Clinical and molecular genetic studies in hereditary hair loss

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. *GJB*4 (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