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Epidermolysis bullosa with severe anemia in an infant

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ABSTRACT :
Epidermolysis bullosa (EB) is a group of rare inherited connective tissue diseases that lead to blister formation on the skin**.** The lesions, which may develop after birth or at any point until early adulthood, often appear in dense blisters that might rupture and leave scars. Blisters are most commonly found in areas of trauma or pressure, such as the hands, feet, and diaper area in children, but they can also appear in the mouth, gastrointestinal tract, or genitalia. This study reports a case of Epidermolysis bullosa with clinical manifestations of anemia and lower respiratory tract infection.

Keywords: epidermolysis bullosa, blistering skin diseases, skin fragility.

I. INTRODUCTION:

The genetic skin disorder epidermolysis bullosa (EB) results in skin fragility, where even minor friction or injury can split the skin layers, leading to blisters and open soresand, in some cases, harming mucosal membranes and internal organs. Children with EB generally have skin that is as thin as a butterfly wing. The prevalence and incidence of EB were estimated to be around 8 per million people and 19 per million live births among this Robust study population(1). Epidermolysis bullosa has been classified into fourtypes that aresimplex, junctional, dystrophic, andKindler syndrome. Although the three kinds of EB have distinct causes, all three present with the same symptoms painful blisters and sores (2).

Most cases of epidermolysis bullosaare due to The type I and type II intermediate filament (IF) proteins K14 and K5, which are responsible for forming a pancytoplasmic network of 10-nm filaments in basal keratinocytes of the epidermis and in other stratified epithelia, are mutated dominantly in the majority of cases of epidermolysis bullosa simplex. Basal keratinocytes become brittle and rupture due to trauma due to defects in the K5/K14 filament network topology andhow keratin biology-focused laboratory studies have advanced our knowledge of the etiology and pathophysiology of EB simplex(3).

It can be diagnosed through genetic testing, prenatal testing, and biopsy or skin sampling for immunofluorescent mapping. Blister care, daily skin cleaning, daily dressing with therapeutic agents, cooling, a suitable diet, pain and itching control, antibiotics, surgery (esophageal stenosis excision, gastrostomy tube installation for skin grafting), and physical therapy are some of the treatments. One of the repercussions of this illness is esophageal blisters and sores, as well as stenosis, dysphagia, and infection.(4)

Case presentation:

A 17-month-old female patient born of a non-consanguineous marriage presented with chief complaints of fever, cold and wheezing on breathing, blistering, crusted and scarred lesions all over the body and with known complaints of Epidermolysis bullosa simplex and LRTI was hospitalized in the pediatric department. She had no history of Tuberculosis, genetic disease and allergies. She was apparently asymptomatic 1 week before later she developed a fever; acute onset,high grade subsided with medication again relapsed no associated with rash, ear discharge, burning micturition, constipation, pain in the abdomen, vomiting, seizures. Cold for a week associated with the nasal block. She had a similar history in the past and was admitted to the hospital at age 4 months (bronchopneumonia for 12 days) and at age 6 months (epidermolysis bullosa simplex with LRTI for 14 days).She was delivered full-term through normal vaginal delivery without any difficulties for the mother and fetus. The first child in the patient's family was healthy and her parents have no similar complaints.

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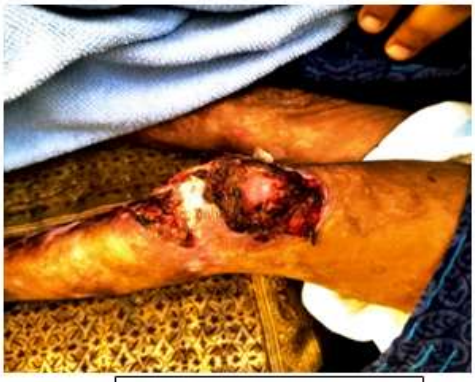

Fig.1.: showing wounds on legs

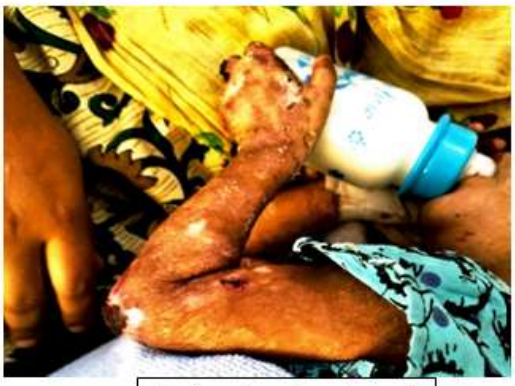

Fig.2.: showing injured skin on hands

Soon after the birth, the child had extensive skin lesions with blisters all over the body. The skin is so delicate that it is prone to trauma even with the stretching of the clothes.The child's sensitive skin led to recurring traumatic harmful sores.

At initial assessment, the baby was moderately active and pallor, she was febrile with high-grade fever (100^0F) and theother vitals are normal.Furthermore, the patient had multiple blistering lesions all over her body, which were crushed and scarred. There were also skin lesions associated with trauma and pressure, as well as multiple skin erosions, ulcers, and oral erosion. The patient's mother had applied mega heal and vita vera lotion on the blisters, dry and itchy skin areas, respectively. Initial laboratoryfindings show that the patient's white blood cell count is abnormally high, suggesting the presence of an infection. She was anemic with a hemoglobin level of 4.6 g/dL, Ig-G antibodies count was also very low which is 0.2 g/dL. The laboratory data suggests that she had uremia, hypoalbuminemia, and hyperkalemia. The CRP levels were high-48mg/l, which shows the severity of the infection.

Treatment: A blood transfusion was done as she was anemic, antibiotics like ceftriaxone and cefixime have been prescribed to rule out infection, syrup paracetamol was given to relieve fever, syrup chlorpheniramine was given to treat runny nose, Z & D drops were prescribed to strengthen the immune system, clotrimazole mouth paint was prescribed to treat mouth ulcers. Ointments, creams and lotions like Veta vera lotion, liquid paraffin, soframycin ointment, and fusidic acid ointment were prescribed to reduce the intensity and severity of epidermolysis bullosa simplex and to help in early recovery. The detailed treatment is as follows;

Table no 1: **Treatment chart:**

Name	Generic name	Dose	Route of administration	Frequency
Inj. Ceftriaxone	Ceftriaxone	75 mg/kg/day	Intravenous	BD
Syp. Cefixime	Cefixime	8 mg/kg/day	Oral	OD
Syp. Chlorpheniramine	Chlorpheniramine	0.35mg/kg/day	Oral	BD
Syp. Paracetamol	Paracetamol	15 mg/kg/day	Oral	SOS
Vita vera lotion	Aloe vera extract	-	External application	TID
Liq. Paraffin	Paraffin	-	External application	TID
Z & D drops	Nutritional supplement	5ml	Oral	BD

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Soframycin ointment	Soframycin	-	External application	BD
Fusidic acid ointment	Fusidic acid	-	External application	BD
Clotrimazole mouth paint	Clotrimazole	-	Gently apply with a cotton pad	BD

II. CASE DISCUSSION :

Both patients and family members experience serious consequences from severe EBS. Obstetricians and pediatricians must be knowledgeable about the mode of inheritance, age-related morbidity, and mortality linked to this uncommon but severe disease in order to promptly counsel the families on the disease's natural history, recurrence risk, and reproductive options. Even though there have been a few reports of uncommon autosomal recessive variants of EBS, autosomal dominant inheritance contributes to the majority of cases. Understanding the precise genetics of EBS aids in advising families on the prognosis of their affected children and the likelihood that the condition will return in future pregnancies(5).

EBS, in contrast to EBD and EBJ, is often a less severe illness with a lower fatality rate(6). Based on the beginning of the disease at birth, distributed friction or trauma-induced blistering, and involvement of the oral mucosa, our patient had severe EBS. Despite these findings, severe EBS symptoms usually get better over time. Septicemia, starvation, and electrolyte imbalances are the primary causes of early morbidity and mortality in severe EB. Therefore, careful consideration must be taken into account for nutrition and skin care. Recurring mucosal sores, eating difficulties, high energy expenditure from increased skin turnover, transcutaneous nutritional loss, and a catabolic state brought on by recurring infections are all causes of malnutrition (7). It is crucial to involve dietitians in the preparation of simple recipes, the identification of high-calorie and protein-fortified foods and beverages to replace protein lost in draining blisters, the suggestion of vitamin and mineral nutritional supplements, and the recommendation of dietary modifications to prevent gastrointestinal problems like constipation, diarrhea, or painful defecation. It is crucial to emphasize the value of a balanced diet while in the hospital.

In this case, the patient with EBS had clinical manifestations of LRTI with severe anemia and blistering, crusted lesions all over the body. laboratory findings WBC & CRP values are high. She was treated with antibiotics to minimize the infections, ointments were applied on to the lesions. The patient had blood transfusions. After 15 days of hospitalization, the patient was discharged as her WBC and CRP values are minimized. Antibiotics, creams, vitamins, and minerals were prescribed to continue after the discharge. The parents were counseled regarding the nutritional diet, recurring infections, skincare,and the likelihood of a condition in future pregnancies.

III. CONCLUSION:

We report an infant with EBS in this article. Managing the disease is traumatic for both parents and the child. Skin needs to be meticulously taken care of to reduce the risk of infections. To stop bleeding and infections, wounds must be dressed regularly. Due to ongoing blood loss, the patient may need regular blood transfusions.These patients are exposed to various types of infections, including sepsis. Therefore, an increase in body temperature, WBC, and CRP should be considered along with other diagnoses.

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