A novel mutation in *PSEN1* (p.Arg41Ser) in an Argentinian woman with early onset Parkinsonism

Autores

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Abstract

Mutations in presenilin-1 (PSEN1) account for the majority of cases of familial autosomal dominant early-onset Alzheimer's disease (AD) as well as in sporadic forms. Atypical presentations are reported including extrapyramidal signs. In the last years, a pleiotropic effect of some PSEN1 variants has been reported in Parkinson's disease (PD).

Palabras clave

Parkinsonism, Presenilin, Genetics, PSEN1