INTRODUCTION

Intracranial xanthomas are rare entity. They occur among patients with hyperlipidemia which consists of several entities such as polygenic hypercholesterolemia, combined familial hyperlipoproteinemia and familial hypercholesterolemia.

Pharmacologic agents along with dietary measurement are the basis of treatment. Medical treatment includes bile resins, niacin and statins that work indirectly to increase low-density receptors and reduce serum low- density lipoprotein levels. These measures stabilize lesions and sometimes may cause regression.

We present a case of intracranial xanthoma associated with type 2 familial hypercholesterolemia.

CASE REPORT

A 20 years female was admitted with complaints of reduced hearing in the right ear with mucopurulent discharge since 3 months associated with occipital headache there; was no history of fever, giddiness, convulsions or vomiting. She was diagnosed as having a cholesteatoma and was posted for modified radical mastoidectomy. She was referred to us for medical fitness, when we incidentally noted cutaneous xanthomas, which were distributed symmetrically over both elbows (Fig. 1), knees, popliteal and antecubital fosse. Her eyes showed evidence of arcus juveniles (Fig. 2). Right ear showed sagging of postero-superior tympanic membrane and ear canal with scanty mucopurulent discharge. Her routine lab investigations were normal except for a high cholesterol level (687mg%). Table 1 shows the lipid profile of the patient. An X-ray of right mastoid (Schuller’s view) showed evidence of sclerosis of mastoid air cells. CT scan brain (Fig. 3) showed a soft tissue density enhancing on contrast (C.T. Value 28 to 36.3 HU on plain, 41 HU on contrast) involving the mastoid air cells, middle ear cavity and external auditory
canal on the right side with erosion of the mastoid, temporal bone, anterior wall and sinus plate; this was thought to be a cholesteatoma. Periossicular soft tissue density was noted, however the ossicular chain was preserved. A possibility of mastoid xanthoma (ectopic occurrence of fat) was also kept in mind. During her tympanomastoid exploration it was noted that, there was erosion of the lateral wall of the mastoid antrum, posterior wall of the external auditory canal. The surgeons tried to remove the disease from middle ear by limited excision. Postoperatively, patient had a right lower motor neuron facial palsy.

Histopathologic examination of the tissue removed during surgery revealed a yellowish gomous material. On microscopy it shows multiple cholesterol crystals surrounded by foamy macrophages, inflammation and calcification suggestive of mastoid- xanthoma. Other family members were also investigated for lipid profile (Table 2).

Thus we had a patient of familial homozygous hypercholesterolemia (Type-II a) with rare presentation of intracranial xanthoma. She was treated with Tab. Atorvastatin 20 mg HS which was increased to 30 mg/day for better control. The patient soon developed statin induced proximal myopathy, hence the dose of atorvastatin was reduced to 20 mg/day and Tab. Ezetimibe 10mg was added.

Her current lipid profile 8 months later is given in Table 1, the cutaneous xanthomas have flattened and the right sided facial palsy which had developed post-operatively has recovered.

**DISCUSSION**

The heritable hyperlipidemia is of six types, I, IIa, IIb, III, IV and V. Subcutaneous xanthomas typically occur in patient with heritable hyperlipidemia. Types II and III hyperlipidemia are caused due to excess circulating lipoproteins and moderately elevated serum cholesterol levels. Accelerated atherosclerosis frequently occurs, resulting in premature coronary artery disease and stroke.

Abnormal lipid storage in the setting of normal serum lipids may occur in conditions such as histiocytosis X, leading to xanthoma formation and needs to be distinguished from xanthomas due to hyperlipidemia.

Xanthomas develop because of lipid leakage from the vascular into the surrounding tissue, where macrophages subsequently phagocyte these lipids. Because cholesterol is not degraded, it accumulates within these cells, creating “foamy” macrophages. The extracellular cholesterol crystallizes into clefts and induces an inflammatory reaction with giant cells and resultant fibrosis. Systemic xanthomas most commonly occur along the Achilles, patellar, and extensor tendons of the hands, buttocks, elbows, eyelids, and hand creases.

Intracranial xanthomas have been reported rarely among patients with hyperlipidemia, most commonly type II. Familial hypercholesterolemia has a dominant inheritance pattern. Combined familial hyperlipoproteinemia occurs in 1% to 2% of the population and presents in the 3rd to 4th decade of life. It is usually not associated with xanthomas. It arises from a reduced number of hepatic low-density lipoprotein receptors, leading to reduced low density lipoprotein clearance from the blood.

Intracranial and extracranial xanthomas can occur...
in the temporal bone,\textsuperscript{3} the skull base (clivus),\textsuperscript{1} and over the cerebral convexities. Most occur in middle-aged and elderly patients, although they have been reported in young patients.\textsuperscript{3,4,6} Because of their slow progression, they tend to present late in life.\textsuperscript{3,4,6} Clinical presentation depends on lesion location and extent. Symptoms may include severe headache,\textsuperscript{1} otorrhea,\textsuperscript{6} cranial nerve palsy, tinnitus,\textsuperscript{1,6} and otitis media.

Although the diagnosis of an intracranial xanthoma may be suggested by its imaging appearance, the diagnosis is usually not considered because of its rarity. These are circumscribed, extraaxial masses that are hypodense to brain on unenhanced CT scans.\textsuperscript{1,3,6} Osseous abnormalities include bony destruction and remodeling.\textsuperscript{1,3,6} The MR imaging appearance of xanthomas is in part due to their high lipid content. On Unenhanced T1- weighted images, most are hyperintense, with corresponding heterogeneous low signal intensity on T-2 weighted images. After the IV administration of contrast material, these lesions do not show significant enhancement.

Pharmacological agents (bile resins, niacin, statins) used alone or in combination with dietary measures are the basis of treatment of the hyperlipidemia. These may stabilize and sometimes, regress the lesions. So early medical intervention is important.

REFERENCES