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CLINICAL INSIGHTS FROM A LARGE BRAZILIAN FRAGILE X FAMILY

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Fragile X syndrome (FXS) involves a large spectrum of cognitive impairment, varying from learning disabilities to all levels of mental retardation. The mechanism that leads to FXS in almost all patients is the amplification of the (CGG)_n repeat at the 5' untranslated region of the *FMR1* gene. We analyzed the segregation of the altered *FMR1* gene in a 6-generation family comprising 325 individuals. We evaluated normal and affected individuals, including clinical, psychological, speech and molecular testing. Clinical evaluation was performed in 98 individuals. Psychological tests were applied to 65 individuals, speech tests to 82, and molecular analysis of the *FMR1* gene was performed in 86 individuals. Pedigree analysis combined with molecular test results led us to the possible carrier of a premutation in the first generation. Factorial Analysis of Multiple Correspondence (SPAD.N software) showed an association between the *FMR1* premutation and speech disorders and an association between the *FMR1* full mutation with mental retardation, speech disorders and high and arched palate. Intrafamilial comparison of noncarriers and affected individuals showed that clinical features included in fragile X checklists might be familial traits without diagnostic significance.

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