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A CASE OF PROGRESSIVE NONFLUENT APHASIA WITH GENETIC SOD1 MUTATION (DO90A) LINKED TO AMYOTROPHIC LATERAL SCLEROSIS

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Introduction

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 NonFluent Aphasia (PNFA) or pure Semantic Dementia or is extremely rare.

Case Report

A right-handed, highly educated, 68-year-old woman was assessed for language problems in the Cognitive Unit of the Deparment of Neurology of Hospital Universitario de Torrejon. Familial history was negative for neurodegenerative and psychiatric diseases.

She describes difficulties in spontaneous speech for 2 years, characterized by poor production with articulatory and phonemic errors.

The first neuropsychological evaluation showed a non-fluent language accompanied by agrammatism, phonemic paraphasias and mild naming disturbances; there has been no evidence of impairment of word or sentence comprehension.

Nonverbal skills, memory and executive functions have also been preserved at the beginning within the range of her very high premorbid level of abilities.

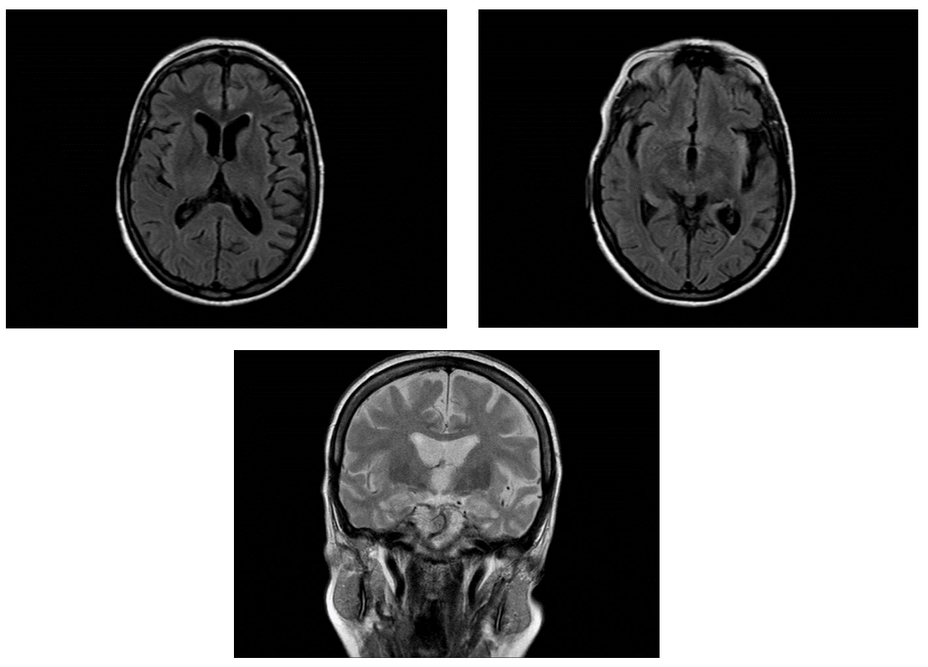
Thereafter, her language progressively worsened and she began memory, attention and orientation problems.

	Initial	2 years later
Attention		
Digit span F/B	5/3	4/2
TMT A	85 sg	115 sg
Executive functions		
TMT B	127 sg	>400 sg
Language		
Phonological fluency (initial letter P)	6	5
Semantic fluency (animals)	7	3
Boston naming test (BNT-15)	5	0
Memory		
MMSE	26/30	13/30
Frontal lobe dysfunction		
Frontal Assesment Battery	16/18	9/18

Table 1. Neuropsychometric test results.

Case Report

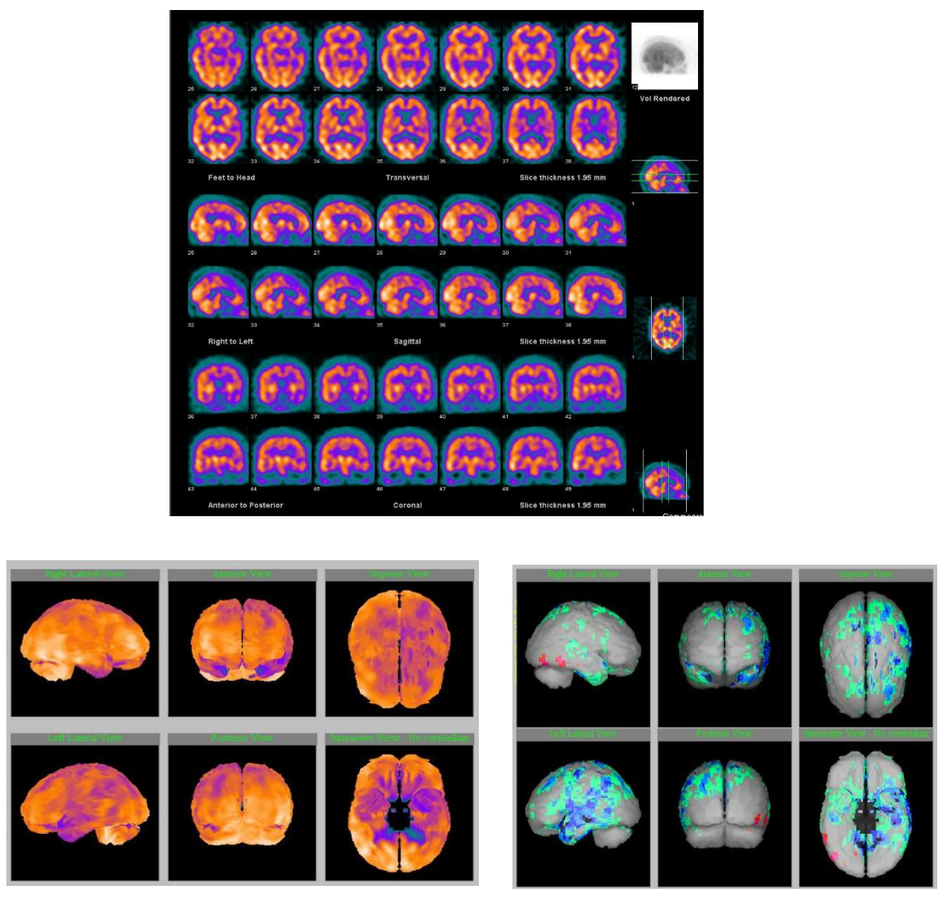
Brain magnetic resonance (MRI) showed a mild atrophy of left frontal and temporal lobes



MRI (axial Flair, coronal T2): mild atrophy of left frontal and temporal lobes

The first 18F-fluoro-deoxy-glucose positron emission tomography (FDG-PET) performed at the beginning of the symptoms did not show alterations.

One year later, the second FDG-PET showed bitemporal and leftfrontal hypometabolism.



FDG-PET. Bitemporal and leftfrontal hypometabolism.

In a research FTDALS 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 ALSFTD will be important in order to attain a greater understanding of the neuropsychological profile of this condition.

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