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Case Report



# Rett Syndrome: Reporting A Case and Review in Literature

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## Abstract

Rett syndrome is a disorder in children's neurological development characterized by an initial normal evolution followed by the loss of voluntary use and characteristic movements of the hands, a delayed growth of the brain and head, difficulty walking, seizures and mental retardation. The syndrome affects almost exclusively girls and was identified by Dr. Andreas Rett in 1966.

**Keywords:** Rett syndrome; Psychomotor retardation; MECP2 Gene; Baden-baden criteria

## Case Report

A 9-year-old female patient who is not aware of the onset of her ailment since she is the product of the penultimate feat of 6 in total and they did not take care of her since her mother was engaged in prostitution, apart from being beaten and sexual attacks. It is seen in the House of Angels in the City of Puebla for children with mental problems. Clinically it is known from 3 years of age, time that the institution lives. Since then it is observed with autoagresiones, psychomotor retardation and of growth, with diminution of the cephalic perimeter, it is known that it has epilepsy from the 6 months of age with a difficult control of its crisis as well as an autistic behavior; the fluttering of his hands is striking in a random, symmetrical way and that even sometimes merits subjection as it becomes self-injuring.

## Discussion

Rett syndrome is diagnosed by observing signs and symptoms during the child's initial growth and development and by periodic evaluations of his physical and neurological status. Recently, scientists developed a genetic test to confirm the clinical diagnosis of this disorder; the test involves looking for the MECP2 type mutation on the child's X chromosome. Given what we know about the genes involved in Rett syndrome, such tests can identify up to 80 percent of cases.

Some children who have characteristics similar to Rett syndrome or who have MECP2 type genetic mutations do not meet the criteria for the diagnosis of the syndrome as specified below. These people are described as having an "atypical" Rett syndrome. Atypical cases constitute about 15 percent of the total number of cases diagnosed.

Examples of essential diagnostic criteria or symptoms include an apparently normal development up to 6 and 18 months of age and having a normal head circumference at birth followed by delays in the rate of growth of the head over time (between 3 months and 4 years of age). Other essential diagnostic criteria include a significant deterioration of speech, repetitive movements of the hand, torso jolts, walking on the tips of the foot or an unstable, rigid and with increased lift base (legs apart).

Support criteria are not required for a diagnosis of Rett syndrome but may occur in some patients. In addition, these symptoms - the severity of which varies from child to child - may not be present in very young girls, but may develop with age. A child who meets the support criteria but does not meet any of the essential criteria does not have Rett syndrome. Support criteria include difficulties in breathing; abnormalities in the electroencephalogram (EEG); convulsions; muscle stiffness, spasticity and / or contractures that worsen with age; scoliosis; grinding or gnashing teeth; small feet in relation to height; delays in growth; decreased body fat and muscle mass (although there may be a trend towards obesity in some affected adults); abnormal sleep patterns, irritability or agitation; difficulties to chew or swallow (swallow); bad circulation in the lower extremities, with cold and bruised feet and legs; decreased

mobility with age and constipation. Diagnostic criteria for classic rett syndrome established by the consensus of the working group [1,2].

### Necessary criteria

a. Prenatal and perinatal period apparently normal.

b. Determal psychomotor development first 6 months of life.

c. Normal cranial blood pressure at birth.

d. Deceleration of the cranial perimeter between 5 months and 4 years of life.

e. Decrease in voluntary hand activity between 6 months and 5 years of age, associated with communication dysfunction and social rejection.

f. Expressive and receptive language very deteriorated with impairment of psychomotor development.

g. Estereotypes of hands: twisting, squeezing, palming / hitting, salivating, hand washing and friction.

h. Anomalies in ambulation or non-acquisition of the march.

i. Possibility of a clinical diagnosis between 2 and 5 years of age

### Support criteria

a. Anomalies of the breathing rhythm in wakefulness.

b. Periodic vigil vigils.

c. Intermittent hyperventilation

d. Period of holding your breath.

e. Forced emission of air and saliva.

f. Abdominal discharge by swallowing large amounts of air.

g. EEG abnormalities such as slow-wake rhythm in wakefulness and intermittent patterns of slow rhythms (3-5 Hz) and paroxysmal discharges with or without clinical crises.

h. Convulsions

i. Anomalies of muscle tone with atrophy of muscle masses and / or dystonia

j. Peripheral vasomotor disorders

k. Scoliosis / xiphosis.

l. Delay in growth (size)

m. Hypotrophic and cold small feet

n. Anomalies in the sleep pattern of the infant, with greater time of day time sleep.

### Exclusion criteria

a. Evidence of an intrauterine growth retardation

b. Organomegaly or other signs of deposit disease.

c. Retinopathy or optic atrophy

d. Presence of a progressive metabolic or neurological disorder.

e. Neurological secondary pathologies due to serious infections or head injuries

## Conclusion

It is recommended to consult a pediatric neurologist, or a pediatrician specialized in the development to confirm the clinical diagnosis of Rett syndrome. The doctor uses very specific guidelines, which are divided into three types of clinical criteria: essential, support and exclusion. The presence of any of the exclusion criteria denies a “classic” or “typical” diagnosis of Rett syndrome.

## References

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