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A Newborn with Epidermolysis Bullosa

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Abstract- Bullous disorders of newborn are a rare entity. Genetically inherited bullous disorders, especially the dystrophic and junctional types have a fatal course (1). Breach of the epidermis in the newborn predisposes them to sepsis.

Index Terms- epidermolysis bullosa, simplex, junctional, dystrophic.

I. CASE REPORT

Baby born to third degree consanguinous parentage had a normal birth history. Immediately after birth, a small raw area of skin 2 x 2 cms was noted in the left foot. Baby was admitted in Neonatal Intensive Care Unit for observation. Within, 6 hours of birth, small bullae were noticed in the cheek. In the next 24 hours, large flaccid bullae developed in the right elbow and heel of right foot. The bullae contained clear fluid and ruptured off with minimal friction leaving large areas of raw skin exposed. The earliest lesions started healing with scarring.

The lesions relentlessly progressed to involve most of the body area, mostly involving the periorificial areas and the extremities, healing with scarring and contracture formation with tightening and narrowing of oral orifice, contracture of elbows and knees.

Child was started on intravenous antibiotics cefotaxime and amikacin and later changed to vancomycin and meropenam .

Dermatologist opinion was obtained and an opinion of epidermolysis bullosa was given.

Cultures from skin and blood were taken. Skin biopsy for electron microscopy and immunofluroscence testing could not be done due to poor general condition of the newborn baby. Adequate feeding, intravenous fluids for hydration, topical antibiotics and emollients for hydration of skin were instituted. Baby developed features of sepsis by day 7 of life. Appropriate antibiotics were started and revised following culture sensitivity report. In spite of antibiotics, baby died of fulminant staphylococcal sepsis by day 32 of life.

II. DISCUSSION

Epidermolysis bullosa refers to a group of inherited disorders characterized by bullos lesions that develop spontaneously or as a result of varying degrees of friction or trauma(2).

E. bullosa may be divided into 3 major inherited forms (simplex, junctional &dystrophic) based on the presence or absence of scarring and skin elements, mode of inheritance, level of skin cleavage(3).

The level of cleavage of simplex variety is within the epidermis and it heals without scarring. Inheritance is Autosomal Dominant and the basic pathology lies with the basal keratinocyte. The junctional variant separates in the Lamina Lucida of dermal epidermal junction and leads to atrophic scarring. Inheritance is AR and basic pathology is with the hemidesmosomes. Dystrophic variant has blister formation in papillary dermis, below the basement membrane. Both AD and AR variants are seen and anchoring fibrils are the abnormal.

Overall incidence is 1 in 50,000 live births(4).

III. SIMPLEX VARIANT

Blister is seen in areas of trauma due to defective keratin filaments and heals without scarring. Major forms are weber-cockayne disease (localized), Kocher variant (generalized), Dowling-meara variant (e.bullosa simplex herpetiformis).

Localized variant has a high threshold for frictional trauma, bullae are confined to hand and feet, seen first in hand and feet, hyperhidrosis is present, lesions heal without scarring(5).

Koebner variant has generalized blistering of skin, most notable in sites of friction. There is extensive blistering in neonatal period and early infancy. There is increased risk of sepsis which could be life threatening. Blistering improves with advancing age. Hyperhidrosis is common and mucosa is mildly affected, nails are not affected. Moderate hyperkeratosis of palms and soles are seen.

Dowling meara variant is the most severe form of simplex which is a differential diagnosis for severe dystrophic or junctional forms, but with overall good prognosis. Small characteristic blisters may be seen in the neonatal period on proximal extremities. Blistering decreases by advancing age and hyperkeratosis increases. Extensive blistering could be life threatening due to sepsis(6).

IV. JUNCTIONAL VARIANT

Cleavage plane is in the lamina lucida at dermo epidermal junction.

Herlitz variant is a lethal variant with 50% mortality within 2 yrs. There is perioral granulation tissue with sparing of lips. Blisters heal with atrophy but no milia. Periungal blistering and erosions are seen, anonychia, dental enamel dysplasia and anemia might be seen.

Non – Herlitz variant is less severe with less mucosal involvement.

Other types are the GABEB type, inversa and e.bullosa associated with pyloric stenosis or muscular dystrophy(7).

V. DYSTROPHIC VARIANT

Divided into Dominant and Recessive forms.

The autosomal dominant variant has its onset at birth. Blistering predominates on dorsum of hands, elbows, knees and lower limbs. This heals with scarring with milia formation. Nail dystrophy is associated.

Recessive form is present at birth with widespread blistering, scarring and milia formation. Deformities like digital fusion and joint contractures are seen. There is severe involvement of mucous membrane and nails. Growth retardation, poor nutritional status and anemia are seen. There are teeth abnormalities. Predisposition to squamous cell carcinoma is present in the heavily scarred areas.

Hallopea-Siemes type, a severe life threatening autosomal recessive type of dystrophic e. bullosa is seen with widespread dystrophic scarring, deformities and severe involvement of mucous membranes. Bullae occur at site of friction. Bullae may be hemorrhagic and lower extremities may be completely devoid of skin. When blister ruptures, raw painful surface is evident. Nikolsky sign is positive. Atrophic scars with milia formation are seen. There is hyperhidrosis and hyperkeratosis. Acquired pseudosyndactyly- claw hand or mitten hand deformities are seen. Fixed flexion deformities of elbows and knees with contractures occur. Bullae, initially sterile, get eventually infected with pseudomonas or staphylococcus which leads to septicemia.

VI. DIAGNOSIS

Skin biopsy for light microscopy does not provide any clue regarding the variant of e. bullosa.

Electron microscopy is the standard criterion for determining the level of blistering. EM can provide additional information on BMZ morphology that can be helpful in differentiating the different types. Immunofluroscence study and Immunomapping with antibodies to a hemidesmosomal antigen, BP230 and antibody to lamina densa protein (ex. type IV collagen) can distinguish epidermolysis bullosa simplex, junctional epidermolysis bullosa, and dystrophic epidermolysis bullosa.

Once the mutations are identified in a family, reliable prenatal diagnosis is possible(8,9,10).

Extensive areas of denuded skin represent loss of the stratum corneum barrier to microbial penetration. Accumulation of serum and moisture on the surface enhances the growth of bacteria.
Patients with severe epidermolysis bullosa subtypes may have immunologic abnormalities, including decreased lymphocyte production or a poor nutritional status that lowers resistance to infections. *Staphylococcus aureus* and *Streptococcus pyogenes* are the usual causative organisms, but gram-negative infections with bacteria, such as *Pseudomonas aeruginosa*, also can occur. Patients also have increased susceptibility to developing sepsis.

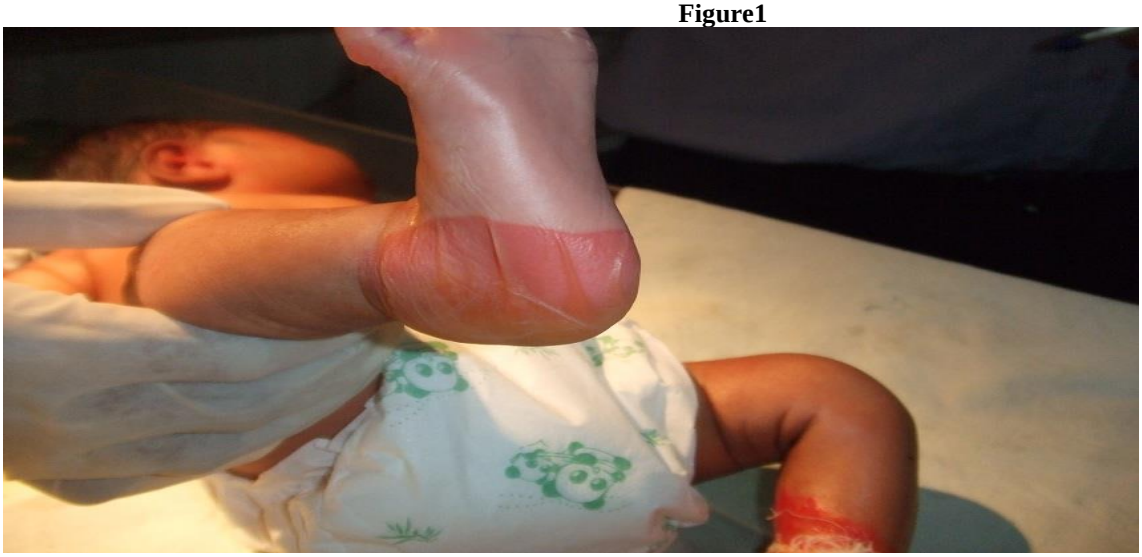
VII. TREATMENT

Parents are to be informed of risk of transmitting genetic abnormalities. Genetic counseling is given. Prenatal diagnosis for future pregnancy can be done id the genetic abnormality in the family is identified. Treatment of e. bullosa is mainly palliative. Avoidance of trauma, friction or pressure, Cool environment is preferred. Prevention of infection is the preferred strategy. With extensive areas of crusting and denudation, a strict wound care regimen should be followed. The wound should be covered with semi occlusive nonadherent dressings. Self-adhering gauze or tape is a better choice for keeping dressings in place. Topical mupirocin is drug of choice. Appropriate parentral antibiotics should be started when necessary.

VIII. CONCLUSION

Epidermolysis bullosa are blistering skin disorders. Course of the disease varies according to the level at which blistering occurs in the skin. Septicemia is the cause of death in most of these cases.

Figure1



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