Cardiac fibrosis and function in primary carnitine deficiency patients evaluated by cardiac magnetic resonance

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Topic(s):
Late Gadolinium Enhancement and Viability

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BACKGROUND
Untreated primary carnitine deficiency (PCD) can lead to sudden cardiac death. It is unknown if patients currently treated with L-carnitine have suffered myocardial damage because of prior long-term carnitine deficiency and thus might be at greater risk of potentially serious arrhythmia. Cardiac evaluation of function and fibrosis is best supported by cardiac magnetic resonance imaging (CMR).

METHODS
In total, 68 subjects were studied – 36 PCD patients treated with L-carnitine according to guidelines, 17 carriers and 17 healthy subjects. Combined cine stacks were used to evaluate left ventricle (LV) systolic and diastolic function and late gadolinium enhancement (LGE) stacks in LV to evaluate fibrosis.

RESULTS
LV end-diastolic (LVEDV), end-systolic (LVESV) and ejection fraction (LVEF) were not significantly different between groups, but LV mass (p=0.037) and LV peak wall thickness (p=0.038) was higher in PCD patients homozygous for the severe c.95A>G mutation (n=17), among whom there were two cases of unexpected mid-wall fibrosis. There were no findings of fibrosis in other PCD patients, carriers or healthy controls (p=0.10).

CONCLUSION
In some cases, PCD patients have myocardial fibrosis shown by LGE. Adherence to recommended L-carnitine supplementation is though preferable in order prevent adverse cardiac remodeling.
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