Clinical Implications of MRI To Assess Cardiac and Pulmonary Function in Patients With Duchenne Muscular Dystrophy

To the Editor:

We read with interest the article in CHEST (January 2005) by Mavrogeni et al about 17 patients with Duchenne muscular dystrophy (DMD), aged 7 to 25 years and without cardiac or pulmonary disease, in whom T2-relaxation time of the sternocleidomastoidei muscles and the myocardium indicated abnormal tissue composition, and why this technique was proposed for monitoring cardiac and pulmonary function in these patients. The presented findings raise the following concerns.

In the light of previous reports, it is unusual that none of the 17 patients had clinical or subclinical cardiac disease. This is particularly curious because 12 patients were between 13 and 25 years old and because the prevalence of cardiac abnormalities in patients with DMD increases with age. Nearly all patients with DMD develop sinus tachycardia and in a series of 328 patients with DMD 25% had subclinical cardiac involvement before age 6 years and 59% between ages 7 and 10 years. Clinical cardiac disease develops after loss of ambulation and is present in all patients after 18 years of age. It is also uncommon that all patients with DMD had reduced T2-relaxation times, but none had any other abnormality on any other instrumental cardiac investigation. Were the cardiac investigations not carried out with proper care or attention? Which abnormalities of the skeletal muscle and myocardium can be determined by the T2-relaxation time, and which cannot be detected by any other method?

In a recent study, dystrophic changes within the myocardium before onset of overt cardiomyopathy or systolic dysfunction were detected on tissue Doppler imaging. Was tissue Doppler imaging carried out and compared with the MRI findings?

To effectively monitor cardiac and respiratory function in patients with DMD, more than a single MRI measurement is warranted. How can therapy be indicated for dilative cardiomyopathy in patients with DMD if only T2-relaxation time is measured? How can the right moment for starting noninvasive positive pressure ventilation be known if no capnographic examinations have been carried out? It is essential and recommended to apply a variety of clinical and instrumental cardiac and pulmonary investigations before eventually starting an appropriate therapy. We regard it as particularly irresponsible to propose a method without comparing it with reliable, well-established techniques beforehand.

Dystrophic alterations in the skeletal muscle of patients with DMD usually follow an inhomogeneous and patchy distribution. If a defined site within the muscle is investigated using MRI, it cannot be determined whether there is an extensive or scarce presence of dystrophy within this area. To correlate the MRI findings with the severity of the neurologic abnormalities, it would be useful to carry out a clinical neurologic investigation of each patient at the time when the MRI is carried out.

Which ECG and echocardiographic abnormalities did the authors particularly look for? If certain ECG and echocardiographic abnormalities are not expected to occur, they may be easily overlooked. This is particularly the case with echocardiographic abnormalities like left ventricular hypertrobracelation, which has been recently described for the first time, to our knowledge, in a patient with DMD.

From 10% to 20% of the patients who are carriers of DMD are cardiologically and neurologically affected. Did the authors also investigate the mother of any patient? Did they find cardiac disease in any of these women?

Overall, monitoring of cardiac and pulmonary function in patients with DMD should not only rely on a single MRI parameter, but should also include various clinical and instrumental techniques. Before proposing a new technique as a means to monitor cardiac or pulmonary disease, the sensitivity and specificity of such a technique in comparison with an established technique should be assessed. Furthermore, the additional value of such a technique should be demonstrated.

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References

Association of Chronic Mountain Sickness With Abnormal Pulmonary Microcirculation

Importance of Adjusting Predicted Diffusing Capacity of the Lung for Carbon Monoxide for Altitude, Hemoglobin, and Lung Volume

To the Editor:

A recent issue of CHEST (February 2010) published findings by Stuber et al. of large increases in pulmonary hypertension with mild exercise in chronic mountain sickness (CMS). It is important, however, to know whether the findings are associated with abnormal pulmonary micorcirculation (using difusing capacity of the lung for carbon monoxide [DLCO] as proxy). To better evaluate whether CMS has abnormal gas exchange, the authors

Response

To the Editor:

Finsterer and Stöllberger present some interesting comments on our article about cardiac and sternocleidomastoid muscle involvement in Duchenne muscular dystrophy (DMD) studied by MRI. We underestimated neither the importance of clinical neurologic and cardiac examination, nor the use of the currently applied techniques in the assessment of DMD patients. Although the technologic progress is of great value, there is no doubt that the patient’s clinical evaluation remains the cornerstone of medicine.

In this article, our purpose was not to underestimate the value of traditionally used approaches; instead, we emphasized the important role of a new technology in the early detection of subclinical cardiac involvement in Duchenne muscular dystrophy. In Ref. 1 we underestimated neither the importance of clinical neurologic and cardiac examination, nor the use of the currently applied techniques in the assessment of DMD patients. Although the technologic progress is of great value, there is no doubt that the patient’s clinical evaluation remains the cornerstone of medicine.

We should also emphasize that DMD patients can nowadays have different clinical presentations, depending on the early use of angiotensin-converting enzyme inhibitors and/or deflazacort, the patient’s supportive treatment, close cardiac and neurologic evaluation, and so forth. Cardiac involvement is not necessarily found in all patients at the time of ambulation loss. Additionally, ECG changes are not necessarily found in all DMD patients. Furthermore, the pathophysiology of cardiac lesions in DMD is more complicated than initially believed. Although it is known that the absence or decrease of dystrophin leads to progressive skeletal muscle and heart failure, it has also been documented recently that abnormal dystrophin can act as a potential susceptibility gene for viral infection of the myocardium and as a factor that markedly increases enterovirus-induced cardiomyopathy.

We did not perform Doppler tissue imaging, which can also offer early information about cardiac involvement, and we did not compare our results with new echo techniques because these techniques are not available everywhere and all echocardiographers are not familiar with them. Our purpose was to emphasize the capability of MRI to perform tissue characterization and its possible use for early detection of subclinical cardiac involvement in DMD. Further comparative studies are needed to establish the role of each technique. However, there is no doubt that MRI proved to be of great importance in the early detection and long-term follow-up of many diseases with cardiac involvement and also was able to detect cardiac lesions missed by echocardiography (myocarditis, cardiomyopathies, thalassemia, and so forth).