Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Frequency of FMR1 premutation in individuals with ataxia and/or tremor and/or parkinsonism

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Genet. Mol. Res. 7 (1): 74-84 (2008)
Received April 20, 2007
Accepted December 15, 2007
Published January 29, 2008

ABSTRACT. A late onset neurological syndrome in carriers of premutation in FMR1 gene was recently described. The condition was named fragile-X-associated tremor/ataxia syndrome (FXTAS) and includes intentional tremor, cerebellar ataxia, parkinsonism, and cognitive deficit. We ascertained the contribution of FMR1 premutation to the phenotypes ataxia, tremor and/or parkinsonism. Sixty-six men over 45 years old presenting these symptoms, isolated or combined, were tested. Also, 74 normal men, randomly chosen in the population, formed the control group. In the patient group, no premutation carrier was found, which is in agreement with other observed frequencies reported elsewhere (0-5% variation).

No significant differences were found when comparing gray zone allele frequencies among target and control groups. The FXTAS contribution in patients with phenotypic manifestations of FXTAS was 15/748 (2%). The presence of gray zone alleles is not correlated with FXTAS occurrence.

Key words: FMR1; FXTAS; Ataxia; Parkinsonism; Premutation; Tremor