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BSc Biology, Carl von Ossietzky University of Oldenburg, Germany 2010
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Current Position

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Keywords

Early-onset dementia - Frontotemporal dementia - Disease causing genes - Advanced genetic profiling

Science

Patients suffering from genetic disorders are in need of a correct diagnosis so that adequate treatment decisions can be made, which is often prevented due to missing knowledge of the genetic disease etiology. This is also the case for many inherited neurodegenerative brain disorders, like dementia. A better understanding of the genetic causes will provide deepened insight into the genesis of dementia, needed for better treatments.

The NBD group has contributed significantly to the identification and characterization of genes involved in neurodegenerative brain diseases. Nevertheless 80% of Dementia patients are still unexplained. To unravel the missing heritability in dementia this PhD research comprises the identification of novel key genes and proteins to enhance our knowledge of the molecular signatures of neurodegenerative dementia. In this context we focus on dementia groups showing first clinical presentation before the age of 65, referred to as early-onset dementia (EOD). Because of the early disease presentation, EOD patients have an extreme phenotype and as such are expected to have a strong genetic heritage.

One of the EOD phenotypes that we focus on is Frontotemporal dementia (FTD). FTD is one of the most heterogeneous brain diseases. Therefore, continued genetic research is vital to fully comprehend the genetic and molecular complexity of this disease. We apply advanced genetic profiling strategies for novel gene discovery, including whole genome, whole exome and gene panel sequencing on FTD patients. A major strength of our research is that it can built on an impressive collection of >4000 patients with different EOD phenotypes (mainly Alzheimer's disease and FTD) ascertained within the *European Early-Onset Dementia consortium*.

Finally, identification of new disease genes and molecular signatures will be instrumental in pinpointing diagnostic biomarkers and drug targets for therapy development and will allow more accurate stratification of patient cohorts for follow-up translational research and clinical trials. This will ultimately offer better perspectives for patients and families affected by dementia.

Research project and fellowship

University of Antwerp – University Research Grant (BOF-doctoral project)
Period: 1.11.2015 – 30.09.2019
Title: 'Next-generation genetics of early-onset dementia to increase our appreciation of the molecular signatures of dementia.' Role: PhD student