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

Generalised severe junctional epidermolysis bullosa (EB)

This information sheet from Great Ormond Street Hospital (GOSH) explains about generalised severe junctional epidermolysis bullosa (previously called Herlitz junctional EB) and how it can be managed. It also contains suggestions for making everyday life more comfortable and contact details for further information and support.

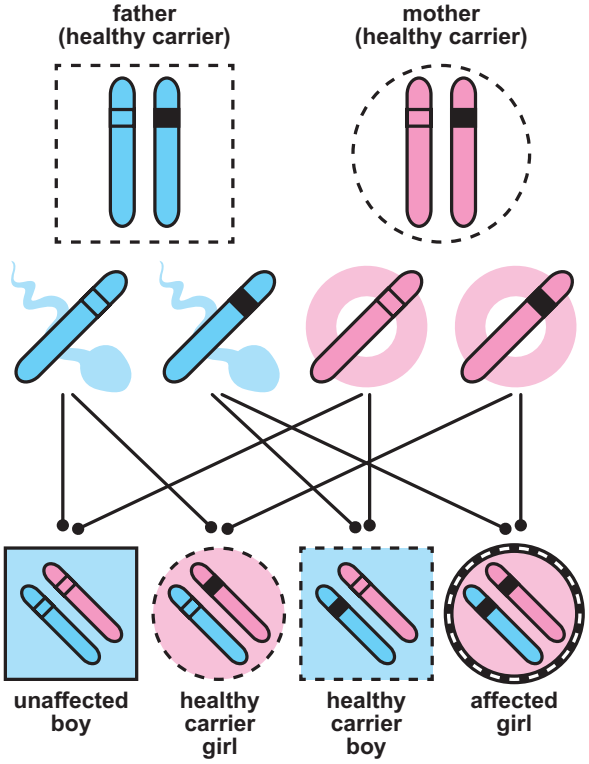
Please note: This information sheet suggests some commercial products that can be helpful in managing generalised severe junctional epidermolysis bullosa – inclusion in this information sheet does not constitute endorsement by GOSH and alternative products may be available.

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.

This information sheet explains about the generalised severe junctional form of EB.

What causes junctional EB?

We have two copies of every gene. If somebody has 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 1 in 4 risk in every pregnancy that the baby will inherit both faulty copies of the gene and therefore have junctional EB. The diagram below shows an example of how junctional EB can be passed on from parent to child – please note that both boys and girls can inherit the condition.



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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 the genes which help to bind the different components of the skin together. Junctional EB results from a defect in an important protein called laminin 332. Laminin 332 is made from three glue-like proteins which help stick the layers of the skin together. Junctional EB generalised severe is caused by a complete loss of one of the parts of this protein.

How is EB diagnosed?

In the majority of cases, the type of EB can be determined by analysis of a skin biopsy (tiny sample of skin). 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 effective treatments to correct the faulty proteins responsible, but present treatment is to manage the symptoms.

How serious is junctional EB generalised severe?

Junctional EB generalised severe is one of the most serious types of EB despite some affected babies appearing to be well and only having a few blisters when they are first born. Over time, the blisters become widespread and blistered areas develop into wounds which can be difficult to heal. The larynx (voice box) also blisters which causes problems with breathing and problems in the lining of the gut make it difficult for the baby to put on weight. The combination of these factors means many babies die within their first few months or years of life.

How can I look after my baby?

Handling

Nurse your baby on a small soft mattress which will be provided by your EB nurse. To lift your baby from the mattress, apply a 'roll and lift' technique to avoid damage from friction and shearing forces – roll your 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.

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 (50 per cent white soft paraffin and 50 per cent liquid paraffin) or Emollin® spray. Blisters should be lanced as above. A layer of 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-Cloth Super-soft® (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.

Give prescribed pain-killers before bathing and have the replacement dressings pre-cut ready to apply.

A mild antiseptic such as Octenisan® wash should be added to the bath water. It is often easier to bathe the baby with the dressings on and change them after bathing. Pat dry with a soft towel.

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Clothing

Initially, a front fastening babygro 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), which is helpful in keeping dressings in place. Skinnies® garments and Skinnies WEB® garments (www.skinniesuk.com) are particularly useful. Other garments include the Tubifast® range.

Wound care

Open wounds must be dressed to encourage healing and stop them sticking 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 wounds in junctional EB. Suitable dressings include PolyMem® (Ferris) and Intrasite® Conformable (Smith & Nephew) which provide protection for the skin and encourage 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 as a top layer similar to 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.

Finger- and toenails are often lost following blistering around the nail bed. Fingertip wounds can be dressed with Mepitel One® as a primary wound dressing, covered with Mepilex® Transfer or PolyMem® and secured with a small strip of adhesive tape, taking care the tape does not come into contact with the skin. 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 mucous membranes 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.

Some babies grow normally for the first few months of life and then weight becomes static or they begin to lose weight. Others struggle to grow from birth.

Increasing the calorie content of the feed can help in the short term but improvement may not be sustained. Changes to the feed should always be made under the guidance of a 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.

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Pain management

Pain management is complex and needs constant readjustment. Your EB team will help with this and will refer to a specialist paediatric pain or palliative care team to ensure your child is as comfortable as possible.

Breathing

Blistering of the voice box (larynx) is a common feature in babies with generalised severe junctional EB. The first sign of this is a hoarse cry which can develop within the first few days or weeks of life. Giving medicines via a nebuliser or steroids by mouth can help breathing difficulties. A small number of older children have benefitted from placement of a tracheostomy to assist with breathing.

Teeth

The enamel on the teeth is poorly developed in children with junctional EB. 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.

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 or ointment 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.

Is it possible to test for EB before birth?

Prenatal testing is available for parents who are known to be carriers of junctional EB. The test is called a 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.

Both tests are only possible if specific gene changes have been identified.

Further information and support

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

DEBRA is the national charity that supports individuals and families affected by Epidermolysis Bullosa (EB) – a painful genetic skin blistering condition which, in the worst cases, can be fatal.

DEBRA provides information, practical help and professional advice 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 Clinical Nurse Specialists for Epidermolysis Bullosa and DEBRA in collaboration with the Child and Family Information Group at GOSH.

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