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Towards precision medicine in genetics and neuroprotection research on Alzheimer's disease in Saudi patients

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Alzheimer's disease (AD) is the most common cause of dementia that represents a major economic and social burden worldwide, affecting more than 47 million people and this number is estimated to double by 2030. As the background of AD in Saudi population is still not investigated, we established Alzheimer's research in genetics and neuroprotective fields permitting the orientation towards personalized medicine. Our study includes finding the genetic basis of AD in Saudi patients, modeling of the disease from fibroblasts of Saudi Alzheimer's patients and drug discovery that is applicable on the patient's model. We referred to 117 sporadic and familial Saudi patients for mutations screening in AD related genes. 76 representative samples were screened for dosage alterations in the same studied genes. Modeling of the disease was produced on human fibroblasts that converted directly to neurons using our novel combination of chemical molecules. We are investigating *in-vitro* if the synthetic analogue of curcumins displays neuroprotective activity against beta amyloid peptide-induced neurotoxicity. In our results, we found one reported missense variation in each of the causative genes of AD: *PSEN1*, *PSEN1* and *APP* and most importantly, we found one possibly damaging novel mutation in *APP* gene and 4 novel nucleotide variations in *SORL1* gene. In copy number variation (CNV) study, loss of heterozygosity was observed in certain regions contains either *SORL1* or *APP* genes. In AD modeling, we identified small-molecule cocktails that converted fibroblasts into neurons without exogenous genetic factors. In drug discovery for AD, we demonstrate the capacity of curcumin and its analogue to correct beta-amyloid neurotoxicity causing Alzheimer's disease. Our study reflects the genetic complexity of AD in Saudi patients. The presence of known and novel mutations of *SORL1* gene in Saudi Alzheimer's patients is in consistence with previous results suggesting the implication of *SORL1* gene in AD. The outcome of this study provides data bases for mutations of AD and favoring in the near future the Saudi patients to benefit from personalized treatments.

Biography

Fadia El Bitar is a Scientist at Department of Genetics at King Faisal Specialist Hospital and Research Center (KFSHRC). She established Alzheimer's research project at KFSHRC. She is studying the genetic basis of Alzheimer's disease (AD) in Saudi population, defining *in-vitro* Alzheimer's modeling issued from patients and working on drug discovery for AD.

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