

University of Groningen

Substantiating atypical phenotypes of epidermolysis bullosa

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STELLINGEN BEHORENDE BIJ HET PROEFSCHRIFT

Substantiating atypical phenotypes of epidermolysis bullosa

1. The plectin 1a isoform is the dominant plectin isoform of the basal cells in the epidermis.
(this thesis)
2. A homozygous nonsense mutation only affecting the plectin 1a isoform results in skin-only epidermolysis bullosa simplex. *(this thesis)*
3. Introns can harbor pathogenic mutations in the known epidermolysis bullosa simplex genes.
(this thesis)
4. Alternate splicing of common plectin isoforms also occurs for exon 8 of the *PLEC* gene. *(this thesis)*
5. Patients with dystrophic epidermolysis bullosa may experience excruciating itch on clinically healthy skin. *(this thesis)*
6. Haploinsufficiency of *LAMA3* manifests as subtle focal enamel pitting in the absence of skin fragility. *(this thesis)*
7. Despite the use of next generation sequencing, the practice of taking skin biopsies for antigen mapping in the diagnostics of epidermolysis bullosa is not obsolete. *(this thesis)*
8. I skate to where the puck is going to be, not where it has been. -**Wayne Gretzky, Canadian hockey legend**
9. Keep your heels, head and standards high. -**Coco Chanel**
10. There's no place like home. -**Dorothy, "The Wizard of Oz" by L. Frank Baum**