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TREM2 homozygous mutation R47C in a patient with sporadic frontal variant AD

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Several nonsense, missense, small deletions, and splice site mutations in gene encoding triggering receptor expressed on myeloid cells 2 (TREM2) on chromosome 6p21.1 has been identified in several neurodegenerative disorders. R47H variant in TREM2 has been associated with increased risk for Alzheimer's disease (AD) in European populations but not in African Americans and Chinese subjects. This association has not been replicated in Indian population. Here for the first time we screened AD (n=160), frontotemporal dementia (FTD, n=75) and mild cognitive impairment (MCI, n=80) subjects coming to the memory and neurobehavioral clinic of SCTIMST, Trivandrum, for R47H variant in TREM2 through direct sequencing analysis. None of the samples carried the R47H variant. However, we report R47C homozygous variant in one sporadic patient presenting with frontal variant AD. The proband was female with age of onset of 55 years and MMSE score of 20/30, carrying APOE ε3ε3 genotype. The proband does not carry any mutations in tau or progranulin. The other genetic risk factors were H1H1 tau haplotype and 3'-UTR variant of progranulin, rs5848 showed CC genotype, and the promoter polymorphism of CD33, rs3865444 showed CC genotype. Earlier R47C was reported in one Caucasian AD patient and in a woman of South Asian ancestry suffering from behavioral variant FTD. MRI showed frontal and temporal lobe atrophy with hippocampal and parahippocampal involvement, consistent with the clinical diagnosis of AD. DNA from family members was not available, precluding segregation analysis.

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