Mothers curse linked to male infertility

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(Medical Xpress) -- Researchers have discovered the first real evidence of the 'mother's curse' and its connection to male infertility due to genetic mutations in mitochondria. Led by Dr. Damian Dowling from Monash University in Melbourne and Paolo Innocenti from Uppsala University, their breakthrough research has been recently published in *Science*.

All animals have two genomes, nuclear and mitochondria, which work together. However, they are inherited in a different way. The nuclear genome comes from both parents while the mitochondrial one is only passed down by the mother. Because of this, mitochondria passed down to males then face an evolutionary dead-end. In this way, changes in mitochondrial genes which can impair males can be passed down. This is what is known as the ‘mother’s curse’ or selective sieve.

In their study, mitochondrial DNA from fruit flies in five different countries were collected and then inserted into a group of flies which had identical nuclear genomes. To show that the mother’s curse is real, the effect of the mitochondrial genomes should be greater in males. Changing the mitochondrial genomes in females only affected seven genes; however, in the males it affected 1,172 genes, including 300 that mainly affect the testes or sperm glands.

Dowling says that while the experiment was done on fruit flies, their genomes are very similar to that of humans, and this same mutation can be linked to infertility in males.

More information: Experimental Evidence Supports a Sex-Specific Selective Sieve in Mitochondrial Genome Evolution, *Science* 13 May 2011: Vol. 332 no. 6031 pp. 845-848 DOI:10.1126/science.1201157

ABSTRACT

Mitochondria are maternally transmitted; hence, their genome can only make a direct and adaptive response to selection through females, whereas males represent an evolutionary dead end. In theory, this creates a sex-specific selective sieve, enabling deleterious mutations to accumulate in mitochondrial genomes if they exert male-specific effects. We tested this hypothesis, expressing five mitochondrial variants alongside a standard nuclear genome in Drosophila melanogaster, and found striking sexual asymmetry in patterns of nuclear gene expression. Mitochondrial polymorphism had few effects on nuclear gene expression in females but major effects in males, modifying nearly 10% of transcripts. These were mostly male-biased in expression, with enrichment hotspots in the testes and accessory glands. Our results suggest an evolutionary mechanism that results in mitochondrial genomes harboring male-specific mutation loads.