EPIDERMOLYSIS BULLOSA

(Case Report)

By

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Epidermolysis bullosa is a rare heriditary condition characterised by development of vesicles and bullae, which may be spontaneous or as a result of mild trauma. The condition is believed to be first described by Goldscheider[1]. The disease is clinically divided into two main groups.

Simple or non-scarring type:—A relatively benign condition transmitted as dominant trait. The lesions may be evident at birth but usually occur during infancy; they are monolocular, varying in size, and contain clear serous fluid. On rupture lesions heal without scarring. Nikolsky's sign is positive. There is usually no involvement of mucous membrane, nails or hairs.

Dystrophic or scarring type:—The lesions are distributed all over body, including mucous membrane and occur soon after birth. Bullae may contain haemorrhagic fluid and on rupture leave marks of scarring, pigmentation or depigmentation. Nails are affected, they get curled up or fall out. Sometimes milia like cysts on dorsum of hands are present. The disease may be transmitted as dominant (when it is not severe) or as recessive trait

Variants of dystrophic type inherited either by dominant or recessive gene were reviewed by Cockayne[2] and divided into three subtypes. The first is inherited as dominant trait and does not interfere with growth and development. The lesions are severe over points liable to trauma or pressure. The finger or toe nails may be lost, the scars may contain small epidermal cysts. The second is more severe and is inherited as recessive gene. The children are under developed and few survive to adulthood. The third variety consist of miscellaneous group showing features of both epidermolysis and congenital ectodermal dysplasia.

A rare, invarilbly fatal subtype named as epidermolysis bullosa hereditaria lethis was described by Herlitz in 1335.[3] These case reports differ from usual simple or dystrophic type and may represent a distinct mutation.

There is no uniformity of views regarding etiology of the condition, It has been shown that there is decreased adherance of epidermis and dermis, but the nature has not been defined as yet. However this is not due to lack of elastic fibres. It is also suggested that the condition may be due to exaggerated irritability of the cutaneous vascular system. An upset in hyaluronidase metabolism due to deficiency of heparin like substance in serum is also postulated.

DIFFERENTIAL DIAGNOSIS

Bullous Impetigo Contagiosa: This inflammatory condition usually appears few days after birth, with bullae anywhere on body except palms and soles. Response to antibiotics is good.

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Bullous syphiloderma: Palms and soles are always involved. Lesions heal without scarring except rhagades. Other syphilitic stigmata are usually present.

Incontinentia pigmenti: Lesions are smaller in size thick walled and are resistant to compression and palpation in contrast to epidermolysis bullosa. Further they are arranged linearly and the content is thick yellow.

Ritter's disease: It starts in first few weeks of extra uterine life, lesions appear first on face, chin and then spread to whole body. There is no exfoliation and mortality is high.

Pemphigus erythematosus: Evolution of lesions is slower, mucosa is more frequently involved, extremities are spared of lesions.

Course and Prognasis: As the disease is inherited natural course perisists through out life. In simple type there is tendency to clear up by puberty and life expectancy is normal. In dystrophic type there is scarring resulting in marked deformities. The condition is usually fatal.

There is no specific treatment, means of alleviation are protection from trauma and secondary infection. Corticosteroids are reported to limit extension of lesions. Antiblotics for secondary infection should be given.

A review of lierature shows that condition is mostly present in Negros of North America. Cases have also been reported from South Africa, In India following first report by Krishnamurty,[4] cases of epidermolysis bullosa have been published by Awachat,[5] and Paul Singh et al.[6] The following five cases of epidermolysis bullosa of dystrophic type observed in two families and followed for a year are being reported.

CASE REPORTS WITH COMMENTS

The clinical findings and laboratory investigations are summarized in the table and the family tree is depicted in figure 1. Case 'S' was first brought to the

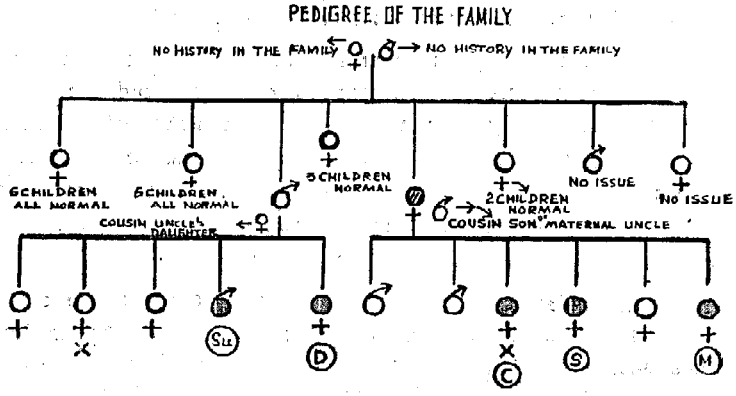

Fig. 1

hospital and subsequent questioning revealed presence of other cases in two families related to each other. Case 'C' died at the age of three years due to intercurrent infection.

Lesions were distributed all over body including mucous membrane of mouth, they were monolocular and contained serous fluid. Evolution of lesions was noticed soon after birth and slight trauma caused vesicles. There was no seasonal variation in the course of disease. Case 'Su' showed corneal opacity and staphyloma due to rupture and healing of lesions (fig. 2). Nails were distorted and in case 'S' were shed off. Lesions were preceded by the sensation of burning at the

Fig. 2
Involvement of cornea. Pigmentation and scarring of healed lesions (case Su).

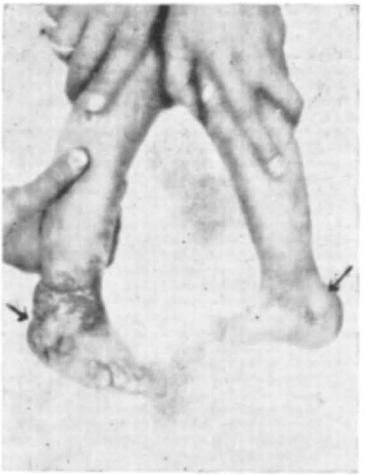

Fig. 2a
Bullae before and after rupture with pigmentation.

site. All cases showed presence of milia like cysts on the dorsum of hands and case 'M' on eyelids also. No case showed involvement of hairs and teeth. Nikolsky's sign was positive.

Urine examination did not reveal porphyria. There was no eosinophilia but leucocytosis was observed. Biopsy of lesions showed bullae at subepidermal level with cleavage between epidermis and dermis. Contents were serous with a few

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TABLE SHOWING CLINICAL FINDINGS

Patient	Age and sex	Lesions	Age at onset	Scarring and pigmentation	Nikolsky's signs	Hair	Nails	Growth	Teeth	Mucous membranes
'S'	7 Yrs. Female	Bullous lesions varying size occurs all over body. Content of lesion clear Milia-like cysts present.	2nd day after birth on hand	Present	Positive	Normal	Absent on toes	Normal	Normal	Lesion positive
'M'	2 Yrs. Female	—do—	1st day after birth on hand	Present	Positive	Normal	Curling & deformed	Normal	Normal	Positive
'Su'	4 Yrs. Male	—do—	4th day on cheek	Present	Positive	Normal	Deformed	Normal	Normal	Positive
'D'	1¼ Yrs, Female	—do—	4th day near ear	Present	Positive	Normal	Deformed	Normal	Normal	Positive
'C'	— Female	Died at the age of 3 years due to diarrhoea and fever. Manifested lesions like above								

AND LABORATORY INVESTIGATIONS

Cornea	Other systems	Urine Porphyrine	L. E. Phenomenon	V. D. R. L.	W. B. C. & D. C.	Skin Biopsy
Normal	Normal	Nil.	Negative	Negative	No eosinophilia leucocytosis.	There is cleavage between epidermis and dermis with formation of vesicle, which contains serous fluid and a few scattered red blood cells. No inflammatory reaction seen in dermis. Elastic tissue normal.
Normal	Normal	Negative	Negative	Negative	—do—	—do—
Opacity left eye	Normal	Negative	Negative	Negative	—do—	—do—
Normal	Normal	Negative	Negative	Negative	—do—	—do—

cases from 2nd day after birth.

(Contd. from page 123)

scattered red blood corpuscles. No inflammatory reaction was seen in dermis (fig. 3).

Fig. 3. Microphotograph showing a part of the vesicle with cleavage between epidermis and dermis.

These cases showed clinical characteristics intermediate between simple and severe type of dystrophic epidermolysis bullosa. They were treated with corticosteriods and antibiotics during hospitalisation.

There was no history of such lesions in parents or other members of the family. Consanguinity is noted in both the families (fig. 1). The simple type is inherited as Mendalian dominant characteristic, where as dystrophic type may be dominant or recessive. Sometimes the disease though clinically similar, may be due to three different genes and have nothing in common genetically, the inheritance is not sex related although incompletely sex related recessive gene was demonstrated in a well studied pidigree.

The absence of lesions in parents and lack of any history in childhood, indicate they may be carriers of recessive gene. Since parents of both the families are related, there is possibility of their inheriting recessive gene from same source. Therefore the present cases in homozygous state are manifesting the disease.

Observations of lesions with slight trauma or spontaneously. healing with pigmentation and scarring, involvement of mucous membrane and nails. histologic evidence of cleavage at subepidermal level fixes the diagnosis of epidermolysis bullosa of dystrophic type. Abscence of such lesions in parents but consanguinity indicate transmission of disease by recessive gene.

SUMMARY

Epidermolysis bullosa and its variants are reviewed in brief. Five cases of epidermolysis bullosa dystrophica observed in two families are reported. Mode of inheritance appears to be due to presence of recessive gene in homozygous state.

ACKNOWLEDGEMENT

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ASSOCIATION ACTIVITIES

The next Conference of our Association will be held jointly with the Association of Physicians of India from 23rd to 26th January 1964 at Patiala. Members will receive the details of accommodation, sight seeing, railway concession form etc from Dr. Chander Parkash; Professor of Clinical Medicine, Medical College, Patiala.

Papers for the joint conference are invited. Papers should be Submitted to Dr. S. C. Desai, M. D., D. V. D., 68, Marine Drive, Bombay–1, by November 15, 1963, so as to be selected by the Joint Programme Committee of the participating Associations.

Symposia for the Conference:

The Central Council at its meeting at Calcutta had suggested holding Symposia or round table discussions on the following subjects: 1. Dyschromia (Pigmentary disturbances of the skin); 2. Serum Protein in dermatologic disorders. Members wishing to participate should get in touch with Dr. S. C. Desai with a summary of their intended presentation by the end of October 1963.

Histopathology Round Table:

Interesting slides may be brought for demonstration by members. Prior information on the clinical data, Photograph of the patient, and histopathologic findings, should be sent to Dr. James C. Fernandez, Y. M. C. A. Building, Lamington Road, Bombay 4. The presenters whose slides are selected will be intimated.

The Association of Physicians have selected a subject of (1) Amyloidosis and (2) Rheumatoid Arthritis for Symposia. Any member wishing to participate should communicate with the moderator Dr. J. C. Patel, Backbay View, New Queen's Road, Bombay 4.

SHARAT C. DESAI, M. D , D. V. D.
General Secretary.