

Stellingen
(Propositions)

behorende bij het proefschrift

Analysis of ABCC6:
Elucidation of the Molecular Pathology of Pseudoxanthoma Elasticum

1. A complete loss of transport function of the ABCC6 protein is the primary cause of PXE (this thesis).
2. A founder effect for the R1141X mutation exists in Dutch patients with PXE (this thesis).
3. Autosomal dominant inheritance in PXE may exist, but is much rarer than previously thought (this thesis).
4. ABCC6 protein is localized on the basolateral membrane of liver and kidney cells in humans (this thesis).
5. The PXE phenotype might be a consequence of calcification of elastic fibers resulting from systemic or local ABCC6 defects (this thesis).
6. Classifying genetic disorders as monogenic or multifactorial might be an oversimplification (Scriber C.R. & Warter P.J. 1999, Trends Genet 15: 267).
7. Predicting all encoded genes continues to be a significant challenge after the sequencing of the human genome (Daly M.J. 2002, Cell 109: 283).
8. Homologous recombination in human embryonic stem cells will be important for transplantation medicine (Rideout W.M. 2002, Cell 109: 17).
9. No species, ours included, possesses a general purpose beyond the imperatives created by its genetic history (Wilson E.O. 1978, On Human Nature).
10. The knowledge of biology will make sense of my self, my genes, and my memes (Blackmore S, 1999, The meme machine).
11. Enlarging and disseminating knowledge are equally important activities and each is done better when both are done in the same place by the same people (Kennedy D. 1997, Academic Duty).

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