15. HOW MENTAL RETARDATION AND AUTISM SYMPTOMS ARE MORE PRONOUNCED IN BOYS WITH FRAGILE X SYNDROME DUE TO LACK OF GENETIC COMPENSATION

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Introduction. Fragile X syndrome is a genetic disorder that is inherited through the X chromosome. It is caused by a mutation in the FMR1 gene, which is located on the X chromosome. When a male inherits an X chromosome with the mutated FMR1 gene, they do not have the genetic compensation that girls have. This lack of compensation makes them more severely affected by the X-linked inheritance of the mutation. The main reason is Absence of Genetic Compensation in Boys: Girls, who have two X chromosomes, have the ability to undergo genetic compensation. The normal X chromosome can partially compensate for the effects of the mutated one, which is inactivated or turned off in each cell. As a result, the impact of the FMR1 gene mutation is more pronounced in boys, leading to more severe intellectual and developmental disabilities. The FMR1 gene contains instructions for the creation of the Fragile X Mental Retardation Protein (FMRP), which plays a crucial role in the brain's synaptic function and plasticity. In individuals with Fragile X syndrome, the mutation causes a decrease or absence of FMRP production, which negatively impacts synaptic communication and contributes to cognitive impairments.

Aim of study. Symptoms of autism, including challenges in social interactions, communication, and repetitive behaviors, are intensified in males affected by Fragile X syndrome. The lack of genetic buffering mechanisms worsens these difficulties, resulting in a more pronounced display of autistic characteristics.

Methods and materials. Hessl D, Nguyen DV, Green C, et al. conducted a study in 2008 in 217 children with FXS (age 6–17 years, 83 girls and 134 boys).

Results. The absence or insufficiency of FMRP in boys has a greater impact on neurodevelopment, resulting in more severe intellectual challenges. Boys with Fragile X syndrome experience more profound effects on cognitive functions, learning, and adaptive behaviors compared to girls, such as limitations in object discrimination learning and reversal tasks, visual-working memory, verbal short-term memory

Conclusion. To sum up, the absence of genetic compensation in males diagnosed with Fragile X syndrome intensifies the severity of cognitive impairment and symptoms of autism. It is essential for healthcare professionals, educators, and families to grasp the complexities of X-linked inheritance and its impact on neurodevelopment. Several factors, including the type of mutation, genetic background, and environmental influences, can influence the variability observed in individuals. Early detection, intervention, and continuous assistance play a critical role in enabling individuals with Fragile X syndrome, irrespective of their gender, to achieve their highest capabilities.