Cardiac manifestations of McArdle's disease

Daniel Jones, Luis Lopes, Perry Elliott, Mohammed Y Khanji

A 69-year-old Caucasian male with McArdle's disease (homozygous R50X mutation in the gene *PYGM*) and a hypertrophic cardiomyopathy (HCM) phenotype presented with episodes of atypical non-exertional chest pain with a satisfactory exercise tolerance and chronic troponin elevation. His brother and sister had mild HCM phenotypes. He had previous ischaemic heart disease with multivessel percutaneous interventions (stented RCA, LAD, OM1 and D1 coronary vessels). Electrocardiography revealed left axis deviation, inferolateral T wave flattening and late progression of QRS in the precordial leads (Panel A). Coronary angiography showed moderate residual distal LAD disease. Cardiovascular magnetic resonance showed good left ventricular function (ejection fraction 66%) with asymmetrical septal hypertrophy. Multiple areas of patchy sub-endocardial adenosine induced perfusion defects were seen (predominantly basal to apical septum and anterior segments) more suggestive of microvascular ischaemia (Panel B) with extensive non-ischaemic pattern of late gadolinium enhancement (LGE) consistent with myocardial fibrosis (Panel C-E), including near transmural LGE in the basal-mid anterior segments, patchy mid wall LGE of mid-septum, mid inferoseptum, inferior and apical-lateral segment.

Our patient displayed all known rarely reported cardiac manifestations of McArdle's disease, including electrocardiographic changes, coronary artery disease and HCM. McArdle's is a rare autosomal recessive glycogen storage metabolic myopathy caused by mycophosphorylase deficiency, predominantly affecting skeletal muscle. Typical features include early fatigue, exercise intolerance with a second wind phenomenon and myoglobinuria. Treatment include those specifically targeted to McArdle's and those to manage the systemic manifestations. Targeted treatments showing some benefit include carbohydrate rich diets and simple carbohydrates prior to aerobic exercise.

Word count 250

1. D. Nicholls, N. Campbell, H. Stevenson and V.Patterson. Angina in McArdle's disease (1996). Heart (76): 372-373.
2. S. Moustafa, D. Patton and M. Connelly. Unforseen Cardiac Involvement in McArdle's disease (2013). Heart, Lung and Circulation (22): 769-771.
3. R. Quinlivan, A. Martinuzzi and B. Schoser. Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V) (2014). Cochrane Database Systemic Review.
4. J. Vissing and RG. Haller. The effect of oral sucrose on exercise tolerance in patients with McArdle's disease (2003). New England Journal of Medicine 349 (26): 2503-9.

Abstract (30 words)

McArdle’s disease is a rare metabolic myopathy and cardiac manifestations are rare. We present a patient with this condition with ongoing chest pain who displays all the known cardiac phenotypes.