THE AUSTRALASIAN COLLEGE
OF DERMATOLOGISTS

Epidermolysis Bullosa Simplex

What is Epidermolysis Bullosa Simplex?

Epidermolysis bullosa simplex (EBS) is one of the main types of epidermolysis bullosa (EB). This group of inherited skin fragility disorders is characterised by blistering of the skin 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, which are joined at the basement membrane zone. In EBS, the skin fragility and blistering occurs above the basement membrane zone within the lowest part of the epidermis.

As the level of blistering in EBS is higher up in the skin compared to other forms of EB, it is often regarded as a more mild type of EB. Approximately 70% of people with EB have EBS, making this the most common type of EB.

What causes Epidermolysis Bullosa Simplex?

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

EBS is almost exclusively inherited in an autosomal dominant pattern. A parent with EBS has a 50% chance of passing EBS onto their child with each pregnancy. EBS can sometimes occur spontaneously where there is no family history. In these cases, EBS is due to a new genetic mutation, which occurs around the time of conception.

What does Epidermolysis Bullosa Simplex look like?

Skin blistering may appear at birth or the first few weeks of life, but it may also manifest later in childhood.

In the mildest and most common form of EBS, blistering is usually confined to hands and feet and may not appear until the child starts walking. Blistering is usually causing by rubbing of the skin, especially by footwear. This is usually worse in warm weather. As this subtype of EBS is mild, affected individuals do not usually require a lot of medical assistance.

In the more severe forms of EBS, blistering can occur all over the body and may be complicated by significant skin inflammation and infection. Blisters can also occur inside the

mouth in EBS. Skin blistering usually heals without leaving scars and the hair and nails are usually not significantly affected.

How is Epidermolysis Bullosa Simplex 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 Simplex treated?

There is currently no cure for EBS. 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
- Choice of footwear
    - Soft, well-ventilated shoes are helpful
    - Ensure shoes are fitted well to avoid excess friction on the feet
    - Natural materials rather than synthetic may be better as they allow the skin to breathe
    - Consider insoles to cushion feet
    - Silver-lined socks may help to keep the skin cool
    - Dusting cornflour in the shoes and socks can help to reduce friction and absorb excess moisture
- 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
    - Corn starch can be sprinkled over the blistered area to soak up excess moisture and dry the blisters out
- 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
    - o EBS blisters and wounds are painful and pain relief is usually required when there are active blisters present
- Multidisciplinary team
    - o Some types of EBS can be a multisystem disorder and a collaborative approach to management across a number of different medical and allied health specialities may be required

What is the likely outcome of Epidermolysis Bullosa Simplex?

As EBS is a genetic condition, the skin fragility and blistering tendency will persist lifelong. This fragility and blistering can be worse in the earlier years of life and there may be some improvement in adulthood. EBS wounds usually heal without leaving permanent scars and patients have a normal life expectancy.

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

Further information about Epidermolysis Bullosa Simplex

www.debra.org.au

www.blisters.org.au

This information has been written by Dr Susan Robertson and Professor Dedee Murrell