Myotonic Dystrophy, Mitral Valve Prolapse, and Cerebral Embolism

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SUMMARY A second case displaying an association between mitral valve prolapse, myotonic dystrophy, and cerebral embolism is described. Mitral valve prolapse may be associated with myotonic dystrophy more often than is recognized.

CARDIAC abnormalities are the rule in hereditary myopathy, and myotonic dystrophy (MD) is no exception. Conduction disturbances are most common, manifested by first degree atrioventricular block,1, 2, 3 intraventricular conductive defect,4 and by evidence of disease in the entire conductive system on His bundle electrogram.5

The association between mitral valve prolapse (MVP) and MD has been reported as a familial trait.6 Cook et al have reported a patient with MD, MVP, and cerebral infarction.7 However, there are grounds for questioning the source of the embolism in their patient. We report a second patient with MD, MVP, and a transient ischemic attack in whom we believe the mitral valve was the source of the embolization.

Case Report

A 41-year-old left-handed white male was admitted to the hospital after the sudden onset of aphasia and clumsiness of the right hand. These symptoms cleared completely in two hours. He was known to have myotonic dystrophy and mitral valve prolapse. There were no other family members with cardiovascular disease or myotonia, although his father had cataracts in his 50's.

Examination on admission showed the characteristic stigmata of myotonic dystrophy with frontal balding, hatchet-like face, mild bilateral facial weakness, and early cataracts. There was grip and percussion myotonia of the hands and minimal weakness. Additionally, a late systolic murmur was heard on auscultation.

M-mode and 2-D echocardiography confirmed mitral valve prolapse. Oculoplethysmography was normal; doppler examination revealed no bruits, and aortic arch and bilateral carotid arteriograms were normal. Electromyography was consistent with myotonic dystrophy. CPK was 67 U/L and glucose 93 mg/dl.

The patient was treated with aspirin and dipryridamole and had no further transient ischemic attacks during his one week hospital course.

Discussion

The association between MVP and MD was initially described in a large kindred by Winters, et al.6 It remains uncertain whether MVP will prove to be another characteristic of MD or only of certain kindreds. Only one similar patient has been reported.7

Cook and his colleagues described the first patient with MVP and MD to suffer cerebral infarction.7 Their patient was chronically ill with hyperthyroidism, polycystic kidney disease, and cardiac arrhythmias including atrial fibrillation. He died as a result of his stroke. His terminal left cerebral infarction had been preceded by a right hemispheric TIA ten days previously supporting the idea of cardiogenic embolization. However, the postmortem clot found in the carotid artery was of platelet-thrombin composition and similar material was not found in the heart. The carotid bifurcation was not examined by angiogram or at postmortem. Our patient had suffered no previous episodes suggesting cardiac arrhythmia, had a normal EKG, and had no evidence of a lesion at the carotid bifurcation that would be likely to generate emboli.

The basic problem in MVP consists of an abnormal enlargement of the superficial area of the mitral leaflets, so that the leaflet tissue becomes redundant and prolapses into the left atrium during ventricular systole.8 It seems likely that constant trauma to valvular surfaces leads to patchy loss of endothelium, formation of small platelet and fibrin thrombi, and subsequent embolization to the systemic circulation.9

Indeed, cerebral ischemic events and strokes associated with the presence of MVP have been reported in patients without evidence of arteriosclerotic cerebral vascular disease, hypertension, or coagulation defect.10-14 Barnett reported a series of 12 patients with recurrent TIA's and partial nonprogressive strokes. Cerebral angiography showed no significant arteriosclerotic vascular disease but angiocardiology and echocardiography demonstrated mitral valve prolapse. The average age was 35 years, compared with 62 years in a larger series of patients with TIA associated with arteriosclerosis. Similarly, a wide range of ophthalmologic manifestations, including amaurosis fugax, have been reported in young people with MVP and normal noninvasive cardiac and systemic testing.15

MVP is not the only cause of cerebral symptoms in MD. The conductive system abnormality, which predisposes these patients to suffer asystole or ventric-
ular fibrillation, may produce syncope or ischemic cerebral infarcts, especially in patients with coexistent carotid stenosis. These abnormalities were not present in the patient reported here.

We conclude that the association of MVP and MD is not a fortuitous one, and therefore should be sought in all patients with MD. Since the presence of MVP in patients with MD represents a potential source of neurologic problems, the symptomatic supportive treatment of these patients should include periodic cardiological evaluation and prophylaxis against infective endocarditis at the time of oral surgery. The use of antiplatelet agents should be considered in patients over 40 years of age with MVP.

References

Problems in Design of Stroke Treatment Trials

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SUMMARY Critical evaluation of the literature was used to identify remediable flaws in the design of clinical trials of stroke treatment. Trials of dexamethasone, dextran, and glycerol were reviewed. Available studies have in common major weaknesses in case selection (failure to exclude arteriolar strokes due to hemorrhage or lacunar infarction), and failure to estimate required sample size. Problems of case selection can be avoided with computerized tomography; the sample size required to show superiority of active treatment over placebo can be estimated using standard formulas. Prognostic stratification is suggested as a method of overcoming problems of unbalanced allocation. Further studies with improved design are required to evaluate the prospects for medical limitation of cerebral infarct size.
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