

Propositions belonging to the dissertation:

Improving Medical Care for Adults with Complex Rare Genetic Syndromes

A multidisciplinary approach

1. Multidisciplinary healthcare is essential to prevent under- and overdiagnostics and under- and overtreatment in adults with complex rare genetic syndromes (*this dissertation*).
2. Central adrenal insufficiency is rare in adults with Prader-Willi syndrome (*this dissertation*).
3. Growth hormone treatment in adults with Prader-Willi syndrome improves body composition without major safety concerns (*this dissertation*).
4. Fatigue in adults with Neurofibromatosis type 1 and Turner syndrome cannot solely be explained by somatic health disorders (*this dissertation*).
5. Height, body mass index, type of health problems and medication use differ between adults with and without intellectual disabilities (ID), which may aid in the development of an artificial intelligence driven “ID alert” to improve ID awareness among healthcare professionals (*this dissertation*).
6. Collaboration between basic and clinical science is crucial to improve medical care for adults with complex rare genetic syndromes.
7. Improving transition from pediatric to adult healthcare reduces the personal and financial burden of fragmented and suboptimal care (*Cassidy et al. BMJ Open. 2022. 12(12): e051314*).
8. Rare diseases are not that rare when you realize that one in every twenty persons is affected (*Nguengang Wakap et al. Eur J Hum Genet 2020. Feb;28(2):165-173*).
9. Negative findings are an essential part of scientific literature, because they encourage us to think critically, avoid publication bias, and move us towards a more balanced scientific landscape (*Matosin et al. Dis Model Mech. 2014. Feb;7(2):171-3*).
10. It is not his possession of knowledge, of irrefutable truth, that makes the man of science, but his persistent and recklessly critical quest for truth (*Karl Popper*).
11. Het spelen van spelletjes kun je niet serieus genoeg nemen.

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