

Epidermolysis bullosa simplex: severe type: information for families

This information sheet from Great Ormond Street Hospital (GOSH) explains about severe type of epidermolysis bullosa simplex and how it can be managed. It also contains suggestions for making everyday life more comfortable.

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.

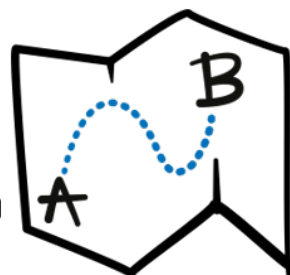
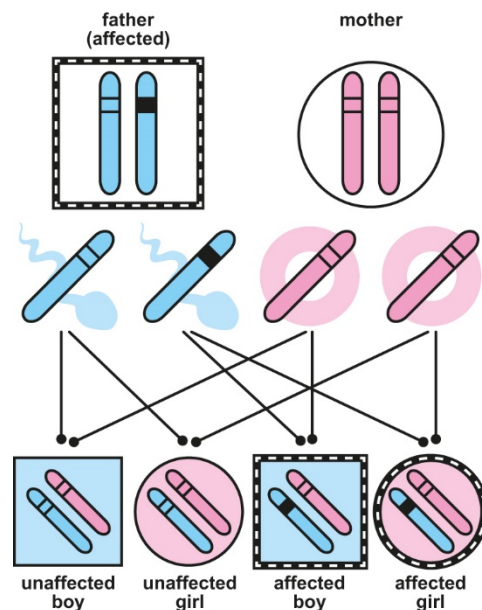
Epidermolysis bullosa (EB) is the term used to describe a number of rare genetic conditions which cause the skin to blister and shear in response to minimal friction and trauma. There are four broad categories of EB: EB simplex, junctional EB, dystrophic EB and Kindler syndrome. Within each of these categories there are several subtypes. If someone has one type of EB then they cannot develop one of the other.

What causes EB simplex?

EB is a genetic condition. Genes determine characteristics such as eye colour and also our health. A genetic mutation means a change similar to a spelling mistake has happened and this change makes the gene faulty.

We have two copies of every gene, each inherited from one of our parents. EB simplex is almost exclusively a dominantly inherited condition. This means that one parent has EB simplex. The other

parent is usually unaffected. Each time the affected parent has a child there is a one in two chance that the child will be affected. There is no carrier status in a dominantly inherited condition.



Sometimes an affected baby is born to unaffected parents. This is the result of what is called a 'new mutation'.

The problem lies in the genes that hold the instructions necessary for production of certain proteins in the skin. These instructions have a fault, rather like a typing error, with the result that the proteins are incorrectly formed, and unable to fulfil their role in attaching the layers of skin together.

How is EB simplex severe type diagnosed?

EB simplex has a characteristic appearance so is usually diagnosed soon after birth. Often one parent will know they have EBS but a more precise diagnosis can be made.

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?

Not yet, but research continues. There is still a long way to go, but an effective treatment to prevent the skin problems may ultimately be possible.

How is EB simplex severe type managed?

Babies can be born with a few blisters at birth or develop them shortly afterwards. They may be born with widespread blistering and open wounds. A hoarse cry is often characteristic of this condition – this is a result of small blisters on the larynx (voice box). This does not usually have any long term harmful effect but may carry on into childhood.

Very occasionally, infants with EB simplex are unwell in the first few weeks of life and may require intensive care. Infection can be a serious problem during this time and may need treatment with antibiotics.

Feeding

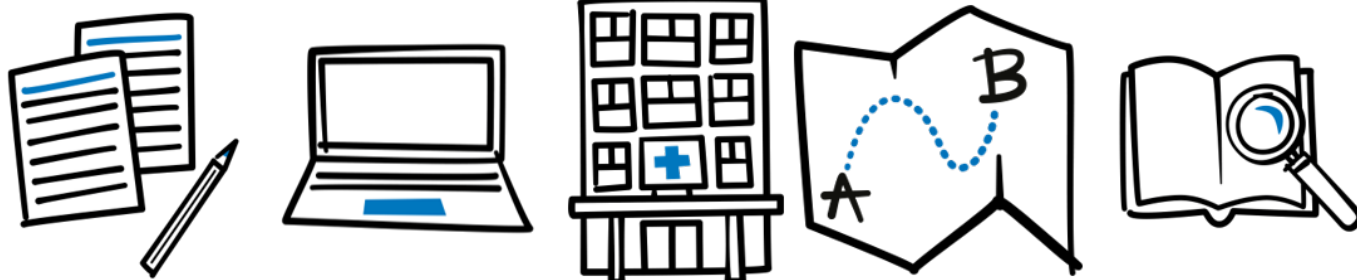
In the first few weeks, some babies have problems with blisters inside the mouth. This can make the baby reluctant to feed. Use of a special teat called a Haberman® or Special Needs Feeder can help the baby to feed. Application of teething gels to the mouth before feeding can also help.

The teat must be moistened with cooled, boiled water or teething gel before feeding, as a dry teat may stick to the blistered areas and cause further damage. Protect the baby's lips with a layer of Vaseline® petroleum jelly before feeding. Breastfeeding can be successful but the baby's face may need to be protected with a layer of greasy ointment to protect existing blisters from further friction.

Some of the baby's nutritional intake will be diverted to wound healing so the dietitian may recommend a high calorie feed to make sure the baby puts on weight.

A few infants are unable to take sufficient feed by mouth so nasogastric feeding (using a tube passed down the nose and into the stomach) is required for a short time. The tube selected should be one recommended for long term feeding, as these are soft and less likely to cause damage to the oesophagus (swallowing tube).

Adhesive tape should not routinely be used to secure the tube because of the risk of tearing the skin on removal. We recommend securing the tape with Mepitac® (Mölnlycke Health Care) or Siltape® (Advancis Medical), a soft silicone tape recommended for fragile skin.



If there are copious secretions making the skin too moist for Mepitac® or Siltape® to stick, a tape with greater adhesion can be used but this should be removed with a non-sting medical adhesive remover such as Appeel® Sterile Sachet (from Clinimed).

Soft silicone tape is also useful for securing intravenous drips if fluids or antibiotics are necessary.

Gastro-oesophageal reflux

Milk coming back up the swallowing tube from the stomach (reflux) is a common problem in many healthy babies and especially common in babies with EB.

Gastro-oesophageal reflux may cause a reluctance to feed as acid from the stomach causes pain when it comes into contact with blisters. Sometimes milk is brought up effortlessly and milk is seen in the mouth.

Reflux should also be suspected if the baby takes the first part of the feed well but then becomes distressed, shakes their head and refuses the remainder of the feed.

Coughing is another sign of reflux and milk may also be seen in the mouth between feeds. Anti-reflux medication is sometimes necessary to prevent damage to the fragile linings of the throat and mouth. Your doctor can prescribe medicines to control the reflux if necessary.

Constipation

This is a common problem in children with all types of EB, and results from pain and blistering at the bottom. They can be reluctant to open their bowels and can become very constipated. Laxatives can be prescribed by your doctor and may need to be continued long term.

Skin

In most children with the severe type of EB simplex, reduction in the amount of blistering is noted as they grow older. The widespread damage gradually settles down and is often limited mainly to the hands and feet. In time, areas of hard skin develop on the soles and palms, which help to protect these areas from blistering. Soreness of the feet can cause problems with walking. However, the extent of long term problems cannot be predicted at an early age.

Blisters tend to occur in clusters, often with an inflammatory appearance giving the impression that the lesions are infected. Unless your child is clinically unwell and skin swabs show an infection, antibiotic therapy is not necessary.

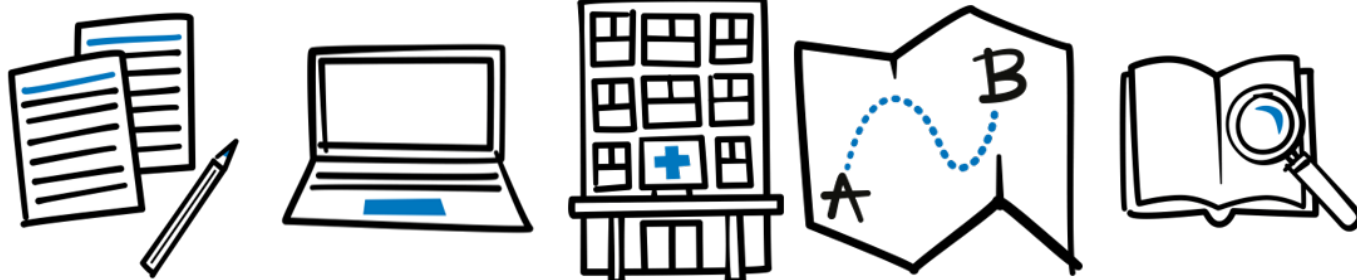
The blisters are not self-limiting and can become very large. We recommend inspecting the skin regularly and lancing the blisters. Gently expel the fluid from the blister using a soft piece of gauze or tissue. Some children prefer to have a 'V'-shaped cut made in the blister with sterile scissors. The roof should be left on the blister to protect the skin underneath.

Dust blistered areas lightly with cornflour to help them dry up and limit the spread. To soften scabbed areas apply a moisturising ointment.

As the skin is broken, its barrier protection is lost and infection can be a problem. If your child is unwell then your GP may prescribe a course of antibiotics. For minor skin infections, we recommend treatment with antiseptic creams or ointments.

Using dressings

Children with EBS severe type tend not to use many dressings as these can cause blisters, particularly at the edges of the dressing. When dressings are needed they should be removed



frequently to lance any new blisters which have formed underneath them. Dressings should be stopped as soon as the open wounds have healed.

Heat and humidity can cause more blistering. This has an adverse effect on people with EB. For this reason, dressings should be kept light and to a minimum. Every effort must be made to keep the environment cool, including the use of air conditioning units in extremes of hot weather.

Avoid bandaging the dressings in place or blisters will appear at the edge of the bandage. Use mittens and socks on hands and feet and tubular bandage on limbs or trunk to secure dressings.

Bathing

Bathing babies who have lots of blisters or wounds in the early days is delayed until the birth damage has healed. The bath should be lined or a towelling sling used to prevent damage to the skin from the sides and base of the bath. Antiseptic bath oil can be added to the water.

Nappy changing

Disposable nappies can be used, but must be lined to reduce friction and blistering. Your EB nurse will show you how to do this. When the bottom is badly blistered, we recommend cleaning with 50/50 (50 per cent white soft paraffin and 50 per cent liquid paraffin) ointment or Emollin® spray rather than water. Raw or blistered areas should then be covered with a barrier ointment and a dressing if necessary.

Eyes

Some children with EBS severe type have problems with sore eyes. This may be caused by a condition called blepharitis (inflamed eyelids). The eyelids, especially the lower lids, contain tiny

glands which make the substances that mix with tears and help the tears spread across the eye.

This thin layer of tears lubricates the eyes, stops them drying out and keeps them comfortable. Without treatment, the dryness can cause the cornea (covering of the eye) to blister. This is very painful. Any eye problems should be reported to your doctor or EB team so the condition can be treated following referral to an ophthalmologist (eye doctor).

Teeth

In most children with EBS severe type, the teeth form normally but blisters and sores inside the mouth can make cleaning difficult. We recommend regular visits to your dentist and cleaning with a soft toothbrush when the mouth is sore. A few children have poorly developed enamel on their teeth and will need restorative treatment.

Pain relief

Initially infants may need strong analgesia (pain relieving medicines) before dressing changes and regularly throughout the day if they are distressed. Older children may also need pain relief before skin care or regularly throughout the day if their feet are very sore.

Soreness, particularly of the feet, can result in delayed walking. This can be helped by giving medicine to help with chronic (background) pain as prescribed by your doctor. Pain management should be discussed at each appointment at your specialised EB Centre and a referral made to the Pain Team if control cannot easily be achieved.

Finger and toenails

Finger and toenails often become thickened and discoloured. This is nothing to worry about, however we advise keeping them short and soft to



prevent them scratching their face and causing potential damage.

If they become impossible to cut, application of a cream containing urea over a period of several days will soften the nail which can then be cut with scissors or clippers. Thickened nails can be brought under control using a coarse nail file and filing a little every day. If necessary, seek the help of a chiropodist.

Clothing

Initially, a front fastening baby grow is the most suitable item of clothing as it protects from external friction resulting from handling and general baby movements such as kicking the legs together. Remove any labels which may rub and avoid clothes with bulky seams when you are buying new clothes for older children.

We recommend that clothing and blankets are made from a soft cotton and non-abrasive. Some families also find materials made with bamboo are suitable for EB skin.

Silk garments are available on prescription and can be worn under regular clothes to reduce blistering.

Footwear

When your baby gets older, you may be thinking about their footwear choices.

Young children receive better protection from wearing a soft boot rather than a shoe as this protects their ankles from being knocked.

Shoes should be soft with minimal internal seams. External seams are preferable. Geox® footwear (www.geox.co.uk) allows sweat to evaporate and cools the feet. These have proved suitable for some children with EB. Older children can wear

trainers if they are well ventilated, such as Climacool by Adidas® (www.adidas.co.uk) and sometimes soft canvas shoes. It helps to have several pairs of shoes of different styles and to change one pair for another regularly to alter sites of friction.

Shoes in line with the uniform policy of schools are often unsuitable and permission needs to be granted to wear more suitable footwear such as black trainers. Many children find Crocs® (www.crocs.co.uk) comfortable as the wide fit prevents contact of the shoe with the top and sides of the feet.

Socks containing a silver thread such as Carnation® Silversocks (www.silversock.co.uk) help to regulate temperature through heat transfer to keep the feet cool which may reduce the rate of blistering and help reduce odour by controlling the level of bacteria.

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 recessive dystrophic 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 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.

