Epidermolysis Bullosa

On the basis of clinical, histologic, and electron microscopic findings, three groups of EB are recognized.
In all types of EB, the blisters form as a result of minor trauma. Because of the great differences in prognoses, families with the potential of having an infant born with one of the frequently or potentially fatal forms of EB, such as EB letalis or generalized EB dystrophica-recessive, a prenatal biopsy at 18 to 20 weeks of gestation is recommended. On electron microscopy, EB letalis shows abnormalities of the hemidesmosomes while generalized EB dystrophica-recessive shows absence of anchoring fibrils.

There is more variability in the clinical course in some forms of EB than was previously appreciated. For example, within the epidermal type of EB, which usually has a good prognosis, some cases of EB herpetiformis, which is known also as the Dowling-Meara variant, may show generalized blistering that at times is associated with mortality during early infancy. Also, the junctional form of EB can result in scarring as cicatricial junctional EB. Occasionally, dermal EB may be transient and heal within a few months. It is likely that the majority of cases published as Bart’s syndrome, which was originally described as congenital absence of the skin, belong in this group.
Histopathology
If a fresh blister is available, a specimen for biopsy may be taken from its edge.
Even though electron microscopic examination (discussed later) is informative, the light microscopic features seen in the various forms of EB are of diagnostic value.

In epidermal EB, which includes EB simplex, EB foot and hands of Weber and Cockayne, and EB herpetiformis (Dowling-Meara), the basement membrane zone is stained with the PAS technique. The PAS-positive basement membrane zone is located on the dermal side of the blister.
In *junctional EB*, the trauma of having a specimen taken for biopsy generally is sufficient to induce separation. This separation is located between the epidermis and the dermis, with the PAS-positive basement membrane zone usually remaining with the dermis. In some cases of EB letalis, autopsy has revealed... are no morphologic or enzymatic abnormalities to distinguish the atrophic benign form of *junctional EB* from EB letalis.

In *EB dystrophica-dominant* and *EB dystrophica-recessive* light microscopy shows dermal-epidermal separation. A PAS stain is of little help in ascertaining the exact level of cleavage because the PAS-positive basement...
membrane zone often appears hazy. If recognizable, it is seen in contact with the detached epidermis or appears split.

*EB acquisita* is not a genodermatosis but an autoimmune disorder.
Pathogenesis. If possible, all specimens of artificially induced blisters should be subjected to electron microscopic examination. In the epidermal types of EB, electron microscopic examination shows that cleavage is the result of degenerative cytolytic changes occurring in the lower portion of the basal cells between the dermal-epidermal junction and...
nucleus (EM 4). Immunofluorescence mapping shows that all three antigens (type IV collagen, laminin, b...
abnormalities of the hemidesmosomes are a secondary phenomenon and that the basic cause of the junctional types of EB is an epidermal intracellular deficiency. A new mutation of the gene encoding beta integrin has been found in the subset of EB letalis with pyloric atresia.
The dermal types of EB, on electron microscopy, show abnormalities in regard to their anchoring fibrils.