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What Does We Know of Chromosome X Fragile

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Abstract

Chromosome X Fragile is the prevalent hereditary cause of intellectual disability. It is an inherited disease linked to the X chromosome, Fragile X syndrome (S X F). Its main clinical manifestation is intellectual disability and affects mainly males, with a prevalence of 1/4000 and females 1/6000. Women are the transmitters. The cause is a mutation on chromosome X9 27.3 and consists of an abnormal expansion of the trinucleotide citicin-guanine-guanine (CGG) in the FMR 1 gene (Fragile X Mental Retardation) in an area not coded at the beginning of the gene.

Introduction

Female of 16 years of age, cesarean birth due to lack of progression and descent, Apgar 9-10 E.G, 38 and 1/2 weeks by clinical examination and Date of last menstruation, placenta with calcifications, Birth weight 2850 gr, size 50 cm. Neonatal examination within normal limits. Normal facies. In the post natal period there is a delay of psychomotor development, walking at 21 months of age, good adaptation to the environment with moments of apparent disconnection of the same. She does not speak but is understood by gestures. The head pediatrician is consulted who does not give importance to the facts and diagnoses that it is a "Slow Maturity".

At 4 years of age she entered the kindergarten, where it observed: aggressiveness towards his classmates and rejection of all school and play activities. Inter consulence is indicated with pedagogy and neurology where mild mental retardation is diagnosed. It begins later, with enuresis and treatment with psychologist, neurologist and psychomotrocist. Schooling was not achieved in spite of numerous attempts in different institutions.

The following EEG, NMR TAC studies were performed; Neuroendocrine, Neurometabolic and genetic, all being normal. At age of 12, and coinciding with menarche, she presented a tonic seizure and pathological electroencephalogram. She is medicated with anticonvulsants, continues with psychological treatment and a specific study is requested for fragile X chromosome that gives altered. At present there is slight mental retardation, stereotyped hand movements, oscillating behaviors between normality, aggressiveness and autism. Problems of social integration and rejection of new stimuli.

Conclusion

Although the fact of having had a diagnosis since of 580 children investigated none had, the fact of knowing or simply thinking by the pediatrician that there are always physical pathognomonic traits of a genetic disease. While this will not cure the child so far, it will help him to carry out activities to improve his adaptation to the environment avoiding frustrations in the parents and family, which in many cases, ends up disintegrating. Making smart decisions in medicine involves being aware of the process involved and its effects.