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SCREENING FOR FRAGILE X SYNDROME IN A MENTALLY RETARDED POPULATION

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The Fragile X syndrome is the most common cause of inherited mental retardation. It is caused by triple nucleotide expansion at CGG at *FMR1* gene at FRAXA locus. Since 2000 we screened 702 mentally retarded subjects at the genetics clinics of Hospital de Clínicas de Porto Alegre for Fragile X syndrome using PCR according to Fu et al. (1991). The aim of this study was to evaluate the clinical data of the subjects screened for fragile X syndrome. Of the 702 cases we fully reviewed 470 unrelated males (420 FRAXA negative and 50 FRAXA positive). There was no difference between the mean ages of positive cases versus negative cases (10.16 years X 10.47 years). Using chi-square analysis we did not find significance between the FRAXA positive and negative groups for the following data: family history of mental retardation (not X-linked), consanguinity, microcephaly, perseverative speech, hyperactivity and seizures. The clinical data showed statistical significance for positive versus negative FRAXA test for X linked mental retardation (36% x 18.1%; p=0.005); macrocephaly (26% x 10.5% p= 0.004); long face (46% x 15%; p= 0.000); long ears (40% x 7.1%; p= 0.000); prominent ears (44% x 13.8%; p= 0.000); hyperextensible joints (38% x 7.9; p= 0.000); macroorchidism (20% x 1%; p=0.000); avoidance of eye contact (14% x 3.3% p=0.004); hand flapping (10% x 1.4%; p=0.003); hand calluses (16% x 0.7%; p= 0.000). Using at least two of the significant clinical data we were able to diagnose the majority of the positive cases (sensitivity 66% and specificity 81%). If the subject had 5 or more clinical findings all but one were FRAXA positive. This study showed the importance of appropriate clinical data to screen subjects with mental retardation in our population.