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Cold cases in epidermolysis bullosa: not the usual suspects

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Cold cases in epidermolysis bullosa: not the usual suspects



1. Heterozygosity for a novel missense mutation c.433G>T, p.Asp145Tyr in the *ITGB4* gene is associated with autosomal dominant epidermolysis bullosa. (*this thesis*)
2. A mottled pigmentation clinical phenotype and epidermolysis bullosa simplex are associated with homozygous nonsense mutations in *EXPH5*. (*this thesis*)
3. Mutations in *EXPH5* result in disruption of the keratin filament cytoskeleton, such event underscores a role for keratins in pigmentary disturbances. (*this thesis*)
4. A distal truncation of the BPAG1-e molecule caused by a homozygous *DST* mutation can result in an intermediate generalized phenotype with prurigo papules. (*this thesis*)
5. Epidermolysis bullosa simplex resulting from *DST* mutations has various patterns of heritability. (*this thesis*)
6. *PLEC* may function as a genetic modifier for *DST*. (*this thesis*)
7. The lamina lucida shows two distinctive cleavage patterns in junctional epidermolysis bullosa, contingent to which molecules are involved. (*this thesis*)
8. Laminin 322 co-localizes with the hemidesmosomes in the blister roof in junctional epidermolysis bullosa skin. (*this thesis*)
9. Even if man were nothing but a piano key, even if this was proved to him by natural science and mathematics, even then he would not become reasonable, but would purposely do something perverse out of sheer ingratitude, simply to have his own way...then, after all, perhaps only by his curse will he attain his object, that is, really convince himself that he is a man and not a piano key! If you say that all this, too, can be calculated and tabulated...then man would purposely go mad in order to be rid of reason and have this own way.
(*Notes From Underground by Fyodor Dostoyevsky*)
10. There are some games you don't get to play unless you are all in. (*Jordan B. Peterson*)
11. You can never hold back spring. (*Tom Waits*)