

SUBMISSION TO THE NUFFIELD COUNCIL FOR BIOETHICS

Preventing the Transmission of Inherited Mitochondrial Disorders.

Submission from Family member

1. My niece
2. Meddling with nature.
3. Psychological effects
4. Transplants
5. Appeal to the Nuffield council for bioethics

1. My niece has "MELAS", a devastating, horrendous syndrome, which has resulted in development of symptoms from the age of 15 years, until the present, she is now 29. At first she appeared normal, small in stature, but big in personality and character, fun loving, generous, kind. She has always had intestinal problems and problems with eating, but at age 15 she had an episode of status epilepticus, and since then her symptoms have deteriorated involving episodes of strokes, severe migraines, optical disturbance, gradual decline of cognitive skills and motor function, profound deafness, further episodes of status epilepticus until the present; she now suffers intractable seizure-like episodes and drop attacks on a daily basis; she cannot read or spell anymore, she cannot be left on her own, she has become a shell of her former self. She bravely finished school and went to college, however the episodes became more frequent and after each episode, she fought back, only to be struck down again by some new manifestation of this terrible disease; she fought back again, and again, and was struck down each time. At first we mistakenly believed we could find some way to cure her, we were in denial, surely, we believed, there must be something that could be done. The diagnosis of MELAS after several years left us with little hope, but there is now hope for others. To watch this vibrant child be totally destroyed by this disease is more than any relative and especially parent should have to bear; I have also watched the lives of my sister and her husband and other daughter be ruined by this disease; only through their constant vigilance and care have they, on many occasions together with dedicated medical professionals, pulled my niece back from the brink of death, only to have to watch her suffer further.

2. Meddling with Nature. -Perhaps I am being over simplistic, but every manmade thing has usually some toxic effect or by product. Paradoxically, this new proposed research/preventative treatment is about an ability to "fix" and improve something in nature which has gone horribly wrong. I am an ordinary person who doesn't think much about ethics and morality, but I view the arrogance of persons who claim to protect the unborn child and be against this miraculous pioneering technology as staggering - if successful, this procedure seeks to protect the unborn by the elimination of a set of diseases which at best are worrying and frightening, and at worst, are totally devastating to a young life, resulting in our case in the gradual destruction and death of a much loved, loving and talented child

3. Psychological Effects -The desire for children is inherent in most individuals. My niece has a younger sister who also has MELAS, so far she has remained largely symptom free, and if this treatment is given permission it will give hope for the first time to her, of having her own healthy children, and hope also to countless other families. I do not see any significant psychological effects for children born using this procedure, if they indeed need to be informed at all. We do not tell our children everything but I am sure if someone was at an age of understanding, knew they were much wanted, knew their parents took advantage of this preventative transplant mechanism in order for them to be spared the horrendous consequences of these mitochondrial disorders then they could only be glad. As a parent, if I knew I carried this defect, I would travel anywhere in order to find a cure or prevent and protect my child from suffering these diseases.

4. Transplants and Ethical concerns - I do not really feel qualified to comment as to the ethics of this procedure, however as I understand it, inherited characteristics are governed by the nuclei of parent cells, and in the proposed procedure the nucleus is intact. Any transplanted genes, small in number, seem to be of concern for mitochondrial function only, therefore I would liken this to a mitochondrial transplant, and I do not see an ethical dilemma, or the need to say any resultant child has 3 parents. This seems to me like a transplant procedure with a donor, the difference being that no-one has to die in order for the transplant to occur, as in the case of e.g., a heart transplant. I also see a danger of "paralysis by analysis", in that we become so bogged down by the whys and wherefores that we lose track of the initial goal, that is, elimination of a set of devastating conditions with little or no risk to third parties.

5. Appeal to the council for Bioethics -My little niece is the bravest person I know, she has battled against the devastating effects of this disease for half her short life, to no avail. Mutations in nature occur, in this instance there seems to be a way of preventing the cataclysmic effects of some of these negative mutations- to know that there is a preventative technique to eliminate this type of disease and to do nothing would be a travesty, there may be problems along the way, but nothing could be worse than watching this "living death", this deterioration and suffering in a talented beautiful child who never had a chance to properly live; and to know that it could have been prevented. Please do not let my niece have suffered in vain, by wasting this opportunity.

I would also like to thank Doug Turnbull and his team for the genuine compassion and support shown for my family over the past number of years.