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REGRESSING DEVELOPMENTAL MILE STONES IN A 3 AND HALF YEAR OLD CHILD: A CASE REPORT

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ABSTRACT
Rett syndrome was described by Professor Andraes Rett in Vienna in 1966 as cerebral atrophy and hyperammonemia. Rett Syndrome is a chromosome X-linked genetic pediatric neurological development disorder characterized by developmental regression, particularly in relation to expressive language and use of the hands. It is also associated with low intelligence and almost exclusively affects females. It is seen mostly after 2-3 years of life. Early diagnosis of such disorders is extremely important along with treatment of patients' problems with love and care to prevent them from further pain and stress. In this case report we describe a 3 year 6 months old girl who was referred from pediatric opd with history of gradual regression in developmental mile stones and learned behaviors like verbal communication (the child could speak in short sentences which regressed to monosyllables and at times incoherent sounds) and use of hands(she could use hands to hold things properly and eat by self without much spillage which was regressed gradually, needing help in feeding and inability to hold any objects without external support. She also had regression in bowel and bladder control. She had repetitive stereotypical wringing and to and fro hand movements with continuous wetting of hands to the extent that she started developing lesions on the dorsum of her right hand. Parents were distressed about the regressing mile stones and behavioral difficulties when they were seen in the Psychiatry OPD with the child.

KEYWORDS: Rett's syndrome, pervasive developmental disorder, developmental disability, learning disability, speech regression, growth retardation.

INTRODUCTION
- Rett's syndrome was first described by Professor Dr. Andraes Rett in Vienna in 1966 as cerebral atrophy and hyperammonemia.
- Rett's Syndrome is a chromosome X-linked genetic pediatric neurological developmental disorder characterized by severe neurodevelopmental regression, particularly in relation to expressive language and loss of muscle tone, low intelligence and almost exclusively affects females.
- Usually born at term after an uneventful pregnancy and delivery, the symptoms are seen mostly after 2-3 years of life although it can be seen after 6 months period of normal development.
- Early diagnosis and treatment is extremely important to prevent them from further pain and stress.

Review of Literature
- First described as a clinical entity in the German article in 1966.[1]

Hagberg and colleagues increased awareness of the disorder in the English literature in 1983 with a further description of the condition in 35 girls with strikingly similar clinical features of ''progressive autism, inability to perform motor functions (eg. loss of purposeful hand use, difficulty in walking), head and brain growth defect, loss of intellectual functioning and communicative abilities occurred after a period of normal development.[2]

Etiology
- Caused by MECP2 gene mutation on the child's X chromosome(Xq28) in majority of cases.[3] Atypical cases may result from mutations in CDKL5, particularly the early onset seizure variant.

Stages
- There are four stages used to describe the symptoms of Rett's syndrome. Stage 1. Earlyonset, Stage 2. Developmental regression or rapid destructive, Stage 3. Pseudostationary stage and Stage 4. Late motor deterioration stage.
- Exclusion Criteria: if there is evidence of a storage disorder, retinopathy, cataract, or optic atrophy, an identifiable metabolic or neurodegenerative

disorder, an acquired neurological disorder, or evidence of peri natal or post natal brain injury. RS may occur coincidently with other disorders.[4,5]

Diagnosis

- Disorders associated with intellectual disability, cerebral palsy, and epilepsy. These diagnostic criteria for RS should encourage the development of reliable communication among investigators and enhance the epidemiological and clinical research of this important disorder.[6]

CASE

- In this case report we describe a 3 year 6 months old girl who was referred from pediatric OPD with history of gradual regression in developmental mile stones and learned behaviors like verbal communication (the child could speak in short sentences which regressed to monosyllables and at times incoherent sounds) and use of hands(she could use hands to hold things properly and eat by self without much spillage which was regressed gradually, needing help in feeding and inability to hold any objects without external support).
- She also had regression in bowel and bladder control. She had repetitive stereotypical wringing and to and fro hand movements with continuous wetting of hands to the extent that she started developing lesions on the dorsum of her right hand.
- Parents were distressed about the regressing mile stones and behavioral difficulties when they were seen in the Psychiatry OPD with the child.
- She was seen in pediatric OPD and was referred in view of the odd behaviors and regressions in mile stones.
- After coming to a provisional diagnosis the parents were psycho-educated and counseled about the nature of the illness, the further course and management. Through counseling we tried to allay the distress they had about the child's illness and behavioral issues and their queries were answered. We advised a baseline IQ assessment to monitor if any further decline. We educated them regarding the behavioral management and speech training at home. We discussed with the pediatric unit which referred the child about the diagnosis and plan of management. Patient is yet to come for follow up with the IQ report. No neuro-imaging or EEG were planned as the child had no neurological sequel. A genetic testing was advised but the parents declined it due to financial constraints.

DISCUSSION

- Diagnostic criteria were developed by representatives of the International Rett's Syndrome Association and the Centers for Disease Control. Criteria are: 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.
- Supportive criteria include breathing dysfunction, epilepsy, spasticity, scoliosis, teeth-grinding, abnormal sleep patterns, growth retardation, abnormal muscle tone, intense eye communication, inappropriate laughing or screaming and decreased response to pain. The diagnosis of RS is considered tentative until 2 to 5 years of age.[6]
- Similar presentation were found to be there in our case with impaired intellectual functions, abnormal hand movements, communication difficulties.

CONCLUSION

- This case is reported, firstly for the rarity of RS and secondly for its clinical interest to neuro-psychiatrists. Assessing children with pervasive development disorders poses interesting challenges in diagnosis and management.

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