Research Paper

Volume : 5 | Issue : 1 | JANUARY 2016 • ISSN No 2277 - 8179

Medical Science

KEYWORDS : Xanthoma, Predominant Hypercholesteromia, Xanthelasma

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

Dr.Basawaraj Puranik — Professor, Department of General Medicine, Shadan Institute of Medical Sciences, Teaching Hospital and Research Centre, Himayathsagar Road, Hyderabad, Telangana, India.

Dr.Shashank Kumar Srivastav — House-Surgeon, Shadan Institute of Medical Sciences, Teaching Hospital and Research Centre, Himayathsagar Road, Hyderabad, Telangana, India.

ABSTRACT

Introduction: Xanthomas are a common presentation of disorders of lipid metabolism usually associated with abnormalities of cholesterol metabolism. They are cutaneous manifestations of lipidosis 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 hypercholesteromia. Histopathology from a lesion showed showed skin with mild atrophic epidermis and overlying orthokeratosis. Dermis contain 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 hypercholesteromia in a 7-year-old girl child with no history of any similar lesions in any of the family members.

Introduction:

Xanthomas are common presentation of disorders of lipid metabolism usually associated with abnormalities of cholesterol metabolism.[1,2] They are cutaneous manifestations of lipidosis in which lipids accumulate in large foam cells within the skin. They are associated with hyperlipidemias, both primary and secondary types.[3] We hereby report a case of 7-year-old girl child presenting with various morphological types of xanthomas with xanthelasma and associated predominant hypercholesteromia with no history of any such lesions in any of the family members which is a rare in occurance.

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), 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 hypercholesterolaemia. The patient was adviced 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.

Volume : 5 | Issue : 1 | JANUARY 2016 • ISSN No 2277 - 8179

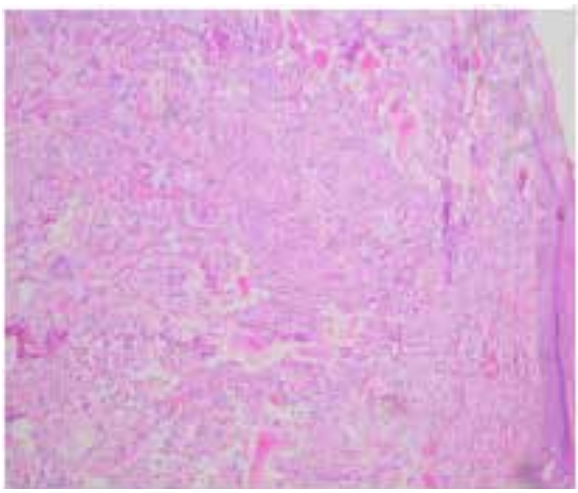

Figure 7: Showing mild atrophic epidermis and overlying orthokeratosis. Dermis contains sheets of foamy macrophages with admixed touton type giant cells.

Discussion:

Predominant Hypercholesterolaemia:

Hypercholesterolemia (also spelled hypercholesterolaemia also called dyslipidemia) is the presence of high levels of cholesterol in the blood.[4] It is a form of "hyperlipidemia" (elevated levels of lipids in the blood) and "hyperlipoproteinemia" (elevated levels of lipoproteins in the blood).[4] Cholesterol is a sterol; see the diagrammatic structure at the right. It is one of three major classes of lipids which all animal cells utilize to construct their membranes and is thus manufactured by all animal cells. Plant cells do not manufacture cholesterol. It is also the precursor of the steroid hormones, bile acids and vitamin D. Since cholesterol is insoluble in water, it is transported in the blood plasma within protein particles (lipoproteins). Lipoproteins are classified by their density: very low density lipoprotein (VLDL), low density lipoprotein (LDL), intermediate density lipoprotein (IDL) and high density lipoprotein (HDL).[5] All the lipoproteins carry cholesterol, but elevated levels of the lipoproteins other than HDL (termed non-HDL cholesterol), particularly LDL-cholesterol are associated with an increased risk of atherosclerosis and coronary heart disease.[6] In contrast, higher levels of HDL cholesterol are protective.[7] Elevated levels of non-HDL cholesterol and LDL in the blood may be a consequence of diet, obesity, inherited (genetic) diseases (such as LDL receptor mutations in familial hypercholesterolemia), or the presence of other diseases such as diabetes and an underactive thyroid.[4] Reducing saturated dietary fat is recommended to reduce total blood cholesterol and LDL in adults.[8] In people with very high cholesterol (e.g. familial hypercholesterolemia), diet is often insufficient to achieve the desired lowering of LDL and lipid lowering medications which reduce cholesterol production or absorption are usually required.[9] If necessary, other treatments such as LDL apheresis or even surgery (for particularly severe subtypes of familial hypercholesterolemia) are performed.[9]

In this case, the patient's 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].

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.[10] While they are neither harmful nor painful, these minor growths may be disfiguring and can be removed.[10] 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 xanthelasmata, 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.[11] Still, xanthelasma is often classified simply as a subtype of xanthoma.[12] 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 yellowish-brown papules surrounded by an erythematous halo.[13]

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

Xanthoma tendinosum:

Xanthoma tendinosum (also tendon xanthoma or tendinous xanthoma[14]) is clinically characterized by papules and nodules found in the tendons of the hands, feet, and heel.[3] Also associated with familial hypercholesterolemia (FH).[15] 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 hypercholesteromia 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 lipid 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 adviced strict dietary control regarding the intake of fats.

REFERENCE

1. Mohan KK, Kumar KD, Ramachandra BV. Tuberous xanthomas in type IIA hyperlipoproteinemia. Indian J Dermatol Venereol Leprol 2002;68:105-6. | 2. Parker F. Xanthomas and hyperlipidemias. J Am Acad Dermatol 1985;13:1-30. | 3. James, William D.; Berger, Timothy G. et al. (2006). Andrews' Diseases of the Skin: clinical Dermatology. Saunders Elsevier. ISBN 0-7216-2921-0. | 4. Durrington, P (August 2003). "Dyslipidaemia". The Lancet 362 (9385): 717–31. doi:10.1016/S0140-6736(03)14234-1. PMID 12957096. | 5. Biggerstaff KD, Wooten JS (December 2004). "Understanding lipoproteins as transporters of cholesterol and other lipids". Adv Physiol Educ 28 (1–4): 105–6. doi:10.1152/advan.00048.2003. PMID 15319192. | 6. Carmena R, Duriez P, Fruchart JC (June 2004). "Atherogenic lipoprotein particles in atherosclerosis". Circulation 109 (23 Suppl 1): III2–7. doi:10.1161/01.CIR.0000131511.50734.44. PMID 15198959. | 7. Kontush A, Chapman MJ (March 2006). "Antiatherogenic small, dense HDL--guardian angel of the arterial wall?". Nat Clin Pract Cardiovasc Med 3 (3): 144–53. doi:10.1038/ncpcardio0500. PMID 16505860. | 8. Hooper L, Summerbell CD, Thompson R et al. (2012). Hooper, Lee, ed. "Reduced or modified dietary fat for preventing cardiovascular disease". Cochrane Database Syst Rev 5: CD002137. doi:10.1002/14651858.CD002137.pub3. PMID 22592684. | 9. Ito MK, McGowan MP, Moriarty PM (June 2011). "Management of familial hypercholesterolemias in adult patients: recommendations from the National Lipid Association Expert Panel on Familial Hypercholesterolemia". J Clin Lipidol 5 (3 Suppl): S38–45. doi:10.1016/j.jacl.2011.04.001. PMID 21600528. | 10. Lang, Gerhard K (2000). Ophthalmology. Stuttgart: Thieme.[page needed] | 11. Shields, Carol; Shields, Jerry (2008). Eyelid, conjunctival, and orbital tumors: atlas and textbook. Hagerstwon, MD: Lippincott Williams & Wilkins. ISBN 0-7817-7578-7.[page needed] | 12. "xanthelasma". Mosby's Medical Dictionary (8th ed.). 2009. Retrieved November 8, 2012. | 13. "Xanthoma". Medical Dictionary - Dictionary of Medicine and Human Biology. Retrieved 2015-02-05. | 14. Rapini, Ronald P.; Bolognia, Jean L.; Jorizzo, Joseph L. (2007). Dermatology: 2-Volume Set. St. Louis: Mosby. pp. 1415–16. ISBN 1-4160-2999-0. | 15. Van Den Bosch, Harrie C.M.; van den Bosch, Harrie C.M. and D. Vos, Louwerens (May 28, 1998). "Achilles'-Tendon Xanthoma in Familial Hypercholesterolemia". New England Journal of Medicine 338 (22): 1591. doi:10.1056/NEJM199805283382205. PMID 9603797. Retrieved 5 April 2013. |