Epidermolysis bullosa (EB) is a general term for a group of diseases characterized by bullae, otherwise known as blisters, of the skin induced by minor mechanical trauma. These diseases, in one form or another, all involve defective adherence of the epidermis or dermis. In other words, the top layer or layers of the skin break loose from deeper layers of the skin. Patients may complain of developing many blisters after starting a new type of training (e.g., walking or running long distances for a job or new sport), or blisters may occur after brushing up against a wall. In the most aggressive forms, blisters and losing the top layer of the skin may occur early in life starting with the process of being born.

Epidermolysis bullosa dates back to 1870 and over the years with the progress of technology has become better understood. To date, three major types of EB are characterized down to the molecular basis of their pathology. EB simplex involves defective structures that hold the epidermis (top layer of skin) together. Consequently, the epidermis breaks apart in two. Junctional EB is characterized by defective structures that hold the epidermis to the dermis. Patients with junctional EB experience the top layer of the skin coming apart from the bottom layer of the skin. Finally, dystrophic EB is categorized by having the epidermis and part of the top layer of the dermis coming apart from the bottom part of the dermis. While there are exceptions to this generalization, EB severity is partially due to the main subtype of EB. The severity generally correlates to the depth of the faulty skin structures; the deeper the skin does not adhere, the worse the disease can be.

EB simplex is one of the major three types of EB, and mostly characterized by faulty structures called keratin filaments (K5 and K14) which hold the epidermis together. The prevalence of EB simplex is about 4.6 per 1 million people in the United States. Most forms of EB simplex are inherited in an autosomal dominant fashion. Consequently, if one parent has the gene and the disease, then each child of that parent has about a 50% of inheriting the disease. There are multiple subtypes of EB simplex, but the most frequently encountered subtypes are generalized EB simplex and localized EB simplex. Localized EB simplex, also known as the Weber-Cockayne subtype, is the most frequently occurring type of EB simplex. Most people start to experience symptoms when they are a child or later in life. Symptoms include tense blisters of the feet, hands, or other areas after repetitive trauma like running or working with the hands. Patients may also report excessive sweating of the palms and soles of the feet. Generalized EB simplex, also known as the Koebner variant, is characterized by symptom onset from birth to early infancy. As the name implies, blistering occurs all over the body and especially the knees, feet, hands, and elbows. Thickening of the skin on the hands can occur. For the most part EB simplex does not cause any scarring, but other complications such as infection can occur.

Junctional EB, another of the major types of EB, commonly entails malfunctioning structures within the space between the epidermis and dermis called the lamina lucida. Junctional EB exalts a prevalence of about 0.4 per 1 million people in the U.S. The inheritance is autosomal recessive. In other words, one copy of the gene needs to be present in both parents for the disease to pass along. Each child of those parents has about a 25% chance of getting the disease, while 50% have a chance of being a carrier. The three major subtypes of junctional EB are the Herlitz, Mitis, and the non-Herlitz types. The Herlitz (also known as Gravis) subtype is the most severe of the subtypes and patients usually do not live past infancy. Infants have blistering with loss of skin all over their bodies. Fingernails are usually lost and mucosal surfaces throughout the body blister and can develop scars. Because of the disease, the lungs, genitourinary, and digestive system including the esophagus also undergo blistering and possibly subsequent scarring. The Mitis subtype also starts during the infancy period, but children usually survive this period and the disease severity lessens with age. Patients commonly develop blisters and erosions around the mouth, nose, and eyes during childhood. Finally, the non-Herlitz subtype of junctional EB (sometimes referred to by its preceding name of generalized atrophic benign EB) also presents after being born with blisters all over the body. Patients always survive to be.
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Adults, but the diffuse blistering over their skin surface continues to occur. Some scarring can occur along with dystrophic nails, hair loss, mucous membrane involvement, and damage to the enamel of the teeth.

Dystrophic EB is the most severe of the three major types of EB, and develops from malfunctioning anchoring fibrils in the upper portion of the dermis. The prevalence is about 0.9 per 1 million people in the US, and passed along to children in an autosomal dominant or autosomal recessive distribution. In patients with dominant dystrophic EB, symptoms usually start to appear in infancy. Patients experience blisters around the toes, fingers, ears, and nose. Nail dystrophy and scarring is also common. Recessive dystrophic EB tends to be more severe than dominant dystrophic EB, and can range from localized disease to diffuse blisters and scarring. The recessive form is present at birth, and progressively becomes more debilitating as the patient ages due to diffuse scar formation of the skin and mucosal surfaces throughout the body. Deformities and contractures of the hands and feet can develop from the repeated scarring. Enamel erosions predispose to cavities of the teeth, and strictures often form in the esophagus or urethra. Secondary consequences of the disease include malnutrition with subsequent growth slowing, anemia, constipation, osteopenia, osteoporosis, and dilated cardiomyopathy. Some patients are also at an increased risk of multiple squamous cell carcinomas of the skin.

Diagnosis of EB is by clinical history and presentation combined with biopsy. Biopsy can help further characterize the specific diagnosis by processing for microscopic examination. In some cases, tests that characterize the DNA for a specific gene mutation may be used.

Currently, there is no cure for EB, although research is gaining momentum for treating specific forms of EB. Generally, the severity of the disease determines the treatment. Patients with severe active disease are commonly treated in burn units. These patients often experience a multidisciplinary approach to treatment. Others with less severe disease practice more preventative measures to blistering such as keeping the skin cool and dry, loose clothing, and avoiding trauma directly or with soft support of body parts. Most all patients need proper wound treatment combined with nutritional maintenance and prevention of infection. Some patients may accomplish reduction of infections with bleach baths.