Rita Cacace

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

BSc Biotechnology for Healthcare, University of Naples "Federico II", 2007
MSc Medical Biotechnology, University of Naples "Federico II", 2009
PhD Biomedical Sciences, University of Antwerp, 2015

Current Position

Post-doctoral researcher
VIB Department of Molecular Genetics
E-mail: rita.cacace@molgen.vib-ua.be
Phone: +32 3 265 1039

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Alzheimer disease (AD) – early onset dementia (EOD) – next generation sequencing – missing genetic etiology

Science

Dementia is the term used to classify a syndrome characterized by progressive deterioration of cognitive functions including memory, learning, language, comprehension and judgment and it is caused by a pathological affliction of the brain. Currently, an estimated 46.8 million people worldwide are living with dementia and up to 75% has a diagnosis of Alzheimer disease (AD). The estimate is indicative of a global epidemic, with AD in a leading position. AD is primarily characterized by loss of memory and deterioration of additional cognitive functions; it mainly affects older people (late-onset (LO) AD) but up to 10% of patients presents the clinical symptoms at younger age (<65 years, early-onset (EO) AD), even in their thirties. LOAD is a complex disease with an estimated heritability of 70-80%, EOAD is almost entirely genetically determined. In fact, families with inherited form of disease were pivotal for the identification of three causal AD genes: amyloid precursor protein (*APP*), presenilin 1 (*PSEN1*) and presenilin 2 (*PSEN2*) and one major risk factor for both EOAD and LOAD: apolipoprotein E (*APOE)*. These discoveries featured the early 1990s and served as basis to the current knowledge of the disease mechanism. Unfortunately, mutations in these genes explain only 5% of EOAD patients, thus the majority of EOAD patients remains genetically unexplained.

AD is still an incurable disease and the available treatments can only alleviate some symptoms. There is a pressing need to improve our understanding of AD pathogenesis to enable the development of disease-modifying therapies.

The aim of this PhD project is to explore the missing genetic etiology of AD in order to gain insights in novel genes and pathways able to contribute to a better understanding of AD pathogenesis. In order to achieve this goal, state-of-the-art genetic and genomics technologies are applied to patients with high genetic load, both families with inherited form

of disease and EOAD patients in general. This gene hunting approach aims to identify novel candidate gene(s) for neurodegenerative dementia and, additionally, to investigate the candidate gene(s) in follow-up studies. The final goal is to provide novel insights in the disease mechanism and to identify additional molecular targets for the development of more effective therapeutic strategies.

Recent Fellowships

UA-BOF
Period 01.10.2015 - 31.12.2015
Title: 'Identification of new genes for Alzheimer's disease using advanced molecular genetic technologies.'
Role: Fellow

All projects and fellowships

Selected publications

Cacace R.*, Van Cauwenberghe C.*, Bettens K., Gijselinck I., van der Zee J., Engelborghs S., Vandenbulcke M., Van Dongen J., Bäumer V., Dillen L., Mattheijssens M. Peeters K., Cruts M., Vandenberghe R., De Deyn P.P., Van Broeckhoven C., Sleegers K.: *C9orf72* G_4C_2 repeat expansions in Alzheimer's disease and mild cognitive impairment. Neurobiology of Aging 34(6): 1712 e1-e7 (2013) Epub: 2013 (I.F.: 6.189) (PMID: 23352322).
*Equally contributing first authors.

Cacace R., Van den Bossche T., Engelborghs S., Geerts N., Laureys A., Dillen L., Graff C., Thonberg H., Chiang H.-H., Pastor P., Ortega-Cubera S., Pastor M.A., Diehl-Schmid J., Alexopoulos P., Benussi L., Ghidoni R., Binetti G., Nacmias B., Sorbi S., Sanchez-Valle R., Llado A., Gelpi E., Almeida M.R., Santana I., Tsolaki M., Koutroumani M., Clarimon J., Lleo A., Fortea J., De Mendonca A., Martins M., Borroni B., Padovani A., Matej R., Rohan Z., Vandenbulcke M., Vandenberghe R., De Deyn P.P., Cras P., van der Zee J., Sleegers K., Van Broeckhoven C., BELNEU consortium, EU EOD consortium: Rare variants in *PLD3* do not affect risk for early-onset Alzheimer disease in a European consortium cohort. Human Mutation (Epub 2015) (I.F.: 5.144) (PMID: 26411346).

All publications