

Multidisciplinary approach in the diagnosis of frontotemporal lobar degenerations and tauopathies: new insights into pathogenetic mechanisms

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Multidisciplinary approach in the diagnosis of frontotemporal lobar degenerations and tauopathies: new insights into pathogenetic mechanisms

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Research Abstract

Genetic aspects of neurodegenerative diseases and a multidisciplinary approach are of increasing importance. In this study we will enroll patients fulfilling clinical diagnostic criteria of different forms of frontotemporal lobar degenerations (FTLD). Detailed neuropsychological

evaluations, cerebrospinal fluid analysis, MR imaging, and DNA isolation from peripheral blood will be performed for all subjects. Molecular genetic exploration will be oriented towards analyses of genes involved in FTLD pathogenesis including direct exon sequencing in selected cases. Postmortem neuropathological examination will allow us to confirm the clinical diagnosis and follow the expression of specific selected proteins related to neurodegeneration. More precise diagnosis and eventually findings of new pathogenic mutations, in clinically less pronounced neurodegeneration, will profoundly modify the accuracy of prognoses and improve genetic consulting for relatives of patients with a proven genetic mutation.

Further information available at:

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