



Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Junctional epidermolysis bullosa (EB) with pyloric atresia

This information sheet from Great Ormond Street Hospital (GOSH) explains about junctional epidermolysis bullosa with pyloric atresia and how it can be managed. It also contains suggestions for making everyday life more comfortable and contact details for a support organisation.

Please note: This information sheet suggests some commercial products that can be helpful in managing junctional epidermolysis bullosa with pyloric atresia – inclusion in this information sheet does not constitute endorsement by GOSH and alternative products may be available.

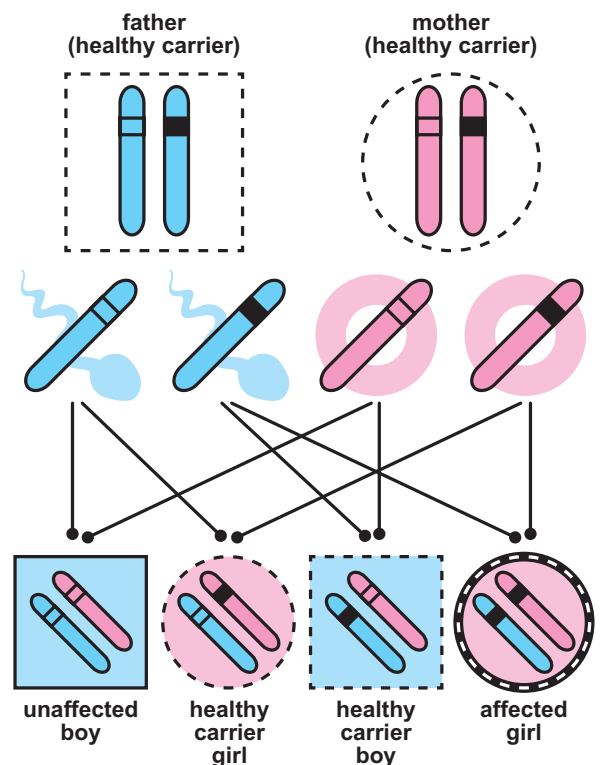
Epidermolysis bullosa (EB) is the term used to describe a number of rare genetic conditions which cause the skin to blister and shear in response to minimal friction and trauma. There are four broad categories of EB: EB simplex, junctional EB, dystrophic EB and Kindler syndrome. Within each of these categories there are several different subtypes.

What causes junctional EB?

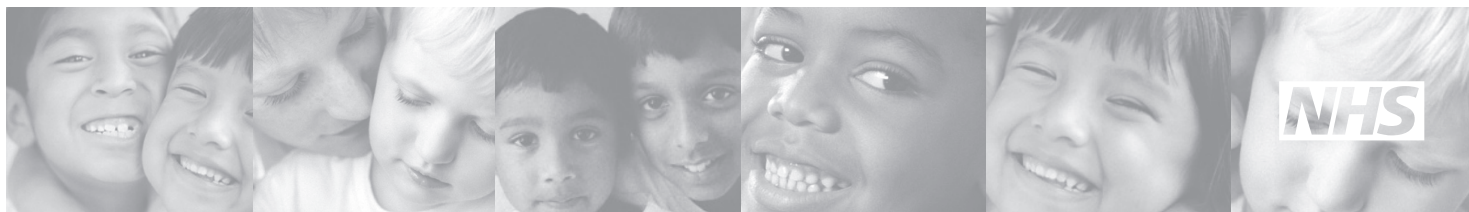
We have two copies of every gene. If somebody has a one faulty copy and the other is unaffected they are a carrier of the condition, in this case a carrier of junctional EB. If their partner is also a carrier for junctional EB there is a 25 per cent (1 in 4) risk in every pregnancy that the baby will inherit both faulty copies of the gene and therefore have junctional EB.

There are three main types of junctional EB:

- Junctional EB generalised severe (previously called Herlitz junctional EB)
- Junctional EB generalised intermediate (previously called Non-Herlitz junctional EB)
- Junctional EB with associated pyloric atresia



All forms of junctional EB reflect mutations (new changes) in genes which help to bind the different components of the skin together. This information sheet is only about Junctional EB with pyloric atresia. Information about the other types of Junctional EB are available on the GOSH website at www.gosh.nhs.uk.



Junctional EB with pyloric atresia (JEB PA)

This is a rare type of junctional EB that affects the skin, digestive and urogenital tracts (used to pass urine). It is caused by faults in special proteins called alpha-6-beta-4 integrins. These proteins help to attach cells to their surroundings, making the skin strong. In addition to the skin, these integrins are found in the lining of the gut and urogenital tracts. These proteins are therefore very important and any faults in them result in weaker skin susceptible to blisters and wounds and problems with digestion and passing urine. Babies with JEB PA are often born prematurely so may have added complications.

Affected babies are often born with missing areas of skin typically over their limbs and blisters soon develop resulting from friction and handling. The inside of the mouth is also affected and blisters and sores develop.

Babies with JEB PA are also born with pyloric atresia, which is a blockage to the lower part of the stomach. This prevents the stomach from emptying. This needs to be repaired by an operation shortly after birth.

Sometimes babies are also born with malformations of the bladder, kidneys and ears.

How is EB diagnosed?

In the majority of cases, the type of EB can be determined by analysis of skin biopsy (tiny sample of skin) under a microscope. Blood samples are also taken from the child and parents to look for the specific gene changes.

Is there a cure or treatment?

Sadly at present there is no cure for EB. Research is progressing towards finding treatments to correct the faulty proteins responsible, but present treatment is to manage the symptoms.

How serious is JEB with pyloric atresia?

This type of junctional EB is often very serious. As the symptoms are so serious, many babies die within the first few weeks or months of life despite successful repair of the pyloric atresia.

In those who have a less serious type, the condition may improve over time and later in life, this group have minimal skin problems. Many will be affected by serious problems with their urogenital tract and lining of the bladder leading to difficulty passing urine. This problem can require a lot of medical intervention and sometimes surgery.

Practical care

Handling

Nurse your baby on a small soft mattress which will be provided by your EB nurse. To lift the baby from the mattress, apply a 'roll and lift' technique to avoid damage from friction and shearing forces – roll the baby on their side, place one hand behind their head, the other under the bottom, allow them to roll back onto your flat hands and lift. As you become confident in handling you may not need the mattress.

Blisters

Blisters must be lanced with a hypodermic needle to prevent them from spreading. The roof should be left on the blister.

Nappy area care

Cleaning with water can sting the blisters and sores and therefore we recommend cleansing with 50/50 ointment or Emollin® emollient spray. Blisters should be lanced as above. A layer of barrier cream such as Proshield® Plus barrier cream, reapplied at each nappy change, should protect blistered and vulnerable areas. Open wounds are covered with the dressing Intrasite® Conformable. The nappy is lined with a soft material such as Conti® SuperSoft (available on prescription) to cover the edges of the nappy to prevent friction.



Bathing

If there is extensive skin loss at birth we recommend delaying bathing until healing has taken place. This is because bathing can be a painful process and it is difficult to protect the baby from further skin damage when all the dressings are removed at once.

Many babies enjoy bathing once the initial wounds are healed. Give prescribed painkillers before bathing and have the replacement dressings pre-cut ready to put in place.

A mild antiseptic such as Octenisan® as prescribed by your EB team can be added to the water instead of commercial baby products. If you find it difficult to hold your baby in the bath then use a towelling baby seat rather than a plastic one.

If there are a lot of dressings it may be easier to bathe the baby with the dressings on and change them after bathing. Pat skin dry with a soft towel rather than rubbing.

Clothing

Initially, a front fastening baby grow is ideal but unless the seams are flat it will need to be worn inside out to prevent the seams from rubbing and causing blisters. Care must be taken to ensure the fasteners do not rub. Older children can wear ordinary clothes if they are easy to put on and take off. Choose clothes which do not have to go over the head and without prominent seams.

A range of garments is available on prescription via your family doctor (GP) to help keep dressings in place. Skinnies® garments and Skinnies® WEB garments (www.skinnies.co.uk) are particularly useful. Other garments include the Tubifast® range.

Wound care

Open wounds must be dressed to encourage healing and prevent adherence to clothing. Some dressings, although described as 'non-adherent' and suitable for those with other types of EB, may cause blistering or extension of the wound. Suitable dressings include PolyMem® (Ferris) and Intrasite® Conformable (Smith & Nephew) which provides protection

for the skin and encourages wound healing. The dressing is applied directly to the skin and secured with a small piece of tubular bandage such as Tubifast®. Intrasite® Conformable will need to be changed at least once a day and more frequently in hot weather to prevent it drying out and adhering to the wound. PolyMem® is changed daily or when staining is seen on the outside of the dressing. Urgotul® (Urgo) is a wound contact layer which can be used under the Intrasite® Conformable and PolyMem® if the dressings are sticking. Other dressings which can be used over dressings such as Urgotul® include Mepilex® Lite, Mepilex® Transfer and Mepilex® (Molnlycke Healthcare). Your EB team will advise you on the appropriate dressing.

As the barrier function of the skin is reduced, some wounds become infected. These can be treated with creams and ointments or specialised dressings. If your child is unwell with fever, contact your GP as treatment with antibiotics may be needed. Please ask your GP to take a wound swab before prescribing antibiotics.

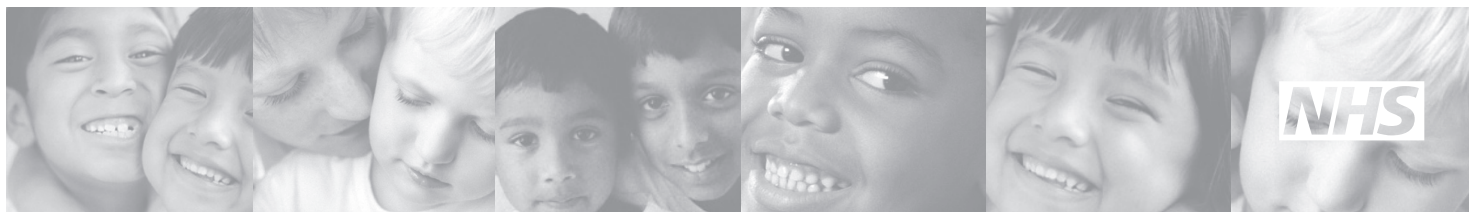
If products accidentally adhere to the skin, or dressings become stuck, a Silicone Medical Adhesive Remover such as Appeel® (CliniMed), will remove the product painlessly and without causing further damage.

Wounds may look deep red, lumpy and bleed easily. This is called over-granulation tissue, which is very common in babies and children with junctional EB. A steroid cream can be applied to reduce this.

Nutrition

Infants may be reluctant to feed due to soreness from blisters in the mouth. A Haberman® (Special Needs) feeder is often helpful. Application of teething gels to the teat or directly to the mouth prior to feeding or a preparation such as Gelclair® further reduces pain. Breastfeeding is often possible although the face may need to be protected with a layer of emollient to reduce friction from rooting.

Due to increased nutrition needs from wound healing and sometimes poor



absorption of feeds, additional protein and calories may be prescribed under the guidance of a specialist dietitian.

Constipation is a common problem with all types of EB, often resulting from blistering and soreness around the bottom. Constipation may be further compounded by side effects from pain medicines. Treatment with laxatives may be needed.

Pain management

Blisters and wounds can be painful but children's need for pain relieving medicines varies greatly.

Pain management is complex and needs constant readjustment. Your EB team will help with this and will refer to a specialist paediatric pain team if extra assistance is needed.

Teeth

The enamel on the teeth is poorly developed in children with junctional EB. Brushing with a soft toothbrush should be encouraged and fluoride supplements may be prescribed. The teeth may be very sensitive to extremes of temperature so give tepid fluids and food if this is the case. Regular assessment by a specialist dentist experienced in EB care will ensure optimal preventative care and treatment is given.

Eyes

Blistering of the surface of the eye can be a problem. Rubbing the eyes can trigger this painful condition, but often results from dry eyes caused by a reduced tear film. Eye drops and ointments prescribed by an ophthalmologists experienced in the care of children with EB can help keep the eyes moist and reduce the incidence of blistering. Once an abrasion has occurred, the lubricants should be replaced with antibiotic eye drops. Abrasions in the eye are painful and pain-relieving medicines should be

given whilst waiting for assessment by the ophthalmologist. The child may prefer to keep their eyes closed and avoid bright light while the abrasion is healing.

Is it possible to test for EB before birth?

In most cases, prenatal testing is available for parents who are known to be carriers of junctional EB. The test is called chorionic villous sampling (CVS) and involves a small piece of the placenta being taken for genetic testing after the 11th week of pregnancy. It may also be possible to test before pregnancy is established using IVF techniques and testing the embryos before they are implanted.

Both tests are only possible if specific gene changes have been identified. In a very small minority where there is insufficient information to interpret the test, a later test (after the 15th week of pregnancy) may be offered. This involves analysis of a small piece of skin taken from the baby.

Further information and support

EB team at Great Ormond Street Hospital (GOSH) – 020 7829 7808

DEBRA is the national charity that supports people living and working with Epidermolysis Bullosa (EB) – a painful genetic skin blistering condition. DEBRA is a registered charity in England and Wales (1084958) and Scotland (SC039654).

DEBRA provides information, practical help and support to anybody living or working with EB, including individuals, families, carers and healthcare professionals, and funds research into the condition. To find out more about how DEBRA can support you please visit www.debra.org.uk.