

Stellingen

Bij het proefschrift

Thyroid Hormone Transporters: from Molecules to Patients

1. The neurocognitive features and consequences of the chronic thyrotoxicosis in peripheral tissues comprise the two main clinical entities of MCT8 deficiency, which both predispose patients to significant morbidity and premature death (dit proefschrift).
2. The T3-analogue Triac holds therapeutic potential in MCT8 deficiency by reducing serum T3 concentrations and alleviating the hyperthyroid state in peripheral tissues (dit proefschrift).
3. Thyroid function tests, including serum T3 concentrations, should be measured in all children with unexplained intellectual disability, as MCT8 deficiency is not detected in current neonatal screening programs (dit proefschrift).
4. Novel missense mutations in MCT8 should always be functionally evaluated *in vitro* and/or *ex vivo* to substantiate a causal relation with the observed clinical phenotype (dit proefschrift).
5. Although many transporters have been shown to mediate thyroid hormone transport *in vitro*, most of these transporters likely have a minimal contribution to cellular thyroid hormone homeostasis *in vivo* (dit proefschrift).
6. Collaborations are invaluable to achieve advances in research to rare disorders (KE Foley *Nat. Med.* 2015; 21:1242-1243).
7. The wide implementation of next generation sequencing in clinical practice will facilitate early diagnosis of (novel) rare genetic disorders, but will also increasingly demand collaboration between basic science and clinical practice to safeguard correct interpretations of the results (M Tarailo-Graovac *et al. N. Engl. J. Med.* 2016;374:2246-2255).
8. The increasing application of personalized medicine and development of individualized therapeutic approaches may improve the effectiveness of medical care, but will increase the pressure on the medical system from an ethical and financial perspective (J Woodcock and P Marks *N. Engl. J. Med.* 2019;381:1678-1680).
9. False facts are highly injurious to the progress of science, for they often endure long; but false views, if supported by some evidence, do little harm, for everyone takes a salutary pleasure in proving their falseness (Charles Darwin).
10. Creativity in research, through logic, genius, and zeitgeist, needs an open and liberal culture, which is currently threatened by over-management and over-regulation of health care (based on R Sullivan *Lancet Oncol.* 2008;9(1):2-3).
11. Op de schouders van een reus kan zelfs een dwerg ver zien.

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