Early detection of cardiomyopathy in Duchenne muscular dystrophy, is there a role for MRI?

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Muscular dystrophies are progressive myopathic disorders resulting from defects in a number of genes required for normal muscle function [1]. Duchenne muscular dystrophy is caused by a defective gene located on the X chromosome, that is responsible for the production of dystrophin [2] and is related to other inherited myopathies like Emery–Dreifuss muscular dystrophy, Becker muscular dystrophy, limb-girdle muscular dystrophies, fascioscapulohumeral muscular dystrophy and myotonic dystrophy. Duchenne muscular dystrophy has a recessive inheritance. The disease is clinically associated with muscle fiber degeneration. The incidence is one in 3,500 live male births, with a prevalence of 3 per 100,000 [3]. The clinical onset of weakness usually occurs between 2 and 3 years of age [4]. One of the major complications of Duchenne muscular dystrophy is a dilated cardiomyopathy. The main cardiac expression of the disease is extensive fibrosis of the posterobasal and lateral left ventricular wall. In patients with Duchenne muscular dystrophy, the incidence of the cardiomyopathy increases from zero under the age of 10, to one-third at the age of 14 years to one half at the age of 18 years and nearly 98% in patients over 18 years [5]. The incidence of cardiac failure in female carriers is

reported to be 10% even in the absence of skeletal muscle involvement [6]. However, in these females carriers, life expectancy is not affected by the presence of the abnormal gen [7]. The cardiomyopathy can be detected by an abnormal ECG characterized by tall right precordial R waves with an increased R/S ratio and deep Q waves in the lateral leads and by echocardiography showing localised lateral thinning and akinesia and noncompaction of the endocardium ultimately progressing in a dilated cardiomyopathy [8–10]. For early and appropriate detection of cardiac involvement of muscular dystrophy, a recent workshop on the management of cardiac involvement in muscular dystrophy gave the following recommendations:

All patients with Duchenne muscular dystrophy should have a cardiac investigation (ECG and echocardiogram) at diagnosis.

Next to this, patients with Duchenne muscular dystrophy should have cardiac investigations before any surgery, every 2 years to the age of 10 and annually after the age of 10 [11].

In female carriers an ECG and echocardiogram should be performed at diagnosis or after the age of 16 years and at least every 5 years thereafter, or more frequently in patients with abnormalities on investigation or carriers with severe skeletal muscle symptoms or cardiac symptoms.

The current study from Puchalski et al. and other studies show that gadolinium enhanced MRI is able to

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detect cardiac fibrosis in patients with Duchenne muscular dystrophy [12, 13]. The study showed, that late gadolinium enhancement was negative related to left ventricular ejection fraction. The cardiac involvement was typically in inferolateral free wall and the basal inferior and anterolateral region of the left ventricle with sparing of the interventricular septum and the right ventricle. Unfortunately, the authors did not compare the MRI findings with electrocardiographic or echocardiographic findings. This is of clinical importance, because if MRI would be able to detect cardiac involvement in patients with Duchenne muscular dystrophy, early treatment with angiotensin converting enzyme inhibitors or beta blockers could be initiated. Therefore, there is a need for a comparative study with electrocardiography, echocardiography and MRI for early detection of cardiac involvement in Duchenne muscular dystrophy. After this the main effort has to be early treatment and prevention of left ventricular dysfunction in this particular group of patients.

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