Epidermolysis Bullosa

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- Epidermolysis bullosa is a group of >30 inherited genetic conditions that cause fragile skin.
- The fragility of the skin means it is easily injured, particularly from things like heat, or rubbing, resulting in blisters and erosions.
- Epidermolysis bullosa occurs when there is a problem with the proteins that act to give the skin its usual structure. Gene mutations cause the proteins to be either missing or reduced in number, which results in fragile and easily damaged skin.
- EB is rare, and there is often a family history of the condition.

Types of EB

Epidermolysis bullosa has 4 types. The classification is based on the level at which blistering occurs in the skin, and each may occur on a spectrum of severity within these groups:

- **Epidermolysis Bullosa simplex**

  # This is the most common form of epidermolysis bullosa, and is also the mildest in severity.

  # In this type blisters are formed on the top layers of skin, referred to as the Epidermis

  # Most cases are mild, with intermittent blistering on the hands and feet, more commonly in hot weather

- **Junctional Epidermolysis Bullosa**

Blistering in this type occurs in the dermal-epidermal junction, which is essentially the middle layer of skin

Some forms of this EB are more severe than others.

Herlitz junctional EB is the most severe type of junctional epidermolysis bullosa.

Individuals may be affected over large regions of their body, and blistering can also affect the mucous membranes (for example in the mouth and digestive tract).

Blistering in these areas makes it difficult to eat and digest food, and can lead to malnutrition and slow growth

Nail and teeth abnormalities may also be present

Due to the severity of symptoms, this type of EB has a higher mortality rate, causing death in infancy or early childhood.

- **Dystrophic EB**

This type involves the deepest layer of skin (the dermis), and dystrophic describes the scarring that may occur following healing of the blisters.

Halloppeau-Siemens is one of the most severe forms of dystrophic EB, and is caused by a genetic defect of collagen VII.

In this type of EB even the most minor of skin trauma can cause excessive blistering.

Patients may also suffer from protein loss, anaemia, secondary infections, and hand deformities.

- **Kindler syndrome**

Rare form of EB in which blistering occurs in infancy, but reduces over time.

Signs and Symptoms

- Normally present from birth, or within the first few weeks of life
- Fragile skin indicated by increased tendency to develop blisters and wounds after minor injury, such as a bump to the hand, or friction from shoes rubbing on the feet.
- Most common locations for blisters to occur is on the hands and feet, but may also occur on other parts of the body
- Blisters, wounds, and erosions are slow to heal, and can become infected easily

Treatment

- Specific treatment options will be advised by your general practitioner or dermatologist
- The general aim of management is:
    - Prevention of blistering by reducing trauma and friction, for example wearing appropriate footwear and clothing for protection
    - Identify and treat wounds quickly to decrease risk of infection
    - Treat infections with appropriate antibiotics [topical and/or oral]
    - Gene therapies and bone marrow transplantation are new approaches to treatment that are currently being researched

Tips for blister prevention:

- Dress children in soft clothing
- Turning clothing inside out if the seam is rubbing on the skin
- Using soft padding [such as foam] on surfaces that are prone to cause friction such as chairs, or infant car seats.

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