LIPOID PROTEINOSIS

(Urbach Wiethe)

By

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Lipoid proteinosis is a rare disorder, about 50 cases having been reported until 1957 (Ungar and Katzenellenbogen, 1957). The entity was first described in 1929 by Urbach and Wiethe and consists pathologically of disseminated areas of hyalinosis and lipoid deposits in the dermis, the oral mucosa, the larynx and and other viscera. To our knowledge this disease has not previously been reported from India.

Report of a case:—V. B., Hindu male, aged 23 years presented with skin lesions since he was 10 months of age, and hoarseness of voice since the age of 4. The lesions appear in crops, each attack involving only a few areas of the skin. During the course of 22 years, the lesions have appeared on all parts of the body surface. The lesion starts as a small solid swelling (papule) which evolves into a bleb (vesicle) within the course of 4–5 days, these develop into lesions containing pus-like fluid which burst leaving behind ulcers which heal in 10 to 15 days, resulting in varioliform scars. The scars were more prominent on the face. Exacerbation of lesions were noticed during extremes of weather in the summer and winter and itching occurs only occasionally. The lesions had no relation to food or sunlight. There was no fever during the episodes of the skin lesions. The frequency of these had lessened since he was 17 years. Sweating was minimal but he could tolerate heat. Hoarseness of voice was first noticed when he was 4 years old. This become progressively worse till the age of 18.

He was normal at birth and the mile stones were normal. He had no infection during infancy. Parents are first cousins and are alive. He has 3 brothers and 3 sisters who do not have similar disease. No history of similar disease in ancestral members of his family is available.

The patient weighs 38 3 Kgs. and the height is 156 cms. Hairs on the scalp were sparse. Several depressed scars were present on the face and scalp (fig. 1,2). Skin is irregularly thickened, waxy and dry. Warty lesions were present on the elbows. The eye lashes had fallen off and the margins of eye lids were thickened and beady (fig. 3). The mucous membranes of the lips and cheeks showed irregular yellowish thickening. The tongue had yellowish nodules, and its margins were thickened and show several pitted scars (fig. 4,5). The movements of the tongue was restricted. All the teeth were carious. Laryngoscopic examination showed no lesions on the vocal cords. X-ray skull did not reveal any area of calcification. His basal metabolic rate was 9%. Serum Cholesterol was 125 mg. and serum total phospolipid was 292

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mg per 100 cc. which was slightly raised (normal range 150–274 mg. per 100 cc.). Chemical extraction studies on the urine showed no evidence of porphyria. Repeated V. D. R. L. tests were negative.

Biopsy of the skin from the forearm showed hyperkeratosis and acanthosis of the epidermis. The granular layer was prominent. With the phospotungstie acid haematoxylin stain a very prominent stratum lucidum was seen, which varied in thickness, and had a wavy outline (fig. 6). Similar hyaline was seen surrounding dermal appendages. Where the hyaline was dense clefts were seen in the materiai (fig. 7). These, to us, appear to be shrinkage artifacts. Frozen sections stained for fat showed only sparse collections of sudanophilic material in the hyaline and around the appendages. Only one biopsy was examined for fat and the scantiness of fat is probably because a longstanding lesion was examined. Alcian blue stain revealed acid mucopolysaccharide material in the wall of the blood vessels. An increase of reticulin fibres was seen with Gomori's reticulin staining in the upper dermis as described by Ungar and Katzenellenbogen (1957). Verhoeff's elastic stain brought out irregularly clumped and fragmented fibres especially in the vicinity of the hyaline material surrounding adnexae (fig. 8).

Discussion :—The clinical and pathological features are clearly those of lipoid proteinosis. Consanguinity of parents as in our cases has been noted to be frequent (Butterworth, 1962). The condition is transmitted as a recessive trait.

The disease is characterised by the deposition of a hyaline "Lipoid Protein" substance in the mucous membrane and skin (Butterworth 1967). The lesions are yellowish white papules, discreet or confluent, and are usually present on the face, neck, hands and eyelids. The margins of eyelids may become beaded. Bullous, pustular and crusting lesions may also occur and these may result in scars of variolform variety or appearing as large, smooth depressed and atrophic areas (Wood et al, 1956). Alopecia of tne scalp and the beaded area of the face in males, and loss of eyelashes have been noted. Lesions have been noted in the submucosa of the mouth, tonsils, larynx, pharynx, trachea, oesophagus, stomach, vagina, testes, eye and brain. Caplan (1967) obtained visceral biopsies and also studied an autopsy. He found lipid material in capillaries and small arterioles of subepithelial tissues of jejunum, appendix urinary bladder and interstitial fibrous tissue of the pancreas. Holtz and Schulze (1951) thought that the disease was related to a combined disturbance of protein and fat metabolism with an endocrine basis. Braun and Weyhbrecht (1952) regarded this as a paraproteinosis on morphological and histological grounds. Hanig and Kramer (1955) concluded that the disease was due to a disturbance of the protein metabolism on the basis of an increase of the globuln fraction of the serum and a decrease of albumin. However, others believe that the disturbances of protein metabolism may be secondary.

The disease has been considered to be related to porphyria (Lundt 1949). However there has been no confirmation of this view. A view has been expressed that amyloidosis and lipoid proteinosis are related disorders, though no unequivocal

evidence has been presented. The hyaline in our material could be stained with Congored but did not show the green birefringence after Congored stain. Green birefringence (dichroism) after Congored stain is one of the most reliable evidences for amyloid.

Urbach and Wiethe (1929) maintained that the sudanophil deposits and hyaline changes result from seepage of fat and protein material from the blood plasma. Weyhbrecht and Korting (1954) postulated an increased permeability of the terminal blood vessels in the dermis. However Ungar and Katzenellenbogen (1957) point out that the neutral fat is seen in location of plaques of degenerated elastica and along the course of hyalinized fibres in areas of dermis which are poorly vascular. These to them suggest an origin from degenerated collagen and elastica which normally contain bound fat. Further they point out that the hyaline does not give a Ninhydrin reaction which would be expected to be positive if the hyaline was due to seepage of the serum from the blood vessels. Evidence of degeneration of elastic fibres were found in our case and the arguments of Ungar and Katzenellenbogen (1957) appear very reasonable to us.

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