

Familial hypercholesterolemia: Clarifications

To the Editor: The article by Shah and colleagues¹ is an excellent review of familial hypercholesterolemia (FH) and highlights an underdiagnosed condition on which clinicians can make a significant impact. I would like to clarify two points:

First, as the authors describe, tendon xanthoma is mostly pathognomonic for FH. Xanthelasma, however, is nonspecific for this condition and does not appear in any of the diagnostic criteria.

Second, the American Diabetes Association (ADA) was one of the societies involved in the 2018 American Heart Association/American College of Cardiology multisociety guidelines,² and the 2020 ADA Standards of Care still reflect a low-density lipoprotein cholesterol (LDL-C) threshold for intensification of 70 mg/dL in patients at very high risk.³ I believe the authors meant to refer to the 2017 American Association of Clinical Endocrinologists/American College of Endocrinology guidelines that introduced a new category of “extreme risk” with an LDL-C treatment goal of less than 55 mg/dL, which includes patients with heterozygous FH and established atherosclerotic cardiovascular disease.⁴ This treatment goal was mirrored by the 2019 European Society of Cardiology/European Atherosclerosis Society guidelines.⁵

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In Reply: We thank Dr. Modarressi for bringing up those points, and we are grateful the review was found to be informative.¹

We agree that, although tendon xanthomas are pathognomonic for familial hypercholesterolemia (FH), xanthelasmas are not. Xanthelasmas are rich cholesterol deposits in the skin of the eyelids that occur in the setting of hypercholesterolemia.² They are nonspecific, but can be seen in patients with FH because these patients often have extreme hypercholesterolemia. Therefore, we suggest in the article that xanthelasmas could be present in patients with FH as a physical finding, and we specifically state that xanthomas are the pathognomonic lesions. To the same effect, it is also possible to see a patient with FH without xanthomas or xanthelasmas.

We also agree with Dr. Modarressi that the low-density lipoprotein cholesterol goal of less than 55 mg/dL was based on the 2017 American Association of Clinical Endocrinologists/American College of Endocrinology guideline recommendations (reference 66).³

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