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Hereditary Epidermolysis Bullosa (A Two Cases Report)

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Abstract: Congenital epidermolysis bullosa are rare genodermatoses, clinically characterized by epithelial fragility leading to the formation of bubbles and cutaneous erosions by cleavage between the epidermis and the dermis. We report the observation of two children in care at pediatric plastic surgery department of the Children's Hospital of RABAT for syndactylies involving the fingers of both hands secondary to this pathology and we will focus on the particularity of the management of this rare pathology.
Keywords: epidermolysis bullosa, hereditary, genodermatosis, child.

INTRODUCTION

Epidermolysis bullosa (EB) is a heterogeneous group of about thirty skin and mucosal conditions, which is accompanied by bubble formation, caused by minor trauma (mechanical fragility). These are rare diseases. For the hereditary form, the cause is the deficiency of one of the proteins involved in anchoring the epidermis to the dermis. There are three types of epidermolysis bullosa: simple EB (intra-epidermal), junctional EB (within the lamina lucida), dystrophic EB (under the lamina densa). The management consists in treating the wounds so that they heal well and as quickly as possible and prevent complications and pain related to skin damage. No curative treatment of HEB is available to date.

CASE REPORT

We report 2 cases treated in the department of Surgery "C" - Unit of Pediatric Plastic Surgery and burns at Rabat Children's Hospital.

Case 1

11 year old boy; having a sister who died at day 15 of life; who presented syndactylia involving the fingers of both hands (mitten hands) (Figure 1) associated with multiple erosions, esophageal stenosis, stunting and anemia.

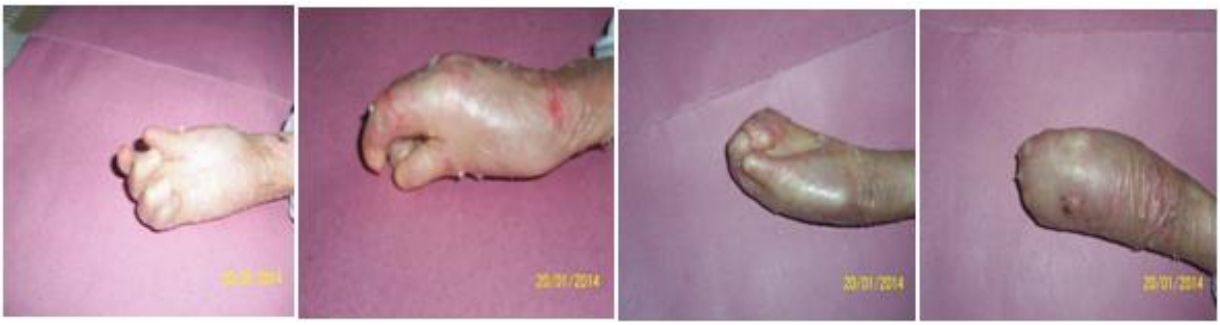

Fig-1: Image showing the appearance of hands in mittens of our first case

Case 2

8-year-old girl, 1st degree consanguinity, who presented cutaneous and mucosal bubbles and erosions associated with syndactyly of both hands (mitten hands) (figure 2), and a failure to thrive.

The management of both cases was multidisciplinary with collaboration between the intensive care unit and the pediatric surgery department (Figure 3 and 4).

Fig-2: Image showing the appearance of muffled hands in our 2nd case

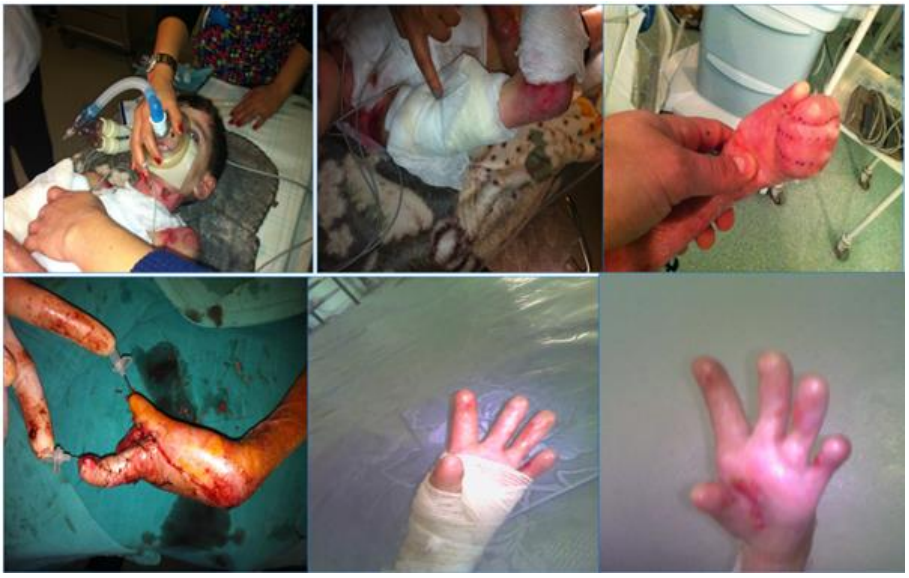

Fig-3: Image showing the management (resuscitation, surgical and results) of our first case

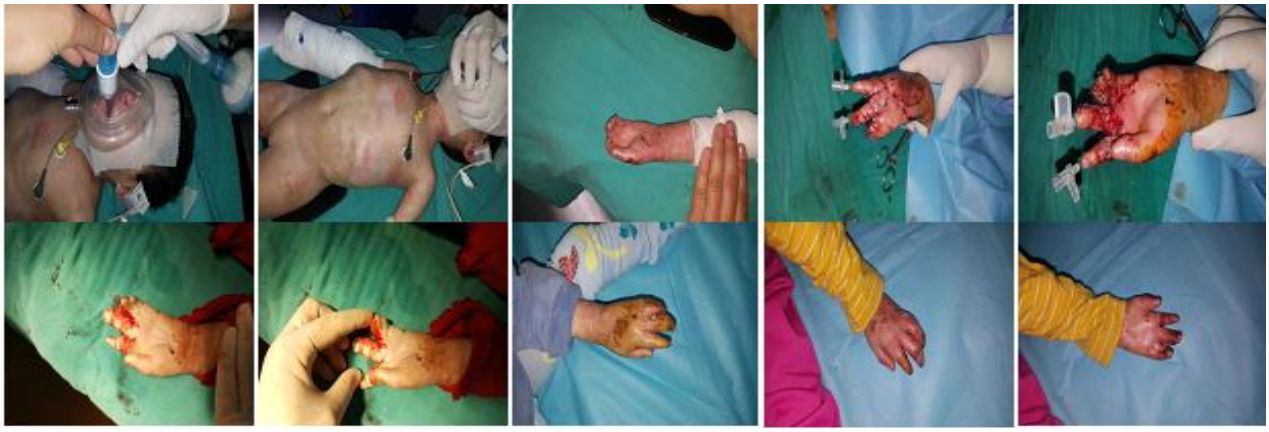

Fig-4: Image showing the management (resuscitation, surgical and results) of our 2nd case

Surgical management consisted in treating syndactylies of the hands, synechiae, skincare as well as the prevention of superinfections of the skin, and the dental care.

DISCUSSION

Hereditary epidermolysis bullosa (HEB) is a heterogeneous group of rare genetic diseases characterized by cutaneous and / or mucosal fragility, localized or generalized [2,3]. Derived from mutations in genes encoding proteins that are mostly involved in the structure and function of the epidermal junction (Nineteen genes are currently identified) [4]. Depending on the level of cleavage in the skin (from top to bottom), simple, junctional and dystrophic forms can be distinguished, to which is added Kindler syndrome [5].

The clinically suspected diagnosis is confirmed by skin biopsy (most centers recommend 3 biopsies) [1] for immunohistological examination. This examination is eventually followed by a genetic analysis [6]. The severity of the pathology is very variable, ranging from

localized forms allowing a near-normal life to rapidly lethal forms.

In generalized severe forms, the extent of wounds, the mucosal involvement, the repeated cicatrizations are sources of multiple systemic complications: undernutrition, pain, joint retractions, chronic inflammatory syndrome, amyloidosis, cutaneous squamous cell carcinoma [7,8]. HEB require multidisciplinary medical management, of which plastic surgery is particularly important for correction of the [1].

No curative treatment of EBH is available to date, the management is to best support patients and their families in daily skin care, prevent, detect and treat complications as soon as possible [9], and empower the patient and family with therapeutic education programs.

Social support is essential to enable community integration at all ages of life. Protein, cell or gene substitution therapies, allogeneic bone marrow [10], cord blood or induced pluripotent stem cell transplantation are therapies under evaluation and are excessively expensive, which will likely represent, in the long term , the main obstacle to their development [1].

A published experiment had shown how deficient skin stem cells were genetically repaired before being grafted onto the child [11]. The experiment consisted of removing the patient's skin cells, including stem cells, and then a healthy gene is introduced into these cells using a virus (the healthy gene replaces the mutated gene in the DNA of stem cells that multiply and transform into skin cells). A surface of 85 cm2 of skin is genetically modified is obtained then grafted on the patient and after 8 months the skin had only the repaired cells [11].

The surgery of our patients has posed specific problems related to mucosal fragility. The cleavage plane is easy since the fingers are surrounded by an epidermal coating. The general anesthesia of the patients has encountered difficulties in terms of the venous route (Venous capital often very small, placement of difficult central catheters, tourniquet setting, fixation of the venous route), intubation (Fragility of the oral mucosa, nasal and ENT, Laryngo-Tracheal Stenosis), and finally the management of per- and postoperative pain.

Deep sedation and local anesthesia should be promoted whenever possible, but all techniques of general anesthesia (intubation, mask) epidural, spinal anesthesia can be envisaged with appropriate precautions.

The expertise of the surgical and anesthetic teams effectively completes the procedure for the management of EBH, especially since adapted equipment (non-disposable silicone knife plates) some patients require several hours of daily dressing with adapted medical devices (silicone dressings).

In most hospitals, the surgical services do not have staff available or trained for such dressings or the necessary equipment. This can pose serious complications in case of emergency surgery. Prevention is about protecting the skin, preventing trauma and any risk of infection as a result of an injury. Air conditioning is a means of prevention in a context of high heat, to limit the drying of the skin.

In light of these data it is concluded that the management of this very rare pathology is multidisciplinary with close collaboration between the pediatric surgeon, the pediatrician, the resuscitator as well as the psychological support as what has been done in our 2 patients.

Our management meets the criteria described in the literature, as well as the surgical management that must be performed in early childhood to prevent recurrence. Unfortunately, our care was delayed because of the low socioeconomic status of our patients, which delayed their access to care, hence the presence of complications, namely synechiae and certain occasions, as well as infections of the skin.

CONCLUSION

Hereditary epidermolysis bullosa is a very serious disabling disease because of infectious, nutritional, neoplastic, cicatricial and functional complications.

The psychological and social impact of the patient and his family is major with difficulties of integration in community. The care is multidisciplinary or heavy in terms of surgery and anesthesia. Prevention prevents complications and recurrences.

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