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WORKSHOP PRESENTATION Open Access

Characterization of both myocardial extracellular volume expansion and myocyte mypertrophy by CMR detect early signs of myocardial tissue remodeling in Friedreich's ataxia patients without heart failure.

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Background

Heart Failure (HF) is the most common cause of death in Friedreich's ataxia (FRDA), a mitochondrial disease characterized by neurodegeneration, hypertrophic cardiomyopathy, caused by homozygous GAA expansions in the *FXN* gene. Recent report demonstrates that specific-gene therapy may prevent and reverse the cardiomyopathy in a mice model of FRDA. Myocardial interstitial fibrosis is a hallmark of FRDA's cardiomyopathy and a potential substrate for arrhythmias and HF. Myocardial tissue characterization by cardiac magnetic resonance (CMR) allows access to tissue-based phenotypes that may better describe LV remodeling in FRDA's cardiomyopathy.

Methods

The aim of this study was to perform direct quantification of myocardial extracellular volume fraction (ECV) and intracellular lifetime of water (τ_{ic}), a measure of cardiomyocyte hypertrophy, using T1-weighted CMR imaging in cohort of patients with FRDA without HF.

We investigated 27 FRDA patients without HF (mean age 26.8 ± 9, 12 female) and in 30 healthy volunteers as control subjects (mean age 49 ± 15) using a 3T CMR system. The T1 quantification by Look-Locker gradient-echo before and after contrast applying a 2-site model for transcytolemmal water Exchange was used for ECV and τ_{ic} quantification. Cine CMR and LGE imaging in matching locations were also performed.

Results

FRDA patients revealed normal LVEF with increased LV Mass-index compared with health controls (for LVEF 67.3% ± 11.5 vs. 62.5% ± 6.8, P = NS; for LVMASSi 62.7 ± 23 vs. 45.1 ± 6.8 g/m^2, $p < 0.05$). In 4 out 27 FRDA patients a non-ischemic LGE pattern was present. Both ECV and intracellular lifetime of water (τ_{ic}) were significantly higher FRDA patients (ECV: 0.36 ± 0.04 vs. 0.28 ± 0.03, $p < 0.0001$; τ_{ic}: 0.12 ± 0.08 vs. 0.08 ± 0.03, $p < 0.005$).

Conclusions

ECV and intracellular lifetime of water (τ_{ic}) determined by T1 measurements characterized early signs of myocardial tissue remodeling in FRDA with normal LVEF. Early changes in tissue-phenotypes are detectable by novel-CMR methods in FRDA patients, and may be useful to track effects of new genetic therapies for FRDA cardiomyopathy.

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