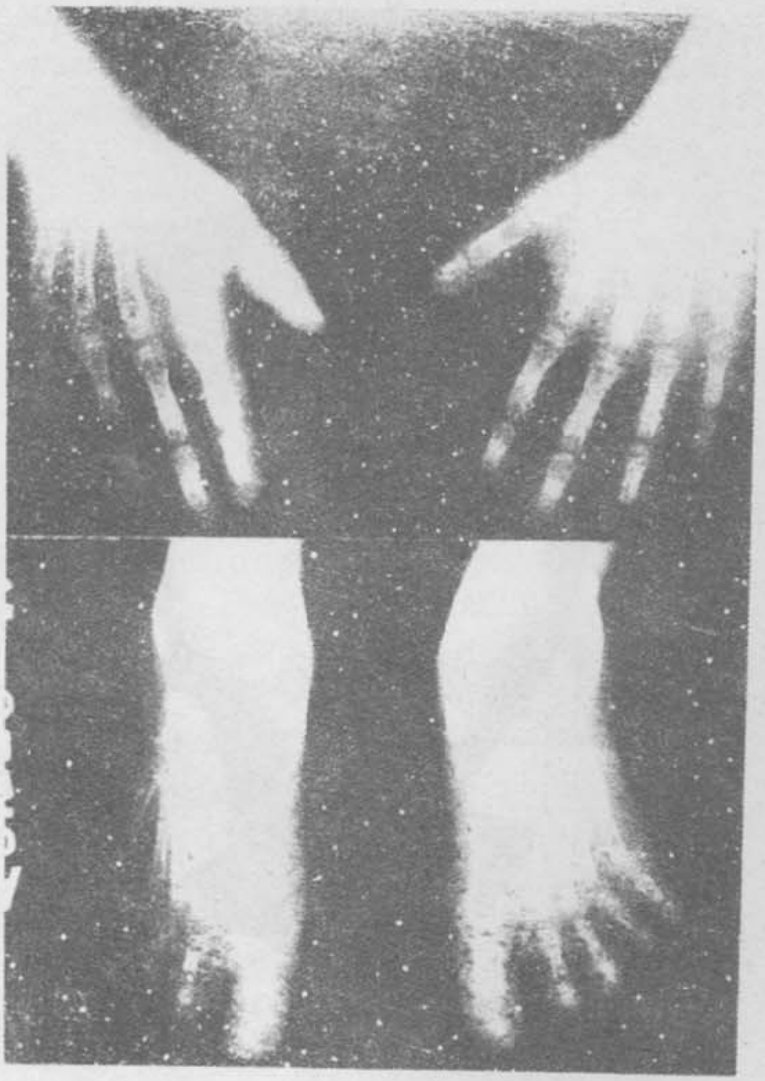

Fig. 3. Roentgenograph showing absence of distal phalanges.

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Lipoid Proteinosis (Urbach-Wiethe Syndrome) with Dwarfism

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Urbach-Wiethe syndrome also known as lipoid proteinosis or hyalinosis cutis et

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Received for publication May 7, 1990; Accepted December 4, 1990

mucosae is a rare recessively inherited disorder due to wide spread deposition of a lipoprotein complex in skin and mucous membrane(1). It has equal sex distribution and all reported patients have been of caucasian race(2). More than 200 cases have been recorded(3), but associated dwarfism has not been reported earlier. We present here a case of Urbach-Wiethe syndrome with dwarfism.

Case Report

A 10-year-old boy was admitted to the Pediatric unit of North Bengal Medical College, Darjeeling with complaints of gradually increasing hoarseness of voice since infancy along with unusual papulonodular infiltration in skin and mucous membrane. Interrogation revealed consanguinity of marriage with uneventful antenatal and postnatal periods. His milestones were within the normal range. His elder sister showed similar voice change without any skin manifestation.

Skin changes consisted of papulo-nodular lesions on the face and areas of diffuse infiltration along with hyperkeratosis and scarring on forehead, nape of the neck, mucocutaneous junction of mouth (*Fig. 1*), extensor aspects of hands, elbows and knees. The pinnae were rough, irregular and thick. There were fine papular deposits on the margin of the eyelids. The skin all over the trunk and limbs were infiltrated giving it a rough irregular appearance. Skin of the shaft of penis and scrotum was normal while the prepucial and perianal skin showed thickening and puckering.

There was characteristic eversion of lips. The tongue was firm with a cobblestone appearance and had loss of mobility due to involvement of lingual frenum.

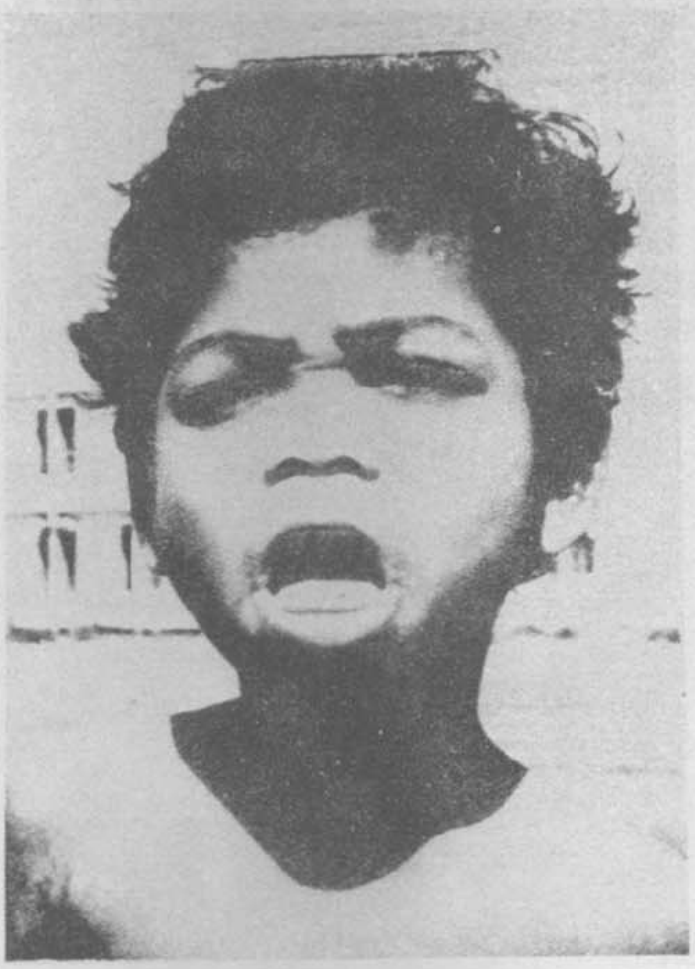

Fig. 1. Photograph showing skin lesion over forehead, angle of the mouth and anterior axillary skin fold.

There was gross restriction of movement of facial muscle, giving rise to a peculiar facial expression. The mucosa of pharynx, soft palate, pillars of tonsils and uvula showed nodular appearance. Hairs and nails were normal. The total number of teeth was 25, 12 being in the upper jaw and 13 in the lower jaw. There was hypoplasia of the upper and lower canines. All the incisors had serrated margin.

The anthropometric measurements were: weight 18 kg, height 117 cm (less than 3rd percentile), span 111 cm; and the ratio of upper segment and lower segment was 1 : 1.03. There was moderate degree of

hepatosplenomegaly. Respiratory, cardiovascular and nervous system examination revealed nothing abnormal.

On direct laryngoscopy under general anesthesia, the pharynx, larynx and vocal cords were studded with irregular papulonodular masses. Movement of vocal cords were restricted.

Laboratory studies including blood count, blood glucose and cholesterol and roent-genography of chest and skull showed no abnormality. Radiological bone age was consistent with chronological age.

Histopathological examination of skin showed deposition of eosinophillic hyaline material in the dermis mainly in relation to blood vessels and sweat glands (*Fig. 2*). Biopsy taken from laryngopharynx showed similar hyaline eosinophilic deposits around blood vessels in the subepithelial zone (*Fig. 3*). Special stains for amyloid were negative.

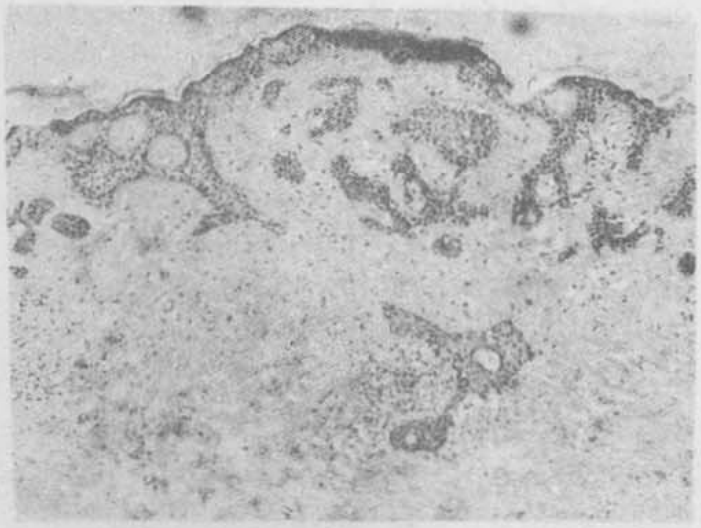

Fig. 2. Microphotograph of the skin showing deposition of eosinophilic hyaline material in the dermis mainly in relation to blood vessels (H & E × 80).

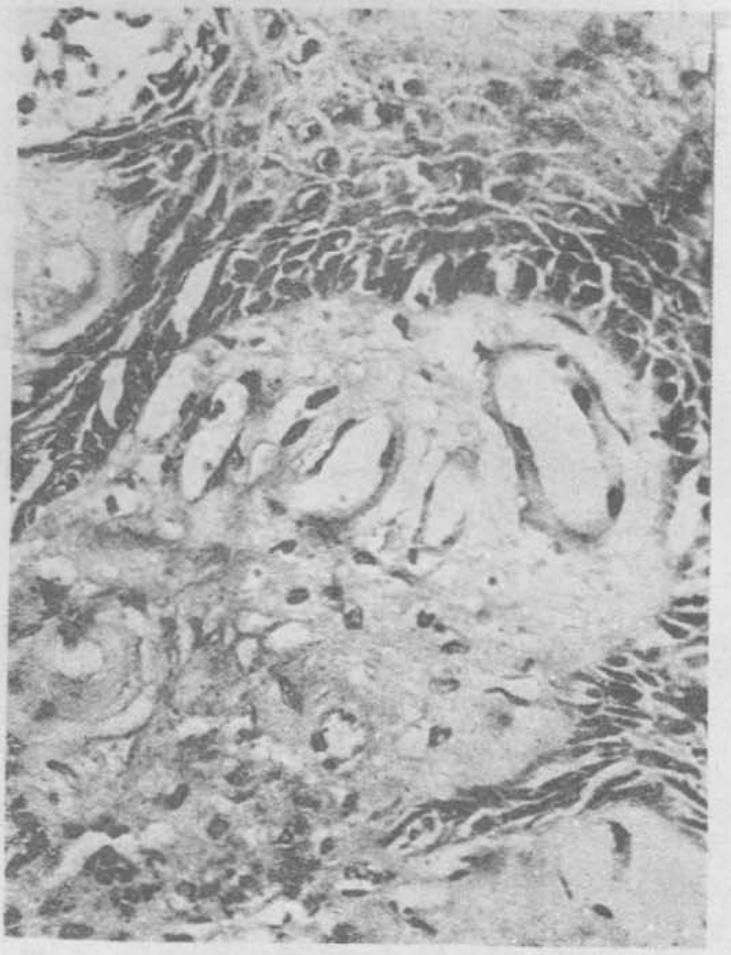

Fig. 3. Microphotograph of the laryngeal mucosa showing thickening of vascular walls with deposition of eosinophilic hyaline material in the submucosal zone (Stains for amyloid were negative) (H & E × 320).

Discussion

Lipoid proteinosis is inherited as a monogenetic autosomal recessive disorder of normal chromosome pattern(4). Geographic isolation with intermarriage brings the gene frequency to at least 0.5%(5). The condition is now recognised as a systemic disorder with generalised visceral involvement(6,7). The present case had moderate degree of hepato-splenomegaly without any gross signs of dysfunction of these two organs. The most prominent symptom is hoarseness of voice, which is present at birth or develops in early infancy as a result of involvement of the larynx and upper respiratory tract by the hyaline deposits(3,5). Lesions show a prediliction for the lips,

oral and pharyngeal mucosa, tongue, tonsils, epiglottis, vocal cord, eyelids, face, nape of the neck, hand, fingers, elbow, knee, axillae, gluteal cleft and scrotum(6).

The mucosal lesions consist of pale-pink to yellow papules and nodules which may impart a pebbly or cobblestone appearance to the surface(5). These are seen mainly in the oral, pharyngeal and laryngeal mucosa, though lesions have been reported in the esophagus, stomach, rectum, labia majora, and vagina. In our patient the skin over the scrotum and penile shaft was normal but there were distinct lesions present at the mucocutaneous junction of prepuce and anus. Ankyloglossia and loss of tongue mobility due to involvement of the lingual frenum, as seen in our case have been reported in the literature(3,5,8). The oral lesions are most often seen in the lower lip. Aplasia or hypoplasia of the upper lateral incisors has been observed and the upper cuspids and upper and lower second bicuspids may also be affected. Our patient had distinct teeth anomaly. A less regular though integral part of lipoid proteinosis is epilepsy of temporal lobe type, traceable to specific vascular changes with calcification in the hippocampus(4).

The basic defect in lipoid proteinosis is metabolic(4). There is excess formation of basement membrane like material, *i.e.*, lipo-protein complex by the genetically defective pericytes of blood vessels and fibroblasts of the skin and mucous membrane(8,9). Ultrastructural and histochemical studies have clearly documented a metabolic defect and the older concept that the disease is due to degeneration of the mesenchyme is no longer tenable.

The cause of dwarfism could not be elicited in our patient. It may possible be due to defective osteoblasts which are biologically similar to fibroblasts.

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