Case Report

Novel presenilin 1 mutation associated with early-onset Alzheimer's disease in a Saudi patient

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ABSTRACT

We report a 60-year-old Saudi patient with the clinical diagnosis of Alzheimer's disease (AD) and a novel mutation in the presenilin gene. We investigated mutations in the presenilin-1 gene in Saudi patients with AD using polymerase chain reaction and direct DNA sequencing methods. We extracted genomic DNA from the whole blood of both patients and normal control individuals. We sequenced and compared amplicons with the sequences of the respective exons of normal individuals as well as data available in GenBank. We detected a homozygous mutation (g→c) in exon 12, resulting in the missense mutation (Arg377Thr), in the DNA of a 60-year-old patient. We located this mutation in the cytoplasmic loop near the transmembrane domain 7.

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