The Turkish Journal of Pediatrics 30:187 - 191, 1988

A CASE OF RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA*

*Kenan Kocabay MD***, Hüseyin Güvenç MD**, Ayşenur Ökten MD**
Sırrı Bektaş MD****

Key words: dystrophic recessive epidermolysis bullosa, serum zinc, bone-age retardation

Recessive dystrophic epidermolysis bullosa is a rare inherited disease characterized by blister formation in the upper dermis at the sites of trauma. Deformities of the hands and feet occur with the formation of mitten-like enclosures of scarred skin that result in the fusion of the digits[1,2]. We present a case of recessive dystrophic epidermolysis bullosa that had mitten-like deformities accompanied by two additional findings; a high serum zinc level and bone-age retardation.

Case Report

A 7½-year-old boy was admitted to the hospital because of denuded areas on the hands and feet. Shortly after birth, he developed blisters on the elbows, hands, trunk and in the mouth. Later blisters formed on his hands and feet but only after being exposed to trauma. His history disclosed that he was a low-birth-weight infant whose third sister had died of a similar skin lesion when she was twenty days-old. He was the result of a consanguinous marriage between two healthy first cousins. He began to speak at three years of age. His mother was 28 years of age and his father was 29 years of age.

Physical examination revealed an afebrile, growth retarded child weighing 14 kg (< third percentile) whose height was 100 cm (< third percentile). He had ulcerations of the mouth, lips, nose, and face and could not open his mouth because of infected ulcerations on his lips. The child had annular, red maculopapular lesions three cm in diameter on the auricle of his left ear. His fingers and toes were fused and contracted because of cicatrices which formed mitten-like deformities on his hands and feet. Several scars left by healing blisters could be seen on the extremities (Figs. 1,2).

* From the Department of Pediatrics, Fırat University Faculty of Medicine, Elazığ.
** Assistant Professor of Pediatrics, Fırat University Faculty of Medicine.
*** Professor of Pediatrics, Fırat University Faculty of Medicine.

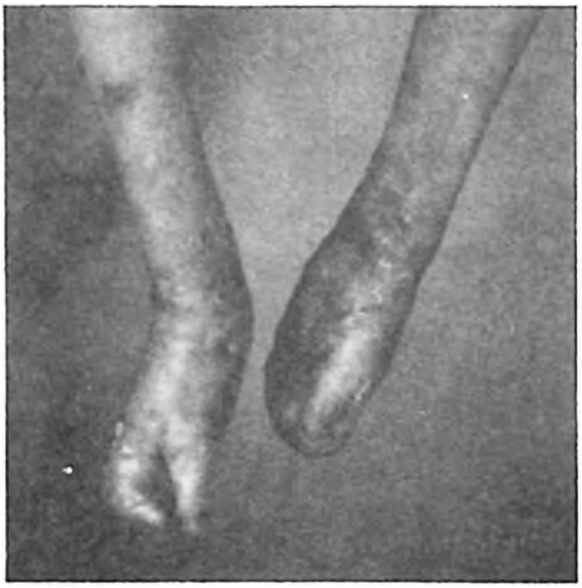

Fig. 1: Contracted and fused fingers.

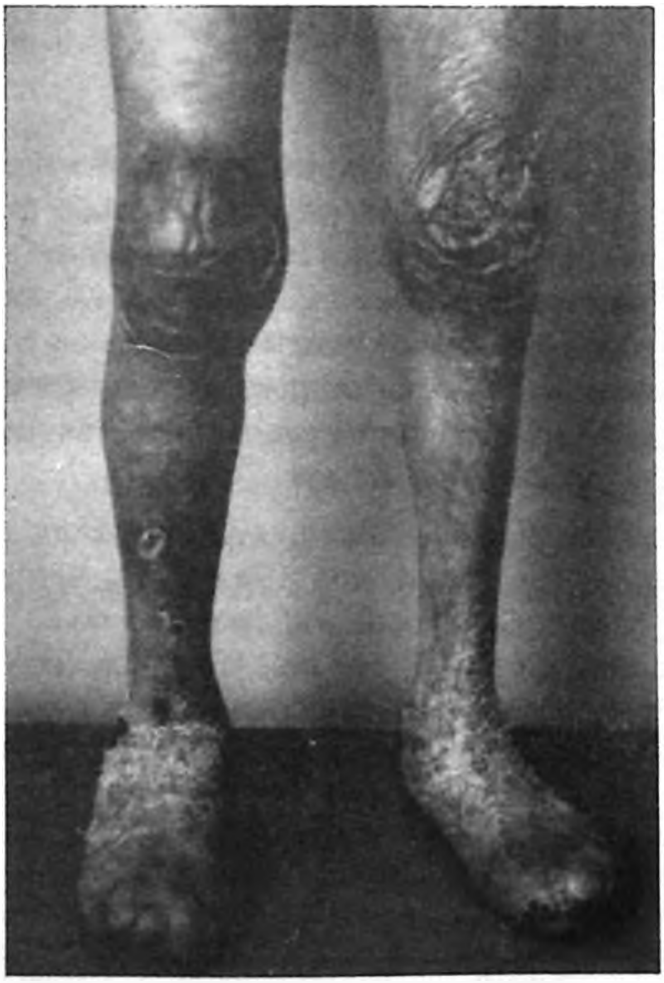

Fig. 2: Contracted and fused toes.

Laboratory studies revealed hemoglobin 8.9 g/dl, with a normal differential, peripheral blood smear with normocytic and hypochromic erythrocytes, urinalysis normal. Serum zinc was 133 microgram/dl (normal < 110 mg/dl according to the Perkin Elmer model 370 atomic absorption method). A roentgenogram of the wrist bone showed it to be seriously osteoporotic and bone age was that of a two-year-old boy. Roentgenograms of the upper gastrointestinal tract were normal. A skin biopsy by light microscopy was characteristic of epidermolysis bullosa.

Discussion

Epidermolysis bullosa herediteria is a rare group of inherited mechanobullous disorders characterized by blistering induced by minimal trauma. The classification of mechanobullous diseases into various subtypes is possible on clinical, genetic, biochemical and histopathological grounds. According to histopathological features, there are three distinct forms: epidermal, junctional, and dermal. Dystrophic epidermolysis bullosa is inherited by either an autosomal dominant or recessive trait. Blister formation is located in the upper dermis. The recessive form, occurring during the first twenty years of life, is usually the most severe of the mechanobullous disorders resulting in severe mutilation, debility and early death in many instances[1,2].

The onset of blister formation and erosion is usually present at birth or shortly thereafter. The entire skin surface and the mucous membranes are susceptible. The erosions heal very slowly with fine parchment-like scarring. Deformities of the hands and feet occur with the formation of mitten-like enclosures of scarred skin that bind the digits together. Contractures are common. The erosion of the oral mucous membrane and esophagus may result in a bound-down tongue and severe esophageal stricture. In addition to the skin and mucous membrane lesions, growth retardation, anemia and life-threatening infections often occur[3-7].

The anchoring fibrils are reduced in the lesioned skin. It has been found that there is an increase in human collagenase activity in the blister fluid. This may be a primary event explaining blister formation in the upper dermis[1,8].

This case was considered an example of recessive dystrophic epidermolysis bullosa because of the onset of the disease at birth, its chronic course, mitten-like scarring and secondary skin infections. The differential diagnosis between recessive dystrophic epidermolysis bullosa and the other forms of the illness was arrived at by clinical findings, histological features, and inheritance patterns.

There is no definitive therapy for recessive epidermolysis bullosa. The most valuable measures are preventive. Avoidance of trauma to the skin is essential. The availability of excellent antibiotics makes it possible to control secondary

infections which used to be fatal. Surgical release of scarred adhesions and mitten-like deformities may prolong life in severe forms of the illness. Some investigators have reported that the use of high doses of vitamin E, cortisone, phenytoin and pipamperone resulted in an improvement in patients with dystrophic epidermolysis bullosa[1-2, 9-11]. An antibiotic and vitamin E were begun in this case. Ampicillin was given in a daily oral dosage of 150 mg/kg body weight for one week and vitamin E, 70 mg b.i.d for ten days. On the tenth day of therapy secondary infections on the ulcerations of the extremities and face improved. Surgical release of the mitten-like deformities will be performed in the future.

This case, with typical mitten-like deformities, had two unusual additional findings: a high serum zinc level and bone-age retardation. Other causes of bone retardation were excluded. Bone retardation occurred in this patient because of impaired growth and development due to nutritional deprivation. It is of interest that the serum zinc was high and not low, as expected. In order to explain this finding, more research into this disorder is necessary.

Summary

A case of recessive dystrophic epidermolysis bullosa with mitten-like deformities accompanied by additional findings of a high serum zinc level and bone-age retardation is presented and the literature is reviewed.

REFERENCES

1. Domonkos AN, Arnold HL, Odom RB. Epidermolysis bullosa. In Andrew GC (ed). *Diseases of the Skin: Clinical Dermatology.* Philadelphia: WC Saunders Co, 1982, p 699.
2. Esterly NB. The vesiculobullous disorders. In Vaughan VC III, Mckay RJ Jr, Behrman RE (eds). *Nelson's Textbook of Pediatrics.* Philadelphia: WB Saunders Co, 1987, p 1402.
3. Gence E, Kotogyan AL, Ergenel A, Tümay G, Cenani A. Epidermolysis bullosa herediteria. *XVI. Türk Pediatri Kongresi, Tebliğleri Cilt II.* İstanbul 18-22 Temmuz 1977, p 207.
4. Wojnarowska FT, Eady RA, Wells RS. Dystrophic epidormolysis bullosa presenting with congenital localized absence of skin: report of four cases: *Br J Dermatol* 108:477, 1983.
5. Kero M. Occurrence of epidermolysis bullosa in Finland: *Acta Derm Venereol* (Stockh) 64:57, 1984.
6. Winship I. Epidermolysis bullosa in South Africa. *S Afr Med J* 69:743, 1986.
7. Eklöf O, Parkkulainen K. Epidermolysis bullosa dystrophica with urinary tract involvement. *J Pediatr Surg* 19:215, 1984.
8. Kero M, Palotie A, Peltonen L. Collagen metabolism in two rare forms of epidermolysis bullosa: *Br J Dermatol* 110:177, 1984.
9. Bauer EA, Cooper TW, Tucker DR, Esterly NB. Phenytoin therapy of recessive dystrophic epidermolysis bullosa. Clinical trial and proposed mechanism of action on collagenase. *N Engl J Med* 303:776, 1980.

10. Bonnetblanc JM, Bouquier JJ. Response to pipamperone in case of epidermolysis bullosa herpetiformis [letter]. *Lancet* 1:1327, 1986.

11. Cooper TW, Bauer EA. Therapeutic efficacy of phenytoin in recessive dystrophic epidermolysis. A comparison of short-and long-term treatment. *Arch Dermatol* 120:490, 1984.