

The lability of ESR in the present case seems to have been correlated to the disease activity. Similar labile ESR results have previously been recorded in 2 out of 10000 presumably healthy blood donors (1). In these cases, the lability occurred only at room temperature and not when the tests were carried out at 4°C and 37°C.

It would be of interest when investigating patients with bullous diseases of the DH/BP/pemphigus group and other disorders with autoimmune phenomena, and particularly those with a positive Coombs test, to look further into the question of labile ESR by more routinely carrying out a multitude of ESR tests at the same time.

ACKNOWLEDGEMENT

The indirect immunofluorescence tests were kindly performed by the Institute of General and Rheumatological Immunology, Rikshospitalet, Oslo.

REFERENCES

1. Heistö, H., Godal, H. C., Grøttum, K. A., Madsen, G. & Uggerud, B.: Increased erythrocyte sedimentation rate probably due to a red cell factor. *Scand J Haematol* 6: 427, 1966.
2. International Committee for Standardization in Haematology: Recommendation for measurement of erythrocyte sedimentation rate of human blood. *Am J Clin Pathol* 68: 505, 1977.
3. Jablonska, S., Chorzelski, T. P., Beutner, E. H., Maciejowska, E. & Rzeza, G.: Dermatitis herpetiformis and bullous pemphigoid—Intermediate and mixed forms. *Arch Dermatol* 112: 45, 1976.
4. Lever, W. F.: *Histopathology of the skin*, 5th ed., Lippincott Co, Philadelphia, J. B., 1975.
5. Pehamberger, H., Konrad, K. & Holubar, K.: Circulating IgA anti-basement membrane antibodies in linear dermatitis herpetiformis: Immunofluorescence and immunoelectronmicroscopic studies. *J Invest Dermatol* 69: 490, 1977.
6. Samter, M.: *Immunological Diseases*, vol II, 3rd ed., pp. 1005–1009. Little, Brown & Company, Boston, 1978.
7. Solheim, B. G., Albrechtsen, D., Thorsby, E. & Thune, P.: Strong association between HLA-Dw3 associated B cell alloantigen and dermatitis herpetiformis. *Tissue Antigens* 10: 114, 1977.
8. Uses for immunofluorescence tests of skin and sera. *Arch dermatol* 111: 371, 1975.
9. Westergren, A.: Die Senkungsreaktion. Allgemeine klinische Ergebnisse. Praktische Bedeutung bei Tuberkulose. *Ergeb Inn Med Kinderheilkd* 577, 1924.

Congenital Localized Skin Defect and Epidermolysis Bullosa Hereditaria Letalis

Inger Skoven and K. T. Drzewiecki

Department of Dermatology, Department of Plastic Surgery and Department of Pathology, Odense University Hospital, DK-5000 Odense C, Denmark

Received April 17, 1979

Abstract. Epidermolysis bullosa hereditaria letalis (Herlitz) rarely appears with all the clinical characteristics originally described as belonging to the syndrome. Besides the blistering of the skin and mucous membranes in the oral cavity, the case presented showed dystrophic nails, congenital localized skin defects with hypoplasia of underlying structures and a rare but characteristic malformation of the foot of the affected extremity. No scar formation occurred before death at the age of 6 months. Histological examination of the blisters showed separation between the basement membrane and the cell membrane of the basal cells. In areas of skin defects, normal appearing hair follicles and sweat glands were seen. The relation of the syndrome to Bart's syndrome is discussed.

Key words: Congenital skin defect; Epidermolysis bullosa hereditaria letalis; Electron microscopy

Congenital localized absence of skin is a rare defect. In the majority of cases there is a solitary coin-sized lesion near the vertex of the scalp, usually involving the skin and subcutaneous tissue (8, 9). Occasionally, congenital skin defects are localized on the trunk or the extremities, where they tend to be more superficial, with a symmetrical distribution most frequently affecting the lower legs. This form may be associated with epidermolysis bullosa. In 1966 Bart (2) reviewed the literature on the subject and found 19 reports of congenital localized absence of skin associated with blistering of skin or mucous membranes or both. The skin defect involved the extremities in all but one patient; no patients had scalp lesions. Twelve cases presented in four of the nineteen reports were examples of the epidermolysis bullosa hereditaria letalis (Herlitz, 1935) type of epidermolysis. This disease entity was re-established in 1974 by Pearson (7) who presented the clinical and histological characteristics of five cases. However, in only four cases originally described by Herlitz has the syndrome been as-

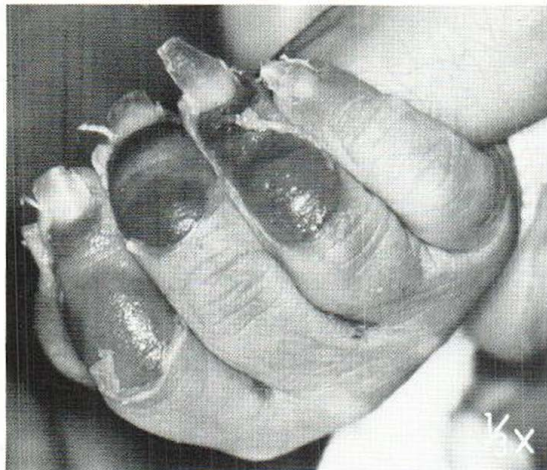


Fig. 1. Skin defects on fingers. The nails are elongated and dystrophic.

sociated with congenital hypoplasia or malformations of the structures underlying the skin defect.

The following is a report of another child with this syndrome. The relation to one of the other forms of mechano-bullous diseases, Bart's syndrome, is discussed.

CASE REPORT

A male infant born January 1977. Normal pregnancy. Birth weight 2750 grams. Delivery by vacuum extraction. No known consanguinity between the parents. One nor-

mal female sibling was born 1½ years later. Father's male cousin had shown a tendency to develop clear blisters following slight trauma between the age of 3 days and 3 months, but no other abnormalities were associated.

At birth the skin was defective in the following areas (Figs. 1, 2): the distal half of the dorsal aspects of second left finger and second, third and fourth right finger; a narrow ring around umbilicus; the lower third of right femur; the right knee; the right lower leg and foot except for a patch of normal skin on the lateral surface of the lower leg and normal plantar surface. The skin defects were sharply demarcated and covered by a red, velvety, glistening membrane beneath which the vasculature was easily seen. Hypoplasia of the right lower extremity and a 20 degree dorsal contracture in the ankle joint were evident. Nails were deformed, and most of them were shed after a few days, leaving crusted areas. On the 2nd day post partum, skin biopsies were taken and the defects were partly covered with split skin transplants. On the third day a few blisters were noticed on the buttocks. No blisters were seen in the suction area on the scalp after vacuum extraction. New erosions continued to appear secondary to mild trauma, blisters or erythema. Epithelialization progressed slowly without scar formation. Inward growth was seen from the border of the defects and the transplanted areas. Recurrent skin infections occurred despite topical and systemic antibiotic treatment. Systemic corticosteroid treatment did not affect the course. From the age of 2 months, blisters developed in the oral cavity. The patient's general growth was retarded and the weight levelled off at about 4.5 kg. Blood eosinophilia was continuously present. For approximately one week before death the blistering had become haemorrhagic, and respiration rendered difficult by acute tracheitis. Death occurred at the age of 6 months. No congenital defects in the internal organs were found at necropsy.

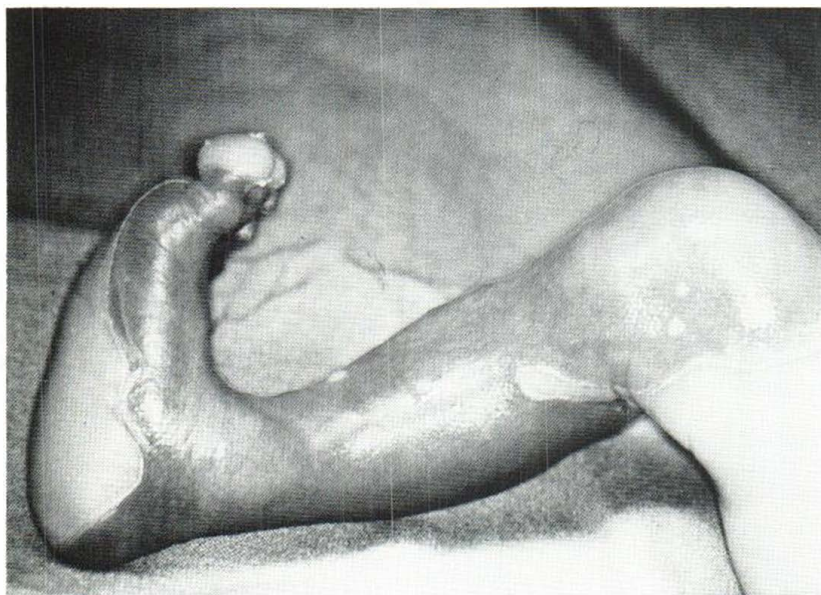


Fig. 2. Skin defect of the right lower extremity. Note dorsal flexion of the foot.



Fig. 3. Biopsy from the area of skin defect. Epidermis is absent. Dermis is covered by PAS-positive membrane. Reticular and elastic fibres are scanty. Apparently normal hair follicles are visible. Note the moderate, acute inflammatory changes in the dermis. HE, $\times 175$.

Light microscopy

Specimens taken 2 days post partum from areas of skin defects (*Fig. 3*) showed the surface to consist of a PAS-positive membrane covering the dermis and sub-cutis, with normal appendices. The collagen elastica and reticulin seemed thin and sparsely represented. Moderate early acute inflammatory changes were seen in the dermis. No signs of vesiculation between the epidermis and dermis

were seen in sections of the skin which were visibly unaffected. The collagen, elastica and reticulin appeared normal.

Specimens of the apparently normal skin taken 3 weeks later (*Fig. 4*) showed areas of clean separation above the basement membrane, the epidermis being normal in appearance. No regeneration of epidermis had occurred from the skin appendices in the areas of skin defects.



Fig. 4. Skin biopsy taken 3 weeks after birth. Separation between epidermis and dermis along dermo-epidermal junction is evident. Epidermis appears normal. HE, $\times 280$.

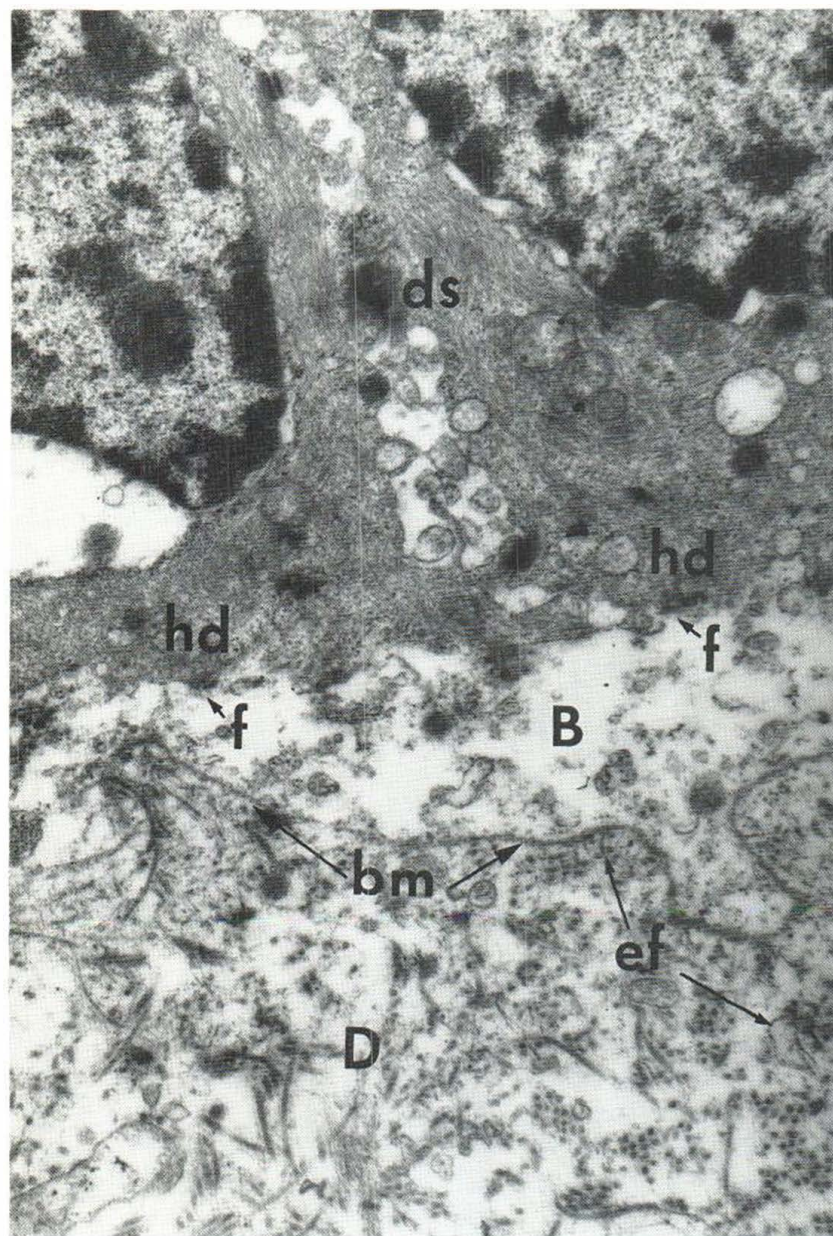


Fig. 5. TEM of the dermo-epidermal interphase. (B) blister. Hemidesmosomes (hd) are detached from the basement membrane (bm), along the extracellular fibrils (F). Desmosomes (ds) make a firm contact between the basal epidermocytes. Elastic fibres (ef) are scanty. Slight swelling of the dermis (D) is present. TEM, $\times 19\,170$.

Electron microscopy

Examination of the blisters showed separation occurring between the basement membrane and the cell membrane of the basal epidermocytes (Fig. 5). The intact basement membrane remained attached to the dermis. Elastic fibres were scanty and slight swelling of the dermis could be noticed. In some sections vesiculation between basal cells of epidermis was present, though no vesiculation was seen between the cells higher up in the epidermis. Hemidesmosomes were normal, with normal appearing fibrils radiat-

ing into the lumen of the blister. The desmosomes were unaffected.

DISCUSSION

The case presented shows the clinical and histological picture of epidermolysis bullosa hereditaria letalis. Except for the unilateral manifestation of the defects on the lower extremities,

this case is identical with four cases described by Herlitz in 1935 (5). Herlitz also noted that the feet and, in two cases, the toes underlying the skin defects were abnormally small and the feet turned upward in a remarkable way. In our case the right fifth toe, devoid of epithelium, was rudimentary at birth and the right foot was turned upwards 20 degrees at the ankle joint. The skin defect extended further up the leg in our case, and subsequently it seems there was hypoplasia of the foot, lower leg and thigh. This was verified by X-ray examination, demonstrating the absence of the right nucleus of Béclard and proximally decreasing hypoplasia of the osseous structures. As in Herlitz's description the syndrome seems to be inherited autosomal recessively, but mutation cannot be excluded. The syndrome shows some variability in disease manifestation, but the disease spectrum does not seem wide enough as to include the transitory tendency of blistering displayed by the father's cousin in our case report (3, 5, 6, 7).

Current classification of epidermolysis bullosa separates the scarring from the non-scarring forms. In 1966 Bart et al. (2) recognized a new non-scarring form. Bart's syndrome was found in a large kinship with 25 affected members. A similar family from the Faroe Islands was reported by Joensen (4) in 1973, and furthermore two single cases have been reported (1, 10). Bart's syndrome has many clinical characteristics in common with the syndrome here described and histopathological findings are identical. Four patients in Bart's kinship even showed similar malformations of the feet affected with congenital skin defects. Bart's syndrome differs from Herlitz's syndrome by the clearly dominant mode of inheritance, the greater variability in disease expression and by the far more favourable prognosis. However, when isolated cases occur, differentiation between the two syndromes at birth may be impossible and the diagnosis must depend upon the course.

REFERENCES

1. Bart, B. J.: Epidermolysis bullosa and congenital localized absence of skin. *Arch Dermatol* 101: 78, 1970.
2. Bart, B. J., Gorlin, R. J., Anderson, V. E. et al.: Congenital localized absence of skin and associated abnormalities resembling epidermolysis bullosa. *Arch Dermatol* 93: 296, 1966.
3. Cross, H. E., Wells, R. S. & Esterly, J. R.: Inheritance in epidermolysis bullosa letalis. *J Med Genet* 5: 189, 1968.

4. Joensen, H. D.: Epidermolysis bullosa dystrophica dominans in two families in the Faroe Islands. *Acta Dermatovener (Stockholm)* 53: 53, 1973.
5. Herlitz, G.: Kongenitaler, nicht syphilitischer pemphigus. *Acta Paediatr* 17: 315, 1935.
6. Klunker, W.: Zur nosologischen Stellung der Epidermolysis Bullosa Hereditaria Herlitz (mit Kasuistik). *Arch Klin Exp Dermatol* 216: 74, 1963.
7. Pearson, R. W., Potter, B. & Strauss, F.: Epidermolysis bullosa hereditaria letalis. *Arch Dermatol* 109: 349, 1974.
8. Pers, M.: Congenital absence of skin: Pathogenesis and relation to ring-constriction. *Acta Chir Scand* 126: 388, 1963.
9. Rauschkolb, R. R. & Enriquez, S. I.: Aplasia cutis congenita. *Arch Dermatol* 86: 54, 1962.
10. Smith, S. Z. & Cram, D. L.: A mechanobullous disease of the newborn. Bart's syndrome. *Arch Dermatol* 114: 81, 1978.

Pustulosis Palmaris et Plantaris: The Value of Routine Radiographic Examinations in the Detection of an Infectious Focus

Ove Jensen

*Department of Dermatology, The Finsen Institute,
DK-2100 Copenhagen Ø, Denmark*

Received May 25, 1979

Abstract. A three-year clinical material of 163 patients with pustulosis palmaris et plantaris is presented. The study was designed to assess the value of routine radiographic examinations of teeth, nasal sinuses and chest in the detection of an asymptomatic infectious focus. 66 patients underwent 151 examinations. Infectious lesions were found in the lungs in 0 of 53 examinations, inflammation in the nasal sinuses in 3 of 52 examinations (6%), and abscesses at the teeth roots in 8 of 46 examinations (17%). The 3 patients with sinusitis had symptoms leading to the diagnosis. The root abscesses were all asymptomatic. Elimination of the infectious foci did not affect the course of the disease. The high incidence of asymptomatic tooth root abscesses is discussed. From studies on clinically healthy persons, similar results have been reported. It is concluded that routine radiographic examinations are of little or no value, possibly with the exception of the dental examination.

Key words: Pustulosis palmaris et plantaris; A three-year clinical material; Focal infection; Routine radiographic examination

Putulosis palmaris et plantaris (PPP) is a chronic pustulous disease of uncertain etiology, affecting