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Letter to the Editor

Diagnose Kearns-Sayre Syndrome According to the Clinical Diagnostic Criteria and a Pathogenic Variant

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Letter to the Editor

We read with interest the article by Moustaine *et al*. on a 16-year-old male with Kears-Sayre syndrome (KSS), which was diagnosed based on the clinical presentation, ophthalmologic examination, and muscle biopsy [1]. The patient benefited significantly from bilateral surgical ptosis correction under local anaesthesia [1]. The study is impressive, but some points require further discussion.

The first point is that the diagnosis KSS was not genetically confirmed. Since KSS is genetically heterogeneous, the phenotype is heavily dependent on the mutated gene and the specific mutation, genetic counselling is highly dependent on the underlying mutation and whether the variant was inherited or occurred sporadically, KSS may not only be due to single mtDNA deletions, but also due to point mutations, for example in *POLG1* [2], it is imperative to clarify the underlying genetic defect in every KSS patient.

The second point is that the family history is missing. We should know whether another first-degree family member was clinically affected and whether a causative mutation was identified in any of these family members. Assuming that the causative defect was a single mtDNA deletion, it is important to know it, because 4% of these deletions are inherited through the maternal line. Additionally, if KSS is due to a mutation in *POLG1* or mutation in other nDNA genes, it is quite likely that the defect was transmitted via an autosomal dominant or recessive way.

The third point is that the index patient did not meet the established diagnostic criteria for KSS [2, 3]. According to these criteria, KSS is diagnosed if the three core features external ophthalmoplegia, onset <20 years, and pigmentary retinopathy are present and at least one of the additional features of complete heart block, cerebrospinal fluid protein >100 mg/dL, or cerebellar ataxia [2, 3]. The index patient did not meet these criteria and did not carry a causative mutation. Therefore, the clinical diagnosis is highly speculative. Ptosis and ophthalmoparesis, along with retinopathy and ragged red fibers, can also occur in other syndromic and non-syndromic mitochondrial disorders (MIDs).

The fourth point is that it is contradictory to mention that the patient had no medical history and then continue a few sentences later that the patient had had ptosis for five years. This discrepancy should be resolved.

In summary, the interesting study has limitations that put the results and their interpretation into perspective. Removing these limitations could strengthen conclusions and support the study's message. Before diagnosing KSS, its diagnostic criteria must be met. In addition, the genetic background of KSS must be clarified to support the diagnosis.

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References
1. Moustaine MO, Azemour Z, Mohammed F, Benlanda O, Nassik H, Karkouri M. Management of Ptosis in Kearns-Sayre Syndrome: A Case Report and Literature Review. Arch Plast Surg, Apr 8, 2024; 51(2):182-186. Doi: 10.1055/a-2207-7587
2. Tsang SH, Aycinena ARP, Sharma T. Mitochondrial Disorder: Kearns-Sayre Syndrome. Adv Exp Med Biol. 2018; 1085:161-162. Doi: 10.1007/978-3-319-95046-4_30
3. Shemesh A, Margolin E, Kearns-Sayre Syndrome, Jul 17, 2023. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing, Jan 2024.