CASE REPORT

Tuberous Sclerosis With Chorea

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Abstarct

Here, we report a case of tuberous sclerosis with mulberry lesion of retina and chorea. The mulberry lesion of retina is considered to be the characteristic retinal lesion of this neurocutaneous syndrome.However, presence of chorea in tuberous sclerosis is a rarity.

Key Words

Tuberous Sclerosis, Chorea,Hamartoma, Mulberry Lession

Introduction

Tuberous sclerosis is a neurocutaneous syndrome with a prevalence of 1 per 6000-9000 individuals.It is inherited as an autosomal dominant trait with variable penetrance. Two genes responsible for tuberous sclerosis include TSC1(located at chrosome 9q34) and TSC2(located at chromosome 16p13.3).TSC1 gene is more responsible for familial cases of tuberous sclerosis (1).The disorder is characterized by a triad of mental retardation,seizures and facial angiofibromas.However, the diagnosis of tuberous sclerosis is established by following the revised criteria for tuberous sclerosis complex (2). Retinal hamartomas have been reported in over 75% cases of tuberous sclerosis but "mulberry lesion" of retina is characteristic of this syndrome(3,4). Here, we report a case of tuberous sclerosis with characteristic mulberry lesion of retina and chorea for its rarity (5,6).

Case Report

A14 years old boy presented with complaints of generalized tonic-clonic seizures and poor scholastic performance for the last nine and six years respectively. His birth and developmental history were unremarkable. Patient,s mother has been suffering from generalized tonic-clonic sezures since early childhood. His younger sib too has been having mixed seizure disorder(generalized tonic-clonic & myoclonic seizures) for the last eight years. Apart from mental retardation and appendicular choreiform movements, his neurological examination was unremarkable. Dermatological examination revealed presence of adenoma sebaceum on face. Ophthalmological evaluation revealed mulberry lesion of retina between optic disc and fovea. Electroencephalography(awake record) revealed background slowing with intermittent sharp transients arising bihemispherically. Cranial computerized tomographic scan(noncontrast) showed multiple

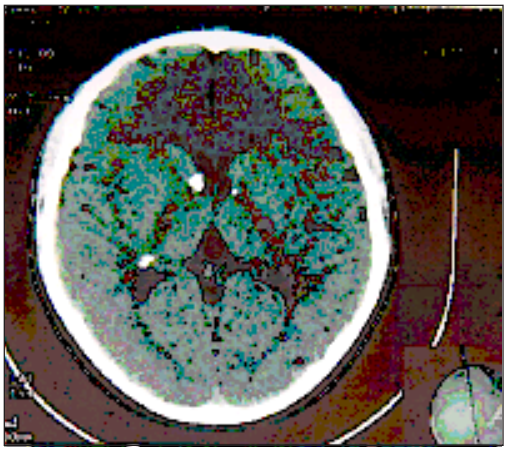

Fig 1. CT Scan (noncontrast) of Head Showing Multiple Subependymal Nodules

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subependymal hyperdense nodules along lateral borders of lateral ventricles(*Fig.1*).

Abdominal ultrasonography, chest skiagram and echocardiography were noncontributory. Examination of mother revealed presence of facial angiofibromas whereas his younger sib had mental retardation which was corroborated by his poor scholastic performance. There was no clinical or laboratory evidence pertaining to any other cause of chorea. With aforementioned constellation of clinical and neuroimaging features,a diagnosis of definite tuberous sclerosis was entertained as the patient fulfilled the requsite criteria for the entity.2

Discussion

Tuberous sclerosis, the second commonest neurocutaneous syndrome, is characterized by a constellation of major and minor features2.Major features include facial angiofibromas,nontraumatic ungula or peri-ungual fibromas,hypomelanotic macules,shagreen patch,retinal hamartomas,cortical tubers,subependymal nodule,subependymal giantcell astrocytoma,cardiacrhabdomyoma,lymphangiomyomatosis and renal angiomyolipoma.Minor features include dental enamel pits,hamartomatous rectal polyps,bone cysts,gingival fibromas,nonrenal hamartomas,retinal achromic patch,confetti skin lesions,renal cysts and cerebral cortical dysplasia.Retinal hamartomas constituting the commonest ocular abnormality in patients of tuberous sclerosis are found in 50% to 80% patients.Three different types of retinal hamartomas have been described in patients of tuberous sclerosis.One form is characterized by a smooth, semitransparent, oval, salmon-coloured hamartoma overlying the retinal vessels. The second type of retinal hamartoma is an opaque, multinodular lesion with yellowish-white colour. It resembles a pile of salmon eggs and has been referred to as "mulberry lesion" or astrocytic hamartoma of retina.These lesions have a tendency to calcify with time and are located in the posterior pole of retina adjacent to the optic disc,obscuring the disc as well as its vessels.The third type of retinal hamartoma shares features of previously described two types of retinal hamartomas.The retinal hamartomas arise from the ganglion cell layer of the retina and usually do not interfere with the vision3,4.The occurrence of chorea in tuberous sclerosis can be attributed to the disrupted neuronal migration resulting in an aberrant cellular interaction and synaptic transmission in the basal ganglia and their connections in general and caudate nucleus in particular.

Conclusion

Chorea can be an unusual neurological manifestation of Tuberous sclerosis. Its occurrence can be attributed to the neuronal migration disorder associated with Tuberous sclerosis. Neuronal migration disorder can cause disruption of synaptic transmission in the basal ganglia and their connections, thereby resulting in chorea.

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