New technique could eliminate inherited mitochondrial disease

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Researchers funded by the National Institutes of Health have developed an experimental technique with the potential to prevent a class of hereditary disorders passed on from mother to child. The technique, as yet conducted only in nonhuman primates, involves transferring the hereditary material from one female's egg into another female's egg from which the hereditary material has been removed.

The resultant eggs, which were fertilized with donor sperm, implanted in females and carried to term, produced offspring free of the mother's mitochondria, but which instead possess the mitochondria from the donated egg cell. Mitochondria are tiny structures within cells that help provide energy to power the cell's activities.

They are passed on from mother to child, in the fluid (called cytoplasm) contained inside the egg cell. In recent years, defects in mitochondria have been linked with a variety of conditions, such as diabetes, cancer, infertility, and such neurodegenerative disorders as Alzheimer's, Parkinson's and Huntington's diseases. The technique raises the possibility that mitochondria associated with a hereditary disorder could be prevented from being passed on to the next generation.

"Recent findings suggest that mitochondrial disorders play a role in at least some proportion of many human disorders," said Duane Alexander, M.D, director of the Eunice Kennedy Shriver National Institute of Child Health and Human Development, which provided funding for the study. "Pending further research, the findings hold the potential of allowing a couple to have a child who is biologically their own, but is free of any conditions associated with defects in maternal mitochondria."

Mitochondria are passed on to subsequent generations only through egg cells and not transmitted through sperm. In addition to the DNA found in the chromosomes, mitochondria have their own DNA. Mutations in mitochondrial DNA have been associated with a variety of human disorders.

The study was conducted by researchers at the Oregon Health Science University in Beaverton and was published online in *Nature*.

Using the technique, the researchers created fertilized eggs and achieved three successful pregnancies in rhesus monkeys, which have resulted in four healthy newborns. Recent advances in the transfer of hereditary material and in microscopy facilitated the achievement, they wrote.

The researchers said that the technique did not appear to pose any risk of chromosomal damage. Analysis of 5-6-day-old embryos (blastocysts) resulting from the fertilized eggs, and of embryonic stem cell lines established from them, did not uncover any evidence of damage to the chromosomes. Analysis of cells from the infant monkeys born after the procedure failed to detect any mitochondrial DNA from the mother.

Source: NIH/National Institute of Child Health and Human Development (news : web)