

From Tuberculosis Suspicion to Fabry Disease: A Challenging Case for an Internist

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Background: Fabry disease, is the second most prevalent lysosomal disease after Gaucher. It is an X-linked inborn error of the glycosphingolipid metabolic pathway that results in accumulation of glycosphingolipids.

Case History: A 58-year-old man presented to the Emergency Department with facial edema and painful right lateral-cervical swelling. He had a medical history of chronic ischemic heart disease and type 2 diabetes mellitus. Axial tomography showed bilateral multiple lymph nodes along the neck region suspicious for tuberculosis diseases. Quantiferon test was positive but the bacterium was not isolated at cultural test. Echocardiogram indicated hyper refringence of the myocardial tissue so a cardiac

MR was requested due to suspicion of amyloidosis. Result showed a cardiopathy of a lysosomal disease like Fabry. Patient was invited to the reference center [1].

Discussion: The present case underscores the importance of a thorough clinical evaluation for our patient. This allowed us to suspect, and then confirm the diagnosis of Fabry disease, when initial presentation was misleading.

References

1. Germain DP (2010) Fabry disease. *Orphanet J Rare Dis* 5: 30.

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