AUTISM IN TUBEROUS SCLEROSIS

RAKESH KHANNA¹, MAMTA SOOD²

SUMMARY

A case of Tuberous Sclerosis with infantile autism, misdiagnosed as mental retardation with seizure disorder is being discussed in light of the implication of the misdiagnosis.

Autism is now generally considered to be a biologically determined behavioural disorder. A number of medical conditions have been shown to be etiologically related to autism. Tuberous sclerosis (T.S.) is perhaps the best documented genetic disorder known to be associated with autism (Wing and Gould, 1979; Coleman and Gillberg, 1987). About 5% of all autistic patients have T.S. The majority of children with T.S. have infantile spasms. The association between infantile spasms and infantile autism is particularly impressive (Rikonen and Amnell, 1981; Hunt and Dennis, 1987).

CASE REPORT

L.M., a 12 year old girl from rural background was brought for psychiatric consultation with a long standing history of delayed development and bizarre behaviour. She was the sixth of seven sibs, born of a non-consanguineous union after a prolonged labour. Her development was normal for the first four months, when she developed infantile spasms. These spasms were first noticed only during sleep. Later the frequency increased to around 20-25 spasms a day. Around the age of one year she started having generalized tonic clonic seizures. All milestones of development were delayed. Intellectual, language and social development were particularly poor. She hardly understood any instructions and needed constant help for even self care. She remained aloof, exhibited poor eye to eye contact and laughed without reason. She could utter only two words. She kept on moving almost continuously throughout her waking hours and kept on touching and manipulating household articles. She had received intermittent treatment for her generalized tonic clonic seizures. The seizures remitted when she was 11 years old.

On examination she had adenoma sebaceum on the face, a shagreen patch on the lateral side of the left chest and early diastolic murmur in the mitral area. Mental status examination revealed a hyperactive, distractible child with very poor eye to eye contact and stereotyped repetitive movement such as clapping, body rocking, touching her face and the examiners hands repeatedly. She emitted incomprehensible sounds. On T.S. questionnaire, she scored 10/13 (score 7+ regarded as autistic behavior). There was no family history of any neurological or psychiatric illness. However, very few of the family members have been examined for T.S.

Investigations revealed numerous areas of calcification on X-ray skull and an enlarged cardiac shadow on X-ray chest. Electro-cardiogram showed a low voltage record with ST segment elevation.

¹ Assistant Prof. of Psychiatry
² Resident in Psychiatry

Central Institute of Psychiatry, Kaske, Ranchi-834006.
and T wave inversion. Electroencephalogram showed normal background activity with generalized sharp waves. I.Q. assessment put her in the category of moderate mental retardation. Multimodal treatment with low dose haloperidol and behavioural techniques led to significant improvement in hyperactivity and stereotype.

**DISCUSSION**

Psychiatric manifestations in T.S. are not uncommon. They have been variously described as a primitive form of catatonic schizophrenia (Crichtley and Earl, 1932), catatonic schizophrenia (Ziestow and Kleiner, 1965), severely socially impaired behaviour (Wing and Gould, 1970), or autism (Riikonen and Amnell, 1981; Hunt and Dennis, 1987) with considerable overlap in clinical presentation. Hunt and Dennis (1987) found 40 of 57 mobile children with T.S. (70%) to show autistic behaviour at five years of age and 59% showed hyperkinetic behaviour. There appears to be a strong correlation between seizure disorder and mental retardation (Gomez, 1979) and between infantile spasms and autistic behaviour (Hunt and Dennis, 1987; Ohlsson et al., 1988).

In this case there was clear evidence of autism along with mental retardation. Evidence suggests that even in severely mentally retarded children, the specific pattern of childhood autism can be reliably identified (Wing and Gould, 1973; Riikonen and Amnell, 1981). Questionnaires specifically aimed at elucidating social impairment and autistic behaviour may be particularly helpful (Hunt and Dennis, 1987). Administration of haloperidol has been shown to result in statistically and clinically significant decrease in hyperactivity, withdrawal, stereotypies and manurism (Anderson et al., 1984). Misidentification of autistic behaviour among severely handicapped children may deprive them of the potentially beneficial effect of neuroleptics.

Young children showing the combination of autism, mental retardation and typical or atypical infantile spasms should be suspected of underlying tuberous sclerosis (Gillberg, 1988). This suspicion should be entertained even in cases without a family history of T.S. Prompt and adequate treatment of infantile spasms and all seizure disorders may prove to be a crucial factor in determining the outcome of these cases.

**REFERENCES**

Anderson, L. T.; Campbell, M.; Grega, D. M.; Perry, R.; Small, A. M. and Green, W. H. (1934). Haloperidol in infantile autism: effects on learning and behavioural symptoms. American Journal of Psychiatry, 141, 1195-1201.

Coleman, M. and Gillberg, G. (1987). The biology of autistic syndromes. 2nd Ed., New York: Praeger.

Crichtley, M. and Earl, C. T. G. (1932). Tuberous sclerosis and allied conditions. Brain, 55, 311-346.

Gillberg, G. (1988). The neurobiolology of infantile autism. Journal of Child Psychology and Psychiatry, 29, 257-266.

Gomez (1979). Tuberous sclerosis. New York: Raven press.

Hunt, T. A. and Dennis, J. (1987). Psychiatric disorder among children with tuberous sclerosis. Developmental Medicine and Child Neurology, 29, 190-198.

Ohlsson, I.; Steffenberg, S. and Gillberg, G. (1988). Epilepsy in autism and autism-like conditions. Archives of Neurology, 45, 566-568.

Riikonen, R. and Amnell, G. (1981). Psychiatric disorders in children with earlier infantile spasms. Developmental Medicine & Child Neurology, 23, 747-760.

Wing, L. and Gould, J. (1979). Severe impairment in social interaction & associated abnormalities in children: epidemiology and classification. Journal of Autism & Developmental Disorders, 9, 11-29.

Ziestow, M. and Kleiner, S. (1965). Catatonic schizophrenia associated with tuberous sclerosis. Psychiatric Quarterly, 39, 466-475.