

★ Mitochondrial DNA copy number (Guyatt, 2019)

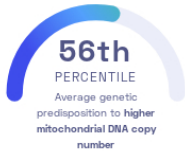
Anna Guyatt, et al.
Human Genomics

Heart Metabolism

STUDY SUMMARY

Discovery of 3 variants associated with mitochondrial DNA copy number.

YOUR RESULT



STUDY DESCRIPTION

The mitochondria have been nicknamed “the powerhouse of the cell” because they are responsible for producing up to 90% of a cell’s energy. Though mitochondria are part of a cell, they have their own, small genomes. Different types of cells in the body have different energy requirements, and as a result, have varying numbers of mitochondria inside them. For example, cells that require more energy for their functions, such as muscle cells, generally have more mitochondria. This genome-wide association study attempted to identify variants in the human genome that influence the number of mitochondria per cell, which was measured as mitochondrial DNA copy number. The study examined the genetic data of nearly 6800 individuals of European ancestry, and found three regions of the genome associated with mitochondrial DNA copy number. One of the implicated genes identified in this study was MED24 which plays a role in activating other genes to produce proteins.

DID YOU KNOW?

Heart muscle cells are thought to have more mitochondria than all other cell types, with roughly 40% of the cell’s mass being mitochondria.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to higher mitochondrial DNA copy number we summed up the effects of genetic variants that were linked to higher mitochondrial DNA copy number in the [study that this report is based on](#). These variants can be found in the table below. The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to higher mitochondrial DNA copy number. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to higher mitochondrial DNA copy number. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to higher mitochondrial DNA copy number. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for higher mitochondrial DNA copy number to be 0.00**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for higher mitochondrial DNA copy number is in the **56th percentile**. This means that it is higher than the polygenic scores 56% of people. We consider this to be an **average genetic predisposition to higher mitochondrial DNA copy number**. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our [Nebula Library tutorial](#).

| VARIANT [Ⓞ] | YOUR GENOTYPE [Ⓞ] | EFFECT SIZE [Ⓞ] | VARIANT FREQUENCY [Ⓞ] | SIGNIFICANCE [Ⓞ] |
|--|----------------------------|--------------------------|--------------------------------|---------------------------|
| rs12873707_C NEW | T / T | 0.16 (-) | 10% | 9.27×10^{-8} |
| rs709591_A NEW | T / T | -0.08 (-) | 37% | 1.54×10^{-7} |
| rs150387260_A NEW | G / G | 0.41 (-) | 5% | 1.65×10^{-6} |

