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Mitochondrial heteroplasmy (Nandakumar, 2021)

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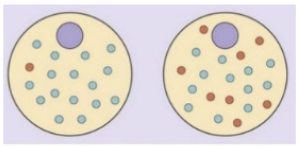
Science Advances

Metabolism | Mitochondria



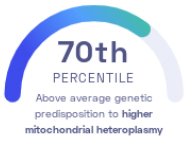
Low Heteroplasmy High Heteroplasmy

A single cell contains many mitochondria with many mitochondrial genomes. Heteroplasmy describes a state where the genomes of the mitochondria differ.

STUDY SUMMARY

This report is based on a study that discovered 20 genetic variants associated with mitochondrial heteroplasmy.

YOUR RESULT



Above average genetic predisposition to **higher mitochondrial heteroplasmy**

STUDY DESCRIPTION

Mitochondria are commonly known as "the powerhouse of the cell". They also have their own small genomes that are distinct from the nuclear genome of the cell. Mutations in the genomes of mitochondria can lead to a state called "heteroplasmy", which means that multiple versions of mitochondrial DNA exist within the same cell or person. Heteroplasmy is caused by mutations of mitochondrial DNA and can result in disease if the mutations disrupt the function of mitochondria. This study of over 980,000 individuals of European ancestry sought to identify regions of the nuclear genome that are associated with the rate of heteroplasmy of mitochondria. The researchers found 20 genomic regions that may collectively explain one-third of the heritability of mitochondrial heteroplasmy. One of the identified variants is located near the TFAM gene, which plays a role in the repair of mitochondrial DNA.

DID YOU KNOW?

The total number of mitochondria per cell varies greatly depending on the cell's function. Red blood cells are the only cells in the human body that don't have any mitochondria, while other cells, such as muscle cells, contain hundreds to thousands of mitochondria each.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to higher mitochondrial heteroplasmy we summed up the effects of genetic variants that were linked to higher mitochondrial heteroplasmy 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 heteroplasmy. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to higher mitochondrial heteroplasmy. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to higher mitochondrial heteroplasmy. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for higher mitochondrial heteroplasmy to be -0.09**. 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 heteroplasmy is in the **70th percentile**. This means that it is higher than the polygenic scores 70% of people. We consider this to be an **above average genetic predisposition to higher mitochondrial heteroplasmy**. 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	GENE	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs1049432_T NEW	G / G	TFAM	0.04 (–)	18%	2.00×10^{-223}
rs28539606_G NEW	A / G	HLA-DQB1	0.02 (↑)	15%	4.00×10^{-23}
rs28602228_T NEW	C / G	CSF2RA	0.01 (–)	37%	8.40×10^{-16}
rs4251979_T NEW	T / C	IL1RN	-0.01 (↓)	73%	1.20×10^{-15}
rs73081554_T NEW	C / C	RPP14	-0.01 (–)	7%	3.20×10^{-14}
rs12461806_G NEW	A / G	TINCR	-0.01 (↓)	91%	1.80×10^{-13}
rs370209610_T NEW	T / T	IFNL4	-0.02 (↓)	98%	6.30×10^{-13}
rs58678340_T NEW	NA	TWNK, MRPL43	-0.03 (–)	1%	3.20×10^{-12}
rs7319964_T NEW	A / A	KLF5	-0.01 (–)	54%	7.40×10^{-12}
rs2149642_T NEW	T / C	BMP2	0.01 (↑)	78%	1.50×10^{-11}
rs143803034_G NEW	G / G	CCZ1B	-0.01 (↓)	96%	1.80×10^{-11}
rs10063311_G NEW	C / C	NDUFS4	-0.01 (–)	22%	9.40×10^{-10}
rs4933661_G NEW	C / C	HECTD2	0.01 (–)	36%	2.20×10^{-9}
rs2286639_G NEW	G / A	SLC6A7	-0.01 (↓)	21%	1.00×10^{-8}
rs11064881_G NEW	G / G	CIT, PRKAB1	-0.01 (↓)	93%	1.40×10^{-8}
rs681343_T NEW	T / T	FUT2, MAMSTR	0.01 (↑)	48%	2.00×10^{-8}
rs11679052_G NEW	G / G	CYP26B1	-0.01 (↓)	58%	5.00×10^{-8}

N/A indicates variants that could not be imputed using the 1000 genomes project datasets and variants that have a frequency of < 5%. Your genome was sequenced at 30x/100x coverage and is not imputed. However, to calculate percentiles, we need to compare your data with other users imputed data. To make the data comparable, we need to exclude some of the variants from your data.