

Stroke Secondary To Atrial Paralysis In A Patient With EDMD

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Citation

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Abstract

We report stroke as a complication of cardiac involvement in Emery Dreifuss muscular dystrophy. A 30-year-old man with an existing diagnosis of this rare form of muscular dystrophy presented with symptoms suggesting cerebral ischaemia. Cranial imaging confirmed multiple cerebral infarctions. Atrial paralysis was discovered, implicating a cardioembolic stroke aetiology. Treatment with anticoagulation and permanent pacemaker insertion was instituted.

CASE REPORT

A 30-year-old man presented with a history of five episodes of expressive dysphasia and sensorimotor disturbance affecting the right arm. Each attack lasted between one and seven hours and had occurred over the preceding 12 months. Emery Dreifuss muscular dystrophy (EDMD) was diagnosed at the age of 16 years and subsequently confirmed by identification of a single nucleotide polymorphism in exon 3 of the emerin gene, Gln86X. This previously recognised mutation leads to premature truncation of emerin.

Examination revealed a short man with generalised muscle wasting, symmetrical weakness of the arms, legs, trunk and face, and contractures at the elbows and ankles - all features typically associated with EDMD. As there were no persistent neurological deficits associated with the thrombo-embolic episodes, a diagnosis of recurrent transient ischaemic attacks was made and aspirin commenced at 300 mg daily.

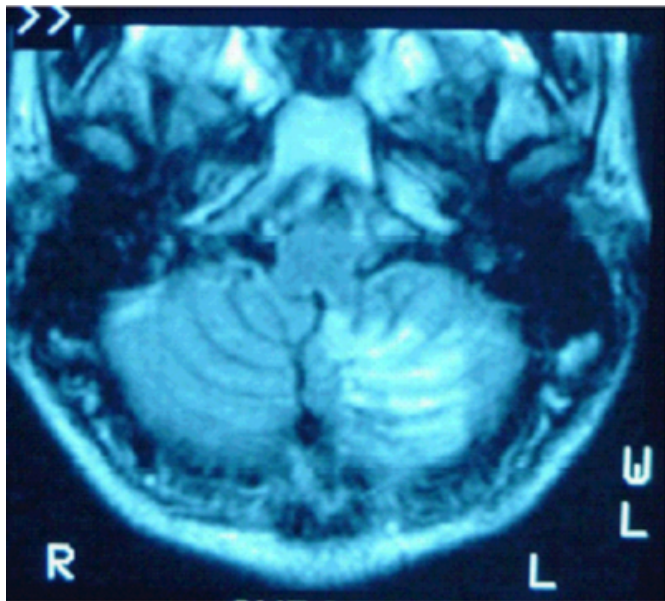
Before completing investigations he returned describing a two-hour episode consisting of vomiting, vertigo and occipital headache. Examination showed a regular pulse at a rate of 56 beats per minute and a soft systolic murmur at the left lower sternal edge. There were no signs of heart failure. Finger clubbing was present, but there were no other stigmata suggestive of infective endocarditis.

An ECG showed a regular heart rhythm with a complete absence of p waves. Review of his notes revealed an ECG showing similar findings dating back four years. Cranial computerised tomography showed established infarction of the right temporal and occipital white matter and an acute

left cerebellar hemispheric infarct in the territory of the posterior inferior cerebellar artery. These findings were confirmed by magnetic resonance imaging (figure 1). Imaging of the cervical vessels with magnetic resonance angiography and duplex ultrasound showed no focal arterial lesions. Blood testing for inherited and acquired prothrombotic disorders was unremarkable. Transthoracic and transoesophageal echocardiography revealed paralysis of a non-dilated left atrium. No intra-cardiac thrombus or valvular lesions were demonstrated. Cardiac dimensions and left ventricular contraction and filling were normal. Twenty-four hour ambulatory ECG monitoring showed occasional ventricular ectopic beats.

Figure 1

Figure 1: MRI (FLAIR sequence) demonstrating a high signal abnormality suggestive of infarction in the left cerebellar hemisphere.



A diagnosis of cardioembolic stroke was made and treatment was initiated with warfarin, aiming for a target International Normalised Ratio of 2.5. A ventricular demand pacemaker was inserted. A paralysed and electrically unexcitable right atrium was confirmed at pacing.

DISCUSSION

The patient described suffered from a potentially preventable complication of cardiac involvement in EDMD.

Cardioembolism appears to be the most likely stroke mechanism in this case, although we recognise that embolic stroke is infrequently thought to cause recurrent ischaemia affecting the same arterial territory as seen in this man's initial presentation.

EDMD was first recognised in 1966 [1]. Although uncommon, the precise incidence is unknown. The disease is characterised by progressive humeroperoneal weakness and contractures at the elbow and ankle developing during the second decade of life. The most common form, EDMD 1, displays an X-linked recessive inheritance pattern, although 10 to 20% of females manifest clinical features due to skewed X-inactivation [2]. The disease was mapped to Xq28 in 1994 and a gene coding for emerin was identified [3]. Diagnosis can be confirmed by the absence of emerin in muscle, skin or buccal smears.

Cardiac complications of EDMD are common, though rarely the presenting feature. Adipose and fibrous replacement of

myocardium leads to atrial arrhythmias and a dilated cardiomyopathy. Cardiac involvement is the major determinant of prognosis. Merlin [4] described sudden death in 20 of a cohort of 73 patients with EDMD aged between 25 and 59 years. Paroxysmal atrial fibrillation or flutter is frequent and often degenerates to atrial paralysis [5]. The atria are characteristically electrically unexcitable once atrial paralysis develops, with electrocardiography showing a junctional escape rhythm at a rate of 40 to 50 beats per minute and an absence of p waves. A third of all cases of permanent atrial paralysis are accounted for by those associated with EDMD [6]. Stroke complicating EDMD has been reported recently, occurring in 4 subjects with EDMD between the ages of 26 and 70 years [5]. However, unlike the case described, in each of these reports cardioembolic stroke resulted from atrial fibrillation or flutter.

The ability of screening electrocardiography and echocardiography to identify those who are at risk of cardioembolic events and sudden death in EDMD is uncertain. The incidence of cardiac arrhythmias increases with age [2] and can occur in the absence of skeletal muscular abnormalities in EDMD, therefore the most appropriate age to commence screening and test frequency is unclear. Dual chamber pacemaker implantation does not prevent the development of atrial arrhythmias in EDMD [5]. Though the risk of embolic phenomena in EDMD complicated by cardiac arrhythmias is likely to be high, there are no trials examining whether the use of anticoagulation or anti-platelet agents in this situation lead to improved clinical outcome.

The objective of this case report is to highlight the risk of stroke in patients with EDMD. Early recognition of atrial arrhythmias and paralysis may enable preventative interventions to be instituted. A prospective study to validate a cardiac screening and treatment protocol for patients with EDMD is needed.

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