



Are Female Patients More Overlooked in the Diagnosis of Fabry Disease?

Muhammet Gürdoğan, Ömer Ferudun Akkuş

Department of Cardiology, Trakya University School of Medicine, Edirne, Turkey

We have read the article by Barman et al. titled as “The Prevalence of Fabry Disease Among Turkish Patients with Non-Obstructive Hypertrophic Cardiomyopathy: Insights from a Screening Study” with interest. (1) In their descriptive study, the researchers found that the frequency of Fabry disease (FD) was 2.5% by screening 80 patients (53 males) diagnosed with hypertrophic cardiomyopathy without left ventricular outflow tract obstruction. The authors emphasized that FD should be kept in mind in the differential diagnosis of patients with unexplained left ventricular hypertrophy (LVH). (1)

FD is an X-linked recessive lysosomal storage disorder, first described in 1898, characterized by a deficiency of α -galactosidase A resulted from a genetic mutation. In FD, glycosphingolipid accumulation occurs in many tissues, especially in the nervous system, heart, kidney and skin. (2,3) As stated by Barman et al., different results were found in the scanning performed in different parts of the world to investigate the incidence of FD. (1) The actual incidence of FD is not exactly known, but it is estimated to be much higher than seen in daily cardiology practice. It is known that males are affected more frequently and more seriously because of X-linked inheritance in FD. However, it has been reported that heterozygous women are also affected by the disease, but the disease symptoms such as LVH appear later in life (about 10 years later). (3,5) Myocardial fibrosis, which is a guide in determining the prognosis of FD and the patients who will benefit from enzyme replacement therapy, is reported to be seen in women without LVH, unlike men. (3,4) It is reported in the literature that the variant of FD characterized only by cardiac involvement is more common in

women. FD screening based solely on LVH means that the disease is detected less frequently in females. The fact that LVH has been reported in only one out of every three women diagnosed with FD in the literature supports this idea. (3-5)

The study by Barman et al., is valuable for being the first study investigating the incidence of FD in patients with unexplained LVH in Turkey. However, under-representation of female gender in the study group (two out of three patients are males) may cause the FD frequency to be detected lower than it actually is. In the future, large-scale studies that minimize the effect of gender difference will help to determine the true prevalence of FD.

Keywords: Diagnosis, gender differences, fabry disease

References

1. Barman HA, İkitimur B, Kılıçkiran Avcı B, Durmaz E, Atıcı A, Aslan S, et al. The Prevalence of Fabry Disease Among Turkish Patients with Non-Obstructive Hypertrophic Cardiomyopathy: Insights from a Screening Study. *Balkan Med J* 2019;36:354-8
2. Hsu TR, Niu DM. Fabry disease: Review and experience during newborn screening. *Trends Cardiovasc Med* 2018;28:274-81.
3. Yousef Z, Elliott PM, Cecchi F, Escoubet B, Linhart A, Monserrat L, et al. Left ventricular hypertrophy in Fabry disease: a practical approach to diagnosis. *Eur Heart J* 2013;34:802-8.
4. Biegstraaten M, Arngrímsson R, Barbey F, Boks L, Cecchi F, Deegan PB, et al. Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. *Orphanet Journal of Rare Diseases* 2015;10:36.
5. Hagège A, Réant P, Habib G, Damy T, Barone-Rochette G, Soulat G, et al. Fabry disease in cardiology practice: Literature review and expert point of view. *Arch Cardiovasc Dis* 2019;112:278-87.

Address for Correspondence: Muhammet Gürdoğan, Department of Cardiology, Trakya University School of Medicine, Edirne, Turkey

Phone: +90 284 235 10 41 e-mail: drmgurdogan@gmail.com

Received: 28 November 2019 Accepted: 10 December 2019 • DOI: 10.4274/balkanmedj.galenos.2019.2019.11.148

Available at www.balkanmedicaljournal.org

Cite this article as:

Gürdoğan M, Akkuş ÖF. Are Female Patients More Overlooked in the Diagnosis of Fabry Disease? *Balkan Med J*.

©Copyright 2020 by Trakya University Faculty of Medicine / The Balkan Medical Journal published by Galenos Publishing House.