Role of Team Work in The Management Of Mitochondrial Disease In Children (Case Report)

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Seyed Hassan TONEKABONI MD ^{1,2} Mitochondrial disorders affect people from neonatal period to adulthood. The clinical pictures also vary from single to multiple involved organs. Many of these disorders are heteroplasmic meaning that in every patient mitochondria, cells and tissue may have different proportion of wild-type and mutant mitochondrial DNA. The most common affected tissue in this group of patients is the one with the highest energy requirements.

Mitochondrial DNA is made of 37 maternally inherited genes in a compact genome state, and other primary mitochondrial diseases are due to mutations in nuclear DNA. So far about 1000 nuclear gene are involved in mitochondrial function. Most Patients will so-called syndromic mitochondrial disease is the results of mutations in mitochondrial DNA.

As similar disorders with a wide spectrum of clinical phenotype a close cooperation between different disciplines of medical and laboratory staff is necessary to reach to a definite diagnosis.

I will present some patients and I will try to show some challenging situations we are faced with in mitochondrial disorder.

Keywords: Mitochondrial disease; Diagnosis; Children; Chronic External Ophtalmoplegia; Team work

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