



Research Article

## A CASE OF TYPE IIA HOMOZYGOUS FAMILIAL HYPERCHOLESTOLEMIA WITH CUTANEOUS XANTHOMAS

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### ABSTRACT

Familial Hypercholesterolemia (FH) is characterized by presence of various types of cutaneous xanthomas with a deranged lipid profile. An 8 year old non-obese girl presented with multiple planar, tuberous and tendinous xanthomas and had associated abnormal lipid profile with elevated LDL cholesterol levels.

**KEYWORDS:** Familial Hypercholesterolemia, Xanthomas

### INTRODUCTION

Familial Hypercholesterolemia (FH) is an inherited disorder of lipid metabolism and is commonly associated with cutaneous xanthomas[1]. We report a case of a young girl with a variety of cutaneous xanthomas in association with type Iia familial hypercholesterolemia.

### CASE REPORT

An 8 year old non-obese girl, born off non-consanguineous marriage presented to us with skin coloured swellings over her buttocks, hands and feet. These swellings were first observed at the age of about 4 years and they have been growing steadily since then. They are asymptomatic in nature. Her father had died suddenly a few years back at the age of 40 years and her elder brother died of an unknown cause at the age of 9 years.

On clinical examination, her general and systemic examination was unremarkable. Cutaneous examination showed multiple skin coloured to yellowish papules coalescing to form plaques of varying sizes were seen over

bilateral buttocks and extending into the posterior aspect of upper thighs (Figure 1). Similar lesions were seen over the interdigital spaces of both hands and inner aspect of upper arms. Yellowish to skin coloured nodules, 2x2cms were present over opposing sides of the natal cleft. Multiple skin coloured nodules were seen over dorsal aspect of fingers and toes on both sides and behind the right ankle (Figure 2& Figure 3). They were non tender, firm in consistency and not attached to underlying structures. Skin overlying was normal. There were no lesions over palmar creases, arcus juvenalis or organomegaly.

On investigation, her routine blood work was normal except for slight elevated liver enzymes. Lipid profile showed a total cholesterol level of 575 mg/dl, Triglycerides 165 mg/dl, HDL cholesterol level 49mg/dl and LDL cholesterol level of 492 mg/dl suggestive of type Iia hypercholesterolemia. ECG, chest X-ray and ultrasound of abdomen were normal. Echocardiography was normal. However Carotid Doppler showed a 30% nonobstructive plaque in the right common carotid artery.

She was started on Atorvastatin 20mg daily and advised strict diet control. A thorough examination and investigations of other members of the family was advised.

**Figure 1:** Plane xanthomas seen over bilateral buttocks and extending into the posterior aspect of upper thighs. Tuberosus xanthomas seen over the natal cleft



**Figure 2:** Multiple tendinous xanthomas seen over dorsal aspect of fingers and toes on both sides



**Figure 3:** Plane xanthomas seen over interdigital spaces of both hands



## DISCUSSION

Familial hypercholesterolemia (Type IIa of the Fredrickson classification) is a codominant disorder of lipid metabolism characterized by strikingly elevated levels of low density lipoprotein (LDL) cholesterol levels with normal triglycerides, cutaneous xanthomas and premature coronary atherosclerosis [1].

FH is due to mutation in the gene for LDL receptors as a result of which the total and LDL cholesterol levels are elevated at birth. However the triglyceride levels are normal and High Density Lipoprotein (HDL) cholesterol levels are normal or reduced. In homozygous state, LDL is removed at only one-third of normal rate and hence there will be plasma LDL levels five to six times normal value [1],[2]. They present with unique yellowish xanthomas in the interdigital spaces of hands, natal cleft and tuberous xanthomas over extensor surfaces subject to stress or trauma especially over the hands, elbows, knees and buttocks [2]. Our patient had a combination of plane, tuberous as well as tendinous xanthomas.

There have been such cases reported from amongst the Indian population [4],[5],[6]. All of these reports indicate the risk of early cardiovascular consequences in homozygous state. Our patient had an abnormal lipid profile with high levels of LDL cholesterol levels as well as atherosclerotic plaque in her carotid artery which puts her at a higher risk for catastrophic coronary sequelae. There has been a case report of giant tuberous xanthomas in a 26 year old with type IIa hypercholesterolemia [7].

The most devastating complication of homozygous familial hypercholesterolemia is accelerated atherosclerosis which can result in death in early childhood. Leung C-H et al. reported a case of familial hypercholesterolemia with xanthomatosis associated with Diabetes Mellitus which further increased the risk of premature coronary heart disease [8]. The clinical importance in a homozygous patient is to foresee a very early development of severe coronary artery disease, and myocardial infarction which can occur in the first and second decades of life. A careful evaluation of first degree relatives and extended family members with special attention to lipid profile plays an important role in early detection of the condition and treatment [1].

## **CONCLUSION**

It is important for physicians and especially dermatologists to recognize xanthomas as they are the earliest clinical indicators of this condition and an early diagnosis and treatment will help in preventing the development of early coronary artery disease.

## **CONFLICT OF INTERESTS**

The authors declare no conflict of interests regarding this Research Article.

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