To the Editor,

We would like to thank authors for their interest in our paper (1) and their valuable comment. In the authors’ letter to the editor, they mention potential concerns about the diagnosis of Fabry disease in the female patients.

The main aim of our study (1) was to screen adult patients with hypertrophic cardiomyopathy without left ventricular outflow tract. In the diagnosis of Fabry disease; Plasma α-galactosidase A activity measurement, determination of glycosphingolipids in plasma and urine and molecular genetic analysis methods of GLA gene are used (2). Fabry disease is an X-linked disorder. According to the Lyon hypothesis, in case of X chromosome inactivation, women can be affected as much as men (3). Plasma α-galactosidase A activity can be determined close to normal values in heterozygous. Definitive diagnosis should be made by molecular genetic analysis in female patients. Only measuring plasma α-galactosidase A activity in female patients may lead to a missed diagnosis. Therefore, as stated in our study, GLA gene sequence analyses were performed in all patients.

Cardiac symptoms and signs may be delayed in female patients with Fabry diagnosis compared to male patients. In a study by Linhart et al., pacemaker implantation, myocardial infarction, Left ventricular hypertrophy, and conduction defects were seen 8 to 10 years later in female patients diagnosed with Fabry disease (4). According to FOS (Fabry Outcome Survey) study data, the mean time between onset of symptoms and diagnosis was 13,7 and 16,3 years for males and females, respectively (5). Left ventricular hypertrophy, considered to be high-risk for Fabry disease, was observed in 46% of males and 28% of females in FOS. The age at onset of LVH in males and females was 38,0 and 55,4 years, respectively (5). According to the FOS database, female patients are usually (often severely) affected. Female patients with chronic kidney disease and/or LVH and/or stroke, considered to be high-risk for Fabry disease, should be performed by molecular genetic analysis. However, screening for FD in our country remains suboptimal. In the future, a countrywide approach to identify FD among patients with high-risk for Fabry disease seems warranted.

References