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RETT SYNDROME - A CASE REPORT

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ABSTRACT

Rett syndrome is a condition of unknown cause, so far reported only in girls, which has been differentiated on the basis of a characteristic onset, course, and pattern of symptomatology. Typically, apparently normal or near-normal early development is followed by partial or complete loss of acquired hand skills and of speech, together with deceleration in head growth, usually with an onset between 7 and 24 months of age. A case of 3 year old girl with Rett syndrome is presented here. This girl had normal development till the age of two and half years. However, gradually over the next few months, she developed features suggestive of Rett Syndrome.

Keywords: Rett syndrome, pervasive developmental disorder, neurodevelopmental disorder

INTRODUCTION

Rett syndrome is not actually a degenerative disease, but a disorder of early brain development[1]. It is characterized by normal development till 6 months to 48 months of age followed by gradual loss of purposeful hand movements and development of characteristic, stereotypical hand movements; loss of previously acquired speech; psychomotor retardation; ataxia; deceleration of head circumference and autistic symptoms[3]. It occurs predominantly in girls. The frequency is 1/ 15,000-1/22,000[1].We report a girl child who presented with regression of language milestones and a diagnosis of Rett syndrome was considered.

CASE REPORT

A 3 year old female child born out of second degree consanguineous marriage to a primigravida mother at 28 weeks of gestation with birth weight of 1.3Kg presented to us with complaints of decreased communication with parents since last 4-5 months. This started after an episode of generalized tonic clonic seizures 5 months back. Child is on oral Phenobarbitone and no h/o further seizures after that. Child at present was able to stand with support and palmar grasp was present. Prior to the episode of GTCS, child was able to speak 2-3 words sentences with meaning which she had attained at around two and half years of age. Now the child was able to speak only some random irrelevant words.

The clinical examination revealed continuous purposeless movements of both upper limbs. Weight and height were below the 3rd centile and microcephaly was present. Child had convergent squint with hypertonia of both upper limbs and lower limbs with exaggerated deep tendon reflexes. Other systems were within normal limits.

Ophthalmologic evaluation revealed right esotropia with normal fundus examination. Audiological evaluation was normal.

MRI: There is mild reduction in cerebral and cerebellar volume with mildly prominent sulci and few deep gyrations. Bilateral lateral and third ventricles are mildly prominent with subtle undulated margins.

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DISCUSSION

Rett syndrome is a disorder of the nervous system that leads to developmental reversals, especially in the areas of expressive language and hand use[3]. Rett syndrome occurs almost exclusively in girls. Studies have linked many Rett syndrome cases to a defect in the methl-CpG-binding protein 2 (MeCP2) gene a transcription factor that binds to methylated CpG islands and silences transcription[3]. This gene is on the X chromosome. Females have two X chromosomes, so even when one has this significant defect, the other X chromosome is normal enough for the child to survive. Males born with this defective gene do not have a second X chromosome to make up for the problem[3]. Therefore, the defect usually results in miscarriage, stillbirth, or very early death[2]. Development may proceed normally until 6-18 months age, when regression of language and motor milestones and acquired microcephaly becomes apparent. An ataxic gait or fine tremor of hand movements is an early neurologic finding[3].

Diagnostic criteria for Rett syndrome were developed by representatives of the International Rett Syndrome Association and the Center for Disease Control[3].

Necessary criteria include normal prenatal and perinatal period; normal psychomotor development through the first 6 months of life; normal head circumference at birth, with subsequent deceleration of head growth; loss of purposeful hand skills; severely impaired expressive and receptive language; apparent severe mental retardation; and gait apraxia and truncal apraxia/ataxia[3].

Supportive criteria include breathing dysfunction, seizures, spasticity, scoliosis, and growth retardation. The diagnosis of RS is considered tentative until 2 to 5 years of age[3].

The differential diagnosis includes other disorders associated with mental retardation, cerebral palsy, and seizure disorders.

Symptoms may include apraxia, excessive salivation, intellectual disabilities, generalised tonic clonic seizures, loss of purposeful hand movements and severe language development problems[3].

The hallmark of Rett syndrome is repetitive hand-wringing movements and a loss of purposeful and spontaneous use of hands; these features may not appear until 2-3 years age[1]. Autistic behavior is a typical finding in all patients. After the initial period of neuroregression, the disease process appears to plateau, with persistence of the autistic behavior[1].

Four stages of Rett's syndrome have been defined to help characterize the disorder and improve its recognition and diagnosis[2]. These stages may be described as follows:

- Stage I: Early onset (6–18 months of age) – affects early development first with stagnation

- Stage II: Regressive/rapid deterioration stage (1–4years) – devastating cognitive and motor regression

- Stage III: Relative stabilization/ plateau (2–10 years) – partial recovery

- Stage IV: Late motor impairment (after 10 years) – cognitive stability with motor impairment

Rett syndrome may be classified as atypical, classical (meets the diagnostic criteria) or provisional (some symptoms appear between ages 1 and 3).[3] Rett syndrome is classified as atypical if it begins early (soon after birth) or late (beyond 18 months of age, sometimes as late as 3 or 4 years old); if speech and hand skill problems are mild or if is appears in a boy (very rare).[3]

Treatment may include assistance with feeding and diapering and physical therapy to help prevent the hands from contracting. Medications such as carbamazepine may be used to treat seizures. Other medications or supplements that have been used or studied include Bromocriptine, Dextromethorphan, folate, L-carnitine and L-dopa[3].

The disease slowly progresses until the patient is a teenager. Then, symptoms may improve. Developmental regression or delays vary. The average life expectancy of a girl with Rett syndrome may be mid-40s. Death is often related to seizure, aspiration pneumonia, malnutrition, and accidents.

CONCLUSION

In conclusion, it is important for clinicians to be aware of this disorder because increased identification will help in greater understanding of this disorder and proper guidance will help the patient and family, and reduce the burden of care on the parents. Thus, it is suggested that all female children presenting with low intelligence and autistic symptoms should be suspected of having Rett syndrome until proved otherwise.

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