DOI: 10.5152/turkjnephrol.2026.251299

Turkish Journal of Nephrology

Striking Lip Angiokeratomas in a Fabry Disease Patient

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Cite this article as: Türkmen E, Köse MÖ, Çınar E, Çınar BP. Striking lip angiokeratomas in a Fabry disease patient. *Turk J Nephrol.* 2026;35(4):331-333.

Fabry disease (FD) is an X-linked hereditary lysosomal storage disorder resulting from a deficiency of the alpha-galactosidase enzyme. It is characterized by extensive systemic involvement, affecting kidneys, cardiac, and neurological systems.[1] Skin involvement is prevalent and frequently assists the clinician in establishing the diagnosis.[2] Additionally, cranial involvement typically manifests as premature strokes and white matter hyperintensities on neuroimaging, reflecting progressive cerebral vasculopathy.[3] This report presents the development of acute stroke and the progression of skin findings in a Fabry patient who declined treatment.

A 43-year-old male patient was referred to the clinic for proteinuria and chronic kidney disease (CKD). His brother had CKD and hearing loss as well. Angiokeratomas were noted around his waist and belt on the physical examination. He had nephrotic range proteinuria and a creatinine level of 4 mg/dL (GFR 18 mL/min/1.73 m^2). Echocardiography revealed left ventricular hypertrophy. Low levels of the alpha-galactosidase enzyme, as well as a mutation consistent with FD were identified.

The patient refused cranial imaging and enzyme replacement therapy (ERT). He started hemodialysis a year later and did not return for a follow-up. He suffered an undocumented stroke 2 years later. Two years after that, he had a second stroke which made it difficult for him to swallow, causing dizziness and weakness in his right arm and leg (Figures 1 and 2). The number of angiokeratomas on his body had increased since his last visit (Figure 3). Additionally, a striking line of angiokeratomas was observed on the lower lip (Figure 4). The patient also exhibited neuropathic symptoms and has been diagnosed with sensorimotor polyneuropathy. After commencing ERT with agalsidase beta, his neurological and neuropathic symptoms improved within a month.

Angiokeratomas are the second most common findings of FD skin involvement, after sweating problems.[2] It is noteworthy that the patient has skin lesions all over the body. Even more interesting is photographic evidence of increased angiokeratomas on the lower lip. As shown in Figure 4, the prominence of the lesions indicates that angiokeratomas are not only a sign of the disease but also an important indicator of patients who may be at risk of globotriaosylceramide buildup and severe systemic disease progression.[2] During the untreated period, the blood vessels in the skin deteriorated more quickly, changing shape in the process.[1,2]

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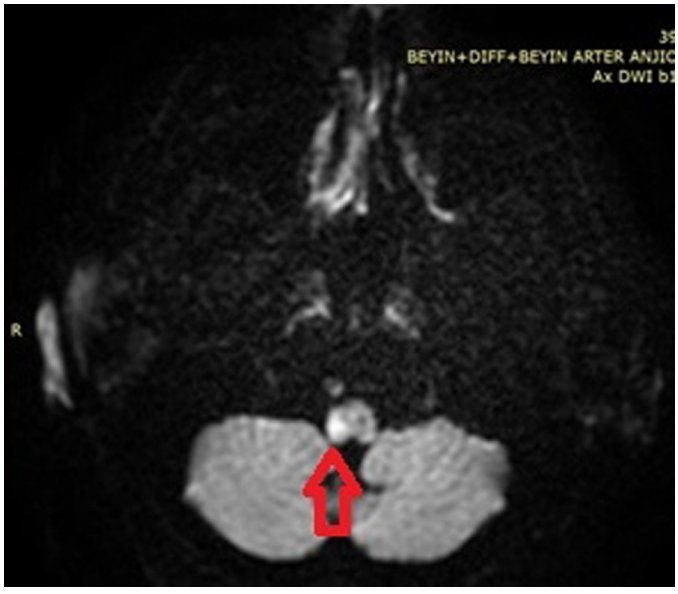

Figure 1. Diffusion magnetic resonance imaging demonstrates an acute right medullary infarction (red arrow).

The prompt initiation of agalsidase beta post-stroke resulted in the partial alleviation of neurological symptoms within a month. This rapid clinical response is important because ERT could protect organs over the long term and facilitate the recovery of FD patients from severe complications and acute cerebrovascular events.[1,3] This case highlights the fact that skin manifestations can serve as clinical indicators of disease progression and emphasizes the importance of early diagnosis and prompt ERT

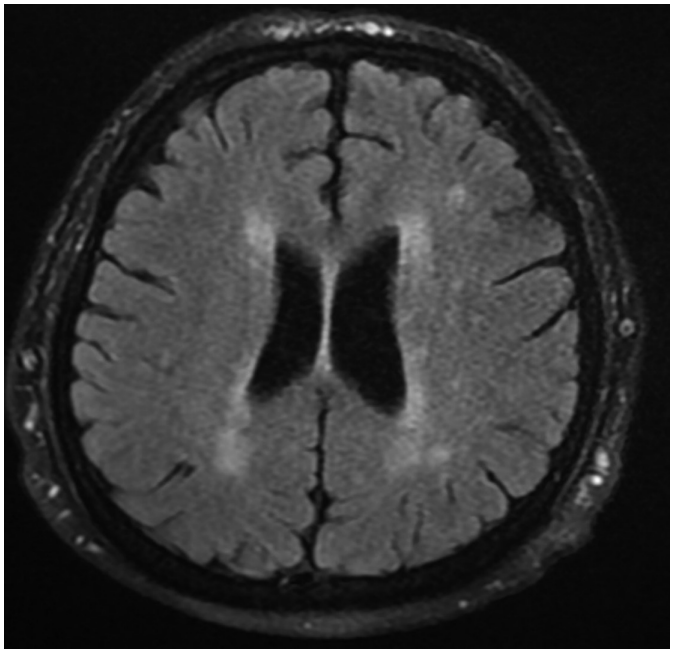

Figure 2. FLAIR magnetic resonance imaging shows chronic periventricular and subcortical white matter hyperintense lesions.

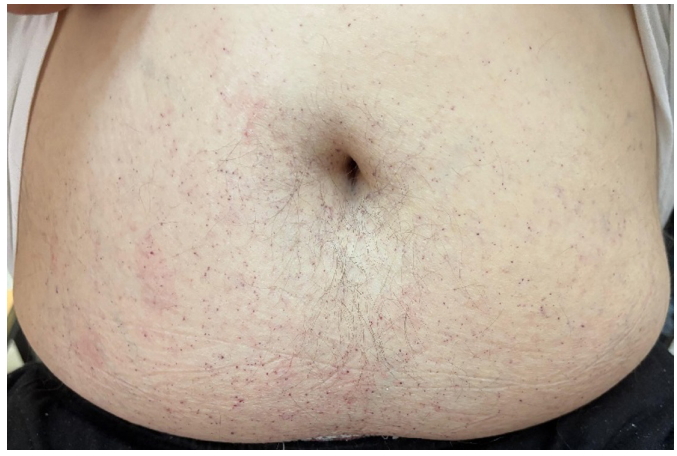

Figure 3. Widespread and prominent angiokeratomas on the patient's abdomen and umbilical region, reflecting the progression of cutaneous involvement during the untreated period.

Figure 4. Comparative images of the patient's lips: The top image shows the initial presentation, and the bottom image shows the presentation 4 years later, demonstrating a striking increase in linear angiokeratomas.

to prevent organ damage and life-threatening complications. Written informed consent was obtained form the patient.

Data Availability Statement: The data that support the findings of this study are available on request from the corresponding author.

Artificial Intelligence Usage Statement: The authors declared that no Artificial Intelligence Tool was used in the preparation of the manuscript.

Informed Consent: Written informed consent was obtained from patient who agreed to take part in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – E.T., M.Ö.K., E.Ç., B.P.Ç.; Design – E.T., M.Ö.K., E.Ç., B.P.Ç.; Supervision – E.T., M.Ö.K., E.Ç., B.P.Ç.; Resources – E.T., M.Ö.K., E.Ç., B.P.Ç.; Data Collection and/or Processing – E.T., M.Ö.K., E.Ç., B.P.Ç.; Analysis and/or Interpretation – E.T., M.Ö.K., E.Ç., B.P.Ç.; Literature Search – E.T., M.Ö.K., E.Ç., B.P.Ç.; Writing Manuscript – E.T., M.Ö.K., E.Ç., B.P.Ç.; Critical Review – E.T., M.Ö.K., E.Ç., B.P.Ç.

Declaration of Interests: The authors have no conflict of interest to declare.

Funding: The authors declared that this study has received no financial support.

REFERENCES

1. Mignani R, Biagini E, Cianci V, et al. Effects of current therapies on disease progression in Fabry disease: a narrative review for better patient management in clinical practice. *Adv Ther*. 2025;42(2):597-635. [CrossRef]
2. Al-Chaer RN, Folkmann M, Mårtensson NL, et al. Cutaneous manifestations of Fabry disease: a systematic review. *J Dermatol*. 2025;52(4):571-582. [CrossRef]
3. Tapia D, Kimonis V. Stroke and chronic kidney disease in Fabry disease. *J Stroke Cerebrovasc Dis*. 2021;30(9):105423. [CrossRef]