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FRAGILE X SCREENING FOR FRAXA AND FRAXE MUTATIONS USING PCR BASED STUDIES AMONG MALES WITH MILD/MODERATE MENTAL RETARDATION OF UNKNOWN ORIGIN: A BRAZILIAN EXPERIENCE OF COMMUNITY GENETICS

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Background: Fragile-X-syndrome (FXS) is the most common type of inherited cognitive impairment. The underlying molecular alteration consists of a CGG-repeat amplification within the FMR-1 gene (FRAXA). The two most common fragile sites with clinical significance are FRAXA at Xq27.3 comprising CGG repeat and a more distal FRAXE associated with amplification of a GCC repeat, located at Xq28. **Aims:** The aim of this study was to evaluate institutionalized patients of APAE (Associação dos Pais e Amigos dos Excepcionais) Limeira, Sao Paulo State, southeast Brazil. **Material and methods:** We surveyed 159 male patients through a clinical protocol and cytogenetical evaluation, 119 of them were selected to molecular screening (FRAXA and FRAXE) and G-banded chromosome analysis. **Results:** 09 were positive cases for X-Fragile syndrome (FXS - FRAXA). No FRAXE patients were identified. Two patients diagnosed had no family history of mental retardation. Southern blotting confirmed the carrier status of the FMR1 premutation in the respective mothers. **Conclusion:** The detection of FXS mutations has allowed us to offer more informed genetic counseling, prenatal diagnosis and reliable patient follow-up. This kind of screening can be an important step towards the inclusion of genetics in the context of the whole primary and secondary health care, working along with pediatricians, obstetricians, child neurologists, psychologists, physical/ speech and language therapists, nurses and social care workers.

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