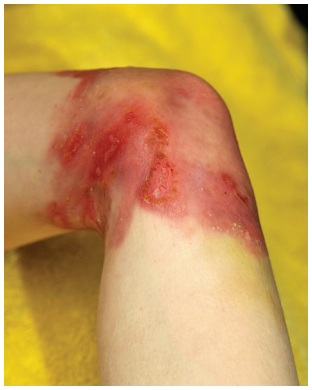
Epidermolysis bullosa: the worst condition you've never heard of



In people with epidermolysis bullosa, any trauma or friction to the skin can cause painful blistering

This new column focuses on the rare and painful genetic skin blistering condition epidermolysis bullosa (EB). It is produced in association with DEBRA, the charity supporting people with the condition, and will feature commentary from specialist nurse practitioners, enabling readers to better identify and manage this condition.

THE MAIN TYPES OF EB

There are three main types of EB — EB simplex (EBS), dystrophic EB (DEB) and junctional EB (JEB) — and each type has multiple subtypes; 30 subtypes of EB are recognised (Fine et al, 2014; McGrath, 2015).

EBS, is characterised by a lack of adhesion of the skin directly above the basement membrane. About 70% of people with EB have EBS (Intong and Murrell, 2012; NHS, 2015a; DEBRA, 2016), with most forms inherited as dominant traits (NHS, 2015b). EBS typically manifests as blistering to hands and feet, but in some forms blisters occur all over the body (NHS, 2015c). Blistering generally appears during the neonatal period, but may manifest later. Painful skin blisters are accentuated by rubbing, which is worse in warm weather, and infections are common (NHS, 2015c).

DEB is characterised by a lack of adhesion of the skin under the basement membrane. Approximately 20–25% of people with EB have DEB, which may be inherited as a dominant or recessive trait (NHS, 2015a; DEBRA, 2016). DEB blisters tend to heal with scarring and can lead to contraction of the joints, fusion of the fingers and toes, contraction of the mouth membranes and narrowing of the oesophagus. The severity varies widely. Often the dominant inherited type is the least severe and the patient can lead an almost normal life. People living with recessive DEB often have to drain and dress blisters every day and they have a high chance of developing a squamous cell carcinoma before the age of 35 (DEBRA, 2016; NHS, 2015c).

Approximately 5–10% of people living with EB have JEB, which is characterised by a lack of adhesion of the skin through the basement membrane (Fine et al, 2014; NHS, 2015a; DEBRA, 2016). JEB is an autosomal recessive condition (NHS, 2015b). JEB generalised severe, which affects around half of those diagnosed with JEB, is usually fatal in infancy with few surviving to the age of five (NHS, 2015c; DEBRA, 2016). Infants usually die from sepsis and respiratory failure due to blistering and narrowing of the airways (NHS, 2015c). JEB generalised intermediate, does not impose life restrictions, but can cause life-long pain and disability.

Although EB is typically inherited, it can arise through a sudden mutation; whereby a gene mutates spontaneously before conception (DEBRA, 2016). Rarely, a severe form of EB can be 'acquired' as the result of autoimmune disease (NHS, 2015a).

DEBRA: THE EB CHARITY

DEBRA is a UK charity that supports the 5,000 people living with EB and their families. It was founded in 1978 and provides an enhanced EB healthcare service, in partnership with the NHS, ensuring that those with the condition have access to the many different specialists that they need — nurses, podiatrists, dietitians, occupational therapists etc. The charity also funds outreach clinics to bring specialist care closer to patients' homes, as some patients have to travel miles to their closest EB Centre of Excellence (Great Ormond Street, Guy's and St Thomas', Birmingham Children's and Solihull Hospital).

To ensure holistic care, DEBRA funds community support staff to work with individuals and families, gives grants to help families with the day-to-day difficulties of living with EB as well as respite in the form of days out, events and the access to specially adapted holiday homes. The charity also funds pioneering research to find effective treatments and, ultimately, a cure for EB.

Further information

If you would like more information on DEBRA or EB, or if you are interested in attending a course or study day, please visit www.debra.org.uk/healthcare-professionals

debra
for people whose skin doesn't work we do

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