Epidermolysis bullosa

What is epidermolysis bullosa?

Epidermolysis bullosa (EB) is a group of rare inherited skin fragility disorders which are characterised by blistering of the skin with minor injury.

What causes epidermolysis bullosa?

EB is a genetic disease. So far, mutations in 18 different genes have been identified as causes for EB. These EB genes produce proteins that are responsible for holding the skin and linings of the body together. Mutations in these genes cause the proteins to be weak or even absent resulting in skin fragility and blistering with minor injury.

EB may be inherited in an autosomal dominant pattern from one parent who already has EB. In these cases there is a 50% chance of the affected parent passing this type of EB onto the child. EB can also be inherited in an autosomal recessive pattern, if both parents are carriers for the condition. Parents who are carriers for EB do not have symptoms of the condition. If both parents are carriers for the same type of EB, there is a 25% chance that their child will develop the condition. EB can also occur spontaneously where there is no family history of EB and neither of the parents are carriers for the condition.

What does epidermolysis bullosa look like?

EB usually presents at birth or early in life and is characterised by skin fragility and painful blistering. These skin wounds are frequently inflamed and itchy and may become infected.

The distribution, extent and severity of skin blistering will be dependent on the particular subtype of EB. In milder forms of EB the blistering may be confined only to the hands and feet and usually heals without leaving scars. In the most severe forms of EB extensive chronic wounds can develop which may be life-threatening or heal to leave disfiguring scars and cause significant disability.

Some types of EB can also affect the nails and hair and lead to their damage or loss. The eyes, ears, mouth, teeth, throat, gullet, genitals and urinary tract may also be affected. Damage and scarring to these structures may lead to difficulty with vision, hearing, eating, swallowing, speaking and toileting.

Complications arising from severe EB can also include anaemia, kidney disease, osteoporosis and squamous cell carcinoma (a form of skin cancer).

There are four main types of EB, which are defined by how deep down in the skin the blistering occurs:

- Epidermolysis bullosa simplex (EBS)
- Junctional epidemolysis billosa (JEB)
- Dystrophic epidermolysis bullosa (DEB)
- Kindler syndrome (mixed EB)

Within these main EB types, multiple subtypes are recognised which can affect the skin and body in different ways, leading to different potential complications.

How is epidermolysis bullosa diagnosed?

Proper diagnosis of the EB type is important as each type has different severity and outcomes.

- A detailed family history is taken to check for symptoms of EB.
- The child and both parents are examined for signs of EB.
- A skin biopsy is usually required to look for structural abnormalities in the skin.
- Genetic testing may be performed after skin biopsies have narrowed down the likely affected gene.
- Prenatal diagnosis can be performed where the genetic mutation is already known.

How is epidermolysis bullosa treated?

There is currently no cure for EB. The aim of treatment is to reduce the development of new blisters, promote skin healing and prevent infection. Treatments are tailored to each individual.

Local treatments for the skin include:

- Protecting the skin from damage
    - Gentle handling to avoid damage by rubbing or friction
    - Use of soft clothing and nappies
    - Avoidance of adhesive (sticky) tapes and dressings
- Blister management
    - New blisters should be pierced (with sterile needles) and drained as they arise as this helps to relieve pain and prevents blister enlargement and spreading
- Wound care
    - Bathing should be performed every 1 to 2 days to keep the skin clean and soak off dressings that require changing (adding salt to the bath can help to make them less painful)
    - Specialty non-stick dressings should be applied over any open wounds with secondary absorbent dressings and bandages to aid healing
    - In Australia, these dressings can be accessed through the government-subsidised National Epidermolysis Bullosa Dressing Scheme

Other treatments include:

- Pain management
    - EB blisters and wounds are painful and pain relief is usually required especially during bathing and dressing changes
- Multidisciplinary team
    - Some types of EB can be a multisystem disorder and a collaborative approach to management across a number of different medical and allied health specialities may be required.
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What is the likely outcome of epidermolysis bullosa?

As EB is genetic condition the skin fragility and blistering tendency will persist lifelong.

There are more than 30 different subtypes of EB, all of which have different features, severity and outcomes. This can range from a normal life expectancy with minimal or no disability in the mildest forms

of EB, to being fatal in infancy or severely life limiting with significant disability in the most severe forms of EB.

There is currently no known cure for EB, although research is actively being conducted worldwide to find better treatments for people living with this condition.