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Case Report

Rett Syndrome

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Abstract

Rett sindrome is a severe neuro developmental disorder that is a leading cause of mental retardation in females, characterized by an apparently normal psycho motor development through the first 6 months of life, followed by stagnation and growth regression in different are as like motor, language and social skills; patients often exhibitautistic behaviors in the early stages. Other symptoms include seizures, breathing problems when awake such as hyper ventilation, apnea, and swallowing air; ataxia and stereotypich and movements. It is caused by mutations in the X-linked gene encodingmethyl-CpG-binding protein 2 (MECP2) [1,2]. One case is presented with positive molecular study.

Keywords: Autism; Intellectual Disability; Rett Syndrome

Introduction

Rett síndrome was first described in 1966 by the austrian neurologist Andreas Rett. It usually affects women and is the second cause of profound mental retardation. Its cause has been associated with a mutation located on the X chromosome in Xq28 in the MECP2 gene that codes for the proteinmethyl-CpG-binding protein [3,4].

Case Report

Female 2 years old without significant family or perinatal history. At 6 months old, she presented neurodevelopment regression, not achieving a sitting position and losing the cephalic support with epileptic spasms in flexion, 10 times a day, with 7clusters, in wakefulness. At the current physical examination with head circumference 16 in, with little visual contact, there are stereotypies in the hands, spasticity, screaming battacks, no language, there is only babbling, with episodes of hyperventilation and sleep disturbance. MRI of the skull with ventriculomegaly. Electro encephalogram hypsar rhythmia with burst supression. Molecular study positive for MECP2 in exon 4c.1233C>T. Current treatment with vigabatr in and magnesium valproate [5].

Discussion

In 2004, Kammoun et al. [6] Proposed four basic criteria for diagnosis:

a) severe psychomotor delay, with impairment of receptor and expressive language.

b) cephalic perimeter inferior to normal.

c) Loss of useful employment of the hand, associated with a dysfunction in communication and social isolation.

d) Appearance of stereotyped movements of the hands. Other characteristic symptoms are walking on tiptoe, sleeping problems, bruxism, seizures, apnea and / or hyperventilation [5,6].

Conclusion

The case present edisstage 2, late destructive, with loss of acquired skills, global neurodevelopmental delay, respiratory alterations and epileptic seizures. There is no specific treatment that can reverse or stop the course of the disease. The rapeutic management revolves around the restoration of synaptic function and maturation, since it has been shown that the deficitis at the level of micro circuits that involve synaptic transmission. The management is symptomatic and individualized. A multidisciplinary and dynamic approach is essential. The knowledge of the genetic causes allows a diagnostic confirmation, a family genetic counseling, an evolutionary prognosis and the application of a therapy to the disease in the very near future [7,8] (Figure 1).

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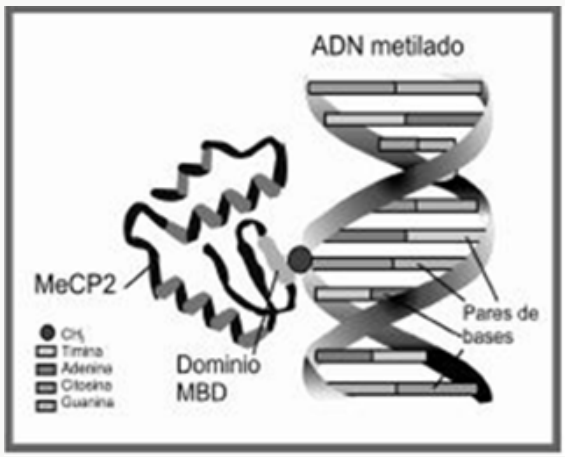

Figure 1: MECP2 gene.

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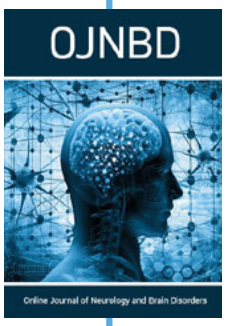

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