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Hydrocephalus and tremor in a man with tetrasomy 48, XXXY

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Introduction: Sex chromosomal aneuploidy is the most common disorder of sex chromosomes in humans (1:400) while chromosome tetrasomies, like addition of more than one extra X and/or Y chromosome to a normal male karyotype, are very rare conditions causing syndromes characterized to varying degrees of tall stature, hypergonadotropic hypogonadism, developmental delays, cognitive impairments and behavioural disorders. Brain MRI studies showed in some of these patients nonspecific T2/Flair white matter hyperintensities, which ranged in size and degree.

Objective: We describe neurological manifestations in a 45 years old man with aneuploidy 48, XXXY.

Methods: Subject underwent serial neurological, neurosurgical and neuropsychological evaluations, brain magnetic resonance imaging (MRI), [123I]-FP-CIT-SPECT and electroencephalogram.

Results: We present the case of a man with 48, XXXY aneuploidy who manifested movement disorders. Syndromic state included exotropia, overweight, tall stature, cognitive development delay and psychiatric disorders (impulse-control disorder and mood alterations). At the age of 35 years, he manifested postural and telekinetic tremor of the four limbs and of the head; after two years progressively worsening gait, balance disorder and urinary incontinence appeared. Brain MRI showed hydrocephalic dilatation of the lateral ventricles and small non-specific hyperintense alterations in T2 sequences in the white matter mainly of the frontal lobes. Neuropsychological evaluation highlighted impairment of verbal and visuospatial memory functions, of frontal functions and of selective attention. Electroencephalogram revealed diffuse slow waves compatible with mild encephalopathy without epileptic abnormalities. The DAT-SPECT was found to be normal. On neurological evaluation ataxic walking, postural tremor and telekinetic were evident, while other extrapyramidal signs were absent. Neurosurgeons decided not to perform derivative surgery. Propranolol therapy was not tolerated for episodes of blood hypotension. However, movement disorders and brain MRI remained unchanged over the next 10 years.

Conclusions: The case described increases the knowledge about the possible neurological manifestations of 48,XXXY aneuploidy.