

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.

Anonymous 15

1. Current understanding and background to mitochondrial DNA disease and its effects
2. Current options available
3. The benefits of nuclear transfer techniques to prevent transmission of mitochondrial DNA disease to future generations
4. Summary

1. Currently there is a relative lack of treatments available for individuals affected by mitochondrial disorders, which affect many of the major organ systems and can seriously impact upon health. Furthermore due to the way in which we inherit our mitochondria from our mothers affected women planning to start a family face the excruciating decision when thinking about starting a family. At present genetic counselling is predominately used in order to help patients understand the potential risks posed to future children they may bare, which is usually heavily dependent upon the mutation they carry. Through genetic counselling they are usually also made aware of genetic screening procedures which are available to help assess the likelihood of having an affected child which can be performed either prior to or following establishment of a pregnancy.

2. However for those select group of patents who carry high mutation loads, genetic testing whether it be through prenatal genetic testing (PND) or preimplantation genetic testing (PGD) are relatively invalid options as any child they bare is likely to also inherit a similar high mutation load. Therefore techniques which would allow for a lady with a high mutation load to have a child which is genetically her own offers great hope to families which have been affected by mitochondrial disease for several generations.

3. The use of nuclear transfer techniques to prevent the transmission of mitochondrial disease to future generations can therefore in my opinion only be viewed as a helpful intervention to help affected couples have healthy children. All children born would carry the genetic information from their two parents and <1% of their total genetic information (mtDNA) from an egg donor, which would predominately contribute genes which are involved in energy generation. What is more is that this technique would allow for healthy babies to be born to families which have been blighted by mitochondrial disease for successive generations. The prospect that preimplantation sex selection could also then be offered to parents wishing to limit the potential risk to future generations is another question entirely and would most definitely have to be treat in its own right most likely dependent upon the patients situation and circumstances at the time.

4. In summary I believe that the use of nuclear transfer techniques to help prevent transmission of mitochondrial disease to successive generations in affected families should most definitely be considered a worthwhile and beneficial procedure. Intervention through this technique could potentially allow for mitochondrial DNA disease to be banished for generations to come in affected families, which could only be viewed as a positive outcome.