

Clinical judgment and decision making

Extended Abstract

The heart in Fabry's disease

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Introduction: Fabry disease is rare X-linked, recessive lysosomal storage disorder expressed as deficiency in enzyme α -galactosidase A that leads to progressive accumulation of globotriaosylceramide and related glycosphingolipids in various tissues. In cardiac tissues, progressive globotriaosylceramide accumulation leads to irreversible cardiac damage.^{1,2} Males are primarily affected by Fabry disease, but female heterozygotes may also have symptoms.³

Case report: Our patient is 47-years-old who has been suffering from chronic kidney disease since he was 23 years old. Only twelve years later he developed end-stage renal disease and has been undergoing regular haemodialysis in Dialysis Centre Prijedor (Bosnia and Herzegovina). He started his journey towards kidney transplantation in 2014 in University Hospital Centre Rijeka, which was performed in May 2018. In 2014, during pretransplantation workup, echocardiography was performed revealing concentric cardiac hypertrophy without left ventricle outflow tract obstruction (**Figure 1** and **Figure 2**) associated with contractility and diastolic filling impairment. Following suspicion on Fabry disease, diagnosis was made by measuring α -galactosidase enzyme activity in leukocytes and molecular genetic testing of GLA gene mutation. Enzyme replacement therapy was started with intravenous infusion of recombinant α -galactosidase A (agalsidase beta).

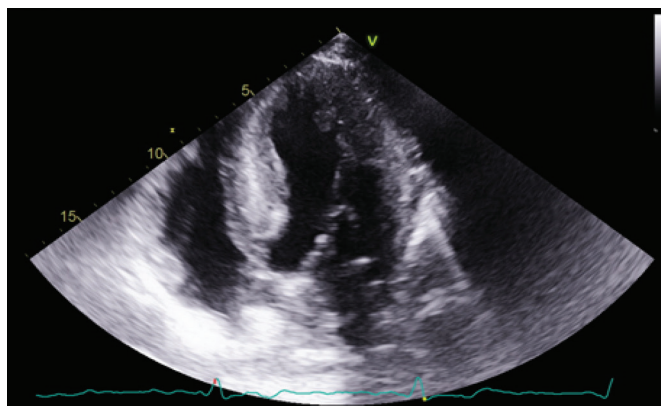


FIGURE 1. Echocardiography image, apical four-chamber view showing thickened intraventricular septum and lateral wall of left ventricle.

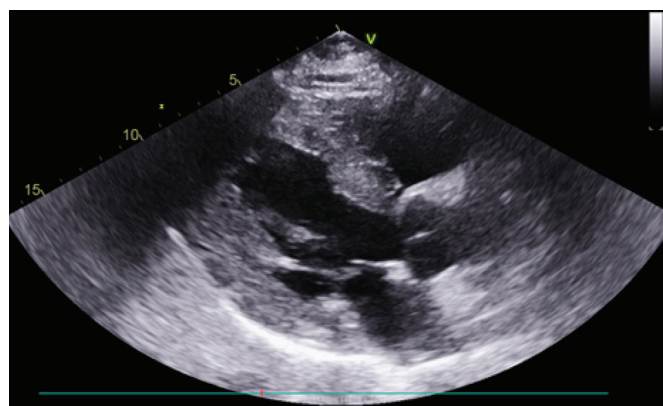


FIGURE 2. Echocardiography image, parasternal long axis view showing thickened intraventricular septum and inferolateral wall of left ventricle with prominent papillary muscle.

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Conclusion: Renal disease and echocardiographic features of hypertrophic cardiomyopathy combined with electrocardiographic and clinical criteria should be considered as "red flags" for Fabry disease.

LITERATURE

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