Classical triad of Kearns-Sayre syndrome

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DESCRIPTION

We describe the images of a 27-year-old man presenting with recurrent syncope due to complete heart block (figure 1). The patient, on further examination, showed ophthalmoplegia, bilateral ptosis (figure 2) and pigmentary retinopathy (figure 3). Kearns-Sayre syndrome (KSS) is a rare mitochondrial genetic disorder with multisystem involvement. KSS usually presents before the age of 20 years. The triad of external ophthalmoplegia, pigmentary retinopathy, and cardiac conduction defects are sufficient to make a confident diagnosis of this rare entity. Cerebrospinal fluid protein of more than 1 g/L or cerebellar ataxia are also important characteristics of KSS. There is no definitive treatment for this condition, but pacemaker implantation can prevent syncope, and reduce morbidity in patients presenting with advanced atrioventricular (AV) conduction abnormalities.

Around 57% of patients with KSS have cardiac involvement, including recurrent syncope, bundle branch blocks, fascicular blocks and non-specific intraventricular conduction disturbances.¹ Around 20% of deaths in these patients is attributed to cardiac causes. The American College of Cardiology/American Heart Association/Heart

Figure 3 Fundus examination of the right and left eye showing pigmentary retinopathy.

Rhythm Society 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.

Histopathological examination of the skeletal system characteristically shows ragged red fibres. Endocrinopathies occur commonly in these patients, and around 13% develop diabetes.² Although diabetes, hypothyroidism, and parathyroid disorders do occur, our patient did not suffer from these conditions; however, follow-up is needed. Ventricular dysfunction has been reported as a part of neuromuscular disease.³

Figure 1 Twelve-lead ECG showing complete atrioventricular block with an incomplete right bundle branch block type of escape rhythm at a rate of 40/min.

Figure 2 Photograph of the patient showing bilateral ptosis with compensatory wrinkling on the forehead.

Learning points

- ▶ Kearns-Sayre syndrome is the differential diagnosis when a young patient presents with cardiac conduction disorders.
- ▶ Permanent pacemaker implantation is only a part of the treatment; such patients need lifelong follow-up as they carry the risk of having ventricular dysfunction as part of their disease, and the risk is enhanced due to right ventricular pacing.

Competing interests None declared.

Patient consent Obtained.

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