A Case Report of Complete Heart Block in an Uncommon Disease Entity: Kearns Sayre Syndrome

Deepak Kumar Mishra¹, Anil Sharma¹, Shalima Gautam¹, Ramesh Kawar¹, BK Goyal²

Abstract
Complete heart block has a varied aetiology; commoner being ischaemia and senile degeneration of AV node. In this article we report a case of complete heart block (AV nodal) in patient of Kearns Sayre Syndrome who has incidently SA node disease also which is further rarer in this disorder.

Case and Discussion
An 18 year old male presented with complaints of giddiness associated with blurring of vision for one day. On evaluation he was bradycardic with a pulse rate of 41 beats per minutes (bpm) regular. His BP was 90/60 mmHg. ECG showed complete heart block with ventricular rate of about 40 bpm. His atrial rate was around 40 bpm but both atria and ventricular rate were dissociated in rhythm. One thing which was obvious at the time of examination was his drooping eyelids. Injection atropine one ampoule was given but without much variation in heart rate and without improvement in symptoms. Fluid boluses were repeated along with repetition of atropine but heart rate rose to 44 with patient being still symptomatic. So immediate temporary transvenous pacing (Bard pacing wire) was done through right femoral route and patient was stabilised haemodynamically. The patient was planned for permanent pacemaker implantation and was worked up. General physical examination revealed significant finding in the form of severe bilateral ptosis and complete external ophthalmoplegia. Bilateral hearing loss was present. Pectus Carinatum at lower sternal border was seen. Generalised muscle wasting was present with power in proximal muscle group lesser than distal. Special investigation showed bilateral pigmented retinopathy, bilateral sensorineural deafness, normal echocardiographic findings. Blood Fig. 1: 12 lead ECG showing complete heart block with ventricular escape rhythm of approx 40 bpm. For a few moment there was loss of capture seen as spikes only..!
Kearns Sayre syndrome is a mitochondrial inherited multisystem disorder affecting multiple organ systems which usually presents before 20 years of age. The patient refused muscle biopsy. Based on these investigations we suspected probably we are dealing with multisystem disease. We thought of some possibility of Duchennes (DMD) and Beckers Muscular dystrophy (BMD) but the natural history of these diseases include early presentation (first decade of life) and rarely the patient survives to late second decade or third decade of life plus there was no calf pseudohypertrophy in our case which was a strong point against DMD and BMD so it was almost excluded. Patient had no diurnal variation of ptosis so Myasthenia Gravis was also excluded almost with certainty (Figures 1, 2, 3, 4 and 5).

Single chamber (VVI mode) permanent pacemaker (Medtronic) was inserted by making a right infraclavicular pocket and rate was kept at 70 bpm. The patient was advised physiotherapy for proximal myopathy to be started after healing of pacemaker pocket wound and was planned for surgical ptosis correction at sometime in near future. ENT opinion was taken for possible correction of sensorineural deafness so as to improve his quality of life. It is of prime importance to mention here that only permanent pacing therapy even prophylactic as in case of fascicular blocks (AAC/AHA/NASPE class 2b) has been shown to be of mortality benefit. Rest form of therapies are all supportive including Coenzyme Q and multivitamins.

Kearns Sayre syndrome is a mitochondrial inherited multisystem disorder affecting multiple organ system which usually presents before 20 years of age. It affects eye (ophthalmoplegia, pigmentary retinopathy, cataract, etc), nervous system (ataxia, sensorineural hearing loss), musculoskeletal system (proximal myopathy, ptosis), cardiovascular (AV blocks), Endocrine (including diabetes) and so on and so forth.

It is a rare entity with a prevalence of 1.17 per 100,000 patients. It is inherited from maternal side because mitochondria being part of cytoplasm in contributed by maternal ova. It is the mutation or deletion of these
A Rare Association of Parachute Mitral Valve with Double Outlet Right Ventricle and Severe Pulmonary Hypertension in an Adult

K Meenakshi *, Sundar Chidambaram **, VE Dhandapani, R Rameshwar †

Abstract

Congenital mitral stenosis (MS) is a rare congenital cardiac malformation and the obstruction to the flow across the mitral valve can be caused by supramitral ring, commissural fusion, short chordae, anomalous mitral arcade, anomalous position

*Professor of Cardiology, **3rd year D.M Post Graduate in Cardiology, ***Professor of Cardiology, †CRRI, Department of Cardiology, Madras Medical College, Chennai 600003, Tamil Nadu

Received; 01.03.2013; Accepted: 23.03.2013

Cardiovascular involvement includes AV blocks, fascicular blocks, LV dysfunction secondary to prolonged Right Ventricular (RV) pacing, heart failure. There have been a few reports of even sinus node dysfunction and prolonged sinus pauses. Non specific ST-T changes and T wave changes have also been described.

Looking carefully at the ECG of the patient it can be noticed that our patient also had involvement of sinus node as his sinus rate (i.e corresponding AA interval) was also lesser than expected and it was actually 40 beats per minute. So the ideal therapy for him would have a dual chamber pacemaker say DDD type; but due to financial constraints single Chamber (VVI) pacing was done.

He was advised for periodic follow ups for the pacemaker interrogation at routine interval and was told to follow up at ophthalmology OPD (for ptosis correction), ENT OPD for possibility of hearing aid.

References