

## Myocardial work analysis in screening of familial dilated cardiomyopathy

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**Introduction:** Early detection of affected family relatives of patients with dilated cardiomyopathy (DCM) is essential in order to guide follow up, outcomes and initiate early treatment. Myocardial work analysis is a novel method which integrated strain imaging and blood pressure and has the potential to identify patients with subclinical disease.

**Purpose:** We analysed myocardial work in family relatives of DCM patients with positive genotype but negative phenotype in order to identify whether myocardial work can identify early changes.

**Methods:** Seventy-four family relatives of DCM patients attending for screening were examined. All individuals were asymptomatic with either positive (45/74, G+) or negative (29/74, G-) genotype and no echocardiographic evidence of left ventricular dilatation or systolic impairment. Non-invasive myocardial work analysis using two-dimensional (2D) speckle tracking echocardiography was analysed. Global longitudinal strain (GLS) was measured by the same vendor specific software used for myocardial work analysis. Left ventricular (LV) ejection fraction (EF) was measured with the Simpson's biplane method. The peak systolic arm cuff blood pressure (BP) measurement at the time of echocardiography was used for the myocardial work study.

**Results:** In total we included 74 individuals (37±15 years old, 50.7% women) with mean systolic and diastolic BP of 121.3±14 and 73.2±10 mmHg respectively, mean EF was 58±5% and mean GLS at 18.4±2.5%.

G+ individuals had pathogenic and very likely pathogenic mutations in 8 different genes (TTN, BAG3, DSP, FLNC, LMNA, DMD, RBM20, TPM1). There was no difference in age, systemic hypertension, diabetes or medical treatment between the 2 groups.

No significant difference was found among G+ and G- individuals in mean systolic and diastolic BP (121.2±14.7 vs 121.2±15.2 mmHg), mean EF (57.3±5 vs 59.1±4%), GLS (-18.2±1.5 vs -18.6±2.9%), mean global work index (1818±403 vs 1928±295 mmHg%) and global constructive work (2192±464 vs 2260±318 mmHg%).

However, we found significant reduction of the global work efficiency (GWE) with a GWE of 94.4±2.7% in the G+ versus 95.9±1.6% in the G- individuals (p 0.02). Moreover, the global wasted work (GWW) was increased in the G+ with a GWW of 111±58 mmHg% versus 82±41 mmHg% in the G- individuals (p 0.03).

**Conclusion:** DCM gene carriers show, early on, decreased myocardial work efficiency and increased wasted work compared to unaffected family members, which appears to be earlier than other parameters such as EF and GLS. Myocardial work analysis could potentially recognize individuals showing early cardiac involvement and guide closer follow up and early initiation of treatment.