JUNCTIONAL EPIDERMOLYSIS BULLOSA

What are the aims of this leaflet?

This leaflet has been written to help you understand more about junctional epidermolysis bullosa. It tells you what it is, what causes it, what can be done about it, and where you can find out more.

What is junctional epidermolysis bullosa?

Junctional epidermolysis bullosa (JEB) is a rare inherited (genetic) skin disorder. It is different from the other forms of epidermolysis bullosa (EB), which include epidermolysis bullosa simplex, dystrophic epidermolysis bullosa and Kindler Syndrome. Individuals who have JEB will not develop one of the other types of epidermolysis bullosa at a later date.

The skin of those who have JEB is fragile and minor everyday knocks and friction cause blisters or raw areas. There are 3 main types of JEB (Herlitz JEB, Non-Herlitz JEB and JEB with pyloric atresia). JEB varies in severity, from relatively mild disease with normal lifespan to the most severe form, Herlitz JEB, in which babies may not live beyond their first birthday. JEB is not an infection, it is not contagious and it is not due to an allergy.

What causes junctional epidermolysis bullosa?

The two outermost layers of skin, the epidermis and dermis, are held together by a variety of proteins, known as anchoring proteins. In those who have JEB, the structure of one or more of these anchoring proteins is faulty, weakening attachment of the epidermis to the dermis beneath. When the skin of JEB sufferers is subjected to friction and shearing forces, the epidermis and dermis separate, fluid accumulates in the gap between them and a blister forms. As the same anchoring proteins are found within the respiratory, digestive and urinary tracts, these organs are sometimes affected as well. Weakness of the anchoring proteins is caused by faults (mutations) in the

genes bearing instructions for their assembly. A variety of such mutations have been identified in JEB. Some are common but others are specific to individual families.

Abnormalities have been identified in genes encoding three proteins known as laminin-332 (previously called laminin 5), type XVII collagen and α6β4 integrin.

Is junctional epidermolysis bullosa hereditary?

Yes, JEB is recessively inherited. Everyone has two sets of genes, one inherited from the mother and the second from the father. In recessively inherited disorders such as JEB, if both genes are faulty, no normal anchoring protein can be made and the skin is fragile. If only one of a pair of genes is abnormal, the second is usually able to produce enough normal anchoring protein for the skin to be unaffected. The parents of a child with JEB each have one normal and one abnormal gene but because they have healthy skin, parents are usually unaware that they carry a faulty gene until after an affected child is born. Each time parents of an affected child conceive, there is a 1 in 4 chance that the new baby will have fragile skin. Both males and females can be affected.

It is very unlikely that those with JEB will themselves have affected children. This can only happen if the sufferer's partner also has a fault in one of the genes controlling attachment of the epidermis to the dermis. The risk of this occurring is very small indeed.

As other more unusual inheritance patterns are possible, detailed genetic testing may be necessary before genetic counselling can be offered.

What are the symptoms of junctional epidermolysis bullosa?

Blisters usually start to appear within hours or days of birth. Many babies with JEB have a characteristic hoarse cry. Painful blisters or open sores develop at sites of minor trauma to the skin. The inside of the mouth is often affected, causing pain during feeding and tooth brushing. As JEB can affect dental enamel, tooth decay occurs if dental care is neglected. Blisters can affect the surface of the eye, causing pain but leaving vision unaffected. Persistent vomiting during the early weeks of life may indicate narrowing of the outlet of the stomach which occurs in some forms of JEB. Less frequent problems include bladder and kidney disorders and difficulty passing urine.

Babies with the most severe forms of JEB are generally very unwell.

What does junctional epidermolysis bullosa look like?

The blisters vary in size, are fragile and rupture easily leaving raw moist areas which are difficult to heal. The most troublesome areas in JEB are usually on the face and legs. Occasionally, a newborn baby may have an area of missing skin especially on the face. Some sufferers of JEB have sparse scalp hair. Nails may be normal but sometimes they are absent or become thickened, or chronically inflamed. Usually, JEB does not cause scarring but some variants do leave small pinkish-purple scars at sites of healed blisters. Scars gradually become more subtle with time. Sometimes small, white pin-head sized spots, known as milia, arise on areas of skin where previous blisters have healed. Skin cancer may develop in adult patients with chronic non healing wounds. Regular examinations by a dermatologist are important to ensure that skin cancer is detected and treated at an early stage.

How will junctional epidermolysis bullosa be diagnosed?

Specialised investigations are usually necessary to make the diagnosis of JEB and to distinguish the different subtypes. Different types of EB may look very similar during the early months of life.

Investigations usually involve removing a small piece of skin from the affected baby. This is a simple procedure involving an injection of local anaesthetic into the skin. The skin sample will then undergo a number of detailed tests. Blood samples will be taken from the affected baby and if possible from both parents for genetic analysis. Pre natal diagnosis is also possible for families in whom the causative genes have been identified.

Can junctional epidermolysis bullosa be cured?

In the past 20 years, there has been exciting and rapid progress in the understanding of JEB but at the moment it cannot be cured. Several laboratories around the world are exploring strategies which they hope will ultimately lead to a cure.

How can junctional epidermolysis bullosa be treated?

As blisters can be induced by even the most gentle skin contact, affected babies need careful handling, but inevitably, some blisters and raw areas will

still occur. Special feeding techniques are often necessary to avoid blisters developing on the lips or inside the mouth.

The obstetric and paediatric teams caring for the newborn baby will contact the dermatologists and DEBRA nurses who will organise appropriate investigations and will demonstrate how to care for the baby's skin.

Blisters should be burst with sterile needles and antiseptic creams should be used to prevent infection. Special non-stick dressings are available to protect the skin. Conventional sticky tapes and plasters must be avoided as these will tear the skin when they are removed. Careful choices of clothing and lifestyle, to reduce friction and protect vulnerable areas of skin, will help reduce the number of new blisters. If the eyes are affected, simple lubricant ointments are helpful. Adequate pain relief is important.

Self Care (What can I do)?

- A healthy diet, regular dental checks (as soon as the first teeth appear), and careful skin care are important.
- If you have an affected child, tell the teachers about JEB and make sure they understand that your child may not be able to take part in some of the more physical activities of the school curriculum.
- If any form of surgery is necessary, it is important to warn the surgical team that their usual dressings and skin care regimes are unsuitable for JEB sufferers. They should also be warned that careful handling is required if an affected person is to be lifted. You should discuss your skin problems well before the date of a planned operation.
- It can be helpful to carry a special wallet-sized information card giving details of JEB and the relevant precautions to be taken should you or your child need help in an emergency. These cards are available from DEBRA.

Where can I get more information about junctional epidermolysis bullosa?

Advice and practical support for epidermolysis bullosa sufferers is available from DEBRA. This charity also funds epidermolysis bullosa research projects and produces a regular magazine with up to date information about new developments. DEBRA’s specialist nurses can visit people in their own homes and in hospital to demonstrate skin care regimes and they are available for telephone advice. If necessary, the nurses will visit schools to talk to staff and students. DEBRA staff can also offer advice and practical support to those applying for a disability living allowance and help with mobility.

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Other useful websites:

http://www.ncbi.nlm.nih.gov/books/NBK1125/
http://ghr.nlm.nih.gov/condition/junctional-epidermolysis-bullosa

For details of source materials used please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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