

FAMILIAL ATYPICAL FRONTOTEMPORAL LOBAR DEGENERATION



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INTRODUCTION

Frontotemporal Lobar Degeneration (FTLD) shows heterogeneous clinical phenotypes. Mutations in the PGRN, MAPT and C9ORF72 genes are described in familial cases. We report two siblings (brother and sister) who presented with atypical clinical phenotypes.

CASE PRESENTATION

Case 1: 77 year-old, right-handed woman complaining a three year history of behavioral disinhibition, hyperorality, repetitive movements, stereotypic speech and delusional misidentification. Disturbances of gait with recurring falls, sphincter incontinence and occasional swallowing difficulties were also reported. Neurological examination showed a bilateral akinetic extrapyramidal syndrome with postural instability, abolition of vertical eyes movements and pyramidal signs. Neuropsychological evaluation revealed a frontal syndrome associated with an asymmetrical (> right) ideomotor apraxia. MRI scan showed frontotemporal cortical atrophy.

Case 2: 66 year-old, right-handed man who presented, in the last four years, a language disorder characterized by mispronunciation of words and slowed speech. Verbal disinhibition and aggressiveness, increased consumption of sweet foods, apathy, neglect of personal hygiene were also reported. Neurological examination showed bradykinesia, postural instability, dysarthria, hypophonia, limitation of vertical eyes movements. Neuropsychological evaluation revealed a non fluent progressive aphasia associated with a disexecutive syndrome. MRI scan showed cortical atrophy. FDG-PET revealed decreased metabolic activity in bilateral frontal and parietal areas and in the right temporal lobe.

It was reported that their father suffered from an unspecified form of dementia.

Genetical analysis, including C9ORF72, MAPT, PGRN, were negative in both siblings.

CONCLUSION

The present cases represent two atypical clinical phenotypes of FTLD: case I shows an association of behavioral variant Frontotemporal Dementia (bvFTD), Progressive Supranuclear Palsy (PSP) and Corticobasal Syndrome; case II shows an association of bvFTD, nonfluent variant Primary Progressive Aphasia and PSP. These overlapping syndromes are described in subjects with mutations in MAPT, PGRN or C9ORF72 genes. Likely the two patients described carry a still unknown genetic mutation.

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