THE AUSTRALASIAN COLLEGE OF DERMATOLOGISTS

Junctional Epidermolysis Bullosa

What is junctional epidermolysis bullosa?

Junctional epidermolysis bullosa (JEB) 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 (the inner layer of skin). In JEB the skin fragility and blistering occurs through the basement membrane zone that lies between the epidermis and the dermis. Approximately 5-10% of people affected with EB have JEB.

What causes junctional epidermolysis bullosa?

JEB 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 this gene cause the proteins to be weak or even absent resulting in skin fragility and blistering with minor injury.

JEB is almost exclusively inherited in an autosomal recessive pattern. This occurs if both parents are carriers of the condition. These parents are often not aware that they are carriers as they do not show physical signs and there may not be a family history of the condition. The birth of a child with JEB is usually completely unexpected.

What does junctional epidermolysis bullosa look like?

Skin blistering usually appears at birth or in the first few weeks of life. Painful and difficult to heal skin erosions can then develop on the face and nappy areas. Significant ulceration of the mouth, throat and gullet may develop leading to a hoarse voice, poor feeding and failure to thrive. Nails may be absent or grow abnormally. Dental enamel defects and tooth decay are common.

How is junctional epidermolysis bullosa diagnosed?

Correct diagnosis of the EB type is important as each 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 for future pregnancies can be performed where the genetic mutation is already known.

How is junctional epidermolysis bullosa treated?

There is currently no cure for JEB. The aim of treatment is to reduce the development of new blisters, promote skin healing and prevent infection. Treatments are tailored to the 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 or nappy liners
    - Avoiding 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 1-2 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
    - JEB blisters and wounds are painful and regular pain relief is usually required.
- Multidisciplinary team
    - JEB is often a multisystem disorder and a collaborative approach to management across a number of different medical and allied health specialities is usually required.

What is the likely outcome of junctional epidermolysis bullosa?

As JEB is a genetic condition, the skin fragility and blistering tendency will persist lifelong. Babies born with the severest from of JEB may die in the first year of life due to complications from severe blistering of the skin and digestive tract such as malnutrition, anaemia and overwhelming infection. Babies born with the milder form of JEB can expect to live into adulthood but may suffer with lifelong pain and disability.

There is currently no known cure for JEB, although worldwide research is being conducted 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