The genetics of spinocerebellar ataxia and dystonia

Nibbeling, Esther

Stellingen bij ‘The genetics of spinocerebellar ataxia and dystonia’ – Esther Nibbeling

I. Spinocerebellar ataxia and dystonia should not be seen as separate entities, but rather as a disease continuum (this thesis)

II. Alterations in synaptic transmission and transcriptional regulation are shared mechanisms underlying different spinocerebellar ataxia types (this thesis)

III. Combining whole exome sequencing, targeted re-sequencing, and gene network analysis is a successful approach to identify novel disease genes (this thesis)

IV. A promising new mechanism in dystonia pathogenesis is disturbed calcium signaling (this thesis)

V. Functional analyses are essential to confirm mutations as definitely pathogenic (this thesis)

VI. Glutamate signaling may be the molecular link between spinocerebellar ataxia and intellectual disability (this thesis)

VII. Genetic research becomes easier with the help of family members (this thesis)

VIII. The list of genetic variant prediction tools will soon be longer than the list of variants per patient obtained by whole exome sequencing

IX. There are no perfect human specimens – we are all genetically flawed in some way
Francis Collins

X. The saddest aspect of life right now is that science gathers knowledge faster than society gathers wisdom
Isaac Asimov