Atypical Case of Various Types of Xanthoma in a 7-Year-Old Baby Girl.

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ABSTRACT

Introduction: Xanthomas are a common presentation of disorders of lipid metabolism usually associated with abnormalities of cholesterol metabolism. They are cutaneous manifestations of lipidoses in which lipids accumulate in large foam cells within the skin. They are associated with hyperlipidemias, both primary and secondary types. Observation: A 7-year-old female child presented to us with lesions of Xanthelasma on the left upper eyelid and multiple types of xanthomas on rest of the body. Routine investigations and systemic examination was normal. Lipid profile revealed raised serum cholesterol levels, serum triglycerides was within normal range, high low density lipoproteins, normal levels of high density lipoproteins and very low density lipoproteins suggestive of predominant hypercholesterolemia. Histopathology from a lesion showed showed skin with mild atrophic epidermis and overlying orthokeratosis. Dermis contains sheets of foamy macrophages with admixed Touton type giant cells. Conclusion: To our knowledge this is first case report of various morphological types of xanthomas with xanthelasma and associated predominant hypercholesterolemia in a 7-year-old girl child with no history of any similar lesions in any of the family members.

Case Report:
A 7-year-old female child presented with complaints of multiple asymptomatic raised lesions over the body since past 2-years. Lesions were initially noticed as small lesions over buttocks and they gradually increased in size and number and involved different parts of the body. There was no history of similar lesions in any of the other family members but the birth history shows III Degree consanguinity. Father of the child died at age of around 40 years due to some uneventful incidence and mother is asymptomatic, even her lipid profile is in normal range. There was no history suggestive of systemic involvement. There was no history suggestive of myocardial infarction or stroke in any of the family members. On examination, there were multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter distributed symmetrically around Gluteal region (Figure 1), elbows (Figure 2), extensor digitorum (Figure 3), knees (Figure 4) and Achilles’ tendon (Figure 5). The lesions were flat around left eyelid suggestive of Xanthelasma (Figure 6). Multiple, asymptomatic, firm, smooth, symmetrical, subcutaneous swellings were noted at bilateral elbows, around bilateral extensor digitorum tendons, bilateral knees and around bilateral Achilles’ tendon. Slit lamp examination and fundus examination was normal. Routine hematological investigations including complete blood counts, liver and renal function tests, blood sugar, thyroid profile, chest radiographs and urine examination were within normal limits. Lipid profile revealed total serum cholesterol of 511mg/dl [200 mg/dl], serum triglycerides 159mg/dl [60-165mg/dl], low density lipoproteins (LDL) of 250mg/dl [<130mg/dl], high density lipoproteins (HDL) of 48mg/dl [>35mg/dl], very LDL of 32mg/dl [5-40mg/dl]. The electrocardiogram was normal, without any abnormal waves. Ultrasonography of abdomen revealed multiple renal calculi, the largest measuring 6.4mm with no obstructive changes on right side, rest of the ultrasonography is normal. Histopathological examination of biopsy sample from lesion over left elbow showed skin with mild atrophic epidermis and overlying orthokeratosis. Dermis contain sheets of foamy macrophages with admixed Touton type giant cells (Figure 7). PAS stain and FiteFaraco stains are negative for organisms. The diagnosis was thus established as a case of Xanthelasma, Extensor digitorum xanthomas, Achilles’ tendon xanthomas and Eruptive xanthoma suggestive of Predominant hypercholesterolemia. The patient was advised strict dietary control regarding the intake of fats. Dietary management involves restriction of dietary saturated fat to <7% of calories, minimization of trans fats, restriction of dietary cholesterol to <200mg/day, replacement of animal fats by vegetable oils, and increasing intake of fish intake.

Figure 1: Showing Eruptive Xanthoma near the Gluteal Region (Buttocks)
Figure 2: Showing multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter around right and left elbows.

Figure 3: Showing multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter around Extensor digitorum.

Figure 4: Showing multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter around right and left knees.

Figure 5: Showing multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter over Achilles’ Tendon.

Figure 6: Showing Xanthelasma on left upper eyelid.
In this case, the patient’s lipid profile revealed total serum cholesterol of 511mg/dl[200 mg/dl], serum triglycerides 159mg/dl[60-163mg/dl], low density lipoproteins (LDL) of 250mg/dl[<130mg/dl], high density lipoproteins(HDL) of 48mg/dl[>35mg/dl], very LDL of 32mg/dl[5-40mg/dl].

Xanthelasma(or xanthelasma palpebrarum)
Xanthelasma (or xanthelasma palpebrarum) is a sharply demarcated yellowish deposit of fat underneath the skin, usually on or around the eyelids.20 While they are neither harmful nor painful, these minor growths may be disfiguring and can be removed.19 They are common in people of Asian origin and those from the Mediterranean region. Because of the hereditary component, they may or may not indicate high blood levels of cholesterol. Where there is no family history of xanthelasma, they usually indicate high cholesterol and may correlate with a risk of atheromatous disease. A xanthelasma may instead be referred to as a xanthoma when becoming larger and nodular, assuming tumorous proportions.21 Still, xanthelasma is often classified simply as a subtype of xanthoma.22 In this case on examination the patient showed flat lesions around left eyelid which is suggestive of Xanthelasma.

Eruptive Xanthoma:
Eruptive xanthoma usually appearing on the back and buttocks, or the extensors of knees and elbows of hyperlipemic individuals, it is the sudden appearance of groups of yellow-brown papules surrounded by an erythematous halo.23

In this case, on examination there were groups of yellowish papules all over the gluteal region(buttocks).

Xanthoma tendinomus: Xanthoma tendinomus (also tendon xanthoma or tendinous xanthoma)4 is clinically characterized by papules and nodules found in the tendons of the hands, feet, and heel.4 Also associated with familial hypercholesterolemia (FH).24 In this case, on examination there were multiple, non-tender, smooth surfaced soft papule and nodules ranging from 1cm to 5cm in diameter distributed symmetrically around elbows, extensor digitorum, knees and Achilles' tendon.

Conclusion: To our knowledge this is first case report of various morphological types of xanthomas like Extensor digitorum xanthomas, Achilles’ tendon xanthomas and Eruptive xanthoma with xanthelasma and associated predominant hypercholesteremia in a 7-year-old girl child with no history of any similar lesions in any of the family members. We hereby conclude that xanthomas in any clinical presentation act as a marker for the underlying lipoprotein abnormalities which should be diagnosed and managed as early as possible to decrease the risk of coronary artery disease and pancreatitis in later years of life. The patient is advised strict dietary control regarding the intake of fats.

REFERENCE

Figure 7: Showing mild atrophic epidermis and overlying orthokeratosis. Dermis contains sheets of foamy macrophages with admixed tontype giant cells.