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CASE

Rett's syndrome: a case report

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Abstract

Rett's syndrome is a neurodevelopmental disorder that occurs almost exclusively in females and has a typically deteriorating course. We describe a case of Rett's syndrome in a girl of three years. She had normal development till two years of age. She was presented to Psychiatry outpatient department (OPD), Assam Medical College and Hospital (AMCH) with loss of her acquired hand skills and speech since last one year. She also developed purposeless stereotypic movements of hands and unable to hold objects in her hands. Gradually she has developed broad based gait and teeth grinding. This has been the first case of Rett's syndrome reported in North East India and West Bengal.

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Introduction

Rett's syndrome is a rare neurodevelopmental disorder[1] of the grey matter of the brain that almost exclusively affects females,[2] but has also been found in male patients. Head circumference is normal at birth, and early developmental milestones are unremarkable. Between five and 48 months (usually between six months and one year), head growth begins to decelerate. Purposeful hand movements are lost and characteristic midline hand-wringing or hand-washing stereotypes develop. Expressive and receptive language skills become severely impaired and are associated with marked mental retardation. In the preschool years, gait apraxia and truncal apraxia, and ataxia develop. A loss of social interactional skills is frequently observed during the preschool years, but social interest often increases later.

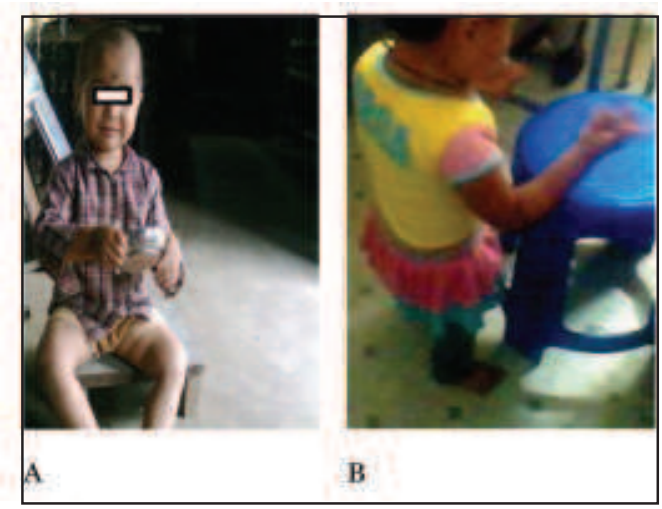
The case

A three-year-old female child has attended to Psychiatry outpatient department (OPD), Assam Medical College and Hospital (AMCH), escorted by mother, being accompanied by accredited social health activist (ASHA), with history of gradual loss of speech, hand skills, and stereotyped movements of the hand and body since last one year. She also had teeth grinding, spitting out saliva, screaming since last four months. She had normal development till two years of age, and suddenly the development became stagnant. Gradually her milestones' achievements were delayed by three years. By three years, she could not walk properly and could not speak any meaningful words. Parents reported her inability to hold, pick up, or grasp things in her hands, which she previously could do. They also noticed that she moved her hands without any purpose. She remains aloof and had decreased attachment to parents. She screamed and smiled without any apparent reason, and moved around aimlessly. For last four months she develops teeth grinding, spitting out her saliva forcefully making a sound by her tongue. There was no history of seizure.

Birth and developmental history: She was delivered at home by spontaneous normal vaginally, cried immediately after birth, developmental milestones were normal till two years of her age. Regular immunisations were carried out as per National Immunization Schedule. At birth her weight (2800 gms) and height were normal.

Family history: History suggestive that her elder brother had delayed developmental milestones.

On examination: Eye contact was poor, most of the times it was absent. Her motor activity was increased. There was stereotypic hand movements, repeated screaming with spitting out her saliva by folding her tongue. She also had bruxism, broad based gait, and truncal ataxia.



Diagnosis: According to the text revision of the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR),[3] the case is diagnosed as Rett's syndrome.

Differential diagnosis: The condition differs from autism in its unusual pattern of developmental loss and characteristic clinical features. The potential for incorrect diagnosis of autism is greatest in the preschool years, when many patients with Rett's syndrome may have some degree of deterioration of social skills. The onset of childhood disintegrative disorder is typically later than that of Rett's syndrome, and the period of normal development is usually much more prolonged in that condition, which also usually affects males. In Asperger's syndrome, cognitive and language skills are preserved and there is not a marked loss of abilities.

Discussion

In 1966, Andreas Rett first reported cases of Rett's syndrome. His initial report noted the characteristic history and clinical findings, such as gait and truncal ataxia and apraxia, breathing difficulties, electroencephalography (EEG) abnormalities, and occasional seizures. He also observed some features suggestive of autism. The presence of some symptoms suggestive of autism, particularly in the preschool years, is the major rationale for placement of this condition in the pervasive developmental disorders' class of disorders in both DSM-IV-TR and the tenth revision of the International Staitstical Classification of Diseases and Related Health Problems (ICD-10).[4] A simple system classifies autism spectrum under developmental disorders along with mental retardation and language disorders.[5]

Prevalence of the condition ranges from one in 15,000 to one in 22,000 females. Several thousand cases are now registered with the International Rett's Syndrome Association. Rett originally speculated that the condition was associated with high peripheral ammonia levels, but this did not prove to be the case. The role of genetic factors was initially suggested by reports of the condition in monozygotic twins and, rarely, extended family members, but most cases appeared to be sporadic in nature.

In 1999, the gene MECP2 was cloned and has subsequently turned out to be responsible for the vast majority of cases. This molecule binds to methylcytosines and is known to be involved in transcriptional repression. More recent data have demonstrated a specific role for this protein in regulating glutamatergic synapse formation during development. Direct or strong genetic influences have been identified for autism.[5] Family and twin studies demonstrate that genes play a greater role in autism than any other neuropsychiatric disorders.[6]

The onset of the condition may be insidious and follow a period of developmental stagnation and delay recognition slightly. Over time, the developmental delay, decelerated head and body growth, and diminished interest in the environment become quite striking. Previously acquired abilities are lost, including purposeful hand movements. The lack of social interest and potential for misdiagnosis of autism is greatest in the preschool years, as usually by the time the child reaches school age, the autistic-like features are less prominent and development plateaus for a time.

At this point, severe mental retardation, seizures, and motor problems are areas of major concern. During this plateau, or "pseudo-stationary" phase, breathing difficulties, bruxism, motor problems, and early scoliosis may be noted. Apnoeic episodes may alternate with hyperventilation. Most children remain ambulatory until a final period of motor deterioration. EEG is frequently abnormal and seizures are common.

Four stages of Rett's syndrome have been defined to help characterise the disorder and improve its recognition and diagnosis:

Stage I: Early onset (six to 18 months of age) – affects early development first with stagnation

Stage II: Regressive/rapid deterioration stage (one to four years) – devastating cognitive and motor regression

Stage III: Relative stabilisation/plateau (two to ten years) – partial recovery

Stage IV: Late motor impairment (after ten years) – cognitive stability with motor impairment

Various nonspecific abnormalities have been reported in Rett's syndrome and include elevated levels of copper and ammonia in the blood, cortical atrophy on brain scan, EEG abnormalities. In some neuropathological studies, decreased brain weight and loss of neurons have been noted with changes in the substantia nigra and caudate nucleus.

The results of neurochemicals (e.g. of endorphins, cortisol, and dopamine) have been contradictory. As previously noted, the observation of a gene associated with Rett's syndrome in at least some cases does raise the possibility of more specific diagnostic tests. Rett's syndrome is a progressive neurodegenerative condition. As adults, patients may be non-ambulatory due to motor problems and scoliosis. There is increased risk of sudden death.

There are no specific treatments for Rett's syndrome. Special education, behaviour modification, and physical and respiratory therapies may be useful. No specific pharmacological treatments are available. Given the high rate of seizure disorder, caution is needed with use of medications that lower the seizure threshold. As with other pervasive developmental disorders, consideration should also be given to supporting the parents and siblings of affected individuals.

Though a few cases had been reported from India, our case has been the first case of Rett's syndrome reported from North East India and West Bengal. Our patient was at stage II, i.e. regressive/rapid deterioration stage (one to four years) – devastating cognitive and motor regression. Our case was similar to all the cases reported previously from India.[1,2,7-11] All the cases were presented with loss of acquired, purposeful hand skills, and expressive and receptive language following a period of normal development. Those cases were also associated with broad based gait and stereotyped hand movements which were similar to our case. All cases were associated with decrease head circumference which we found in our case also.

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