CASE REPORT

Suspected Epidermolysis Bullosa Simplex in a 5 Week Old Nigerian: A case Report.

B.E. Otaigbe FWACP, **B. Datubo-Brown** MBBS
Department of Paediatrics, University of Port Harcourt Teaching Hospital, Port Harcourt, Nigeria

ABSTRACT

Background: *Epidermolysis bullosa is a rare non scaring autosomal dominant disorder characterized by recurrent blistering of the skin and mucous membrane. The skin is fragile and minor rubbing may cause blistering. It's epidemiology in our environment is unknown probably because of paucity of information on the clinical presentation and management with resultant mortality within the first few months of life.*

Method: *A case report of a 5-week old female who presented with generalized blistering and denudation of the skin first noticed on the left foot at birth and a review of the literature on the subject using Medline and online search was used. She was treated at various traditional medicine homes and clinics before referral to the teaching hospital. She was managed initially for bullous pemphigus with antibiotics for proven septicaemia, and the wound infection ignorantly managed with daily sofratulle dressing alternating with closed dressings before a definitive clinical diagnosis of was made.*

Result*: The patient was referred late to the tertiary centre. She was initially treated for bullous pemphigus and sepsis with antibiotics and wound dressings with poor response before the diagnosis of epidermlysis bullosa was made. The patient died from severe foot bleeding before blood could be transfused.*

Conclusion: *Delay in making the right diagnosis hence the appropriate treatment even in a tertiary health is being highlighted.*

KEYWORDS: Suspected Epidermolysis Bullosa; Delayed diagnosis; Complications.

Paper accepted for publication 8th July 2006.

INTRODUCTION

Epidermolysis bullosa (EB) is a genetic disease characterized by the presence of extremely fragile skin and recurrent blister formation resulting from minor mechanical friction or trauma[1,2]. It is classified into three main categories including 1) Epidermolysis Bullosa simplex (EBS), 2) junctional Epidermolysis Bullosa (JEB), 3) dystrophic epidermolysis bullosa (DEB). In the United States, it is estimated to affect 12,500 people with 50 cases occurring per million live births. It affects all sex and race[2].

Epidermolysis bullosa simplex (EBS) is usually inherited in an autosomal dominant manner and rarely in cases of consanguity, as autosomal recessive. It is associated with little or no extracutaneous involvement unlike the other forms which can produce significant multi-organ involvement. Four clinical forms of EB are known, ranging from a mild blistering of the hands and feet (EBS, Weber-Cockayne type), to a generalized blistering EBS (Kobner type), EBS with mottled pigmentation and the fatal EBS (Dowling Maera type). The defect in all types is in the central á-helical coil of keratin 5 and 14 which makes up intermediate filaments of the basal keratinocytes. Keratin 5 and 14 genes are located on chromosome 17q and 12q respectively. The intraepidermal bullae result from cytolysis of the basal cell [3]. The aim of this report is to highlight the clinical presentation, complication and the management of this disorder which can be fatal if not properly diagnosed and managed.

CASE REPORT

A 5-week old female referred to the University of Port Harcourt teaching hospital(UPTH) from a Federal Medical centre in Bayelsa state with a history of extensive skin excoriations, shedding of nails of both fingers and toes and a high grade fever of all of four weeks duration. Blisters were first noticed on the dorsum of the left foot to mid shin. It subsequently involved the chest, abdomen, gluteal region and back. Fever which started one week later was high grade and intermittent. The Mother had visited various clinics where the infant was dressed with gauze and bandages. She also received oral ampiclox, paracetamol, and chloroquine with no improvement. This prompted referral to the University of Port Harcourt Teaching Hospital. Pregnancy was unsupervised and uneventful. Mother did not ingest any drugs or herbs.

There was no history of consanguity or history of blistering in the family. Review of the systems revealed nothing significant. The mother was unmarried.

On examination the patient was ill looking, febrile (38.2 ^{0c}), not dehydrated and weighed 3.4kg. She was moderately pale with tightly packed occlusive dressing over the trunk and lower limbs (Fig. 1) which when removed revealed extensive areas of denuded skin

Correspondence to: **Dr. B.E. Otaigbe**
E-mail barbiejoe64@yahoo.com

extending over cheek, chest, abdomen, axilla, elbow, knees and both upper and lower limbs. There was hypopigmentation of the interscapular area and a flexor deformity of the left foot with an ulcer and scarring on the ankle of same foot (Fig. 2). There was no area of scarring over other parts of the body. There was shedding off the nails on all fingers and toes (Fig. 3). Other systems were normal.

A provisional diagnosis of bulbous pemphigus with congenital absence of nails was made. She was commenced on intravenous Ceftriaxone 50mg/kg in two divided doses, Genticin 5mg/kg in three divided doses with daily sofratulle dressing after a saline bath. Dermazine and Genticin cream were also applied. She received 750 IU of Anti tetanus sera (ATS). On review by the Burns and Plastic team closed dressing alternated with sofratulle gauze was prescribed. Results of investigation showed leucocytosis with neutrophilia and a haematocrit of 27%. Retroviral screening and VDRL were seronagative and non reactive. Blood culture yielded heavy growth of *klebsiella spp* while the wound swab isolated a heavy growth of both *pseudomonas aeriginosa* and *staphylococcus aureus*. Same antibiotics were continued as the organisms were sensitive to them. She sucked actively on the breast on demand and received water orally. Fever settled after 5 days and child appeared to be stable. On the eleventh day of admission a diagnosis of suspected Epidermolysis simplex- Dowling Meara type was made and hence the occlusive was discontinued. Haemorrhagic areas were noticed on the same day of removal of bandages. She subsequently bled actively overnight (unnoticed by mother and nurses) from an ulceration on the left foot and died before she could be transfused.

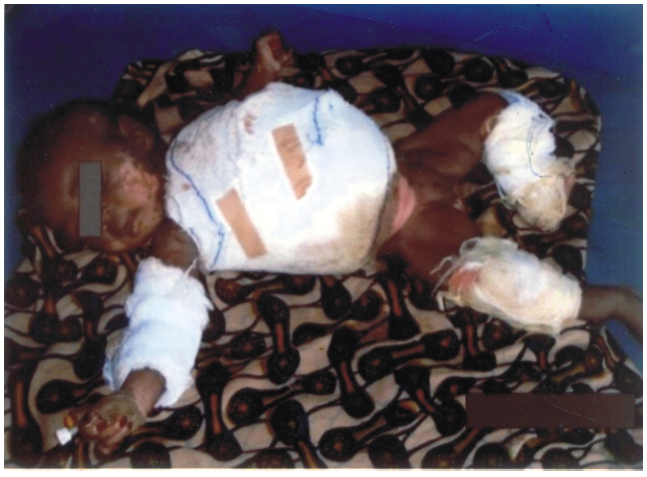

Fig. 1. Five week old female with Epidermolysis Bullosa simplex with closed occlusive dressing on admission

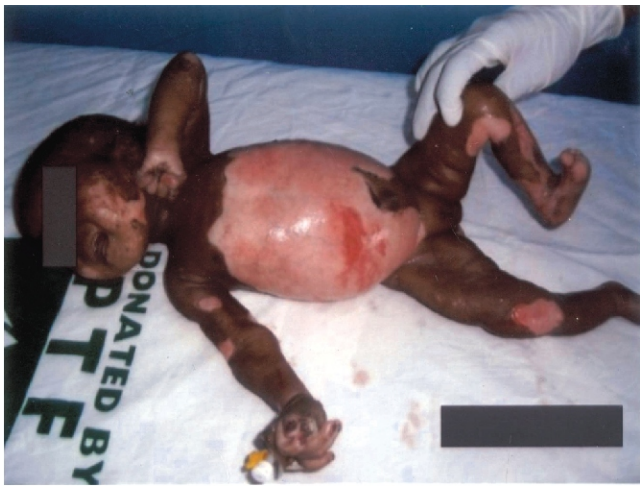

Fig. 2. Extensive skin denudation in patient with EBS-DM

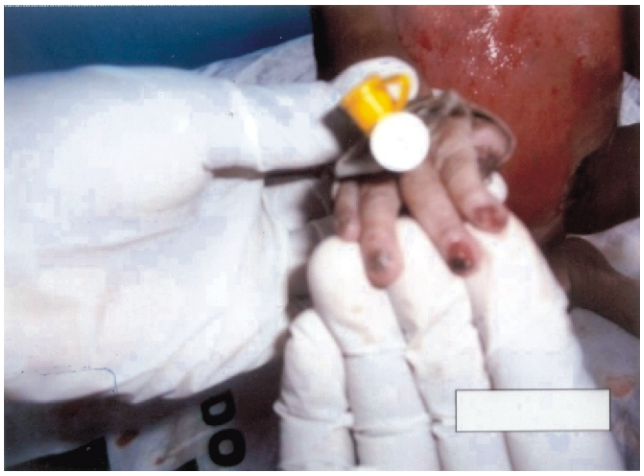

Fig. 3. EBS-DM showing absence of fingernails

DISCUSSION

Fragility of the skin in EBS can result from little or no trauma[4]. In this index case the presence of flexor deformity and blisters forming on the left foot at birth may suggest intrauterine mechanical trauma, though a history of breech presentation and oligohydranminos was denied by the mother. In EBS Dowling Maera type (EB-DM), onset is usually at birth, as seen in our patient, and widespread blistering is typical. Sites of predilection include the hands, feet, elbows; knees, legs and scalp[3]. Bullae heal with minimal or no scaring. Unlike in other forms of EBS, nail dystrophy is common in EBS-DM as are hyper and hypo pigmentation. The nails were shed and absent on all limbs in our patient and there was evidence of hypo pigmentation on the back between the scapulae. Intraoral lesions are minimal but were not noticed in our patient who sucked well at the breast throughout admission. Herpetiform blisters though often seen were also not noticed in this patient. Haemorrhagic blisters are present mainly in EB-DM and are known to be severe enough to result in neonatal

death.

Transmission electron microscopic examination of a skin biopsy or immunofluorescent antibody/antigen mapping is the sine qua non for diagnosis of EBS[4]. This is not possible in our center. Though examination of a skin biopsy is often required to establish the diagnosis, it may not be necessary in some individuals especially those with a family history of blistering.[4] This was however denied by the mother a single parent, who did not know much about the father and his family.

Management involves evaluation of the sites of blister formation including oral and oesophageal blisters and erosions which may indicate the severity of the disease. The extensive blistering at presentation was an indication of severe disease in this patient.

Supportive care to protect the skin from blistering, appropriate dressing that will not further damage the skin and the prevention and treatment of secondary infections is the mainstay of treatment[4]. Closed dressing is contraindicated but unfortunately was ignorantly used in the various clinics the mother had visited and initially also in UPTH. This resulted in further skin erosion with possible erosion into a blood vessel with resultant bleeding. Dressing in three layers is advocated[4] ; primary nonadhensive dressing impregnated with petrolatum or topical antiseptic, roll of gauze to provide stability to primary layer, as the secondary layer and a tertiary layer with some elastic property to ensure the integrity of the dressing e.g. a band net. Cyprohepatadine (periactin) can substantially reduce blistering in some individuals with EBS-DM.[6] Twenty percent aluminum chloride applied to palm and soles can reduce blister formation in some individuals, presumably by decreasing sweating[4].

Failure to thrive may be a problem requiring additional nutritional support. Fluid and electrolyte derangement may be enough to threaten life in the neonatal period and requires careful management[3]. Our patient was monitored to ensure adequate caloric and fluid intake through breastfeeding and oral administration of water.

Infancy is an especially difficult time for EB patients as the generalized blistering may be complicated by infection, sepsis and death[2]. Infection is due to loss of the stratum corneum barrier to microbial penetration, accumulation of serum and moisture which enhances bacterial growth and associated immunological abnormalities in severe forms which lowers resistance to infections. *Staphylococcus aureus* and *streptococcus pyogenes* are the usual causative organisms, but Gram-negative infection with *pseudomonas aeruginosa* can occur. In our patient, blood culture yielded heavy growth of *klebsiella spp* while the wound swab isolated a heavy growth of both *pseudomonas aeruginosa* and *staphylococcus aureus*. She was managed aggressively with appropriate antibiotics. Treatment with topical antibiotics for wound infection or the use of silver impregnated dressings or gel is helpful.[5] Prevention of infection entails strict wound care, regular whirlpool therapy followed by application of topical antibiotics[2].

Excessive heat may exacerbate blistering and infection in all forms of EB except with EB-DM which appears to improve with heat or warmth in some individuals. This may have been the case with our patient who remained stable until she bled. Death from exsanguinations was the secondary cause of death in our patient and could have been prevented with early detection. Generally the condition of patients with EBS-DM improves with time.

Genetic counseling should be offered to affected parents.

CONCLUSION

EBS is a rare disease whose diagnosis can be easily missed. Certainty of diagnosis in our environment is hindered by the absence of immunoflorescent antibody/antigen mapping and transmission electron microscopy of skin biopsy. The prognosis is good with early detection, appropriate wound management, infection control and nutritional support.

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