Introduction

C9orf72-associated frontotemporal dementia usually presents with the behavioral variant but cases of primary progressive aphasia (PPA) have also been described. However, the phenotype of this is poorly understood.

Materials and Methods

We undertook a longitudinal single case study of a patient with a C9orf72 expansion who presented with progressive speech and language problems consistent with a diagnosis of PPA. We performed a range of cognitive and neurolinguistic assessments at two time-points. Neuroimaging with volumetric T1-weighted imaging on a 3T Siemens Trio MR scanner was also acquired.

Results

A 54-year-old gentleman presented with a two year history of progressive speech problems and a one year history of behavioural change. His first symptom was increasing effortfulness of speech production. He had also developed a decrease in motivation, emotional lability and increased appetite. In the family history it was noted that his mother had had dementia in her 60s. Genetic testing revealed an abnormal expansion in the C9orf72 gene.

Detailed neuropsychological and linguistic assessments showed an apraxia of speech, impaired repetition and an orofacial apraxia but with intact naming, single word and sentence comprehension, and normal performance in other cognitive domains apart from impaired executive function, social cognition and processing speed. At his follow-up visit his speech production had deteriorated and he now had impaired performance on tasks of single word and sentence comprehension but with intact naming.

MRI showed relatively generalised symmetrical cortical atrophy with a frontal predominance and involvement of the caudate and hippocampus bilaterally. Atrophy had spread more posteriorly at the follow-up visit.

Conclusion

C9orf72-associated PPA in this gentleman was associated with an initial AOS with early behavioural changes, executive dysfunction and impaired social cognition. C9orf72 is a rare cause of PPA but should be tested for in those particularly with ALS and early behavioural change.