The New Baby with EB

Information for parents

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DEBRA is the national charity that supports individuals and families affected by Epidermolysis Bullosa (EB).
A charity registered in England and Wales (1084958) and Scotland (SC039654)

What is EB?

Epidermolysis Bullosa (EB) is a group of skin disorders in which the skin blisters or breaks down extremely easily.

There are 4 main types of EB. Each group has several subtypes.

What is the cause?

The problems lie in the genes that hold instructions necessary for production of certain important 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 do you get it?

EB is an inherited disorder.

It can be dominantly or recessively inherited.

Dominant inheritance

Everyone has 2 copies of every gene, one inherited from their mother and one from their father. In dominantly inherited EB a defect in one of these genes 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 1 in 2 chance in every pregnancy that the child will be similarly affected. However, dominant forms of EB can sometimes be seen as a “new mutation” when there is no family history.

Recessive inheritance

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

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

Diagnosis is made by examination of a small piece of skin called a biopsy. This is taken using local anaesthetic and does not require stitches. Blood samples are also taken from the baby and parents for DNA analysis to find the specific mutation on the gene, this information can be used to offer prenatal testing in future pregnancies for those who are carriers of severe forms of EB.

Is there a cure for EB?

Not yet, but research continues towards finding an effective treatment by replacing the faulty proteins in the skin.

How can I look after my baby?

You can do all the things any newborn baby needs but will have to change the way some of these things are done – such as handling, feeding and changing nappies and also some extra things like blister and wound care. Your EB nurse will help teach you how to do this.

It is not usually necessary for the baby to be in an incubator unless there are other medical reasons, such as the baby being born prematurely. In fact, the heat and humidity provided by the incubator may make the blistering worse.

Wherever possible nurse the baby in a cot, lie your baby on a soft pad and lift on this.

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

Never lift from under the arms. Remember, friction and shearing forces may cause blisters and skin loss, direct pressure is safe.

Clothing

Naked babies with EB may damage their skin by kicking their legs together and rubbing with their arms. We recommend dressing the baby in a front fastening babygro initially. If these are not flat seamed you will need to turn them inside out to prevent the raised seams from rubbing and causing blisters.

Nappies

Disposables are fine. Make sure the nappy is a good fit to reduce friction. It may be necessary to line the nappy with a soft liner to protect the skin from the edges of the nappy. Creams and dressings may be prescribed on the advice of your EB nurse.

Feeding

Sometimes the skin lining the inside of the mouth becomes blistered and sore. If the baby is finding it hard to feed because of the soreness we recommend a special teat called a Haberman feeder which contains a valve, reducing the need for strong sucking and allowing a good delivery of milk. Breast feeding is often possible although when the baby routes for the nipple the skin on the face can be rubbed and needs to be protected with cream or ointment.

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

Arise from friction or sometimes spontaneously in response to heat.

Blisters must be lanced with a sterile needle as they are not self-limiting and will get bigger if left unchecked. 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 dressing. Choice is limited as many dressings described as non-adherent behave differently on the skin of those with EB.

Your EB nurse will advise on appropriate dressings for your baby.

Types of EB

There are 4 major types of EB; these are simplex, junctional and dystrophic and Kindler syndrome.

Within each group there are many different sub types and therefore each type of EB has a wide range of symptoms.

It is not possible for one type to change to another within the family.

EB simplex

This is generally a dominantly inherited condition, although there are cases of recessively inherited EB simplex, but these are very rare.

There are 3 main types of EB simplex.

Localised EB simplex generally affects only the hands and feet and is at its most troublesome during warm and humid conditions.

More widespread blistering can occur and this is called **generalised EB simplex**.

Dowling Meara

Dowling Meara EB simplex causes more widespread blistering particularly in young children. Babies with Dowling Meara simplex may be very ill for a while, but the majority recover and the

extensive blistering will gradually settle. Blistering and thickened skin on the hands and feet can cause problems in the longer-term.

Junctional EB

Junctional EB is recessively inherited. This can be a mild condition, causing little disability and few long term problems. However, in its most severe form (Herlitz junctional EB) problems with breathing and weight gain can lead to death as a baby or young child.

Occasionally babies with junctional EB have a blockage in the gut requiring an operation.

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, and problems in the longer term caused by scarring.

Kindler syndrome

Kindler syndrome is recessively inherited. Blisters and wounds are common at birth and in the first few weeks. After that blistering reduces but some skin problems remain and there may be complications associated with scarring.

How can I find out more about EB?

DEBRA produce a range of booklets specific to each type of EB which will be available to you once the type of EB is confirmed. Your EB nurses and doctors will be able to answer many of your questions.

What help is available?

DEBRA is the national charity that supports individuals and families affected by Epidermolysis Bullosa (EB).

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**, email **membership@debra.org.uk** or call 01344 771961.

Your local community nurses and health visitor will be able to able to help you with daily care.