ORIGINAL ARTICLE

Periorbital Xanthomas in a Case of Familial Hypercholesterolemia

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SUMMARY

Background: Familial hypercholesterolemia is a genetic disorder responsible for high levels of blood cholesterol and its further consequences. As a public health issue, its proper diagnosis and management is important for healthcare systems.
Case Report: A 42-year-old patient was presented with high levels of cholesterol while having fatty liver and coronary artery disease. Her relatively extensive periorbital xanthomas were noted. Familial hypercholesterolemia was diagnosed and statin therapy was initiated with desirable follow-ups. **Conclusion:** Periorbital xanthomas should be kept in mind as a sign of high blood cholesterol in different diseases including familial hypercholesterolemia. Early diagnosis and proper management should be considered not forgetting the capability of Persian Medicine.
Keywords: Familial Hypercholesterolemia; Xanthoma; Cholesterol; Case Report

INTRODUCTION

Familial hypercholesterolemia (FH) is a monogenic autosomal dominant (90% of cases) genetic disease characterized by high levels of blood low-density lipoprotein cholesterol (LDL-C)[1]. It could result in some clinical features such as arcus senilis (corneal arcus), tendon xanthomas, and xanthelasmas[2,3]. Despite the fact that FH is the most common cause of premature coronary heart disease, it has been underdiagnosed and undertreated globally[4].

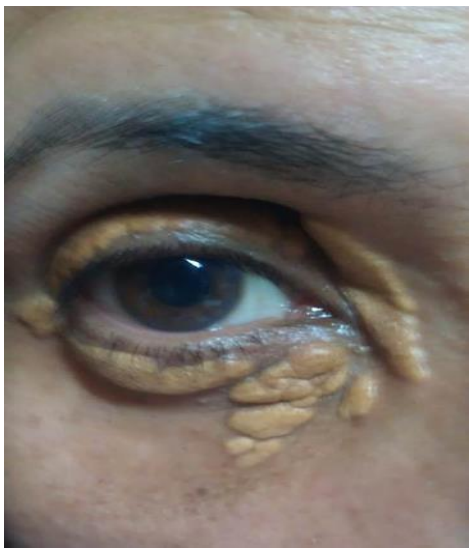
It is estimated that prevalence of heterozygous and homozygous forms of FH are 1 in 500 (even as common as 1 in 200 in some studies) and 1 in million, respectively[5]. In addition, in some ethnic groups such as Afrikaners and French Canadians FH is more prevalent[4]. Hypercholesterolemia is also known as a major risk factor for atherosclerosis, obesity, and other related metabolic disorders[6]. All in all, FH is considered as a major public health issue which requires more attention.

CASE REPORT

A 42-year-old woman was admitted to Faghihi hospital (affiliated with Shiraz University of Medical Sciences) with chief complaint of yellowish lesions around right eye. Her physical examination was quietly normal. Only tendon xanthomas (on the elbow, Achilles tendon, and dorsum of hands and feet) and xanthelasmas (Fig. 1) were noted. In past medical history, she had three-vessel coronary artery disease (CAD); therefore, coronary artery bypass graft (CABG) surgery had been done for her about six years prior to admission. Fatty liver disease had also been diagnosed for her at that time. In her family history, she reported premature ischemic heart disease in her brother, which was the cause of his death in the age of 28 years. In her lab data, high levels of total cholesterol: 670 mg/dl (reference range: 120-200 mg/dl), and low-density lipoprotein cholesterol (LDL-C): 617 mg/dl (reference range: 0-130 mg/dl). Despite high levels of total cholesterol and LDL-C, total triglyceride and high- density lipoprotein (HDL) levels (132 and 52 mg/dl respectively) were within the reference ranges.

Considering the clinical presentation and lab data, Familial hypercholesterolemia was diagnosed according to the American Heart Association diagnostic criteria for FH [7]. Consequently, statin therapy was stated for the patient in addition to advising for life style modification. She was finally discharged uneventfully. In her follow-ups, she had relatively a desirable condition while skin manifestations (xanthelasmas) had been decreased remarkably.

Fig. 1: Right sided periorbital xanthomas (xanthelasmas).



DISCUSSION

Familial hypercholesterolemia is often diagnosed by routine blood testing and usually no clinical manifestation is seen at the time of diagnosis. In heterozygous forms (HeFH), blood cholesterol level exceeds 300 mg/dl and in homozygous forms (HoFH), blood cholesterol is >600

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mg/dl. Prolonged severe types of FH may present with a spectrum of manifestations from premature cardiovascular disorders (e.g., atherosclerosis, coronary artery disease, and myocardial infarction) to cosmetic (xanthomas and xanthelasmas) or orthopedic issues (tendon xanthomas). While Achilles tendon is the most common site for tendon xanthomas, such clinical manifestation may occur in other sites such as elbow and extensor tendons of hands and feet[8].

American Heart Association has introduced following criteria for clinical diagnosis of FH: LDL-C >190 mg/dL and either premature coronary heart disease or LDL-C >190 mg/dL in a first degree relative[7].

Main differential diagnosis of FH includes: 1) sitosterolemia, 2) cerebrotendinous xanthomatosis, 3) familial combined hypercholesterolemia, and 4) polygenic hypercholesterolemia[8].

To treat FH, we should focus on reduction of blood cholesterol level. Dietary modification and a potent statin regimen along with combination of other cholesterol reducing agents are the most common used therapeutic strategies. Liver transplantation, portocaval shunting, and LDL apheresis are other treatment options[8].

It is worth mentioning that Persian Medicine (PM) as a holistic medical system has its own nutritional recommendations which have shown promising evidence for lipid lowering effects [9]. In this regard, lifestyle modification including revision of eating habits (e.g., chewing well and avoiding indulgence in eating [10]) is the main initial step even before using proper nutraceuticals or herbal formulations.

CONCLUSION

As a type of xanthoma, xanthelasmas are raised yellowish macules in eyelid and periorbital skin. While it can occur in other metabolic disorders and even in those with normal cholesterol levels, xanthelasmas usually regress by the decrease of cholesterol level. However, as it could be a sign of some important differential diagnoses, such a clinical manifestation should always be observed carefully.

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Conflict of interest: None declared.

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