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PRELIMINARY ANALYSIS OF PTPRS AND PTPRD GENETIC MARKERS IN ALZHEIMER'S PATIENTS IN PALESTINIAN NURSING HOMES: PILOT STUDY

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Abstract: Alzheimer's disease (AD) is a progressive neurodegenerative disorder characterized by amyloid β (A β) plaques, tau tangles, synaptic dysfunction, and cognitive decline. Genetic susceptibility factors strongly influence disease onset and progression, but population specific patterns remain underexplored, particularly in Middle Eastern cohorts. This study investigates single nucleotide polymorphisms (SNPs) within the protein tyrosine phosphatase receptor sigma (PTPRS) and delta (PTPRD) genes, key regulators of neuronal plasticity, signaling, and tau and A β homeostasis, in Palestinian AD patients residing in nursing homes. These genes were selected due to emerging evidence linking PTPRS to tau aggregation, synaptic integrity, and amyloid precursor protein processing, and PTPRD to neurofibrillary tangle accumulation and AD risk. A cross sectional genetic association design was employed. AD severity and neuropsychiatric symptoms were assessed via a caregiver completed questionnaire adapted from the Alzheimer's Questionnaire (2014) for the Palestinian context, which enabled staging of dementia severity and evaluation of symptoms including delusions, hallucinations, agitation, anxiety, and depression. Buccal swabs were collected from participants, and DNA was extracted, PCR amplified, and sequenced using Illumina NGS. Bioinformatic analysis was performed on the Galaxy platform, and statistical associations were evaluated in SPSS using chi square tests. Two SNPs were examined: rs560380 (PTPRD) and rs10415488 (PTPRS). While direct tau pathology assessment was not conducted, the risk alleles, rs10415488 T and rs560380 A, were selected based on prior evidence linking them to tau related pathogenesis and neurofibrillary tangle accumulation. According to Poirier et al. (2024), PTPRS serves as a novel marker for early tau pathology and synaptic integrity in Alzheimer's disease. Results revealed enrichment of these risk alleles in AD cases, with modest protective effects of the C alleles. However, statistical significance was not reached, largely due to the small sample size. Despite methodological constraints, this first Palestinian focused neurogenetic analysis highlights population specific risk alleles and demonstrates the potential utility of these SNPs as diagnostic markers for early tau pathology and synaptic integrity. The findings provide groundwork for future studies integrating molecular and clinical data into personalized AD diagnostics and therapeutics.