

## Prevalence of Left Ventricular Hypertrophy in Patients with Fabry's Disease at the Time of Diagnosis

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Fabry disease (FD) is a lysosomal storage disease linked to the X chromosome caused by mutations in the gene that encodes the enzyme  $\alpha$ -Galactosidase A ( $\alpha$ GAL A) resulting in reduced or absent activity. This produces the pathological accumulation of glycosphingolipids (mainly globotriaosylceramide -GL3- and its deacylated form globotriaosylsphingosine -LisoGL3-) in a wide variety of cells throughout the body, and the most relevant in the clinical picture of this disease are organs such as the heart skin, kidneys, blood vessels, peripheral nerves and central nervous system [1,5].

At cardiac level, one of the main manifestations is Left Ventricular Hypertrophy [2,3] (LVH), but the same is evident years after the beginning of the symptoms in the classical forms or even almost the only manifestation in the late forms (Late-onset, cardiac variant) [1].

### Objective

To assess the presence of LVH in a series of patients from Argentina at moment of the diagnosis of FD with clinical manifestations and genetic variants associated with the classic form.

### Materials and Methods

We included 12 patients, 4 (33%) men with age  $25 \pm 16$  yo and 8 (66%) women, age  $33.9 \pm 13.8$  yo, without arterial hypertension or Valvular Disease, with a diagnosis of FD and before starting specific treatment: Enzyme Replacement Therapy (ERT). It was considered normal a mass of up to  $88 \text{ g/m}^2$  in women and  $102 \text{ g/m}^2$  in men [4].

The images were obtained with a GE Vivid 7 ultrasound, and the subsequent analysis with software GE Echopac 12© General Electric, taking measurements in 2 D mode on the Parasternal long axis view. Calculation of the Left Ventricle (LV) mass using the Devereux formula ( $0.8 \times 1.04 (((\text{LVEDD} + \text{IVSd} + \text{PWd})^3 - \text{LVEDD}^3))) + 0.6\text{g}$ ) according to ASE and EAC4 criteria.

For the analysis of data, the Chi square test ( $\chi^2$ ) and significance  $p < 0.05$  were used.

### Results

The LV mass in men was  $100.25 \pm 34.3 \text{ g/m}^2$  and in women  $85 \pm 28.2 \text{ g/m}^2$ . The 50% (2 cases) of men presented ventricular hypertrophy, at an average age of  $38.5 \pm 7.8$  years, while the remaining 50% that did not present HVI, had an age of  $12 \pm 5.7$  years. According to these data, the age over 30 years was a significant data for the development of LVH ( $p < 0.05$ ). In women, 37.5% (3) had LVH with an average age of  $44.3 \pm 4.2$  years, for which an age of 40 years or more had a high probability of LVH (75%).  $p = 0.028$ .

### Conclusions

The presence of LVH in the classic forms of Fabry disease is related to the age, occurring in men mainly from the age of 30 and in women from 40 years.

### Bibliography

1. Politei J., *et al.* "Recomendaciones para el diagnóstico, tratamiento y seguimiento de la enfermedad de Fabry en Argentina". *Revista Nefrología Argentina* 16.2 (2018).
2. Kampmann C., *et al.* "Onset and progression of the Anderson-Fabry disease related cardiomyopathy". *International Journal of Cardiology* 130.3 (2008): 367-373.
3. Perry Elliott., *et al.* "Classification of the cardiomyopathies: a position statement from the European society of cardiology working group on myocardial and pericardial diseases". *European Heart Journal* 29.2 (2008): 270-276.
4. Lang RM., *et al.* "Recommendations for Cardiac Chamber Quantification by Echocardiography in Adults: An Update from the American Society of Echocardiography and the European Association of Cardiovascular Imaging". *Journal of the American Society of Echocardiography* 28.1 (2015): 1-39.
5. Alberto Ortiz., *et al.* "Fabry disease revisited: Management and treatment recommendations for adult patients". *Molecular Genetics and Metabolism* 123.4 (2018): 416-427.

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