

The new baby with Epidermolysis Bullosa (EB): information for families

Epidermolysis Bullosa (EB) encompasses a group of rare genetic fragile skin conditions, which cause the skin to blister or shear in response to minimal friction or trauma. This information sheet from Great Ormond Street Hospital (GOSH) describes how a newborn baby with EB will need to be cared for and what to expect in the first few days, weeks and months.

Please note: This information sheet suggests some commercial products that can be helpful in managing epidermolysis bullosa – including them in this information sheet does not mean that they are recommended by GOSH and alternative products may be available.

There are four main types of EB: Simplex, Junctional, Dystrophic and Kindler Syndrome, and within each type there are various subtypes, ranging from mild to severe. The type of EB will not change over time, nor are children with EB infectious or contagious in any way.

Below some of the most common forms of EB are outlined, this is by no means an exhaustive list, however gives a general idea of the various presentations.

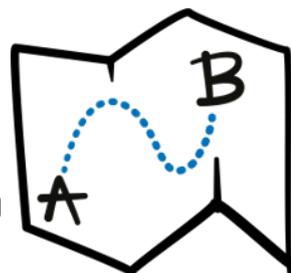
EB Simplex (EBS)

This is generally a dominantly inherited condition, although there are cases of recessively inherited EBS, however these are very rare. EBS tends to affect the proteins in the top layers of the skin and causes superficial blistering that heals without scarring.

There are two subtypes of EBS:

EBS Localised which tends to present mainly on the hands and feet in the form of blisters. More widespread blistering can occur and this is EBS. EBS Severe (previously called Dowling Meara EBS) causes more widespread blistering.

Management of these babies can sometimes be tricky as dressings can make skin damage worse. These babies can be very poorly in the neonatal period, particularly with reflux (where stomach contents travel back up the oesophagus or foodpipe). Symptoms tend to improve as they get older and children then present with hardened skin on the hands and feet.



Junctional EB

Junctional EB is recessively inherited and the subtypes range very much from mild to severe. Occasionally babies with Junctional EB have a blockage in the gut requiring an operation. This is called Junctional EB with Pyloric Atresia. Sadly, in its severest form Junctional EB Severe, babies do not tend to live past their second birthday so care is aimed at making them as comfortable as possible. These babies are missing essential proteins in their gut and their oesophagus and a combination of these two factors leads to death in early infancy.

Dystrophic EB

Dystrophic EB can be dominantly or recessively inherited. In common with many genetic disorders, those with the dominant form may be more mildly affected. However, recessive dystrophic EB varies in severity from minor symptoms to severe loss of skin at birth. There may also be problems in the long term associated with scarring (sometimes in the oesophagus). However this varies depending on the type of dystrophic EB your baby has and how severely.

Kindler Syndrome

Kindler syndrome is recessively inherited. Symptoms may include increased sensitivity to light, patchy/discoloration of their skin and/or hardening of the skin on the palms of the hands and soles of the feet (hyperkeratosis). Sometimes the gums and teeth are also affected as they get older.

More detailed information about the different types of EB is available from the GOSH website at www.gosh.nhs.uk or DEBRA the support organisation.

How do you get EB?

EB is a genetically inherited condition, meaning that there are abnormalities in a person's DNA/genes. Everyone has two copies of every gene, one inherited from their mother and one from their father. There are three ways in which EB can be inherited:

Dominant inheritance

In dominantly inherited EB, a fault in one copy of the gene can lead to fragile skin and blistering, even though the other gene is normal. This means anyone who has a dominantly inherited form of EB can pass the condition onto his or her children. There is a 50 per cent (1 in 2) chance in every pregnancy that the child will be similarly affected.

Recessive inheritance

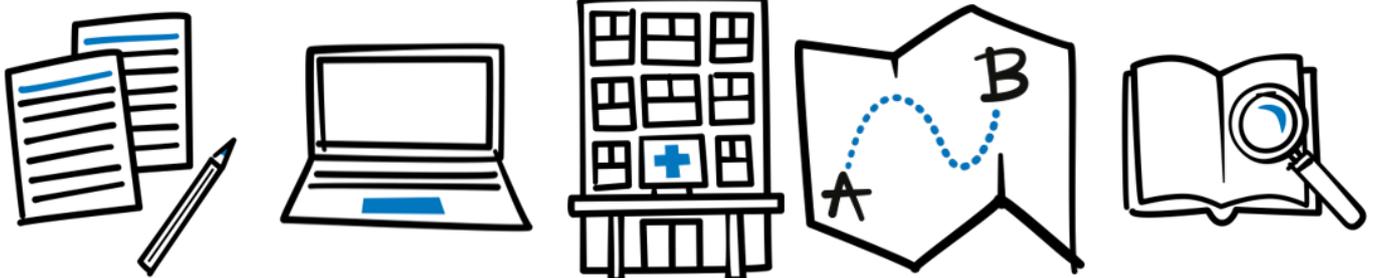
In recessively inherited forms of EB, both copies of the gene have to be faulty in order for the baby to be affected. A person with one faulty copy of the gene is healthy and said to be a carrier of the disorder. However, if two such people who carry the faulty gene have children there is a 25 per cent (1 in 4) risk that the child will inherit both faulty copies of the gene (one from each parent) and will have EB.

New mutation (fault)

However, dominant forms of EB can sometimes be seen as a 'new mutation' when there is no family history. This is where the gene fault occurs sporadically ('out of the blue') after conception.

How can I find out which type of EB my baby has?

A skin biopsy will confirm whether your baby has EB, and if so what type. This will involve taking a small sample of good skin (not blistered) using a local anaesthetic. This is a relatively quick



procedure and does not require any stitches. The sample will then be sent to the National EB Laboratory in London where a diagnosis will be made. Your EB nurse will deliver the result in person in around one to two weeks.

The inheritance of the EB (whether it is dominant or recessive) will need to be worked out using a blood sample from both parents and baby. This information is useful to have as it can be used to determine whether testing is needed for future pregnancies. Blood results can take up to three to six months to be finalised.

How can I look after my baby?

You can do all the things any newborn baby needs you to do but some adaptations may be necessary in the form of handling, feeding and dressing. Your baby may also need protective dressings which your EB nurse will teach you how to apply.

It is not usually necessary for the baby to be in an incubator unless there is a medical reason, such as the baby being born too soon (prematurely). In fact, sometimes heat can make blisters worse.

Wherever possible, nurse the baby in a cot laid on a soft pad and lift them using the pad. If your baby has severe skin loss then your EB nurse may be able to provide you with a soft mattress.

When it is necessary to lift using your hands, roll the baby onto their side, place your hand behind their head, and the other hand under their bottom and allow the baby to roll back onto your hands and lift in one movement.

Never lift from under the arms. Remember, rubbing forces may cause blisters and skin loss, whereas direct pressure is safe. If your baby is very fragile, you may want to use a gentle emollient (such as 50/50) to grease your hands before you handle them. However, please

remember that emollients will make your hands slippery so take precautions and use a firm direct grip.

Clothing

Naked babies with EB may damage their skin by kicking their legs together and rubbing with their arms. We recommend dressing babies in a front fastening baby grow. If the seams of the baby grow rub, you may need to turn it inside out. Baby grows that are soft and made from soft cotton, bamboo or even with silk are recommended. Your EB nurse will be able to give advice on where to purchase these items. If you find you are spending a lot of money on baby clothes, please speak to your EB nurse or DEBRA community support worker who may be able to help.

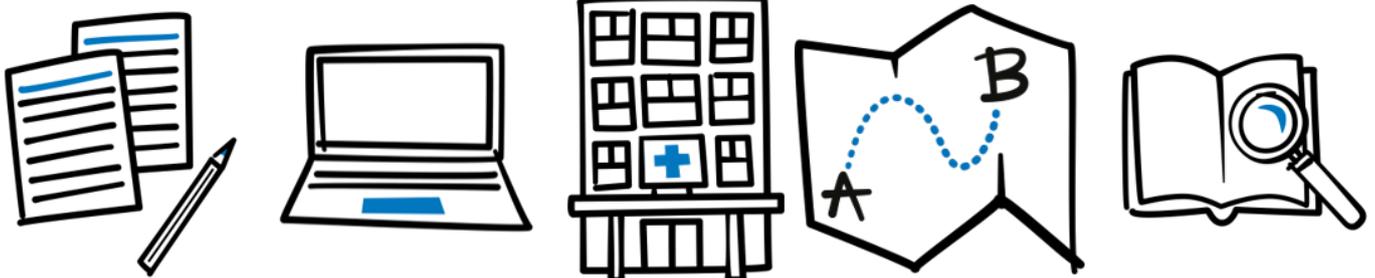
Nappies

Standard, disposable nappies are perfectly suitable for babies with EB. Make sure the nappy is a good fit to reduce friction; nappies that are too loose or tight may cause skin damage. It may be necessary to line the nappy with a soft cloth liner to protect the skin from the edges of the nappy. Barrier creams and dressings may be prescribed on the advice of your EB nurse.

Feeding

EB can also affect the internal lining of the body, especially inside the mouth and throat. If your baby has a lot of oral blisters, this may be quite painful and disrupt their feeding regime. We recommend that some pain relief is given before feeding. Again, your EB nurse will be able to advise on suitable bottles and teats that are gentle on the mouth.

Breast feeding is often possible although when the baby roots for the nipple, the skin on the face can be rubbed which may cause some skin damage. For this reason, we recommend lubricating the



breast and nipple with Vaseline™ or an emollient cream. Teething gels or specialised preparations such as Gelclair® can be applied to the teat or nipple or directly to the mouth to reduce pain when feeding.

Blisters

Blisters develop from friction or sometimes spontaneously in response to heat. Some types of EB are more 'blistery' than others. Blisters must be lanced with a sterile needle as they may spread or enlarge without getting better. Where the roof remains on the blister there is no need for a dressing. Dusting with simple cornflour will help to dry up the blister and reduce further friction.

Wounds

Wounds must be dressed with a non-adherent (non-stick) dressing. Your EB nurse will advise on

appropriate dressings for your baby and will arrange for all appropriate supplies to be available on prescription for when you are discharged home.

If your baby has extensive damage from birth, we will not bathe them until this has healed. Your EB nurse will carry out this first bath with you and demonstrate how to hold and what to use.

What help is available?

Your EB nurses and doctors will be able to answer many of your questions. Depending on the type of EB that baby has, your EB nurse will be able to carry out routine home or hospital visits. You will also be under the care of the specialist team at GOSH, where you will be required to attend regular appointments. Your local community nurses and health visitor will also be able to help you with daily care.

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.

