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Case report on Cerebrotropic Hyperammonia with Autism Spectrum Disorder (RETT)

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ABSTRACT:

Background: Rett syndrome is a rare genetic neurological and developmental disorder that causes progressive mobility and speech difficulties as the brain grows abnormally. The disease has the greatest impact on young women. Rett syndrome (RTT) is a neurodevelopmental disorder characterised by a delay of spoken language and the development of discrete hand stereotypies. It affects about one in every 10,000 live female births. Symptoms such as loss of learnt intentional hand skills, autistic behaviours, motor dysfunctions, seizure disorders, and gait abnormalities have all been used to make clinical diagnosis.

Diagnostic Evaluation: A physical exam and thorough information about the child's growth and medical history are used to identify the conditions. A significant component of a Rett syndrome diagnosis is the lack of normal hand function. Speech is being suppressed.

Therapeutic Intervention: Tab. Carbamazepine 100mg BD, Tab Clonazepam 0.5 mg Sos, Tab phenobarbital 5mg OD, Tab topiramate 25mg TDS.

Outcome: A physical exam and thorough information about the child's growth and medical history are used to identify the condition. A significant component of a Rett syndrome diagnosis is the lack of normal hand function. Speech is vanishing.

Conclusion: The patient has been admitted to AVBRH's Pediatric department, where she will receive ongoing medical and nursing treatment. His health is stable and being monitored.

Keywords: CT Scan, USG, MRI Autism spectrum disorder, Rett , Fever

INTRODUCTION:

Rett syndrome is commonly identified in the first two years of life, and the news can be distressing for a kid.(1) Although there is no cure for Rett syndrome, girls and their families can benefit from early discovery and treatment. It was previously thought to be on the Autism Spectrum. is a female specific

genetic disease that typically manifests between the ages of 6 and 18 months. Repetitive movements, as well as language and coordination issues, are common signs. (2) Movement inconsistencies in addition to the basic movement abnormalities identified in RTT - hand stereotypies and gait dyspraxia - affected individuals have a wide spectrum of movement difficulties. The majority of people who are affected are hypotonic at birth and in their early adult years, but their ankles and lower extremities develop dystonia. The orometer and choreiform actions of the limbs. (3)

The majority of RTT cases have a genetic cause, which has led to the development of numerous RTT mice models. These models have given researchers insight into the disorder's origins as well as future treatment approaches. Furthermore, the animal model has shown that the sickness can be reversed, implying that there is hope for the future. (4) RTT is mostly a female disease since the gene that causes the majority of instances, MECP2, is situated on the X chromosome. 3 RTT is usually caused by interrupting one copy of MECP2. The disease reveals itself as a regression with a loss of hand skills and spoken language, as well as the onset of characteristic repetitive hand movements, after a period of normal growth. (5)

The majority of Rett cases have a genetic cause, which has led to the development of numerous RTT mice models. 7 to 12 years old These models have shed light on the disorder's aetiology and pointed to prospective treatment options. Furthermore, the animal model has shown that the sickness can be reversed13, implying that there is hope for the future.(6) Rett is mostly a female disease since the gene that causes the majority of instances, MECP2, is situated on the X chromosome. A rare inherited condition that inhibits female brain development. The Rett syndrome is rarely inherited, despite the fact that it is caused by a gene development Rett is a genetic disease that affects young females between the ages of six and eighteen months. Language and cognitive difficulties are common symptoms. Following a time of normal development, is a severe neurodevelopmental disorder characterised by increasing loss of intellectual functioning, fine and gross motor skills, and communicative abilities, as well as slowed head growth and the development of stereotypic hand movements. Seizures, a disrupted respiratory pattern with hyperventilation and occasional apnea, scoliosis, growth retardation, and gait apraxia are common in girls with RTT (Hagberg et al., 1983).

CLINICAL FINDINGS :

Doctors looked for indications and symptoms in a child's early growth as well as conducting routine physical and neurological testing to diagnose Rett syndrome. RTT is usually caused by interrupting one copy of MECP2. After a period of normal growth, the disease manifests itself as a regression with a loss of hand skills and spoken language, as well as the onset of odd repetitive hand gestures.(7)

STAGES OF AUTISM -

Level 1: Assistance is required.

Level 2: Requires a great deal of help.

Level 3: Completely reliant on others.

Limitations of ASD Levels

CAUSES : An X chromosomal mutation is found in the majority of children with Rett syndrome. It's unclear what this gene does or how Rett syndrome is caused by its mutation. Many additional genes involved in development are thought to be influenced by this one gene, according to researchers., having an autistic member of one's own family mutations in the human genome .

- Fragile X syndrome is another genetic disorder.
- being the son or daughter of old parents
- The weight of the baby during birth is low.
- Metabolic irregularities
- Virus infections have occurred in the past as a result of heavy metal and toxin exposure in the environment

SIGN AND SYMPTOMS:

- Screaming fits are common.
- uncontrollable sobbing
- avoiding direct eye contact.
- sleep regression • sensory issues
- Inappropriate body posture or facial expressions
- A voice with a peculiar timbre.
- Poor eye contact or avoidance of eye contact
- Disturbances in behaviour.
- Deficits in Language Comprehension
- outbursts of rage; inconsolable weeping; avoidance of eye contact
- difficulties with the senses
- Regression in sleep
- Abnormal body posture or facial expressions.• A voice with an unusual tone.
- Flaccid muscles, difficulty to connect muscle motions, muscle weakness, coordination issues, tight muscles, or rhythmic muscle contractions are all examples of muscular disorders.
- Breathing disorders include no-breathing episodes, fast breathing, and shallow breathing.
- Developmental: failure to thrive or delayed development
- Irritability or repeated movements are examples of behavioural symptoms.
- Cognitive: inability to communicate or understand words, as well as sluggishness of movement
- Seizures, constipation, drooling, scoliosis, teeth grinding, and tremor are also prevalent.

PRESENT HISTORY :

In June 2021, a 7-year-old girl patient was hospitalized to AVBRH with Fits, nausea, and vomiting, and his family said that he was unable to walk because to her Rett Syndrome. Cerebrotropic Hyperammonia and Autism Spectrum Disorder have been the subject of several studies.

Family history:
The family consists of four members. (Rett) Cerebrotropic Hyperammonia with Autism Spectrum Disorder was discovered in my patient. Except for my patient, who was admitted to the hospital, none of the other members had any health issues.

Past history:

The patient has no major medical history.

CLINICAL FINDING :

Social interaction and communication difficulties, as well as repetitive or stereotyped behaviour patterns and typically limited interests, characterize this neurodevelopmental condition.

ETIOLOGY:

The majority of instances of Rett syndrome are caused by a single gene mutation (also known as a mutation).According to studies funded by the National Institute of Child Health and Human Development, the majority of instances of classic Rett syndrome are caused by a mutation in the Methyl cytosine-binding protein 2 (MECP2) gene (NICHD). MECP2 is a gene found on the X chromosome. Despite the fact that there is no known cause for autism spectrum disease, it is usually assumed that abnormalities in brain structure or function are to blame.

According to brain scans, autistic children's brains have distinct forms and architectures than neurotypical children.

Physical Examination: My patient has not able to walk and edema, parotoid swelling, and soreness in the mid arm area

Diagnostic assessment : HB-6.7%,total protein-6.9, total platlet count-15, 000

Therapeutic Intervention –

Inj. Emset: 1.8mg TDS, Tab. Allopuraznol 300mg BD Inj. Pantop 12mg BD, Inj. Ceftraxon 20ml
NS 1 mg BD, Inj. Pantop 12mg BD, Inj. Pantop 12mg BD, Inj. Pantop 12mg BD,

DISCUSSION:

On June 5, 2021, a 7-year-old girl patient was admitted to AVBRH's paediatric unit with complaints of pallor and edema and was diagnosed with Rett syndrome. Rett syndrome is one of the most common causes of complex impairment in women. Early neurological regression is characterised by major impairments in motor, cognitive, and communication skills, as well as autonomic dysfunction and, in some

cases, seizure disorder. This X-linked dominant neurodevelopmental condition is caused by a mutation in the MECP2 gene, which codes for the methyl-CpG-binding protein MeCP2.

Several mouse models based on conditional knockout of the Mecp2 gene or a truncating mutation have been developed. We examine clinical features, with a focus on behavioural phenotypes, and explore current clinical management strategies as well as future perspectives on gene therapy.

ASD is a multifaceted developmental disorder characterised by persistent difficulties in social interaction, speech, and nonverbal communication, as well as confined and repetitive activities. Each person's ASD experience and the severity of their symptoms are different. In 2013, the term "autism spectrum disorder" was coined to define as persistent difficulties in social communication and engagement, as well as limited and repetitive patterns of behavior, interests, or hobbies, characterised this developmental disease. A number of related studies were reported[8-12].

The severity of each person's ASD symptoms and their experience with the disorder are unique. The term "autism spectrum disorder" was first coined in 2013 to describe a developmental disorder marked by chronic difficulties in social communication and interaction, as well as limited and repetitive patterns of behavior, interests, or hobbies. Each person's ASD symptoms are different, as is their experience with the illness. RTT has been found in a small percentage of guys, although it is much more common in girls. Movement inconsistencies in addition to the basic movement abnormalities identified in RTT - hand stereotypies and gait dyspraxia - affected individuals have a wide spectrum of movement difficulties. The majority of persons affected are hypotonic at birth and in early adulthood, but their ankles and lower extremities develop dystonia. The limbs' orometer and choreiform motions.

RTT is mostly a female disease since the gene that causes the majority of instances, MECP2, is situated on the X chromosome. RTT is usually caused by interrupting one copy of MECP2.After a period of normal growth, the condition manifests itself as a regression with a loss of hand skills and spoken language, as well as the commencement of characteristic repetitive hand movements.

Behavioral difficulties include inappropriate social interaction, poor eye contact, compulsive behavior, impulsivity, repetitive motions, self-harm, or repeated words or behaviors. Insensitivity to others' feelings or despair are psychological issues.

CONCLUSION:

The patient has been admitted to AVBRH's paediatric department, where he will receive continued medical neurology and nursing treatment, and her condition is stable and being monitored.

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