Stefanie Smolders

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

Education

BSc Biochemistry and Biotechnology, University of Antwerp, 2011
MSc Biochemistry and Biotechnology, University of Antwerp, 2013

Current Position

PhD Student at University of Antwerp
VIB Department of Molecular Genetics

Email: stefanie.smolders@molgen.vib-ua.be
Phone: +32 3 265 1029

Keywords

Massive parallel sequencing (MPS) – Functional genomics – Parkinson Disease (PD) – Gene identification – Molecular mechanisms

Science

Positional cloning strategies and massive parallel sequencing (MPS) in families with a monogenic form of Parkinson Disease (PD) have established a total of 7 genetically validated genes and 8 putative causal genes. Intense molecular research indicated, when connecting the different gene functions, that neuronal demise in PD originates from the interconnection of different processes, i.e. synaptic transmission, endosomal protein sorting and recycling, mitochondrial quality control, stress response, ubiquitin-proteasome proteolysis and lysosome mediated autophagy. However, less than 10 % of familial PD is explained by protein altering mutations in known PD and related Parkinsonism genes, suggesting that over 90 % of the genetic etiology of familial PD is still unclear. Identification of novel causal genes will therefore be instrumental in further unravelling the molecular basis and disease processes underlying both familial and sporadic PD.

With my project I aim to generate a more detailed mechanistic view of PD by studying the disease from different angles simultaneously. We aim to (1) identify exonic or regulatory variants with a high probability to contribute to PD etiology using whole genome sequencing (WGS). In addition, we will simultaneously (2) explore the potential of RNASeq on lymphoblasts to identify gene networks and pathways affected by the disease-related genetic defects. We aim (3) to speed up the identification of the culprit genetic defects and their biological impact by integrating information on affected pathways and gene networks with detailed information on the underlying genomic variation. Once we identify novel candidate genes, we will (4) explore the mutation spectrum in PD and clinically overlapping brain disorders to estimate its contribution to neurodegeneration. Finally, the integrative approach will (5) guide further deciphering of the mutation mechanisms in cellular models, and will give us, combined with clinical information, the opportunity to define relevant genotype-phenotype correlations.

Recent Fellowships

University of Antwerp – Academic Assistant Molecular Genetics
Period: 01.10.2015 – 30.09.2017
Title: 'Enquiry of the molecular basis and mechanisms of Parkinson disease'
Supervisor: Prof. Dr. Christine Van Broeckhoven
Role: PhD Fellow

University of Antwerp – Doctoral Fellowship
Period: 01.01.2014 – 30.09.2015
Title: 'Enquiry of the molecular basis and mechanisms of Parkinson disease'
Supervisor: Prof. Dr. Christine Van Broeckhoven
Role: PhD Fellow

All Fellowships

Selected Publications

Theuns,J., Verstraeten,A., Sleegers,K., Wauters,E., Gijselinck,I., **Smolders,S.**, Crosiers,D., Corsmit,E., Elinck,E., Sharma,M., Krüger,R., Lesage,S., Brice,A., Chung,S.J., Kim,M-J., Kim,Y.J., Ross,O.A., Wszolek,Z., Rogaeva,E., Xi,Z., Lang,A.E., Klein,C., Weissbach,A., Mellick,G., Silburn,P.A., Hadjigeorgiou,G., Dardiotis,E., Hattori,N., Ogaki,K., Tan,E., Zhao,Y., Aasly,J., Valente,E.M., Petrucci,S., Annesi,G., Quattrone,A., Ferrarese,C., Brighina,L., Deutschländer,A., Puschmann,A., Nillson,C., Garraux,G., LeDoux,M.S., Pfeiffer,R.F., Boczarska-Jedynak,M., Opala,G., Maraganore,D.M., Engelborghs,S., De Deyn,P.P., Cras,P., Cruts,M., Van Broeckhoven,C., on behalf of the GEO-PD Consortium,: Global investigation and meta-analysis of the C9orf72 (G4C2)n repeat in Parkinson disease. Neurology. Epub: 2014 (I.F.: 8.303) (PMID: 25326098)

All Publications