UNUSUAL PHENOTYPIC PATTERNS IN FAMILIAR FRONTOTEMPORAL DEMENTIA

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BACKGROUND

Frontotemporal lobar degeneration (FTLD) shows heterogeneous clinical phenotypes. Approximately 40% of FTLD patients have a history that is suggestive of familial transmission and the most common genes associated are GRN, MAPT and C9ORF72. We describe an Italian family showing different clinical phenotypes of the FTD spectrum without a known genetic mutation.

CASE REPORT

The family was native of Ancona, Italy. Three siblings, two sisters (S1 e S2) and one brother (B1) came to our attention and are described in details. History of dementia characterized by prominent behaviour problems was reported in other seven members of the family (Fig.1).

S1 is a 64 year-old, right-handed lady with an atypical semantic dementia. She presented with a two year history of progressive language disturbance, mainly characterized by world finding problems, semantic and phonological paraphasias, associated with behavioural alterations such as impulsiveness and verbal aggressiveness. The *neuropsycological evaluation* revealed an atypical language disturbance characterized by a prevalent semantic impairment together with phonological and syntactical errors. Semantic breakdown was present also in the non-verbal domain with associated agnosia and prosopoagnosia. *MRI* showed predominantly left temporal atrophy; *PET* revealed decreased metabolic activity in left frontal and temporal areas.

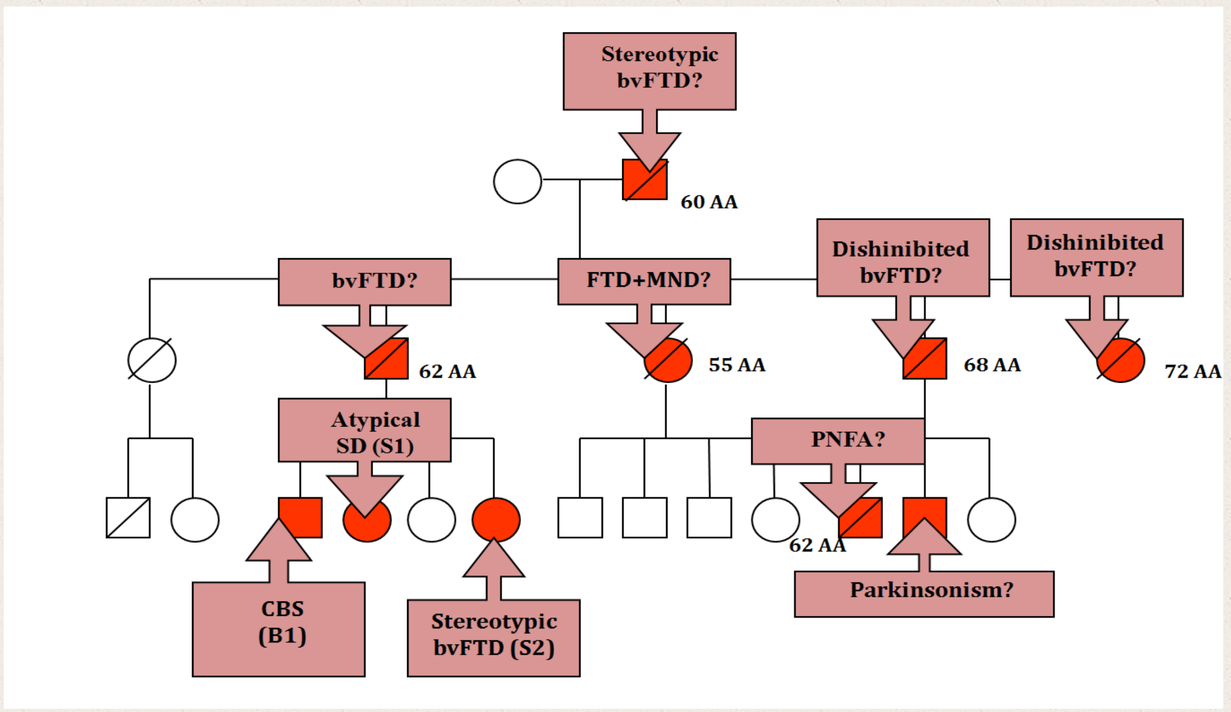

Fig.1. Family tree of the three siblings S1, S2, B1.

B1 is a 61 year-old, right-handed man with Corticobasal syndrome. He referred a two year history of progressive motor impairment in his left arm associated with mild behavioural alterations such as verbal dishinibition and irritability. Neurological examination showed an extrapyramidal syndrome. *Neuropsycological evaluation* revealed ideomotor apraxia and astereognosia in his left upper limb. *MRI* scan showed right fronto-parietal cortical atrophy (Fig. 2A-B)**; *FDG-PET* revealed decreased metabolic activity in the same regions** (Fig. 2C)**.**

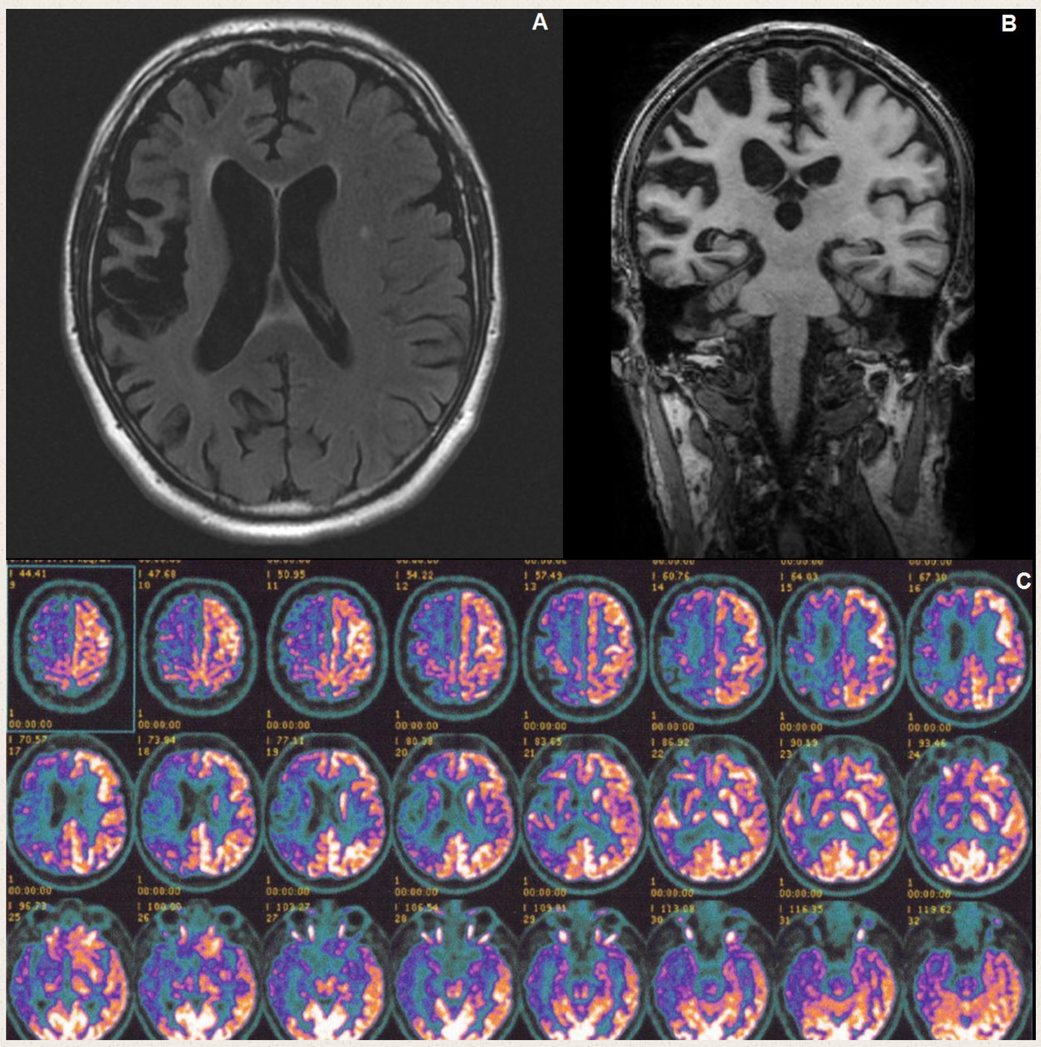

Fig.2. Patient B1: MRI scan showed right fronto-parietal cortical atrophy (A, B) and PET revealed decreased metabolic activity in the same areas (C).

S2 is a 65 year-old, right-handed lady with bvFTD. It was referred a two year history of attention disturbance and behavioural problems including verbal aggressiveness, impulsiveness, binge eating, compulsive attitudes together with psychosis (delusions and visual hallucinations). *Neuropsycological evaluation* showed sever frontal behavioural syndrome. *CT* scan showed predominantly right fronto-temporal atrophy (Fig.3).

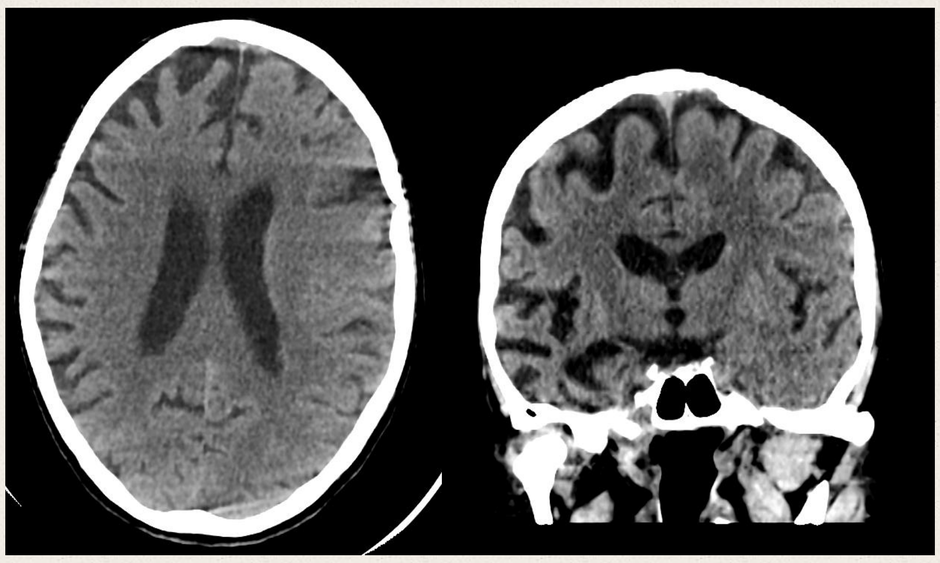

Fig. 3. Patient S2: CT scan showed right fronto-parietal cortical atrophy

***Genetical analyses* of the known mutations of C9ORF72, MAPT and GRN were negative, but low plasma progranulin levels were detected in the siblings.**

CONCLUSION

Likely the family described carries still unknown genetic mutation and permit to infer that FTD Italian families should carry different mutations from the ones described in other countries.

XLVII CONGRESSO NAZIONALE
22-25 OTTOBRE 2016 – VENEZIA