Direct molecular analysis of FMR-1 gene mutation in patients with fragile Xq syndrome and their families Análisis molecular directo de mutaciones en el gen FMR-1 en pacientes con Síndrome de Xq frágil y sus familias

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Background: The unequivocal diagnosis of fragile Xq syndrome is based in the direct analysis of the underlying FMR-1 gene mutation, that consists in an increased number of trinucleotide CGG repetitions. Aim: To study families with fragile Xq syndrome, using the Southern technique for the analysis of the mutation. Subjects and methods: Fifteen individuals, pertaining to 6 families with fragile Xq syndrome, were studied. Clinical, cytogenetic and molecular analysis using Southern technique, were done. Results: Five male individuals had a clinically evident syndrome, confirmed by cytogenetic analysis that showed fragility in 10 to 29% of studied cells. One subject with a clinical picture suggesting fragile Xq had a normal cytogenetic study. The other studied subjects were the mothers of the five subjects with the syndrome, that must be carriers, and four brothers. Molecular analysis showed that seven subjects (5 males) had a complete mutation, five (4 females) were carriers of a pre mutat