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Quantitative perfusion mapping in Fabry disease

Authors:
K Knott¹, JB Augusto¹, S Nordin¹, R Kozor², C Camaioni³, H Xue⁴, RK Hughes¹, C Manisty¹, LAE Brown⁵, U Ramaswami⁶, D Hughes¹, P Kellman⁴, S Plein⁵, JC Moon¹, ¹University College London - London - United Kingdom of Great Britain & Northern Ireland, ²University of Sydney - Sydney - Australia, ³Barts Health NHS Trust - London - United Kingdom of Great Britain & Northern Ireland, ⁴National Institutes of Health - Bethesda - United States of America, ⁵University of Leeds - Leeds - United Kingdom of Great Britain & Northern Ireland, ⁶Royal Free Hospital - London - United Kingdom of Great Britain & Northern Ireland,

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Background

Fabry disease (FD) is an X-linked lysosomal storage disease resulting in tissue sphingolipid accumulation. Although storage is important, other processes occur and can be measured including hypertrophy, inflammation and fibrosis. These can be quantified by multi-parametric CMR including cine imaging, T1 and T2 mapping and late gadolinium enhancement (LGE).

Purpose

Recent developments in CMR perfusion mapping allow rapid in-line perfusion quantification permitting the assessment of microvascular dysfunction. We hypothesized that there is microvascular dysfunction in FD which is associated with storage, fibrosis and oedema.

Methods

A prospective, observational study of 44 FD patients (49 years, 43% male, 24 (55%) with left ventricular hypertrophy (LVH)) and 27 healthy controls. Multi-parametric CMR included vasodilator stress perfusion mapping. Myocardial blood flow (MBF) was measured and its associations with other processes investigated.

Results

Compared to LVH- FD, LVH+FD had a higher LV ejection fraction (73% vs 67%, p=0.04), lower T1 (937 vs 985ms, p=0.007), more LGE (85% vs 15%, p<0.001) and a lower stress MBF (1.76 vs 2.36 ml/g/min, p<0.001). The reduction in stress MBF was more pronounce in the subendocardium than subepicardium. LVH-FD had lower stress MBF than controls (2.36 vs 3.00 ml/g/min, p=0.001). Across all FD, LGE and low native T1 were independently associated with reduced stress MBF. On a per-segment basis segment basis, stress MBF was independently associated with increasing wall thickness, decreasing native T1, increasing T2, increasing ECV and LGE.

Conclusions

FD patients have reduced perfusion, particularly in the subendocardium with greater reductions with LVH,
storage, oedema and scar. Perfusion is reduced even without LVH suggesting it is an early disease marker.

Figure 1. Multiparametric CMR in Fabry disease and healthy controls. Left to right – Cines (diastole), native T1 maps, T2 maps, stress myocardial blood flow (MBF) maps, late gadolinium enhancement (LGE). (A): Healthy control (all normal) . (B) Fabry disease, no LVH, low T1, normal T2, no LGE. (C) FD patient with severe LVH, low T1(some pseudonormalization in LGE areas), high T2 in LGE areas, and extensive LGE. MBF falls with increasing disease severity, particularly in the endocardium.