Epidermolysis Bullosa: An Overview of Current and Future Potential Treatment Options

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Epidermolysis Bullosa (EB) is a rare inherited condition of which the most common clinical finding is blistering and erosions of the skin following minor trauma. While this is the most well-known manifestation of EB, there are many other complications, including: gastrointestinal malabsorption, various genitourinary fusions of skin, glomerulonephritis, contractures and fusions of joints, dental abnormalities, corneal scarring, respiratory tract scarring, skin cancer, and cutaneous infections, which can sometimes lead to sepsis and eventually death. EB consists of four main classifications, and 34 distinct subtypes. The four classifications are Epidermolysis Bullosa Simplex (EBS), Junctional Epidermolysis Bullosa (JEB), Dystrophic Epidermolysis Bullosa (DEB), and Kindler Epidermolysis Bullosa (KEB). These classes are distinguished by the layer of skin in which the formation of blisters occurs: EBS is formed in the intraepidermal layer, JEB is formed in the lamina lucida of the basement membrane, DEB is formed below the basement membrane, and Kindler's EB can involve multiple or all these layers. Treatment for EB until recently has largely been supportive care, focusing on symptom management and proper nutritional support. Treatments that are appearing now include gene therapy, stem cell therapy, and placental-based therapies. Gene therapy involves the actual splicing of affected genes that are responsible for adhesions of skin layers associated with certain types of EB. Stem cell and placental-based therapies focus more on regenerating skin for the EB patient, although studies are ongoing. Research continues for treatments to combat this rare genetic disease.