

Mitochondrial DNA Mutations and Age-Related Muscle Loss

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What are Mitochondria?

- **Definition:** Mitochondria are **membrane-bound organelles** present in the cytoplasm of most eukaryotic cells, often called the “**powerhouses**” of the cell.
- **Maternal inheritance:** Mitochondria are inherited **exclusively from the mother** via the egg cell.

Key functions:

- **Energy production:** Generate **adenosine triphosphate (ATP)**, the primary energy carrier required for muscle contraction, protein synthesis, and cell division.
- **Cellular respiration:** Facilitates the breakdown of food into energy in the form of ATP.
- **Regulating cell death:** Control **apoptosis**, ensuring the maintenance of healthy tissues.
- **Mitochondrial DNA (mtDNA):** Contains its DNA, which is prone to mutations that significantly affect cellular functions.

Key Findings of the Study

- **Deletion mutation's role:** Deletion mutations in mtDNA reduce its size and functionality, gradually **eroding mitochondrial efficiency**.
 - These mutated mtDNA molecules **outcompete healthy ones**, leading to a decline in ATP production.
- **Chimeric genes:** Deletion mutations create **chimeric genes** by fusing different mitochondrial genes, forming abnormal sequences.
 - Chimeric genes disrupt mtDNA expression, accelerating mitochondrial dysfunction and muscle degradation.
- **Age-related changes:** Older individuals exhibit a **two-fold increase in chimeric mitochondrial mRNA**, correlating with biological ageing.
 - Mutations also affect **brain and muscle tissues**, emphasizing their systemic impact on ageing.
- **Biomarkers for biological ageing:** **mtDNA deletion mutations** and **chimeric mRNA** are key indicators of biological ageing.
 - Understanding these markers could pave the way for therapies to **prevent or repair mutations**, delaying age-related muscle loss.

Source: TH

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