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RETT SYNDROME : A CASE REPORT

Paediatrics

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ABSTRACT

Rett syndrome is a neurodevelopmental disorder that should be considered in a child who demonstrates regression in previously acquired skills after a period of normal development. Here, we report a case of a female child with history of gross motor and speech regression with generalized tonic-clonic convulsions. On examination ,microcephaly , stereotypic hand movement, intense eye gaze , teeth grinding and drooling of saliva with diminished deep tendon reflexes and hypertonia was present. Hence,on the basis of these features and investigatons, we have diagnosed the patient as Rett syndrome.

KEYWORDS

Rett syndrome, neurodevelopmental, regression, stereotypic hand movement

INTRODUCTION

Rett syndrome is a neurodevelopmental X-linked dominant disorder in which regression of previously acquired skills follows a period of typical development. It is one of the most frequent causes of mental disability in females, with an incidence of 1 in 10,000 to 15, 000. Rett syndrome can present with a multitude of symptoms including but not limited to a deceleration in head growth, gait abnormalities, loss of purposeful hand movements often replaced with repetitive stereotypical movement (hand-wringing), loss of speech and breathing abnormalities. Rett syndrome is associated with a complex phenotype and has been classified into typical, atypical, and variant presentations. Approximately 90% of reported cases have mutations of the methyl-CpG-binding protein 2 (*MECP2*) gene.

Some atypical cases may result from mutations in cyclin-dependent kinase-like 5 (*CDKL5*). Mutations in *MECP2* have been associated with impacting the development of neurons and axodendritic connections. Jellinger and Seitelberger (1986) were the first neuropathologists to identify and describe the pathology behind it. They found that the brain in patients of rett syndome weighed less and the neurons of the substantia nigra pars compacta contained less melanin in comparison to the age-matched controls. Although it was thought to be exclusive to females, males with the phenotype and *MECP2* mutations are now being defined.(1)(2)(3)

CASE REPORT

Here, we report a case of 7 year old female child , who was brought with complaints of inability to walk and speak. Child had normal development till 8 months of age, was able to stand with support. There is history of speech regression. From 3 years of age, child is having generalized tonic-clonic type of convulsions. History of loss of previously acquired milestone present. Birth history was normal.

On examination, microcephaly with stunting and wasting was present with bilateral ankle contracture. Stereotypic hand movement and intense eye gaze was present. Teeth grinding and drooling of saliva was seen. Hypertonia seen in bilateral upper and lower limb. Deep tendon reflexes were diminished in both upper and lower limb except right knee and ankle , which were absent. Bilateral plantars were flexor.

Ophthalmology and hearing evaluation was within normal limits. Metabolic workup was normal. EEG was done, suggestive of right sided epilepsy. MRI brain was normal. Patient was started on oral valproic acid. Occupational therapy for oromotor insuffiency and fine motor movements, and limb physiotherapy was started.

So, on the basis of history and examination with investigations , we have diagnosed the patient as Rett syndrome.

DISCUSSION

Rett syndrome is a neurodevelopmental disorder, which means that the course of the disorder changes over time when motor and cognitive development should be progressing. These changes can be evidenced by a sequence of "stages" which encapsulate specific changes in the girls as the disorder progresses(4). Girls with classical rett syndrome have an apparently normal period of early development, and at about 6-18 months of life, the early signs of the disorder start to emerge. Stage I is the early-onset stagnation period and appears between 6 and 18 months of age. Features include a delay or stagnation in development. This is followed by Stage II, the rapid-developmental regression period between 1 and 4 years of age when they lose acquired skills such as communication skills, including language and socialisation, as well as losing some fine and gross motor skills. In addition, deceleration of head growth may be noted . It is during this time that stereotypic hand movements become apparent . Stage III is called as pseudostationary stage. In this post-regression stage, where the phenotype stabilizes, they develop an intense eye gaze and increased social awareness. They may also partially regain some of the skills lost during stage II. The last stage, stage IV, is known as the late motor deterioration stage and can last for years or decades. This stage is characterised by reduced mobility, muscle weakness, rigidity and spasticity with the development of dystonia and hand and foot deformities as they grow older. Walking may cease but eye gaze usually improves, repetitive hand movements may decrease and cognition, communication or hand skills generally do not decline.

However, the spectrum is broad, with girls developing comorbidities at different stages which include seizures, breathing disturbances with hyperventilation and/or apnoeas, gastro-intestinal complications, gait disturbances and scoliosis, which in combination make the management very complex.

Among the most characteristic features is the deceleration of head growth, which is frequently seen between 6 and 24 months of age and occurs in 80% . (5)Most patient typically start walking in their early years, albeit with an abnormal gait which is often described as dyspraxic. One third of this,lose this skill as the disorder progresses . Between 60-80% develop seizures, usually towards the end of the regression period or post-regression, which in some can be difficult to control .(6)Some have gastrointestinal problems including gastro-oesophageal reflux, air swallowing with abdominal distension, and

chronic constipation. Some experience abdominal pain occasionally due to gallbladder disease . The lack of oral motor control frequently results in feeding difficulties, and poor weight gain, which may lead to nutritional deficiencies that require close monitoring and in some gastrostomy tube placement is required to maintain body weight and general health . Bone health is a major concern as most patient develop a scoliosis. Approximately 10% with a scoliosis require surgical intervention.

Overall survival and quality of life are improving with the development of guidelines for the management of specific comorbidities. Behavioural abnormalities have long been recognised as a fundamental feature , particularly autistic behaviour which arises during the period of regression and can persist into the post-regression period.

CONCLUSION

Rett syndrome is X-linked dominant neurodevelopmental disorder with incidence of 1 in 10,000 to 15,000. On the basis of history and examination, we diagnosed the patient as Rett syndrome. Currently , patient is on syrup valproic acid and occupational therapy and physiotherapy is being continued. She is currently between stage III/IV. Supportive and symptomatic treatment is required for Rett syndrome.

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