Chloroquine Toxicity Misdiagnosed as Fabry Disease Associated with Systemic Lupus Erythematosus and Hashimoto Thyroiditis

To the Editor:

Fabry disease or Anderson-Fabry disease is an X-linked recessive lysosome storage disease resulting from the deficiency of the α-galactosidase enzyme (α-Gal A)1. Low activity of the enzyme leads to abnormal lysosomal accumulation of globotriaosylceramide in many organs such as kidneys, and arterial walls, cardiac muscles, and nerve cells. While men with < 1% α-Gal A enzyme typically experience symptoms of acroparesthesias, and the appearance of angiokeratomas, hypohidrosis, characteristic corneal and lenticular opacities (cornea verticillata), and proteinuria, women (homozygous and heterozygous) can range from being asymptomatic to having severe symptoms1. Literature reports a coexistence of Fabry disease with autoimmune diseases such as systemic lupus erythematosus (SLE), rheumatoid arthritis, and IgA nephropathy⁵. The coexistence of Fabry disease, SLE, and Hashimoto thyroiditis is extremely rare. As a result of an overenthusiastic chase for the infrequent disease, we were mistaken in our diagnostic reasoning and failed to take into account all other possible causes of renal damage.

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To our knowledge, few cases of the coexistence of Fabry disease and SLE have been reported in the literature³,⁴,⁵. Likewise, there are only a few cases of chloroquine toxicity misdiagnosed as Fabry disease in patients with different rheumatic disease⁶,⁷,⁸,⁹. However, tissue damage induced by chloroquine has many morphological similarities to Fabry disease, leading to misinterpretations of pathological findings in the case of an ambiguous medical history, or in a situation in which all relevant data regarding medical therapy are not available to the interpreting pathologist⁹,¹⁰.

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