Sara Van Mossevelde

Neurodegenerative Brain Diseases Group
Department of Molecular Genetics, VIB
Laboratory of Neurogenetics, Institute Born-Bunge
University of Antwerp

Education

BSc Medicine, University of Ghent, 2008
MSc Medicine, University of Ghent, 2012

Current Position

PhD Student at the University of Antwerp
VIB Department of Molecular Genetics

Email: sara.vanmossevelde@molgen.vib-ua.be
Phone: +32 3 265 1038

Keywords

Early-onset dementia - translational research – genotype-phenotype correlation studies - frontotemporal lobar degeneration (FTLD) – amyotrophic lateral sclerosis (ALS)

Science

Frontotemporal lobar degeneration (FTLD) is a term covering a heterogeneous group of neurodegenerative disorders with predominant atrophy of the frontal and/or the temporal lobes of the brain. It is the third most common form of dementia after Alzheimer's disease and vascular dementia, and accounts for 5-10% of all dementia patients.
FTLD is remarkably heterogeneous in clinical presentation, neuropathological characteristics and underlying genetic causes. Clinically, frontotemporal dementia (FTD) is characterized by a progressive deterioration in behaviour, personality and/or language with relative preservation of memory function.
About 15% of FTD patients also develop amyotrophic lateral sclerosis (ALS), a motor neuron disease clinically characterized by progressive muscle weakness, muscular atrophy, fasciculations and spasticity. FTLD and ALS are suggested to be two extremes of a continuum of neurodegenerative disorders.
Genetically, *C9orf72* repeat expansions are the most common cause of FTLD and ALS, explaining 25% of familial FTLD, 37% of familial ALS and up to 88% of familial FTLD-ALS patients. Recently the TANK-binding kinase 1 (TBK1) gene was identified as a second genetic cause of both FTLD and ALS.

The main objective of this PhD project comprises the in-depth description of phenotypical characteristics of C9orf72 repeat expansion carriers in various fields (clinical, biochemical, pathological and imaging) in order to identify specific biomarkers correlating with current and expected future disease status, as well as their temporal evolution. Second, we want to identify factors that are able to modify the phenotype of C9orf72 repeat expansion carriers.

Research projects and fellowships

University of Antwerp – Doctoral Fellowship
Period: 1.05.2014 – 30.09.2017

Title: 'In-depth research of C9orf72 repeat expansion carriers and families to identify core clinical phenotypes, biomarkers and disease modifiers.'
Role: PhD student

Selected Publications

Gijselinck I, **Van Mossevelde S**, van der Zee J, Sieben A, Philtjens S, Heeman B, *et al*. Loss of TBK1 is a frequent cause of frontotemporal dementia in a Belgian cohort. Neurology 2015; in press.

Gijselinck I, **Van Mossevelde S**, van der Zee J, Sieben A, Engelborghs S, De Bleecker J, *et al*. The C9orf72 repeat size correlates with onset age of disease, DNA methylation and transcriptional downregulation of the promoter. Mol Psychiatry 2015; in press.