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Case Report

RETT SYNDROME: A CASE REPORT

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ABSTRACT

Many childhood diseases are diagnosed a couple of years after birth. This article presents a similar condition resulting in regression and slowing down of brain growth after a normal course of development during neonatal period. Rett syndrome is such a condition that presents itself after some months of apparently normal development. This disease is usually manifested with deceleration of head growth, loss of purposeful hand movements which are replaced by hand wringing or hand washing stereo types. This is also followed by marked mental retardation with severe impairment in expressive and receptive language skills. The child presented in this case report had a normal development till 18 months of age after which she developed delay in gross motor development and further in other areas of development. The child is now on symptomatic treatment and presents with recurrent respiratory tract infections which require hospitalization.

INTRODUCTION

Rett syndrome (RTT) is an X-linked dominant pervasive neurodevelopmental disorder. This syndrome is not strictly speaking a degenerative disease, but a disorder of early brain development marked by a period of developmental regression and deceleration of brain growth after a relatively normal neonatal course. It occurs predominantly in girls. This syndrome was originally described in 1960 by Andreas Rett.[1,2]

Clinical case

We report the case of a 15-year-old girl admitted with complaints of fever, irritability while passing urine and breathlessness for 2 days. She is a known case of Rett Syndrome.

Child was born at term out of consanguineous marriage. Antenatal period was uneventful. But a history of prolonged labor noted. Child was born with congenital Talipus Equinovarus which was corrected before the age of one year. Her development including gross motor, fine motor and language were within the normal limits till 18 months of age. At approximately 18 months of age, the parents noted marked delay in her ability to walk. They also noted unusual hand wringing stereotyped behaviors. At the age of three years the child had her first episode of seizures. Onexamination, abnormal EEG wave patterns were noticed. Purposeful hand movements were markedly impaired. Child was then started on antiepileptic. Expressive and receptive language skills were severely impaired. The child also has scoliosis. There is a history of repeated hospitalizations due to lower respiratory tract infections.

Subsequently, the child developed feeding difficulties for which Nasogastric feeding tube was inserted. The child also presented with autistic behavior but shows love for music especially Carnatic Music.

The treatment is symptomatic and supportive. As the child has now presented with symptoms of Lower respiratory tract infection, she is now managed with antibiotics, bronchodialators, mucolytic and antipyretics.

DISCUSSION

Sporadic mutations in the gene methyl CPG-binding domain protein 2 (MECP2) located on the X chromosome contributes to majority of the RETT syndrome cases. Girls are the most affected ones and male fetus diagnosed with RETT syndrome wouldn't survive to term. Recently, MECP2 mutations have also been identified in some mentally retarded males. Methyl – CPG-binding domain protein 2 is found near the end of the long arm of the X chromosome at Xq28.

Jeffrey et al revised the diagnostic criteria for RTT in 2010 and is as follows;

RTT Diagnostic Criteria 2010

Consider diagnosis when postnatal deceleration of head growth observed.

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Required for typical or classic RTT

1. A period of regression followed by recovery or stabilization*
2. All main criteria and all exclusion criteria
3. Supportive criteria are not required, although often present in typical RTT

Required for atypical or variant RTT

1. A period of regression followed by recovery or stabilization*
2. At least 2 out of the 4 main criteria
3. 5 out of 11 supportive criteria

Main Criteria

1. Partial or complete loss of acquired purposeful hand skills.
2. Partial or complete loss of acquired spoken language**
3. Gait abnormalities: Impaired (dyspraxic) or absence of ability.
4. Stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and washing/rubbing automatisms

Exclusion Criteria for typical RTT

1. Brain injury secondary to trauma (peri- or postnatally), neurometabolic disease, or severe infection that causes neurological problems***
2. Grossly abnormal psychomotor development in first 6 months of life#

Supportive Criteria for atypical RTT##

1. Breathing disturbances when awake
2. Bruxism when awake
3. Impaired sleep pattern
4. Abnormal muscle tone
5. Peripheral vasomotor disturbances
6. Scoliosis/kyphosis
7. Growth retardation
8. Small cold hands and feet
9. Inappropriate laughing/screaming spells
10. Diminished response to pain
11. Intense eye communication - "eye pointing"

Since MECP2 mutations have recently been spotted in many individuals prior to any clear evidence of regression, those individuals under 3 years' old who have not lost any skills but otherwise have clinical features suggestive of RTT are also attributed to have diagnosis of "possible' RTT. It is ideal to have these individuals reassessed every 6-12 months for evidence of regression. If regression manifests, the diagnosis should then be changed to definite RTT. However, if the child does not show any evidence of regression by 5 years, the diagnosis of RTT should be questioned.

**Loss of acquired language is based on best acquired spoken language skill, not strictly on the acquisition of distinct words or higher language skills. Thus, an individual who had learned to babble but then loses this ability is considered to have a loss of acquired language.

***There should be clear evidence (neurological or ophthalmological examination and MRI/CT) that the presumed insult directly resulted in neurological dysfunction.

#Grossly abnormal to the point that normal milestones (acquiring head control, swallowing, developing social smile) are not met. Mild generalized hypotonia or other previously reported subtle developmental alterations during the first six months of life is common in RTT and do not constitute an exclusionary criterion.

##If an individual has or ever had a clinical feature listed it is counted as a supportive criterion. Many of these features have an age dependency, manifesting and becoming more predominant at certainages. Therefore, the diagnosis of atypical RTT may be easier for older individuals than for younger. In the case of a younger individual (under 5 years old) who has a period of regression and ≥2 main criteria but does not fulfill the requirement of 5/11 supportive criteria, the diagnosis of "probably atypical RTT" may be given. Individuals who fall into this category should be reassessed as they age and the diagnosis revised accordingly.[3]

Management

Management is mostly symptomatic and supportive**.** Major problems include motor function, environmental contact, feeding and irritability. Music therapy decreases hand wringing and apparently improves orientation and contact. Seizure control is usually by carbamazepine, which is the treatment of choice because of minimal side effects, particularly in younger children.[4]

Nursing Management

Nursing management of this girl child with Rett syndrome is discussed below using the nursing process approach;

Delayed child development related to neurodevelopmental disorder

Expected outcome

- Achieve age-appropriate growth, behaviors, and skills to the fullest extent possible.

Nursing Interventions

- Reassess developmental levels at intervals appropriate for illness
- Explore family's feelings regarding child's health conditions and required treatments
- Provide positive encouragement to the child while doing her activities
- Encourage age-related play and other activities especially music to strengthen the development possible

Risk for injury related to seizure activity

Expected outcome

- Child will be protected and free from injury

Nursing interventions

- Explain to the parents about the different stimuli that can trigger a seizure activity
- Provide a safe environment for the child
- Institute seizure precautions such as keeping padded side rails up with the bed in its lowest position
- Administer antiepileptic as prescribed
- Coordinate with a physiotherapist for strengthening exercises

Risk for aspiration related to feeding difficulties

Expected outcome

- Child will be protected from aspiration

Nursing Interventions

- Periodically review results of swallowing studies as ordered
- Auscultate bowel sounds to assess for gastric motility
- Keep head of bed elevated when feeding and at least for one hour after feeding
- Provide food with consistency that the child can swallow
- Keep suction machine available when feeding the child and educate mother about the use of suction machine
- Eliminate distracting stimuli when feeding the child

Interrupted family process related to a critically ill child in the family

Expected outcome

- Family will identify internal and external resources to deal with the situation

Nursing Interventions

- Encourage family members' expression of concerns about seriousness of condition, possibility of death, or incapacitation of the child
- Listen for expressions of hopelessness and helplessness
- Support family grieving for the critically ill child. Acknowledge normality of wide range of feelings and ongoing nature of process.
- Stress importance of continuous open dialogues among family members
- Identify individual roles and anticipated and perceived role changes

CONCLUSION

Children with Rett syndrome often presents with normal growth and development during infancy and early childhood. After this they develop a period of regression which results in a global developmental delay. It is noted that majority of the children who present with Rett syndrome are females. Careful and meticulous care by the primary caregiver is very important for improving the quality of life of these children presenting with Rett syndrome.

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