



**HAL**  
open science

## Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up

Josef Finsterer, Sinda Zarrouk-Mahjoub

► **To cite this version:**

Josef Finsterer, Sinda Zarrouk-Mahjoub. Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up. Chinese Medical Journal, 2016, 129 (20), pp.2518-2519. 10.4103/0366-6999.191835 . pasteur-01439282

**HAL Id: pasteur-01439282**

**<https://hal-riip.archives-ouvertes.fr/pasteur-01439282>**

Submitted on 29 Jun 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 Attribution - NonCommercial - NoDerivatives | 4.0 International License

# Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up

Josef Finsterer<sup>1</sup>, Sinda Zarrouk-Mahjoub<sup>2</sup>

<sup>1</sup>Krankenanstalt Rudolfstiftung, 1030 Vienna, Austria

<sup>2</sup>Genomics Platform, Pasteur Institute of Tunis, 1002 Tunis, Tunisia

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

To the Editor: With interest, we read the article by Yu *et al.*<sup>[1]</sup> about the clinical presentation and central nervous system (CNS) imaging in 19 patients with Kearns-Sayre syndrome (KSS), of whom 12 were genetically confirmed. We have the following comments and concerns.

The main disadvantage of the study was that the diagnosis was not genetically confirmed in all patients and mitochondrial DNA (mtDNA) deletion was detected in only 63% patients.<sup>[1]</sup> What was the reason for seven KSS patients not undergoing genetic testing? Did not all patients undergo muscle biopsy?

If only 12 of 19 patients underwent muscle biopsy, how can KSS be diagnosed? Among the seven patients who obviously did not undergo muscle biopsy, upon which clinical criteria were these patients diagnosed as KSS?

Among the seven patients who were not genetically confirmed, did the authors look for mtDNA point mutations in lymphocytes? It has been repeatedly reported that KSS may not only be due to mtDNA deletions but also due to mtDNA point mutations, such as m.3249G>A in the tRNA (Leu) gene, m.3255G>A in the tRNA (Leu) gene, or m.3243A>G in the tRNA (Leu) gene.

We should also be informed about the results of biochemical investigations to know about the activity of respiratory chain complexes. Complex I, complex II, complex IV, complex I and IV, complex I, III, and IV, complex I, IV, and V, and complex I, III, IV, and V deficiency or normal respiratory chain activity has been previously described in KSS.<sup>[2,3]</sup>

Among the 15 patients who underwent cerebral magnetic resonance imaging, six patients showed hyperintensities on diffusion-weighted imaging in the white matter or basal ganglia.<sup>[1]</sup> We should be informed about the nature and pathogenesis of these lesions. Were these lesions on the corresponding apparent diffusion coefficient map hyper-, hypo-, or iso-intense? Were these lesions interpreted as cytotoxic edema or as vasogenic edema? Can these lesions be interpreted as stroke-like lesions, the morphological correlate of a stroke-like episode?

Cognitive functions can be impaired in KSS patients.<sup>[4]</sup> Did the 19 patients also undergo neuropsychological testing to assess their cognitive abilities and if they were impaired or not?

Did the authors consider implantation of an implantable cardioverter defibrillator in any of their patients since four patients died from sudden cardiac death in a series of 35 KSS patients?<sup>[4]</sup> Did the patients undergo long-term electrocardiogram recordings, and in particular were loop recorders implanted to see if any of the patients had a tendency to develop prolonged QT-interval, early repolarization, or ventricular arrhythmias?

It was interesting to see that only 3 of 19 had short stature.<sup>[1]</sup> Short stature is one of the clinical criteria for diagnosing KSS.<sup>[5]</sup>

Overall, these interesting case series should be supplemented by more detailed clinical, instrumental, and genetic data. The more information about KSS patients is collected, the more we can learn about the phenotypic and genotypic variability of these patients, and the better will be the management and outcome of KSS patients.

## Financial support and sponsorship

Nil.

## Conflicts of interest

There are no conflicts of interest.

## REFERENCES

1. Yu M, Zhang Z, Wang QQ, Liu J, Zuo YH, Yu L, *et al.* Clinical and brain magnetic resonance imaging features in a cohort of Chinese patients with Kearns-Sayre syndrome. *Chin Med J* 2016;129:1419-24. doi: 10.4103/0366-6999.183417.

**Address for correspondence:** Prof. Josef Finsterer, Krankenanstalt Rudolfstiftung, Postfach 20, 1030 Vienna, Austria  
E-Mail: [fipaps@yahoo.de](mailto:fipaps@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:** 15-06-2016 **Edited by:** Xin Chen

**How to cite this article:** Finsterer J, Zarrouk-Mahjoub S. Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up. *Chin Med J* 2016;129:2518-9.

### Access this article online

Quick Response Code:



Website:  
[www.cmj.org](http://www.cmj.org)

DOI:  
10.4103/0366-6999.191835

2. Marin-Garcia J, Goldenthal MJ, Sarnat HB. Kearns-Sayre syndrome with a novel mitochondrial DNA deletion. *J Child Neurol* 2000;15:555-8. doi: 10.1177/088307380001500812.
3. Reichmann H, Degoul F, Gold R, Meurers B, Ketelsen UP, Hartmann J, *et al.* Histological, enzymatic and mitochondrial DNA studies in patients with Kearns-Sayre syndrome and chronic progressive external ophthalmoplegia. *Eur Neurol* 1991;31:108-13.
4. Khambatta S, Nguyen DL, Beckman TJ, Wittich CM. Kearns-Sayre syndrome: A case series of 35 adults and children. *Int J Gen Med* 2014;7:325-32. doi: 10.2147/IJGM.S65560.
5. Rivner MH, Shamsnia M, Swift TR, Trefz J, Roesel RA, Carter AL, *et al.* Kearns-Sayre syndrome and complex II deficiency. *Neurology* 1989;39:693-6. doi: 10.1212/WNL.39.5.693.