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EPIDERMOLYTIC HYPERKERATOSIS WITH MENTAL RETARDATION, BREAST HYPOPLASIA AND VENTRICULAR SEPTAL DEFECT

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Two cases of epidermolytic hyperkeratosis are described, one having a generalised variety along with mental retardation, nystagmus, unilateral breast atrophy and intermittent divergent squint, and the other having the localised variety associated with mental retardation, ventricular septal defect, sensory-neural deafness, hypoplasia of first left metacarpal and extensive dental caries.

Key words : Epidermolytic hyperkeratosis, Mental retardation, Ventricular septal defect.

Epidermolytic hyperkeratosis (EH) was the term first used by Frost and Van Scott[1] to describe an autosomal dominant trait—congenital bullous ichthyosiform erythroderma (CBIE) and its localised forms with a similar histopathology. The localised forms[2] may be limited as nevus unius lateris, or systematized as ichthyosis hystrix.[3] Klaus et al[4] described a variant of EH confined to the palms and soles. EH has also been reported in association with mental and physical retardation, and skeletal defects.[5,6] We report two cases of EH with mental retardation. In addition, the first patient had unilateral breast hypoplasia, strabismus and nystagmus, while the second patient had ventricular septal defect (VSD), nerve deafness and physical retardation.

Case Reports

Case 1

A 13-year-old Kashmiri girl was normal at birth except for nystagmus. At seven months of age, she developed erythema followed by vesiculation all over the body. On subsiding, it left behind gradually progressive verrucous lesions over the dorsa of hands, feet, forearms, legs and trunk. Pruritus and pustulation was more frequent in summers. Nystagmus remained unchanged since birth. The right breast was noted to be smaller than the left since three years. She had attained menarche six months before reporting and was having normal periods.

Cutaneous examination revealed dark brown, verrucous, linear, plaques present all over the body except the face, presternal area, upper back, and palms and soles. The lesions were longitudinally disposed over the postero-medial aspect of the extremities and were arranged transversally over the trunk. The lesions on the chest were extending on to the right breast which was atrophic and were encroaching on to the left breast (Fig. 1). The lesions were more verrucous over the flexures. Multiple pustular lesions were present in between the verrucous plaques. Oral and genital mucosae, teeth, hairs and nails were normal. Ophthalmological examination showed congenital ocular nystagmus with intermittent divergent squint. Otorhinolaryngeal examination and audiometry were normal. Average I.Q. was 58. No other family member had similar skin lesions or mental retardation.

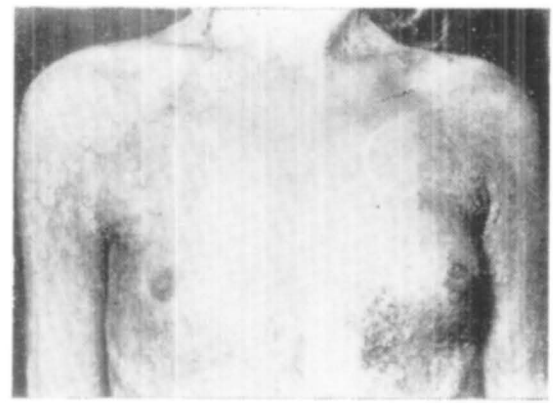

Fig. 1. Hypoplastic right breast and verrucous lesions over the chest, neck and upper arms (case 1).

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Case 2

A 7-year male child was apparently normal at birth except for skin-coloured, linear plaques over the left upper and lower extremities (Fig. 2), and a few lesions over the same side trunk. The lesions gradually became hyperpigmented and warty. There was no history of vesiculation or pustulation. No other family member had similar skin disease or mental retardation. The child used to become easily fatigued and dyspnoeic for the last three years and there was history of frequent chest infections. He was an ill-looking, physically stunted, mentally retarded child with a height of 95 cm, weight 10.4 kg and head circumference 46 cm. There was mild anemia, central cyanosis and all the fingers showed clubbing. Simian crease or epicanthic fold were not present. Cardiac examination showed evidence of ventricular septal defect with pulmonary hypertension.

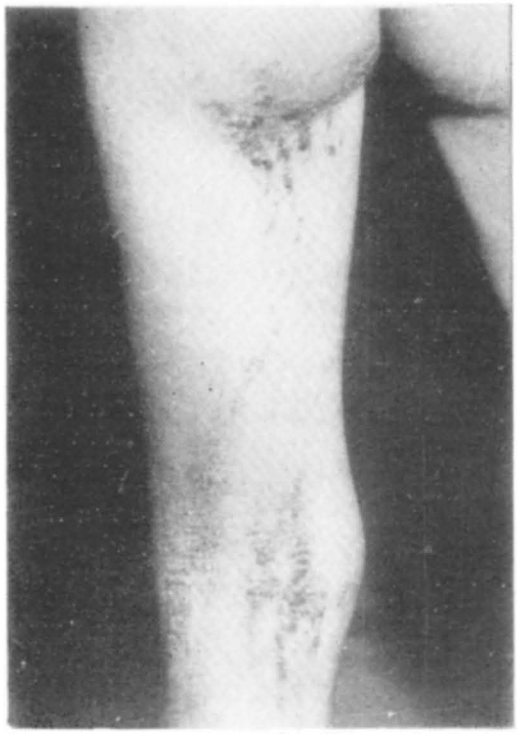

Fig. 2. Linear verrucous plaques over the back of lower extremity (case 2).

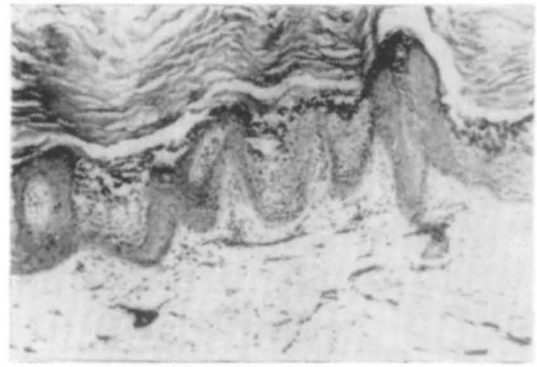

Fig. 3. Hyperkeratosis, hypergranulosis and vacuolisation of granular and prickle cell layers (Haematoxylin and eosin ×140).

Other positive findings were sensory-neural deafness, hypoplasia of the left first metacarpal and extensive dental caries.

Haemogram, urinalysis, serum biochemistry, hepatic and renal functions and skiagrams of chest were normal in both patients except for a mild anaemia and cardiomegaly in the second patient. Spine in both the cases was normal. Skin biopsy from the verrucous lesions in both the cases showed identical features consisting of compact hyperkeratosis, hypergranulosis and vacuolisation of cells with indistinct cell boundaries in the granular and prickle cell layers. There was focal lympho-mononuclear infiltrate in the upper dermis (Fig. 3).

Comments

The clinical picture of the first case was consistent with the generalised variety of epidermolytic hyperkeratosis (EH), and in the second case with nevus unius lateris or localised EH. Epidermolytic hyperkeratosis is a rare type of ichthyosis and there are no published reports of EH or its variants from India except for a report of ichthyosis hystrix in a mother and her two children, sans histopathological documentation.[7] The cases of EH reported in literature

and associated findings are summarised in table I.

Table I. Associated defects with epidermolytic hyperkeratosis.

Authors	Number of cases		Associated defects
	Described	With associated defects	
1. Barker and Sachs[5] (1953)	Reviewed 19	5	4 Mental retardation 2 Physical retardation 1 Dental caries
	Reported 2	Nil	Nil
2. Reed et al[8] (1964)	4	1	Mental and physical retardation
3. Zeligman et al[2] (1965)	5	1	Mental retardation Schizophrenia
4. Frost and Van Scott[1] (1966)	4	Nil	Nil
5. Solomon et al[6] (1968)	4	4	4 Skeletal defects of spine
6. Adam and Richards[9] (1973)	2	Nil	Nil
7. Allen[10] (1978)	3	Nil	Nil
8. Bhutani[11] (1982)	2	1	Mental retardation
9. Zachary and Wells[12] (1983)	1	Nil	Nil
10. Ayala and Donfrio[13] (1984)	1	Nil	Nil
11. Lookingbill et al[14] (1984)	1	Nil	Nil
12. Kanerva et al[15] (1984)	1	1	Undescended tests Spondylolisthesis

Mental retardation was present in seven (14.28 per cent) of forty nine cases. Dental caries was present in one out of nineteen cases reviewed by Barker and Sachs.[5] Skeletal defects confined to spine were present in all four cases described by Solomon et al,[6] but congenital metacarpal hypoplasia has not been reported with EH. Unilateral breast hypoplasia, VSD, sensorineural deafness and strabismus have also not been reported in association with EH. A case of unilateral breast hypoplasia with histopathologically documented nevus unius lateris was reported in a thirteen year old girl by Pack and Sunderland[16] and she developed carcinoma breast and died at the age of twenty three. Sensorineural deafness and strabismus have been described in epidermal nevus syndrome by Solomon et al but none of the patients had histopathology suggestive of EH.

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