MOLECULAR SCREENING FOR FRAGILE X SYNDROME IN THAILAND

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Running Title: Fragile X syndrome in Thailand

Abstract

Fragile X syndrome (FXS) is the most common form of inherited mental retardation. We screened for FXS in 237 Thai males (age ≤ 15 years) with developmental delay of unknown cause. We found 16 (6.8%) to have FXS using standard molecular analysis. We then studied the extended families of these 16 FXS subjects and 4 other independently ascertained FXS cases. We found that there were at least 35 affected males and 8 affected females. In addition we found that there were at least 31 premutation carrier females and 4 premutation males. The CGG repeats numbers in these premutation individuals ranged from 60 to 125. By comparison, the normal CGG repeats were 19-50 with a heterozygosity of 67.2% in 337 randomly selected males. This study provides insight into the high incidence of FXS in developmentally delayed Thai males and points the way toward the means of prevention of mental retardation by genetic counseling and prenatal diagnosis.