

## Mitochondrial Disorders in Two Children of a Family: A Case Report

Mohadeseh Azadvari <sup>a</sup>, Hossein Ahmadi <sup>b</sup>, Maryam Rafiei <sup>c</sup>, Leila Gholamhosseini <sup>b,d</sup>, Seyede Zahra Emami Razavi <sup>a,\*</sup>

<sup>a</sup> Physical Medicine and Rehabilitation department, Imam Khomeini Hospital, Tehran University of medical sciences, Tehran, Iran

<sup>b</sup> Department of Health Information Management, School of Health Management and Information Sciences, Iran University of Medical Sciences, Tehran, Iran

<sup>c</sup> Physical Medicine and Rehabilitation specialist, Khatamol Anbia Hospital, Tehran, Iran

<sup>d</sup> Department of Health Information Technology, School of Paramedical Sciences, AJA University of Medical Sciences, Tehran, Iran

\* Corresponding author email address: [zemamirazavi@gmail.com](mailto:zemamirazavi@gmail.com)

### Abstract

Mitochondrial disorders belong to the group of metabolic diseases, that may cause various symptom. It may present at any ages and have harmful effects on different tissues. We present two children of a family, a 5-year-old boy and an 8-year-old girl, that came with complaint of gait imbalance and repeated fallings. Both of them had normal history of birth and developmental status, but when the girl was 6 years old, gait imbalance and ataxia started, that slowly progress. And the same sign started in a boy when he was 4-year-old. In electrodiagnostic study, we found peripheral polyneuropathy (demyelinating type) with a proximal myopathy. All findings include history and physical exam, laboratory data and electrodiagnostic study were in favour of mitochondrial disorders that confirmed by muscle biopsy. Mitochondrial disorders should be considered any time if a disorder present with more than one organ involvement. The prognosis of patients varies impressively from one patient to the other. With timely diagnosis and treatment of symptoms, better outcome will be expected.

Keywords: Mitochondrial, Disorders, Family, Children, Case report

### 1. Introduction

Mitochondrial disorders are a group of metabolic diseases, which may cause various symptoms, may present at any ages, have harmful effects on any tissues (Menezes & Ouvrier, 2012). Mitochondria is a vital organelle and due to its dysfunction may cause various features in cells, tissues, organs and systems. The final result of mitochondrial diseases is failure in cells to produce energy in the form of adenosine triphosphate (ATP) and leading to multi system dysfunction (Chinnery, Elliott, Hudson, Samuels, & Relton, 2012). The organs that is most related to production of energy by mitochondria will be the most symptomatic in mitochondrial diseases, including the central nervous system, heart, skeletal muscle, endocrine organs and kidney. Organs with completed active mitosis at birth, including the brain, muscles nerve, retina, pancreas, liver, and kidney, may be vulnerable for several reasons.

The prevalence is estimated around 1 in 5000 in patients tested for deletions and for common mutations of mtDNA

which account for 5-40% of cases, depending on the study (Bannwarth et al., 2013; Thorburn, 2004).

These diseases can present in childhood to adulthood and progress subsequently with significant suffering that result in heavy burdens on affected families (McFarland, Taylor, & Turnbull, 2010).

Signs, symptoms and organ involvement are greatly heterogeneous from one individual to the other, which is the reason for a high degree of inter- and intra-familial variability. This is the cause why each affected subject will have a different prognosis. Early recognition and treatment of symptoms are crucial for improving the prognosis.

### 2. Case presentation

Two siblings, 5 years old boy and an 8 years old girl, came with complaint of gait imbalance and repeated fallings tour clinic. Both of them were born on time, 39-40 weeks of pregnancy, and had normal development. When the girl was 6 years old, gait imbalance and ataxia started, that was slowly progressive. The same sign started in boy