

PROGRANULIN PLASMA LEVELS AS POTENTIAL BIOMARKER FOR THE IDENTIFICATION OF GRN DELETION CARRIERS. A CASE WITH ATYPICAL ONSET AS CLINICAL MILD COGNITIVE IMPAIRMENT CONVERTED TO ALZHEIMER'S DISEASE

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Progranulin gene (GRN) mutations are associated with different clinical phenotypes, including Frontotemporal Dementia (FTD), Corticobasal Degeneration and Alzheimer's disease (AD). The range of age at onset is very wide and patients presenting initial symptoms around eighty years have been described. Previous studies demonstrated that progranulin plasma levels determination may be a reliable method to identify GRN deletion carriers. To evaluate progranulin plasma levels in all patients followed at our Alzheimer's Centre whose plasma was available (n=176). Progranulin levels were evaluated by ELISA in 75 patients clinically diagnosed with AD, 45 with FTD and 56 with Mild Cognitive Impairment (MCI) or subjective memory complaints. Four patients displayed low values. GRN Sequencing confirmed that three of them carried the CACT deletion in exon 7, and their clinical diagnosis was FTD. We also identified a patient carrying a previously reported CAGT deletion in exon 5 clinically diagnosed with AD. The onset of symptoms was at 77 years and the initial diagnosis was of amnesic MCI, which converted to AD six months later. In the following years, the patient also developed behavioral disturbances, gait apraxia and parkinsonian symptoms. At present, she is 84 years old. Progranulin plasma level evaluation is a reliable biomarker to identify GRN deletion carriers and discriminate between FTLD and other dementias which may mimic it. We thus encourage the inclusion of this non-invasive and easy test in clinical practice.