

**Open Access** 

### WORKSHOP PRESENTATION

# 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.

Otavio R Coelho-Filho<sup>1\*</sup>, Ravi V Shah<sup>2</sup>, Thiago D Venancio<sup>1</sup>, Alberto R Martinez<sup>1</sup>, Tomas G Neilan<sup>2</sup>, Irene Righetti<sup>1</sup>, Cynthia B da Silva<sup>1</sup>, Ingrid Faber<sup>1</sup>, Iscia Lopes-Cendes<sup>1</sup>, Marcondes França Jr<sup>1</sup>, Michael Jerosch-Herold<sup>2</sup>

*From* 19th Annual SCMR Scientific Sessions Los Angeles, CA, USA. 27-30 January 2016

#### 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 ( $\tau_{ic}$ ), 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  $\pm$  9, 12 female) and in 30 healthy volunteers as control subjects (mean age 49  $\pm$  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  $\tau_{\rm ic}$ 

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<sup>2</sup>, p < 0.05). In 4 out 27 FRDA patients a non-ischemic LGE pattern was present. Both ECV and intracellular lifetime of water ( $\tau_{ic}$ ) were significantly higher FRDA patients (ECV: 0.36 ± 0.04 vs. 0.28 ± 0.03, p < 0.0001;  $\tau_{ic}$ : 0.12 ± 0.08 vs. 0.08 ± 0.03, p < 0.005).

#### Conclusions

ECV and intracellular lifetime of water ( $\tau_{ic}$ ) 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.

#### Authors' details

<sup>1</sup>Medicine, State University of Campinas - UNICAMP, Campinas, Brazil. <sup>2</sup>Harvard Medical School, Boston, MA, USA.

Published: 27 January 2016

<sup>&</sup>lt;sup>1</sup>Medicine, State University of Campinas - UNICAMP, Campinas, Brazil Full list of author information is available at the end of the article



© 2016 Coelho-Filho et al. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http://creativecommons.org/licenses/by/4.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/ publicdomain/zero/1.0/) applies to the data made available in this article, unless otherwise stated.

doi:10.1186/1532-429X-18-S1-W7 **Cite this article as:** Coelho-Filho *et al.*: 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.. *Journal of Cardiovascular Magnetic Resonance* 2016 18(Suppl 1):W7.

## Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar

**BioMed** Central

• Research which is freely available for redistribution

Submit your manuscript at www.biomedcentral.com/submit