LIPOID PROTEINOSIS - A CASE REPORT

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ABSTRACT

Lipoid Proteinosis (LP) is a radiologically rare inborn error of metabolism in which abnormal deposition of lipids and proteins in multiple systems of the body in the presence of an absolutely normal lipid profile.[1,2] Even though any systems of the human body is affected by it, the most predominant manifestations to a dermatologist`s eye are lipid laden skin lesions and to a radiologist`s eye are symmetric intracranial calcifications. To one who is aware of this entity, LP is usually an incidental diagnosis. Because of its rarity and to spread awareness about it in the scientific community, a case of LP diagnosed incidentally in a 45 years male who was referred for CT scan of head following a road traffic accident is being reported here.

Keywords: Lipoid Proteinosis; Intracranial calcification; Skin lesions, CT scan

Case Report

A 45 years male was referred for CT scan examination of head as he was found lying on the road following a road traffic accident.

Clinically he was conscious, co-operative and well oriented to time, place and person. He had no prior history of any systemic diseases. Yellowish patches (Fig.1) were seen on the skin over the leg. There was absence of any hyperkeratosis of skin and depigmentation of lips. The tongue was freely mobile. There was no hoarseness of voice. There was no significant family history or consanguineous marriage. His blood counts and the lipid profile were within normal limits. Serum cholesterol was 126 mg/dl (normal < 200 mg/dl), serum triglycerides were 82 mg/dl (normal < 170 mg/dl) and serum HDL cholesterol was 41 mg/dl (normal < 70 mg/dl).

Plain radiograph of skull was unremarkable (Fig. 2). Plain CT scan brain window images showed subtle symmetric comma shaped calcifications in medial aspect of the temporal lobes (Fig. 3).

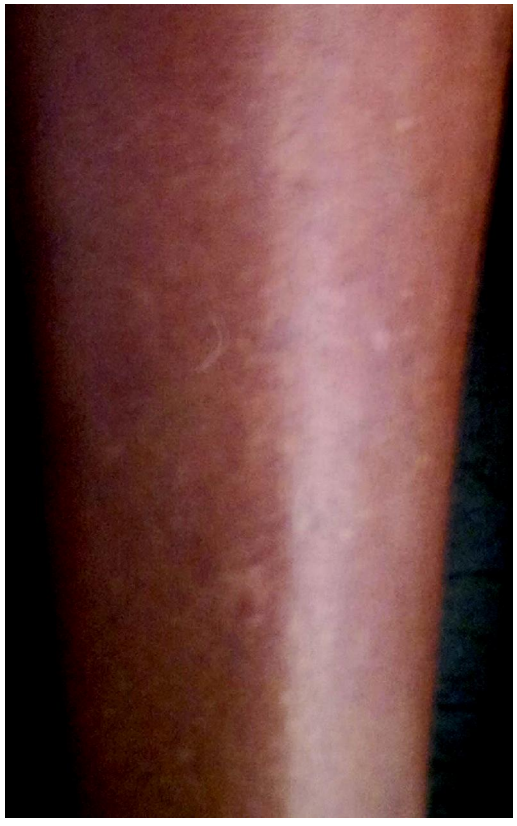

Figure 1: Yellowish patches are seen on the skin over the leg.

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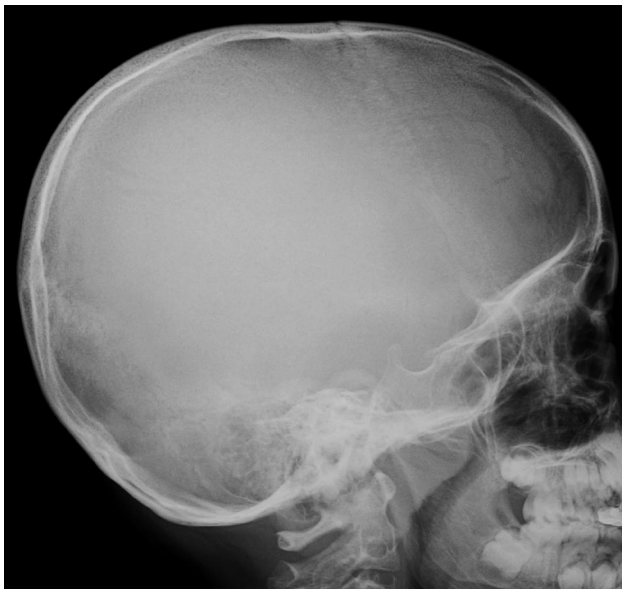

Figure 2: Plain radiograph of skull appears within normal limits.

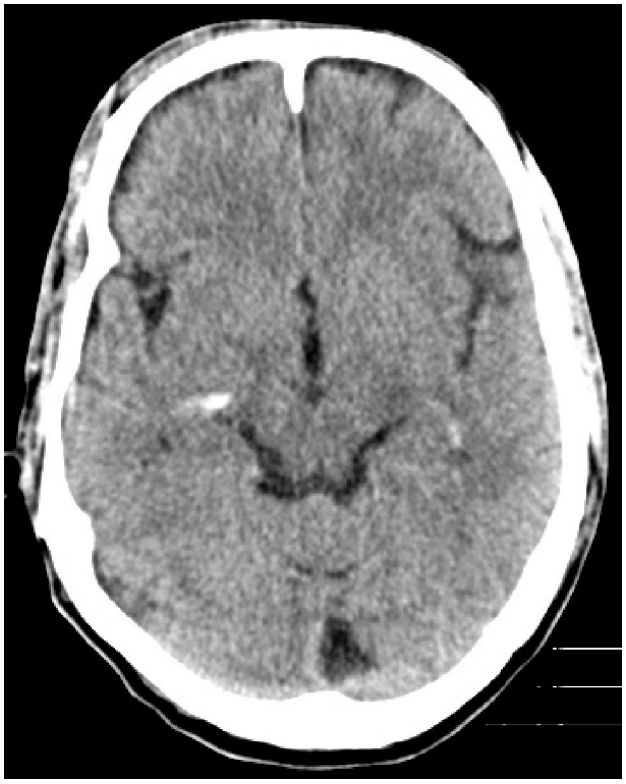

Figure 3: Plain CT scan at brain window images shows subtle symmetric comma shaped calcifications in medial aspect of the temporal lobes that is classically seen in LP

Hence a radiological diagnosis of LP was made. Histopathology of punch biopsy from yellowish plaques on skin showed abundant deposition of amorphous eosinophilic material surrounding the sweat glands, capillaries and in the thickened papillary dermis. The hyaline like material was diastase resistant and PAS positive (Fig. 4).These findings were consistent with the radiological finding of LP.

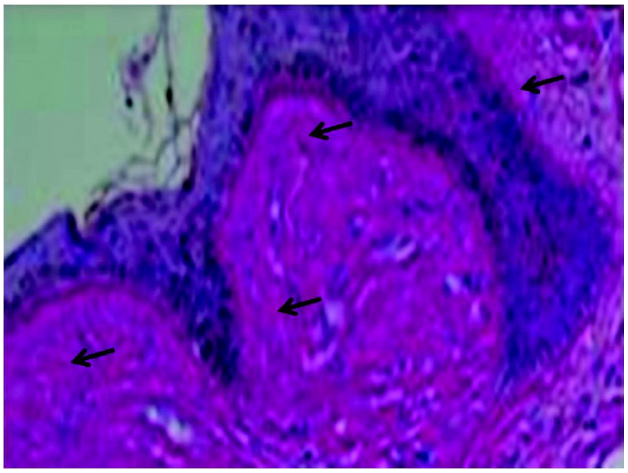

Figure 4: Histopathology of punch biopsy from yellowish plaques on skin showed abundant deposition of amorphous PAS positive eosinophilic material surrounding the sweat glands, capillaries and in the thickened papillary dermis.

Discussion

It was Seibman who first reported Lipoid Proteinosis in 1908.[3] Urbach and Weithe subsequently named it as "Lipoidosis cutis et mucosae" in 1929.

Hamada et al[4] mapped LP to chromosome 1q21 at D1S498[5] and concluded that mutations outside exon 7 exhibit more severe mucocutaneous LP phenotype.[6] LP has a slow but steady and progressive course. Histologically the material deposited in the tissues resembles lipids and proteins. Hence this entity was erroneously labeled as LP.

Cases reported from Asia are very few, although most cases have been seen in the Caucasian races in South Africa and Central Europe.[3,8] The entity is very rare and till date not more than 500 cases have been reported from all over the world. Therefore, the actual data about incidence and prevalence is not completely known. No definitive age, sex or race predilections exist. There is documented autosomal recessive inheritance as there is usually history of consanguinity among unaffected parents.[9] The subtype of parent disorder manifesting with mental sub normality has been reported to be transmitted by autosomal dominant inheritance due to mutant genes.[3]

In the gene encoding extracellular matrix protein 1 (ECM1) on band 1q21, loss of function mutations are

seen. Patients with exon 7 mutations display slightly milder clinical features, while mutations in exon 6 result in a more severe phenotype.[5,6] Recently a novel nonsense mutation in the ECM1 gene in a Pakistani family has been identified.[7]

A glycoprotein produced by the normal ECM1 gene is expressed in skin, mucosa and the entire human viscera. Mutation in this gene leads to deposition of hyaline like material in the skin and viscera in abnormal amounts which is the cause of clinical manifestations. These deposits stain positive with Periodic Acid-Schiff stain, are diastase resistant and negative for Congo red.[10]

On barium swallow examination the deposits of hyaline like material can be seen as filling defects in upper aero-digestive tract. Radiological hallmark is the presence of bean to comma shaped intracranial calcifications in the temporal lobes in amygdala which is more evident in patients with prolonged affliction with LP. Epilepsy, when present, may be related to these calcifications. Hence complete evaluation by performing MRI/CT scan in patients with LP is recommended in order to identify these abnormalities.[11] Radiographs and CT are adequate for a definitive diagnosis, while MRI is useful to assess the brain in totality.

The differential diagnoses to this entity are not many. Although the skin lesions in Erythropoietic protoporphyria (EPP) have similar appearance, the deposits are not seen around sweat glands. Increased protoporphyrin levels in erythrocytes are also seen in EPP.[11] Deposits in amyloidosis and xanthomas have different chemical composition although externally skin might appear similar as in LP.

LP is not always asymptomatic. When extensive and prolonged, complications of LP that are known are airway obstruction due to laryngeal involvement, hoarseness of voice and impaired speech due to vocal cord involvement, gastrointestinal bleed if small bowel is involved and corneal opacities and secondary glaucoma due to ocular involvement.[3,4]

There is no permanent cure for LP. Symptomatic medical treatment for skin lesions includes dimethyl sulphoxide, etretinate, intralesional heparin and D-penicillamine. However none of these medicines have shown consistent good results. Dermabrasion for skin lesions is done in cases which show no improvement by oral medications. Carbon dioxide laser for lesions of eyelids and aero digestive tract is also under consideration. Anticonvulsants are used when the patient has seizures.[1,2]

To conclude, although LP is not deadly, it can at times be quite distressing. Appropriate radio-pathological workup can lead to early detection so that a team effort of multiple specialties can render proper management.

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