Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Nephrology

Case Report: Initiation of Hemodialysis in a Child with Fabry Disease at Mohammed VI University Hospital (CHU) in Marrakech

S. Bouhjar[1*], S. Boujnane[1], N. Hamouche[1], M. Chettati[1], W. Fadili[1], I. Laouad[1]

[1]Mohammed VI University Hospital Center of Marrakesh, Cadi Ayyad University, Nephrology, Marrakesh, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i03.015 | **Received:** 29.01.2025 | **Accepted:** 05.03.2025 | **Published:** 11.03.2025

***Corresponding author:** S. Bouhjar
Mohammed VI University Hospital Center of Marrakesh, Cadi Ayyad University, Nephrology, Marrakesh, Morocco

Abstract **Case Report**

Introduction: Fabry disease is the most common lysosomal storage disorder. The classic form, which occurs in males with α-Gal A enzyme activity below 1%, typically begins in childhood or adolescence. Progressive deterioration of renal function, leading to end-stage renal disease (ESRD), usually occurs between the third and fifth decades of life. We report the case of a child diagnosed with Fabry disease at the age of 6 years, who developed chronic kidney disease (CKD) at the age of 9 years, and subsequently started hemodialysis at the age of 15 years. ***Medical Observation:*** The patient, Y.Y., a 6-year-old child, had a family history of Fabry disease, with an older brother undergoing treatment while maintaining normal renal function, and a maternal uncle who was on hemodialysis for an undocumented nephropathy before passing away. The patient was referred to the Med VI UHC in Marrakech for recurrent fever with splenomegaly. Clinical examination revealed: Hyperthermia at 39.1°C with anhidrosis, Abdominal distension with diffuse tenderness and a palpable spleen, Acroparesthesias and angiokeratomas, General deterioration in health and growth retardation. An electrocardiogram (ECG) showed sinus rhythm with signs of left ventricular hypertrophy (LVH), which was confirmed by echocardiography. Initial biological tests revealed normal renal function, but with a severely reduced plasma galactosidase level (0 µmol/L/h) and a Plasma Lyso-GL3 level of 123 ng/ml, confirming the diagnosis of Fabry disease. Over the following nine years, the patient's renal function progressively deteriorated, leading to end-stage renal disease (ESRD) and the initiation of hemodialysis at the age of 15. ***Conclusion:*** End-stage renal failure due to Fabry disease is exceedingly rare but possible in children. A detailed family history should be obtained, and Fabry disease must be considered in the differential diagnosis as a potential cause of unexplained renal failure in young boys.

Keywords: Fabry Disease, Hemodialysis, Chronic renal failure, Children, End stage renal disease.

INTRODUCTION

Fabry disease is the most common lysosomal storage disorder. A mutation in the GLA gene located on the X chromosome (Xq22.1) causes a deficiency in the enzyme alpha-galactosidase A (α-Gal A), leading to the progressive lysosomal accumulation of globotriaosylceramide and its derivatives in all cells of the body [1].

The classic form, which occurs in males with α-Gal A enzyme activity below 1%, typically begins in childhood or adolescence. Progressive deterioration of renal function, leading to end-stage renal disease (ESRD), usually occurs between the third and fifth decades of life.

We report the case of a child diagnosed with Fabry disease at the age of 6 years, who developed chronic kidney disease (CKD) at the age of 9 years, and subsequently started hemodialysis at the age of 15 years.

MEDICAL OBSERVATION

The patient, Y.Y., a 6-year-old child, had a family history of Fabry disease, with an older brother undergoing treatment while maintaining normal renal function, and a maternal uncle who was on hemodialysis for an undocumented nephropathy before passing away.

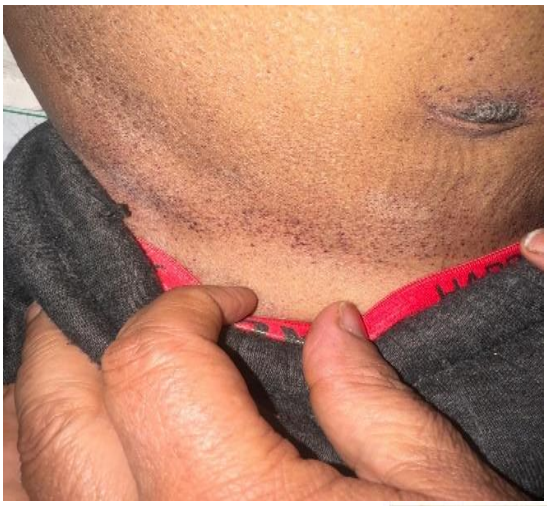
The patient was referred to the Med VI UHC in Marrakech for recurrent fever with splenomegaly. Clinical examination revealed: Hyperthermia at 39.1°C with anhidrosis, Abdominal distension with diffuse tenderness and a palpable spleen, Acroparesthesias and angiokeratomas, General deterioration in health and growth retardation. A urine dipstick test was negative. An electrocardiogram (ECG) showed sinus rhythm with signs of left ventricular hypertrophy (LVH), which was

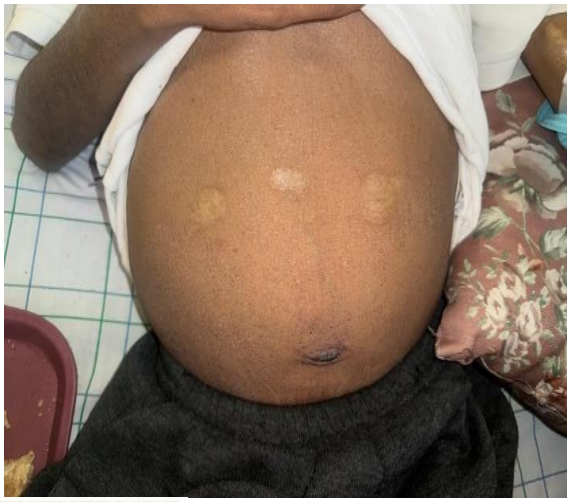
confirmed by echocardiography. The ejection fraction was normal.

Initial biological tests revealed normal renal function, but with a severely reduced plasma galactosidase level (0 µmol/L/h) and a Plasma Lyso-GL3 level of 123 ng/ml, confirming the diagnosis of Fabry disease.

Over the following nine years, the patient's renal function progressively deteriorated, leading to end-stage renal disease (ESRD) and the initiation of hemodialysis at the age of 15.

The patient was subsequently started on Agalsidase beta (1 mg/kg every two weeks). After one year of treatment, there was general clinical improvement, with no further impact on renal function.





DISCUSSION

Fabry disease (FD) is a lysosomal storage disorder caused by an abnormality in glycosphingolipid metabolism. It is X-linked and results from mutations in the gene encoding alpha-galactosidase A (Xq22.1), with over 700 mutations identified to date. These mutations lead to the accumulation of globotriaosylceramide in various cells throughout the body, particularly in the skin, heart, nervous system, and kidneys [1]. This accumulation in the endothelium and smooth muscle cells of renal blood vessels narrows the lumen, leading to ischemia and degenerative changes in the glomeruli. This process typically culminates in renal failure during the fourth or fifth decade of life [2-4]. The slow progression to renal failure may accelerate with additional kidney damage. In our patient, the absence of sweating combined with high body temperature and an

inability to concentrate urine may have predisposed him to kidney injury [5] and potentially contributed to early-onset renal failure.

CONCLUSION

End-stage renal failure due to Fabry disease is exceedingly rare but possible in children. A detailed family history should be obtained, and Fabry disease must be considered in the differential diagnosis as a potential cause of unexplained renal failure in young boys.

REFERENCES

1. Hagège, A. A., & Germain, D. P. (2015). Adult patients with Fabry disease: what does the cardiologist need to know?. *Heart*, *101*(12), 916-918.
2. Wise, D., Wallace, H. J., & Jellinek, E. H. (1962). Angiokeratoma corporis diffusum: a clinical study of eight affected families. *QJM: An International Journal of Medicine*, *31*(2), 177-206.
3. Gubler, M. C., Lenoir, G., Grünfeld, J. P., Ulmann, A., Droz, D., Habib, R., ... & Grandhomme, C. (1978). Early renal changes in hemizygous and heterozygous patients with Fabry's disease. *Kidney international*, *13*(3), 223-235.
4. Desnick, R. J., Klionsky, B., & Sweeley, C. C. (1978). Fabry's disease: a-galactosidase A deficiency. In Stanbury lB, Wyngaarden m, Frederickson DS (eds): The Metabolic Basis of Inherited Disease (ed 4). New York, McGraw-Hili, pp 810-840.
5. Schrier, R. W., Hano, J., Keller, H. I., Finkel, R. M., Gilliland, P. F., CIRKSENA, W. J., & TESCHAN, P. E. (1970). Renal, metabolic, and circulatory responses to heat and exercise: Studies in military recruits during summer training, with implications for acute renal failure. *Annals of Internal Medicine*, *73*(2), 213-223.