

Junctional Epidermolysis Bullosa Information for Parents

Epidermolysis bullosa is the name given to a large group of inherited blistering disorders.

There are many different types of epidermolysis bullosa (EB) but the majority come into one of the three main types:

simplex

dystrophic

junctional

The common factor in all types of EB is the fragility of the skin and its tendency to blister and shear away in response to minimal everyday friction and trauma. This is because some of the sticky proteins which give the skin its strength to resist damage are absent or reduced.

Within each type of EB there is a wide scale of severity.

There are several different types of junctional EB (JEB). These are Herlitz, non-Herlitz and junctional EB with pyloric atresia.

Herlitz JEB is usually the most serious type. Sadly, many children die within the first few weeks or months of life, with the majority not surviving beyond their first birthday.

Blistering on the voice box (larynx) can result in breathing difficulties and problems within the digestive tract often reduce the absorption of food leading to poor weight gain and eventually to loss of weight.

Non-Herlitz JEB tends to be a milder form than Herlitz, although many similarities are present in the young infant. In the older child or adult blistering and ulceration is troublesome on the shins and scalp.

Junctional EB with pyloric atresia can also carry a poor prognosis and many of those affected die in infancy. In addition to skin fragility the babies have an intestinal blockage requiring immediate surgery. A small number of children who have junctional EB with pyloric atresia survive, sometimes with minimal skin problems.

What has caused your baby to have junctional EB?

Junctional EB is a recessively inherited genetic disorder. In order to have a child with a recessive disorder such as JEB, both parents must be carriers. Every one of us has two copies of every gene, one inherited from their mother, and the other from their father. As long as one of those copies is normal the person will have no symptoms of the disease. Therefore a person with one faulty copy of the gene is healthy and is said to be a carrier of the disorder.

However, if 2 people with a faulty copy of the gene have a child and the child inherits two faulty copies, the child will be affected with the recessive disorder.

Every time two parents who are carriers of a faulty gene for junctional EB have a child, there is a 1:4 chance that the child will be affected.

Unfortunately, at present it is not possible to test everyone to see if they are a carrier of the defective gene and we only know parents are carriers when they have a baby with junctional EB.

How is EB diagnosed?

When a baby is born with blistered skin or shows signs of skin fragility the diagnosis of EB is suspected and a skin biopsy is taken. A small piece of skin is taken using local anaesthetic and examined in a specialist laboratory. This is a complex test and you may have to wait for several weeks before the results of the test become available.

Is there a cure for junctional EB?

Sadly, at this time there is no cure available. However, scientists all over the world are working towards gene therapy treatments, which will one day be available to help correct the faulty genes and strengthen the skin.

Caring for your baby

Caring for a baby who has junctional EB can be very hard. It is important that you make use of any available help from your GP, community nurses, health visitor and hospital staff.

Once you have got to know the people involved you can make as much or as little use of the support network as you wish.

You must not view this option with a sense of personal failure or feel guilty if you are not the sole carer for your baby all of the time. Family life must be

considered and you may also need to spend time with other children. Staff at DEBRA can give non-medical advice together with social and emotional support. You may wish to be put in contact with the parents of other children who are similarly affected and this can be arranged through DEBRA.

You will quickly learn how to handle your baby. Pick the baby up by rolling him away from you onto his side. Place one hand behind the head, and the other under the bottom. Allow the baby to roll back onto your hands and lift up. Use this method as an alternative to sliding your hands under the baby or lifting him up under the arms and risking damage to the fragile skin. If you are anxious about hurting the baby, or friends and family want to hold him, place the baby on a small pillow or mattress and carry him on this. This makes it easier to transfer the baby from one person to another. Remember that however careful you are there will always be new blisters; so do not blame yourself or other carers.

Feeding

Often the baby will have sores and blisters inside the mouth. This can make feeding more difficult but most babies are able to breast or bottle-feed successfully. Always moisten the teat with cooled boiled water as a dry teat can stick to the blisters. It will also help to apply Vaseline to the lips before every feed. If the mouth is very sore teething gels can be applied to the teat, which will act as a local anaesthetic to make feeding more comfortable.

Care of the skin

The blisters are not self-limiting and will grow if left. We recommend you lance the blisters with a sterile needle as soon as they appear. Any open wounds require dressings, which are usually changed every few days. The specialist DEBRA nurses will train your community nurses how to dress the wounds and you can participate in this as soon as you feel able. In our experience parents quickly become the experts!

Are the blisters and wounds painful?

If the baby appears to be in pain or discomfort then pain-relieving medicines can be given.

Sometimes these are needed just for dressing changes, if the baby is comfortable throughout the day. If the baby is in pain between the dressing changes then medicines can be given on a regular basis.

Prenatal testing

Testing is available in future pregnancies to see if the baby has junctional EB. At present there are two techniques used to assist prenatal diagnosis.

If the specific mutations have been identified from analysis of the DNA, diagnosis can be made from analysis of a chorionic villous sample (a small piece of the placenta) taken after the 10th week of the pregnancy.

The sample is taken using guidance by ultrasound imaging and the route used is either via the vagina or through a fine needle passed through the abdominal wall. The genetic material is extracted from the sample and compared with that of the affected child and his parents.

Not all the mutations responsible for junctional EB have been identified. If the specific mutations have not been found in a particular family, then prenatal diagnosis can be made from analysis of a foetal skin biopsy.

Using local anaesthetic, a hollow needle is introduced through the mother's abdomen and into the amniotic cavity. Forceps are then passed down the needle to remove tiny pieces of skin from the baby.

This test is performed after the 15th week of the pregnancy.

Both tests carry a small risk of miscarriage, approximately 1%.

A booklet for professionals caring for children with junctional EB is available from DEBRA.

When you feel ready to learn more, please ask your nurse to explain the information to you.