

Considerations Regarding the Family Members in the Management of Patients with Familial Hypercholesterolemia

Kazuhiko Kotani^{1,2}

Learning Point of the Article:

When encountering patients with familial hypercholesterolemia in orthopedic practice, the considerations regarding the family members are necessary.

To the Editor,

We read with great interest the report written by Dr. Thakare et al., which indicated that the management of underlying lipid disorders, especially familial hypercholesterolemia (FH), should be considered when orthopedic physicians encounter patients with Achilles tendon xanthomatosis [1]. The learning points of their report are valuable for identifying patients with FH since FH tends to be underdiagnosed while a treatment regimen is being established [2]. Furthermore, we would like to add some instructive comments to their learning points.

FH is an autosomal dominant inherited disorder, which prevalently affects approximately 1 in 250–500 people, and the affected patients present with elevated low-density lipoprotein levels in the blood, which is associated with a high risk of coronary artery disease [2]. Thus, the early management of FH is crucial [2]. Although Achilles tendon xanthomatosis is an essential physical sign for FH [1], this sign is generally asymptomatic. In our experience, patients with FH visit orthopedic clinics with complaints of suspected disorders related to their ankles or discomfort (e.g., wearing the shoes) due to Achilles tendon xanthomatosis. There are many non-specific opportunities to identify patients with FH in daily practice [3],

and orthopedic physicians are therefore encouraged to participate in such opportunities. Establishing systems to easily consult lipid specialists is also needed after identifying such patients [3].

Of note, based on the dominant inheritance of FH, the general practice should not be just about the patient whom the physician sees. Instead, physicians should also examine as many family members of the patient as possible (i.e., children, siblings, and parents) to identify any other potential patients with this disease. Physicians are thus able to ask a family history of FH and premature development of coronary artery disease [2], even though the family history may be ambiguous in some cases. In addition, the physicians could suggest family cascade screening for FH [4,5], while ethical concerns need to be carefully adhered to.

From the viewpoint of earlier management, screening for pediatric FH has recently made substantial advances [4,5]. The treatment is not too late for any patient in an era when many adult patients can live longer [6]. At least, orthopedic physicians should keep in mind the importance of taking a family history of their patients and explaining to such patients the need for the timely management of FH, including family cascade screening.

Author's Photo Gallery



Dr. Kazuhiko Kotani

Access this article online

Website:
www.jocr.co.in

DOI:
<https://doi.org/10.13107/jocr.2026.v16.i03.7008>

¹Division of Community and Family Medicine, Jichi Medical University, Tochigi, Japan,
²Division of Preventive Medicine, National Hospital Organization Kyoto Medical Center, Kyoto, Japan.

Address of Correspondence:

Dr. Kazuhiko Kotani,
Division of Community and Family Medicine, Jichi Medical University, 3311-1 Yakushiji, Shimotsuke-City, Tochigi 329-0498, Japan.
E-mail: kazukotani@jichi.ac.jp

Submitted: 18/12/2025; Review: 03/01/2026; Accepted: February 2026; Published: March 2026

DOI: <https://doi.org/10.13107/jocr.2026.v16.i03.7008>

© The Author(s). 2026 Open Access. This article is distributed under the terms of the Creative Commons Attribution 4.0 International License (<https://creativecommons.org/licenses/by-nc/4.0/>), which permits unrestricted use, distribution, and non-commercial reproduction in any medium, provided you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons license, and indicate if changes were made. The Creative Commons Public Domain Dedication waiver (<http://creativecommons.org/publicdomain/zero/1.0/>) applies to the data made available in this article, unless otherwise stated.

Again, the considerations regarding the family members should be emphasized when encountering patients with FH.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

References

1. Thakare A, Bhandari H, Patwa B. A rare case of bilateral xanthomatosis of tendon of achilles: A case report. *J Orthop Case Rep* 2025;15:75-9.
2. Representatives of the Global Familial Hypercholesterolemia Community, Wilemon KA, Patel J, Aguilar-Salinas C, Ahmed CD, Alkhnifsawi M, et al. Reducing the clinical and public health burden of familial hypercholesterolemia: A global call to action. *JAMA Cardiol* 2020;5:217-29.
3. Kotani K. Primary care systems with interprofessional collaboration in the diagnosis and management of familial hypercholesterolemia. *Aust J Gen Pract* 2022;51:e1-2.
4. Pettit AR, Klaiman T, Kersting RC, Johnson C, Ogbuefi N, Moran M, et al. A qualitative study of perceptions of the care pathway for familial hypercholesterolemia: Screening, diagnosis, treatment, and family cascade screening. *Implement Sci Commun* 2024;5:135.
5. Tsai HH, Young JL, Cherny S, Ahmed CD, Khan SS, Duquette D. "I don't think people should die young": Perspectives of parents with children diagnosed with familial hypercholesterolemia. *J Community Genet* 2024;15:549-58.
6. Besseling J, Hovingh GK, Huijgen R, Kastelein JJ, Hutten BA. Statins in familial hypercholesterolemia: Consequences for coronary artery disease and all-cause mortality. *J Am Coll Cardiol* 2016;68:252-60.

Conflict of Interest: Nil
Source of Support: Nil

Consent: The authors confirm that informed consent was obtained from the patient for publication of this article

How to Cite this Article

Kotani K. Considerations Regarding the Family Members in the Management of Patients with Familial Hypercholesterolemia. *Journal of Orthopaedic Case Reports* 2026 March;16(03):429-430.

