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Molecular research on Alzheimer's disease of Saudi patients and its applications

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Introduction & Aim: Alzheimer's disease (AD) is the most common form of dementia and neurodegeneration. Cerebral atrophy, beta amyloid aggregation and intra-neuronal neurofibrillary tangles are associated with AD. Although AD is largely sporadic occurring in the elderly, a minority of cases belongs to early onset form that appears before age of 65 and is genetically inherited. As little is known on the background of AD in Saudi population, we established research on the genetic basis of AD in Saudi patients and initiated in vitro cellular model derived from the patients that is applicable for drug discovery.

Method: To find out the genetic cause of Alzheimer's disease in Saudi patients, we recruited 100 AD belonging to familial and sporadic cases and screened them by direct sequencing for possible pathogenic mutations in AD related genes. 76 representative samples were examined for copy number of variants. Modeling of the disease was studied by direct conversion of human fibroblasts to neurons using our novel combination of chemical molecules to be applicable on fibroblasts issued from Saudi patients genetically inherited AD.

Results: We found 2 out of 24 novel variants to be potentially pathogenic mutations in exons 23 and 26 of SORL1 gene. We got out of 72 known variants, probably damaging mutations in the following genes: SORL1 exon11 (c1582; A528T); APOEe4 exon4.1 (c487; R163C) and APOEe4 exon4.2 (c526; R176C). Certain index cases displayed loss of heterozygosity on chromosomal regions that include genes causing Alzheimer's disease. We identified small-molecule cocktails that converted fibroblasts into neurons without exogenous genetic factors.

Conclusion: The outcome of this study is providing data bases for mutations of AD and favoring in the near future the Saudi patients to benefit from personalized treatments.

Biography

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