

## **Abstract**

Echocardiography plays a key role in the evaluation of functional and structural changes of the heart. Fabry disease (FD) is a genetic (X-linked) lysosomal storage disease caused by mutations in the gene for alpha-galactosidase A. Cardiovascular involvement is a major cause of morbidity and mortality in FD. The aim of our work was to refine the diagnosis and risk stratification of Fabry cardiac involvement using echocardiography.

In the first part of our retrospective project, we analysed the relationship between the uric acid level, development, and progression of left ventricular (LV) hypertrophy and adverse events. Our results showed a strong relationship between uric acid level and progression of LV hypertrophy, mortality, and cardiovascular events. Uric acid can be used as a marker of cardiovascular risk in FD.

In the second part of our retrospective project, we analysed the diagnostic value of tissue doppler imaging (TDI) of mitral annular velocities in Fabry cardiomyopathy. Despite the lower TDI mitral annular velocities in FD, the overlap with the general population is too large to allow reliable diagnosis of FD in routine clinical practice.

In the prospective study, we analysed the prevalence, characteristics, prognosis and validity of the current echocardiographic criteria for heart failure (HF) in FD. The study found a high prevalence of symptomatic HF in FD. Heart failure with preserved ejection fraction caused by LV concentric hypertrophy was the dominant phenotype. Left ventricular mass index,  $E/e'$  and global longitudinal strain yielded the highest diagnostic utility for HF diagnosis and were significantly correlated with levels of natriuretic peptides and prognosis. Echocardiographic criteria proposed by current HF guidelines apply to Fabry patients and predict cardiovascular events.