Dystrophic epidermolysis bullosa

What is dystrophic epidermolysis bullosa?

Dystrophic epidermolysis bullosa (DEB) is one of the main types of epidermolysis bullosa (EB). This group of inherited skin fragility disorders is characterised by blistering of the skin and body linings with minimal injury.

There are four main types of EB which are defined by the depth at which the skin becomes blistered. There are two main layers of the skin, the epidermis (the very top layer) and the dermis. These two layers are joined at the basement membrane zone. In DEB the skin fragility and blistering occurs below the basement membrane zone (deeper down in the skin compared with the other types of EB). Approximately 20-25% of people with EB have DEB.

What causes dystrophic epidermolysis bullosa?

DEB is a genetic disease. It is caused by a mutation (like a spelling mistake) in one of the genes that makes the proteins that hold the skin together. Mutations in this gene cause the proteins to be weak or even absent, resulting in skin fragility and blistering with minor injury.

DEB may be inherited in either an autosomal dominant or autosomal recessive pattern. Dominant DEB (DDEB) is usually inherited from one parent who already has DEB and is generally regarded as a milder form of DEB. Recessive DEB (RDEB) occurs when both parents are carriers for the condition but do not have symptoms themselves. DEB can also occur spontaneously where there is no family history of DEB and neither of the parents are carriers for the condition.

What does dystrophic epidermolysis bullosa look like?

The different subtypes of DEB can vary greatly in severity. Skin blistering usually appears at birth or the first few weeks of life.

In DEB the blisters and skin wounds tend to heal with scarring and milia (small white lumps). Scarring can lead to significant deformity with joint contractures and fusion of the fingers and toes. The nails often grow abnormally or may be absent. 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 RDEB can also include anaemia, kidney disease, osteoporosis and squamous cell carcinoma (a form of skin cancer).

How is dystrophic epidermolysis bullosa diagnosed?

Correct diagnosis of the EB type is important as each type has different levels of 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 dystrophic epidermolysis bullosa treated?

There is currently no cure for DEB. 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
    - 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
    - DEB blisters and wounds are painful and regular pain relief is usually required

- Multidisciplinary team
    - DEB is often a multisystem disorder and a collaborative approach to management across a number of different medical and allied health specialities is usually required
    - The multidisciplinary team may include dermatologists, specialist nurses, plastic surgeons, dentists, gastroenterologists, dieticians, endocrinologists, pain specialists, psychologists, physiotherapists, occupational therapists, podiatrists, speech therapists and social workers.

What is the likely outcome of dystrophic epidermolysis bullosa?

As DEB is a genetic condition, the skin fragility and blistering tendency will persist lifelong. Scarring to the skin and body linings is progressive and therefore the severity of DEB may increase over time with progressive disability. Milder forms of DEB (usually DDEB) may have a near normal life expectancy, whereas the more severe forms (usually RDEB) may have a reduced life expectancy with a high chance of developing aggressive and life threatening skin cancer at a relatively young age.

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