Peroneal muscular atrophy associated with cardiac conducting tissue disease: further observations

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Summary

Two further cases of peroneal muscular atrophy associated with cardiac conduction abnormalities are described.

KEY WORDS: cardiomyopathy, cardiac conduction, hereditary neuropathies.

Introduction

Peroneal muscular atrophy (PMA) seldom involves the heart (Braunwald, 1980). However, an association between peroneal muscular atrophy (PMA) and cardiac conduction defects has been previously recorded (Littler, 1970). We report two further cases.

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Case 1

A 54-year-old man was diagnosed as having PMA 15 years earlier. His legs had an 'inverted bottle' appearance with marked wasting of the peroneal muscles and bilateral pes cavus. The tendon jerks were absent in the lower limbs whilst those in the upper limbs were diminished. The plantar responses were equivocal. There was no sensory deficit for the appreciation of light touch, pain or temperature and posterior column sense was intact. There was no evidence of cerebellar dysfunction. The optic discs were normal. He had no kyphoscoliosis or chest deformity.

His father probably had PMA; neither of his 2 sons have evidence of PMA or heart disease.

His first electrocardiogram in 1977 showed sinus rhythm with right bundle branch block and left axis deviation. Six months later, he developed atrial fibrillation (Fig. 1).

At the present time he is well, in atrial fibrillation at 70 beats/min with a blood pressure of 120/80 mmHg. There is no ventricular enlargement and his heart sounds are normal. His latest electrocardiogram shows coarse atrial fibrillation with right bundle branch block and left anterior hemiblock. An echocardiogram showed no evidence of valvular heart disease; the left ventricular end diastolic dimension measured 4-3 cm and the left ventricular end systolic dimension 3-1 cm, with an ejection fraction of 0-63. Systolic time intervals were measured and showed a pre-ejection period (PEP) of 100 ms and a left ventricular ejection time (LVET) of 760 ms, giving a ratio of 0-38. All these results are within normal limits.

Fig. 1. Twelve lead ECG (Case 1) demonstrating atrial fibrillation with right bundle branch block and left anterior hemiblock.

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Case 2

An unmarried 27-year-old man in whom a diagnosis of peroneal muscular atrophy was made a year earlier had pes cavus bilaterally and typical 'inverted bottle' legs with weakness of the peroneal muscles. The biceps muscles were thin bilaterally and weak and there was weakness of extension of the wrists and fingers with some small muscle atrophy in both hands. The knee jerks were feeble and the ankle jerks absent, the plantars giving no response. The arm jerks were all absent. There was no cutaneous sensory deficit in the limbs and there was no loss of posterior column sense. Apart from myopia, the cranial nerves were normal. There was no evidence of cerebellar dysfunction. The optic nerves were normal. He had no skeletal thoracic abnormalities.

A maternal second cousin had 'muscular dystrophy'.

In August 1979 he was admitted to a local hospital with central chest pain. His heart rate was 42/min and regular; blood pressure was 158/80 mmHg. His apex beat was displaced laterally with a soft, mid-systolic murmur at the apex. There was no evidence of cardiac failure.

A chest radiograph showed generalized cardiomegaly. The electrocardiogram demonstrated junctional escape rhythm with complete absence of atrial activity, a QRS frontal axis of -30° and normal QRS duration. There was no evidence of acute myocardial infarction or ischaemia (Fig. 2).

He was subsequently referred for coronary angiography which demonstrated normal coronary arteries including the branch to the sinus node. The left ventricle was slightly dilated with an ejection fraction of 0.33 and a left ventricular end diastolic pressure of 10 mmHg rising to 20 mmHg after angiography. The atrial standstill was confirmed.

Discussion

Although it is sometimes difficult to distinguish between cases of peroneal muscular atrophy, Friedrich's ataxia and the Roussy-Levy type of hereditary polyneuropathy, our two patients were considered by a consultant neurologist to fit in to the group of peroneal muscular atrophy. Though electrocardiographic changes are common in Friedrich's ataxia, heart block is not particularly associated with this condition and cardiac involvement is extremely rare in the Roussy-Levy syndrome (Lascelles, Baker and Thomas, 1970).

The evidence suggests that the cardiac conduction disorders associated with PMA are not necessarily secondary to a cardiomyopathy but represent a primary degeneration of the conducting tissue (Littler, 1970).

As well as having evidence of bifascicular block, Case 1 was in atrial fibrillation and atrial fibrillation was reported in one case by Ponder, Chatterjee and Sutton (1971); their second case being in atrial flutter. Leak (1961) described a case of paroxysmal atrial flutter associated with PMA. Our second case had complete atrial standstill. Although it is not possible on available evidence to exclude a cardiomyopathy confined to atrial muscle, the evidence suggests that the specialized conducting tissue in the atria is also involved in the degenerative process:

The evidence against a cardiomyopathy in PMA comes from the available data on cardiac muscle and its function. Cardiac biopsies were taken at the time of insertion of an epicardial pacemaker in two of Littler's original patients. One showed a normal myocardium on light microscopy alone whilst the second showed normal myocardium on light microscopy and electronmicroscopic sub-cellular changes compatible with simple cardiac hypertrophy (Kay, Littler and Meade, 1972). Ponder et al. (1971) demonstrated normal cardiac function and normal coronary arteriograms in one patient with complete heart block and PMA. The echocardiographic and systolic time interval findings on our first patient indicated normal ventricular function, whilst in our second case, the depressed ejection fraction and
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elevated left ventricular end diastolic pressure could be explained on the basis of atrial standstill.

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References