PSYCHOLOGICAL INTERVENTION IN TUBEROUS SCLEROSIS: A CASE REPORT

PUJA SHARMA & KIRAN RAO

ABSTRACT

Tuberous sclerosis (TS) is characterized by the clinical triad of epilepsy, mental subnormality and adenoma sebaceum. TS is a multi-system disorder resulting in severe distress for the individuals and their family members. The present study illustrates an eclectic psychosocial management of a patient with TS and normal intelligence.

Key Words: Tuberous Sclerosis, Depression, Psychological intervention

Tuberous Sclerosis (TS) is a genetic condition transmitted as an autosomal dominant trait. It is characterized by the classical clinical triad of epileptic seizures, mental retardation and adenoma sebaceum. In addition, a variety of associated problems including autistic behaviour, hyperactivity and aggressive behaviour may be present (Hunt, 1993). Several clinical studies suggest that almost 50% of the patients with TS may have normal intelligence (Harrison & Bolton, 1997). More recently, neuropsychological deficits in patients with normal intelligence have been reported in the form of impairment in planning, working memory and executive function (Harrison et al., 1999; Hunt, 1999). The view of TS has changed from that of a degenerative condition to a multi-system disorder.

In the Indian setting, several studies have highlighted the clinical presentation of TS (Khanna et al., 1991; Narayanan et al., 1987; Somasundaram & Vaidyalingam, 1969). However, none have dealt with the psychosocial issues involved. Patients with TS, who have normal intelligence, may benefit from psychological intervention directed at helping them and the family cope with the illness. This paper illustrates the use of an eclectic psychological intervention in a patient with TS.

CASE REPORT

Ms. P is a 20 years old, unmarried Hindu female from an urban lower middle, socioeconomic background and nuclear family. She was referred to the psychiatry outpatient unit in November 1998 by the Neurologist, with complaints of anger outbursts, multiple, impulsive deliberate self-harm attempts, increase in frequency of seizures, impaired social and occupational functioning, of two years duration. These symptoms had a gradual onset and a progressive course. She had a history of complex-partial seizures and adenoma sebaceum since the age of seven years. She was diagnosed as having TS, and was on anti-epileptic treatment. Complete seizure control had not been achieved. There was no family history...
of TS. Patient was the eldest of two siblings with a younger brother. Father was the sole wage earner and under stress of losing his job due to impending closure of the factory in which he worked. Family environment was characterized by high level of expressed emotions in the form of critical comments and hostility directed at the patient. Personal history revealed the client to be sociable and outgoing, but also demanding and impulsive. She had discontinued her studies two years ago, after failing twice in the 10th standard examination. On physical examination, the presence of ash leaf spots, facial angiofibroma on the left cheek and a shagreen patch in the lower back were recorded. Detailed mental status examination revealed depressed mood and ideas of worthlessness and hopelessness. A psychiatric diagnosis of Dysthymia was made and the client was started on SSRI. In view of the psychosocial stressors and history of academic failure, the patient was referred to the clinical psychologist for further management.

RESULTS

A detailed baseline cognitive and mood assessment was carried out using the Bhatia’s Performance Test of Intelligence (Bhatia, 1955), the NIMHANS Neuropsychological Battery (Mukundan et al., 1991), the Beck Depression Inventory (BDI, Beck et al., 1967), and the Dysfunctional Attitude Scale (DAS, Weissman, 1979). These tests were repeated after 1 year at the end of active intervention (post-treatment) and six months later (follow up assessment). Table 1 depicts the significant findings. Psychological testing ruled out mental retardation and specific learning disability. Neuropsychological evaluation indicated a predominantly frontotemporal involvement with deficits in attention and concentration, working memory, reasoning, planning, verbal encoding, recall, integrative ability, set shifting and presence of confabulation.

PSYCHOLOGICAL INTERVENTION

Based on the evaluation, a management plan was drawn up, with the initial phase comprising of supportive therapy and cognitive retraining. In the middle phase, cognitive therapy techniques were added. Details of the intervention are shown in Table 2.

| Tests/ function | Baseline | Post | Followup |
|-----------------|----------|------|----------|
| I.Q.            | 104      | 119  | 112      |
| Attention & concentration | Mild Impairment | Adequate | Adequate |
| Distractibility | Mild impairment | Adequate | Adequate |
| Set Shifting    | Severe Impairment | Mild | Mild |
| Planning        | Moderate Impairment | Mild | Mild |
| Working memory  | Severe impairment | Moderate | Moderate |
| Kinetic melody  | Mild impairment | Adequate | Adequate |
| Verbal encoding | Moderate impairment | Mild | Mild |
| Confabulation (Verbal) | Present | Absent | Absent |
| Perseveration (motor) | Present | Absent | Absent |
| Delayed verbal memory | Moderate Impairment | Mild | Mild |
| Visual integration | Moderate Impairment | Mild | Mild |
| BDI (Range 0-63) | 35      | 08   | 11       |
| DAS (Range 40-280)| 223     | 197  | 167      |

A supportive approach was adopted, ventilation of problems was facilitated, and the therapist (PS) maintained a warm and non-judgmental stance. The patient reported feeling anxious about her future, expressed negative body image because of her facial scar and plump appearance and had high rejection sensitivity. Reassurance was given regarding her normal intellectual functioning and...
# Table 2: Details of the Psychological Intervention

| Period       | Pharmacotherapy | Psychological Intervention | Sessions | Outcome                                      |
|--------------|-----------------|---------------------------|----------|----------------------------------------------|
| Phase I      |                 |                           |          |                                              |
| (0-3 months) | Phenobarbital (60mg) | Individual Work:           | individual sessions (daily)=76 | Reduction of: |
|              | Carbamazepine (900-1200mg) | 1. Supportive Therapy:     |          | Seizure frequency                           |
|              |                 | Ventilation, reassurance,  |          | Depressed mood crying                        |
|              |                 | guidance, activity        |          | spells suicidal ideation                     |
|              |                 | scheduling               |          | DSH attempts impulsivity                     |
| Initial Phase| SSRI (50-100mg) | 2. Cognitive Retaining:    |          | Improvement in:                             |
|              |                 | grain sorting, letter     |          | Attention, Concentration                     |
|              |                 | cancellation, colouring   |          | Reasoning, Planning                         |
|              |                 | tasks, verbal fluency     |          |                                              |
|              |                 | and encoding              |          |                                              |
|              |                 |                           |          |                                              |
|              |                 | 3. Psycho-education       |          |                                              |
|              |                 |                           |          |                                              |
|              |                 | Family Work:              |          |                                              |
|              |                 | 1. Intake & Family        | Family sessions (fortnightly)=6 | Father more responsive, |
|              |                 | Assessment                |          | mother & brother still have                 |
|              |                 | 2. Psycho-education &    |          | difficulty in accepting                      |
|              |                 | support                  |          |                                              |
|              |                 | 3. Recognizing high      |          |                                              |
|              |                 | expressed emotions,      |          |                                              |
|              |                 | coping skills             |          |                                              |
| Phase II     |                 | Individual work:          |          |                                              |
| (4-10 months)| Phenobarbital (60 mg) | 1. Supportive Therapy:     | individual sessions=32 | Reduction in: |
|              | Carbamazepine (1200mg) | (added) Environmental     |          | Anger outbursts,                            |
|              |                 | Social Skills training    |          | Behavioural problems at home                |
|              |                 | with anger management:    |          | Seizure frequency leading to reduction in    |
|              |                 | Life skill management:    |          | the medication                              |
|              |                 | diet and exercise         |          | Anxiety about body image                    |
| Middle Phase | SSRI (< to 50mg & stopped) | 2. Cognitive Retraining:  |          | Improvement in:                             |
|              |                 | Home based tasks with    |          | Abstraction, set shifting,                   |
|              |                 | increase in complexity of |          | Verbal Encoding,                            |
|              |                 | earlier tasks,            |          | Reasoning, Confidence,                      |
|              |                 | Go-no-Go tasks for set    |          |                                              |
|              |                 | shifting and cognitive    |          |                                              |
|              |                 | flexibility, Precis       |          |                                              |
|              |                 | writing for verbal        |          |                                              |
|              |                 | encoding                  |          |                                              |
|              |                 |                           |          |                                              |
|              |                 | 3. Cognitive Therapy:     |          |                                              |
|              |                 | Identifying cognitive     |          |                                              |
|              |                 | errors, Cognitive         |          |                                              |
|              |                 | restructuring             |          |                                              |
|              |                 |                           |          |                                              |
|              |                 | Family work:              |          |                                              |
|              |                 | Psychoeducation Reduce    |          | Improvement in:                             |
|              |                 | expressed emotions        |          | Communication at home,                      |
|              |                 | Contingency management,   |          | less fights greater                         |
|              |                 | use of positive           |          | acceptance of patient                       |
|              |                 | reinforcement and time out|          |                                              |
| Follow-up    |                 | Individual Sessions       |          |                                              |
|              | Clobazam (15mg)  | (monthly)=12              |          |                                              |
|              | Family Work:    |                           |          |                                              |
|              | Family Counseling & support | Family sessions (Once in 3 months)=2 |          | Improvement in: communication, Family       |
|              |                 |                           |          | actively involved in making future plans    |
that, if she worked on her cognitive deficits through retraining exercises, it may be possible for her to pass the 10th standard examination. Cognitive retraining, using attention enhancing and verbal encoding tasks, was initiated. As the patient spent most of her time watching television, activity scheduling was introduced so that she could plan her day with both work and leisure activities. This reduced preoccupation with her problems. The family assessment helped to identify the communication patterns in the family and the family’s negative perception and response to the patient’s illness. Psycho education regarding TS was provided and myths and fears regarding seizures clarified. Parents were given a feedback about the findings from the psychological assessment.

In the middle phase, cognitive techniques were introduced. The patient reported that she was a “fat, ugly girl and a failure” and that her family would be better off without her. By maintaining a diary of her daily thoughts, mood and behaviour, the patient was able to recognize her negative cognitions and how they influenced her mood and behaviour. She observed how she magnified the negatives, minimized the positives, personalized and over generalized certain statements made by her mother. These dysfunctional attitudes and negative automatic thoughts were identified and dealt with through cognitive restructuring.

During this period, high expressed emotions in the form of critical comments from the mother and younger brother continued. The mother had very high expectations and was unable to positively reinforce the efforts that the patient was making. In order to reduce face-to-face contact with the mother, develop a work habit and increase peer interaction, the patient was referred to the day care centre at NIMHANS. She worked in the Crafts section and was an eager and skilled worker. However, within one month at the day care centre, patient faced interpersonal problems with the staff and co-workers. She had difficulty in maintaining boundaries, had poor frustration tolerance and impulsive behaviour which were then taken up in the sessions. Social skills training in the form of role play and rehearsal and anger management were introduced. Life style changes were also discussed and a diet chart with an exercise regimen was initiated.

In the family sessions, the focus was on helping the family understand the link between stress and seizures and on reducing the level of expressed emotion. Patient’s mother was given a feedback about how her comments, even though well intentioned, were perceived as a rejection by the patient. Parents were helped to become more aware of the patient’s emotional needs and how her DSH attempts were a cry for help. Conjoint sessions with patient and parents were held to enable open communication within the family. Unfortunately, due to academic pressures, the younger sibling could not be part of these sessions.

At the end of 1 year, the family reported significant treatment gains in the form of reduced seizure frequency and improvement in her social and occupational functioning. Post assessment was carried out and the patient was given a feedback of the gains made by her and the need to plan for the future. She expressed the desire to study and write her tenth grade examination. She decided to stop attending the day care centre and started preparing for her exams. She was able to make a study plan for herself as well as balance her leisure time. She did minimal household chores to help the mother. A monthly follow up was agreed upon.

During the first month, there was resurgence of behavioural problems, particularly anger outbursts. The therapist recognized that the patient was quite tense and anxious due to her examination. The neurologist started her on tablet Clobazam. Regular monthly contact was maintained to reassure the patient and she was taught breathing exercises and coping self-statements to manage her examination anxiety. She passed the matriculation exam and reported feeling “on top of the world”. This was a major turning point for the patient and the family. The patient reported feeling more confident of herself, as well as having a more comfortable relationship.
with her parents, especially her mother. After an unsuccessful attempt at laser therapy for her facial angiofibroma, she had better acceptance of her physical appearance. Parents also acknowledged the efforts and the gains made by the patient. As she had been seizure free for the past six months, the father was able to find her a job as an apprentice at a photocopying centre close to the house. The parents are currently looking for a regular job for the patient as well as considering self-employment options.

**DISCUSSION**

Although time intensive, psychological interventions combined with pharmacotherapy, brought the seizures under control and reduced disability. Cognitive retraining enabled the patient to complete her school leaving examination successfully, as well as benefit from the other psychological interventions. Tuchman (1994) suggested that TS might be a good model for understanding the relations between epilepsy, language and behaviour. Although depression remitted, DAS scores continue to be high and mild cognitive deficits in planning and working memory persist. Since there are continuing stressors, in terms of the issue of marriage and the lack of a stable job that would help her to be economically self-sufficient, the need for supportive therapy on a long-term basis is recognized. The patient has been advised to continue the home-based cognitive exercises. She is currently on a three monthly follow-up during which she visits both the neurologist and the clinical psychologist. This case report illustrates the complex psychosocial issues present in patients with TS and normal intelligence that necessitate a multidisciplinary approach. Majority of persons with TS have a normal life span (Hunt, 1999) and ensuring a better quality of life for them is important.

**REFERENCES**

Beck, A.T., Ward, C.H., Mendelson, M., Mock, J. & Erbaugh, J. (1961) An inventory for measuring depression. *Archives of General Psychiatry*, 4, 561-571.

Bhatia, C.M. (1955) Performance tests of intelligence under Indian conditions. Bombay: Oxford University Press.

Harrison, J.E. & Bolton, P.F. (1997) Annotation: Tuberous Sclerosis. *Journal of Child Psychology & Psychiatry*, 38, 603-614.

Harrison, J.E., O’Callaghan, F.J., Hancock, E., Osborne, J.P. & Bolton, P.F. (1999) Cognitive deficits in normally intelligent patients with Tuberous Sclerosis. *American Journal of Medical Genetics (Neuropsychiatric Genetics)*, 88, 642-646.

Hunt, A. (1993) Development, behaviour and seizures in 300 cases of tuberous sclerosis. *Journal of Intellectual Disability Research*, 37, 41-45.

Hunt, A. (1999) Psychiatric and Psychological Aspects in: Tuberous Sclerosis Complex (Eds) Gomez, M.R., Sampson, J.R. & Whittemore, V.H. Chapter 5, pp 47-62, London: Oxford University Press.

Khanna, R.K., Sharma, L.N. & James, M.X. (1991) Tuberous sclerosis: report on an unusual family. *Indian Journal of Psychiatry*, 33, 314-317.

Mukundan, C.R., Rao, S.L., Jain, V.K., Jayakumar, P.N. & Shailaja, K. (1991) Neuropsychological assessment: A cross validation study with neuroradiological-operative findings in patients with cerebral hemisphere lesions. *Psychacopsychologia*, 4, 33-39.

Narayanan, H.S., Gandhi, D.H. & Girimaji, S.R. (1987) Tuberous sclerosis: clinical experience with 19 cases. *NIMHANS Journal*, 5, 91-96.

Somasundaram, O. & Vaidyalingam, M.
(1969) Tuberous sclerosis: A report of 12 cases. Journal of Child Neurology, 9, 95-102.

Indian Journal of Psychiatry, 10, 142-148.

Weissman, A. (1979) The Dysfunctional Attitude Scale: A validation study. Dissertation Abstracts International, 40, 1389B-1390B.

Tuchman, R.F. (1994) Epilepsy, language and behaviour: Clinical models in childhood.

PUJA SHARMA, M.Phil. Ph.D. Scholar, Department of Clinical Psychology. KIRAN RAO*, M.Phil, Ph.D., Additional Professor, Department of Clinical Psychology, National Institute of Mental Health & Neuro Sciences, Hosur Road, Bangalore-560 029. (email: kiranr@nimhans.kar.nic.in).

* Correspondence.