

Mitochondrial DNA copy number and brain MRI markers: Implications for brain health and neurological disorders.

Harry Warner*

Department of Neurosciences, University of Exeter, United Kingdom

Abstract

Mitochondrial DNA (mtDNA) copy number has been found to be associated with various health conditions, including neurological disorders. In recent years, researchers have explored the relationship between mtDNA copy number and brain magnetic resonance imaging (MRI) markers, which can provide important information about brain structure and function. This article reviews two studies that investigated the association between mtDNA copy number and brain MRI markers in older adults and individuals with mild cognitive impairment (MCI). The first study found that higher mtDNA copy number was associated with larger brain volume, particularly in the gray matter, and better white matter integrity in older adults. The second study found that lower mtDNA copy number was associated with smaller brain volume, particularly in the hippocampus, and greater white matter hyperintensities in individuals with MCI.

Keywords: Mitochondrial DNA copy number, Brain MRI markers, Gray matter, White matter, Brain volume, Mild cognitive impairment, Neurodegeneration.

Introduction

Mitochondrial DNA (mtDNA) is the genetic material found in mitochondria, the energy-producing organelles in our cells. The copy number of mtDNA has been found to be associated with various health conditions, including neurological disorders. In recent years, researchers have explored the relationship between mtDNA copy number and brain magnetic resonance imaging (MRI) markers, which can provide important information about brain structure and function [1].

A study published in the journal *Neurology* in 2018 investigated the association between mtDNA copy number and brain MRI markers in a large sample of older adults. The study included 1,164 participants who underwent brain MRI and had their mtDNA copy number measured from blood samples [2].

The researchers found that higher mtDNA copy number was associated with larger brain volume, particularly in the gray matter. Gray matter is the part of the brain that contains most of the neuronal cell bodies and is involved in processing information. The study also found that higher mtDNA copy number was associated with better white matter integrity, as measured by fractional anisotropy (FA). White matter is the part of the brain that contains myelinated nerve fibers, which facilitate communication between different parts of the brain. The findings of this study suggest that higher mtDNA copy number may be a marker of better brain health in older adults. However, the study was cross-sectional, which means that it only captured a snapshot of the relationship between mtDNA copy number and brain MRI markers at one point in time [3].

Longitudinal studies are needed to determine whether changes in mtDNA copy number over time are associated with changes in brain MRI markers and cognitive function. Another study published in the *Journal of Alzheimer's Disease* in 2019 investigated the association between mtDNA copy number and brain MRI markers in individuals with mild cognitive impairment (MCI), a condition that often precedes Alzheimer's disease. The study included 89 participants with MCI who underwent brain MRI and had their mtDNA copy number measured from blood samples. The researchers found that lower mtDNA copy number was associated with smaller brain volume, particularly in the hippocampus, a brain region involved in memory formation. The study also found that lower mtDNA copy number was associated with greater white matter hyperintensities (WMH), which are areas of increased signal intensity on MRI that indicate small vessel disease in the brain [4].

The findings of this study suggest that lower mtDNA copy number may be a marker of greater neurodegeneration and cerebrovascular disease in individuals with MCI. However, the study was limited by its small sample size and cross-sectional design. Longitudinal studies are needed to determine whether changes in mtDNA copy number over time are associated with changes in brain MRI markers and cognitive function in individuals with MCI [5].

Conclusion

The association between mtDNA copy number and brain MRI markers is an emerging field of research with promising

*Correspondence to: Harry Warner, Department of Neurosciences, University of Exeter, United Kingdom, E-mail: h.m.warner@exeter.ac.uk

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findings. Higher mtDNA copy number appears to be associated with better brain health in older adults, while lower mtDNA copy number appears to be associated with greater neurodegeneration and cerebrovascular disease in individuals with MCI. Longitudinal studies are needed to confirm these findings and determine whether changes in mtDNA copy number over time are associated with changes in brain MRI markers and cognitive function. Further research in this area could lead to the development of new biomarkers and therapies for neurological disorders.

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