

Deep learning for polygenic score analysis for Alzheimer's disease risk prediction in the Chinese population

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Abstract

Background: Alzheimer's disease (AD) is a leading cause of mortality in the elderly. Genetics studies have identified variants associated with AD. Moreover, Polygenic score analysis can infer the risk of developing AD solely on the basis of genotype information.

Method: Whole-genome sequencing was performed in 2 AD cohorts recruited from mainland China ($n = 2,340$) and Hong Kong ($n = 1,009$). Genotype and phenotype data were input into a neural network model that output a polygenic score. Regression analysis was subsequently conducted to examine the associations between the resultant polygenic scores and AD plasma biomarkers.

Result: The resultant polygenic scores were associated with AD pathogenesis and the levels of specific AD plasma biomarkers in the Chinese population.

Conclusion: The results suggest that deep-learning methods have utility for investigating the underlying mechanisms of AD and promote the possibility of developing a genotype-based strategy for AD risk prediction.