CASE REPORT

A Case Report on Rett's Syndrome

Soumitra Ghosh[1], Jyotirmoy Sarma[2], Debdutta Nayak[3], Tanveer Dabria[4]

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Abstract

Introduction: Rett's syndrome is a neurodevelopmental disorder that occurs almost exclusively in females and has a typically deteriorating course that is mostly caused by an identifiable mutation of the *MECP2* gene located in the X chromosome. The most characteristic features are loss of purposive hand movements and acquired fine motor-manipulative skills along with lack of language development, distinctive stereotyped movement, episodes of hyperventilation, etc.

Case description: Here, we are presenting a case of Rett's syndrome in a girl of 4 years 10 months who had presented to the psychiatry OPD of Tezpur Medical College and Hospital on being referred from the Department of Pediatrics. There was a history of loss of hand skill and speech with purposeless stereotypic movement of hand, as a result of which she was unable to hold objects in her hand. She had broad-based gait with teeth grinding and a history of episodic hyperventilation. To our knowledge, this has been the second case of Rett's syndrome from the eastern zone of India. The first case was reported by Ghosh S et al. from a medical college in Assam.

Conclusion: Rett's syndrome is a relatively rare disease with a deteriorating and progressing course without any specific treatment for it. Management plans should be focused on early detection and symptomatic treatment in the form of seizure control, behavior therapy, physiotherapy, etc., along with proper genetic counseling and parental counseling.

Keywords: Autism spectrum disorder, Case report, Executive cognitive functioning deficits, Genetic.

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Introduction

Rett syndrome is an extremely rare (1 in 15,000–22,000 in every female live birth) progressive neurodevelopmental disorder that almost invariably affects females. The infant normally develops up to the age of 6–18 months, and then there is a decline in cognitive skills.

The most characteristic features are the loss of purposeful hand movements and the loss of all the previously acquired skills. The accompanying symptoms include loss, partial loss, or lack of language development; characteristic stereotypical wringing or "handwashing" movements, with the arms flexed in front of the chest or chin; stereotypical salivary hand-wetting; improper food-chewing; frequent episodes of hyperventilation; nearly always an inability to gain control over the bowels and bladder; frequent excessive drooling and tongue protrusion; and a loss of social engagement. The youngsters usually maintain a sort of "social smile" in early childhood, looking at or "through" people without engaging in social engagement (though social interaction often develops later).

The stance and gait tend to become broad-based, the muscles are hypotonic, trunk movements usually become poorly coordinated, and scoliosis or kyphoscoliosis usually develops. Spinal atrophies, with severe motor disability, develop in adolescence or adulthood in about half the cases.

The *MECP2* gene, which can be found on the X chromosome, is primarily responsible for Rett syndrome.

Here, we are presenting a girl with Rett's condition, 4 years 10 months, who had presented to the Psychiatry OPD of Tezpur Medical College and Hospital on being referred from the Department of Pediatrics. There was a history of loss of hand skill and speech with purposeless stereotypic movement of hand, as a result of which she was unable to hold objects in her hand. She had a broad-based gait with teeth grinding and a history of episodic hyperventilation.

[1,2,4]Department of Psychiatry, Tezpur Medical College and Hospital, Sonitpur, Assam, India

[3]Department of Psychiatry, Silchar Medical College and Hospital, Silchar, Assam, India

Corresponding Author: Jyotirmoy Sarma, Department of Psychiatry, Tezpur Medical College and Hospital, Sonitpur, Assam, India, Phone: +91 9085579050, e-mail: jsarmajmc29@gmail.com

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This is, as far as we are aware, the second instance of Rett's syndrome in India's eastern region. The first case was reported by Ghosh S et al. from a medical college in Assam.[1]

Case Description

Miss X is a 4-year 10-month-old girl in the Psychiatry OPD of TMCH, being referred from The Department of Pediatrics, with complaints of continuous unprovoked crying for the last 3–4 days and purposeless movement of the neck with excessive salivation. According to the mother, she was delivered by cesarean section due to decreased fetal movement at the 8th month of gestation. Her birth weight was below Indian average (1.9 kg). She cried immediately after birth but had difficulty in sucking. According to the mother, she had delayed developmental milestones, when she started walking at the age of 18 months, they noticed it was a wide gait, and she had a hand-wringing movement. They also gave the history of hyperventilation of the child episodically. There was a lack of development of language and excessive salivation, and she was not able to chew food. So, she was always given smashed

food or semisolid food. There is also no bowel and bladder control present. Gait was broad-based without any vertebral deformity. The girl has been clinically provisionally diagnosed to be a case of Rett syndrome (ICD F84.2). Parental counseling has been done regarding the management and its provable outcomes have been done. For further evaluation and management, physiotherapy consultation, genetic testing, and regular follow-up have been recommended.

Discussion

Andreas Rett, an Austrian neurologist and physician, originally reported it in 1966.[2] Rett syndrome had been classified by the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) as a pervasive developmental condition prior to the identification of a genetic etiology.[3] However, DSM-5[4] states that it should be classified as an autism spectrum disorder and that this is how it should be diagnosed.

The DSM-5 changes regarding autistic disorders and the classification system used in the International Classification of Diseases, Tenth Revision (ICD-10) are incompatible. ICD-10 still includes separate designations for Rett syndrome (ICD-10).[5]

According to ICD-11,[6] Rett syndrome has been separated from autism spectrum disorder (6A02) and is now classified as LD90 disease with abnormalities of intellectual development (LD90.4 Rett syndrome).

The major distinguishing characteristics of Rett syndrome (F84.2), which has a typical start between 7 and 24 months, course, and acquired fine motor-manipulative skills, are described in the diagnostic standards of ICD-10 along with lack of language development, distinctive stereotyped movement, episode of hyperventilation, loss of social engagement with retained social smile, epileptic fits, etc.

In our situation, the patient displayed the typical signs of lost intentional hand movements and improved fine motor-manipulative abilities. This shortfall was accompanied by a loss of language development, a recognizable stereotyped tortuous "handwashing" motion with the arms flexed in front of the chest or chin, as well as a stereotypical wetness of the hands with saliva. Additionally, it was linked to episodes of hyperventilation, virtually usually the inability to regain control of one's bowels and/or bladder, excessive drooling, tongue protrusion, and loss of social engagement. The muscles are hypotonic, the gait is wide-based, and the trunk movements are disorganized. Kyphoscoliosis came into existence. All these signs and characteristics lead us to believe that this is a case of Rett's syndrome according to ICD-10 diagnostic guidelines.

Differential Diagnosis

The patient's condition differs from autism in that it has a peculiar pattern of developmental loss and distinctive clinical symptoms. Early infancy is where autism misdiagnosis occurs most frequently, whereas children with Rett's syndrome may experience some social ability decline.

Childhood disintegrative disorder typically manifests later than Rett's syndrome, and typically affects males. Also, there occurs some improvement of the previously acquired skills after they are lost.

The period of normal development in a case of autism is typically much longer. Deliberate self-injurious behavior and complex stereotypic movements are rare in Rett's syndrome unlike Autism spectrum disorder.

Language and cognitive capabilities are not significantly lost in Asperger's syndrome, instead, they are intact.

Conclusion

Rett syndrome is a condition that is rather uncommon, with a deteriorating and progressing course without any specific treatment for it. Management plans should be focused on early detection and symptomatic treatment in the form of seizure control, behavior therapy, physiotherapy, etc., along with proper genetic counseling and parental counseling. Early diagnosis is critical since Rett syndrome has a poor prognosis and a deteriorating course. To reduce the rate of progression and deterioration as well as comorbid impairments, we should concentrate on the management strategy, which includes early identification, diagnosis, and symptomatic treatment.

Given the variety of clinical aspects associated with Rett syndrome, including seizures, aberrant behavior, and other neurological symptoms, a multidisciplinary strategy encompassing pediatrics, psychiatry, neurology, and physiotherapy is a viable choice for the appropriate care of the condition.

Despite the lack of a specific treatment at this time, research is currently being done to identify the best course of action by concentrating on the genetic basis of the problem.

A recent study by Neul JL et al. published in June 2023 finds a new molecule Trofinetide for the treatment of Rett syndrome, but this study is still in phase III of RCT.[7] Another recent study by Ehinger Y et al. showed that gene therapy and X-chromosome reactivation can be newer treatment modalities of Rett syndrome.[8] More scientific studies are anticipated in the coming days to better understand the disorder.

Orcid

Jyotirmoy Sarma https://orcid.org/0009-0006-7746-2117
Debdutta Nayak https://orcid.org/0000-0001-7330-4441

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