Left hand dystonia as a recurring feature of a family carrying C9ORF72 mutation.

Girelli F.[1], Fiori C. [1], Ranaldi V. [1], Baldinelli S. [1], Cameriere V. [1], Silvestrini M. [1], Provinciali L. [1], Rollinson S. [2], Pickering-Brown S. [2], Mann D., Snowden J. [2], Luzzi S. [1]

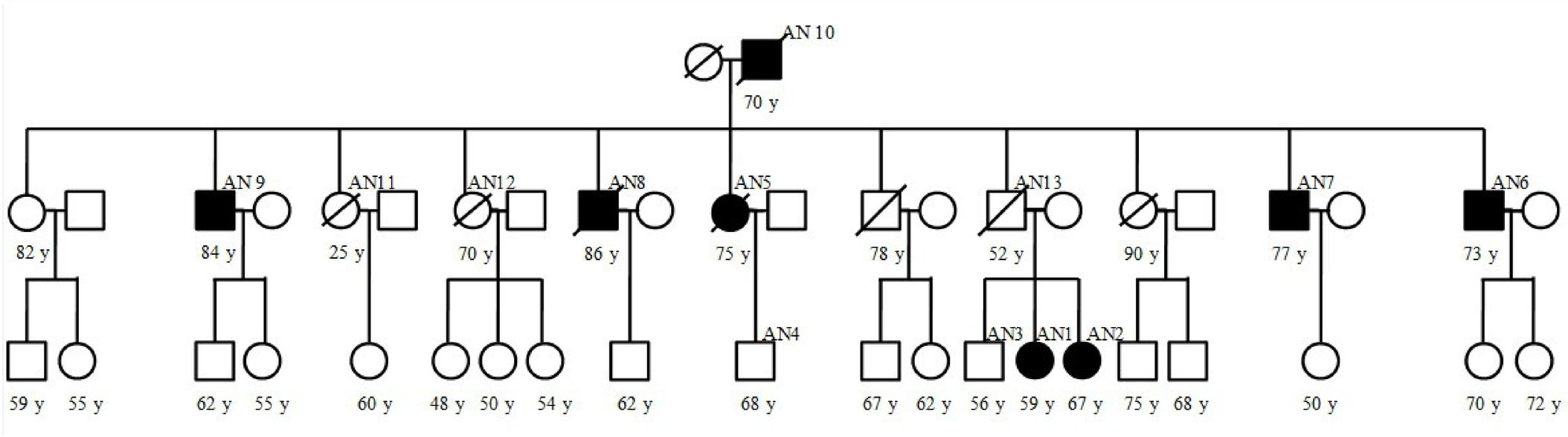
[1]Marche Polytechnic University – Department of Experimental and Clinical Medicine
[2] Institute of Brain, Behaviour and Mental Health, University of Manchester, Manchester, UK

Objectives

The clinical phenotype associated with mutations in the C9ORF72 gene is known to be variable. We report an unusual and previously unreported association between C9ORF72 and the presence of left-hand dystonia.

Methods

We describe the presenting symptoms and progression of members of a native Italian family carrying the C9ORF72 mutation.



Results

In three family members behavioural problems developed consistent with frontotemporal dementia followed, a few years after disease onset, by a left hand dystonia. In two of these family members neurological and neuropsychological evaluations excluded more widespread signs of corticobasal syndrome such as apraxia and extrapyramidal signs.

Conclusions

The present cases add further evidence of the puzzling and challenging phenotypic variability associated with frontotemporal dementia.

References

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