FAMILIAL INCIDENCE OF TUBEROUS SCLEROSIS (A Case Report)

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Introduction

Tuberous sclerosis (TS) was first recognized by Von Recklinghausen in 1862 but the term was coined by Bourneville in 1880. It is the second most common neurocutaneous disorder after neurofibromatosis. The diagnostic triad as described by Vogt [1], consists of seizures, mental retardation and adenoma sebaceum. Cutaneous lesions like hypopigmented macules [2,3], adenoma sebaceum (4), shagreen patches (5), ungual fibromas and cafe-au-lait spots (1) are often pointers of diagnosis in a child with epilepsy and mental retardation. TS is a dominantly inherited disorder with variable penetration. The incidence of vertical transmission varies between 30 to 57 per cent [6,7]. Multiple (more than one member) sibling affection has been reported rarely [8]. The series reported by us includes a family of five (mother and four children) and this appears to be one of the largest number reported in literature.

CASE REPORT

The family members affected were mother (29 years old), two sons (10 and 7 years old), and two daughters (5 and 4 years old). Detailed inquiry into the family members on the paternal and maternal sides of the patients as well as their ancestors did not reveal any definite history of TS in any of them. The family tree is shown in Fig 1. The affected lady gave history of having the disease from the age of 8 years with manifestations of epilepsy and skin lesions. She did not have mental retardation. However, all the four children had epilepsy, skin lesions and mental retardation. The skin lesions are detailed in Table 1 and shown in Fig 2. There were no abnormalities in cardiovascular and respiratory systems. Except for epilepsy and mental retardation, CNS examination also was normal in all patients. Laboratory investigations were essentially normal. Chest posteroanterior radiogram, electrocardiogram, echocardiogram and ultra-

sound study of abdomen did not reveal any abnormality. Skull radiogram showed calcified spots in two patients (40%), EEG was abnormal in 4 (80%), and CT scan showed abnormality in all the patients. The electroencephalographic and CT scan abnormalities are described in Tables 2 and 3.

Fig. 1 : Family tree of the Tuberous sclerosis patients.

Fig. 2 : The family of patients. Lesions on the faces of the mother and the child on extreme right are clearly visible.

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TABLE 1
Skin lesions : family members

Type of lesion	Patient	Total
Adenoma sebaceum	M S1 D1	3
Shagreen patch	M S1 S2 D1	4
Hypopigmented macules	S1 D1 D2	3
Cafe-au-lait spots	M D2	2
Total skin lesions (neurocutaneous markers)		5

M = Mother, S1 = Elder son, S2 = Younger son, D1 = Elder daughter, D2 = Younger daughter

TABLE 2
EEG findings : family members

EG findings	Patient	Total
Generalised epileptic discharge	S1 S2 M D1	4
Generalised theta response	M S1	2
Rt. temporal lobe sharp wave	S1	1
Slow wave parieto junctional areas	S1 D1	2
Total	5	

M = Mother, S1 = Elder son, S2 = Younger son, D1 = Elder daughter, D2 = Younger daughter

TABLE 3
CT Scan results

CT Results	Family members (X)	Total
Multiple periventricular calcification	M S1 S2 D1 D2	5
Prominent cortical sulci	M S1 S2 D1	4
Ventricular dilatation	S1 S2	2
Cortical calcification	M S1 D1	3

(X) : M = Mother, S1 = Elder son, S2 = Younger son, D1 = Elder daughter, D2 = Younger daughter

All the five patients were being treated symptomatically including antiepileptic drugs and have been under follow-up for the last four years. A gradual increase in the mental retardation was observed in all children and the epileptic attacks recurred occasionally despite therapy.

Discussion

TS is a disease of the childhood [9]. The family being reported by us consists of four children - all under the age of 10 years and their mother (age 29 years) who also suffered from TS from the age of

8 years. The incidence of four siblings in addition to their mother with the disease of TS is one of the largest family reported in literature [1]. The vertical transmission of the disease reported to be 30-40 per cent in literature [6,7] was also recorded in our series - from mother to her four children. There was no other case of TS among the ancestors both on the paternal and maternal sides of the patients. It is possible that some cases might have been missed because of ignorance or mild atypical presentation. Skin lesions in one form or the other were seen in all (100%) of the family members affected. TS cases without skin lesions are very rare and often remain undiagnosed [7,10]. Adenoma sebaceum (60%), shagreen patch (80%) and hypopigmented macules (60%) were the main neurocutaneous markers detected in our series. Adenoma sebaceum and hypopigmented macules are usually present from birth and shagreen patch develops after 10 years of age [2,4,5]. None of our cases had other system involvement, though lesions in eye, heart, lungs and kidney have been found frequently in other series [1]. The neurological manifestations of seizures and mental retardation have been universal finding in our series as in most others [1,7,9]. The CT scan has been most rewarding and informative, showing diagnostic features like periventricular calcification in all (100%) and prominent cortical sulci, ventricular dilatation as well as cortical calcification. We however did not detect brain tumours in any of our cases, an association reported in about 8-15 per cent of TS patients [9].

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