LIPOID PROTEINOSIS
(A case report with review of literature)

R. T. BAGALE *

Summary

A case of lipoid proteinosis is being reported because of its rare occurrence. A brief review of literature is given.

Introduction

Lipoid proteinosis (also known as hyalinosis cutis et mucosae, Urbach-Wiethe disease, nevus ichthyosiformis) was first described by Siebenmann in 1908. Urbach-Wiethe reported on the clinical and histopathological findings in nine cases in 1929. Approximately 200 cases have been reported in world literature. Age at which the cases were diagnosed varied from 6 months to 67 years but most of the lesions appeared in infancy. Skin and mucous membranes are mainly affected but systemic nature of the disease also has been documented[1]. The disease is recessively inherited with variable expressivity, affects all populations and shows equal sex distribution. Pathogenicity of this disease is not understood. Microangiopathy and hypersensitivity to physiological trauma have been postulated as possible etiological factors[2]. Personal and family history of diabetes mellitus has been reported.

Case Report

An eight year old boy from Osmanabad (Marathwada) area was seen in the skin O.P.D. with complaints of multiple skin eruptions on the body and hoarseness of voice since infancy.

The child was born after full-term normal delivery at home and cried well after birth. By the end of first year the mother noted gradual onset of hoarseness of voice. About 2-3 months later the child developed papular lesions initially over face which were followed by similar lesions as well as occasional blisters, erosions, nodules and verrucous plaques on the trunk and proximal extremities many of which had resolved to form atrophic scars. There was no predeliction in sun exposed areas. There was no history of convulsions.

There was no family history of similar illness or diabetes mellitus. The parents were consanguinous.

On examination physical stature was normal for age, but the child appeared to have mild mental retardation. The

* Lecturer in Skin & V.D.
S. R. T. R. Medical College and Hospital
Ambajogai, Maharashtra - 431517

Request for reprints:
Dr. R. T. Bagale,
Lecturer in Skin & V. D.
S. R. T. R. Medical College & Hospital
Ambajogai, Maharashtra - 431517

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scalp hair was sparse especially on the vertex. Nails showed longitudinal striations. There was characteristic beading at the margins of both eyelids. Varioliform scars were seen on the face especially on the forehead, nose and cheeks. Reticular and keratotic papular lesions were noted over the sides of the neck, anterior axillary folds, antecubital and popliteal fossae. Few irregular erosive lesions of 2-3 cm. size were seen over the back but most of the back lesions were ivory-white atrophic scars, surrounded by hyperpigmented, verrucous collar. Similar lesions were also seen on the rest of the trunk and proximal part of the extremities. Yellowish white papules of about 1-2 mm size were seen on the inner aspect of the lower lip. Similar lesions were noted on the buccal mucosa and soft palate. The tongue appeared swollen and the frenum was bound-down so that patient could not protrude his tongue properly. The voice was hoarse and hollow. Indirect laryngoscopy showed beading and irregularity of the vocal cord margins with impairment of vibrations. Fundoscopic examination did not reveal any abnormality. Systemic examination was normal. A clinical diagnosis of lipoid proteinosis was made.

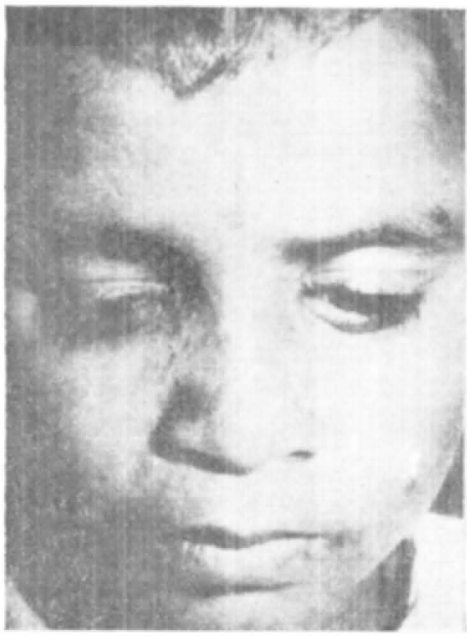

Fig. 1 Photograph showing characteristic beading at lid margins and pockmarks over face.

Hemoglobin, leucocyte count, ESR, blood VDRL, LFT, routine urine examination, X-Ray Chest and X-Ray skull revealed no abnormalities. Skin biopsy from one of the verrucous plaques showed hyperkeratosis and papillomatosis with extensive deposition of PAS positive amorphous hyaline material in the papillary dermis and around the capillaries.

Patient was treated with framycetin sulphate 1% cream for the erosive lesions and tablet predinisolone 5 mg. twice daily for ten days which was tapered gradually over a period of three weeks and then stopped. The skin lesions improved and the patient was subjectively better, but there was no improvement in the voice.

Discussion

Lipoid proteinosis is a hereditary metabolic disorder of fibrinocytes which, because of a presumed enzyme deficiency leads to deposition of functionless amorphous hyaline material consisting largely of glycoproteins but also a lipoid component (hence lipoid proteinosis) in the skin and mucous membranes. Mucous membrane lesions usually precede the skin lesions. Grosfeld et al have described the three cardinal features of the disease-beaded thickening of eyelid margins, hoarseness of voice and intracranial calcifications[3]. The latter feature was not present in our patient. Hoarseness is generally present at birth or appears in early infancy as seen in our case. This results from deposition of hyaline material in the vocal cord margins

which hampers its vibrations. Pale pink to yellowish papules are also seen mainly in the oral, pharyngeal and laryngeal mucosa. Such infiltrations also have been reported in the mucous membranes of oesophagus, stomach, jejunum, rectum, bronchii, lungs, pancreas, testis, kidneys, labia minora and vagina. Recurrent pain and swelling may develop in the parotid glands due to obstruction of Stensen's duct.

Fundal changes such as drusen of Bruch's membrane reported in about half of the patients[4] were absent in our case.

The beanshaped hippocampal calcification on either side of the sella turcica may be associated with variable degree of mental retardation and epilepsy of temporal lobe type[5]. Akinetic, psychomotor, uncinate and major and minor epileptic attacks may develop in later childhood. These intracranial calcifications were seen in eight out of twenty two patients where skull films were taken but only two of these had epilepsy[6]. Our patient who was assessed to have mild mental retardation showed no evidence of calcification in the brain. He also had no other sign of CNS involvement.

The disease is extremely chronic but benign. Severe respiratory tract infection may pose threat to these patients' life and may require an urgent tracheostomy to prevent asphyxia. There is no specific treatment available. Prednisolone may be used with some objective clinical improvement and shift of the mucopolysaccharide pattern in the skin towards normal. Removal of the vocal cord infiltrations may be attempted for symptomatic relief from hoarseness. Dermabrasion of facial lesions may help in removing some infiltrations and pockmarks[7].

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