

## Case Report



## Rett Syndrome – Masquerading Autism

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### ABSTRACT

Rett syndrome is a rare genetic disorder which mostly affects the girls. We present a case of three year old girl with Rett syndrome who had normal development till the age of one and a half year. Gradually over the next few months she lost her speech both expressive and receptive, acquired purposeful movements and motor control. Physiotherapy and occupational therapy brought partial improvement.

**Keywords:** Rett syndrome, Developmental disability, Pervasive developmental disorder, developmental regression.

### INTRODUCTION

**R**ett syndrome, a recently developed progressive neurodevelopment disorder exclusively affects girls. This disorder is characterized by normal development followed by a progressive loss of acquired functions. The most common symptoms of Rett syndrome are deceleration of head growth, small hand and feet, deterioration of motor and language skills, repetitive stereotypic purposeful hand movements such as hand wringing, hyperventilation and abnormal sleeping pattern. Rett syndrome was first recognized in 1966. The prevalence rate is 1 in 10,000 to 1 in 22,000.<sup>1</sup> The first reported case from India was in 1994.<sup>2</sup> Since then a few cases have been reported from Indian subcontinent.<sup>3,4,5</sup> The incidence of recurrence in a family and the likelihood of having another child with Rett syndrome is less than 1%.<sup>6</sup>

### CASE REPORT

A three year Bengali girl was brought to the outpatient department of pediatrics, IMS and SUM hospital by her parents with complains of poor mental development, loss of speech and motor development, homolateral stereotypic hand movement, loss of purposeful hand activity since last eighteen months. Parents reported that she was born from a nonconsanguineous marriage, full term normal delivery at hospital without any perinatal complications. There was no history of any major illness or seizure in past. Her head size was 34 cm with a weight of 2.7 kg at birth. She had a normal development until one and a half year than gradually started deteriorating. Her somatic and head growth became slow. Her hands and feet were small. She could not sit and stand with support or walk. She lost her acquired speech. She could not indicate her wants, defecating and urinating in clothes. She could not recognize her parents; family members, even her play materials and had no reaction to others. Belly breathing was noticed followed by frequent

vomiting and hyperventilation. She exhibited self mutilating behaviors like hair pulling, teething, and self biting and thumbs sucking. At present her head circumference was 45.6 cm. At the age of 17 month she was diagnosed as a case of autism and was receiving therapy elsewhere. Her CT scan, MRI and EEG, audiometric reports were normal and genetic report for MECP2 was positive. She was diagnosed as Rett syndrome.

She was referred for developmental evaluation. Developmental assessment indicates the presence of profound degree of delay in all areas of development. Her developmental age was found to be 6 months with a developmental quotient of seventeen. Parents counseled about the condition and its deteriorating course. The girl was on occupational therapy, physiotherapy, speech therapy and music therapy since last three months. She can say papa, can sit with support, extending hand for grasping and moving her eyes and started responding to music.

### DISCUSSION

Hagberg described rett syndrome as a progressive syndrome of autism, dementia, ataxia and loss of purposeful hand use in girls.<sup>7</sup> This disorder leads to developmental reversals and often misdiagnosed as autism because of marked impairment in social interaction, communication and stereotyped behavior. In rett syndrome a child start out normally, in between 6 to 18 months shows slow progressive regression in developmental milestones, head circumferences and overall growth where in autism aberrant development is usually present in early childhood. Gait abnormality and breathing difficulty are specific in rett syndrome unlike autism. In rett syndrome repetitive stereotypic hand movements are always present while in autism it may or may not be seen. In rett syndrome both receptive and expressive communication is usually completely lost but



an autistic child can use aberrant language. Rett syndrome affects only girls and caused by mutations in an X linked MECP2 gene which is not in case of autism, cerebral palsy and childhood integrative disorder.<sup>8</sup>

Unlike rett syndrome there are no normal developmental period and stereotypic hand movement and belly breathing in cerebral palsy.<sup>9</sup> In childhood disintegrative disorder normal development occurs for first two years then sudden loss of speech and social skills is marked. It is an extremely rare disorder, affecting mostly males. Usually these children exhibit very low level of intelligence and develop seizure disorders. In our case, the girl was apparently normal with average birth weight and head size, and normal developmental milestones until one and half year of her life. The symptoms started by slow head growth associated with loss of purposeful repetitive homolateral stereotypic hand movements, autistic behaviors and severe psychomotor retardation. Later the genetic report confirmed the diagnosis.

### CONCLUSION

The aim of therapeutic intervention is to slow the loss of abilities, improve the movements and encourage communication skills, social interaction and cognitive functioning. Behavior therapy may help to control self injurious behavior and regulate breathing difficulties. Therefore correct diagnosis is an important factor for specific intervention.

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