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

Junctional epidermolysis bullosa (EB) generalised intermediate type

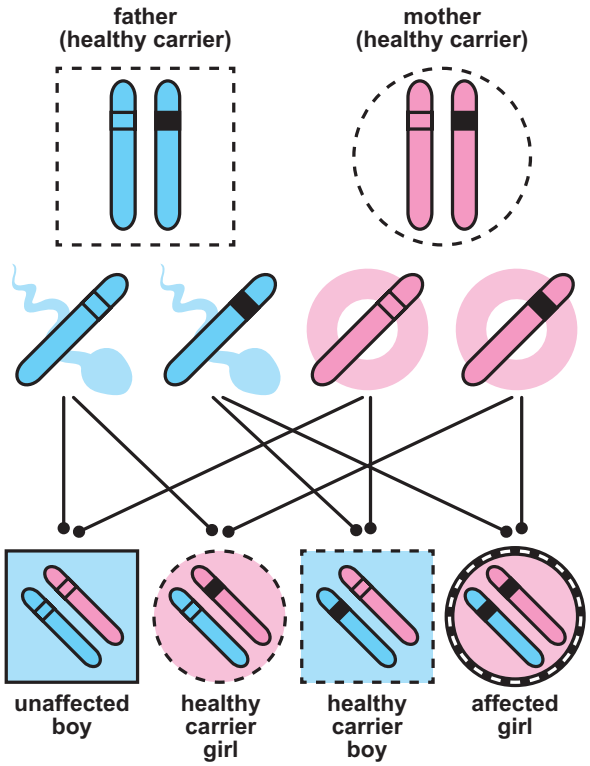
This information sheet from Great Ormond Street Hospital (GOSH) explains about junctional epidermolysis bullosa generalised intermediate type and how it can be managed. It also contains suggestions for making everyday life more comfortable and contact details for a support organisation.

Please note: This information sheet suggests some commercial products that can be helpful in managing junctional epidermolysis bullosa generalised intermediate – 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 said to be a 'carrier' of the condition, in this case a carrier of junctional EB. They do not usually have any symptoms of the condition. 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 generalised intermediate. Information about the other types of Junctional EB are available on the GOSH website at www.gosh.nhs.uk.

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Ref: 2016F1950

Junctional EB generalised intermediate

Most types of junctional EB intermediate result from a defect in important proteins called laminin 332 and type XVII collagen. Laminin 332 is made from three glue-like proteins, which help to stick the layers of the skin together. In addition to the skin, type XVII collagen is present in the lining of the mouth, the surface of the eye, upper oesophagus (swallowing tube) and lining of the bladder. These organs can therefore be affected in individuals with junctional EB.

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 generalised intermediate?

The majority of those affected survive to adulthood and some into old age. However, symptoms can be severe in the newborn baby and a lot of supportive care required.

The main longer-term problems include skin fragility leading to continual blistering and wounds, damage to the eyes and teeth and extra nutritional needs.

Wounds may remain a lifelong problem and areas of previous wounding can become permanently damaged and scarred. Thickened finger and toe nails and hair loss (alopecia) are common in older children and adults.

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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, which is 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 if needed 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 from 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

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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 it often results from dry eyes caused by a reduced tear film. Eye drops and ointments prescribed by an ophthalmologist 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 while waiting for assessment by the ophthalmologist. The child may prefer to keep their eyes closed and avoid bright light while the abrasion is healing.

Later complications

Wounds may become chronic (non-healing). These are typically over the shins and scalp.

Some older children and adults suffer from hair loss (alopecia) over scarred areas of their scalp. Avoid tying back long hair tightly as this can encourage hair loss.

More serious complications

There is an increased risk (up to 25 per cent) of developing a type of cancer called squamous cell carcinoma after the age of 25. It is important that all the skin is examined regular by an EB specialist to detect these early so treatment can be offered.

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.

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Compiled by the GOSH Epidermolysis Bullosa department and DEBRA in collaboration with the Child and Family Information Group at GOSH.

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