

What is EB?

Epidermolysis Bullosa
and DEBRA UK



Epidermolysis Bullosa



Genetic condition

Epidermolysis Bullosa (EB) is a complex and rare group of genetic skin conditions that cause the skin to be extremely fragile. Blisters and wounds occur from pressure, friction or heat on the skin and/or mucous membranes.

EB may be inherited as either dominant or recessive. There are an estimated 5,000 people affected by EB in the UK and 500,000 worldwide.



Types of EB

There are over 30 subtypes of EB, grouped into four main types: EB Simplex (EBS) accounting for around 70% of patients, Dystrophic EB (DEB) 25%, Junctional EB (JEB) 5% and other rare types including Kindler Syndrome.



Symptoms and complications

Symptoms vary and range in severity depending on the type of EB, from relatively minor disability (such as limitations of walking distance because of blistering of the feet) to severe forms of EB which can be fatal.

Blisters continuously form and have to be drained and dressed daily, a painful process that can take several hours. Blisters can form internally such as inside the mouth and create difficulty swallowing. There can be narrowing of the oesophagus and airways, contracture of the mouth and reduced movement of the tongue. Eyes may be affected by blistering. Dental enamel may also be affected, with overcrowding, malalignment of teeth and tooth decay. The build-up of scar tissue can cause fingers and toes to fuse together. It can mean a higher risk of developing skin cancer. The most common challenge amongst all types of EB is the pain and itching that occurs as a result of the blistering.



Treatment

People living with EB may require treatment from a range of medical specialists aimed at symptom relief and to prevent complications such as infection. It is currently incurable.

“ My son Jamie was born with no skin on his feet, knees and hands and even where there was intact skin, it blistered. It was so hard to bond – I didn’t hold my baby for the first six months.



He is the first person in my family to have EB so it came as an enormous shock. Jamie has a full skin check every day, which involves me lancing all blisters that have occurred overnight. I dress all the wounds and put protective bandages on before dressing him as well as giving him pain killers. The daily routine is quite structured to ensure he feels safe and secure at all times. He has his large dose of morphine before the evening meal so he’s ready for his bath and can cope with more dressing changes. The most difficult thing about EB is seeing your child in pain, knowing that the care you are giving is causing so much distress.

The support I have received from DEBRA has been outstanding and makes a real difference. DEBRA has provided me with a changing table where I keep all the dressings and has signposted me to disability living allowance to help soften the blow of not being able to return to work as I am now a full time carer. The information booklets and website are a good reference to anyone wanting to know more. Meeting others with EB through DEBRA has made my outlook on the future less daunting. Seeing children with EB playing happily and managing daily activities gave me positivity and hope. The DEBRA days give me a boost of mental energy I so often need.



I feel extremely proud and in awe of Jamie, his happiness and joy keeps me motivated to stay strong. I am keeping positive that Jamie will improve with age and become less fragile so his pain levels will reduce.

Katie White, mother of Jamie White

Our work

DEBRA was the world's first EB patient support group, founded in 1978 by Phyllis Hilton whose daughter Debra had Dystrophic EB. When told that nothing could be done for her baby, she set out to find ways to treat Debra's skin using cotton dressings. We have come a long way since then. From discovering the first EB genes to funding cutting-edge clinical trials of therapies to control symptoms and better ways to manage complications like cancer, the research journey has been remarkable.

We have made significant progress in advancing diagnosis, treatment and daily management of EB. DEBRA is now international in scope with a network of DEBRA groups worldwide sharing information and best practice. We are committed to making sure that people with EB and their families and carers get the vital and wide-ranging support they need so that the devastating symptoms of EB can be reduced while we work towards improved treatment and a cure.



Specialist healthcare

We work in partnership with the NHS to deliver an enhanced multidisciplinary EB healthcare service for people with all types of EB. We have invested in the development of EB centres of excellence, support clinical teams to provide advice to improve health and wellbeing, fund training and clinical best practice guidelines and connect patients with the expert, specialist services they need across the UK, from diagnosis onwards.



Diagnosis

EB may be diagnosed or flagged as a possibility by the neonatal team or by clinicians later in life. Further laboratory testing such as skin sampling and genetic testing is undertaken to confirm the diagnosis and EB type. Prenatal testing is also possible. We can support families by providing information about EB, helping with referrals to specialists, accessing medical supplies and equipment.

Lifelong EB support

We provide individualised support, information, guidance and practical help for those living with EB and their carers, connecting them with the specialist services they need and a support network across the EB community. This includes finance maximisation, benefits and education advocacy, grants, bereavement support, events, social opportunities and respite holiday homes.

Pioneering EB research

The research journey for EB over the past 40 years has been remarkable – from gene discovery to cutting-edge trials of therapies to control symptoms and manage complications like cancer. During this time, we have invested over £20m and been responsible for establishing much of what is now known about EB. We are the largest UK funder of EB research. We have an ambitious, patient-centred research strategy that will enable us to fund science of the highest quality across the world, including prioritising investment in drug re-purposing, to develop a treatment pipeline to slow, stop and/or reverse EB.

Visit our website to find out more about clinical practice guidelines and healthcare services as well as contact details for the specialist EB teams in hospitals in Glasgow, Birmingham and London. <https://www.debra.org.uk/specialist-eb-healthcare>

Research



We want to accelerate the pace and breadth of our research. To achieve this we are committed to increasing our funding and driving a programme of world-class, innovative and collaborative research that will bring hope and improved outcomes for everyone living with EB. Together we can achieve this ambitious and essential journey to change lives and end suffering.

Our new strategy puts patient outputs front and centre, with a focus on translational research that will have a positive impact on those with EB today. This strategy also embraces partnerships with specialists in fields that relate to EB, including dermatologists, ophthalmologists, podiatrists, and oncologists, in addition to strengthening our relationships with the pioneering EB centres of the UK.

Our four overarching research priorities are those we view most likely to deliver outputs for people living with EB. They are:

- invest in drug re-purposing and develop drug discovery programmes to accelerate finding treatments.
- increase investment in patient-centred research themes
- continue to invest to understand better the causes and progression of EB and the role of the immune system
- invest heavily in the next generation of EB researchers.

To find out more about our research please contact: research@debra.org.uk or visit our website: www.debra.org.uk

Our funding programmes

Applications are welcome from all disciplines committed to improving the lives of people with EB. Funding for researchers will be provided through the following schemes.

Project grants

Awards up to three years will be available to researchers through our research calls. Funding will be judged on relevance to EB, scientific merit and novelty. Applicants must demonstrate the potential benefit to EB patients in the research proposal.

Programme grants

Awards will be provided where collaboration between several groups is required such as clinical projects adopting a multicentre approach.

PhD studentships

PhD grant awards for 3-year projects to develop degree graduates into EB researchers. The criteria will be the same as for project grants with the addition of quality of research environment and training for students.

Clinical research fellowships

Awards for registered healthcare professionals to establish themselves as independent investigators to run their own group and develop their own research interest in EB. The aim is to create future leaders in the EB field.

Non-clinical research fellowships

Awards to support post-doctoral researchers at early and intermediate stages of their career to establish themselves as independent investigators to run their own group and develop their own research interest in EB. The aim is to create future leaders in the EB field.

Small grants

Small grant awards support small pilot studies such as generation of preliminary data or feasibility studies which would not normally attract funding. The aim is to make novel ideas more competitive for larger follow up funding grants.

“We are calling on the scientific community to join us on this journey to accelerate EB research innovation.”

Dr Sagair Hussain, Director of Research, DEBRA UK

- ▶ **DEBRA** is the national charity supporting those living with or directly affected by EB. We have achieved global recognition as a leader on EB, and are the only UK charity specifically aimed at continuously supporting the EB community.

DEBRA funds pioneering research, specialist healthcare, international best practice and lifelong care and support to the EB community.



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