Utility of Cardiac MRI in Anderson Fabry Disease

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Indications and Purpose of the Scan

- CMR is an excellent way to non-invasively diagnose cardiac involvement in Anderson Fabry disease (AFD). CMR can help differentiate Fabry disease from other potential causes of cardiomyopathy. CMR is especially valuable in diagnosing patients mislabelled as hypertrophic cardiomyopathy.

Why CMR (specific advantages)

- CMR provides a non-invasive method to assess structure, function, and abnormal scar in the extracellular tissues via late gadolinium enhancement (LGE). Concentric thickening and inferolateral mid-myocardial scar are the most common manifestations of AFD, but apical and asymmetric septal hypertrophy has also been described. Significant hypertrophy is associated with ventricular arrhythmia. Male patients have higher incidence of hypertrophy compared to female patients. However, among men, myocardial fibrosis generally occurs in those with left ventricular hypertrophy. In women, myocardial fibrosis can occur without left ventricular hypertrophy.
- Noncontrast T1 mapping was found to be a useful measure in patients with Fabry disease. The values are significantly lower in Fabry disease, compared to healthy volunteers and patients with other confounding diseases (aortic stenosis, hypertrophic cardiomyopathy, hypertension) causing left ventricular hypertrophy. T1 values showed pseudonormalization or elevation in the left ventricular inferolateral wall, correlating with the presence or absence of late gadolinium enhancement. (1, 2) Female subjects had lower LV mass and wall thickness, longer myocardial T1 values and larger extracellular volume suggesting a key sex difference in cardiac remodeling (2).

Evidence

- Deva et al. Cardiovascular magnetic resonance demonstration of the spectrum of morphological phenotypes and patterns of myocardial scarring in Anderson- Fabry disease. JCMR 2016 18:14
  - 39 patients were included in this study. Majority of the patients had wall thickening, with concentric wall thickening being the most common pattern. Majority had pathologic scar, mid myocardial inferolateral scar being the most common.
  - Fabry cardiomyopathy in male patients is characterized by left ventricular hypertrophy, impaired myocardial function, and subsequent progressive myocardial fibrosis. In contrast to male patients, the loss of myocardial function and the development of fibrosis do not necessarily require myocardial hypertrophy in female patients with Fabry disease.
  - 32 patients were studied over 3 years regarding disease progression and clinical outcome under enzyme replacement therapy. Patients without fibrosis showed a significant reduction in left ventricular mass and improvement in myocardial function, compared to those with mild or severe fibrosis.
  - Pathological correlation of scar with delayed gadolinium enhancement in a patient with Fabry disease.

Contraindications

- Any implanted device that is not MRI compatible
- Inability to lie flat
- Inability to tolerate the scan
- Altered mental status
- Severe arrhythmias
References

1. Deva et al. Cardiovascular magnetic resonance demonstration of the spectrum of morphological phenotypes and patterns of myocardial scarring in Anderson-Fabry disease. JCMR 2016 18:14