

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)