Stellingen

‘Finding Genes for Parkinsonism

1) The next generation sequencing technologies, including exome and whole-genome sequencing, offer unprecedented opportunities to dissect the genetic architecture of the complex traits. *(this thesis)*

2) Because of its geographical, historical, and cultural characteristics, the population of Sardinia has a unique genetic background. *(this thesis)*

3) The association between the TARDBP mutation and Parkinson’s disease supports an involvement of the TDP-43 protein in a broader neurodegenerative spectrum, including not only motoneuron disease and FTLD, but also PD. *(this thesis)*

4) The genetic defect in the SYN1 gene delineates a novel form of human Mendelian parkinsonism, and provides further evidence for abnormal synaptic vesicle recycling as a central theme in the pathogenesis. *(this thesis)*

5) The identification of SLC30A10 mutations delineates the first primary disease of manganese metabolism. *(this thesis)*

6) Conquering genetic disorders requires genetic thinking: mapping and genome sharing among affected individuals continue to be *sine qua non* tools for establishing a link between a genomic variant and its phenotypic consequences. *(Stylianos Emmanuel Antonarakis, 2010)*

7) The only way to have complete confidence in a finding is independent replication. *(David Botstein, 2003)*

8) The positional cloning allows one to find where a gene is, without knowing what it is. *(Gonçalo Abecasis)*

9) The human genome has a certain tendency to incite passion and excess. *(Eric Lander, 2001)*

10) Truth is what stands the test of experience. *(Albert Einstein)*

11) Between the idea and the reality, between the motion and the act, falls the shadow. *(Thomas Stearns Eliot, 1925)*