CASE REPORT

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Epidermolysis bullosa in a twins infant: a rare case

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ABSTRACT

Background: Epidermolysis bullosa (EB) is a rare hereditary genodermatosis characterized by blisters due to trauma and temperature. Cases of EB in twin infants are rare. This report will discuss EB in twin infants to improve our knowledge about this genodermatosis.

Case: A baby boy with a twins history aged 4 months was consulted by the Pediatric Department with complaints of fluid-filled blisters that have been present since birth. During examination, bullae appeared on the upper and lower extremities. The gram examination and culture results showed Staphylococcus aureus infection and gentamicin sensitivity. The histopathology results showed a subepidermal blister with the dermis layer showed lymphocytic infiltration, which was in accordance with EB. The baby was hospitalized for 5 days and then came back to the outpatient unit with his twin, who had the same complaint. Examination of the second infant revealed multiple erosions and hypopigmented macules on the superior and inferior extremities. Both babies were born at term, normal, adequate weight, and are the first twins. Direct immunofluorescence did not show immunoglobulin G (IgG) and complement C3 deposits in the basement membrane zone. Both infants received symptomatic therapy.

Conclusion: Epidermolysis bullosa is a rare case, especially in twins. Electron microscopy is a gold standard for determining EB type. Symptomatic treatment is the main therapy in this population of EB.

Keywords: Epidermolysis bullosa, genodermatosis, pediatric dermatology, twins infant.
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INTRODUCTION

Epidermolysis bullosa (EB) is a rare genodermatosis disease, especially in twins. Epidermolysis bullosa is characterized by skin conditions vulnerable to the skin after trauma.[1,2] Epidermolysis bullosa is currently included in an incurable genetic inherited disease.[3] Cases of EB in infants are known to be rare. However, there have been lethal cases of EB in twins that occurred in Korea.[4] Classification of EB is divided into four parts that is simple EB (EBS), junctional EB (EBJ), dystrophic EB (DEB), and mixed EB (Kindle syndrome).[2] Knowledge of bullous disease is still challenging because of its rare cases and limited diagnostic tools. In this paper, we report a case of EB in infant twins.

CASE DESCRIPTION

A 4-month-old baby boy with a history of having a twin was consulted by the pediatric health department with complaints of fluid-filled blisters that have been present since birth, and no mucosal lesions were found. Complaints of blisters at birth appeared first on the right and left legs, then complaints of blisters often appeared, and the location of the lesions moved. Currently, complaints of blisters appear on the upper and lower extremities scattered. According to the patient's mother, initially, the blisters filled with clear fluid grew larger and then the fluid turned cloudy and red-black. After that, the blisters burst, and a black scab appeared, which then came off and left white patches underneath. The patient's mother denied the previous complaint of fever. The patient's mother denied the family history from both parents, but similar complaints appeared in the patient's twin brother. The first complaint of blisters in the patient's twin brother also appeared at the same time as the patient. The patient's mother also said that the blisters tended to appear when both babies were exposed to high-temperature air. Both babies had received treatment because of the patient's skin condition, but the complaints did not improve, and then the patient was referred to Dr. Soetomo General Hospital. On examination, multiple bullae were found with tense walls, filled with cloudy fluid with varying diameters, and brownish crusts on the upper and lower extremities. Multiple erosions were also found in the lower extremities (Figure 1).

There were no complaints of fever, cough, runny nose, or diarrhea during the examination. There was no scar tissue picture, and Nikolsky's signs on both babies were negative. Based on history and physical examination, the diagnosis was first suspected with bullous impetigo and differential diagnosed with bullous epidermolysis and bullous pemphigoid. The temporary management for the twins was an administration of 0.9% sodium chloride (NaCl) compress on the crusted and wet lesion, topical antibiotic sodium fusidate 2% cream, and gentamicin 1% cream from the pediatric department on

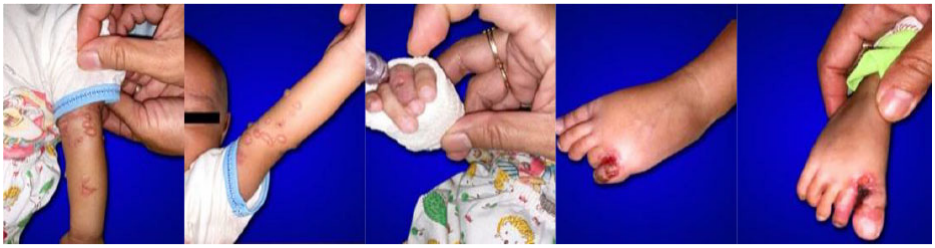

Figure 1. The first baby showed multiple bullae with tense walls, multiple erosions in the upper and lower extremities, and no nail or mucosal damage was present.

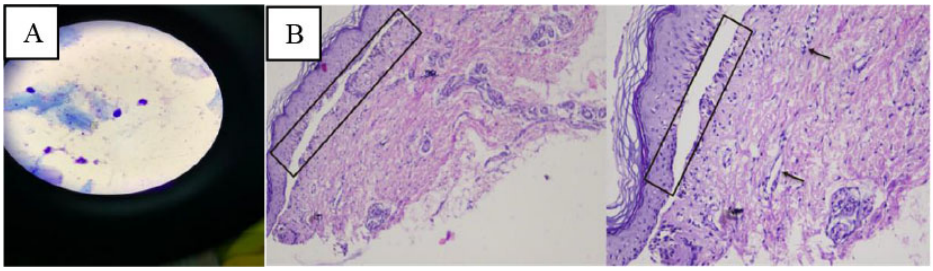

Figure 2. Supporting examination results. Cocci bacteria were found on gram staining (**A**), and histopathology examination found a subepidermal blister (black rectangle) with the dermis layer showing a slight infiltration of lymphocyte cells (black arrow) (**B**).

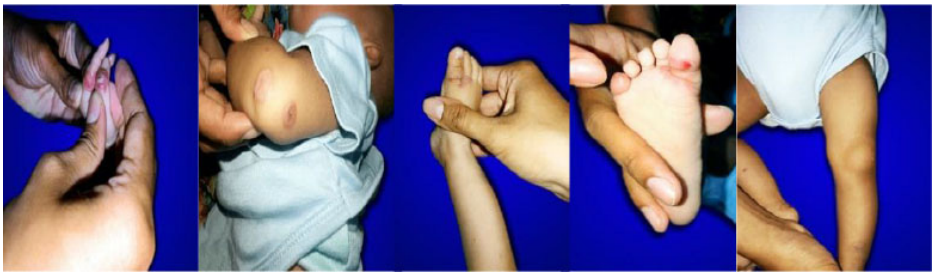

Figure 3. The second baby showed multiple erosions and scattered hypopigmented macules on the superior and inferior extremities, and there was no nail and mucosal damage.

the first baby. The first baby was planned for gram examination, swab culture, and biopsy. Cocci bacteria was found in the first baby, then on the culture examination, the results were *Staphylococcus aureus* bacteria and had sensitivity to gentamicin. The biopsy results showed an empty subepidermal blister with the dermis layer showing a slight infiltration of lymphocyte cells. The picture was obtained in accordance with bullous epidermolysis (Figure 2).

An examination of the patient's twin brother was carried out 1 week later. On examination of the second baby, multiple erosions and hypopigmented macules were found on the upper and lower extremities (Figure 3). No bullae were found at this time. The biopsy results in the second baby were the same as in the first, but the type of epidermolysis could not be determined. Therefore, the examination was continued by direct immunofluorescence in both infants. The results of the direct immunofluorescence examination did not reveal IgG and C3 deposits in the basement membrane zone (Figure 4).

Both babies were born at term, normal delivery, adequate weight, and are twins first. They received bacillus calmette guerin (BCG), polio, hepatitis B, diphtheria, tetanus, and pertussis (DPT) immunizations. The patient's mother had no complaints during pregnancy and performed routine examinations. There was no history of food or drug allergies in infants and parents. The next therapy for the second baby is wound care with 0.9% NaCl in the area of the ruptured blister. Besides that, sodium fusidate cream can be given. During control, the first baby's complaints of blisters had dried up, but in the second baby, new blisters appeared on the right knee (Figure 5A, B). We also provide education about disease, wound care, and disease prognosis to both parents of patients.

DISCUSSION

Epidermolysis bullosa in infants is a very rare case. The etiology and pathogenesis of EB in infants is still unknown. Hereditary factors and genetic mutations are known to predispose to EB. Almost as many as 16 genetics are thought to have a role in the occurrence of EB. The characteristic features of EB include mucocutaneous bullae caused by the mechanism of damage that occurs in skin tissue. This tissue damage also causes erosion and ulceration, usually formed due to frictional trauma.[2]

Epidemiological data indicate that EB simplex is the most common type of EB, with the number of cases more than 70% of all types of EB. Epidermolysis bullosa simplex is inherited in an autosomal dominant manner in most cases. In some cases, EB simplex can be inherited in an autosomal recessive manner. Junctional type EB is often inherited in an autosomal recessive manner. The appearance of bullae between the lamina lucida layer and the basement membrane zone area usually characterizes it. Dystrophic EB can be inherited in an autosomal dominant and autosomal recessive manner, and the latter is mixed EB (Kinder syndrome), which is inherited in an autosomal recessive manner. Kindler syndrome is characterized by extensive shedding of the skin layer and the appearance of bullae in the acral area, photosensitivity, atrophy of skin tissue, and extensive poikiloderma.[2] Based on the latest classification, EB simplex is divided into three major parts, namely localized EB simplex (Weber–Cockayne), intermediate (generalized intermediate/Koebner), and severe (Dowling-Meara).[3] Typical characteristics that often occur in EB simplex are symptoms that often appear in early life (early childhood), and bullae symptoms are often triggered by an increase in air temperature. In this group, mucosal symptoms and nail damage are rare. Symptoms in other types are more

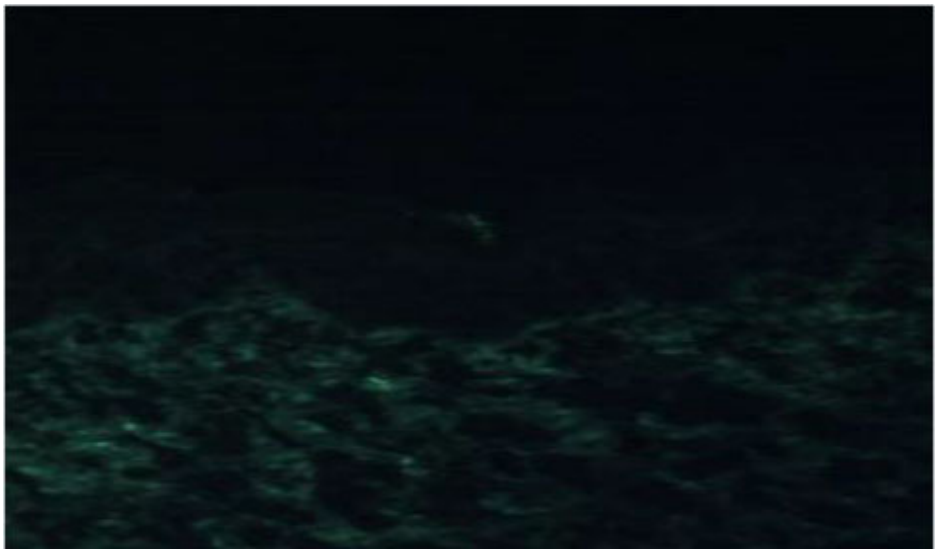

Figure 4. Direct fluorescence in both babies showed negative results.

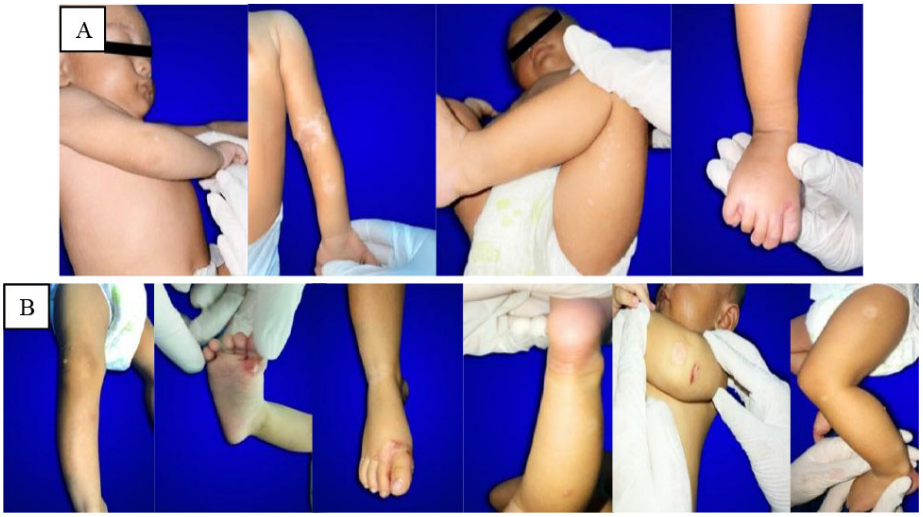

Figure 5. Follow-up examination on both babies showed improvement (**A, B**). New blisters were found on the second baby (B).

severe and cause mucosal abnormalities.[5] Typical characteristics that often occur in EB simplex are symptoms that often appear in early life (early childhood), and bullae symptoms are often triggered by an increase in air temperature. In this group, mucosal symptoms and nail damage are rare. Symptoms in other types are more severe and cause mucosal abnormalities.[6]

Electron microscopy is the gold standard examination of EB. Electron microscopy examination can see the level of blistering in EB more precisely than biopsy and immunofluorescence examination. Besides that, electron microscopy is also able to see cells and subcells that ordinary (ultrastructural) microscopes cannot reach.[1,5,7] In this case, electron microscopy examination cannot be carried out due to limited equipment and high cost. We also did a direct immunofluorescence examination on both babies. In this case, direct immunofluorescence aims to rule out the differential diagnosis of EB. The results of the direct immunofluorescence examination did not reveal IgG and C3 deposits in the basement membrane zone, so the diagnosis of bullous pemphigoid and epidermal bullous acquisita could be ruled out.[8,9] Another additional test that can be done to establish the type of EB is genetic testing. Studies report different causes of genetic mutations in each type of EB.[1,9,10] Most EB therapy is carried out in a supportive manner. Treatments, including wound care, infection control, surgical therapy, and adequate nutrition must also be considered. Providing education to the family must also be considered, considering that EB is a life-long disease.[1] The prognosis of epidermolysis in infants depends on the location of the bullae.[11]

CONCLUSION

EB is a very rare case, especially in twins. Determining the type of EB is still difficult to do. Microscope electron is a gold standard for determining the type of EB. Symptomatic treatment still holds as the main therapy of EB.

PATIENT'S CONSENT

The parents of the patient received consent and agreed to share the clinical picture and history of the patient for education and publication.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest regarding the publication of this.
Article.

AUTHOR CONTRIBUTIONS

Author DMH constructed the concepts, searched the literature, data collection, main writer of the manuscript, and edited the manuscript. Author DI searched the literature, collecting data, and prepared and edited the manuscript. Authors IZ, S, IC, and YW reviewed the manuscript. The author constructed concepts and reviewed the manuscript.

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