EPIDERMOLYSIS BULLOSA

When being a butterfly is PAINFUL

FRAGILE

Makes the skin so fragile that children with it are referred to as **"butterfly children"**.

RARE[1,2,3]

Inherited from parents who may not be aware they are carriers.[1]
It is estimated that **1 in 20,000** babies born will have some form of EB.[4]
Worldwide there are thought to be **~500,000** people living with EB.[1,3]
In the UK there are known to be 5,000 people who have the condition.[1]

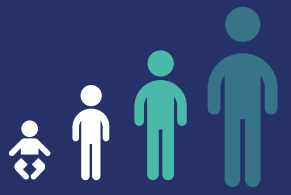
CAN BE DEVASTATING[5,6]

Intensely painful, recurrent blistering and chronic wounds of the skin can result in limited mobility. Not only does EB affect the external skin, but also the lining of the mouth and throat, the eyes and digestive system, leading to malnutrition, anaemia, intolerable itch and pain.[2]

NO CURE[3,6]

Current treatment is focused on wound care which involves **daily or regular** dressing changes to protect the skin, encourage healing and prevent infection.[2]
Depending on disease severity, dressing changes can take several hours each day and can be extremely painful, often requiring the use of prescription pain medication.[2]

LIFE-LIMITING[3]



People with the most severe forms often don't survive past childhood.[1,2,3]

Those with more severe EB have a higher chance of developing an aggressive form of squamous cell carcinoma (SCC), a type of skin cancer.[1,2,6]

References 1. Debra UK (2018). What is EB?. https://www.debra.org.uk/faqs/faqs. Last Accessed February 2021. **2.** Denyer J, Pillay E, Clapham J. Best practice guidelines for skin and wound care in epidermolysis bullosa. An International Consensus. Wounds International, 2017. **3.** EB Research Network (2021). https://www.eb-researchnetwork.org/research/what-is-eb/. Last Accessed February 2021. **4.** Debra. EB in Depth. https://www.debra.irg/about-eb/eb-depth. Last Accessed February 2021. **5.** DEBRA Ireland (2018). What is EB? Available at: https://debraireland.org/about/what-is-eb/. Last Accessed February 2021. **6.** Hernández-Martín A, Torrelo A. Inherited Epidermolysis Bullosa: From Diagnosis to Reality Actas. Dermosiiliogr. 2010;101(6):495–505. **7.** Genetics Home Reference (2018). Dystrophic epidermolysis bullosa. Available at: https://ghr.nlm.nih.gov/condition/dystrophic-epidermolysis-bullosa#inheritance. Last Accessed February 2021. **8.** Intong et al. Inherited epidermolysis bullosa: New diagnostic criteria and classification. Clinics in Dermatology 2012;30:70–77. **9.** Marinkovich MP (2018). Epidermolysis Bullosa. Available at: https://emedicine.medscape.com/article/1062939-overview#a2. Last Accessed February 2021. **10.** DEBRA UK (2018). Epidermolysis Bullosa: A Guide for Parents, Schools and Playgroups. Available at: https://www.debra.org.uk/downloads/community-support/parents-schools-and-playgroups.pdf. Last Accessed February 2021. **11.** Debra International. FAQ. https://www.debra-international.org/faq. Last Accessed February 2021. **12.** Bodan RC. Epidermolysis Bullosa: An Insider's Perspective to a Rare Genetic Connective Tissue Disorder. Journal of the Dermatology Nurses' Association. 2016;8(1):46-56. Available at: http://www.nursingcenter.com/cearticle?an=01412499-201601000-00006&Journal_ID=849729&Issue_ID=3353323. Last Accessed February 2021. **13.** Davila-Seijo P et al. Current dystrophic epidermolysis bullosa research does not match research needs perceived by patients and clinicians Journal of the American Academy of Dermatology 2014;71(5):1008–1011.

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THREE MAIN TYPES OF EB[1]

SIMPLEX (EBS)

Although often life-altering, this is the most common subtype of EB,[3] but can be less severe compared to other EB sub-types.[2] With the exception of its most severe types, EB simplex only affects the skin; blistering is limited, sometimes just restricted to the hands and feet and where clothing causes friction. Heat and humidity can also cause the condition or make it worse.[2]

DYSTROPHIC (DEB)

DEB is one of the major forms of epidermolysis bullosa.[7] In mild cases, blistering may primarily affect the hands, feet, knees, and elbows.[7] Severe cases involve widespread blistering and wounding that can lead to blindness, scarring and deformity and rigidity of joints, resulting in poor mobility.[1,7,8] In such cases, there is also a high chance of developing squamous cell carcinoma of the skin.[3]

JUNCTIONAL (JEB)

A rare form of EB.[1] JEB has a broad spectrum of severity from milder forms to the lethal form of generalised severe JEB with most (87%) not surviving beyond the first year of life.[9] In all forms of JEB, the most problematic wounds occur on the scalp and lower legs.[2]

PSYCHOLOGICALLY CRUSHING

Not only do children live with significant physical pain and discomfort, but they also face deep social and psychological impacts.[10,11] Many children are able to attend school, although they require extra care and are often not able to take part in all the same activities. However, some children are not able to attend school at all, which can have a huge impact on their social life and education.

Carers are often parents who face the very real and unthinkable prospect of never being able to properly cuddle their children. They regularly find themselves as their child's nurses, having to go through the difficult routine of changing bandages and dressing wounds while watching their child in pain.[12]

INTO THE FUTURE[13]

Children living with EB and their caregivers' greatest need is for better ways to manage the daily consequences of their condition, especially their wounds, unbearable itch and pain. There is increasing focus on prevention and treatment of the aggressive skin cancers associated with EB.

With the breadth of research on genetic therapies, there is great hope and expectation for a cure to be developed for some sub-types or EB in the longer term.

AMRYT PHARMA

References 1. Debra UK (2018). What is EB?. https://www.debra.org.uk/faqs/faqs. Last Accessed February 2021. **2.** Denyer J, Pillay E, Clapham J. Best practice guidelines for skin and wound care in epidermolysis bullosa. An International Consensus. Wounds International, 2017. **3.** EB Research Network (2021). https://www.eb-researchnetwork.org/research/what-is-eb/. Last Accessed February 2021. **4.** Debra. EB in Depth. https://www.debra.irg/about-eb/eb-depth. Last Accessed February 2021. **5.** DEBRA Ireland (2018). What is EB? Available at: https://debraireland.org/about/what-is-eb/. Last Accessed February 2021. **6.** Hernández-Martín A, Torrelo A. Inherited Epidermolysis Bullosa: From Diagnosis to Reality Actas. Dermosiiliogr. 2010;101(6):495–505. **7.** Genetics Home Reference (2018). Dystrophic epidermolysis bullosa. Available at: https://ghr.nlm.nih.gov/condition/dystrophic-epidermolysis-bullosa#inheritance. Last Accessed February 2021. **8.** Intong et al. Inherited epidermolysis bullosa: New diagnostic criteria and classification. Clinics in Dermatology 2012;30:70–77. **9.** Marinkovich MP (2018). Epidermolysis Bullosa. Available at: https://emedicine.medscape.com/article/1062939-overview#a2. Last Accessed February 2021. **10.** DEBRA UK (2018). Epidermolysis Bullosa: A Guide for Parents, Schools and Playgroups. Available at: https://www.debra.org.uk/downloads/community-support/parents-schools-and-playgroups.pdf. Last Accessed February 2021. **11.** Debra International. FAQ. https://www.debra-international.org/faq. Last Accessed February 2021. **12.** Bodan RC. Epidermolysis Bullosa: An Insider's Perspective to a Rare Genetic Connective Tissue Disorder. Journal of the Dermatology Nurses' Association. 2016;8(1):46-56. Available at: http://www.nursingcenter.com/cearticle?an=01412499-201601000-00006&Journal_ID=849729&Issue_ID=3353323. Last Accessed February 2021. **13.** Davila-Seijo P et al. Current dystrophic epidermolysis bullosa research does not match research needs perceived by patients and clinicians Journal of the American Academy of Dermatology 2014;71(5):1008–1011.

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