A case of progressive non-fluent aphasia with genetic SOD1 mutation (DO90A) linked to amyotrophic lateral sclerosis

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Background: The association between Amyotrophic Lateral Sclerosis and behavioral form of Frontotemporal Dementia is well known. Conversely, the association between ALS and FTD language variants, Progressive Non-Fluent Aphasia (PNFA) or pure Semantic Dementia or is extremely rare. Case report: A case study of a 68 year old woman is described with a two-year history of progressive primary non-fluent aphasia accompanied by agrammatism, phonemic paraphasias and mild naming disturbances; there has been no evidence of impairment of word or sentence comprehension. Non-verbal skills, memory and executive functions have also been preserved within the range of her very high premorbid level of abilities. Fluorodeoxyglucose positron emission tomography (FDG-PET) was consistent with bitemporal and left-frontal hypoperfusion. In a research FTD-ALS study a mutation in SOD1 (p.Asp90Ala; D90A) was discovered. There was no family history of ALS or FTD; his son also is an asymptomatic carrier of the mutation. Up to the present time, she has not developed motor neuron symptoms. Conclusions: The association between amyotrophic lateral sclerosis (ALS) and language variants of FTD is uncommon. D90A SOD1 mutation is related to a heterogeneous ALS clinical phenotype. In our knowledge, this is the first case of PNFA with a genetic mutation linked to ALS. Further study of the nature of the language changes found in ALS-FTD will be important in order to attain a greater understanding of the neuropsychological profile of this condition. KEYWORDS: Amyotrophic Lateral Sclerosis; Frontotemporal Dementia; Progressive Non-Fluent Aphasia