Archivos de Cardiología de México

PERMANYER

Check for updates

IMAGES IN CARDIOLOGY

Pompe disease: a lesson to learn

Enfermedad de Pompe: una lección por aprender

Claudia A. Pavón-Flores, Leonardo Rivera-Rodríguez, and Juan E. Calderón-Colmenero*

Departamento de Cardiología Pediátrica, Instituto Nacional de Cardiología "Ignacio Chávez", Ciudad de México, México

Pompe disease (PD), was first described by Johannes Pompe in 1932. It is also known as acid α-glucosidase (GAA) enzyme deficiency, a severe progressive myopathy with autosomal recessive inheritance. GAA deficiency produces excessive glycogen storage in lysosomes, with secondary cell dysfunction, and progressive damage to cardiac, respiratory, skeletal, and smooth muscles. The incidence is estimated to be 1:40,000-1:300,000[1,2].

The most severe form begins in the 1st months of life. It includes massive cardiomegaly, muscle hypotonia, mild hepatomegaly, and delayed motor milestone. Without treatment, these patients do not survive beyond the 1st year of life[3].

The enzyme replacement therapy with recombinant human acid alpha-glucosidase is the specific treatment and is available at all ages[4].

We present the case of a 5-month-old male, with positive family history for hypertrophic cardiomyopathy. The patient was diagnosed with hypertrophic cardiomyopathy at 3 months of life. The clinical presentation was characterized for dyspnea on exertion, diaphoresis, and heart failure.

The echocardiogram showed concentric biventricular hypertrophic without and decreased left ventricular function. Late intramyocardial low-septal, middle, and apex enhancement were detected in the magnetic resonance (Figs. 1 and 2). The Genetic screening reported Gene GAA variant c.1987del (p.Gln663serfs*33) compatible with PD. Fifteen days after admission, he died of ventricular fibrillation.

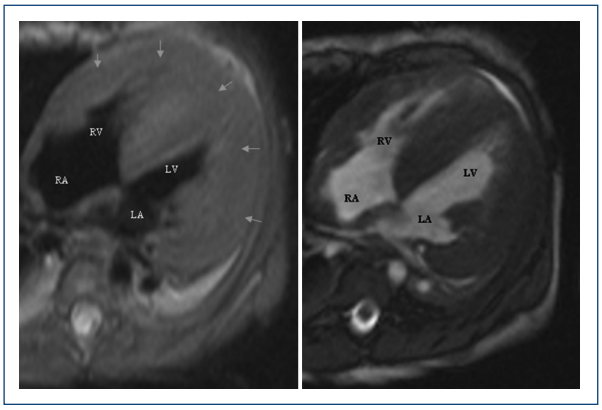

Figure 1. Magnetic resonance in four chambers shows biventricular hypertrophic cardiomyopathy (arrows). RA: right atrium; RV: right ventricle, LA: left atrium; LV: left ventricle.

***Correspondence:**
Leonardo Rivera-Rodríguez
E-mail: rivleonard@gmail.com

Date of reception: 23-06-2023
Date of acceptance: 03-04-2024
DOI: 10.24875/ACM.23000139

Available online: 31-10-2024
Arch Cardiol Mex. 2024;94(4):529-530
www.archivoscardiologia.com

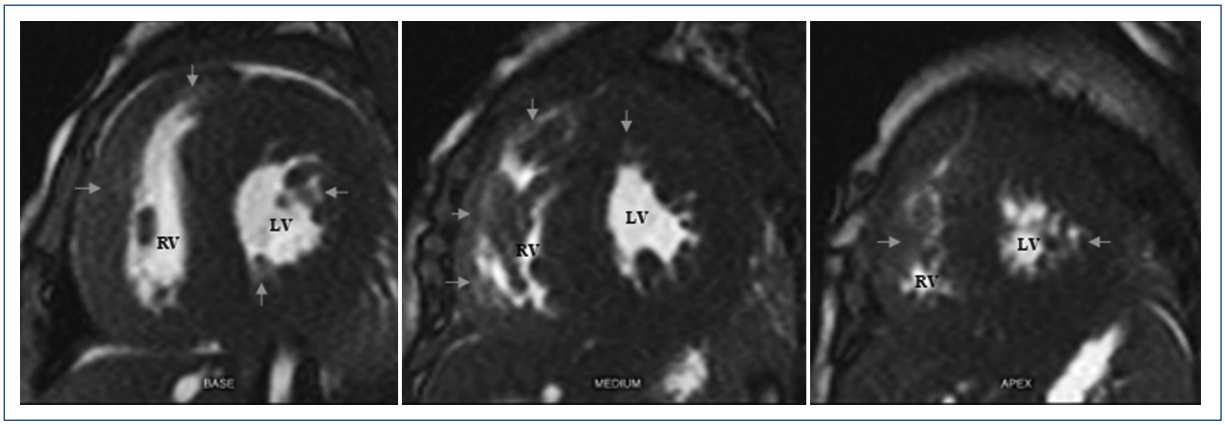

Figure 2. Magnetic resonance images in short-axis plane, present cardiomyopathy with late low, middle, and apex intramyocardial enhancement (arrows). RV: right ventricle; LV: left ventricle.

Performing timely detection is vital for patient survival, with the current support of imaging studies and genetic tests, the diagnostic is achieved. We suggest monitoring patients and performing interventions to avoid complications. Treatment is available in Mexico, so prompt action must be taken.

Funding

None.

Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that no patient data appear in this article.

Right to privacy and informed consent. The authors declare that no patient data appear in this article.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

References

1. Almasri M, Sanders E, Zakaria D. An usual cause of infantile failure to thrive. J Am Coll Cardiol. 2022;79:2633.
2. Zhong Y, Zhang W, Fan W. Hypertrophy cardiomyopathy: a case of infantile-onset Pompe disease in cardiac magnetic resonance findings. Heliyon. 2023;9:e18330.
3. Lim JA, Li L, Raben N. Pompe disease: from pathophysiology to therapy and back again. Front Aging Neurosci. 2014;6:177.
4. Bay LB, Denzler I, Durand C, Eiroa H, Frabasil J, Fainboim A, et al. Childhood Pompe disease: diagnosis and treatment. Arch Argent Pediatr. 2019;117:271-8.