

## Abstract 33

### TOTAL CLINICAL ASCERTAINMENT OF DAUGHTERS WHO HAVE FATHERS WITH FRAGILE X ASSOCIATED TREMOR ATAXIA SYNDROME

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There is an increased risk for neurological, medical, endocrinological and psychological involvement in female individuals with the *FMR1* premutation. They often have concerns about these issues in particular neurological problems, as they get older and/ or if their fathers experienced Fragile X-associated Tremor/ Ataxia Syndrome (FXTAS). We therefore determined the prevalence and age of onset (O) of neurological problems, medical conditions, menstrual dysfunction and psychological issues in daughters of men with FXTAS to help address their concerns. We conducted phone or in person interviews with 136 females known to be carriers of the fragile X premutation and who had fathers with definite or probable FXTAS. Mean ( $\pm$  SD) age of the subjects was 44.90 ( $\pm$  9.65) years (age range 15-80 years). We found history of tremor in 15.4%, (O 41.63  $\pm$  15.90 years) and difficulty walking in 21.5%, (O 44.26  $\pm$  15.26 years). However, only 6 of 136 (4.4%) daughters had been diagnosed with FXTAS at the time of the study (O 45.92  $\pm$  7.56 years). Migraine headaches (36.9%, O 29.28  $\pm$  11.68 years), thyroid problems (20%, O 32.77  $\pm$  10.31 years) and hypertension (16.2%, O 44.13  $\pm$  10.05 years) were three most common medical conditions. Anxiety and depression was seen in 66.7% and 52.2% of the subjects. Of these individuals, 46.2% were in counseling and 49.6% were taking psychiatric medications. The spectrum of clinical involvement is quite broad and appears to be similar to other females with the premutation reported in previous studies. However, medical and psychological surveillance should be administered to provide appropriate management and holistic care for these premutation carriers. Additionally, background gene effects and biological markers that predispose individuals with the premutation to FXTAS need to be developed. The relationship of early psychological symptoms and the eventual development of FXTAS also requires further investigation.