

Clinical Genetics

AB010. Aberrant behavior profile of fragile X syndrome in medically underserved population

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Background: Fragile X syndrome (FXS) is the most common cause of inherited intellectual disability and it is associated with autism spectrum disorders. Abnormal neurological symptoms are believed due to the lack fragile X mental retardation protein (FMRP). By understanding the role of FMRP in the brain, some therapeutic approaches have been tried with the hope to reduce or prevent FXS symptoms. In Indonesia, a number of children with FXS are diagnosed and treated as a developmentally disabled child without etiologic diagnosis of FXS. None of them get community-based services such as language, occupation, and behavioral therapy as a basic intervention. This study was aimed to describe baseline aberrant behavior profile of FXS individuals in Central Java and Yogyakarta province, Indonesia.

Methods: PCR-based and Southern blot analyses were carried out to determine FXS diagnosis. The Aberrant

Behavior Checklist-Community (ABC-C) was used to measure aberrant behavior profile. This was done by caregiver who spent at least 6 hours/day with FXS individual. Fifty-eight items were used to assess maladaptive behaviors across five subscales: irritability, lethargy/social withdrawal, stereotypic behavior, hyperactivity, and inappropriate speech.

Results: Forty-six FXS individuals were enrolled including 30 males and 16 females, with range of 7 to 60 years (6 children; 40 adults). The analysis of this study was focusing on adulthood manifestations, as the number of children was small. Adult males had significantly higher scores of irritability ($P=0.02$) and hyperactivity ($P=0.001$) compared to females. Irritability in males and females were remarkably decreased with age. Interestingly, in males, lethargy/social withdrawal and stereotypic behavior were gradually increased with age. In contrast, stereotypic behavior was gradually decreased with age in the female patients.

Conclusions: Males have more severe degree of irritability and hyperactivity compared to females and the social withdrawal is worsening with age in adult males with FXS.

Keywords: Aberrant behavior profile; fragile X syndrome (FXS); intellectual disability; Indonesia

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