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CASE STUDY: EPIDERMOLYSIS BULLOSA

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Background:

Epidermolysis Bullosa is a group of rare genetic skin conditions, which is characterized by extremely fragile skin and recurrent blister formation, resulting from minor mechanical friction or trauma. Discoveries of the molecular basis of epidermolysis bullosa have resulted in the development of diagnostic tools, including prenatal and preimplantation testing. Historically, epidermolysis bullosa subtypes have been classified according to skin morphology. Based on a better understanding of the basement membrane zone (BMZ) and the genes responsible for its components, newer treatments (e.g., gene or protein therapy) may provide solutions to the skin fragility found in patients with epidermolysis bullosa.

Abstract:

Epidermolysis bullosa is a large rare group of genetically determined disorders that are inherited. As autosomal dominant or recessive.

Most common feature of this disease is blisters formation on the skin and oral mucosa at different

Levels following trauma or traction. Exact cause still unknown, but mostly appears to be related to abnormal enzyme activity and collagen.

The most common oral manifestations involve, enamel hypoplasia, rampant caries, microstomia

Tongue atrophy, oral ulceration with scaring and constriction of buccal vestibules. Dental treatment of these patients is a great challenge that involves preventive measures by rein-

Forcing oral hygiene and regular fluoride applications, in addition to treatment of carious teeth and

Extraction of teeth with poor prognosis.

In this study two cases of Epidermolysis Bullosa, simplex and dystrophic types are reported and their specific clinical Manifestations with their dental clinical treatment are described.

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INTRODUCTION:

Epidermolysis Bullosa (EB) is a group of rare genetic skin conditions, which is characterized by extremely fragile skin and recurrent blister formation, resulting from minor mechanical friction or trauma.

The skin has two layers, the outer layer is called the epidermis and the inner layer the dermis. Normally, there are 'anchors' between the two layers that prevent them from moving independently from one another. In people with EB, the two skin layers lack the anchors that hold them together, and any action that creates friction between the layers (like rubbing or pressure) will create blisters and painful sores. Sufferers of EB have compared the sores to third-degree burns.

Over the past 15-20 years, 13 major genes responsible for the majority of cases of EB have been identified. The genetic errors in EB result in defects in the proteins that adhere the epidermis to dermis.

In many countries, Butterfly Children is a term often used to describe younger patients because their skin is as fragile as a butterfly's wings. Sometimes, children with the condition are also described as Cotton Wool Babies. And in South America, Crystal Skin Children is the term used.

CASE REPORT:

This case about a female neonate with blistering of the skin during the immediate neonatal period. A 1.2 kg term child was born to 29 weeks old primigravida mother by means of SVD at General Hospital. The mother was reserved at 20 weeks of development; hence she had ordinary antenatal visits and an evidently uneventful antenatal period. There was no history of skin issue in either the mother's or father's families. After delivery Mother's baby was with APAGAR score of 8 at first moment and 9 at fifth moment. Examination following birth was unremarkable with no cetaceous appearance. At 3 hours, the infant created unconstrained stripping of skin with sub-conjunctiva hemorrhage. Steadily there was development of rankles which at first included the neck, ear, lower leg and lips. The child was moved to nursery. The examination in the nursery baby body containing straw-shaded liquid the rankles would crack to uncover seriously erythematous basic skin. Insignificant erosion over the skin inspired new rankles. Complete blood checks (CBC), Blood Culture were sent these tests were inside typical extents. The infant was seen by a dermatologist who affirmed the analysis of Epidermolysis bullosa and exhorted nursing care, delicate dealing with, Misstating oral drops, Fuci eat cream, Cloxacillin, Vaseline and fluid paraffin as an emollient. Quiet was on preservationist the executives infusion, ceftriaxone 60mg I/v B.D, infusion amikacin 10mg I/v B.D, infusion 10% D/W, infusion calcium gluconate in 50ml N/Saline. Infusion linezolid 12mg IV TDS. Treatment centers around strong consideration and the counteractive action of inconveniences for instance, the decrease of rankling by wearing delicate attire, greasing up skin, and staying away from grinding; thinking about rankled and contaminated skin using dressings, wound-recuperating creams, and depleting rankles.

INVESTIGATION:

Indirect immunofluorescence staining was performed with 17 antibodies on a skin biopsy and the result was consistent with Epidermolysis bullosa. Two intraepidermal splits were found in the granular and basal layers. Diagnosing Epidermolysis bullosa can be made by histology on skin biopsies but at the time of investigation the clinical team chose to only take one biopsy due to the risk of infections, and this biopsy was used for immunofluorescence.

Molecular genetic analysis by direct sequencing of DNA extracted from peripheral blood lymphocytes from the proband identified a KRT5sequence variant: c.1429G>A; p. (Glu477Lys) in heterozygous form. This missense mutation has previously been reported in 2 (both sporadic) of 10 cases with generalized severe Epidermolysis bullosa. The parents of the proband were both clinically unaffected and, as most cases of Epidermolysis bullosa are autosomal dominant, this case was most probably sporadic. The sequence variant was not present in DNA from the mother, but a blood sample for molecular genetic analysis from the father for the familial KRT5 mutation was not available.

Can be caused by mutation in either KRT5or KRT14, thereby disturbing the normal K5/K14 keratin filament assembly.

DICUSSION:

Epidermolysis bullosa (EB) is a group of rare inherited bullous skin disorders that differ in nature and severity. Currently, there is no cure for the disease. One of the complex problems of EB is the repetitive and painful care of skin wounds. The purpose of this study was to explore how adult patients the impact of wound care during childhood and which coping strategies they considered as helping. The impact, physically, psychologically and on daily life, was apparent for patients and parents. Helpful coping strategies were transferring care, regulating emotions, and dyadic strategies, such as supporting each other by

distraction, encouragement, using rituals and collaboration. EB is due to a mutation in at least one of 18 different genes. [2] Some types are autosomal dominant while others are autosomal recessive. [2] The underlying mechanism is a defect in attachment between or within the layers of the skin.[5] There are four main types: epidermolysis bullosa simplex, dystrophic epidermolysis bullosa, junctional epidermolysis bullosa, and Kindler syndrome.[2] The diagnosis is suspected based on symptoms and confirmed by skin biopsy or genetic testing.[6]

There is no cure for the condition.[4] Management involves wound care, pain control, controlling infections, nutritional support, and prevention and treatment of complications.[2] About half a million people are affected globally.[5] It occurs equally commonly in males and females.[7] Children who are affected may be bullied by other children or experience inappropriate comments from adults.[5].A group of restorative masters will enable you to choose what treatment is best for your kid and offer counsel about living with the condition. Drugs can be utilized to treat contamination or to diminish torment. Medical procedure might be required if EB causes narrowing of the nourishment pipe or issues with the hands.

CONCLUSIONS:

Epidermolysis bullosa equiseta is a rare disease in childhood. Mucosal involvement is frequent and severe. Because the clinical features are misleading, the use of immune electron microscopy and Western blot analysis is essential to making a diagnosis. Treatment with a combination of prednisone and daps one is often effective. The prognosis in children is better than it is in adult patients.

MATERIALS AND METHODS:

Patients and biological materials:

Families were recruited in Lahore over a period of 5 years by actively searching all available registries at our institutions as well as by publicizing our efforts during professional meetings. All participants Or their legal guardian provided written and informed consent According to a protocol previously approved by the local Committee and by the National Committee for Genetic Human Research of the Lahore Ministry of Health in accordance with the Declaration of Helsinki Principles. Blood samples were drawn from all family members and DNA was extracted according to standard Procedures. Epidermolysis Bullosa diagnosis and subtyping were determined based on Light microscopy and electron microscopy examination of skin

Biopsies as described previously (Bergman, 1999; Petronius et al.2003).

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