

# EB

# is Epidermolysis Bullosa

'Hard to say. Hell to live with'

**FRAGILE SKIN CONDITION** Friction causes blistering, skin loss, open wounds, sores and can lead to skin cancer

**WHY** Skin proteins that 'glue' skin layers together are defective or missing



**1 in 227** of us carry a defective gene that causes EB **Affects 500,000** people worldwide

**ANYBODY** Equally affects males and females across every ethnic group

**GENETIC** Inherited, though a small number may be acquired

**NOT CONTAGIOUS** You cannot 'catch' EB as it is genetic

**NO TREATMENT NO CURE** yet! But Research is progressing fast

**DIAGNOSIS** Microscopic evaluation of a skin biopsy to see which layer is affected

**SKIN**

**TYPES OF EB** The layer of blistering determines the type of EB:

**EPIDERMIS**

**SIMPLEX** Mildly affected may have blistering on hands and feet. In more severe cases, whole body blistering can be extensive. *Whilst painful and debilitating the majority lead long lives*

**BASEMENT LAYER**

**JUNCTIONAL** Blistering can be extensive over the whole body and internal organs such as mouth, oesophagus and trachea. Herlitz Junctional is rapidly fatal with 87% dying in the first year of life. It is extremely painful due to extensive internal blistering to mouth, nasal passages, trachea, oesophagus, stomach, intestine, making even breathing very painful

**DERMIS**

**DYSTROPHIC** Deep wounds leading to scarring and contractures around joints. Mitten deformities of fingers and toes, contracture of mouth, narrowing of oesophagus and corneal abrasions, severe pain and increasing disability leading to malignant aggressive skin cancer (squamous cell carcinoma) in early adulthood

**HOW**



**RECESSIVE**

Both parents carry the gene but unaffected and usually don't know **25% chance of child having it**



**SPONTANEOUS MUTATION**

Neither parent carries EB. The gene mutates spontaneously in either the sperm or egg before conception



**DOMINANT**

One parent carries the gene for EB and is affected by the condition themselves **50% chance of passing on**



**DAILY MANAGEMENT**

**BLISTERS** Have to be popped with a needle and dressed

**BANDAGING** To cover open wounds to prevent infection and protect skin from further friction

*Daily bandaging can take hours and is very painful*

**NUTRITION** Small mouth opening due blistering and scarring. Swallowing impaired from narrowing and pain

*Stomach tube and supplements often needed*

**EYE CARE** Regular eye ointments to prevent corneal abrasions which are extremely painful



**HELP**

**FIGHT FOR A LIFE FREE OF PAIN. TO END EB. RESEARCH THE CURE**

Research is being translated into clinical trials. Gene modification and gene correction technologies make cures a realistic possibility. The challenge is bringing forward effective treatments for all types of EB. **PLEASE DONATE**

**#cureEB 100% OF DONATIONS GO TO FUND RESEARCH**

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This is a generalisation of the three main types of EB. For accurate diagnosis, if you suspect your child may have EB please contact your local doctor. Sohana Research Fund is a charity registered in the England & Wales 1158672

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