

University of Groningen

The genetics of spinocerebellar ataxia and dystonia

Nibbeling, Esther

IMPORTANT NOTE: You are advised to consult the publisher's version (publisher's PDF) if you wish to cite from it. Please check the document version below.

Document Version

Publisher's PDF, also known as Version of record

Publication date:

2017

[Link to publication in University of Groningen/UMCG research database](#)

Citation for published version (APA):

Nibbeling, E. (2017). The genetics of spinocerebellar ataxia and dystonia. [Groningen]: Rijksuniversiteit Groningen.

Copyright

Other than for strictly personal use, it is not permitted to download or to forward/distribute the text or part of it without the consent of the author(s) and/or copyright holder(s), unless the work is under an open content license (like Creative Commons).

Take-down policy

If you believe that this document breaches copyright please contact us providing details, and we will remove access to the work immediately and investigate your claim.

Downloaded from the University of Groningen/UMCG research database (Pure): <http://www.rug.nl/research/portal>. For technical reasons the number of authors shown on this cover page is limited to 10 maximum.

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