

## Abstract 81

### STUDY OF 5'UTR OF THE FMR-1 GENE IN MEN FROM SALVADOR, BAHIA, BRAZIL

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The Fragile X Syndrome (FXS) is a genetic disorder characterized by CGG expansion in FMR-1 gene and the gene variation in subjects from Bahia is still unknown. The aim of this study is to estimate the frequency of FMR-1 gene allelic classes in a representative group of male subjects from Salvador, the Bahia capital city. The allele frequency was determined by Polymerase Chain Reaction (PCR) according to Haddad et al 1996. Each sample was classified based on the relationship between the PCR fragment length and the number of CGG repeats. These categories are present as follows: classes 1 to 5 represent the normal alleles and class 6, 7 and 8 represents the Gray Zone, the premutation class and the mutation class, respectively. In this study, were analyzed 511 samples without FXS. There was a predominance of normal alleles: class 2 (39.0%), class 3 (34.6%), class 4 (25%) class 1 (1%) and class 5 (0.4%) corresponding to: 11 to 18 CGG repeats, 19 to 26 CGG repeats, 27 to 34 CGG repeats, <10 repeats CGG and 35 to 40 CGG repeats, respectively. No allele was found in class 6 (41 to 60 CGG repeats), class 7 (> 60 CGG repeats), or class 8 (> 200 CGG repeats) among analyzed samples. This is the first study using the PCR technique to FMR-1 gene alleles detection into population of Salvador. Through the molecular biology techniques used, the class allelic distribution from the FMR-1 gene was detected, turning possible its use as a screening diagnostic to FXS in this population.

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