This response was submitted to the call for evidence by the Nuffield Council on Bioethics on *Emerging techniques to prevent inherited mitochondrial disorders: ethical issues* between January 2012 and February 2012. The views expressed are solely those of the respondent(s) and not those of the Council.

# Carolyn Appleby – Techniques to prevent inherited mitochondrial disorders – response.

My submission regarding the recent advances in mitochondrial research and how this will be very beneficial to me and members of my family.

My family and I are absolutely delighted with the recent funding and support for the research into mitochondrial genetic diseases. I am a 46 year old lady who inherited this devastating and life changing genetic defect from my mother. It has also been proved that I have also passed a form of this genetic defect on to my 2 sons now aged 22 and 19 years old.

Maybe in years to come if the research into mitochondrial genetic diseases is allowed to go ahead as planned by professor Doug Turnbull and his team at Newcastle, this genetic default may be one day eliminated in order to help future members of not just my family but other families who have been unfortunate to contract one of the many forms of this mitochondria.

At this moment in time some of the health issues that I have that is attributed to my mitochondrial disease are:-

- Problems with shortness of breath upon undertaking activities
- Problems with swallowing certain foods and digestion
- Chest infections
- Problems with vision
- Weak arms and legs
- Fatigue on a daily basis being a major factor, severely restricting my day to day function
- Other medical issues are made worse because of these factors, one of which could be the fact that I now have a heart condition but this cannot be attributed to the mitochondria yet.

As of yet my 2 sons do not have any obvious symptoms of the mitochondria but that may change as they both become older, following in their mum's footsteps. That is why we as a family are so hopeful that this pioneering research is allowed to go ahead and receive as much funding and positive backing as is possible so that my boys may one day have a family of their own with the peace of mind that one day this debilitating genetic disease will be eliminated altogether or controlled to such a degree that symptoms of the disease are prevented from escalating.