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Short Communication

Role of Genetics in Primary Myopathies and Muscle Weakness

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DESCRIPTION

Different from mitochondrial encephalomyopathy, which affects both the muscles and the neurological system, mitochondrial myopathy is a type of mitochondrial disease that solely affects the muscles. There are hundreds of mitochondria that provide all the energy needed by a typical human cell. The symptoms of mitochondrial illness can vary because a person can have a unique mix of healthy and damaged mitochondria with a unique distribution in the body. Most often, mitochondrial disease is a multisystem disorder that affects several cell, tissue, and organ types. Due to the disproportionately high energy demands of muscle and nerve cells, mitochondrial disease typically manifests as muscular and neurological problems. Some of the more prevalent effects include impaired vision, cardiac arrhythmia (abnormal heartbeat), diabetes, and stunted growth.

Primary mitochondrial myopathies

Primary Mitochondrial Myopathies (PMM) are a group of illnesses that mostly affect the skeletal muscle and are connected to genetic abnormalities (such depletions, deletions, or mutations) in the Mitochondrial DNA (mtDNA) or in genes outside of the mitochondria (nuclear DNA). Every cell in the body contains hundreds of mitochondria, which control how much cellular energy is produced and store the genetic instructions for doing so within their own distinct DNA (mtDNA) [1]. These disorders typically impair the affected cells' capacity to digest food, absorb oxygen, and produce energy. Moreover, individuals with muscle sickness symptoms but affected organs (such as the brain, liver, kidney, etc.) are not typically diagnosed with PMM and may be given a more general clinical diagnosis, such as Kearns-Sayre syndrome, MELAS syndrome, etc [2]. Certain mitochondrial myopathies may also cause problems in other organs, such as the brain. Mitochondrial encephalomyopathies are myopathies that also severely impact the neurological system [3].

The onset of MELAS, also known as mitochondrial encephalopathy, lactic acidosis, and stroke-like syndrome, can occur in children as well as young adults. MELAS can be identified by its lactic acidosis, encephalomyopathy with seizures and/or dementia, and repeated stroke-like episodes. These incidents are not typical strokes, which are sudden neurological symptoms caused by disturbances in the blood supply to the brain. Weakness is a hallmark of the myopathy form of Mitochondrial DNA Depletion Syndromes (MDDS), which eventually affects the respiratory muscles. An example of MDDS is Alpers syndrome, which is characterised by worsening liver disease and abnormalities in the brain. These classes would include forms of mitochondrial myopathy that have been discovered based on genetic sources, such as mitochondrial DNA common mutation syndromes or mitochondrial DNA deletion syndromes [4]. For instance, Kearns-Sayre disease is a form of mitochondrial myopathy as well as a sort of mitochondrial DNA deletion syndrome. Mutations in either the nuclear or Mitochondrial DNA (mtDNA), which both encode for the mitochondrial proteins, can cause myopathy [5].

CONCLUSION

It's interesting to notice that cardiomyopathies with worse outcomes are associated with mtDNA changes, most frequently mt-tRNA mutations. Moreover, Kearnes-Sayre syndrome, which is brought on by a substantial mtDNA genome loss. Muscle symptoms are prominent in many well-known disorders connected to mitochondrial dysfunction. It is typical for symptoms to appear early, with exercise resulting in a heavy feeling in the limbs and sore muscles. After exercising, patients could experience exhaustion, nausea, and shortness of breath. Patients might only be able to accomplish a limited amount of exercise as their symptoms get worse over time. Instead of focusing on the specific challenges of mitochondrial sickness, other medicines under study try to fix or avoid the damaged mitochondria. These drugs are dietary supplements based on three chemical components that are crucial for the production of ATP in our cells.

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