Journal of Cardiovascular Magnetic Resonance

BioMed Central

Meeting abstract

Open Access

2069 Assessment of genetic dilated cardiomyopathy in LMNA-mutation carriers by cardiac MRI

Kristel M Antila*[1], Juha R Koikkalainen[2], Jyrki MP Lötjönen[2], Tiina Heliö[3], Sari M Kivistö[4] and Kirsi Lauerma[4]

Address: [1]Helsinki University, Helsinki, Finland, [2]VTT Technical Research Centre of Finland, Tampere, Finland, [3]Department of Cardiology, Helsinki University Central Hospital, Helsinki, Finland and [4]Department of Radiology, Helsinki Medical Imiging Center, Helsinki, Finland

* Corresponding author

from 11th Annual SCMR Scientific Sessions
Los Angeles, CA, USA. 1–3 February 2008

Published: 22 October 2008

Journal of Cardiovascular Magnetic Resonance 2008, **10**(Suppl 1):A338 doi:10.1186/1532-429X-10-S1-A338

This abstract is available from: http://jcmr-online.com/content/10/S1/A338

Introduction

DCM is characterized by enlargement and impaired contraction of left or both ventricles. With an estimated prevalence of 36/100 000 in adults in the United States, DCM is a significant cause of morbidity and mortality. One third to one half of the causes are familial and the most important risk gene for cardiomyopathy is lamin A/C gene. Thus, we wanted to look for other methods than conventional echocardiography, to recognize this type of serious cardiomyopathy early. We wanted to characterize more precisely the cardiac findings of the Finnish *LMNA* -patients using magnetic resonance imiging (MRI)

Purpose

To characterize the cardiac functional parameters and myocardial late enhancement pattern in LMNA-carriers.

Methods

12 LMNA-carriers (representing 9 families) who were heterozygotes for one lamin A/C mutation (Ser143Pro, T1085deletion, Ala132Pro, Arg190Tpr) that may cause dilated cardiomyopathy and 14 healthy controls were studied with a 1.5 T MR imager. The volumes, wall thickness and motion of both ventricles were assessed from cine images. The LMNA-carriers were evaluated with late enhancement imaging.

Results

All carriers were in NYHA class I and had sinus rhythm. 6/12 carriers presented with first degree atrioventricular block. 5/12 LMNA-carriers were asymptomatic, palpitation being the most common symptom. The carriers had higher BSA corrected LVEDV (93 +/- 13 vs 80 +/- 20, p = 0.005), LVESV(36 +/- 7 vs 24 +/- 7, p = 0.003) and lower EF(61 +/- 6 vs 70 +/- 3, p = 0.0003). The carriers had decreased regional wall motion at basal inferior, anterior, and septal segments, and decreased thickening of apicoseptal segment. 7/12 carriers showed midwall late enhancement that represents myocardial fibrosis. Inferior segments were most commonly involved; fibrosis was also observed in anterior, septal and lateral walls.

Conclusion

Due to the severity of the disease and the possibility of early intervention (pharmacological, device and surgical therapy) to prevent or attenuate progression to cardiac heart failure and to avoid sudden death we wanted to characterize early familial dilated cardiomyopthy using MRI.

LMNA-mutations are the most common genetic defects in familial dilated cardiomyopathy. To our knowledge this is the largest LMNA-carrier group described with MRI in the world. Our conclusion is that MRI can be used for early diagnosis of cardiomyopathy in known gene carriers and for phenotyping different DCM subgroups. Left ventricu-

lar regional dysfunction is consistent with late enhancement in the basal segments.