

## Abstract 65

### GENETIC SCREENING TO DETERMINE AN ETIOLOGICAL DIAGNOSIS IN PATIENTS WITH MENTAL RETARDATION IN A SOUTHEAST REGION OF BRAZIL

Charles Marques Lourenço<sup>1</sup>, Elisabete Giusti<sup>2</sup>; Vanessa Veríssimo Viana<sup>2</sup>; Egeu Bosse<sup>2</sup>, Luciana Benedeti Lavoura<sup>2</sup>, Antonia de Souza Viana<sup>2</sup>, Daniele Candioto Vidal<sup>2</sup>, Ludmila Serafim de Abreu<sup>3</sup>; João Monteiro de Pina-Neto<sup>1,3</sup>

Neurogenetics Unit, Medical Genetics Division, Clinical Hospital of Ribeirao Preto, University of São Paulo, Brazil<sup>1</sup>

Genetics Department, School of Medicine of Ribeirao Preto<sup>2</sup>

APAE (Associação de Pais e Amigos dos Excepcionais), Limeira, São Paulo, Brazil<sup>3</sup>

**Background:** Mental retardation or intellectual disability affects 2% of the general population, but in 60% to 70% of cases the real cause of this retardation is not known. An early etiologic diagnosis of intellectual disability can lead to opportunities for improved educational interventions, reinforcing weak areas and providing a genetic counseling to the family.

**Objective:** To investigate etiological factors in mental retardation in one educational institution APAE (Associação dos Pais e Amigos dos Excepcionais) for mental impaired patients (adults and children) in Limeira, Sao Paulo State, Southeast of Brazil.

**Methods:** A detailed clinical, cytogenetic, biochemical and molecular survey was undertaken on 300 patients with no previously recognized cause of retardation (patients with clinical diagnosis of Down syndrome were excluded from this evaluation). The etiological study was based on a clinical genetic approach with special attention to dysmorphism and neurological findings.

**Results:** The patients had a female: male ratio of 1:2 and their ages varied from 10 months to 48 years. The patients were classified as having mental retardation of unknown origin, i.e. patients with apparently normal pre-, peri- and postnatal histories who had neither dysmorphism nor affected first-degree relatives, and had a normal karyotype and metabolic screen. Among the 193 males, 151 of them were selected for molecular screening (FRAXA and FRAXE) and G-banded chromosome analysis, 11 had positive molecular screening (PCR) and/or Southern blotting for fragile-X syndrome. Of all patients, 15 had abnormal chromosome findings; it was worthy to note that Triple X syndrome was noted in 3 girls with minor dysmorphisms, 35 patients had features of MCA/MR syndromes and 69 had features of non-syndromic X-linked mental retardation based on the family's pedigree. The former group of MCA/MR syndromes may include unidentifiable chromosomal aberrations, uniparental disomy, mutations and multifactorial factors.

**Conclusion:** Accurate determination of the etiology of mental retardation is important not only for genetic counseling purposes, but also in identifying prenatal events which make infants more vulnerable to perinatal and postnatal risk factors.

Acknowledgement: Angela M. Vianna-Morgante (IB/USP) for Southern blot analysis.