Author's response to reviews

Title: De Toni-Debre-Fanconi syndrome in a patient with Kearns-Sayre Syndrome. Diagnostic delay in one case

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Dear Sirs,

As pediatrician involved in care of children with diabetes, I became interested in metabolic and endocrine disorders related to diabetes, in pediatric rare cases and in establishing an early and accurate diagnosis in any particular case.

Mitochondrial myopathies have many different faces, with a complex array of symptoms. Some symptoms can be so mild that they’re hardly noticeable, while others are life-threatening. The possibility of mitochondrial dysfunction needs to be taken into account by every medical subspecialty.

Kearns-Sayre syndrome (abbreviated KSS) is a disease caused by a 5,000 base deletion in the mitochondrial DNA. Unlike most mitochondrial diseases, it is not maternally inherited.

In this case presentation the diagnosis was delayed for many years; more, a wrong diagnosis was made initially, and the parents were very frustrated.

Analyzing the medical literature (PubMed search) regarding the association between Kearns-Sayre syndrome and de Toni Debre Fanconi syndrome I found only 8 articles.

I consider that the practitioners should be aware about the presence of the mitochondrial myopathies, and the broad spectrum of their clinical presentations. Also, the diagnosis of de Toni Debre Fanconi syndrome in a patient should be followed by an extensive research about the possible presence of an associated mitochondrial myopathy.

Respectfully yours,

Mihai Cristina Maria, MD, PhD