Extreme myocardial siderosis in transfusion-dependent thalassaemia; myocardial tissue characterisation pivotal in guiding management

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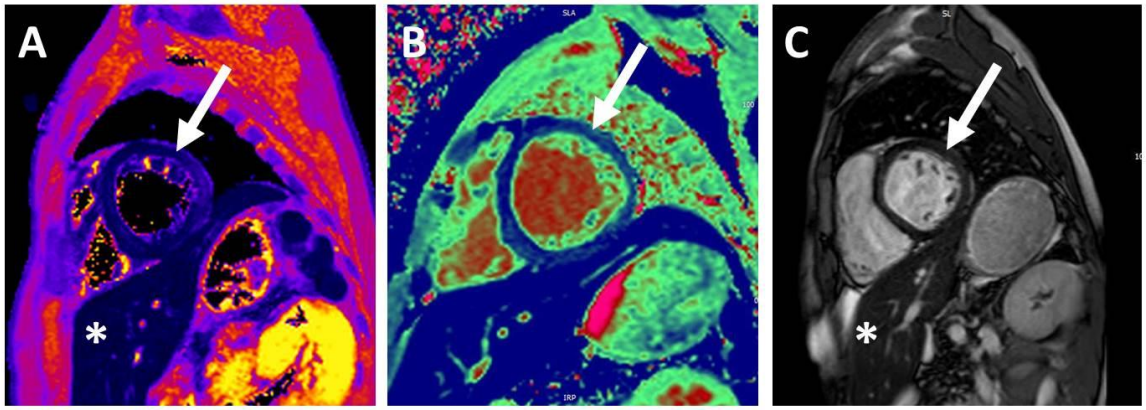
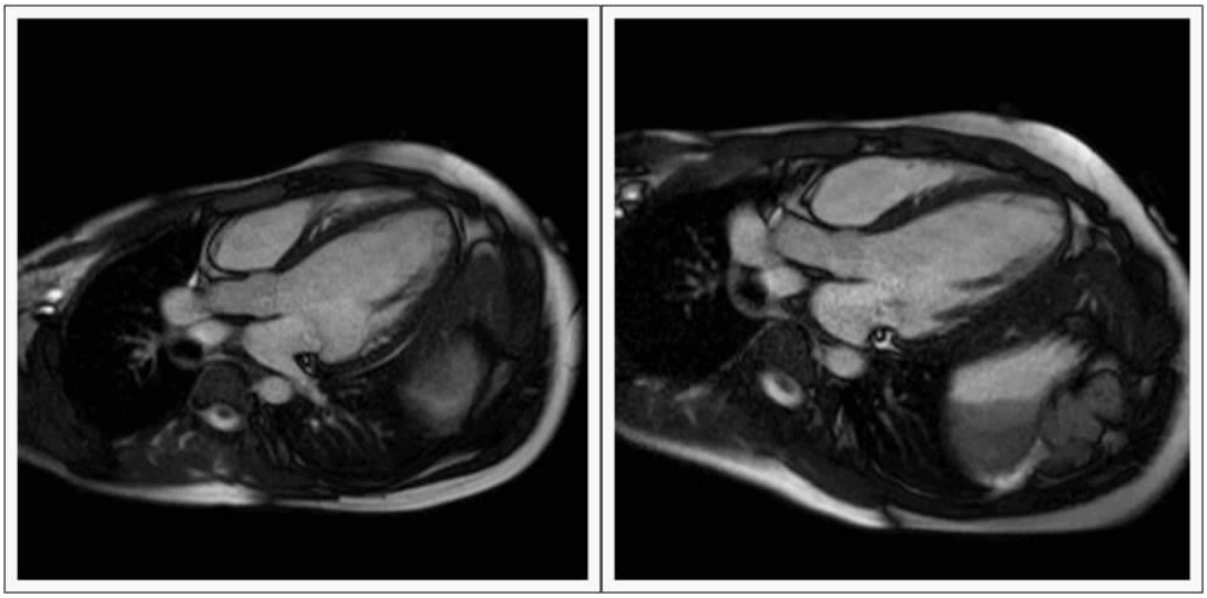
A 22-year old man receiving monthly blood transfusions for longstanding beta-thalassemia major presented with palpitations. He had a history of hypogonadotropic hypogonadism, paroxysmal atrial fibrillation in the context of severe transfusion-related iron overload for the last 10 years, splenectomy in childhood and multiple complications from different chelators; including life-threatening klebsiella sepsis with desferrioxamine, severe agranulocytosis with deferipone and gastric ulceration and renal tubular acidosis with deferasirox. More recently, he was diagnosed with insulin-dependent diabetes and suffered an ischaemic stroke.

Cardiac magnetic resonance (CMR) imaging revealed moderate liver (Panel A, asterix, liver T2* relaxation time 4.2ms, normal >17ms) and severe myocardial iron loading (Panel A, white arrow, myocardial T2* relaxation time of 4.7ms, normal >20ms, severe <10ms), causing left ventricular dysfunction (ejection fraction [EF] 44%, supplementary video 1). Native myocardial T1 (Panel B, white arrow) was 563ms (normal 1020 ± 60ms at 1.5 Tessla). Both liver and myocardium appeared dark on the short axis scout images (Panel C, white arrow and asterix). He received aggressive iron chelation treatment with oral Deferipone and continuous intravenous Desferrioxamine, via a portacath, with improved symptoms, reduced left ventricular end-diastolic volume and normalisation of systolic function on subsequent CMR (EF 60%, supplementary video 2).

Heart failure and arrhythmia due to myocardial iron infiltration carry a poor prognosis and are the main mechanisms of premature death in thalassemia. CMR reliably detects and quantifies myocardial iron and is used to guide chelation therapy with substantial improvement in survival and quality of life for transfusion-dependent patients.

Word count 247





Supplemental videos 1 and 2. Left, three chamber cine showing left ventricular systolic dysfunction with an ejection fraction of 44%. Right, Normalisation of function after intensified chelation therapy.