

## Abstract 94

### FRAGILE X SYNDROME : FRAGILITY IN GUARANTEEING HUMAN RIGHTS

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Nearly 18 years after the cloning of the *FMR1* gene, the FXS remains a health care and social challenge, particularly in developing communities. During the years of 2007 and 2008, we studied 92 male individuals aged 3 to 25 years presenting idiopathic mental retardation (MR), from 81 unrelated families. They attended a genetics and a neuropsychiatric clinic in Salvador city, Bahia, Brazil. Clinical evaluations and structured interviews were performed in multidisciplinary home visits in 64 of these families. A PCR test for FXS was performed in 44 mentally retarded individuals from 41 families, and 18 of them (15 probands, 36,6%) tested positive. The methods of data collection and analysis enabled assessment of the impact of FXS diagnosis on the families, as well as association of their health care status with social and economical parameters. MR was recurrent in about 70% of 58 families for which bona fide data were available. Since the majority of MR individuals in the community are not provided with a causative diagnosis for their MR, and their relatives rarely counseled, it may be inferred that their right to diagnosis was not fulfilled. The growing advances in the diagnosis of the causes of mental retardation and awareness of norms for human rights are not enough, per se, to compensate for the lack of efficient public health policies. In relation to FXS, the major cause of inherited mental retardation, the absence of organized strategies to diagnose the syndrome impacts those who are poorer and more vulnerable more severely than higher income families. This situation places relatives of FXS individuals in the condition of "silent transmitters", a silence with cross-generational repercussions, which affect human rights.