

Neonate with Severe Complications of Epidermolysis Bullosa and Bilateral Clubfoot: An Unusual Case Presentation and Treatment

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Abstract

Epidermolysis bullosa is a term for a heterogeneous group of rare disorders, characterized by extensive blistering of the skin and mucous membranes as the most prominent features. So far association of the disease with extra-cutaneous manifestations and complications has been pointed out. We present an unusual case of a neonate with severe complications of epidermolysis bullosa, associated with bilateral clubfoot.

Key words: epidermolysis bullosa, clubfoot, newborns

Introduction

Epidermolysis bullosa (EB) is a term for a heterogeneous group of rare disorders, characterized by extensive blistering of the skin and mucous membranes^{1,2,3}. More than 30 subtypes of diseases have been classified based on the pattern of genetic inheritance, genetic mutation involved and morphology/topography of lesions⁴. All clinically distinctive phenotypes have skin manifestations as the major features, however, involvement of other systems that may severely affect the health of the patients are not unusual. EB may be associated with: muscular dystrophy, cardiomyopathy, pyloric atresion, urologic abnormalities, oral and eye manifestations, nail dystrophy, alopecia or tracheal epithelial erosion^{5,6,7}.

Clubfoot is one of the most common congenital birth defects, characterized by a complex three-dimensional deformity of foot^{8,9,10,11}. The cause of clubfoot is unknown and various theories have been proposed^{9,11}. Most commonly, clubfoot is an idiopathic isolated birth defect of the musculoskeletal system, however, in approximately 20% of the cases it can be associated with congenital anomalies such as: spina bifida, arthrogryposis, cerebral palsy, myotonic dystrophy, myelomeningocele, amniotic band sequence, or trisomy 18^{9,10,11}.

We are reporting a case of a long-time hospitalised newborn with severe complications of EB, associated with bilateral clubfoot.

The Case

A newborn female, born at full-term, was hospitalized to NICU due to extensive dermal-mucous lesions. She was the sixth child of a non consanguineous couple who denied the presence of any

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hereditary or congenital abnormalities in the family. On admission, the baby was afebrile, normotonic, normoreflexive and had vital functions within normal limits. Dermatological examinations revealed generalized bullous lesions filled with a thick and grey secret, wide-spread skin loss in both anterior crural regions, bleeding ulcerations around major joints and dystrophic nails [Fig 1 & 2]; multiple erosions and bulla on the

oral and nasal mucosa. Systemic examination including respiratory, cardiovascular, gastrointestinal and neurological systems revealed no pathological changes, whereas the orthopaedist confirmed the presence of bilateral clubfoot. Laboratory investigations showed leucopenia, elevated C-reactive protein, presence of streptococcus pneumoniae on the secret, and cleavage of the dermis on histological analysis of the lesional skin.



Fig 1: Photograph showing generalized bullous lesions, skin loss, bleeding ulcerations and dystrophic nails.



Fig 2: Photograph showing extensive cutaneous lesions and clubfoot

Oral antibiotics (*erythromycin*, *cephalexin*) and rectal analgesics were given. Regular cleaning of the wounds and frequent bandaging with *vaseline* gauze

was done. Baby was being fed with breast milk through a syringe.

First month: Bottle feeding was commenced. Therapy improved wound closure, but blisters continued to develop [Fig 3]. Laboratory investigations revealed: hypoalbuminemia; anemia; positive PCR; presence of *providencia*, *citrobacter* and *pseudomonas* on the bulla fluid culture; sterile hemoculture and elevated IgG levels for rubella on both mother and baby. Bilateral bronchopneumonia was detected in chest x-ray.



Fig 3: Photograph showing improvement of wound healing after one month of treatment. Fresh bulla, nail dystrophy and numerous crusts on the skin present.



Fig 4: Photograph showing loss of fingernails and fresh bulla after two months of treatment.

Treatment with *fresh frozen plasma*, *concentrated erythrocytes* and *human albumins* was initiated. Oral antibiotics were continued, *antimycotic (nystatin sol.)* was administered preventively. Topical antibiotic and an epithelisation ointment were applied locally.

Second month: Laboratory results were within normal limits. *Candida albicans* was grown on mouth and umbilical region. Fresh bulla were present on skin, there was a loss of finger and toenails [Fig 4].

Symptomatic treatment continued and the baby was discharged. She was scheduled for clubfoot treatment at an orthopaedist.

Discussion

Although new knowledge about the mutations of genes that encode structural proteins responsible for adhesion between cutaneous structures allows precise classification of patients in subcategories of EB^{12,13}, in our case, we could not classify the type of EB and explain the possible connection with clubfoot due to limited resources.

The diagnosis and classification of EB is made based on: clinical symptomatology, histopathology, electron microscopy, mutation analysis and immunofluorescence mapping.

However, in a resource limited setting, the diagnosis is mainly clinical^{14,15}. Medical surveillance for involvements of other systems should be a part of the routine evaluation. In children, complications such as: cutaneous losses, infections, anemia, respiratory failure, dehydration or digestive problems lead to impaired nutrition and growth and in some cases even death^{4,12,16}.

Despite the progress on the knowledge about the cause of EB, currently, there is no satisfactory treatment available and the long-term prognosis of the disease is poor. Gene, protein replacement and cell-based therapies are being investigated, but these approaches are at the early stages of investigation, and there are a number of uncertainties surrounding them^{13,17}. Treatment is primarily supportive and preventive consisting of: wound healing, infection control, pain management, nutritional support, adequate dressing and other therapies depending on the complications that arise^{4,12,17}.

Our patient suffered from severe complications, however, after intensive care treatment, general condition improved. Clubfoot treatment was scheduled after healing of skin wounds, whereas parents were informed about the delicacy of the disease and instructed about bathing, skin treatment and continuous regular follow-up visits.

Conclusion

We presented an unusual clinical case of neonatal EB associated with congenital clubfoot and other complications that needed long-time hospitalization and treatment. Due to our limited resources, we could not explain the possible connection. Our case serves as a reminder for scientist that more research on neonatal EB is critical for better quality of life and increased life expectancy of these patients.

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