

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 FTL, but also PD. (*this thesis*)
- 4) The genetic defect in the *SYNJ1* 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 Land-er, 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*)