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

¹Marche Polytechnic University – Department of Experimental and Clinical Medicine
²Institute 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**