who wonders if she is next and not wanting a child of her own dying the same way, is an impact I even find hard to imagine and repeatedly pinch myself, hoping to awake from this “dream”. The hope of allowing mitochondrial donation is truly a beacon, a chance that I believe the mito community deserves, rather than being cast aside.....would a potential cancer cure or organ transplants be cast aside if there was an immeasurable remote risk to future generations?

In conclusion, I would like to greatly thank the Senators of the Senate inquiry for their time and consideration of mitochondrial donation, and for opening discussions into the possibility of one day allowing the technique here on Australian shores. Our family awaits with abated breath to see what our future holds.

Yours sincerely,