Caroline Robberecht PhD

Neurodegenerative Brain Diseases Group
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Education

2004 BSc Biomedical Sciences, KU Leuven
2006 MSc Biomedical Sciences, KU Leuven
2012 PhD Biomedical Sciences, KU Leuven

Position

Research Associate

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Keywords

Neurodegenerative brain diseases - biobanking - translational genetics – ethics in genetics – genetic epidemiology

Science

My research interest is focused on the molecular genetics of neurodegenerative brain diseases (NBD) including Alzheimer disease (AD), frontotemporal lobar degeneration (FTLD) and Parkinson disease (PD).

With my work, I aim to contribute to the discovery and characterization of genetic variations i.e. mutations in causal and risk genes as well as their frequencies in different populations and in NBD cohorts. For this purpose, a high-throughput multiplex assay for genetic profiling, based on next-generation sequencing technology, was designed and is being validated. The genetic profiling assay will contribute to our understanding of the genomic heterogeneity in NBD patients and in control populations. The observed genetic variations in NBD genes and their associated phenotype will be captured in mutation databases.

Through our continued biobanking initiative, we invest in the systematic collection of a wide range of human biosamples from thoroughly characterized research participants. This will facilitate the translation of genetic breakthroughs into biologically and clinically relevant approaches for improved diagnostics, prevention and treatment.

Further, I have a special interest in new genomic developments and their potential ethical consequences with respect to the biological privacy of the participants in research projects. In our research group I will take the responsibility for safeguarding the research participant's legal rights in genetic research, biobanking

and data sharing by providing the necessary ethical framework in which the NBD research is being performed. This includes writing research protocols and completing questionnaires needed to obtain ethical assurance from the Ethical Committee at the University of Antwerp and the Antwerp University Hospital for each new research project. I will also maintain an overview of the on-going research projects and their compliance with the respective ethical assurances.

Selected Publications

Robberecht C, Voet T, Zamani Esteki M, Nowakowska BA, Vermeesch JR. Nonallelic homologous recombination between retrotransposable elements is a driver of *de novo* unbalanced translocations. Genome Research. Epub 2012 Dec 3. (PMID: 23212949)

Robberecht C, Pexsters A, Deprest J, Fryns JP, D'Hooghe T, Vermeesch JR. Cytogenetic and morphological analysis of early products of conception following hystero-embryoscopy from couples with recurrent pregnancy loss. Prenatal Diagnosis 2012 Jul; 32(10):933-42. (PMID: 22763612)

Robberecht C, Voet T, Utine GE, Schinzel A, de Leeuw N, Fryns JP, Vermeesch JR. Meiotic errors followed by two parallel postzygotic trisomy rescue events are a frequent cause of constitutional segmental mosaicism. Molecular Cytogenetics 2012 Apr;5:19. (PMID: 22490612)

All publications