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Rett Syndrome

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Rett syndrome is a fairly newly described disorder which consists of a devastating lifelong disability producing severe degeneration of motor skills and intellectual function in girls. We have recently seen six patients of Rett syndrome. To our knowledge Rett syndrome is being reported for the first time from India.

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Case Reports

In the year J.992 and 1993 at the Child Neurology Clinic of the All India Institute of Medical Sciences (AIIMS), New Delhi we encountered six patients who were referred with the diagnosis of cerebral palsy but actually had Rett syndrome. All the six were girls ranging from 2.5 to 6.5 years age, at initial contact. Their clinical profile is summarized in *Table I.* All patients were born after nonconsanguinous marriages with no identifiable antenatal or perinatal insults. The birth weight was normal in 3 and unrecorded in others and they were reported normal till 15 to 24 months age. The complaints included poor mental progress, loss of attained hand skills, and stereotypic hand movements which included hand wringing in 4, mouthing in 1 and inappropriate clapping in 1. In all patients, the movements disappeared in sleep. The older subjects (cases 3, 4, 6) could no longer hold a pencil or turn pages of a book skillfully.

Gait was clumsy and wide based in

TABLE I—*Clinical Profile*

S. No.	Age (mo)	Development normal till (mo)	Head circum-ference (cm)	Altered movement	Loss of psychomotor skills	Shakiness of torso	Cognitive regression
1.	30	18	45	+	-	-	+
2.	36	24	43	+	-	+	+
3.	40	18	48	+	+	-	+
4.	78	20-24	45	+	+	-	+
5.	42	18-20	48	+	-	+	+
6.	48	15	46	+	+	+	+

three, with slow deterioration. The unsteadiness was not associated with any cerebellar, posterior column signs or nystagmus. Behavior pattern included lack of responsiveness, indifference to parents affection or punishment and were reported as "disinterested". Speech was unintelligible in all, though attainment of bisyllables and short phrases in infancy were normal in three. Sleep disturbances were reported in two patients and included getting up[1] at inappropriate hours, playing on their own and going off to sleep. Teeth grinding was observed in 3 patients and hyperventilation episodes in one.

Early milestones were attained normally by all the six, but by 24 months all revealed symptoms of developmental regression. Seizures observed in 2 patients (cases 4 and 5), were generalized tonic-clonic and were well controlled on carbamazepine for over 12 months follow up.

On examination, there was mild increased tone in two: muscle power and deep tendon reflexes were normal and plantar response was flexor. Weight and height were below 50th percentile in 3, only weight was below 50th percentile in 2 and the head circumference ranged from 43-50 cm (*Table I).* Previous records of anthropometry available in 2 patients revealed a normal head circumference, weight and height till 18 and 30 months age (cases 4 and 5). No dysmorphisms or malformations were present. Formal IQ testing was not possible. Investigations performed included CT scan in 2 (both were normal) and EEC? in 1 (which revealed evidence of generalized seizure disorder).

The subjects were misdiagnosed as cerebral palsy but there were subtle differences. The observation of regression after normal early developmental period associated with loss of purposeful hand activity, stereotypic movements and shakiness of torso clubbed with cognitive regression. The course was also characterized by autistic behavior and sleep disturbances. The cessation of head growth, available in only two of our subjects, was remarkable.

Discussion

Rett syndrome, originally reported to affect only girls, was described by Andreas Rett in Vienna as "Cerebral atrophy associated with hyperammonemia". The latter attribute is no longer considered an essen-

tial part of the syndrome. The clinical features have been described as similar to those of a uniform and progressive encephalopathy (1-3). No metabolic or genetic marker has been discovered yet; hence the diagnosis is entirely clinical.

The definitive clinical requirements for the diagnosis are an apparently normal prenatal and perinatal period with normal head circumference and an apparently normal psychomotor development during the first six months of life. All the six patients were reported normal till 15-24 months age. This is followed by deceleration of head growth associated eventually with severe dementia, loss of purposeful use of the hands, autism and severe psychomotor regardation. Head deceleration was identifiable in only 2 patients who had past records of anthropometric data. Stereotypic hand movements, gait apraxia and jerky, akinetic truncal movements develop from 1-4 years of age. These were observed in all our patients; we recognized patients between 2.5 to 6.5 years only. Stereotyped movements and gait disturbances are seen in all patients. Bruxism, oculogyric crises, parkinsonism and dystonia are also.common while myoclonus and choreoathetoid movements are rarely seen. Bruxism was present in 3 of our 6 patients. Fitzgerald *et al.* reported that hyperkinetic movement disorders tended to dominate in younger patients, and bradykinesia in older girls, suggestive evidence of age-related neurodegenerative changes in the basal ganglia(4).

Other supportive symptoms include breathing dysfunction(5), EEG abnormalities^), seizures (70-80%), spasticity with muscle wasting, peripheral vasomotor disturbances, scoliosis (83% over 15 years), growth retardation and hypotrophic feet. We observed hyperventilation in only patient. Because of lack of availability of norms, small feet cannot be commented upon in our subjects. Additional accompanying symptoms include hirsutism, pigmentary dysplasia, gastrointestinal problems, disturbances in sleep with night screaming, constipation, bruxism, impaired responsiveness to pain and susceptibility to infection(l-3). By adolescence, approximately 75% of patients are in wheelchairs or bedridden(7). Most patients are below average for growth and development although puberty occurs at normal ages.

Breathing impairment is a major feature of-this syndrome. The common patterns are periods of both apnea and hyperventilation. We observed episodic hyperventilation in only one patient. It has been suggested that the periods of apnea may contribute to the brain damage because oxygen saturation levels as low as 50% have been noted(5).

Most diagnosed patients till date are female, suggesting that the syndrome is genetic. Recently, there have been reports of a syndrome very similar to Rett syndrome in boys, including the typical movement disorder and regression of mental function(8-10). Recent studies point to two sites for the abnormal gene—a distal Xp21 or proximal Xp22 site. However, the exact gene is yet to be localized.

EEG studies of awake Rett patients have indicated that there is a progressive slowing of the normal activity with appearance of sporadic bursts of epileptiform activity, atypically localized(6). A dysrythmic pattern is observed during wakefulness, during drowsiness and light sleep with bilateral bursts of spike or multispike and wave activity(ll). The EEG changes may

be seen as early as 18 months and decline with age. They tend to correlate with clinical staging, but are not specific to this syndrome.

Pathological findings correlate to the duration and severity. There is diffuse cortical atrophy and acquired microcephaly. Diffuse cortical atrophy mainly in the frontal region is detectable on CT scan. Only 2 patients(l,5) had a CT scan, which did not reveal any significant atrophy. On perfusion study and PET scanning, abnormal/ reduced regional cerebral blood flow and impaired oxidative metabolism has been found.

Biochemical analysis of the cerebrospinal fluid has revealed low levels of the central neurotransmitters-serotonin, dopamine and norepinephrine, and elevated beta-endorphin levels. The diagnostic value of CSF beta endorphin assay and an increased cerebrospinal fluid/plasma beta endorphin ratio is yet to be confirmed. Deficiency of ornithine transcarbamoyl transferase has been postulated to have a role in the etiology.

Therapy is currently only 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.

Despite good appetite, Rett syndrome patients usually have reduced muscle mass and appear poorly nourished. Modification of feeding methods and diets, including the introduction of ketogenic diets, may improve the nutritional state, promote weight gain, and improve control of seizures and EEG abnormalities.

Bromcriptine has been tried(13). Initial trials showed improved communication, regularization of sleep pattern, and improved vocalization. There was decrease in apnea, bruxism, and in the stereotypic hand movements, and improvement in motor abilities. However, a double blind controlled trial in 10 patients by the same group, showed improvement in only two patients, and partial improvement in one.

Scoliosis is a major problem and requires early surgery and posterior spinal fusion. Recently, elbow splints have been tried to decrease the hand movements and improve motor ability; they show some promise, but further research is needed(7).

In conclusion, Rett syndrome should be identified as distinct from cerebral palsy as the condition is seen among Indian children. It is important to recognize the entity because of its therapeutic and prognostic implications.

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The Shaken Baby Syndrome

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Although the term "Shaken Baby Syndrome" has become well entrenched in the western literature on child abuse, it has not been reported in our country. This is more likely due to lack of awareness than absence of this form of baby bashing in our society. Hence, we report this case to highlight its salient features. The syndrome is characterized by a constellation of neurological findings with minimal or absent external marks of injury in the presence of fundal hemorrhages and CT evidence of subarchnoid and subdural hematomas in absence of any bleeding disorder(l). History of shaking is characteristically absent as the perpetrator inflicts the injury unknowingly out of anger or even during rough play. The term shaken baby syndrome was coined by Caffey to describe a group of infants with this typical picture where a nursemaid admitted to roughly shaking the babies by the arm or trunk(l). The mechanism of injury was assumed to be whiplash type motion of the head resulting in tearing of the bridging veins.

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Case Report

An 9-month-old male infant apparently healthy till the evening of the day of admission was brought with complains of vomiting, lethargy, irritability and seizures. There was no history of fever, otorrhea, rash, bleeding from any site or injury. However, on questioning later, the father