# STELLINGEN

behorende bij het proefschrift

## Genetic Causes of Cerebrovascular Disorders in Childhood

1. *COL4A1* and *COL4A2* testing should be considered in porencephaly and/ or germinal matrix hemorrhage of antenatal origin or postnatal origin in infants born after 30 weeks gestation. (this thesis)

2. In familial *COL4A1* or *COL4A2* mutations, modifying factors seem important contributors to the phenotype, postulating that these *COL4A1* or *COL4A2* mutations can be regarded as risk factors. (this thesis)

3. Although an indirect link between up-regulation of the type I interferon signalling pathway and cerebrovascular pathology has been suggested in Aicardi-Goutières syndrome, *USP18* mutations provide the first direct evidence that this link is correct. (this thesis)

4. *USP18* mutation testing should be considered in pseudo-TORCH syndrome with severe cerebral hemorrhage of antenatal or neonatal onset. (this thesis)

5. *USP18* missense mutations, as well as mutations in other genes involved in ependymal dysfunction, are likely to be genetic causes of human congenital hydrocephalus.

6. In exome sequencing, enthousiasm about identified possible pathogenic mutations can lead to tunnel vision, which underscores the importance of looking at what is missed.

7. In view of the "genotyping first" approach that will characterize the near future of clinical genetics, the modern geneticist might need to subspecialize in molecular mechanisms and become a pathway expert that interprets the molecular data in view of this knowledge.

8. The doctrine that we are the product of our DNA leads to the fantasy that by manipulating [it] we could avoid or cure all disease and even escape eventual death. That is indeed a fantasy. All flesh is mortal (Richard Lewontin, 2009)

9. Samenwerken is meer dan "samen werken".

10. Wetenschappelijk onderzoek doen is als zwemmen; het vereist regelmatig zuchten, af en toe een keerpunt en een lange adem.

11. Verhuizen naar België maakt Kafka's "Het Slot" een stuk realistischer.

Marije Meuwissen, 2 april 2014