

Cardiac disease in Kearns-Sayre syndrome requires comprehensive management

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Letter to the Editor

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With interest we read the article by Trivedi et al about a 7-year-old female with multi-system Kearns-Sayre syndrome manifesting as developmental delay, hypotonia, failure to thrive, hypoparathyroidism with consecutive hypocalcemia, ptosis, and cardiac conduction defect.¹ The patient received a prophylactic pacemaker in the absence of a complete atrioventricular block.¹ We have the following comments and concerns.

We do not agree with the statement in the introduction that cardiac complications of Kearns-Sayre syndrome only include left anterior fascicular block, right bundle branch block, and complete atrioventricular block.¹ Cardiac involvement in Kearns-Sayre syndrome is variable and may also include dilated cardiomyopathy² requiring a ventricular assist device,³ bradycardia-related polymorphic ventricular tachycardia,⁴ myocardial scarring,⁴ or ventricular arrhythmias, including QT-prolongation,⁵ ventricular tachycardia,⁵ or torsades des pointes.^{5,6}

We also do not agree with the management of the presented patient. Since patients with Kearns-Sayre syndrome not only develop atrioventricular block but also ventricular arrhythmias⁵ and experience sudden cardiac death,⁷ we should be informed about the results of long-term electrocardiography recordings by means of a reveal recorder. Kearns-Sayre syndrome patients may not only require implantation of a pacemaker but rather implantation of a loop recorder or an implantable cardioverter defibrillator.⁸ In a retrospective study of 35 patients with Kearns-Sayre syndrome, four (11%) died after diagnosis.⁹ Interestingly, all four patients of this study experienced sudden cardiac death.⁹ Thus, prophylactic implantation of an implantable cardioverter defibrillator rather than a pacemaker should be considered.

Recently, it has been shown that subclinical cardiac involvement may be detected by application of cardiac MRI, including late gadolinium enhancement.⁴ Thus, we should be informed about the findings on cardiac MRI in the presented patient.

Though Kearns-Sayre syndrome is due to sporadic, single mitochondrial deoxy-nucleic acid deletions in the majority of cases, there is a 4% risk of inheritance.¹⁰ Thus, we should be informed if Kearns-Sayre syndrome in the presented patient was inherited from her mother or sporadic, if the mother had clinical manifestations, or if she carried the mitochondrial deoxy-nucleic acid deletion as well.

Patients with Kearns-Sayre syndrome may not only present with developmental delay, hypotonia, failure to thrive, hypoparathyroidism, ptosis, or cardiac conduction defect, but also with dementia, dysarthria, dystonia, cataracts, pigmentary retinopathy, hypoacusis, diabetes, growth retardation, myopathy, or elevated cerebrospinal fluid protein.^{11,12} Since these features may not be present at onset or during the first years but may develop during the further disease trajectory, long-term follow-up of these patients is required.

In summary, this interesting case report could be more meaningful by discussing the broad heterogeneity of cardiac disease in Kearns-Sayre syndrome, by genetic investigation of the mother, by investigating the index case with cardiac MRI, and by prospective investigations of the index case for multi-system involvement. Even if a pacemaker is implanted, we should watch out for ventricular arrhythmias, consider loop recording, and eventually implantation of an implantable cardioverter defibrillator.

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Conflicts of Interest. None.

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