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Case report

Symptomatic complete heart block leading to a diagnosis of Kearns–Sayre syndrome

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ABSTRACT

Kearns–Sayre syndrome (KSS) is a rare syndrome characterized by the triad of progressive external ophthalmoplegia, pigmentary retinopathy and cardiac conduction system disturbances; it is a mitochondrial encephalomyopathy with which usually presents before the patient reaches the age of 20. Here we present a case report of a patient with KSS who presented with symptomatic complete heart block.

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1. Introduction

Symptomatic complete heart block (CHB) or complete AV Block is usually a medical emergency and needs immediate pacing. It is a more common diagnosis in the elderly, however some patient do present earlier in life we should try to look for a secondary cause for the CHB. Here we present a case report of a young patient who presented with symptomatic complete heart block and had a diagnosis of Kearns–Sayre syndrome.

Kearns–Sayre syndrome (KSS) is a very rare, genetic,¹ sometimes fatal multisystem disorder which usually present before the age of 20. It is a mitochondrial encephalomyopathy² in which defects of the central and peripheral nervous system, along with poor intellectual development are present.

It is characterized by progressive external ophthalmoplegia, mild skeletal muscle weakness, retinal pigmentation, intra cardiac conduction defect, hearing deficiencies, impaired cognitive dysfunction, cerebellar signs, diabetes mellitus and other endocrine disorders. These patients are generally short statured, have a lax posture, poor musculature, frequent secondary kyphoscoliosis, and hyperlordosis are present.

2. Case report

Mr RS, a 28-year-old male presented to our emergency department with recurrent syncope for last 1 year, whose frequency had increased over the last 1 month. His electrocardiogram (ECG) revealed complete AV Block with a broad

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QRS ventricular escape rhythm of 36 bpm (Fig. 1) and he was immediately paced using a temporary pacemaker. On further evaluation of his history we found that he had symptoms of generalized weakness and listlessness which were first noticed when he was about 10 years old, and till then he had presented normal neuropsychomotor development as compared to his siblings. However after 10 years of age there was deficiency in his stature and mental development and he also began to complain of progressive “dropping eyes” along with diplopia. His clinical examination was unremarkable except generalized motor weakness and reduced mental development. The echo revealed dilated chambers, depressed LV systolic functions with an LV ejection fraction of 40% and with global hypokinesia. Arterial blood gas pH estimation revealed mild metabolic acidosis. An ophthalmology consult was also obtained. The fundus examination was suggestive of pigmentary retinopathy in both the eyes, along with ptosis and restriction of movement of extra ocular muscles (Figs. 2 and 3). Histopathological examination of skeletal muscle was inconclusive, however there was a suggestion of mitochondrial myopathy as a differential diagnosis. A diagnosis of Kearns–Sayre syndrome (KSS) was made based on the clinical triad of progressive external ophthalmoplegia, pigmentary retinopathy and cardiac conduction system disturbances, which in this case was the presence of complete heart block. He subsequently underwent a single chamber permanent pacemaker implantation, with a lead screwed into the Right Ventricular outflow tract and is well on follow up with no further syncopal episodes.

3. Discussion

Kearns–Sayre syndrome (KSS) is a rare, genetic¹ condition characterized by a combination of external ophthalmoplegia, pigmentary retinopathy, progressive degeneration of cardiac conduction system,³ progressive external ophthalmoplegia,⁴ mild skeletal muscle weakness, hearing deficiencies, cerebellar signs, impaired cognitive dysfunction, diabetes mellitus and other endocrine disorders. The most important prognostic factor in KSS patients is the cardiac involvement which can occur in almost 60% of the patients, characterized

Fig. 1 – Complete heart block with wide QRS escape @ 36 b/min.

Fig. 2 – Ptosis and external ophthalmoplegia (permission obtained from the patient to use his photograph for teaching purpose).

by progressive degeneration of the conduction system, syncopal attacks, heart failure and even sudden cardiac death. The various ECG abnormalities reported include complete and incomplete right bundle branch block, fascicular block, left bundle branch block and nonspecific intra ventricular conduction delays.⁵ Patients with KSS who have ventricular conduction defects show a rapid progression to complete AV Block with an associated mortality of almost 20%. Electrophysiological studies generally show an increase in His to ventricular activation time. The ACC/AHA/HRS guidelines give a class I, level of evidence B rating to implantation of pacemakers for third-degree and advanced second-degree AV block at any anatomic level when associated with neuromuscular diseases and AV block.⁶ Our patient presented with most of the features of KSS and had recurrent syncopal attacks due to complete AV Block, which was an indication for pacing. Ventricular dysfunction has also been reported as a part of the neuromuscular diseases, and our patient had depressed LV function, which may be due to the neuromuscular dysfunction or may be due to persistent bradycardia of long duration.

Thus Kearns–Sayre syndrome is a rare multisystem disorder with involvement of the cardiac conduction system and screening of family members is essential considering the genetic nature of the disease and regular long term cardiovascular follow up is essential.

Fig. 3 – Fundus photo showing bony spicules suggestive of pigmentary retinopathy.

Conflicts of interest

All authors have none to declare.

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