Federica Perrone

Neurodegenerative Brain Diseases group
VIB-UAntwerp Center for Molecular Neurology
Laboratory of Neurogenetics, Institute Born-Bunge
University of Antwerp

BSc Biological Science, Sapienza - Univeristy of Rome, 2011

Current Position

PhD Student at University of Antwerp
VIB-UAntwerp Center for Molecular Neurology
E-mail: federica.perrone@molgen.vib-ua.be
Phone: +32 3 265 1953

Keywords

Early-onset dementia (EOD) – Frontotemporal dementia – Alzheimer's disease – Next generation sequencing (NGS) – Molecular genetics – gene discovery

Science

Basic and clinical scientists realized that they have long underestimated the complexity of dementia. With the world population ageing at an unprecedented scale and the absence of curative therapies or preventive strategies, the number of people living with dementia worldwide today is estimated at 47 million and set to almost more than triple by 2050. In the past 30 years, major progress in molecular genetic research allowed the identification of a significant number of monogenic autosomal dominant genes as well as susceptibility genes for dementia, leading to the recognition of key molecular processes underlying neurodegeneration. Notably most of these discoveries were realized in large families with an early disease onset. When dementia occurs before the age of 65 years, it is classified as early-onset dementia (EOD), with the leading causes being Alzheimer's disease (AD) and Frontotemporal dementia (FTD). However, despite these discoveries, 80% of all dementia cases remain still unexplained by mutations in known genes. The aim of this PhD project is to identify novel disease causing and disease-modifying/protective genetic profiles, by focusing on the subgroup of the EOD patients. Because these patients suffer from a disease of the aging brain at a relatively young age, they have an extreme presentation of the disease and as such can be expected to have a strong genetic heritage. Study of this subgroup of dementia patients will therefore more likely lead to identification of novel genes and molecular signatures of dementia. Furthermore, we expect that novel genes and gene products identified in this study will also be relevant for the more common late-onset forms.

Research Projects and fellowships

University of Antwerp - Doctoral fellowship
Period: 15.10.2014-14.09.2016
Title: Understanding the complexity of dementia through novel genes, genetic modifiers and protective profiles using next-generation genetics of early-onset dementia
Role: PhD fellow

Publications

Perrone F*, Nguyen HP*, Van Mossevelde S, [...], Van Broeckhoven C, van der Zee J. Investigating the role of ALS genes CHCHD10 and TUBA4A in Belgian FTD-ALS spectrum patients. Neurobiology of Aging. 2016 Dec 21 S0197-4580(16)30314-1. DOI: 10.1016/j.neurobiolaging.2016.12.008

van der Zee J*, Gijselinck I*, Van Mossevelde S, **Perrone F**, Dillen,[...], Van Broeckhoven C. TBK1 Mutation Spectrum in an Extended European Patient Cohort With Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Hum Mutat. 2016 Dec 23. DOI: 10.1002/humu.23161

van der Zee J*, Mariën P*, Crols R*, Van Mossevelde S, Dillen L, **Perrone F**, [...], Van Broeckhoven CMutated CTSF in adult-onset neuronal ceroid lipofuscinosis and FTD. Neurol Genet. 2016 Sep 16;2(5):e102. DOI: 10.1212/NXG.0000000000000102