Matthias De Wachter

Projects titles:

1)Phenotype-genotype correlation in RORA-related neurodevelopmental disorders'

2)4-AP in GOF or mixed function KCNA2 encephalopathy

3)Precision medicine in epilepsy: opinions from clinicians

Duration	4 months
Short Bio	*I was trained as Pediatrician at Antwerp University Hospital and gained interest in genetic epilepsies during my rotation at the department of Pediatric Neurology, Genetics and in the rehabilitation and epilepsy centre of Pulderbos.* *After that, I started a clinical fellowship in Pediatric Neurology together with a PhD in epilepsy. My clinical work focused on complex epilepsies both in neonates and older children. My research focuses on the deep phenotyping of rare genetic epilepsies and their treatment.*
Home Institution	Antwerp University Hospital
Host institution	Filadelfia Epilepsy Centre
Project description	*During my fellowship, I focused on the epileptic phenotype of individuals with heterozygous variants in RORA. Both LoF and GoF variants are described causing complex phenotypes consisting of ID, ASD, movement disorders and seizures. Because a clear description of the epilepsy phenotype is missing, in this study we aimed to define the epileptic spectrum of individuals with RORA variants.* *As we did this project in collaboration with Gaetan Lesca from Lyon University, there was also room for the projects mentioned above, which focused on the multicentre approach to rare genetic epilepsies and their treatment.*
Personal statement	*As a national reference center of epilepsy in Denmark, it is the only center in Denmark that offers high-specialized care for patients with epilepsy. Because of its unique position, it has patients from all over the country, and with different rare genetic epilepsies. I was part of a high-experienced team, both in clinical practice as well as in research. The unique aspects of*

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	having different specialties in own department, even in one hallway, made it particularly interesting. Specialized nurses, a pharmacist, genetic technician, geneticists and (child) neurologists are working closely together. This experience not only increased my knowledge in genetic epilepsies, but also increased my international network with members of different ERN's, and resulted in collaborations with Germany, France, Denmark and the USA. *Being back in Antwerp, these collaborations are still ongoing, and I often can bring other collaborators in contact with the people I worked with, to stimulate collaboration or to answer specific clinical questions.*