
INTRODUCTION

A male infant was born by spontaneous delivery on February 7, 2020, with a gestational age of 40 weeks and a birth weight of 4.1 kg. After birth, the infant presented with appearance of skin loss on the bilateral lower limbs, feet, left wrist, face, and lips. Large areas of skin defects, erosion, and exudation were noted on the extensor side of the bilateral lower limbs and feet, and some skin loss with a small amount of exudation was observed on the left wrist, face, and lips, which was accompanied by dorsal hyperextension of the right foot and oral mucosal ulceration (Figure 1).

Because the parents refused invasive examinations (skin biopsy, i.e., transmission electron microscopy and immunofluorescence examination (1)) and the child was hospitalized in a period during which the strictest prevention and control measures for novel coronavirus pneumonia were enacted, the hospital canceled the invasive examinations; therefore, skin biopsy was not performed.

The infant’s parents were healthy and nonconsanguineous. They reported that neither of them had skin defects at birth. They also denied nail dystrophy or complete absence of the nail and a history of recurrent oral herpes or ulcers, and no other family members had such symptoms. The mother had multiple scheduled prenatal examinations during the pregnancy, and the sick infant delivered via natural birth was her first child. She did not have a history of previous miscarriage and underwent a thyroid function test and ultrasound B-mode examination in the second trimester suggested bilateral renal sinus separation and excessive dorsiflexion of both feet of the fetus. A nuchal translucency (NT) scan, a noninvasive prenatal DNA test, and an oral glucose tolerance test (OGTT) showed no significant abnormalities.

Ultrasound B-mode findings indicated that the infant had congenital dysplasia, suggesting that he may have a genetic disease. In a subsequent genetic test, compound heterozygous variations of c.C2005T (the nucleotide at position 2005 in the coding region was mutated from C to T) and c.G7922A (the nucleotide at position 7922 in the coding region was mutated from G to A) were detected in the child’s collagen type VII alpha 1 chain (COL7A1) gene, and the mutations were from the child’s parents’ genes (Table 1). The COL7A1 gene is a well-established causative gene for autosomal recessive dystrophic epidermolysis bullosa. Based on these results, COL7A1 gene mutations may have been the cause of the disease in the child; thus, the child was definitively diagnosed with autosomal recessive dystrophic epidermolysis bullosa.

CASE REPORT

After admission, the child received aggressive nutritional support. For treatment, cefmetazole was given for anti-infection, aseptic dressings were applied on the body surface with skin defects, iodophor disinfection was carried out, recombinant human epidermal growth factor gel and chlortetracycline eye ointment were applied externally, petrolatum was used to cover the skin defects, sterile gauze was used to wrap the lesions, and the dressings were changed.

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<th>Gene</th>
<th>Exon</th>
<th>Nucleotide change</th>
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daily or every other day. The wounds were kept dry, prolonged compression was avoided, and secondary bacterial infection was actively prevented and treated symptomatically as necessary. At discharge, the child’s vital signs were stable, some epidermal defects were visible on the extensor side of the bilateral lower limbs, feet, and left wrist as well as on the face and lips with reduced exudation, and fresh epidermal coverage was observed (Figure 2).

DISCUSSION

Congenital epidermolysis bullosa must be differentiated from other diseases such as staphylococcal scalded skin syndrome (SSSS), neonatal impetigo, congenital bullous ichthyosiform erythroderma, congenital syphilis, and neonatal herpes simplex. Among these diseases, SSSS is a severe acute generalized exfoliative pustulosis that occurs in neonates and is characterized by the development of flaccid scalded bullae and large areas of skin exfoliation due to generalized erythema throughout the body (2). SSSS mostly occurs with sudden onset 1-5 weeks after birth. Initially, erythema occurs around the mouth or eyelids and then rapidly spreads to the trunk and proximal extremities or even to the entire body, which usually heals after 7-14 days. SSSS is a blistering and desquamative skin disease caused by the exfoliative toxins of staphylococcus aureus. It is a toxin-mediated condition (3), so the blisters and erosions are usually sterile. In this case, the child had three consecutive negative common bacterial culture test results during hospitalization, enabling exclusion of SSSS. Neonatal impetigo, congenital bullous ichthyosiform erythroderma, congenital syphilis, and neonatal herpes simplex all have associated specific pathogenic infections or are accompanied by other typical clinical manifestations,

Figure 1. After birth, the patient had hyperextension of the right lower limb, skin damage with a small amount of exudation on the bilateral lower limbs, and oral mucosal damage.

Figure 2. When the child was discharged one week after birth, partial epidermal defects were visible on the extensor side of the bilateral lower limbs and feet, but the amount of exudation had decreased, and fresh epidermal coverage was observed.
but in this case the child had no obvious infection manifestations except for specific skin lesions, allowing exclusion of the above diseases.

Autosomal recessive dystrophic epidermolysis bullosa was first reported in 1966 by Bart et al. (4) and was confirmed to be caused by mutations in COL7A1 by Cristiano et al. in 1996 (5,6). The disease takes the form of dystrophic epidermolysis bullosa (7), and patients have congenital local skin defects, mucocutaneous blisters, and nail abnormalities (8). This disease is mostly sporadic, but familial predisposition has also been reported.

In this case, the defective skin had begun to heal without complications at discharge. Based on our experience, nutritional support and infection prevention should be prioritized. The child was isolated from other patients during hospitalization, his blankets and clothes were autoclaved, and strict aseptic practices were carried out (9). Dressings were changed as needed by a designated person, and secretions were managed in a timely manner. The wounds were protected, and the child was carefully monitored and supported to improve his immunity and protect the function of his organs.

This case once again demonstrates the crucial importance of prenatal diagnosis, genetic counseling, and genetic testing, which are effective measures to prevent the birth of children with genetic diseases, and early intervention can minimize the pain of the family.

References: