Chiesi
global rare diseases

KEY FACTS ON Epidermolysis Bullosa (EB)

Introduction

Epidermolysis Bullosa (EB) is an inherited tissue disorder that can cause the skin to be very fragile. In severe cases the skin can be injured just from touch and the wounds may never fully heal.

Currently there is no cure for EB and treatment is heavily based on wound care and pain management.

estimated **500,000**

sufferers worldwide,
many of whom are children

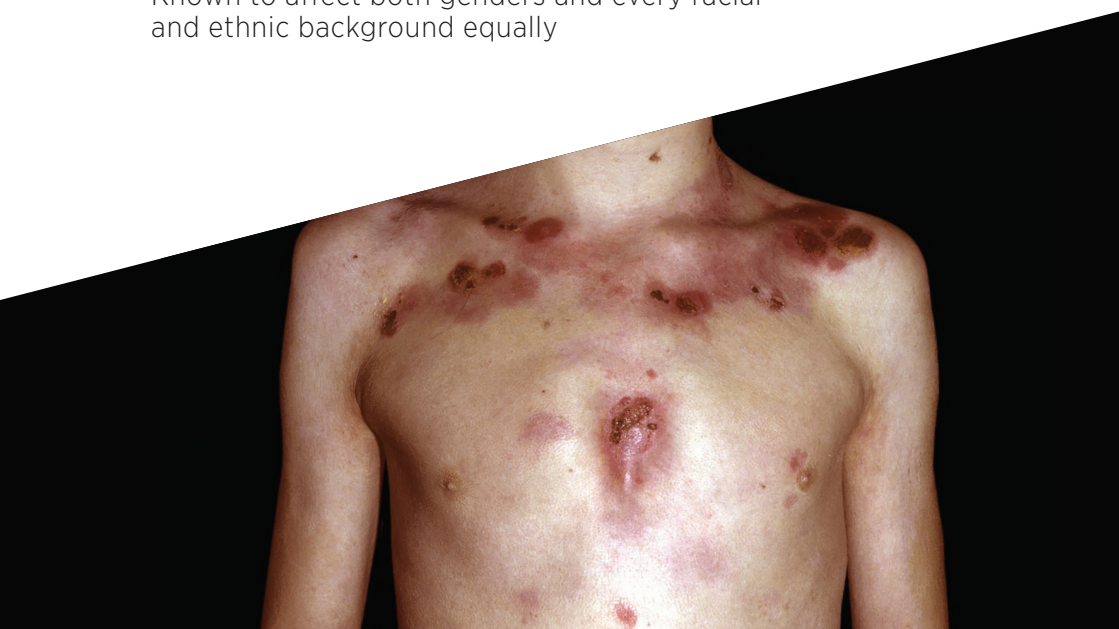
WHAT IS Epidermolysis Bullosa? (EB)

Rare and can be devastating

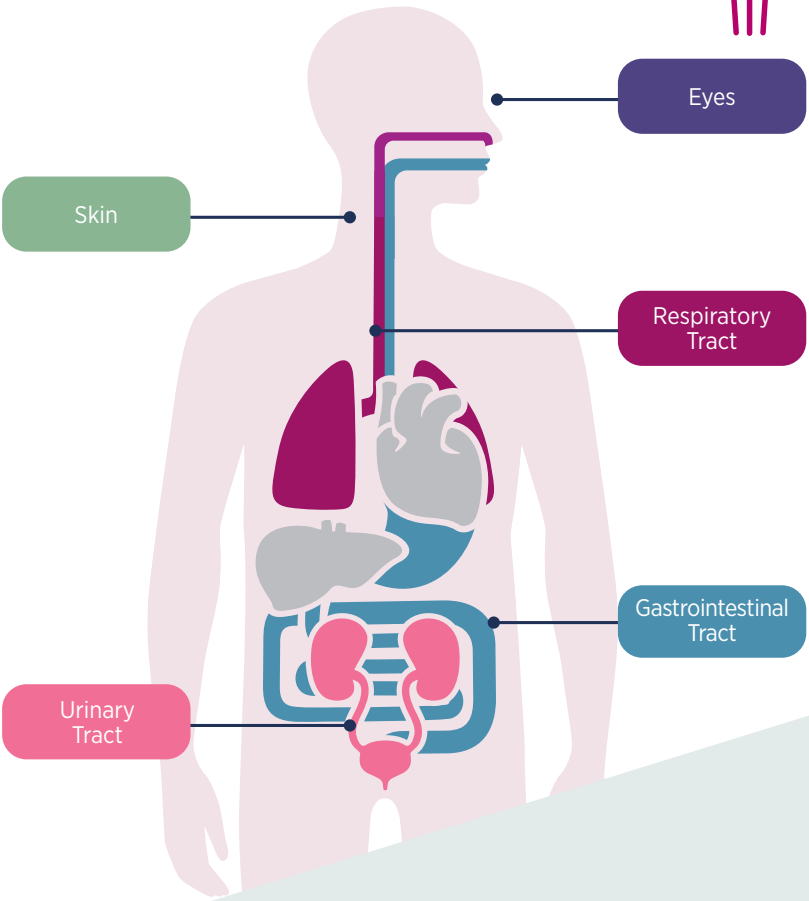
- A rare heterogeneous group of inherited disorders that can be devastating
- Characterised by mechanical fragility of epithelial lined or surfaced tissues, most notably the skin

Genetic

- Comprised of a number of subtypes that vary in clinical presentation as well as in incidence and prevalence
- Known to affect both genders and every racial and ethnic background equally



Body surfaces affected in EB



EB affects not only the externally visible **skin**, but also the **mucous membranes**. The mucous membranes cover internal body surfaces that open to the outside.

Complications associated with severe EB

Joint contractures

Severe, chronic blistering

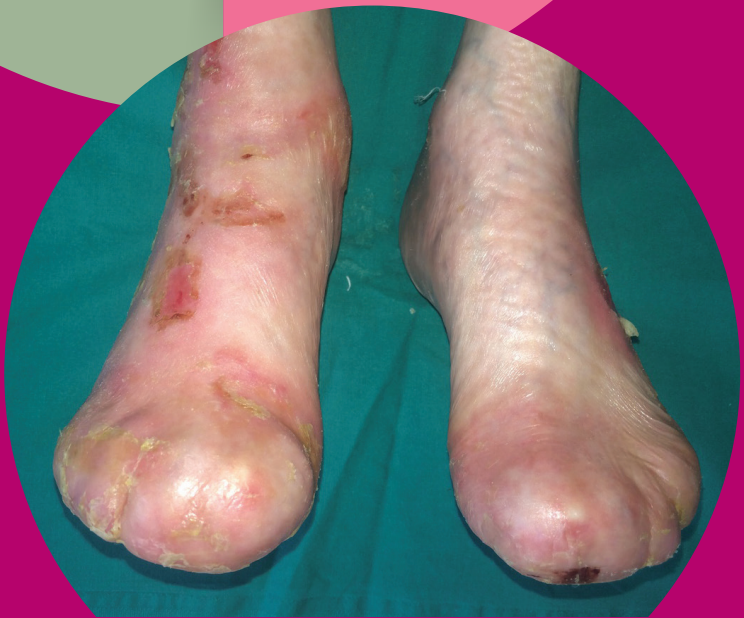
Ulceration and scarring of the feet

Mutilating scarring of the hands and feet

Strictures of the oesophagus and mucous membranes

Risk of premature death

High risk of developing aggressive squamous cell carcinomas and infections



Four EB Sub-Types

Currently, EB is classified into four different subtypes, depending on the location of the abnormal or missing protein in the skin:

1. Epidermolysis Bullosa Simplex (EBS)

EB simplex
(primarily dominant autosomal)

It is the most common EB type, with the majority of mild cases remaining under-diagnosed. Classical presentation of EBS includes:

- Blisters, primarily of the palms and soles
- Blistering is worsened or provoked by trauma and heat
- Blisters heal over a few days with no permanent scarring
- No or very mild mucosal involvement (for example, no mouth blisters)

2. Junctional Epidermolysis Bullosa (JEB)

Junctional EB
(recessive autosomal)

It is the rarest form of EB and characterised by severe blistering all over the body.

The severity varies considerably across the two major subtypes, intermediate and severe, with the latter associated with early death in the first 6-24 months of life.

3.

Dystrophic Epidermolysis Bullosa (DEB)

Dystrophic EB (dominant or recessive)

is caused by a diminished or absent anchoring protein, collagen VII. **Dominant (DDEB)**, which is the second most common form of EB, causes blistering to areas particularly subject to trauma such as the hands, feet, knees and elbows, typically resulting in scarring, milia formation*, and thickening or loss of nails. Mucous membrane involvement, particularly of the mouth and the anal margins, can lead to difficulties with eating and constipation.

Recessive (RDEB) can be one of the most severe types of EB, with extensive and hard to heal blistering and wounding, fusion of fingers and toes, and risk of squamous cell carcinoma (SCC). SCC is by far the most common cause for mortality in RDEB patients with 80% dying as a result of it by the age of 55.

*Milia are tiny white bumps or small cysts on the skin

4.

Kindler Syndrome

Kindler EB
(autosomal recessive)

It is rare, difficult to diagnose and is often confused with other subtypes of EB. Clinical features may include generalised blistering at birth, keratoderma, skin atrophy, poikiloderma, photosensitivity, periodontitis, malabsorption and diarrhoea in early life, and urethral strictures. There is also an increased risk of SCC in later life.

Diagnosis

EB can be a devastating diagnosis for families. Most cases will present at or shortly after birth, however if symptoms are mild, a diagnosis may not be made until later in the patient's life.

Although it will have no effect on the course of the disease, a diagnosis can be useful in establishing the patient's prognosis and offering counselling to the parents if they plan to have more children.

A definitive diagnosis is usually straightforward and most commonly made from analysis of a skin biopsy. Although skin biopsies are useful, they might not always give us an answer. Another method is to do a genetic test using a blood sample from the patient.

Care in EB

EB is a chronic and incurable disease, requiring continual care and monitoring. The principles of care to improve quality of life and minimise complications for all patients includes prevention of skin trauma, relieving symptoms through timely wound management, and the prevention of secondary infections.

However, many forms of EB can be very challenging to manage and require a patient-centred multidisciplinary approach.

The multi-disciplinary team might include a dermatologist, paediatrician and/or neonatologist, pathologist, pain specialist, specialised nurses, psychologist, and nutritionist.

Specific complications may also require involvement from an ophthalmologist, gastroenterologist, dentist, otolaryngologist*, and plastic surgeon.

Wound and skin care

- It is important to regularly undertake careful skin and wound assessment.
- It is recommended that a dressing, such as soft padding, should be applied to prevent blistering, and skin and wound bed damage leading to pain and bleeding on removal.
- Avoid adhesive tape.
- Blisters need careful management as they will extend rapidly if left unchecked. Intact blisters should be lanced at their lowest point to limit tissue damage.
- Different dressings may be appropriate for patients with various EB sub-types.

*ear, nose, and throat specialist

Pain management

Pain can be persistent and severe in EB patients.

Best care practice guidelines recommend that psychological interventions, such as cognitive behavioural therapy (CBT), hypnosis and relaxation training, be used in conjunction with physical and pharmacologic therapies.

Analgesia such as paracetamol and ibuprofen may be sufficient to manage mild pain, while prescription therapies may be needed to treat severe pain and anxiety associated with dressing changes, chronic ulceration, bone pain etc.

Nutritional support

Proper nutrition is crucial for people with EB as it may be compromised due to poor appetite, oral blistering and dysphagia.

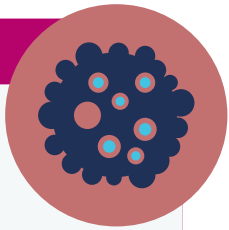
In addition, a diet high in calories, protein and vitamins can aid wound healing and strengthen the immune system to fight infection.

Malnutrition is especially liable in the dystrophic types.

Regular oesophageal dilatations can temporarily improve swallowing when strictures present in patients with dystrophic EB.

Long-term enteral (tube) feeding is recommended in severe EB.

Complications of EB



Chronic wounds should be regularly monitored for changes that may indicate squamous cell carcinoma, which is the most potentially dangerous complication of EB.

Such change often occurs at multiple sites.

A low threshold for biopsy for suspected areas is recommended.

Surgical excision is required, as these lesions tend to be aggressive.

Notes

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