mitoHEART

CMR-based diagnosis of cardiac involvement in patients with mitochondrial myopathy

project home: University hospital Münster

State of the art

Mitochondrial myopathy comprises various clinical subforms of neuromuscular disorders that are characterised by impaired mitochondrial energy metabolism due to dysfunction of the mitochondrial respiratory chain. This is a heterogeneous group of genetic disorders that can be caused by mutations in either mitochondrial DNA (mtDNA) or nuclear DNA. In this regard, after the encephalo-neuromuscular syndromes, cardiomyopathy is the most common pathological phenotype associated with mtDNA point mutations (with an estimated 20% associated with cardiac phenotypes, predominantly hypertrophic cardiomyopathy). Some small-sized studies mainly based on ECG recordings and/or echocardiography have revealed different forms of cardiomyopathy such as hypertrophic or dilated cardiomypathy and suggested incidences varying between ~15-40%. In particular, in one larger study, a significantly higher mortality was documented in patients with mitochondrial myopathy and proof of cardiomyopathy compared to those with mitochondrial disease not affecting the heart. Moreover, cardiac death was identified as the number one cause of mortality. Hence, early identification of those patients with mitochondrial disease who are prone to cardiac involvement is of clinical importance. However, early diagnosis of cardiomyopathy may be difficult using conventional non-invasive methods such as echocardiography as this technique primarily shows wall motion abnormalities which occur only at later stages of myocardial involvement. In contrast, cardiovascular magnetic resonance (CMR) promises to be a more accurate and comprehensive non-invasive tool for cardiac evaluation of patients with mitochondrial disease since this novel technique enables accurate assessment of functional and morphological parameters. Consequently, detection of potential structural abnormalities prior to functional impairment using CMR may enable a more timely implementation of appropriate treatment strategies in these patients. However, no comprehensive and targeted CMR studies have been performed so far in patients with mitochondrial disorders.

Targets

With the study "mitoHEART", we aim at 1) accurately characterising cardiac disease manifestation in patients with mitochondrial myopathy, 2) elucidating the in vivo cardiac damage pattern of patients with different subforms of mitochondrial disease and 3) assess therapeutic success in relation to the underlying cardiac disease by performing comprehensive CMR studies.

Project management and contact address

Ali Yilmaz, Univ-Prof. Dr. med.
University hospital Münster

Cardiology, Molecular Imaging and Cardiomyopathies / Department of cardiology I: Section for Cardiac Imaging
Albert-Schweitzer-Campus 1, 48149 Münster
Phone: +49 251 - 83 - 44948
Fax: +49 251 - 83 - 44907
Web: www.rbk.de/herzbildgebung

Project members

Dajana Küdde,
University hospital Münster

Cardiology, Molecular Imaging and Cardiomyopathies / Department of cardiology I: Section for Cardiac Imaging
Albert-Schweitzer-Campus 1, 48149 Münster
Phone: +49 251 - 83 - 44833

Fax: +49 83 - 8101 - 44907
E-mail: Dajana.Kuedde@ukmuenster.de
Web: www.rbk.de/herzbildgebung

Literature

Jose T, Gdynia HJ, Mahrholdt H, Vöhringer M, Klingel K, Kandolf R, Bornemann A, Yilmaz A, CMR gives clue to "ragged red fibers" in the heart in a patient with mitochondrial myopathy.
Int. J. Cardiol. 2011 May; 149(1):e24-7