

Early detection of iron overload in the heart: a key role for MRI!

Leo H. B. Baur

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Hereditary haemochromatosis is autosomal recessive disorder associated with the HLA-A locus on the short arm of chromosome 6 [1]. It is the most common genetic disorder worldwide [2]. About 56,000 babies are born each year with thalassaemia major, of whom 30,000 require lifelong transfusions to survive [3]. The disease is very prevalent in European countries along the Mediterranean Sea and in South East Asia. In other countries it becomes a problem because of immigration from these areas. Excessive iron deposition occurs mostly in the pancreas, pituitary, but also in the heart. In normal subjects, absorption of iron from the gut is inversely correlated with iron stores. In homozygous hereditary haemochromatosis there is increased absorption of iron, which is not regulated by iron stores [4]. This leads to progressive iron accumulation because there is no normal mechanism for excretion of excess iron once it has been absorbed into the body. The clinical manifestations of iron overload include liver disease in 75% of cases, weakness and lethargy in 74%, skin hyperpigmentation in 70%, diabetes mellitus in 48%,

arthralgia in 44%, impotence in 45%, electrocardiographic abnormalities in 31% and heart failure and conduction disturbances in 15% [5, 6]. Heart failure remains a major cause of death in these patients, frequently people younger than 35 years [7]. In the majority of patients iron induced cardiomyopathy can be reversed by intense chelation therapy with desferrioxamine and deferiprone [3, 8]. At this moment T2* magnetic resonance imaging is the best way for early detection of iron overload in patients with haemochromatosis [9–11]. T2* is an unique magnetic resonance parameter measured in milliseconds and shortened in relation to the level iron in the heart. The parameter can be used for screening haemochromatosis patients and to guide chelation therapy in those with established cardiac haemochromatosis [8]. The paper of Mavrogeni et al. [12] in this issue of the International Journal of Cardiac Imaging shows, that heart failure patients with severe iron overload indicated by a T2* value less than 10 ms had a lower exercise capacity than patients with a T2* value higher than 10 ms. The authors also show, that there was a excellent relation of the extent of iron overload measured by T2* and exercise tolerance in thalassemia patients. As shown by others, thalassemia patients present prolonged phase II on-transient oxygen kinetics during submaximal, constant workload exercise, compared with healthy controls, possibly suggesting a slower rate of high energy phosphate production and utilization and reduced oxidative capacity of myocytes [13]. This could also

Editorial comment on the article of Mavrogeni et al.
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L. H. B. Baur (✉)
Department of Cardiology, Atrium Medical Centre
Parkstad, University of Maastricht, Henri Dunantstreet 5,
6401 CX Heerlen, The Netherlands
e-mail: lbr01@atriummc.nl

account for their significantly limited exercise tolerance. Reduction of exercise tolerance is also highly associated with the limited functional status of peripheral muscles in these patients [14]. This study shows again that T2* MRI plays a crucial role in screening cardiac haemochromatosis and guidance of chelation therapy. Early detection of cardiac iron load and early initiation of treatment are crucial for survival of these patients. Recently the chelator deferasirox has been shown to be more effective in better at improving myocardial iron [15]. This could improve outcome in thalassemia patients. As shown, early effects of therapy can only be detected with dedicated MRI sequences. Unfortunately these MRI scanners are not always available in those areas, where the prevalence of haemochromatosis patients is extremely high.

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