LIPOID PROTEINOSIS

A J Kanwar, Malkit Singh, S C Bharija, B N Dutta and M S Belhaj

A case of lipoid proteinosis presented with multiple papular lesions on the face and hoarseness of voice from early infancy. Five sibs of the patient were affected and one had died early in infancy, probably because of laryngeal obstruction.

Key words : Lipoid proteinosis, Hoarseness.

First described by Urbach and Wiethe in 1929,[1] lipoid proteinosis is an autosomal recessive disorder characterized by deposition of an amorphous eosinophilic material in the skin and other tissues. Deposition of this material results in several characteristic features like thickened eyelids with beaded margins, xanthoma-like plaques on elbows and knees, and hoarseness of voice which is often the first clinical manifestation.[2] The disease is rare and less than 300 cases have been reported so far.[3] The purpose of the present communication is to report a case of lipoid proteinosis in a Libyan male. Six siblings of the patient had also been similarly affected. To the best of our knowledge, this is the first case report of this rare disorder from this part of the world.

Case Report

A 21-year-old Libyan male farm worker had gradually progressive papular lesions and scars on the face and extremities of several years duration. Although he had a life-long history of hoarseness, he denied any episodes of dyspnoea or any other respiratory problem. There was no history of photosensitivity, convulsions or any gastro-intestinal complaints. His parents were first cousins. Five other younger sibs of the patient (one brother and four sisters) had similar skin lesions and hoarseness of voice since very early childhood. Still another sib, had died at the age of 1½ years due to some respiratory problem. Other affected family members were not available for complete evaluation.

The patient was well developed and intelligent. He had several small waxy papules of variable sizes on the forehead, nose, cheeks, eyelids, elbows, knees and abdomen. Atrophic and some varioliform scars were observed on the forehead, cheeks and elbows. On the mucosal surfaces of lower lips, there was a large nodular lesion, in addition to small papular lesions. Though the tongue was firm on palpation, there was no significant macroglossia. However, the patient was unable to protrude it beyond the lip margins. There was patchy alopecia of the beard region. Hair on other parts of the body including the eyelashes were normal. Lesions on the elbows were rather large and had a xanthomatous appearance. There was no hyperkeratosis. Teeth were normal and systemic examination was unremarkable. Secondary sexual characters had been well developed.

Biopsy of a waxy nodular lesion on the elbow revealed a dense eosinophilic hyaline material beneath the basal layer of the epidermis and also surrounding the capillaries, sweat gland ducts and hair follicles. The material was positive with the periodic acid-Schiff stain and negative with stains for amyloid. Laboratory data were within normal limits : erythrocyte

From the Departments of Dermatology and Pathology, Faculty of Medicine, Al-Arab Medical University, Benghazi, Libya.

Address correspondence to: Dr. A. J. Kanwar.

sedimentation rate, complete blood cell counts, serum proteins and lipid electrophoresis, glucose tolerance test and blood, urine and faeces for porphyrins. Skiagrams of chest and skull were normal. No calcification was found on skull roentgenograms.

Comments

The diagnosis of lipoid proteinosis usually presents no difficulties. A combination of hoarseness of voice from early infancy and waxy papulo-nodular lesions which heal with scars are highly diagnostic. Our patient presented typical genetic, clinical and histopathologic features of lipoid proteinosis. The absence of disease in the parents of the patient who were first cousins is compatible with the known recessive autosomal transmission.[3-5]

The other clinical features which may be encountered in patients having lipoid proteinosis are : recurrent pain and swelling in the parotid glands due to obstruction of Stensen's duct,[6] eye changes such as corneal ulcerations[7] and fundal abnormalities.[3] Partial alopecia and retarded hair growth as observed in our patient have been reported by Bansal and Mathur.[9] Intracranial calcification is also an important clinical feature of this disorder.[10] A few patients suffer from epilepsy, but majority exhibit no central nervous system deficit.[11] Epilepsy may not occur before the age of 53 years.[3]

All the clinical manifestations of lipoid proteinosis are due to the deposition of a hyaline material in the skin, mucous membranes and other tissues. The exact nature of this material is unknown, but it appears to consist primarily of neutral mucopolysaccharide with some hyaluronic acid, tryptophan, neutral fat and cholesterol.[12] Lipid appears to be present in lesions due to the affinity between lipoproteins and glycoproteins rather than due to a primary metabolic defect.[2] According to Fleishmajor et al,[13] hyaline material consists essentially of type IV collagen.

The hoarseness of voice which is the most common clinical finding in lipoid proteinosis is due to deposition of hyaline in the vocal cords. These deposits may be quite extensive especially on the larynx and pharynx and severe cases may require tracheostomy.[11] It is probable that the cause of death in one younger sib of the patient was laryngeal obstruction due to the hyaline material.

There is no known therapy for lipoid proteinosis.[10] The cutaneous and mucous membrane abnormalities in lipoid proteinosis although cosmetically and functionally compromising, do not appear to decrease longevity except untreated laryngeal obstruction.[1] The treatment is largely symptomatic, like surgical removal of vocal cord infiltrates, anticonvulsants for epilepsy, dermabrasion of facial lesions and tracheostomy for laryngeal obstruction.

References

1. Urbach E and Wiethe C : Lipoidosis cutis et mucosae, Virchow Arch Pathol Anat Physiol 1929; 273 : 285-294.
2. Shore RN, Howard BV, Howard WJ et al : Lipoid proteinosis : demonstration of normal lipid metabolism in cultured cells, Arch Dermatol, 1974; 110: 591-595.
3. Barthelemy H, Mauduik G, Kanitakis J et al : Lipoid proteinosis with pseudo-membranous conjunctivitis, J Amer Acad Dermatol, 1986; 14 : 367-372.
4. Hofer PA : Urbach Wiethe disease (lipoglycoproteinosis, lipoid proteinosis, hyalinosis cutis et mucosae) a review, Acta Dermato-Venereol, 1973; 53 : 1-5.
5. Rosenthal AR and Duke JR : Lipoid proteinosis, Amer J Ophthalmol, 1967; 64 : 1120-1128.
6. Burnett JW and Marcy SM : Lipoid proteinosis, Amer J Dis Child, 1963; 105 : 81-84.
7. Hewson GE : Lipoid proteinosis (Urbach Wiethe syndrome), Brit J Ophthalmol, 1963; 47 : 242-246.
8. Jensen AD, Khodadoust AA and Emery JM : Lipoid proteinosis : report of a case with electron

microscopic findings, Arch Ophthalmol, 1972; 88 : 273-277.

9. Bansal NK and Mathur GK : Lipoid proteinosis —a case report, Ind J Dermatol Venereol Leprol, 1982; 48 : 209-212.
10. Ramsey ML, Tschen JA and Wolf JE : Lipoid proteinosis, Internat J Dermatol, 1985; 24 : 230-235.
11. Weidner WA, Wenzl JE and Swischuk LE : Roentgenographic findings in lipoid proteinosis, Amer J Roentgenol, 1970; 110 : 457-462.
12. Caro I : Lipoid proteinosis, Internat J Dermatol, 1978; 17 : 388-392.
13. Fleischmajor R, Timpl R, Graves P et al : Hyalinosis cutis et mucosae, a basal lamina disease, J Invest Dermatol, 1981; 76 : 314-320.
14. Finkelstein HW, Mammond HL and Jones RB : Hyalinosis cutis et mucoase, Oral Surg, 1982; 54: 49-53.