

CaseReports

A girl with Rett Syndrome

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Introduction

Although Rett syndrome (RS) was first described by Andreas Rett in 1966, this disorder became internationally recognized only after the report of Hagberg et al. in 1983¹. It is a neurodevelopmental disorder which affects almost exclusively girls and is associated with deceleration of head growth, typical stereotyped hand movements, severe mental deficiency, cortical and extrapyramidal dysfunction including gait disturbance and truncal ataxia as well as loss of purposeful use of the hands^{2,3}.

Case report

A 4 ½ year old girl presented to General Hospital Ratnapura with a history of reduced speech, abnormal hand movements and reduced awareness of surroundings of one year duration (Figure 1).



Figure 1 Child on presentation

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She had normal speech development until the age of 3 1/2 years; she could speak four word sentences, relate a short story and sing songs. Her speech gradually deteriorated over one year and now she can only speak three single words. At 3 1/2 years she could draw a circle although now she cannot scribble due to wringing movements of both hands, more on the left. Child's grasp is abnormal and she is unable to hold the plate or pen properly. Her social development was age appropriate till 3 1/2 years. Now she avoids eye contact and interaction with peers and plays on her own. Her gross motor development is normal for the age. Mother recollected 4-5 episodes of sighing breathing during the last 4 months. Child is grinding her teeth at night. The sleep pattern is normal. Parents sought indigenous treatment for one year. As there was no response to indigenous treatment she was brought to get medical advice.

She never had seizures or fainting attacks. There was no history of head injury or measles in infancy. Excepting three hospital admissions for minor ailments like viral fever and diarrhoea, child was well previously. Child is the younger of two girls, product of a non-consanguineous marriage. Birth weight was 3 kg. There were no perinatal complications and no neonatal jaundice. Elder girl is 6 1/2 years old and is in good health. There are no relatives with epilepsy, psychiatric disorders, mental subnormality or liver disease such as Wilson disease.

On examination, child's weight was 9 kg, well below the 3rd centile for her age; height was 95cms, at 3rd centile; occipito frontal circumference was 46 cm well below 3rd centile. Child was active, conscious, shy, covering half of the face with one hand (Figure 1); there were no dysmorphic features; child was not pale or icteric; hand wringing movements were present, more on left side; she holds left with right hand. There were numerous carious teeth. Dry skin and ichthyosis were present. There was no scoliosis. The cardiovascular and respiratory systems and abdominal examination revealed no abnormality.

Central nervous system i.e. higher functions, cranial nerves, including fundi, were normal. Tone and reflexes were normal. Power-grasp was poor but difficult to assess. Gait was normal.

Blood counts, liver function tests and renal function tests were normal. ECG was normal. Epileptiform changes were present in EEG.

As this girl has a regression disorder with apparently normal development until 3 1/2 years, normal head circumference at birth followed by slowing, repetitive hand movements, reduced speech and growth retardation, a diagnosis of Rett syndrome was made.

Discussion

Prevalence of Rett syndrome is estimated between 1:10,000 and 1:15,000 girls⁴. Until 1999, there was no known biochemical, morphological or genetic marker for RS and diagnosis was established on clinical criteria⁵. Recently, Amir et al. have identified mutations on the gene MECP2, which is located on Xq28 and encodes methyl-CpG-binding protein 2 (MeCP2) in patients with RS⁶. Mutations have been found in about 80% of RS patients⁷.

References

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