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The genetics of Alzheimer's disease in Asia

Alzheimer's disease (AD) being one of the neurodegenerative disorders, which was characterized by a gradual loss of memory, decline in other cognitive functions and decrease in functional capacity. Molecular analyses of families with early onset of the AD (EOAD) made possible to identify dominantly acting mutations in genes, such as amyloid precursor protein precursor protein (APP) and presenilin 1 and 2 (PSEN 1 and PSEN 2). However, the etiology of the late onset of the AD (LOAD) was less straightforward than EOAD. In most Asian countries, the population and the number of AD patients are growing rapidly, highlighting the needs to colligate mutations across populations. DNA extraction, linkage study, next-generation sequencing and Sanger sequencing, bioinformatics, neurological evaluation, diagnostic imaging, APOE gene polymorphism analysis and pathological assessment were performed in all patients. We reported an update (2009–2018) of genetic screening results from large-scale EOAD series ascertained across four Asian countries, Thailand, Malaysia, Philippines and Korea. Over 400 patients, familial and sporadic cases were screened, 22 cases carried a mutation in APP, PSEN1 and PSEN2, which had all three CSF biomarkers—total tau protein (Tau), phospho-tau protein (P-Tau) and amyloid β (Aβ)42 in abnormal ranges. Among the 22 distinct mutations found in the patients and isolated cases in 3 populations, definite pathogenicity established for only 6%, emphasizing the needs to survey variants in larger patient cohorts. Currently, we are exploring their causative functional effects by cloning them into a stable cell line. In addition, the segregation analyses in the family members, as well as targeting deep resequencing in large datasets for validating the role of the variants are in works.

Biography

Van Giau Vo is an Assistance Professor in the Department of Bio-nanotechnology, Gachon University and a Postdoctoral Research Assistant in Seoul National University College of Medicine in Seoul National Bundang Hospital. His background is in biotechnology and neuroimaging genomics and holds a PhD in Biomedical from Gachon University. Currently, his work investigates how neurogenetics-based biomarkers can be used to improve diagnostic accuracy in Alzheimer's disease and to the biology of neurodegenerative diseases, with the goal of accelerating treatment development. He has published over fifteen peer-reviewed scientific papers (excluding the conference ones) fields that cover neurogenetic study on early-onset dementia, molecular biology and cell-material interactions.

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