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# Junctional epidermolysis bullosa

Wing Yan Yuen  
Groningen, 6 juni 2012

1. Junctional epidermolysis bullosa type Herlitz is caused by a total absence of functional laminin-332 and leads to childhood lethality. *(this thesis)*
2. The care for patients with junctional epidermolysis bullosa type Herlitz should be focused on comfort, while conventional invasive procedures aimed for prolongation of life should be avoided. *(this thesis)*
3. Health care professionals should not only provide the best care for epidermolysis bullosa patients, but also for their parents. *(this thesis)*
4. Pathogenic *ITGB4* mutations cause a phenotypic spectrum ranging from a lethal subtype with pyloric atresia to localized blistering. *(this thesis)*
5. Adults with junctional epidermolysis bullosa have an increased risk of developing squamous cell carcinomas and should be annually checked from age 25 on. *(this thesis)*
6. Punch grafting is an easy, inexpensive, safe and effective treatment for chronic non-healing ulcers in junctional epidermolysis bullosa type non-Herlitz patients. *(this thesis)*
7. Haploinsufficiency of the laminin alpha 3 chain can lead to enamel defects. *(this thesis)*
8. Junctional epidermolysis bullosa of late onset is caused by mutations in *COL17A1*. *(this thesis)*
9. Due to the different biological turnover of type XVII collagen and its shed ectodomain, antibodies staining against its endodomain are the most sensitive in detecting a reduced type XVII collagen expression. *(this thesis)*
10. Mutations in the *COL17A1*, *ITGB4* and *DST* genes are rarely found in basal epidermolysis bullosa simplex and should only be screened in cases with abnormalities in immunofluorescence antigen staining, and excluded in cases with an autosomal dominant inheritance. *(this thesis)*
11. A validated and reliable measurement tool for the quality of life in epidermolysis bullosa patients is important in the assessment of future therapies. *(this thesis)*
12. Patience is bitter, but its fruit is sweet. *(Jean-Jacques Rousseau)*
13. All things are difficult before they are easy. *(dr. Thomas Fuller)*
14. The cure for boredom is curiosity. There is no cure for curiosity. *(Dorothy Parker)*