

# From diagnosis to treatment of the first Fabry disease patient at Dubrava University Hospital

 **Nikolina Jurković Dubravčić\***,  
 **Renee Mixich,**  
 **Andrea Pleša,**  
 **Senka Pejčković**

Dubrava University Hospital,  
Zagreb, Croatia

**KEYWORDS:** Fabry disease, cardiomyopathy, amyloidosis.

**CITATION:** *Cardiol Croat.* 2024;19(11-12):612. | <https://doi.org/10.15836/ccar2024.612>

**\*ADDRESS FOR CORRESPONDENCE:** Nikolina Jurčević Dubravčić, Klinička bolnica Dubrava, Avenija Gojka Šuška 6, HR-10000 Zagreb, Croatia. / Phone: +385-91-5771-494 / E-mail: [njurkovicdubravcic@gmail.com](mailto:njurkovicdubravcic@gmail.com)

**ORCID:** Nikolina Jurković Dubravčić, <https://orcid.org/0000-0002-9754-8712> • Renee Mixich, <https://orcid.org/0000-0002-0991-7515>  
Andrea Pleša, <https://orcid.org/0009-0008-2593-7808> • Senka Pejčković, <https://orcid.org/0000-0002-7557-9358>

**Introduction:** Anderson-Fabry disease is a rare, inherited X-linked lysosomal storage disorder. It is the second most common lysosomal storage disorder after Gaucher disease. Diagnosis is typically done through dried blood spot testing, and early detection is crucial for effective treatment. Recognizing Fabry disease in its early stages is challenging, as the initial symptoms in childhood are often subtle and nonspecific, leading to potential misdiagnosis. The hallmark of Fabry cardiomyopathy is left ventricular hypertrophy. Often, diagnosis is made after unexplained strokes in younger patients, cardiac complications, or kidney failure, meaning specialists such as neurologists, cardiologists, and nephrologists are often responsible for diagnosis.<sup>1,2</sup>

**Case report:** The case describes a 65-year-old female patient hospitalized in April 2023 with suspected hypertrophic/infiltrative cardiomyopathy. Her family history was negative for sudden cardiac death (SCD) or heart failure. Extensive diagnostics were performed, including echocardiography, serum protein electrophoresis with immunofixation, renal and abdominal ultrasound, coronary angiography, cardiac scintigraphy, fundoscopy, and cardiac MRI. The MRI results suggested cardiac amyloidosis, leading to further investigation. Enzyme activity testing for Fabry disease showed normal alpha-galactosidase levels but positive lyso-GL3, which prompted genetic testing. This confirmed a pathogenic mutation in a heterozygous state, indicating the classical form of Fabry disease. The patient began receiving Fabrazyme therapy at a collaborating healthcare facility, with a dose of 70 mg (Fabrazyme 35 mg) during a two-hour infusion. After successful nurse training at the facility, she transitioned to receiving treatment at Dubrava University Hospital (DUH), where she is regularly monitored. Her first treatment at DUH was administered successfully in September 2024, without any side effects or allergic reactions, highlighting both the effectiveness of the treatment and the team's preparation.

**Conclusion:** Accurate and timely diagnosis is critical to initiate enzyme replacement therapy, which significantly improves patient outcomes.

**RECEIVED:**  
September 30, 2024

**ACCEPTED:**  
October 31, 2024



## LITERATURE

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