

# **Epidermolysis Bullosa Simplex – Dowling Meara**

## **Jacqueline Denyer - EB Nurse Consultant**

### **INTRODUCTION**

Epidermolysis Bullosa (EB) is the term used to describe a number of rare genetically determined skin conditions characterised by a tendency for the skin to blister or shear away in response to minimal friction and trauma.

### **DIAGNOSIS**

In the majority of cases the type of EB can be determined by analysis of a skin biopsy. Often more precise diagnosis can be made following identification of the specific mutations using DNA analysis from blood samples.

There are three broad categories of EB. These are EB Simplex, Junctional EB and Dystrophic EB. Within each of these categories there are several subtypes. It is not possible for the type of EB to change within an individual.

### **INHERITANCE**

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. Sometimes an affected baby is born to unaffected parents. This is the result of a new mutation occurring on a gene. The genes responsible for EB Simplex are those which make the proteins keratin 5 and keratin 14. Mutations within these genes produce faulty proteins which make the inside skeleton of skin cells weak meaning it breaks easily and blisters result.

The three main types of EB Simplex are Localised EBS (previously called Weber Cockayne, Generalised EBS (previously called Köbner) and Dowling Meara (herpetiformis type – so called because of the tendency for blisters to appear in clusters). In common with the other types of EB, the Dowling Meara form varies in severity.

### **MANAGEMENT OF THE CHILD WITH DOWLING MEARA EB SIMPLEX**

Infants are often born with widespread blistering and skin loss or blistering may appear 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 and even adulthood. Occasionally infants are very 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 intravenous antibiotics.

### **Feeding**

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 special teat called a Haberman Feeder often helps the baby to feed 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. Breast feeding is often successful. Because some of the nutrition is diverted into wound healing, a high calorie feed is sometimes 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 a specialist EB dietitian, if possible.

A few infants are unable to take sufficient feed by mouth, and naso-gastic tube feeding is required for a short period of 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. 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 tube with Mepitac (Mölnlycke Health Care) - a soft silicone tape recommended for fragile skin. If there are copious secretions making the skin too moist for Mepitac to stick, a tape with greater adhesion can be used but this should be removed with a non sting silicone medical adhesive remover such as Appeel (Clinimed) or Niltac (Trio Healthcare). Mepitac 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 and especially common in babies with EB when it may cause a reluctance to feed as the refluxed acid results in pain on the blistered areas. Sometimes milk is brought back up effortlessly but reflux should also be 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 is necessary to control symptoms. Mild reflux responds to the drug ranitidine. For management of severe reflux we recommend triple therapy with lansoprazole, domperidone and ranitidine.

**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. For management of chronic constipation and prevention of faecal impaction the osmotic laxative, Movicol Paediatric (Norgine) is very effective.

### **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 presence of infection; 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 (babies)**

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 lanced 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.

Children with Dowling Meara are frequently unable to tolerate dressings as these exacerbate blistering, particularly around the edges of the dressing. Dressings should be withheld as soon as possible and while in place removed frequently to lance fresh blisters which have formed beneath them. Any open wounds must be dressed with a non adherent dressing. Mepitel (Mölnlycke) and Urgotul (Urgo) are atraumatic on removal but the edges can damage the fragile skin. Where this happens we recommend placing Aquacel (Convatec) spread with a layer of 50/50 (50% white soft, 50% liquid paraffin) under the edges of the Mepitel. Aquacel plus 50/50 can be used directly on superficial wounds. The primary dressing (Mepitel, Urgotul or Aquacel) is covered with a soft secondary dressing to absorb exudate and offer protection from further trauma. Suggested secondary dressings include PolyMem (Ferris) Mepilex, Mepilex Lite and Mepilex Transfer (Mölnlycke). 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 (Mölnlycke Healthcare) on limbs or trunk to secure dressings. The Tubifast and secondary dressing should be changed daily and the area inspected for fresh blisters. The primary dressing may be left in place for several days to encourage healing.

Infection is frequently a problem and should be treated with antibiotic therapy if the child is unwell. For minor skin infections we recommend treated with Crystacide cream (1% stabilised hydrogen peroxide) or products containing silver or honey which are effective in reducing the bacterial load. We do not advise using Silver dressings in small infants and not continued for more than a few weeks in any age group.

Disposable nappies can be used, but must be lined to reduce friction and blistering. When the nappy area is very blistered we recommend cleaning with 50% liquid / 50%white soft paraffin rather than water. Raw or blistered areas should then be covered with Intrasite Conformable dressings (Smith & Nephew) and red or vulnerable areas covered with a thick layer of Bepanthen Ointment (Roche).

### **Skin care (older children)**

Blistering may reduce as the child grows older but hyperkeratosis (hard skin) can be a problem particularly on the palms and soles. Blisters forming under the hard skin are difficult to lance and very painful. Use of Cryogestic spray (ethyl chloride) temporarily freezes the area providing pain relief. Dressings recommended above can be used for all age groups, but often dressings with a cooling effect are helpful. These include Actiform cool (Activa) and Intrasite Conformable (Smith & Nephew). These dressings can be kept in the fridge to make them extra soothing.

### **Eyes**

Some children with Dowling Meara have problems with sore eyes. This may be caused by a condition called blepharitis, or inflamed eyelids. The eyelids, especially the lower lids, contain tiny glands which make 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 eye) to blister which is very painful. We recommend any eye problems are reported to your doctor or EB team so the condition can be treated following referral to an ophthalmologist (eye doctor).

### **Teeth**

In most children the teeth are normally formed, but blisters and sores inside the mouth can make cleaning difficult and 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 require strong analgesia (pain relieving medicines) prior to 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 medicines to help with chronic (background) pain. We recommend using the medicines amitriptyline or gabapentin, both of which are effective in managing chronic pain. Pain management should be discussed at each appointment at your specialised EB centre and a referral made to the Pain Team if control cannot be easily achieved.

**Finger and toe nails** often become thickened and discoloured. If they become impossible to cut, application of a urea cream 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**

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.

### **Footwear**

Young children have better protection from wearing a soft boot rather than a shoe, in order to prevent their ankles from being knocked. Shoes should be made from soft leather with minimal internal seams or preferable external seams. Geox footwear ([www.geox.co.uk](http://www.geox.co.uk)) allows sweat to evaporate and cools the feet and these have proved suitable for some children with EB. Older children can wear trainers if they are well ventilated, such as Climacool (Addidas) 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 leather trainers. Friction relieving products from the Silipos Company can be useful to protect problem areas - Advasil (Advancis) is a silicone sheet which can be used over minor blistering or to protect vulnerable areas. This can be washed and re-used several times. Socks containing a silver thread, 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.