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Early detection of cardiac involvement of desminopathy by cardiovascular magnetic resonance

Martyna Faber[1], Teresa Trenkwalder[2], Federica Montagnese[3], Heiko Stern[4], Christian Meierhofer[4]

[1]Department of Radiology, German Heart Center Munich, Technical Universitiy of Munich, Germany
[2]Department of Cardiology, German Heart Center Munich, Technical Universitiy of Munich, Germany
[3]Department of Myopathology, Friedrich-Baur-Institute, Department of Neurology, LMU Klinikum Munich, Germany
[4]Department of Congenital Heart Disease and Pediatric Cardiology, German Heart Center Munich, Technical Universitiy of Munich, Germany

A 42-year-old man presented with loss of muscle strength and aching in the calves. Skeletal muscle biopsy of anterior tibial muscle showed a myofibrillar myopathy with increased endo- and perimysial connective tissue, centralized myonuclei, and intracellular rimmed vacuoles (Fig. 1C, D). Molecular genetic analysis detected classic R350P-desmin gene mutation, causing an autosomal-dominant desminopathy.

Cardiac examinations were performed. Transthoracic echocardiogram was unremarkable with normal biventricular function, electrocardiogram revealed a slightly prolonged PQ-time but no pronounced arrhythmia.

Cardiovascular magnetic resonance showed no late-gadolinium-enhancement. Pre- and postcontrast T1-mapping was performed using modified look-locker-inversion-sequence (MOLLI, Myomaps, Siemens healthineers). T1-relaxation-times were located in the upper normal range with values between 940 and 1000 ms. Extracellular volume fraction (ECV) was calculated by using T1-map pre and post contrast. The patient's hematocrit was measured on the examination day. ECV-maps (Fig. 1A) and polarmaps (Fig. 1B) were generated to visualize ECV results in left ventricular myocardial segments. ECV ranged between 25.1% and 34.7% (normal up to 29%) in the different cardiac segments, with higher values in the apical myocardium. As these findings are compatible with the myopathological changes of increased deposit of connective tissue in the endo- and perimysium, these are suspicions for early stage of cardiac involvement in desminopathy. Cardiac magnetic resonance proved to be a sensitive diagnostic tool for early cardiac involvement in desminopathy, which have not yet been revealed by other non-invasive methods.

Desminopathies belong to a genetically heterogeneous group of disorders named myofibrillar myopathies, which might be caused by mutations in the desmin gene, but may also affect other adjacent genes.

Conflict of interest: None declared

Address for correspondence: Martyna Faber, MD, Department of Radiology, German Heart Center Munich, Technical Universitiy of Munich, Lazarettstrasse 36, 80636 Munich, Germany, e-mail: martyna.faber89@gmail.com

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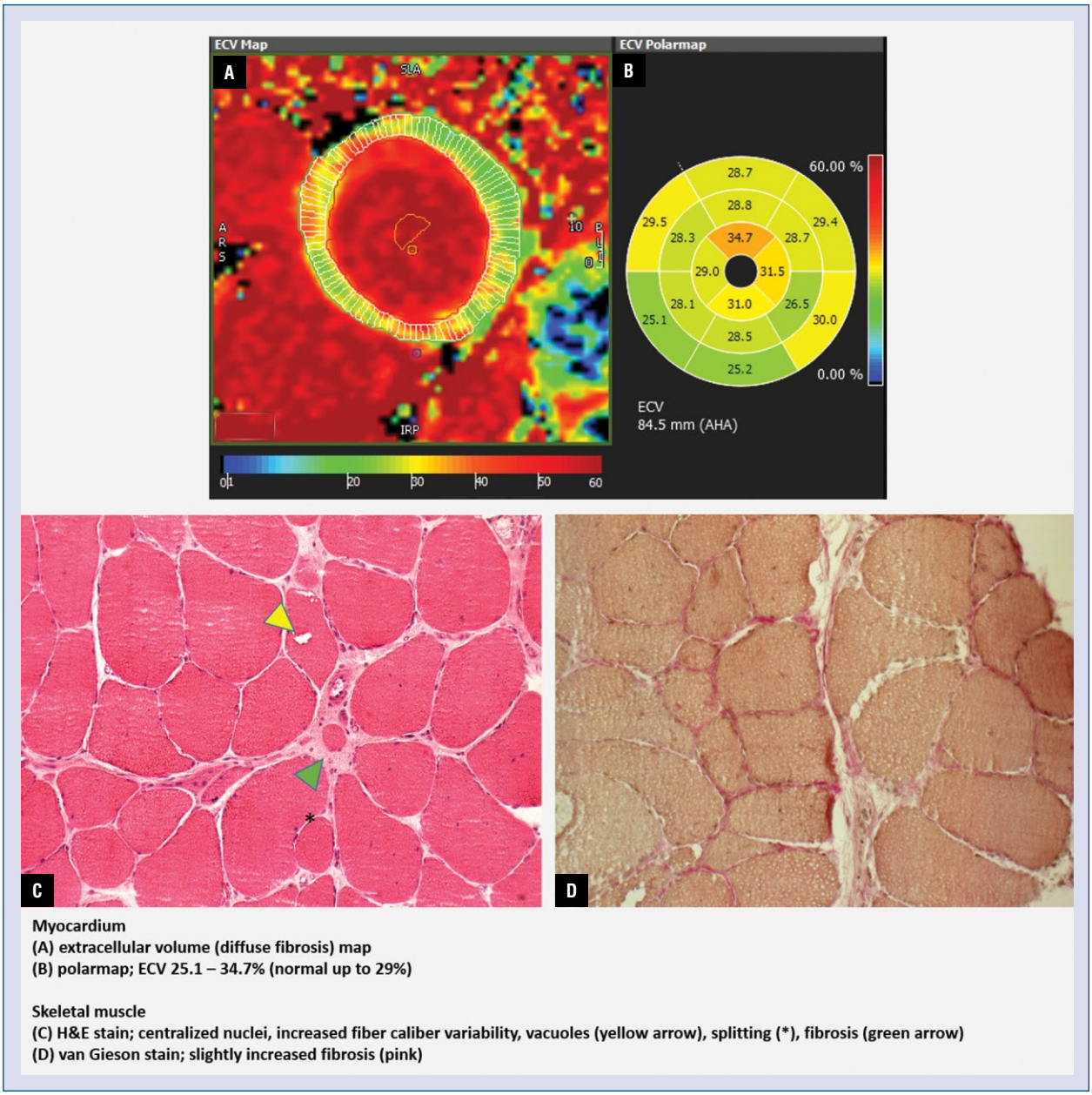

Figure 1. Cardiac and skeletal muscle involvement in autosomal-dominant desminopathy. Cardiac magnetic resonance imaging: Extracellular volume map (**A**); polarmap (**B**). Skeletal muscle biopsy: hematoxylin and eosin stain (**C**) and van Gieson stain (**D**).