

# Characterization of genetic neurodevelopmental disorders at adult age, with a focus on 22q11.2 deletion syndrome

Citation for published version (APA):

Boersma- von Scheibler, E. N. M. M. (2023). *Characterization of genetic neurodevelopmental disorders at adult age, with a focus on 22q11.2 deletion syndrome*. [Doctoral Thesis, Maastricht University]. Maastricht University. <https://doi.org/10.26481/dis.20231114eb>

## Document status and date:

Published: 01/01/2023

## DOI:

[10.26481/dis.20231114eb](https://doi.org/10.26481/dis.20231114eb)

## Document Version:

Publisher's PDF, also known as Version of record

## Please check the document version of this publication:

- A submitted manuscript is the version of the article upon submission and before peer-review. There can be important differences between the submitted version and the official published version of record. People interested in the research are advised to contact the author for the final version of the publication, or visit the DOI to the publisher's website.
- The final author version and the galley proof are versions of the publication after peer review.
- The final published version features the final layout of the paper including the volume, issue and page numbers.

[Link to publication](#)

## General rights

Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.

- Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
- You may not further distribute the material or use it for any profit-making activity or commercial gain
- You may freely distribute the URL identifying the publication in the public portal.

If the publication is distributed under the terms of Article 25fa of the Dutch Copyright Act, indicated by the "Taverne" license above, please follow below link for the End User Agreement:

[www.umlib.nl/taverne-license](http://www.umlib.nl/taverne-license)

## Take down policy

If you believe that this document breaches copyright please contact us at:

[repository@maastrichtuniversity.nl](mailto:repository@maastrichtuniversity.nl)

providing details and we will investigate your claim.

## **Characterization of genetic neurodevelopmental disorders at adult age, with a focus on 22q11.2 deletion syndrome**

Emma N.M.M. Boersma- von Scheibler, 14 november 2023, Maastricht

1. Adults with 22q11.2 deletion syndrome have an increased risk of Parkinson's disease and hearing loss compared to adults in the general population. (this thesis)
2. Results of the studies included in this thesis may indicate precocious aging in adults with 22q11.2 deletion syndrome. (this thesis)
3. Retinovascular parameters are potential biomarkers for neurodegenerative disorders in 22q11.2 deletion syndrome. (this thesis)
4. The co-existence of genetic neurodevelopmental and early-onset neurodegenerative disorders may indicate shared cellular and molecular mechanisms. (this thesis)
5. Natural history studies in adults with 22q11.2 deletion syndrome are important since they may generate knowledge that allows for a personalized approach by health care providers. (valorization)
6. Implementation and improvement of e-health is crucial to provide good health care to a growing population of individuals with (rare) genetic neurodevelopmental disorders.
7. Genetic testing may be beneficial at any age in individuals with an intellectual disability.
8. Intellectual disability medicine should be a standard part of the medical education curriculum.
9. Medicine is a science of uncertainty and an art of probability. (William Osler)
10. Failure is success in progress. (Albert Einstein)
11. Ook door de volwassenheid heen stroomt de rivier van mijn jeugd. (Frans Depeuter)