

## Curriculum vitae Ann Swillen



Ann Swillen is professor at the Department of Human Genetics, KU Leuven and at the Department of Rehabilitation Sciences, KU Leuven (University of Leuven, Belgium). Trained as an educational psychologist, she is also affiliated to CME-UZ (the clinical unit of the Department of Human genetics), an international centre of excellence in the field of clinical and molecular genetics. A particular focus is on individuals with genetic, neuropsychiatric, and neurodevelopmental conditions that affect learning and behavior.

Through a multifaceted collaborative approach with many disciplines, we aim for four goals:

1. Better define the studied neuro-genetic syndromes (e.g. 22q11.2 DS, 22q11 dupl., 22q13.3 DS, other CNV's ...);
2. Identify mechanisms of cognitive impairment;
3. Identify mechanisms for increased psychiatric risk;
4. Using specific neuro-genetic conditions as homogeneous genetic models to better understand the interaction among genetic, behavioral and environmental factors in developmental disorders.

Ultimately, a better knowledge of neuropsychiatric and neurodevelopmental conditions will help us to refine our treatment strategies and improve the life of affected children and their families.

Besides her research, Prof. Ann Swillen is (since 1994) the coordinator of the multidisciplinary 22q11 DS clinic, and is actively involved in sharing knowledge on the implications of genetic syndromes on cognition and behaviour and the practical implications for education/teaching etc... by giving multiple lectures to professionals on national and international meetings, and to national and international parent-patient associations. She is member of the scientific board of different parent organizations.

Prof. Ann Swillen is an international recognized authority in the field of development, cognition and behaviour in 22q11 DS and other CNV's, and she is author of more than 85 peer-reviewed scientific publications in the field of medical genetics and behavioral phenotypes. She is member of different international expert panels (e.g. international 22q11 DS Foundation, European 22q11 Alliance,...), and co-founder of the International Brain-Behavior 22q11 DS Consortium (2011) and the 22q11 DS Society (2014). Since 2017, she is expert member for Belgium on the COST action "Maximising Impact of research of NeuroDevelopmental DisorderS" (MINDDS), a newly funded European COST Action that was proposed by researchers in Cardiff and KU Leuven. The main objective is to create a collaborative network to enhance identification of patients carrying genetically penetrant pathogenic Copy Number Variants (CNV) which are rare cases and this Action presents a unique opportunity to understand neurodevelopmental disorders (NDD), standardise research methodologies and facilitate exchange of information for the benefit of clinicians, researchers and patients.

Finally, Prof. Ann Swillen is PI (Europe A) of the NIH Grant U01 1U01MH101722-01 on "Genomic Risk and Resilience in 22q11 Deletion Syndrome: A Window into the Genetic Architecture of Mental Disorders", the IBBC 22q11 consortium, a collaboration between caregivers and scientists from 22 institutions and five genotyping sites throughout the world.

She received the Angelo DiGeorge Medal (Award) during the The 10th Biennial International 22q11.2 Deletion Syndrome Meeting in Sirmione, Italy (July 2016), for her outstanding and exceptional contribution to the understanding and treatment of the 22q11.2 deletion syndrome and for her commitment to patients.

Prof. Swillen is holder of the recent inaugurated 22q11 DS/Vecarfa fund at the KU Leuven.