

The Role of Precision Medicine in Treating Rare Mitochondrial Diseases

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Description

Mitochondrial diseases, rare and complex, stem from dysfunctional mitochondria the cell's powerhouses responsible for generating energy. These diseases, which can affect multiple organ systems, are primarily caused by mutations in mitochondrial or nuclear DNA. Traditionally, the treatment of mitochondrial diseases has been challenging due to their heterogeneity, lack of comprehensive understanding and limited therapeutic options. However, precision medicine a groundbreaking approach individual genetic, environmental and lifestyle factors offers new hope for effective diagnosis and treatment. Mitochondria play a vital role in cellular energy production through oxidative phosphorylation. Dysfunction in this process can lead to a wide spectrum of clinical manifestations, including neurological, muscular and systemic disorders. These diseases are rare, with an estimated prevalence of 1 in 5,000 individuals worldwide. Mitochondrial diseases are predominantly caused by mutations in mitochondrial DNA (mtDNA) or nuclear DNA (nDNA) that affect mitochondrial function. These mutations may be inherited maternally (mtDNA) or follow Mendelian inheritance patterns (nDNA). Symptoms can range from muscle weakness and fatigue to more severe conditions like seizures, stroke-like episodes and cardiomyopathy. The diversity of symptoms makes diagnosis a complex task.

Diagnosis and treatment

The rarity and variability of mitochondrial diseases pose significant challenges for healthcare professionals. Traditional diagnostic methods rely on clinical observations, biochemical tests and muscle biopsies, which may not provide conclusive results. Treatment is largely supportive, aimed at managing symptoms rather than addressing the root cause. Precision medicine leverages advances in genomics, bioinformatics and biotechnology to revolutionize the approach to diagnosing and treating diseases. In the context of mitochondrial diseases, this approach offers the potential for: High-throughput sequencing technologies, such as Whole-Exome Sequencing (WES) and whole-genome sequencing (WGS), have become integral to precision medicine. These tools enable the identification of specific genetic mutations responsible for mitochondrial diseases, allowing for precise diagnosis and personalized treatment plans. Advancements in biomarker discovery have further enhanced the precision medicine approach. Biomarkers

like lactate levels, specific proteins, or metabolites can aid in the early detection and monitoring of mitochondrial diseases. One of the most potential applications of precision medicine is gene therapy. Techniques like CRISPR-Cas9 allow scientists to edit defective genes, potentially correcting mutations in mtDNA or nDNA. For instance, potential research has demonstrated the feasibility of reducing mutant mtDNA levels in cells using targeted nucleases.

Mitochondrial Replacement Therapy (MRT)

For disorders caused by mtDNA mutations, mitochondrial replacement therapy offers a novel solution. MRT involves replacing defective mitochondria with healthy ones from a donor, ensuring the child inherits functional mitochondria without the associated disease risk. While controversial, this technique has opened avenues for preventing the transmission of mitochondrial diseases. Precision medicine has facilitated the development of drugs customized to target specific genetic mutations. For *e.g.*, certain mitochondrial diseases caused by deficiencies in coenzyme Q10 (CoQ10) can be managed with CoQ10 supplementation. Similarly, therapeutic approaches targeting mitochondrial biogenesis and enhancing oxidative phosphorylation efficiency are under investigation. Leigh syndrome, a severe neurological disorder caused by mutations in mitochondrial or nuclear genes, exemplifies the potential of precision medicine. Gene therapy targeting specific mutations has shown potential in animal models, offering hope for translating these findings into clinical practice. Barth syndrome, a rare mitochondrial disease caused by mutations in the TAZ gene, has seen advancements through precision medicine. Researchers are exploring gene replacement therapy and small-molecule therapies to address the underlying genetic defect. The application of precision medicine to mitochondrial diseases raises ethical and regulatory concerns. Techniques like mitochondrial replacement therapy spark debates about genetic modification and potential long-term effects. Regulatory frameworks must evolve to address these issues while ensuring patient safety and accessibility to therapies. Improving the accuracy, speed, and affordability of genomic sequencing will make precision medicine more accessible. High-throughput drug screening and AI-driven drug discovery can identify new therapeutic candidates. Establishing comprehensive patient registries and data-sharing initiatives will enable researchers to identify patterns and develop targeted interventions.