Treatment of Lipoid Proteinosis with Etretinate

Sir,

Lipoid proteinosis is a rare nosologic entity, also called Urbach–Wiethe disease or hyalinosis cutis et mucosae. The disease is characterised by the deposit of hyaline-like materials in the skin, mucous membranes and other tissues. The etiopathogenesis of lipoid proteinosis has not yet been clearly established, but it is regarded as an autosomal recessive genodermatosis, with variable expression. Parental consanguinity has been described rather frequently (1). The first manifestation of the disease is hoarseness present soon after birth or early in infancy, caused by intrauterine deposits of hyaline material in the vocal cords. Skin lesions such as papules, nodules and plaques appear shortly afterwards, and with time calcifications appear in the brain. Typical papules are present on the upper and lower eyelids in about 50% of the cases (2). We here report a good effect of etretinate on the skin lesions.

CASE REPORTS

Case 1

A 36-year-old woman had been suffering from hoarseness and dysphonia since birth, and from her first year of life she had had papules, vesicles and later scars. There was no family history of skin diseases but consanguinity was present. She is rather short of stature; her face is hypomimic and coarse, with scars on the forehead, cheeks and sides of the head. The eyelids presented along the margins typical white papules. On the elbows and on the knees there were verrucous plaques. There were crusted plaques and scars on the trunk (Fig. 1). The hair was sparse. The tongue was thickened and affixed to the floor of the mouth.

Indirect laringoscopy revealed thickened and uneven vocal cords. Scull X-ray showed intracranial calcifications overlying the sella turcica. A biopsy of the skin showed the characteristic features of lipoid proteinosis with orthohyperkeratosis and hyaline deposits in the dermis, which gave a strongly positive reaction with PAS.

Investigations showed normal blood count, serum protein, blood urea, liver function tests and serum lipids.

Previous therapy with dermabrasion had produced little and only temporary benefit. After a formal verbal consent had been obtained from the patient, she was treated with etretinate 1.0 mg/kg daily for 2 months and then with 0.75 mg/kg. At the follow-up after 4 months the examination revealed a substantial improvement of the verrucous lesions on the knees and elbows and of the ragads on the palms. She felt the skin to be more elastic. As the plasma level of cholesterol and triglycerids increased (6.8 mmol/l and 2.4 mmol/l, respectively) and also the transaminases (ALT 31 μ/l, AST 28 μ/l), the therapy was stopped. After 2 months the lesions relapsed.

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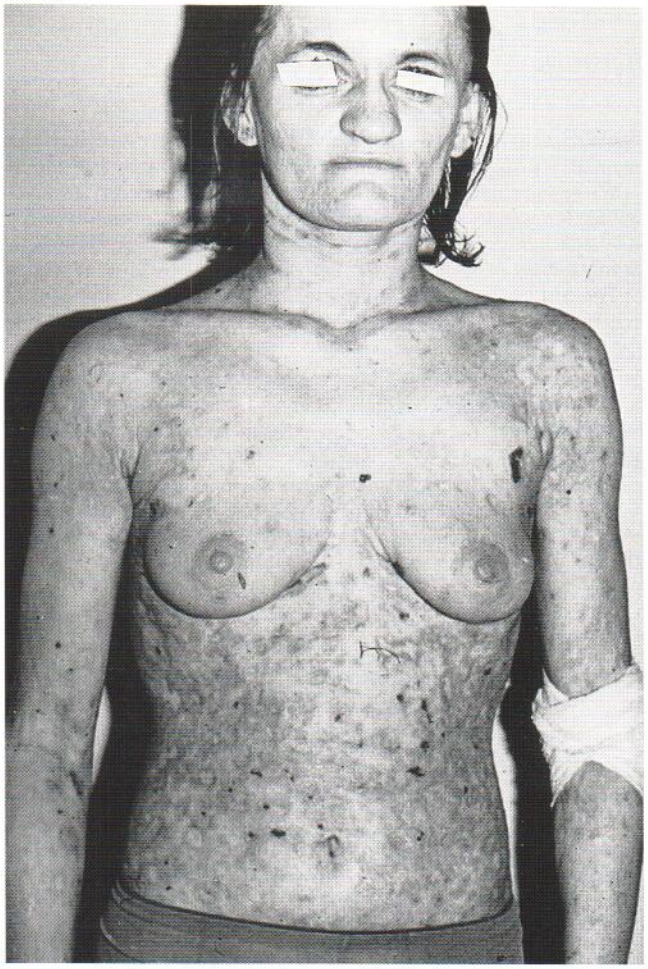

Fig. 1. Skin lesions of case No. 1. There are numerous crusted papules, plaques and atrophic scars on the trunk.

Case 2

A female aged 40 years had had skin lesions present from early infancy and hoarseness from birth. The diagnosis of lipoid proteinosis was confirmed by histology. The family history revealed parental consanguinity, and some members of the family had epilepsy. As a child the patient was diagnosed as having epilepsy, and she had been treated with phenitoin ever since then.

The skin was pale and on the margins of the eyelids were present typical waxy papules. The tongue was pale, depapillated, hard and thickened; along the lips there were whitish papules and fissures (Fig. 2). Indirect laringoscopy showed irregular, thickened vocal cords and X-ray showed calcifications overlying the sella turcica, and in both maxillar sinuses. Blood count and all biochemical analyses were normal.

The patient was treated with etretinate 1.0 mg/kg daily, for 2 months and then with 0.5 mg/kg for 4 months. The lesions on the elbows, knees and on the hands improved. She believed that her skin looked better and became more elastic. Her skin and her lips became dry, which was diminished by emollients.

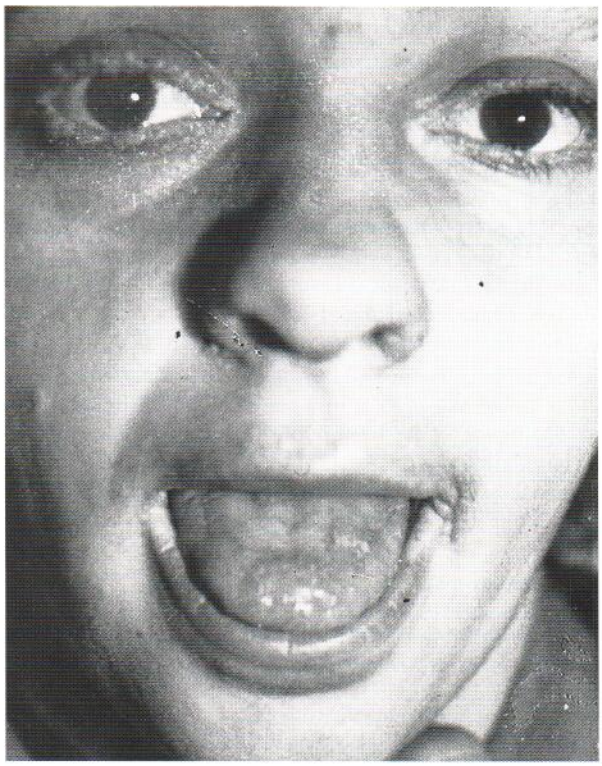

Fig. 2. Skin lesions of case No. 2. Typical small papules on the eyelids, fissures of the lips and thickened tongue.

Although these preliminary observations showed an improvement of the disease, one might not expect a complete regression of it. Nonetheless our trial clearly demonstrated that retinoids may act on the cutaneous lesions in lipoid proteinosis. Indeed, the observation that the skin manifestation responds to such a brief treatment period is encouraging, and perhaps with a longer course of therapy, a better improvement would be seen.

REFERENCES

1. Vukas A. Hyalinosis cutis et mucosae (M. Urbach–Wiethe). Lijec Vjesn 1971; 93: 1039–1044.
2. Blodi FC, Whinery RD, Hendricks CA. Lipoid proteinosis (Urbach–Wiethe) involving the lids. Trans Am Ophtalmol Soc 1960; 58: 158–166.

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