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Editorial

Mitochondria: More than just the cell's powerhouse

The mitochondrion is a key cellular organelle, often called as the "powerhouse" of the cell. It is a dynamic and complex organelle, the influence of which extends far beyond mere energy production. Its primary key function is the generation of most of the cellular energy, adenosine triphosphate (ATP) through oxidative phosphorylation. The mitochondrion also plays a key role in regulation of cellular life and death, metabolism, and signaling.

In this issue of the *Galle Medical Journal*, Munasinghe, *et al.,* present and discuss a case of mitochondrial disease in a 7-year-old child. As mitochondria are considered as the central nexus of cell function, their derangement is implicated in a broad and growing spectrum of diseases. Mitochondrial diseases represent a rapidly evolving and often under-recognized area of medicine with implications across virtually all specialties.

The scope of mitochondrial disease is vast and complex, primarily due to two factors. Firstly, the mitochondrial role in the provision of major part of body's energy requirement, since every cell in the body (except mature red blood cells) relies on mitochondrial energy. These diseases can affect any organ system at any age. Organs with high energy demands - such as the brain, muscle, heart, liver, and endocrine glands - are the most frequently and severely impacted. Secondly, considering the aspect of genetic complexity, these disorders can be caused by mutations in either the mitochondrial DNA (mtDNA) or in the nuclear genes that encode mitochondrial proteins. This dual genetic origin leads to a wide spectrum of inheritance patterns and clinical presentations.

The clinical presentation is heterogeneous, making diagnosis a challenge in medical practice. Patients may present with seemingly unrelated symptoms, including neurological, muscular, endocrine etc. Many patients suffer for years before the underlying mitochondrial dysfunction is identified. Therefore, clinicians need to maintain a high index of suspicion for these diseases, especially in patients with multi-system involvement or symptoms that do not conform to typical diagnostic categories.

The field is advancing rapidly. From the aspect of diagnosis, next-generation sequencing has made genetic diagnosis more accessible, moving beyond invasive muscle biopsies as the first step.

When the treatment aspect is considered, cure remains elusive, treatments currently focus on supportive care, symptom management, and nutritional supplementation (*e.g*. coenzyme Q10). Promising research in gene therapy and strategies to manipulate mtDNA is underway.

In this context, we require to have an understanding of mitochondria-related health problems. Recognizing the signs of mitochondrial dysfunction is essential for early diagnosis and intervention, improving patient outcomes in this complex group of disorders.

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