

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

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## Background

Untreated primary carnitine deficiency (PCD) can cause dilated cardiomyopathy, cardiomegaly and cardiac arrest which are all treated with oral carnitine supplements.

It is unknown whether myocardial disease is still present in a well-treated PCD population.

Cardiac evaluation of function and fibrosis is best investigated by cardiac magnetic resonance imaging (CMR).

## Method

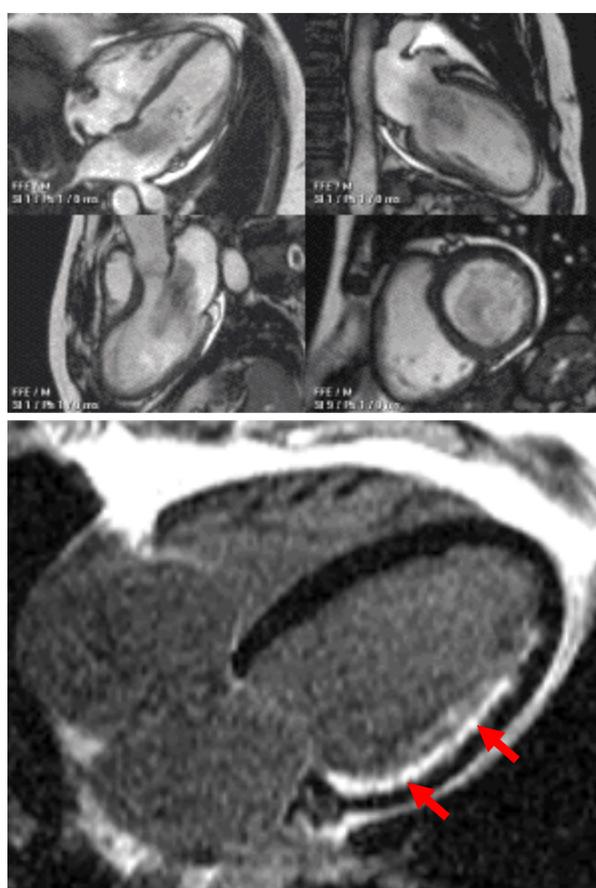
From a population of 28 identified homozygote PCD patients, 17 were included in the present study. Further, 17 haplozygote, 17 heterozygote PCD patients and 17 healthy subjects was age and gender matched and included in the present study.

A total of 68 participants were studied using CMR.

All patients had a cine stack to evaluate left ventricle (LV) systolic and diastolic function and late gadolinium enhancement stack in LV to evaluate myocardial fibrosis.

## Cardiac magnetic resonance imaging

Top: Functional cine imaging; bottom: Late enhancement sequences with fibrosis (arrow)



## Results

Left ventricular size and systolic function were not different between groups.

Left ventricle myocardial mass and left ventricle thickness was higher in homozygote PCD patients.

In homozygote PCD patients, there were two cases of unexplained fibrosis. There were no findings of fibrosis in any of the other patient-groups and neither in the 17 healthy controls ( $p=0.10$ ).

## Conclusion

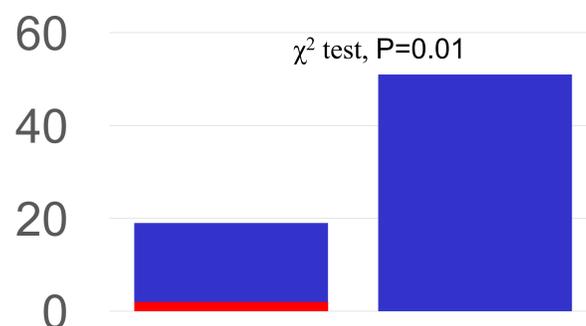
This is the first study to our knowledge, investigating cardiac function of patients suffering from PCD and carriers of PCD using CMR.

There were no differences in the cardiac function among the groups and all were within published normal ranges of cardiac parameters.

There were not significantly more PCD patients with fibrosis, however, since 2 patients suffering from PCD were found to have fibrosis and none of the other patients had fibrosis, there was a clear trend warranting further investigations.

## Fibrosis in the myocardium

Left column: Homozygote PCD patients Right column: All other patient groups; blue: no fibrosis, red: fibrosis



## Left ventricular function related to plasma carnitine levels

Blue: heterozygote, red: haplozygote, grey: homozygote

