Mythbusting Mitochondrial Donation

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Claim

Children with donated mitochondria have three parents.

True or false?

False.

Why?

For the following three reasons.

1. Unlike nuclear DNA, mitochondrial DNA is not thought to encode individually distinguishing characteristics. Instead, it encodes (part of) the *generic* characteristic of having functioning mitochondria.

2. The amount of DNA that a child will inherit from a mitochondrial donor is very small (but see the caveat below).

3. 'Parent' is a long-established term that does not capture a new type of genetic relationship – involving mitochondrial DNA *without* involving nuclear DNA – that has become possible only very recently.

Claim

Children with donated mitochondria inherit only 0.1% (or 0.01%, or similar figures) of their DNA from the mitochondrial donor.

True or false?

Roughly true, but with a caveat.

Why?

When comparing the relative proportions of mitochondrial and nuclear DNA in a person, you must decide the following.

- What exactly you are counting in the DNA. For example, are you counting 'classical' (protein-coding) genes, genes defined more broadly, or base pairs?

- Whether you are comparing genomes, or raw quantities of DNA. This makes a difference, because a typical cell in the human body contains a single copy of the nuclear genome, but multiple copies of the mitochondrial genome.

Using any of these measures, the proportion of mitochondrial DNA to nuclear DNA in a person is vanishingly small. But if you give a very specific figure for this proportion, then you should specify how you arrived at the figure.

Claim

Mitochondrial donation involves moving material (DNA) contained within the nucleus of a parental egg or embryo, into a donor egg or embryo.

True or false?

Almost true, but the term 'nucleus' is misleading, and whether an egg or an embryo is involved will depend on the specific technique used.

Why?

There are several different mitochondrial donation techniques. Of these techniques, only the following two may legally be used in treatment in the UK at present.

- The technique most likely to be used in the UK involves moving material shortly *after* the mother's egg has been fertilised, to create a single-celled embryo.

  This technique is called **pronuclear transfer**. It involves taking nuclear material from one embryo (created from the mother's egg and the father's sperm), and moving this material to another embryo (created from the mitochondrial donor's egg and a sperm) that has had its own nuclear material removed.

  The resulting embryo – which now contains nuclear material from the mother and father, and mitochondria from the donor – is then considered for transfer to the mother's womb.

- The other technique that is permitted in the UK involves moving material shortly *before* the mother's egg is fertilised.

  This technique is called **maternal spindle transfer**. It involves taking nuclear material from an egg (the mother's egg), and moving this material to another egg (a mitochondrial donor's egg) that has had its own nuclear material removed.

  The resulting egg – which now contains nuclear material from the mother, and mitochondria from the donor – is then fertilised with the father's sperm to create an embryo. This embryo is then considered for transfer to the mother's womb.

Although nuclear *material* is moved during both of these techniques, this material is *not* contained within a single nucleus. This is because the single nucleus that would ordinarily be present in a cell is temporarily absent during these particular stages of reproduction.

Instead, the material that is moved during these techniques consists of *two pronuclei* (in the case of pronuclear transfer) or *one maternal spindle* (in the case of maternal spindle transfer).

Claim

Mitochondrial donation involves replacing *all* of a mother's mitochondria with a donor's mitochondria.

True or false?

Almost true – this is certainly the *objective* of mitochondrial donation, but in practice it is not always perfectly achievable.

Why?

The aim of mitochondrial donation is to try to replace a mother's original mitochondria (containing disease-causing mutations) with a donor's healthy mitochondria.

In practice, however, a tiny quantity of the mother's original mitochondria is likely to be 'carried over' with the nuclear material during mitochondrial donation. Scientists and doctors working on mitochondrial donation are looking for ways to reduce this 'carryover' as much as possible, but they may not be able to eliminate it completely.

Additionally, it is possible for the tiny quantity of 'carried over' mitochondria to start to increase in quantity, in a subsequent embryo or child (this phenomenon is sometimes called 'reversion'). If this happens, then it could pose a risk of mitochondrial disease. The reasons why this happens, and whether and how it can be avoided, are still being investigated by scientists and doctors.

Claim

Mitochondrial donation is also known as MDT/MRT/PNT/MST.

True or false?

All true, in certain contexts.

Why?

Mitochondrial donation is a term that has been used in science, medicine, law and regulation, particularly in the UK and also in Australia. This term can describe a female donor deciding that she wishes to provide the donor egg that will be used in the processes described above (where nuclear material is moved from one embryo or egg to another), or alternatively it can describe the processes themselves.

Mitochondrial donation treatment – or the acronym, **MDT** – is a variation of the term 'mitochondrial donation', that has been used by the UK body that regulates this area (the **Human Fertilisation and Embryology Authority**) and by others. It describes the processes described above (where nuclear material is moved from one embryo or egg to another), and also describes the way that these processes are offered as a treatment option, to people who are at risk of passing mitochondrial disease to their children.

Mitochondrial replacement therapy and **mitochondrial replacement technique(s)** – or the acronym for both of these terms, **MRT** – are similar terms to 'mitochondrial donation', that are often used in scientific and medical circles. Again, these terms describe the processes described above (where nuclear material is moved from one embryo or egg to another), and also describe the way that these processes are offered as a treatment option, to people who are at risk of passing mitochondrial disease to their children.

Finally, the two mitochondrial donation techniques that are legally permitted in the UK are also sometimes referred to using acronyms. **Pronuclear transfer** is sometimes referred to using the acronym **PNT**, and **maternal spindle transfer** is sometimes referred to using the acronym **MST**. These terms are used to refer to specific techniques, rather than referring to mitochondrial donation as a whole.

Claim

A child born in Mexico in 2016, following mitochondrial donation, was the first person ever to inherit DNA from three different people in the previous generation.

True or false?

False.

Why?

17 children were born following the use of an experimental fertility treatment technique in the USA between 1996 and 2001, which involved introducing some mitochondria from donated eggs into the mothers' eggs, resulting in a mixture.

The technique used in that instance was very different to the mitochondrial donation techniques discussed above, but it did result in the 17 children inheriting DNA from three different people in the previous generation. Therefore, the child born in Mexico in 2016 was not the first person to have such inheritance.