Epidermolysis Bullosa (EB)
Fact Sheet

debra
Working for a life free of pain.
Pour une vie sans douleur.

What Is Epidermolysis Bullosa?

Epidermolysis Bullosa (EB) is a group of genetic conditions, together affecting approximately 1 in 17-20,000 live births, with an estimated 500,000 people worldwide living with EB. The condition is always painful, frequently very disabling and life threatening and, in its most severe forms, fatal in infancy. EB affects both genders and every race and ethnicity.

There are currently three main types of the condition recognised: Simplex, Dystrophic and Junctional. These are defined by how deeply in the tissue blistering occurs. Each type has multiple subtypes, but as knowledge increases about the condition, classification of types and subtypes is refined. “Hemidesmosomal“ EB is sometimes considered to be a fourth type of EB; this and other previously-known skin blistering conditions, such as Kindler syndrome, are now considered to be part of the broader EB classification.

What Does The Name Mean?

Skin has two layers - the outer is called the epidermis and the inner layer is the dermis. Between the layers is the basement membrane zone, where anchoring fibers connect the two. "Bullosa" refers to the Latin words “bullae” or “bullous,” meaning blisters, and “lysis" in Greek means to break down. Hence, Epidermolysis Bullosa refers to the breakdown and blistering of the skin. Mucosal linings may also be affected in severe types.

What Causes EB?

All forms of EB are genetic in origin, and the genes responsible for most subtypes of the disease are now known. Some still await identification. Proteins affected include Collagen VII and XVII, Keratin 5 and 14, Laminin, Integrin and Plectin. People with Dystrophic EB have a greater incidence of squamous cell carcinoma, an aggressive form of skin cancer. Recessive types are seen more frequently when parenting occurs between close relatives.

What Are Butterfly Children?

The youngest with EB are called butterfly children because their skin is said to be as fragile as a butterfly's wing.

Is Epidermolysis Bullosa Infectious or Contagious?

Definitely not. However, the wound themselves can easily become infected by germs. This causes further pain, and infection delays healing. More severe types are transmitted through recessive inheritance; that is, both parents carry the gene for the disorder but are not themselves affected by it. With each pregnancy, there is a 25% chance the baby will be affected; a 50% chance that the baby will have every appearance of being unaffected but be a “carrier” like the parents; and a 25% chance that the baby will be entirely unaffected. As nature is capricious, some families have the terrible misfortune to have most, or even all, of their children affected by EB. Most people are entirely unaware that they are carriers. Also, fresh mutations of the gene take place from time to time. This is rare – but it could happen.

Does the disorder impair mental health?

Not at all. Despite the considerable disfigurement EB can cause, the mental health of people with EB is normal. Indeed, they often have above-average intelligence, which makes it all the more difficult to come to terms with physical limitations. They are more than ever aware of the restrictions placed on them, and that many of their hopes and ambitions cannot be fulfilled.

Is there a cure?

EB is currently incurable, in all types. There is no effective treatment for EB. Infants are often born with areas of the skin completely missing, and EB wounds do not heal like normal skin. As an “orphan disease” (being rare), it is challenging to fund research toward the development of a cure, but this is one of the missions of the DEBRA organization.

How Can I Help/Learn More?

More information and donations are accepted through the DEBRA Canada website at:
www.debracanada.org

To speak with a representative of DEBRA Canada call 1-800-313-3012, or email:

Erin Hoyos (Project Coordinator/Administrative Officer) at: ehoyos@debracanada.org
Tina Boileau (President) at: tina@debracanada.org
Ryan Hultman (Vice President) at: ryan@debracanada.org