

Clinical and molecular genetic studies in hereditary hair loss

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Stellingen

behorende bij het proefschrift

“Clinical and molecular genetic studies in hereditary hair loss”

1. Hypotrichosis congenita Marie Unna is associated with a 360 kb locus on chromosome 8p21 and may be caused by mutations in a regulatory element (*this thesis*)
2. Keratitis-ichthyosis-deafness syndrome is identical to ichthyosis-like hystrix-deafness syndrome (*this thesis*)
3. *GJB4* (connexin30.3) is not required for normal skin function and for normal hearing (*this thesis*)
4. Clouston syndrome can mimic pachyonychia congenita (*this thesis*)
5. Absence of the C-terminus of *GJA1* leads to severely disturbed transport of the mutant protein and its wild type counterpart. This observation may explain the skin symptoms caused by the truncation (*this thesis*)
6. Dominant mutations in gap junction genes can cause skin symptoms, recessive ones cannot (*this thesis*)
7. The feasibility of gene therapy approaches targeting splice-site mutations is aptly demonstrated by the partial phenotypic rescue of a *plakophilin-1* splice site mutation by use of a cryptic splice site (*this thesis*)
8. Nature’s experiments on human subjects would never have been approved by an ethics committee, hence we have a moral as well as scientific obligation to study them
9. The phenotype resulting from a gene knockout does not reflect the absent protein’s function; rather, it reflects the actions that the protein network that it was in takes to compensate for its absence
10. Without a profound understanding of the proper muscle tension and breathing pattern in Sanchin *kata*, the Sanchin *afficionado* will develop hemorrhoids or worse
11. An Apple a day keeps the help desk away