EPIDERMOLYSIS BULLOSA DYSTROPHICA ET ALBO-PAPULOIDEA PASINI
(A Case Report)

RADHA RANI AGGARWAL, RAJKUMAR GARG AND ADHARSH CHOPRA

Summary

A young intelligent female suffering from Epidermolysis bullosa dystrophica et albo-papuloidea Pasini (EBDAP) is reported[1]. Clinical diagnosis was made on the basis of typical albo-papuloid efflorescences (APE) distributed predominantly on the trunk and extremities. The efflorescence was noted by her parents within few hours of her birth and bullae on pressure points appeared on 11th day. In addition, mucus membrane of soft palate was involved. Diagnosis was confirmed histopathologically. This is the second case report of this kind from India.

Epidermolysis bullosa dystrophica et albo-papuloidea is a rare hereditary disorder, first described by Pasini[1] in 1928 under the title "Dystrophy cutanea bulleuse atrophiante et albo-papuleuse". EBDAP is characterised by small flat, firm, well defined ivory-white papules, some of them perifollicular with visible wrinkling and just raised from skin surface. Whitish papules are distributed mainly in lumbo-sacral regions and rarely on extremities, nape of neck, retroauricular region and scalp. They may also be present on mucus membrane of the mouth. Rook[5] in 1968 reported that continuous light pressure over the albo-papuloid efflorescences may show blister formation. Menter and Patz[2] could see fragility of the skin in albo-papuloid lessions. Albo-papuloid efflorescence has also been seen partly along the creases of skin in bands or plaques form by Goetz et al[3]. Primary APE was clearly demarcated by Pasini from papuloid efflorescence developing after blister formation.

The other features of this entity are typical of hyperplastic form of EBD. Bullae mainly on pressure points, healing with scar formation and onychodystrophy are important associated features. Hair and teeth are usually unaffected. Occurrence of milia isolated as well as with APE is known. Congenital Polykeratosis, psychic abnormalities, Acanthosis nigricans and Porphyria may be associated in rare instances.

EBDAP is regarded as geno-dermatosis transmitted by dominant inheritance though recessive inheritance has also been mentioned by Cerutti[4]. APE usually appears first in later childhood or in adults but rarely in infancy[5] (Kumar and Kumar[6]) but in our patient, APE was present at birth.

Pathogenesis of EBDAP has not been fully explained. APE mainly on covered areas of body may represent a response to mild trauma unobserved by patient and traction on skin through body

Department of Dermatology & Venereology
Government Medical College, Patiala.

Received for publication on 14—5—1975

movements may be effective stimulus as is postulated by Menter and Patz[2]. Ishikawa and Yaoita[7] think that this disease is mainly due to disturbances of acid muco-polysaccharide metabolism. Development of blisters on APE after friction can be explained by disturbed acid muco-polysaccharide metabolism.

Not many reports are available in international medical literature and since only one case from India has been reported so far, we are reporting this case. Interesting points in our patient are appearance of APE at birth, involvement of mucus membrane, presence of milia, presence of lesions in retroauricular region and nape of neck and absence of bullae on knee.

Case Report

History

A 24 year old female, visited the Skin Outdoor with complaints of whitish papular lesions on body since birth. Whitish lesions were noticed by her parents within few hours of birth on trunk and since then they had gradually increased in number becoming generalised. In addition, patient used to get recurrent bullous lesions especially at pressure points but also on the APE after friction or mild trauma inflicted during routine life. Bullae contained usually clear fluid, at times haemorrhagic fluid and also purulent material, and they healed by scar formation. Dystrophy of nails was also present since childhood. No other member was suffering with such disease in her family.

General systemic examination revealed no abnormalities.

Local examination : The lesions were generalised but predominantly on the trunk and in lumbo sacral region, (Fig. 1 Page No. 248) around the umbilicus and lower abdomen, on the flexor and extensor aspect of extremities including hands, feet, fingers, toes, nape of neck and retroauricular region. Lesions were whitish yellow papules of variable size, round or oval in shape, discrete as well as confluent and some of them perifollicular. Lesions were just raised from skin surface and were flat and wrinkling. Skin between the lesions was normal. Similar papules were present on the soft palate. Continuous rubbing of the whitish papular lesions resulted is blister formation. In addition to this, discrete firm milia were present on dorsae of hands and extensor surfaces of forearms.

Bullae of varying sizes were present mainly on extensor surfaces of elbow as well as around the wrist and ankle joints. Discrete bullae were present on rest of the body and some of them contained serous fluid but few contained haemorrhagic and purulent material. At places there was scarring. All the finger and toe nails were dystrophic (Fig. 2 Page No. 248). Hair and teeth were normal.

Routine investigations, STS, Blood urea, total and differential serum proteins, fasting blood sugar, bleeding time, clotting time, urine for porphyrins, blood uric acid, serum Na and K were normal. X-ray skull was normal.

Histopathology

This was consistent with the diagnosis of EBDAP. Skin biopsy showed in upper cutis collagen tissue hyperplasia with homogenisation of the collagen fibres. Epidermis was thin and rete cones flattened (Fig. 3 Page No 248). Bullae at the dermoepidermal junction and upper cutis showed collagen hyperplasia.

Treatment

There is no satisfactory treatment for EBDAP. Our patient took Prednisolone 15 mgm per day for 6 months without any beneficial effect. Patient felt better in the beginning because of control of secondary infection. Treatment with Germanin (Bayer 205) has been recommended by Goetz and Meinicke[3] with improvement of both bullous lesions and APE.

REFERENCES

1. Pasini A: Dystrophie cutanee bulleuse atrophiante et albo-papuleuse. Ann Derm Syph Paris 12 : 1044, 1928.

2. Menter MA and Patz IM: Albo-papuloid variety of Pasini, the pattern of epidermolysis bullosa in Transvaal Bantu. Brit J Derm 85 : 32-36, 1971.

3. Goetz H and Meinicke K: Zur Klinik und therapie der epidermolysis bullosa et albo-papuloidea Pasini. Derm Wschr 131:481-487, 1955.

4. Cerutti P: Contributo allo studio della epidermolysis bullosa. G ital Derm 74:335, 1933.

5. Rook A Wilkinson DS and Ebling FJG: Text Book of Dermatology, Blackwell and Edinburg 2 : 1606, 1970.

6. Kumar K and Kumar R: Epidermolosis bullosa dystrophica et albo-papulodea pasini in an Indian. Dermatologica 146 : 137-143, 1973.

7. Ishikawa H and Yaoita H: Epidermolysis bullosa et albo-papuloidea (Pasini) und ihr untertypus. Jap J Derm 78 : 438, 1968.

True

The Staphylococcal Exfoliatin (S. E.) causes cleavage of the epidermis directly beneath the stratum granulosum in man. The cutaneous responses of several mammalian and nonmammalian species were examined in vitro and vivo. Human and murine skin as well as that of hamsters and monkeys exfoliated while all other species tested (rat, rabbit, guinea pig, dog, frog and chicken) were refractory. Neither dermal elements nor circulating factors interfered with or influenced sensitivity to S.E. Besides species specificity S.E. is also tissue specific. The species and tissue specificity of the staphylococcal scalded-skin syndrome may be attributable to either keratinocyte receptors specific for exfoliatin or the presence of specific, as yet undefined substances in the intercellular space.

Reference: Elias PM, Fritsch P and Mittermayer H: Staphylococcal Toxic Epiderma necrolysis: species and tissue susceptibility and resistance, J Invest Derm, 66 : 80, 1976.