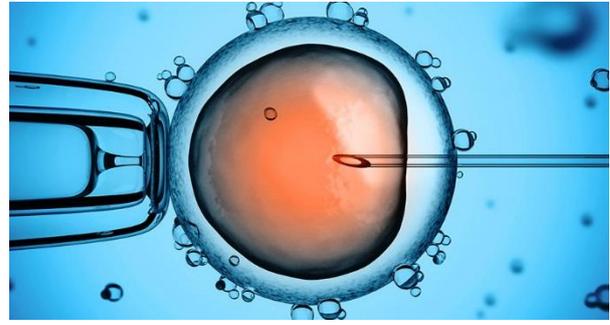


Case study: Genome edited human embryos

Purpose: Using genome editing to correct a genetic mutation in human embryos that can be passed down from parents. One example of this is a gene that causes a thickening of the heart muscle (hypertrophic cardiomyopathy), which may result in heart failure.

Koya79/iStockphoto



The global challenge

Cardiomyopathy is one of many inherited conditions that are passed down to a child from its parents. More than 10,000 inherited diseases might be prevented by correcting harmful genetic mutations. Many such diseases are serious, such as cystic fibrosis, Huntington's disease, and breast cancer linked to mutations in the BRCA gene.

UK facts & figures

- In the UK, cardiomyopathy affects at least one in 500 people of all ages. A mutation in one of several genes can lead to sudden cardiac arrest even in athletes. In some cases the mutation is "dominant" meaning that a single copy (allele) of the abnormal gene is sufficient to cause the problem.
- A recent study suggested that it was possible to use genome editing to correct a dominant mutation in embryos fertilised by sperm from a man with cardiomyopathy. This needs to be verified and further checks on safety must be carried out; therefore it would still be several years before treatments could be available in the UK, if it was deemed to be an acceptable use of the methods.
- It is currently illegal in the UK to implant a genetically altered embryo into a woman's womb; regulators in the US and many other countries also disallow the procedure.

Arguments made in favour of developing genome edited human embryos

- Allow individuals or couples carrying versions of genes that will definitely give rise to genetic disorders, such as Huntington's disease, to avoid passing them on to their children and to subsequent generations
- Reduce, in offspring, the risk of a disease associated with a specific gene variant, e.g. *BRCA1* and breast cancer
- Reduce the lifetime costs of treating people with genetic disorders
- Research involving edited embryos could improve understanding of the role of specific genes in early human embryo development. This basic knowledge could help to improve IVF success rates, and perhaps reduce miscarriages.

Arguments made against developing genome edited human embryos

- Genome editing to improve basic knowledge and/or to avoid disease could open the door to use the methods to make 'designer babies'
- The techniques might unintentionally introduce errors that could put future generations at risk
- The money for this research is better spent on developing cures for people living with genetic conditions

The alternatives

- Parents can choose to adopt, or have gamete (sperm or egg) or embryo donation with the loss of a direct genetic connection to the child.
- Potentially harmful versions of genes can often be identified by screening embryos in the lab following IVF (in vitro fertilisation) – a method termed preimplantation genetic diagnosis (PGD). It is then possible to implant only embryos without the abnormal genes into a woman's womb. However, PGD is not always successful, because it relies on having sufficient numbers of embryos to screen and find at least one that is developing well and lacks the abnormal gene(s). Moreover, in some cases it can't be used to avoid a child developing a genetic disease if one parent only has copies of a gene that causes genetic disease, even if the child receives a non-disease causing copy of the gene from their other parent, or if both parents only have disease causing variants of a gene.
- Alternatively, people with a known predisposition to heart disease can be carefully monitored throughout their lives, can avoid over-exertion and can take medication to help regulate their heart rate.