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Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up

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Cognitive functions can be impaired in KSS patients.¹ 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?²

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.¹

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.

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Conflicts of interest
There are no conflicts of interest.

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There were several articles studying the exact nature of high
a detecting platform has not been established in our laboratory.
for the diagnosis of mitochondrial disease.
chain complexes in muscle tissues, can also help provide evidence
Biochemical investigations, especially activity assay of respiratory
A3243G, A8344G) in their blood, which were negative.
performed common mtDNA point mutations analysis (mtDNA
muscle biopsy, but all of them showed typical KSS features. We
tissue to do the mutation examination although RBF% cannot be
had muscle biopsy in another hospital, and we got a little muscle
were not enough for further gene analyses; one patient (patient 12)
however, mtDNA mutation detection was available in 11 of them
which showed RRF, RBF, and COX‑negative fibers in almost all patients.
reported by us, 15 patients underwent muscle biopsy in our hospital
symptoms, or cerebrospinal fluid protein levels above 1000 mg/L.
progressive external ophthalmoplegia. Eur Neurol
features, muscle pathology and molecular genetic analysis can
The diagnostic criteria have been widely used.
Diagnosis and Management of Kearns-Sayre Syndrome Rely
DNA studies in patients with Kearns‑Sayre syndrome and
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