



Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation

Josef Finsterer, Sinda Zarrouk-Mahjoub

► To cite this version:

Josef Finsterer, Sinda Zarrouk-Mahjoub. Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation. Chinese Medical Journal, 2016, 129 (18), pp.2267-8. <10.4103/0366-6999.189913>. <pasteur-01444419>

HAL Id: pasteur-01444419

<https://riip.hal.science/pasteur-01444419v1>

Submitted on 24 Jan 2017

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



Distributed under a Creative Commons CC BY-NC 4.0 - Attribution - Non-commercial use - International License

Correspondence

Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation

Josef Finsterer¹, Sinda Zarrouk-Mahjoub²

¹Krankenanstalt Rudolfstiftung, 1030 Vienna, Austria

²Genomics Platform, Pasteur Institute of Tunis, 1002 Tunis, Tunisia

Josef Finsterer and Sinda Zarrouk-Mahjoub contributed equally to this work.

To the Editor: With interest, we read the article by Kwon *et al.*^[1] about a single patient with Kearns-Sayre syndrome (KSS), who successfully and without complications underwent peripheral nerve block for multiple muscle biopsies. We have the following comments and concerns.

The main disadvantage of this study is that the diagnosis of KSS was not genetically confirmed. Although the patient underwent muscle biopsy, the authors did not report the results of genetic studies of the mitochondrial DNA (mtDNA) extracted from the muscle.^[1] The cause of KSS is usually a single mtDNA deletion;^[2] however, in rare cases, single mtDNA point mutations might be causative.^[3] KSS should not be diagnosed upon a muscle biopsy alone.

The authors should also provide information if the family history was positive or negative for a mitochondrial disorder (MID). Did any of the first-degree relatives present with a history of a neuromuscular disorder, in particular, an MID, or did any of them have a history of complications during anesthesia?

It is not conceivable why the patient required multiple muscle biopsies.^[1] In general, a single biopsy is sufficient to show typical histological, immune-histological, or ultrastructural abnormalities, such as ragged-red fibers, cytochrome-C-oxidase-negative fibers, abnormal mitochondria with both unusual cristae and paracrystalline inclusions and biochemical abnormalities, such as complex-I-, complex-II-, complex-III-, or complex-IV-deficiency, alone or in combination.

It should be mentioned that general anesthesia or even regional anesthesia might deteriorate myopathy and, thus, weakness in MID patients.^[4,5] Did the patient experience previous complications during anesthesia? Did he ever receive long-duration or high-dose propofol, which should be avoided in MID patients?

Since KSS might be associated with cardiomyopathy, we should be informed about the results of long-term electrocardiogram (ECG) recordings, echocardiography, and

cardiac magnetic resonance imaging. KSS patients might present with hypertrophic cardiomyopathy, dilated cardiomyopathy, or Takotsubo syndrome (TTS). It should be discussed that particularly general anesthesia and surgery in MID patients might trigger a TTS.^[5]

Long-term ECG recordings are important since KSS patients may die from sudden cardiac death (SCD). Since SCD might be even the initial manifestation of cardiac involvement in KSS, it is essential to monitor KSS patients cardiologically as soon as the neurological diagnosis is established. Cardiac conduction defects or arrhythmias preceding SCD in KSS patients include QT-prolongation, Torsade de pointes, or polymorphic ventricular tachycardia. KSS patients rather require an implantable cardioverter defibrillator than a pacemaker.

It is also important to mention that KSS patients usually present with short stature. Which was the height of the presented patient, his parents, and his siblings?

Overall, this interesting case requires genetic confirmation of the diagnosis since MIDs frequently show a wide range of phenotypic mimicry and a work-up of first-degree family members. To profit most from the description of a KSS patient, all aspects of the syndrome should be addressed. Particularly, addressed should be cardiac involvement since these patients might die from SCD.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Address for correspondence: Dr. Josef Finsterer,
KAR, Postfach 20, 1180 Vienna, Austria
E-Mail: fifigs1@yahoo.de

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 3.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms.

© 2016 Chinese Medical Journal | Produced by Wolters Kluwer - Medknow

Received: 05-06-2016 **Edited by:** Peng Lyu

How to cite this article: Finsterer J, Zarrouk-Mahjoub S. Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation. Chin Med J 2016;129:2267-8.

Access this article online

Quick Response Code:



Website:
www.cmj.org

DOI:
10.4103/0366-6999.189913

REFERENCES

1. Kwon WJ, Bang SU, Oh SC, Shim JW. Peripheral nerve block is safely administered in a patient with Kearns-Sayre syndrome. *Chin Med J* 2016;129:1251-2. doi: 10.4103/0366-6999.181971.
2. Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, *et al*. Redefining phenotypes associated with mitochondrial DNA single deletion. *J Neurol* 2015;262:1301-9. doi: 10.1007/s00415-015-7710-y.
3. Zhang Y, Wang ZX, Niu SL, Xu YF, Pei P, Yuan Y, *et al*. Phenotype heterogeneity associated with mitochondrial DNA A3243G mutation (In Chinese). *Acta Academiae Medicinae Sinicae* 2005;27:77-80.
4. Finsterer J, Haberler C, Schmiedel J. Deterioration of Kearns-Sayre syndrome following articaine administration for local anesthesia. *Clin Neuropharmacol* 2005;28:148-9. doi: 10.1097/01.wnf.0000165352.10841.21.
5. Kawagoe I, Odoh M, Koh K, Takada T, Inada E. A case of intraoperative cardiac arrest in a patient with mitochondrial encephalomyopathy undergoing lung resection. *Masui* 2013;62:431-4.

