

Genetic defect suggests high blood pressure may come from mom

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A specific genetic defect in one Chinese family shows that high blood pressure was inherited from the female parent, researchers report in *Circulation Research: Journal of the American Heart Association*.

The defect results from a point mutation — the substitution of a single DNA "base" for another during replication — in the genes of a tiny cellular organ called the mitochondria, which generates a cell's energy. When reproductive cells come together to form an embryo, the mitochondrial DNA from the mother cell is passed on to the offspring. Evidence has suggested a mother-child inheritance link for high blood pressure due to mitochondrial inheritance.

The Chinese-led group, which also included researchers from the Cincinnati Children's Hospital Medical Center and the Medical University of Vienna in Austria, provides clinical, genetic, molecular and biochemical evidence that a mitochondrial mutation designated tRNA^{Ile} 4263A>G is associated with inherited high blood pressure. The DNA substitution researchers discovered is an adenine-to-guanine switch at position 4263 on the mitochondrial genome (4263A>G).

Geneticists identified a large family from northern China in which 15 of 27 members who descended from the same female ancestor had blood pressures above 140/90 mmHg even after treatment. Only seven of 81 non-maternal relatives had high blood pressure.

Researchers compared the family members with 342 Chinese residents

of the same northern area to confirm a maternal link. Analysis of the mitochondrial genome of the maternal relatives and other tests revealed the site of the hypertension-related mutation and showed that it impairs the mitochondrial respiration chain, which increases levels of a reactive oxygen species (i.e., free radicals).

The findings show that inherited mitochondrial dysfunction is involved in [high blood pressure](#) and may provide new insights into maternally transmitted hypertension, researchers said.

Provided by American Heart Association

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