

Published by DEBRA

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Epidermolysis Bullosa (EB) is the name given to a group of rare genetically determined blistering skin disorders. Within this group there are three major types of EB, these are:

junctional

dystrophic

and simplex.

Although they all come under the general heading of epidermolysis bullosa, each type is very different in the symptoms they produce. The common factor being genetic inheritance and blistering of the skin in response to minimal everyday friction and trauma. Apart from this, problems are individual to each type.

Epidermolysis bullosa simplex is almost exclusively a dominantly inherited disorder.

This means that one parent has the condition, and will have blisters on the skin.

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 disorder.

The two main types of EB simplex are Weber Cockayne (localised to the hands and feet) and Dowling Meara (herpetiformis type).

This short booklet will explain some of the problems, care and management associated with the Dowling Meara form of EB simplex.

The **Dowling Meara** sub-type of EB simplex is unusual in that it is often seen as a new mutation, where both parents are unaffected, and the inheritance pattern will commence with this new infant.

Diagnosis is made from analysis of a skin biopsy, taken from clinically unaffected skin. Intra epidermal cleavage occurs within the level of the basilar keratinocytes where clumping of tonofilaments may also be seen.

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In many cases more precise diagnosis can be made following identification of the specific mutations using DNA analysis from a blood sample.

When the skin is subjected to everyday friction, the layers of the skin move across each other, and a blister develops. More intense friction, or a shearing action will cause the skin to be pulled away, leaving a wound. In common with the other types of EB, the Dowling Meara form varies in severity.

Infants

are often born with widespread damage, or blistering appears shortly after birth. A hoarse cry is characteristic of this condition, resulting from small blisters on the larynx (voice box). This does not generally have any long-term harmful effect, but may persist into childhood.

A few infants are very sick in the first few weeks of life and occasionally require management in an intensive care ward. Infection can be a serious problem during this time and may need treatment with intra venous antibiotics.

In those first few weeks, some babies have problems with severe blistering of the mouth and sometimes the oesophagus as well. This can make the baby reluctant to feed. Use of a Haberman Feeder (mail order from tel. 01989 566669) often helps, as does application of teething gels to the mouth prior to feeding. The teat must be moistened with cooled boiled water before feeding as a dry teat may stick to the blistered areas and cause further damage. Lips are protected with a layer of Vaseline petroleum jelly prior to feeding.

Feeding

Because some of the nutrition is diverted into wound healing, a high calorie feed is usually required to ensure adequate growth. Initially, a formula feed such as that recommended for infants who are of a low birth-weight is given. Specific advice should be obtained from Lesley Haynes, dietitian for EB. (0207 405 9200 ext 5761). Lesley will liaise with dietitians in the Child's local hospital or community.

A few infants are unable to take sufficient feed by mouth, and naso-gastic tube feeding is required. The tube selected should be one recommended for long-term feeding as these are soft and less likely to cause damage to the oesophagus.

DEBRA HELPS SUFFERERS FROM ALL TYPES OF EPIDERMOLYSIS BULLOSA

• By raising money for vital research into the disease.

This alone can bring real and lasting benefits to present sufferers and their families, and to children yet to be born, by providing effective treatment and detection of the disease.

• By investigating and implementing all possible ways of helping sufferers in their daily lives.

These measures are many and varied and part of our funds are used for these purposes.

They include the following:

- To give counselling support, especially to the parents of a newborn child.
- To inform and educate sufferers, the medical profession and the public about the disease.
- To publish and distribute material for parents and workers in the community.
- To advise sufferers, where necessary, in obtaining the benefits to which they are entitled.
- To help and advise on all difficulties connected with education and employment.
- To further these aims nationally and internationally by assisting individuals in other countries to promote similar associations.
- To provide regular newsletters to members.
- To encourage sufferers to lead as full a life as possible.

OTHER FREE DEBRA PUBLICATIONS

Epidermolysis Bullosa: An Outline for Professionals
(FOR PROFESSIONALS ONLY)

The Management of Junctional Epidermolysis Bullosa.
(FOR PROFESSIONALS ONLY)

Care and Management of Children with Dystrophic Epidermolysis Bullosa.

A Guide for Parents, Schools and Playgroups

A Guide to Physiotherapy for Children with Dystrophic Epidermolysis Bullosa.

Play and Development Needs of a Child with Dystrophic Epidermolysis Bullosa.

Diet for Epidermolysis Bullosa (for children over 1 year)

Nutrition for Babies with Epidermolysis Bullosa

Bibliography

General Information leaflets for fundraising

Brief History of the Association/List of Research Projects Funded

Can I Go It Alone? A guide to independent living for young people with severe EB.

Coping with Epidermolysis Bullosa – A Guide for Adults

DEBRA IS DEPENDENT ON CHARITABLE FUNDS TO CARRY OUT ITS WORK.

Adhesive tape should not be used to secure the tube because of the risk of tearing the skin on removal. We recommend securing the tube with Mepiform (Molnlycke Health Care). This is an adherent silicone dressing recommended for scar care, it will hold the tube securely but can be removed without risk of skin damage.

Mepiform is also useful for securing intra venous cannulae if fluids or antibiotics are required by this route.

Gastro-oesophageal reflux is a common problem in many healthy babies. In those with epidermolysis bullosa it may cause a reluctance to feed as the refluxed acid results in pain on the blistered areas. Reflux is suspected if the baby takes the first part of the feed well, but then becomes distressed, shakes his head and refuses the remainder of the feed.

Coughing is another sign of reflux, and milk may be seen in the mouth between feeds. Anti-reflux medication in the form of gaviscon and/or ranitidine relieves the discomfort and oral intake should improve. The Dietitian may recommend adding a thickening agent to the feed.

The introduction of weaning foods at the appropriate age will help to control the symptoms.

Constipation is a common problem in those with all types of EB, and results from pain and blistering at the anal margin. If left unchecked this leads to faecal retention, constipation and in extreme cases, faecal soiling and over-flow.

A stool softener such as lactulose may be sufficient, however, in many cases a stimulant laxative and extra dietary fibre are required in addition.

Skin In most children with the Dowling Meara form of EB simplex reduction in the amount of blistering is noted as they grow older. The widespread skin damage gradually settles down until the main affected areas are 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 the lesions are infected.

Unless the child is clinically unwell and skin swabs show growth of pathogens; antibiotic therapy is not indicated.

Children with Dowling Meara simplex are adversely affected by the heat, and in particular by humidity, which can cause spontaneous blistering. For this reason dressings are kept light and to a minimum, and every effort must be made to keep the environment cool, including use of air conditioning in extremes of hot weather.

Skin care

Bathing is delayed until the birth damage has healed, and is then restricted to times of major dressing changes, until the wounds are healed. Bathe in an emollient, such as Oilatum or Dermol 600. The skin must be inspected regularly and blisters burst with a sterile needle. The roof is left on the blister. Dead skin is cut away in order to avoid formation of blisters around the scab or crust.

Blistered areas are lightly dusted with cornflour to help them to dry up and limit the spread. Scabbed areas can have application of Dermol 500, a moisturizer containing antiseptic.

Any open wounds are dressed with Aquacel dressings (Convatec). Superficial wounds are dressed with Mepitel (Molnlycke). Cornflour is dusted over the top of the Mepitel and covered with light gauze dressing.

Avoid bandaging the dressings in place, or blisters will appear at the edges of the bandages. Use mittens/socks on hands and feet, or tubular bandage such as Tubifast (Seton) on limbs or trunk to secure dressings.

The Tubifast and gauze should be changed daily and the Aquacel and Mepitel changed every few days to allow wound healing.

Pain relief

Initially infants may require strong analgesia such as morphine prior to dressing changes. Older children may benefit from paracetamol or ibuprofen given before skin care, or regularly as required.

Chronic pain, which may delay development, can be controlled by use of the drug amitriptyline, which is well documented for its use in nerve pain. 0.5 mg per kg is given

once a day, usually at night. Benefits are subtle and are generally seen after several weeks of treatment.

Finger and toe nails

Often become thickened and discoloured. If they become impossible to cut, application of a urea cream such as Aquadrate or Eucerin over a period of several days will soften the nail which can then be cut with scissors or nail 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

Disposable nappies can be used, but must be lined to reduce friction and blistering. Initially a front-fastening baby suit 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. When buying clothes for older children remove any labels which may rub, and avoid clothes with bulky seams.

If seams create a problem with underclothes, they can be worn inside-out. Avoid shirts with stiff collars. Airtex shirts can be worn in place of a school shirt.

A range of clothing suitable for children with delicate skin is available mail order from Cotton Comfort, P O Box 2406, Bath, BA1 5ZD. Tel. 01225 336559.

Marks & Spencer have a range of under clothes with flat seams and labels stitched on the outside of the garment which they recommend for children with eczema and fragile skin. These are available from larger branches or from their catalogue.

Footwear is

often a problem. Young children have better protection from wearing a soft boot rather than a shoe, in order to prevent their ankles from being knocked. Makes of footwear such as Elephanten (available in small sizes only) and Ecco have proved to be suitable. Older children can wear trainers if they are well ventilated, 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. Friction relieving products from the Silipos Company can be useful to protect problem areas. (Silipos (UK) Limited, 85a Stanmore Hill, Stanmore, Middlesex HA7 3DZ. Tel. 0208 4207007).