PATIENT INFORMATION LEAFLET

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 not an infection; it is not contagious, and it is not due to an allergy. It is different from the other forms of epidermolysis bullosa (EB), which include epidermolysis bullosa simplex, dystrophic epidermolysis bullosa and Kindler epidermolysis bullosa. Individuals who are affected by JEB will not develop one of the other types of epidermolysis bullosa at a later date.

The skin of those who are affected by JEB is fragile. Minor everyday knocks and friction, such as rubbing or scratching, cause blisters or raw areas. There are three main sub-types of JEB: severe, or Herlitz, JEB; intermediate, or non-Herlitz, JEB and JEB with pyloric atresia. JEB varies in severity. It can be relatively mild, and the person has a normal lifespan, or in the most severe form, babies may not live beyond their first birthday.

WHAT CAUSES JUNCTIONAL EPIDERMOLYSIS BULLOSA?

The two outermost layers of the skin, called the epidermis and the dermis, are held together by a variety of proteins, known as anchoring proteins. In individuals who are affected by JEB, the structure of one or more of these anchoring proteins is faulty. This weakens the attachment of the epidermis to the dermis beneath. Our skin is routinely subjected to friction and shearing forces (when two surfaces rub together, such as the skin rubbing against the bed linen). When there is friction in people who are affected by JEB, the epidermis and dermis separate, fluid accumulates in the gap between them and a blister forms. The same anchoring proteins are also found in the respiratory, digestive and urinary tracts, so these organs are sometimes affected as well. Weakness of the anchoring proteins is caused by faults (mutations) in the genes carrying the instructions to keep them attached . 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 an autosomal recessive inherited condition. Everyone has two sets of genes; one set is inherited from their mother and the second from their father. In recessively inherited disorders such as JEB, a faulty gene from both parents will cause abnormal anchoring protein and the skin will be fragile. If only one gene from either parent is abnormal, then the second, good gene from either parent, is usually able to produce enough normal anchoring protein for the skin to be unaffected. The parents of a child affected by JEB will each have one normal and one abnormal gene. This means they will have healthy skin and 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 (25%) chance that their new baby will have fragile skin. Both males and females can be affected.

In individuals affected by JEB, the risk of having affected children is very small. This can *only* happen if the affected person's partner also has a fault in the same gene making the anchoring protein – such risk is extremely small.

As a number of different genes may give rise to JEB, 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 affected by JEB have a characteristic hoarse dry cry. Painful blisters or open sores develop at areas of minor trauma to the skin. The inside of the mouth is often affected, causing pain during feeding and tooth brushing. As JEB weakens teeth enamel, tooth decay is common. Brushing should be encouraged with a soft toothbrush and fluoride supplements may be prescribed. The teeth may be very sensitive to extremes of temperature so give tepid fluids and food. Blisters can affect the surface of the eye, causing pain. Sometimes this affects vision. To keep the eyes moist and reduce the possibility of blistering eye drops or ointments may be prescribed.

JEB with pyloric atresia causes a blockage (obstruction) of the lower part of the stomach (the pylorus). This prevents food from emptying out of the stomach into the intestine. Persistent vomiting during the early weeks of life may indicate narrowing of the outlet of the stomach or blockage. Less frequent problems include bladder and kidney disorders and difficulty passing urine.

Babies affected by the most severe forms of JEB are generally very unwell, struggle to gain weight and are unable to grow normally.

WHAT DOES JUNCTIONAL EPIDERMOLYSIS BULLOSA LOOK LIKE?

The blisters vary in size. They are fragile and burst 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, hands or feet. Some individuals affected by JEB have sparse scalp hair. Nails may be normal but are sometimes absent or become thickened, or chronically inflamed. Usually, JEB does not cause scarring, but some variants do leave small pinkish-purple scars where blisters have healed. Scars gradually become less obvious with time.

It is possible, though not common, for adult patients affected by chronic non-healing wounds to develop skin cancer. Regular examinations by a dermatologist are important to ensure 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 of epidermolysis bullosa (EB). 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 area, known as a biopsy. 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. When prospective parents know that they carry

the JEB gene, the unborn baby can be tested to see if it will be affected.

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 caused by a gentle skin contact, affected babies need careful handling, such as when picking baby up, 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.

DEBRA is a charity set up to help people affected by epidermolysis bullosa conditions such as JEB. The obstetric and paediatric teams caring for the newborn baby will contact the dermatologist and EB nurses. These centres are familiar with caring for babies and children affected by EB. They can organise appropriate investigations and will demonstrate how to care for the baby's skin.

Blisters should be burst with a sterile needle and antiseptic creams may 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/rubbing and protect vulnerable areas of skin, will help reduce the number of new blisters. If the eyes are affected, regular checks by an ophthalmologist are very important to ensure the eyes are healthy. Eye drops, antibiotics and/or pain relief medication may be prescribed as and when necessary.

SELF-CARE (WHAT CAN I DO)?

- Follow the practical care advised by the specialist EB dermatologist and nurses.
- A healthy diet, regular dental checks (as soon as the first teeth appear), and careful skin care are important.
- 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 alert the surgical team that their usual dressings and skin care methods are unsuitable for those affected by JEB. They should also be warned that careful handling is required if the affected person is to be lifted. You should discuss your skin problems well before the date of a planned operation and remind the team just before the 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. Some people wear a medical alert bracelet.

WHERE CAN I GET MORE INFORMATION ABOUT JUNCTIONAL EPIDERMOLYSIS BULLOSA?

Advice and practical support for individuals affected by EB 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. Specialist EB nurses are available for telephone advice. They can visit people in

their own homes and in hospital to demonstrate skin care methods . 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.

DEBRA
The Capitol Building
Oldbury
Bracknell
Berkshire RG12 8FZ
Tel: 01344 771961
Web (DEBRA-UK): www.debra.org.uk
Web: www.debra-international.org

Other useful websites:

http://www.ncbi.nlm.nih.gov/books/NBK1125/

http://ghr.nlm.nih.gov/condition/junctional-epidermolysis-bullosa

https://www.gosh.nhs.uk/medical-information-0/generalised-severe-junctional-epidermolysis-bullosa-eb/

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

BRITISH ASSOCIATION OF DERMATOLOGISTS

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