

Doença de Fabry, o Papel do Electrocardiograma *Fabry Disease, the Role of the Electrocardiogram*

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Palavras-chave: Doença de Fabry/diagnóstico; Electrocardiografia; Hipertrofia Ventricular Esquerda.

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A 46-year-old man, with a smoking history and anemia, presented with pain in the right iliac fossa and fever. The diagnosis of acute appendicitis was made. The preoperative electrocardiogram (EKG) is showed in Fig. 1. After a detailed clinical history, we concluded that he had been suffering from neuropathic pain since childhood. Previous analytical studies showed an estimated creatinine clearance of 50 mL/min/1.73 m².

From the family history we highlight is mother who suffered an extensive ischemic stroke at the age of 65, since then totally dependent (reviewed process her ECG revealed criteria

for left ventricular hypertrophy (LVH) and short P-R interval).

The study progressed with an echocardiogram which confirmed moderate concentric LVH, without any other relevant abnormalities. Serum α -galactosidase activity and genetic study confirmed the suspicion of Fabry disease.

He was the first patient to undergo enzyme replacement therapy in our hospital. The patient's daughter was evaluated in a genetics consultation, confirmed to be positive for the mutation, with no manifestations of the disease to date.

Fabry disease is characterized by errors in glycosphingolipid metabolism due to absence or deficiency of the enzyme α -galactosidase A activity. It is an X-linked genetic disorder.

Hemizygotic patients are more significantly affected, showing characteristic signs and symptoms from infancy.¹⁻⁴ However, the diagnosis is often delayed by several years. Clinicians must keep a high clinical awareness for red flags in the EKG as in this case - short P-R interval associated with LVH criteria and concentric ventricular hypertrophy on

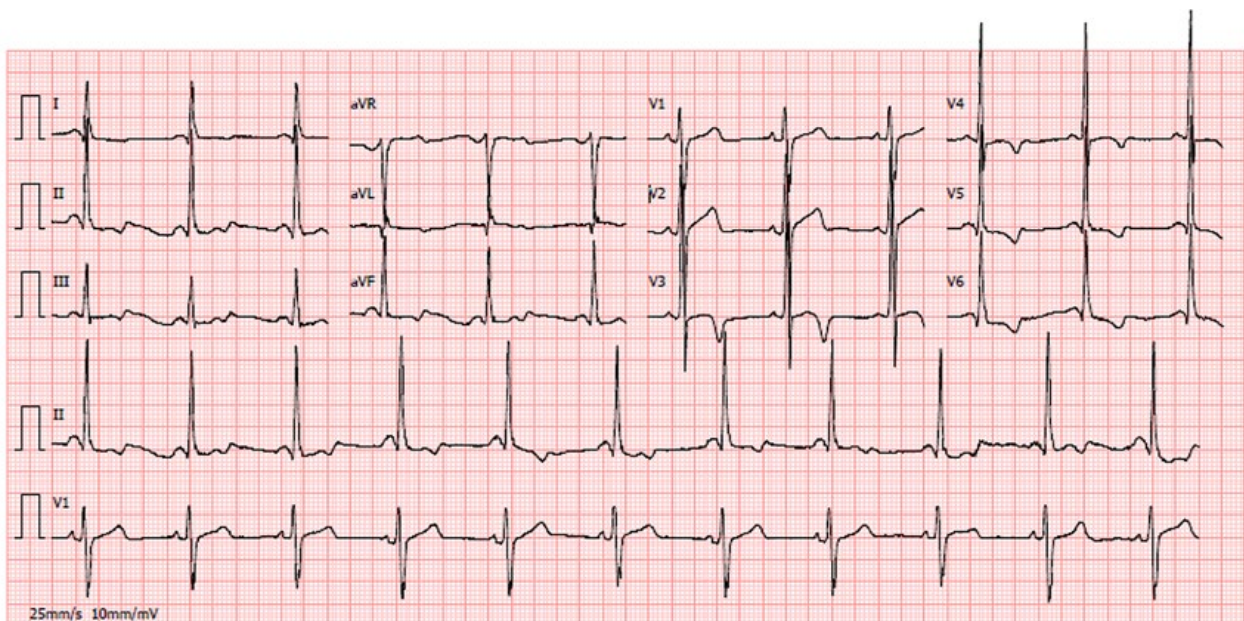


Figure 1: ECG of the patient – sinus rhythm, short P-R interval and criteria for LVH.

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echocardiogram, which raised the clinical suspicion.^{2,3}

The crucial importance of a well conducted interview and the richness of a thorough differential diagnosis is highlighted in this case.

From the assessment of an ordinary acute surgical patient, the diagnosis of a rare disease emerged, which changed the life path of this patient and his descendants. ■

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AP - Escrita do rascunho inicial, redação final
MSS, ASC, RC – Revisão da redação final

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AP - Writing initial draft, final draft
MSS, ASC, RC – Revision of the final draft

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