Original Research Paper

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Cardiology

# A CASE REPORT OF SYMPTOMATIC COMPLETE HEART BLOCK DIAGNOSED AS KEARNS-SAYRE SYNDROME(KSS)

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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 KSS who presented with symptomatic complete heart block.

**KEYWORDS :** Kearns-Sayre syndrome (KSS), Complete AV block, Dual chamber pacemaker.

## 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, sometimes fatal multisystem disorder which usually present before the age of 20. It is a mitochondrial encephalomyopathy in which defects of the central and peripheral nervous system, along with poor intellectual development are present.

## CASE REPORT:

A 34 year old female presented with symptoms of giddiness and pre-syncopal episode, whose frequency increased over a period of time. Her Electro cardiogram (ECG) revealed complete AV block with 36 bpm.

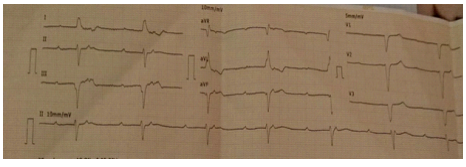

**Fig 1: ECG of complete AV block**

On further evaluation of history we found that she had two similar episodes in past for which she was managed with medication in ER. Around age of 15 she experienced progressive 'dropping eyes with diplopia'. Her clinical examination was unremarkable except mild generalized motor weakness and reduced mental development. MRI brain, audiometry, urine analysis, serum creatinine kinase, lactate and pyruvate levels, basic metabolic panel, calcium, magnesium, plasma cortisol levels and thyroid profile were normal except her hemoglobin which falls into anemic range fluctuates between 8 to 12 mg/dl.. The echo revealed normal LV size, fair LV systolic function with LV ejection fraction of 50% and normal LV compliance. An ophthalmology consult was also obtained. The fundus examination was suggestive of salt pepper retinopathy consistent with retinitis pigmentosa along with bilateral severe ptosis and restricted extra ocular muscle motility in both eyes.

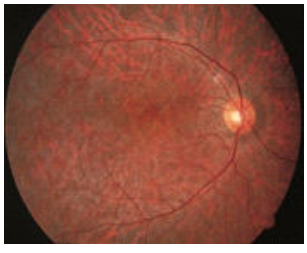

**Fig 2:** Salt Pepper Retinopathy Suggestive Of Retinitis Pigmentosa

No bell's phenomenon observed. She underwent frontalis suspension for ptosis but due to infection it was released afterwards. Histopathological examination of skeletal muscle was inconclusive, however there was a suspicion 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. She subsequently underwent dual chamber permenant pacemaker (DDDR) insertion with one lead in right ventricular apex and other in right atrial appendage. She is well on follow up with no symptoms. The post procedure echocardiography was suggestive of moderate LV dysfunction with LV ejection fraction of 40% and global LV hypokinesia.

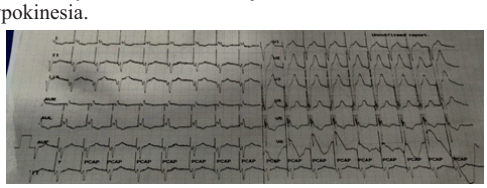

**Fig 3:** ECG Showing Pacemaker Rhythm After Dual Chamber Pacemaker Insertion

## DISCUSSION:

Kearns-Sayre syndrome (KSS) is a rare, genetic condition characterized by a combination of pigmentary retinopathy, progressive degeneration of cardiac conduction system, progressive external ophthalmoplegia, 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 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. Patients with KSS who have ventricular conduction defects show a rapid progression to complete AV Block with an associated mortality of almost 20%. 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. Our patient presented with most of the features of KSS and had recurrent pre-syncopal attacks due to complete AV Block, which was an indication for pacing. She was advised for pacemaker insertion on her previous encounters but never underwent due to financial constraints.

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.

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