

CASE REPORT

A Case of Rett Syndrome

by

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Abstract

A case of Rett syndrome in a 3½ year-old girl is presented. The patient had normal pre and perinatal period and normal psychomotor development till the age of 14 months, followed by behavioural, social and psychomotor regression. Physical examination revealed a below normal head circumference, loss of eye and psychic contact, stereotypic hand movements and gait disturbance. No laboratory test can confirm the diagnosis of Rett syndrome, therefore the diagnosis was established by virtue of history of illness and clinical manifestations. This is the first case of Rett syndrome found and reported in Indonesia.

Introduction

Rett syndrome is a relatively newly recognised disease. It was Andreas Rett in 1966 who first described in detail a disease entity which had never been reported previously. Rett himself, named the syndrome as 'ein cerebral-atropisches syndrome bei hyperammonämie' (cerebral atrophy accompanied by hyperammonemia). The clinical manifestations he noted at that time, consisted of autism, dementia, apraxic gait, loss of expression and stereotypic movements, accompanied by cerebral atrophy and hyperammonemia (Rett, 1966, cited from Hagberg et al., 1983).

The first publication by Andreas Rett was in German. Perhaps due to language barrier, his publication was not widely read. Eleven years elapsed before he made another publication in English. Since that time, the syndrome has been widely recognised (Rett, 1987).

Up till 1987, there have been reports of Rett syndrome cases from 27 countries. In the United States, for example, attention to the syndrome has rapidly increased and parents of children with Rett syndrome have founded an organisation, the International Rett Syndrome Association. However, until recently, there hasn't been any reported Rett syndrome case in Indonesia.

This syndrome affects only girls. The prevalence is 1 in 1500 girls in Sweden (Hagberg, 1985) and 1 in 9000 girls in Scotland (Rett, 1987). This syndrome does not seem to be a rare phenomenon but it is more or less universal (Hagberg et al., 1985).

The etiology and pathophysiology of the disease are not clearly understood. Nomura et al. (1985) suggested that the initial lesion formed in the lower central nervous system, either the brainstem and/or midbrain.

Missliwetz and Depastas (1985), found cerebral atrophy and depigmentation of the substantia nigra in an autopsy of a case with Rett syndrome. Foss et al. (1985), investigated urine protein by chromatography and found that the results were normal. Blood ammonia level may increase but it is not statistically significant (Hagberg et al., 1983; Riederer et al., 1985).

The signs and symptoms of the disease include:

- (1) Severe dementia or loss of intellectual function.
- (2) Loss of contact with environment which is manifested by autistic behaviour.
- (3) Loss of expression or amimia.
- (4) Alalia or lack of ability to talk.
- (5) The most important sign is stereotypic movements of the hands. The patients do repeated movements resembling rubbing, washing, wringing and they lose the ability of normal purposeful hand use such as writing or holding.
- (6) Walking with a wide and unstable gait and stooping.
- (7) Epileptic seizures occur in 70% of the cases.
- (8) CT scan of the head may show normal or reveal atrophy of the brain. (Hagberg et al., 1985).

Until recently there hasn't been any laboratory test to confirm the diagnosis.

The treatment for Rett syndrome is physiotherapy to improve muscle tone and prevent scoliosis. Rett (1987) also suggested music therapy to improve contact with the environment.

Case

H, a female, 3½ years old, Indonesian of Chinese origin. From the history of illness, we found that the girl had a normal growth and development till the age of 13 months. It was at the age of 14 months, when her parents noticed something extraordinary. She moved her hands resembling washing, walked with a wide and unstable gait and began stooping. She was taken to a pediatrician. EEG was performed and the result was abnormal. Some drugs were administered then without any conspicuous improvement. CT scan was also performed and the result was normal. Gradually, her condition became worse. She could hardly communicate with others. Her hands were frequently put into her mouth. Grasping was severely compromised such that she could not hold any object without dropping it. She could not speak nor understand commands. At the age of 3 years, her condition stabilized. There has been no improvement nor deterioration. History of pregnancy and delivery were normal. She was born spontaneously, term, immediately cried, with birthweight of 2500 grams and body length of 49 cms. The head circumference at birth was normal. Developmental milestones were normal till the age of 13 months. Immunization was complete. She was the youngest of 3 siblings, others were normal. No consanguinity existed between the parents.

On physical examination, she was alert, without expression, rather hyperactive. The head circumference was 45,5 cm (below normal). Her body temperature was normal. Her body weight and body length were normal for her age. Heart rate, pulse rate, respiratory rate and blood pressure were within normal limits. Eye and environmental contact were absent. Eye fundi were normal heart, lungs and abdomen were normal. On extremities and gait examination, we found stereotypic hand movements without any purpose, resembling washing and wringing or putting her hands into the mouth. She could not hold any object well. She walked with a wide and unstable gait and stooping. On the left leg, the deep tendon reflex increased and Babinski sign was positive.

Routine blood, urine and stool examination did not show any abnormalities. Blood ammonia was normal. EEG (sleep records) showed left focal irritation.

The working diagnosis was Rett syndrome. We consulted Professor Andreas Rett for further confirmation (by showing him the video-tape of the patient) and it was agreed that the case was a typical Rett syndrome.

The treatment was physiotherapy to improve muscle tone and prevent scoliosis. Music therapy was also introduced and seemed to give a good result.

Discussion

The diagnosis of Rett syndrome in this case was based on the history of illness and clinical manifestations. We found the case at the age of 3½ years. Looking back, the diagnosis could have been established earlier, as several symptoms such as stereotypic hand movements and gait disturbance

have been noticed at the age of 14 months. But perhaps the delay was due to lack of familiarity to such a case.

This patient fulfilled all the diagnostic criteria for inclusion and exclusion stated by Hagberg et al. (1985). The criteria for inclusion are :

- (1) Female sex.
- (2) A normal pre and perinatal period, essentially normal psychomotor development through the first 6, often 12-18 months of life.
- (3) Normal head circumference at birth. Deceleration of head growth (and therefore, by interference, brain growth) between 6 months - 4 years of age.
- (4) Early behavioural, social and psychomotor regression (loss of achieved abilities), development of communication dysfunction and signs of dementia.
- (5) Loss of acquired purposeful hand skill through ages 1-4 years.
- (6) Hand wringing - clapping - 'washing hand' stereotypies appearing between ages 1-4 years.
- (7) Appearance of gait apraxia and truncal apraxia (ataxia) through ages 1-4 years.
- (8) Diagnosis is tentative until 3-5 years of age.

The criteria for exclusion are :

- (1) Visceromegaly, other signs of organ storage.
- (2) Retinopathy or optic atrophy before the age of 6 years.
- (3) Congenital microcephaly.
- (4) Perinatally acquired brain impairment.

The head circumference of this patient was 45,5 cms. It was below normal for her age and was compatible with the head circumference of a 1½ year - old girl. From this data, we try to draw a conclusion that the disease might have occurred at the age of approximately 1½ years, leading to decelerated brain growth since then. This fact corresponded well with the history of illness that the first symptom occurred at the age of 14 months.

It seemed that music therapy gave a good result, marked by a better contact with the environment. However, further follow up was not possible due to parental rejection.

Eventhough this is the first case of Rett syndrome found and reported in Indonesia, we presume that other such cases may exist here but have gone unrecognised.

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