

Genetic Interest Group HFE Bill Briefing

Interspecies Embryo Research

We believe that the cytoplasmic hybrid technology is promising research that deserves to be explored by the scientific community. Work with cytoplasmic hybrid embryos has significant potential to deliver cures and treatments for patients suffering from currently intractable or incurable conditions.

The objective of the use of cytoplasmic hybrids is to create stem cells. Once a stem cell has been harvested, it can be made to divide and produce more identical stem cells. Stem cells can be persuaded to specialise into different cell types e.g. nerve cells, or muscle cells. This property can be used to examine how a disease under study affects these cells. Once the mechanism of the condition is understood, potential treatments and cures can be developed.

The technology allows researchers to create embryos without using the valuable and scarce resource of donated human eggs. GIG believes that this work deserves to be permitted because of its enormous potential to help people whose daily lives are compromised by serious medical conditions. Our members look towards this research with hope, and even expectation. Their strongly held view that this avenue of research should be investigated fully, is a product of their (or their families') healthcare situation: many live with incurable or intractable conditions.

The true potential of this technology will only be understood as research develops. Currently cytoplasmic hybrid embryo research holds massive potential for greater understanding of degenerative neural disorders such as Parkinson's Disease, and Motor Neurone Disease.

GIG would like to see this research permitted under the same level of regulation that the HFEA currently provides to research on embryos.

Research and Potential Treatments for Mitochondrial Cytopathies

Mitochondria are a component of most cells in the human body, essential for metabolism of food into energy; they are often referred to as "cells' powerhouses". Their components are coded for by DNA held both in cell's nuclei (where 99.9% of all human DNA can be found) and by their own extremely tiny quantity of DNA: mitochondrial DNA. Mutations in any gene related to mitochondrial components can lead to devastating conditions; those occurring in mitochondrial DNA are further complicated by unusual inheritance patterns. Conditions are subject to "variable penetrance", where it is difficult to predict the seriousness of a condition.

Mitochondrial Cytopathies are a wide range of disorders, many of which are supported by member groups of GIG.

- **Muscular Dystrophy**
Many Muscular Dystrophies are caused by mitochondrial dysfunction. Muscular Dystrophies are disorders that cause progressive weakness in patients and can lead to premature death. Patients with Muscular Dystrophy are supported by our member group:
Muscular Dystrophy Campaign: www.muscular-dystrophy.org

- **Retinitis Pigmentosa**
Mitochondrial dysfunction has been linked to some forms of Retinitis Pigmentosa, a condition in which the patients' retinas degenerate, restricting vision, leading to tunnel vision and eventually blindness. Support is provided to families with Retinitis Pigmentosa by our member group:
The British Retinitis Pigmentosa Society: www.brps.org.uk
- **CLIMB: Children Living with Inherited Metabolic Diseases** provides support, advice and information to families diagnosed with metabolic disorders, many are mitochondrial cytopathies.
CLIMB: www.climb.org.uk

Mitochondrial dysfunction has also been found to be linked with Alzheimer's disease, heart disease, Parkinson's disease, epilepsy and diabetes.

The best potential cure for this group of conditions is pronuclear transfer, a method which allows the donation of healthy mitochondria. The new HFE Bill allows pronuclear transfer for research purposes only, with a provision for later regulation to allow clinical application.

GIG would like to see the HFEA empowered to license clinical applications of this research, if and when this becomes feasible. The HFEA as an expert group, is well qualified to assess the potential efficacy of this new technology and its safety.

Pre-Implantation Genetic Diagnosis

Pre-Implantation Genetic Diagnosis (PGD) is an important technology that mitigates the devastating effect an inherited condition can have upon a family. It provides a means by which embryos fertilised in vitro can be selected on the basis of not carrying an inherited disorder; and is a safe alternative to the invasive method of ante-natal diagnosis which carries its own risk of miscarriage.

Currently the technology can be used by families with Cystic Fibrosis, Duchenne Muscular Dystrophy, and Huntington's Disease amongst many more. All of the conditions named are serious, life limiting disorders with no cure available.

As the Bill currently stands, this technology is applicable to "serious" conditions. However, it is not possible to draw up a list of conditions deemed sufficiently "serious" as to permit intervention. This is because families' perception of seriousness will differ according to their situation and their experience, and because diseases and disorders vary tremendously in their manifestation. Even apparently treatable conditions such as cleft lip or palate may in some cases be so severe as to be devastating in their impact, and in some cases may be associated with other abnormalities in a syndrome with far reaching consequences for the child. Any list will inevitably be subjective and arbitrary, imposing unnecessary and over rigid constraints where decisions should better be left to clinicians and couples to reach together in the full knowledge of all the relevant facts.

GIG would like to see Pre-Implantation Genetic Diagnosis continue to be supported alongside other diagnosis techniques.