

Severity of myocardial dysfunction in Familial Mediterranean Fever, can it discriminate homozygous from heterozygous cases?

*Dawood O. (1), AbdelMassih A.F. (1,3), El-Hamshary A. (2), El-Said E. (2), Hassan R. (2)
Pediatric, Cardiology Unit, Pediatrics Department, Specialized Children Hospital, Faculty of Medicine,
Cairo University, Cairo, Egypt (1);
Pediatric, Rheumatology Unit, Pediatrics Department, Specialized Children Hospital, Faculty of
Medicine, Cairo University, Cairo, Egypt (2);
Children's Cancer Hospital, Egypt 57357, Cairo, Egypt (3)*

Introduction: Familial Mediterranean fever is the most common auto-inflammatory disorder. Myocardial dysfunction in the context of FMF might be the result of several factors: persistent inflammation, amyloid deposition, medications involved in control of disease activity.

Methods: 40 patients (Group 1: G1) with FMF and 40 healthy controls (Group 2: G1) have been examined with Tissue Doppler Imaging and Speckle Tracking Echocardiography for examination of Left ventricular Systo-diastolic functions, demographic, clinical and genetic data of cases have been recorded as well. Patients have been furtherly subdivided into Group 1A (G1A) with homozygous mutation and Group 1B (G1B) with heterozygous mutation.

Results: Left ventricular diastolic dysfunction have been depicted in cases with FMF, this dysfunction was more profound in cases with homozygous mutation as evidenced by higher LV E/E' ratio in cases with homozygous than heterozygous FMF mutation (G1A: 15 ± 3 vs. G1B: 6.57 ± 1.2 , $P<0.001$). Moreover subtle systolic LV dysfunction has been found in cases with FMF, LV systolic function was even lower in cases with homozygous mutation as evidenced by a lower LV GLS (global Longitudinal Strain) in such cases (G1A: -11.2 ± 2.2 vs. G1B: -20.2 ± 4.2 , $P<0.001$). LV GLS proved even to be highly predictive for homozygosity in cases with FMF with sensitivity of 94%.

Conclusion: The aforementioned findings point towards the occurrence of systo-diastolic dysfunction early in the course of FMF, cases with homozygous mutation are more at risk, this might be related to more severe inflammation and subsequent amyloid degeneration.