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IN THE HEART OF BRAZIL: CLINICAL EVALUATION AND ETIOLOGICAL CHARACTERIZATION OF MENTALLY RETARDED SEMI-INSTITUTIONALIZED PATIENTS IN CENTRAL-WESTERN OF BRAZIL

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BACKGROUND: Mental retardation is a life-long disability occurring in 2-3% of all children. Genetic causes are very heterogeneous and the clinical presentation most often non-specific. Current diagnostic tools can only identify a specific aetiology in about 50% of cases, and more importantly, often do not exclude a genetic cause in apparently sporadic cases of mental retardation, although new genetic technologies are becoming available. A proven genetic aetiology has many implications for the extended family.

OBJECTIVE: To investigate etiological factors in mental retardation in one specialized institution (APAE - Associação dos Pais e Amigos dos Excepcionais) in Porto Nacional City, located 60km from Palmas (state capital of Tocantins, Central-Western of Brazil).

MATERIAL AND METHODS: A clinical geneticist performed the history and physical examination in 83 patients aged between 11 months and 63 years during a week. A blood sample was obtained in 79 of them for a genetic screening that included a standard karyotype, fragile X molecular genetic testing and search for inborn errors of metabolism using aminoacids and organic acid chromatography.

RESULTS: An environmental insult was noted in 18 patients; a specific autosomal genetic disorder in 10 patients; and no diagnosis made in 33 patients (42.5%). Among the 38 males, 13 of them were selected for molecular screening (FRAXA and FRAXE). Four had positive molecular screening (PCR) for X-fragile syndrome. Of all 83 patients, 18 had abnormal chromosome findings, with Down's syndrome noted in 12 of the patients (two cases with Robertsonian translocation) and Turner syndrome noted in 3 girls. 8 patients had features of MCA/MR syndromes and 5 had features of non-syndromic X-linked mental retardation; 3 patients had suggestive features of a hereditary metabolic disorder.

CONCLUSION: Clinical, cytogenetic and molecular screening of mentally retarded patients for the fragile-X syndrome and other causes of mental retardation is helpful in identifying individuals and their families who may benefit from genetic services such as counseling and treatment, specially in poor regions with limited access to the genetic evaluation.