

## Abstract 82

### COGNITIVE AND LANGUAGE PROFILE OF FEMALES WITH FRAGILE-X PREMUTATION

Célia Maria Giacheti, Natalia Freitas Rossi, Débora Aparecida Rodrigueiro,  
Gimol Benzaquen Perosa, Danilo Moretti-Ferreira.

São Paulo State University – Unesp, Botucatu, SP, BRAZIL

Associations of FMR1 premutation with phenotype are controversial. However, premutation carriers can exhibit a mild form of the physical Fragile X phenotype syndrome. Premature ovarian failure and tremor/ataxia syndrome (FXTAS) have also been described. Learning disabilities and mental retardation have been reported in a few females with premutation. This study assessed cognitive, speech, language and academic performance in 7 female premutation carriers. Chronological age was 19-70 years and all females belonged to the same genealogy. Premutation was confirmed by Southern Blotting. Only case had no positive history of academic difficulties and could read and write. Cognitive performance was assessed by Wechsler Scale and receptive language by the Token Test. Peabody Vocabulary Test (PVT) assessed receptive vocabulary. The Consortium to Establish Registry to Alzheimer's Disease (CERAD) to evaluate memory and expressive language praxis, and a general test with the use of the Mini-exam of the Mental State were used. Academic skills were assessed by the School Performance Test (SPT). Cognitive performance was in the average range in all cases but one that was within the inferior average. Results: Mental and chronological ages were similar. Articulatory imprecision and weak vocal intensity associated with speech intelligibility impairment in 5/7 women. A case presenting FXTAS symptoms also showed severe intelligibility deficit with slow speech rate. One woman presented appropriate performance on PVT, Token Test, CERAD and SPT, whereas the other 5/7 showed significant difficulties: receptive semantic vocabulary impairment (PVT); moderate-to-severe difficulty in the comprehension of syntactic forms (Token Test); and impairments in cognitive, language and visuospatial tasks (CERAD). This study supports our clinical impressions that females with the fragile X premutation present language and academic impairments, particularly as compared to cognitive aspects. Further studies with premutation carriers are necessary considering that cognitive and language deficits reflect an emerging neuropsychological, linguistic phenotype of FMR1 premutation.

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