Case Reports

Hypertrophic Peripheral Neuropathy (Dejerine-Sottas Disease) Associated with Heart Block

Case Report Presentation and Review of the Literature

George KOUVARAS, M.D., George CHRONOPOULOS, M.D.,
George PISTIS, M.D., and Dennis COKKINOS, M.D.

SUMMARY

We present a case of a male suffering from hypertrophic peripheral neuropathy (Dejerine-Sottas disease) and severe involvement of the cardiac conductive tissue causing syncopal attacks. It is the first time that an association of this neuromuscular disease with cardiac involvement is described.

Additional Indexing Words:

Hypertrophic peripheral neuropathy Heart block

CARDIAC involvement occurs in several neuromuscular diseases.¹⁾ Severe abnormalities of the conduction system of the heart have been reported in peroneal muscular atrophy,²⁾ scapuloperoneal muscular atrophy,³⁾⁻⁵⁾ limb girdle dystrophy, and dystrophia myotonica.²⁾ However, no reference to cardiac conduction defects in hypertrophic peripheral neuropathy (Dejerine-Sottas disease) has been made.

We report a case of hypertrophic peripheral neuropathy which was associated with severe conductive tissue defects which necessitated permanent pacemaker implantation.

CASE REPORT

A 42-year-old man was admitted to hospital because he had developed dizziness and syncopal attacks 1 month prior to admission. His ECG (Fig. 1) showed a PR interval of 0.30 sec, LBBB, and 2:1 A-V block. Electrophysiological study (Fig. 2) showed that the A-H and H-V intervals were very prolonged (200 msec and 130 msec, respectively) and the second A wave was blocked distal to the bundle of His and was not followed by a V deflection. Cardiac x ray showed a normal cardiothoracic ratio.

From the Department of Cardiology, Tzanio State Hospital, Piraeus, Greece.

Address for reprints: George Kouvaras, M.D., Sparti 127, 185 46 Piraeus, Greece.

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Fig. 1. ECG of the patient showing a PR of 0.30 sec, LBBB, and 2:1 A-V block.

Fig. 2. Electrophysiological study of the patient. Paper speed is 50 mm/sec. Each interval between lines is 50 msec. The A-H interval is 200 msec and the H-V interval is 130 msec. The 2:1 block is Mobitz type 2.

Sinus node recovery time, corrected sinus node recovery time and sinoatrial conduction time calculated according to the method described by Narula et al⁶⁾ were found to be within normal limits.

The patient first noticed a feeling of heaviness in his legs and weakness and numbness in his feet 10 years ago. Gradually his legs became weak and he noticed loss of strength in his arms and hands. As the disorder progressed, the patient showed difficulty in walking, and the intrinsic foot muscles and peronei became very weak and wasted. He also experienced early sensory disturbances in cold and warm and pain sensations in his hands, which gradually increased. He developed an unsteady gait and he has been unable to

walk for the last 3 years without the aid of crutches. Lately, he has been confined to a wheelchair. On examination, he was emaciated and looked ill. There was a generalized muscle wasting in his limbs and the knee jerk and plantar reflexes were absent. There was severe impairment of postural sensation and vibration in the toes. Pain sensations showed impairment distally in all four limbs. The ulnar, peroneal and sural nerves bilaterally were visibly thickened and palpable through the skin. Electromyography showed reduced sensory action potentials, prolonged distal motor latency to the tibialis anterior and signs of denervation. Sural nerve biopsy showed the following results: in light microscopy there was a reduction in the density of myelinated fibers.

Sections stained with toluidine blue showed that the majority of large axons were surrounded by onion bulb whorls. Schwann cells were conspicuous and a few myelin sheaths were reduced in thickness. In electron microscopy the onion bulb whorls were prominent. The whorls were separated from each other by collagen. At the center of each onion bulb formation there was a single axon surrounded by its Schwann cell. Myelin sheaths were very thin (Fig. 3). These findings confirmed the diagnosis of hypertrophic peripheral neuropathy.

Family history: His mother had been treated for some kind of cardiopathy together with an unspecified neuromuscular disease. Her 2 brothers

Fig. 3. Sural nerve. Electron micrograph showing an onion bulb whorl. The axon (A) is thinly myelinated (M).

presented clinically the same neuromuscular disorder, but in a less progressive form. They both died suddenly in their 50's. The patient has one younger brother who is healthy.

A VVI pacemaker was implanted in the patient who has been free of cardiac symptoms since then. An echocardiogram (M-mode and 2D) performed the day of his discharge was normal.

DISCUSSION

Neuromuscular diseases have been associated with cardiac involvement such as cardiomyopathy,^{5),7)-9)} arrhythmias (Mawatari and Katayama 1973,⁴⁾ Leak 1961¹⁰⁾) and conduction disturbances.^{2)-5),8)} They constitute a rare cause of atrioventricular block and sinoatrial heart block.^{3),11)} Lambert and Fairfax³⁾ studied 892 patients with chronic heart block for evidence of neurological disorder. Six of them were found to have neuromuscular disease related to the conduction disturbance. Three of these patients had the scapuloperoneal syndrome, 1 the oculocraniosomatic syndrome, 1 limb girdle dystrophy and 1 dystrophia myotonica. In the last 2 patients cardiomyopathy was present in addition to the conduction disturbance.

Peroneal muscular atrophy has been associated with familial complete heart block.^{2),3)} It has also been reported that 15 of 37 males belonging to 2 families having an X-linked humeroperoneal neuromuscular disease showed a spectrum of electrocardiographic abnormalities that ranged from abnormal P waves to permanent atrial standstill and from first-degree atrioventricular block to complete heart block.¹²⁾

As far as we know, our case is the first one reported in the literature of Dejerine-Sottas disease associated with cardiac involvement. This cardiopathy could be attributed to selective degeneration of the conduction system of the heart.

Hypertrophic peripheral neuropathy is a rare illness inherited as an autosomal recessive trait¹³⁾ but X-linked inheritance cannot be ruled out.¹⁴⁾ It has been recently classified as hereditary motor sensory neuropathy type 3.¹⁵⁾ The underlying pathological process is a segmental loss of myelin.¹⁶⁾ The disease is named after Dejerine and Sottas¹⁷⁾ who described it in detail. The age of onset of the disease varies and symptoms may not develop until middle age. The disease often afflicts several members of the family, but sporadic cases have been reported. It presents with a variety of symptoms and Russel and Garland¹⁸⁾ considered that enlargement of nerves, which characteristically become easily palpable^{16),17)} and wasting of muscles are the only constant clinical features. The clinical picture consists of chronic pro-

gressive peripheral sensory and motor neuropathy with weakness, muscle wasting in the limbs and glove and stocking sensory loss.¹⁹⁾ Some patients become unable to walk without assistance.

The family history of the patient points to the familial type of the disease although the 3 members of his family died without having had a thorough neurological or cardiological investigation. The electrophysiological study, which we performed, disclosed involvement of the whole conduction system of the heart apart from the sinus node, since we found significant prolongation of the A-H and H-V intervals and the impulses were blocked to the bundle of His. We felt that there was an absolute indication for pacing, and indeed the patient has been free of syncopal attacks since pacemaker implantation. A review of the literature of electrophysiological studies carried out in patients with neuromuscular diseases reveals that the most common abnormality of the conductive tissue is prolongation of the H-V interval.^{6),19)-21)}

The patient's echocardiogram which was normal, militates against any cardiac muscle involvement, and considering his general ill-health we did not proceed to cardiac catheterization or myocardial biopsy.

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