Tuberous Sclerosis (Bourneville Disease)

Jugal Kishore Kar, Manoranjan Kar

The patient M.T. was a boy of 12 years and admitted with generalized tonic-clonic seizure (GTCS). Family history of the patient was non contributory. Past h/o GTCS, once at 2nd yr of age. The patient was of average built and nourished. Mild mental retardation was evident. Clinical examination showed many papules of sebaceous adenoma spread out typically like the wings of a butterfly on both the sides of the face and nose (Fig. 1). Fundoscopy, USG Abd, X-ray both hands were normal. NCCT brain revealed subependymal calcified nodules (Fig. 2). However, seizure of the child was controlled well with Eptoin.

The term tuberous sclerosis was given by the French neurologist Bourneville in 1880 is characterised by: (i) retarded mental development (ii) occurrence of epileptiform seizures, (iii) adenoma sebaceum, and (iv) humours in one or more organs. It is usually a neurocutaneous syndrome with an autosomal dominant inheritance, sporadic TSC cases (no family history) are not uncommon. Cutaneous manifestations are facial angiofibromas (adenoma sebaceum), Shagreen patches, hypopigmented macules, ash leaf spots, café-au-lait spots, facial hypoplasia and periungual fibromas. The patterns of epilepsy encountered are generalized tonic clonic seizures, complex partial seizures, simple partial seizures and myoclonic jerks including infantile spasms. The other features are tubers, sub-ependymal nodules (SEN) and giant cell astrocytomas (SEGA5), cardiac rhabdomyoma, retinal hamartomas, renal angiomyolipomas, pulmonary lymphangiomatosus, dental pitting, bone cysts, gingival fibromas, hamartomatous rectal polyps and cerebral white matter radiation lines on MRI. Tuberous sclerosis complex (TSC) results from mutations in one of two genes, TSC1 on chromosome 9q34 and TSC2 on chromosome 16p13, that encode distinct proteins, tuberin and hamartin respectively. The favorable response of seizures in TS to vigabatrin were documented. A definite diagnosis of TSC requires two or more distinct types of lesions rather than multiple lesions of the same type in the same organ system.

References


*Associate Professor, Dept. of Medicine, Midnapur Medical College, West Bengal; **Associate Professor, Dept. of Surgery, Calcutta Medical College, Kolkata, West Bengal

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