










Clinical challenges in diagnosing and follow up of Anderson-Fabry disease patients - Is it indeed so rare or are we not recognizing on time?

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KEYWORDS: Anderson-Fabry disease, left ventricular hypertrophy, cardiomyopathy.

CITATION: *Cardiol Croat.* 2024;19(11-12):450. | <https://doi.org/10.15836/ccar2024.450>

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Introduction: Anderson-Fabry disease (AFD) is a rare X-linked lysosomal storage disorder caused by mutations in galactosidase A gene encoding for the enzyme alpha-galactosidase A, resulting in the progressive accumulation of glycosphingolipids in various tissues. The heterogeneous nature of symptoms, along with its rarity, poses several challenges in diagnosing and managing AFD. Cardiac involvement frequently occurs in AFD patients, manifesting as left ventricular hypertrophy, conduction system impairment, and valvular abnormalities.¹⁻³ **Aim:** To raise awareness and increase the role of cardiologists in the early detection of Anderson-Fabry disease, to initiate early treatment and prevent the progression of this, still underdiagnosed, disease.

Patients and Methods: Two patients from the same family have been diagnosed with AFD, while the rest of the family is still undergoing evaluation. After extensive evaluation, AFD was confirmed in multiple organ systems in both patients, with predominant involvement of the heart. The diagnostic workup of AFD should be based on a stepwise approach, including extracardiac and cardiac "red flags", to recognize AFD as early as possible. AFD-associated cardiomyopathy can be potentially reversible or stabilized after a specific treatment. Therefore, early and timely detection of cardiac "red flags" is important. Furthermore, it is important to highlight the collaboration with leading centres that have more experience in creating a team that deals with the treatment and monitoring of such patients. Through our single-centre experience, we have implemented a structured approach to improving the diagnosis and follow-up of AFD patients, an approach that includes medical staff education, systematic screening, multidisciplinary team approach with personalized treatment and follow-up plans.

Conclusion: Raising awareness of this rare disease is important because, due to its non-specific symptoms, it continues to go underdiagnosed. Early detection can lead to earlier treatment, improvement of patients' quality of life and prevention of fatal complications.

RECEIVED:
October 13, 2024

ACCEPTED:
October 31, 2024



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