Research Paper

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Multiple Xanthomas in a 10 Year Old Girl Child – a Case Report

Medical Science

KEYWORDS : Xanthoma, Familial hypercholesterolemia, Excision

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ABSTRACT

Cutaneous manifestations of hyperlipidemia include xanthelasma and xanthomas. These lesions consist of lipid laden foamy histiocytes under the skin (foam cells). The occurrence of xanthomas in the absence of xanthelasma is quite rare. We report a 10 years old girl child with multiple tuberous xanthomas over the knee,buttock,tendoachilles and elbow in the absence of xanthelasma,who was diagnosed to be a case of familial hypercholesterolemia. The lesions over the knee and buttock were huge and not responding to oral lipid lowering agents. Hence surgical excision of these lesions was done.

CASE REPORT:

This 10 year old non obese girl presented with multiple non tender papulo nodular lesions over the knees, elbow, gluteal region and the ankle since the age of one (fig.1). These lesions were progressively increasing in size. There was no such complaint in the family members. Further investigations revealed a normal blood glucose levels and a high levels of serum cholesterol and low density lipoproteins. Thyroid profile was normal. Ophthalmic examination was done and there was no associated corneal arcus juvenalis and lipemia retinalis. An echocardiogram was done which revealed a normal study. Usg abdomen was normal. Her family members were screened and their lipid profile also revealed the similar findings of elevated cholesterol and LDL. Hence she was diagnosed as a case of Familial Hypercholesterolemia. As per physician opinion she was started on oral lipid lowering agents (atorvastatin). The lesions over the knees and the gluteal region were huge and were of hindrance to her day to day activities. Hence we proceeded with excision of those lesions (fig.2).HPE report was consistent with tuberous xanthoma. The post operative period was uneventful. The patient is on regular follow up. She was adviced to continue the lipid lowering agents regularly with frequent monitoring of the lipid profile and cardiac checkups.

fig.1

fig.2

DISCUSSION:

Xanthomas and xanthelasma are the cutaneous manifestations of the disorders of lipid and lipoprotein metabolism [1]. Familial hypercholesterolemia is an inherited autosomal dominant disorder. It may be homozygous or heterozygous. Homozygous variety is clinically characterized by cutaneous xanthomas,atherosclerosis and corneal arcus usually developing in early childhood.[2] Xanthomas are slowly enlarging subcutaneous nodules attached to tendons,ligaments,fascia and periosteum with normal overlying skin. Extensor surface of hands, feet and ankle are involved more frequently. Various forms of xanthomas are tuberous, plane; eruptive, tendinous, subperiosteal etc. Total cholesterol levels are usually more than 500mg/dl and can be as high as 1200 mg/dl. The major complication of familial hypercholesterolemia is accelerated atherosclerosis which can result in ischemic heart disease even in childhood. These patients often have symptoms of vascular disease before puberty but symptoms can be atypical or go unreported till adulthood [2]. Parents and other relatives should be screened for hypercholesterolemia. Hence theses patients with xanthomas are at increased risk to develop coronary artery disease and occasionally pancreatitis.[3] The main aim of treatment for xanthomas that are associated with an underlying lipid disorder is to identify and treat the lipid disorder initially so that associated systemic complications can be prevented. In most instances treating the underlying hyperlipidemia will reduce or resolve the xanthomas and will reduce the risk of heart diseases [4]. Dietary and lifestyle modifications with oral medications are used to treat the disorder. Medications commonly used are statins/fibrates etc. [4] Surgical excision of xanthomas was done when lesions do not resolve spontaneously or with treatment of underlying cause. However surgical methods may not be a permanent treatment as recurrence rates for xanthomas depend on the underlying serum cholesterol levels.

CONCLUSION:

Tuberous Xanthomas occur as cutaneous manifestation of hyperlipidemia. They usually occur in young adults. Medical management is usually suboptimal. In this case report, we have described a ten year old girl with multiple tuberous xanthomas refractory to lipid lowering agents. Hence surgical excision was done.

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