TUBEROUS SCLEROSIS : A THREE-GENERATION PEDIGREE

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This is a report of tuberous sclerosis in members of three generations originating from parents, both suffering from the disease. Out of 12 family members only two were exempted and clinical expression varied from minimum lesion like adenoma sebaceum only to severe mental retardation and epilepsy.

Key Words: Tuberous selerosis, Autosomal dominant inheritance, Adenoma sebaceum

Introduction

Tuberous sclerosis is a rare congenital disorder characterised pathologically by sclerotic masses in cerebral cortex, adenoma sebaceum and hamartoma formation in various organs; and clinically by mental retardation and epilepsy. Typical skin lesions are ash leaf macules,[1] shagreen patch and angiofibroma. Hypopigmented macules are the earliest signs and may appear in the form of "thumb print" (polygonal), "ash leaf" (lance ovale), or "confetti" (white spots). The inheritance is simple autosomal dominant and it is claimed that gene of tuberous sclerosis is located on chromosome 9P.[2] Parents may have no adonema sebaceum or neurological involvement and cases of fresh mutation may be as high as 40%. Involvement of more than one sibs is relatively common.[3]

Care Reports

We are reporting a family where members of three generations are affected. Family consists of affected couple, their children and grand children. Of 12 family members 10 are affected, two males in second generation are spared. Clinical presentation varied from only benign angio fibroma to severe mental retardation and epilepsy. Members of third generation are most severely affected (Table I).

Discussion

Reports of tuberous sclerosis in three generations are rare. Berg reported one such family in 1913.[4] There are several reports of involvement of two generations.[5]

In this study out of 10 affected in three generations, 90% had adenoma sebaceum, mean age of appearance was between 7-9 years, 30% had hypopigmented macules (this is lower than previous reports);[1] 40% had shagreen patches; 50% had epileptic fits of which one had temporal lobe type of seizure and one had infantile spasm; 70% had mental retardation; 30% had normal intelligence in spite of presence of adenoma sebaceum with or without epilepsy.

Psychiatric problems found other than retardation were paranoid psychosis in girl with temporal lobe epilepsy and hyperkinetic syndrome of childhood in a child with infantile spasm. Extreme degree of variability of expression, characteristic of the disease is clearly noticeable in the pedigree. The family tree suggests dominant mode of inheritance.

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Table I. Family Tree with Manifestations

	Father Adenoma Sebaceum Mild Retardation		Mother Adenoma, Sebacecum Low nomral I.Q. 90+			
Sova age 40+ I.Q. 90+ A.S. Epilepsy, Intracranial	Sarama age 32 Mild MR A.S. Shagreen Patch Epilepsy -Digital fibroma.	Swapan (29) I.Q. 90+ A.S. Confetti sign Ash leaf macule (Married wife Pregnant)	Kalpana Mild MR A.S. -Subungual fibroma patch -Temporal lobe epile -psy (EEG) -Paranoid Psychosis (Died-Suic cide) at age 20)	Jaharlal age 26 Normal I.Q. No apparent problem	Nirapada Normal I.Q. No. apparent problem	Narugopal -Mild retardation A.S. -Shagreen Patch -Subungual fibroma -Dental Epulis -Epilepsy -X-Ray skull Intracranial calicification
Gobinda age 25 Balai (age 7) I.Q. 84 A.S Shagreen Patch Ash leaf Macule I.Q. 70 A.S (Just starting to appear)	Lenin - Severe MR Hypopigmented Macule -Hyper Kinetic disorder - Infantle spasme (Died drowing at age 7)					

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