

## Fabry's Disease Can Mimic Acromegaly

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**Citation:** Stephen Soloway<sup>1\*</sup>, Nicholas L. DePace. *Fabry's Disease Can Mimic Acromegaly*. *Ann Case Rep Clin Stud*. 2025;4(7):1-2.

**Received Date:** 03 July, 2025; **Accepted Date:** 08 July, 2025; **Published Date:** 10 July, 2025

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### CLINICAL IMAGE

A 50-year-old Hispanic male, with proteinuria and congestive heart failure, family history of Fabry's disease, was treated and maintained on enzyme replacement therapy, co-managed by cardiologist, neurologist, rheumatologist. A rare inherited X-linked, lysosomal storage disease, caused by deficiency of alpha galactosidase A, which leads to a buildup of GB 3-Globotriaosylceramide. It may be seen in females due to lyonization, commonly presents as small fiber neuropathy, heat intolerance, congestive heart failure, proteinuria. Seen here bossing of the forehead, macroglossia, digital clubbing, OA of the hip, positive SSB antibody, renal involvement is common with disease of podocytes known as Zebra bodies. The differential diagnoses are Hurlers syndrome, Sanfilippo syndrome, Amyloidosis, and Acromegaly.

**Keywords:** Fabry Disease; Macroglossia; Bossing; SSB



