Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up

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To the Editor: With interest, we read the article by Yu *et al*.[1] 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.[1] 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.[2,3]

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.[1] 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.[4] 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?[4] 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.[1] Short stature is one of the clinical criteria for diagnosing KSS.[5]

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.

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Access this article online

Quick Response Code:

Website: www.cmj.org

DOI: 10.4103/0366-6999.191835

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.

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