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EDITORIAL COMMENT

Duchenne muscular dystrophy; a cardiomyopathy that can be prevented?

Barbara J. M. Mulder · Ernst E. van der Wall

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Cardiac magnetic resonance imaging (CMR) has long been recognized as an accurate and reliable means of evaluating cardiac anatomy and ventricular function. Considerable progress has been made in the field of CMR, providing accurate evaluation of myocardial ischemia and infarction [1–3]. Contrast-enhanced CMR can be used to visualize the transmural extent of myocardial infarction with high spatial resolution [4–6]. Infarcted myocardium appears hyperenhanced compared with normal myocardium when imaged by a late enhancement MRI technique with the use of T1-weighted sequence after injection of gadolinium chelates. Late gadolinium-enhanced CMR can clearly delineate subendocardial infarction and the transmural extent of delayed enhancement potentially predicts functional outcome after revascularization in acute myocardial infarction and chronic ischemic heart disease [7]. Stress first-pass contrast-enhanced myocardial perfusion CMR can be used to detect subendocardial ischemia and recent studies have demonstrated the high diagnostic accuracy of stress myocardial perfusion CMR for detecting significant coronary artery disease [8–10]. Magnetic resonance angiography (MRA) was recently introduced as a method that can provide visualization of all three major coronary arteries, coronary bypasses, and the aorta within a single three-dimensional acquisition [11, 12]. CMR has become the first choice imaging modality in complex congenital heart disease [13–16] and imaging great vessels [17, 18]. In recent years, late gadolinium enhancement CMR has also been used to visualize myocardial interstitial abnormalities. Previous studies have clearly shown late enhancement patterns in patients with different forms of cardiomyopathies, amyloidosis, myocarditis, and storage diseases [19–22]. Silva et al. [20] showed that late gadolinium enhancement can be demonstrated in cardiomyopathy patients, with a mean signal intensity of 390 ± 220% compared with normal regions. The affected areas included papillary muscles (sarcoid), the mid-myocardium (Anderson-Fabry disease, glycogen storage disease, myocarditis, Becker, and Duchenne muscular dystrophy) and the global subendocardium (systemic sclerosis, Loeffler's endocarditis, amyloid, and Churg-Strauss). Focal myocardial late gadolinium enhancement is found in the specific cardiomyopathies, and the pattern is distinct from that seen in myocardial infarction, recent studies have demonstrated right ventricular late gadolinium enhancement in patients with congenital heart disease and right ventricular loading conditions [14, 23]. Myocardial preservation is likely a multifactorial process that may affect the right and left ventricles differently [24–28].

B. J. M. Mulder
Cardiology Department, B2-240, Academic Medical Center, Meibergdreef 9, 1105 AZ Amsterdam, The Netherlands

E. E. van der Wall (✉)
Department of Cardiology, C5-P28, Leiden University Medical Center, Leiden, The Netherlands
e-mail: e.e.van_der_wall@lumc.nl

In an article in the present issue of the International Journal of Cardiovascular Imaging, Puchalski et al. [29] studied late gadolinium enhancement with CMR in 74 patients with Duchenne muscular dystrophy. Duchenne's disease is an X-linked recessive inherited disorder with an annual incidence of 1/3,000 live male births. Pre-clinical cardiac involvement has been found in 25% of patients under 6 years old increasing to 60% between the ages of 6 and 10 years and then declining in incidence with age. Clinically apparent cardiomyopathy is first evident after 10 years of age and increases in incidence with age, being present in all patients over 18 years of age. Unlike in connective tissue disease such as Marfan [30], progressive cardiomyopathy is a common cause of death in Duchenne muscular dystrophy, presumably secondary to fibrosis of the myocardium. The posterobasal and left lateral free wall of the left ventricle are initial sites of myocardial fibrosis pathologically. The purpose of the present study [29] was to assess whether late gadolinium enhancement could identify fibrosis in selective areas of the myocardium. In addition, the relationship between the presence and extent of fibrosis and left ventricular function was established. Of the 74 patients, 24 patients (32%) had late gadolinium enhancement involving the posterobasal region of the left ventricle in a subepicardial distribution. Those patients with more involvement had spread to the inferior and left lateral free wall with progressive transmural fibrous replacement. There was relative sparing of the interventricular septum and right ventricle. As a result, late enhancement was able to detect fibrosis in selective regions of myocardium in patients with Duchenne's disease. The presence of late gadolinium enhancement was associated with diminished cardiac function. Recently, Silva et al. [31] showed in 10 patients with muscular dystrophy that CMR could identify myocardial fibrosis and may be useful for detecting the early stages of cardiomyopathy in myocardial dystrophy. Myocardial fibrosis (midwall and/or subepicardial) was observed in 7 out of 10 patients, and the lateral wall was the most commonly involved segment. There was moderate correlation between segmental myocardial fibrosis and left ventricular dysfunction. Another recent CMR study in 32 young patients with Duchenne muscular dystrophy by Mertens et al. [32] showed a global normal systolic function, but reductions in systolic deformation parameters as well as reduced early diastolic myocardial velocities in the anterolateral and inferolateral left ventricular walls. The prognostic significance of these findings warrants further longitudinal follow-up studies to determine whether late gadolinium enhancement precedes a decrease in cardiac function, and whether early pharmaceutical interventions are useful in preventing progression of Duchenne muscular dystrophy. Recent clinical studies with corticosteriods have already shown promising results by their potential to retard development of left ventricular dysfunction [33, 34]. Mavrogeni et al. [33] showed that Duchenne patients on deflazacort are characterized by better preservation of the CMR T2 relaxation time of the myocardium and an improved left ventricular systolic function. Markham et al. [34] demonstrated that treatment of Duchenne patients with steroids was protective against left ventricular dysfunction. This indicates that steroid treatment, when started prior to ventricular dysfunction, retarded the anticipated development of ventricular dysfunction in Duchenne patients. Larger functional studies—preferably with CMR—are still needed to establish the true value of corticosteroids in delaying the development of cardiomyopathy in patients with Duchenne muscular dystrophy.

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