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TUBEROUS SCLEROSIS IN IDENTICAL TWINS

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Tuberous sclerosis was observed in six-year-old identical twin brothers born with a single placenta and a single amnion. Both had adenoma sebaceum, shagreen patches, ash leaf spots and progressive mental deterioration. One of them had a verrucous pigmented nevus on the left temple and recurrent localised motor seizures.

Key words: Tuberous sclerosis, Identical twins.

Tuberous sclerosis was first recognised as a specific disease in the 19th century.[1] It is believed to be an autosomal dominant trait with great variability of expression.[2] Recently, Eapen et al[3] reported the syndrome in identical twin sisters. We report another case of the disease in identical twin brothers.

Case Reports

Six-year-old identical twin brothers from district Fatehpur had asymptomatic, verrucous, papular lesions on their face since the age of 13 months. Both were the third children of their parents. They were born at home after normal delivery at 7 months of gestation with a single placenta and a single amnion. The gestation period was normal except for the unsuccessful attempt for termination of pregnancy with an unknown injection. Both were small at birth; birth weights not known. Their growth and milestones of development were normal.

Between the ages of 6 months to 12 months, one had recurrent localised motor seizures. The momentary fits used to start with sudden jerking movements of hands and feet and rolling up of eye balls. There was no history of seizures in the other. None had any evidence of focal neurological deficit. There were no behavioural deviations, neither respiratory, cardiac or genito-urinary symptoms. Both of them were studying in class II. They were average in studies. During follow-up over the next three years, there was some deterioration in their mental capacities. The first one was more dull in studies. There was no history of similar illness among family members.

Skin lesions first appeared in the naso-labial folds in both brothers simultaneously and gradually progressed to affect other areas on the face and body. The lesions differed slightly in both of them. Both had a large number of dome-shaped 2 to 5 mm size, reddish brown papules (adenoma sebaceum) on face, nose and neck; more concentrated on the butterfly area of face. Oval, depigmented macules (ash leaf spots) were present on the buttocks, back and thighs (3 in one and 2 in the other). Hypopigmented, yellowish, variably thickened (1 to 2 mm), rough, soft plaques (shagreen patches) varying in size from 4 to 20 mm or more, with irregular margins were also present in both of them in the lumbo-sacral area, forehead and flanks. All the lesions were larger and present in greater numbers in the first than in the second. The first also had a firm verrucous pigmented nevus on the left temple just lateral to the left eye. Cafe-au-lait spots were present in both of them. Periungual fibromata (Koenen's tumours), poliosis, cutaneous tags, ichthyosis, plantar keratoderma and macular atrophy were not seen.

General examination did not reveal any

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abnormality. Both boys were alert and active. Systemic examination also revealed nothing abnormal. Liver and spleen were not palpable. Central nervous system examination was normal. Fundus examination showed normal disc. Phacomas or other tumours were not seen. Routine urinalysis, total and differential leucocyte counts and erythrocyte sedimentation rates were within normal limits. Each had 'O' Rh-ve blood group. No abnormality was detected on skiagrams of the skull and chest. HLA studies were not done.

Comments

Only Eapen et al[3] have, to our knowledge, reported tuberous sclerosis among identical twin sisters. However, they did not give details of the cases. Being an inherited autosomal dominant condition, the disease may occur in a heterozygous twin of the patient just as it may be present in other sibs. Korten and Scholten[4] reported a pair of heterozygous twins, one of whom exhibited the classical symptoms of tuberous sclerosis and the other epilepsy and oligophrenia.

The physical characteristics such as eye and hair colour help in assessing the identical nature of the twins, but it is essential to perform blood grouping, HLA typing or enzyme pattern studies before declaring monozygocity of twins. We could not do HLA typing or enzyme pattern studies. However, the single placenta and the single amniotic sac along with the identical blood groups suggest that our twins are monozygotic.

A report of a disease occurring in a single pair of twins or a series of twins, though interesting, has a limited value for simply inherited conditions. It does not enlighten further on the pattern of inheritance which is recognised even without them. Very occasionally, however, a single discordant monozygotic pair might point to a hitherto unsuspected complication; for example in such a pair, if only one of the members suffered from a disease thought to be recessive, would show that penetrance was incomplete.[5]

Our report is also of limited value. These cases can be attributed to new mutations as there is no history of similar illness in the family. The incomplete penetrance of the disease is apparent from the differences in the clinical patterns of both the cases.

References

1. Morgan JE and Wolfert F: The early history of tuberous sclerosis, Arch Dermatol, 1979; 115: 1317-1319.
2. Feingold M and Pashyan H: Tuberous sclerosis, in: Genetics and Birth Defects in Clinical Practice, Little Brown and Company, Boston, 1983; p 187-188.
3. Eapen EP, Naik RPC and Parameswara YR: Tuberous sclerosis, Ind J Dermatol Venereol Leprol, 1984; 50: 60-63.
4. Korten JJ and Scholten ACM: Tuberous sclerosis (Bourneville pringle disease). Tubereiuze sclerose (ziekle Bourneville pringle), Ned T Geneesk, 1970; 11: 1909-1911. (Abstracted Excerpta Medica, 1971).
5. Roberts JAF and Pembrey EM: Multifactorial inheritance — its contribution to continuous variation and its role in the causation of common diseases, in: An Introduction to Medical Genetics, 7th ed, The English Language Book Society and Oxford University Press, 1978; p 269-272.