Stellingen behorende bij het proefschrift

Enzyme-replacement therapy in classic infantile Pompe disease

long-term outcome, dosing, and the role of antibodies

Carin van Gelder
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1. Now that ERT enables patients with classic infantile Pompe disease to live longer, a new phenotype is emerging that differs from that of children and adults with non-classic Pompe disease. *(dit proefschrift)*

2. New features in long-term survivors are facial-muscle weakness and ptosis, speech disorders, dysphagia, impaired motor function, and distal muscle weakness. *(dit proefschrift)*

3. Although ERT is unlikely to cross the blood-brain barrier in patients with classic infantile Pompe disease treated with ERT, glycogen storage seems to have limited impact on the function of the CNS during childhood. *(dit proefschrift)*

4. Clinical outcome in patients with classic infantile Pompe disease treated with ERT can be improved by dose augmentation, immunomodulation, and neonatal screening. *(dit proefschrift)*

5. The pathophysiological mechanisms of LSDs should be further elucidated, as they might prompt the development of new and ultimately curative therapies. *(dit proefschrift)*

6. Centralisatie van zorg vraagt extra tijd van de patiënt maar leidt tot kwalitatief betere zorg.

7. De klinische relevantie moet leidend zijn bij het beoordelen van de effectiviteit van een geneesmiddel.

8. "As to diseases, make a habit of two things: To help, or at least, to do no harm." *(Hippocrates)*

9. "Nothing's ever for sure. That's the only sure thing I do know." *A beautiful mind.*

10. "They do certainly give very strange, and newfangled, names to diseases." *(Plato)*

11. Een dag niet genieten is een dag niet geleefd. *(G. van den Berg)*