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Familial Hypercholesterolemia: A Case Report

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Introduction

Familial hypercholesterolemia (FH) is an autosomal dominant genetic disorder caused by mutations in several genes involved in low-density lipoprotein (LDL) metabolism [1, 2]. It is characterized by an isolated increase in LDL cholesterol and is associated with a high risk of premature cardiovascular disease [3].

This condition is often underdiagnosed and undertreated, especially in resource-limited countries where genetic testing is less accessible [4]. We report the case of a patient with familial hypercholesterolemia complicated by severe cardiovascular disease.

Clinical Observation

A 44-year-old female from a first-degree consanguineous marriage, with a personal history of hypertension under treatment, family history of early deaths and hypercholesterolemia.

Cutaneous xanthomas and xanthelasmas appeared at age 7. The diagnosis of hypercholesterolemia was established at 33 during family screening. She has been treated with rosuvastatin 40 mg daily.

Her evolution was marked by severe coronary artery disease requiring double coronary artery bypass grafting.

Physical examination revealed tendon and cutaneous xanthomas as well as xanthelasmas on the eyelids (Images 1-6).

Biological Workup

- LDL cholesterol: 5.41 g/l
- HDL cholesterol: 0.34 g/l
- Triglycerides: 0.88 g/l
- Total cholesterol: 5.93 g/l
- Lipoprotein electrophoresis: clear appearance; LDL 83.3%, HDL 12.5%, VLDL 4.2%
- ApoB level: 3.14 g/l

The diagnosis of familial hypercholesterolemia was made based on Dutch Lipid Clinic Network (DLCN) criteria including:

- Family history of early coronary disease in a first-degree relative,
- Personal history of premature coronary artery disease,
- Presence of tendinous xanthomas,
- LDL-C > 3.3 g/l [5].

The vascular workup showed multiple bilateral atheromatous plaques in the carotid arteries and arteries of the lower limbs.

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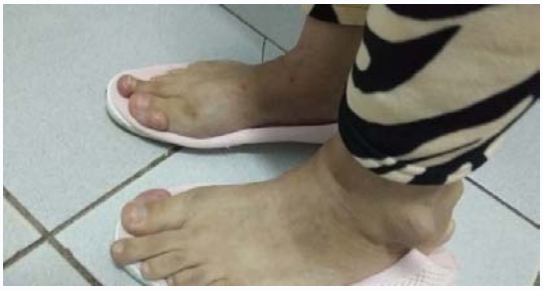

Image 1: The patient presents with a pre-tibial cutaneous xanthoma.

Image 2: Tuberous xanthoma on the thigh.

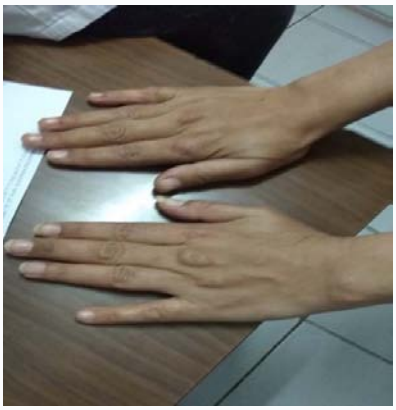

Image 3: Tendinous xanthomas at the metacarpophalangeal joints.

Image 4: Bilateral palpebral xanthelasma.

Image 5: Tuberous xanthoma at the elbow.

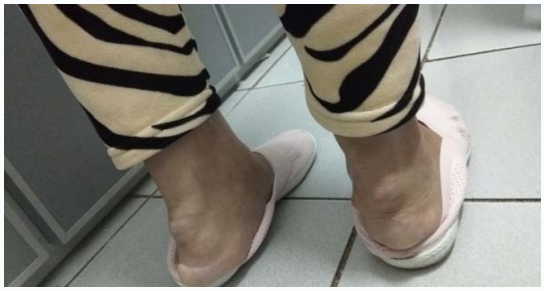

Image 6: Achilles tendon xanthoma.

Management

Treatment consisted of:

- Reinforced hygienic and dietary measures,
- Rosuvastatin 40 mg/day,
- Ezetimibe 10 mg/day,
- PCSK9 inhibitors.

Genetic testing is essential for confirmation and guiding family screening but is not available locally. Cascade screening was proposed for family members.

Discussion

Familial hypercholesterolemia is a major cause of premature atherosclerosis and cardiovascular disease, with an estimated prevalence of about 1 in 250 in the general population [6].

It remains widely underdiagnosed, with fewer than 20% of cases identified worldwide, delaying appropriate management [7].

Diagnosis relies on the DLCN criteria, which integrate family history, clinical signs (xanthomas, xanthelasmas), lipid levels, and cardiovascular complications [5].

High-intensity statin therapy combined with ezetimibe and PCSK9 inhibitors significantly reduces LDL levels and cardiovascular risk [8].

Cascade family screening is recommended to identify affected individuals early and initiate preventive treatment.

Conclusion

Familial hypercholesterolemia is a serious and under-recognized

genetic disorder requiring heightened clinical awareness, especially in the presence of suggestive family history.

Early diagnosis and optimal treatment are essential to prevent severe cardiovascular complications.

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