Tuberous sclerosis: A rare cause of seizure in Nigeria.

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Abstract

**Background:** Tuberous sclerosis is a rare genetic multisystem disorder that is typically apparent shortly after birth. Dermatologic manifestations may be the only clues to the diagnosis of the disorder, which is also marked by childhood seizures and mental retardation. The aim of this report is to present a twelve year old boy with tuberous sclerosis.

**Methods:** A review of the case records of a child with angiofibromas of the face and neck and the relevant literature.

**Results:** An eleven year old primary two pupil of Ijaw tribe, southern Nigeria, presented with recurrent afebrile, generalized tonic-clonic seizures from nine months of age, hyperpigmented papulonodular eruptions on the face and neck with some hypo pigmented patches at the back for six years. He also had a growth on the right index finger of six years duration. There was associated learning disabilities and poor school performance, with sudden outburst of mood swings ranging from laughter to anger. Speech was delayed. He has been on Carbamazepine for the past two years and is seizure free. There was no history of similar illness in the family.

Physical examination showed that he had labile mood with presence of hyperpigmented papulonodular (angiofibromas) eruptions on the malar area of the face and neck. There were also associated hypomelanotic macules on the back, bony cyst on the right index finger. He also had bilateral descended testis. All other systems were essentially normal. Cranial computed tomography showed multiple hyperdense non enhancing nodules in the walls of the lateral ventricles with a hyperdense nodule in the subcortical area of the parietal lobe of the left cerebrum. Multiple areas of non enhancing hypodensities were also seen in the cortical white matter of the frontal and parietal lobes bilaterally with associated thickening of the adjacent gyri. He is being managed by a team of a paediatric neurologist, surgeon, speech therapist and a dermatologist. He is regular at follow up clinic.

**Conclusion:** Tuberous Sclerosis though a rare condition, once diagnosed needs multidisciplinary management to improve the quality of life of the patient.

**Key words:** Tuberous sclerosis, seizure, skin rashes.
neurological manifestations were emotional lability and speech defect. Also our patient had the neuropathological features associated with TS- had subependymal nodules on CT scan. It is worthy of note that subependymal giant cell astrocytoma can occur in 6-14% of individuals with TS and is more likely to occur in childhood. These can enlarge and cause obstruction to the cerebrospinal fluid pathways, thereby resulting in raised intracranial pressure, focal neurological deficits and deterioration in seizure control. Neurological manifestations of TS in African children had been described by few studies, for instance Cisse et al reviewed 18 cases of TS seen over a ten year period at the University Hospital Centre, Conakry, Guinea. A high incidence of epilepsy was found in the cases studied. Chalabi- Benabdallah also reported a high incidence of epilepsy in the 22 Algerian families with TS studied. Pitche et al reported 4 cases of TS from Togo; all showed features of neurological involvement, three had enhancing hypodensities were also seen in the cortical white matter of the frontal and parietal lobes bilaterally with associated thickening of the adjacent gyri.
associated epilepsy while the fourth case showed features of autism. Neuroimaging studies to precisely identify pathologies in the brain were however not carried out in the study. Severe neurological involvement appears to be a frequent finding in Africans with TS.

The dermatological manifestations of tuberous sclerosis tend to be the most common findings. 1 Sebaceous adenomas develop between 4 and 6 years of age. They appear as tiny red nodules over the nose and cheeks and are sometimes confused with acne. Angiofibromas of the lumbrosacral skin region (shagreen patches) usually appear before puberty. Cafe au lait macules, periungual fibromas (koene's tumors), forehead fibrous plaques, skin tags and confletti like macules can also be observed. 1 Our patient had facial angiofibromas, shagreen patches on the back and periungual fibromas.

Ocular manifestations include retinal harmatomas, translucent patch, mulberry lesion and retinal depigmentation spot. Retinal lesion in TS are of two types mulberry tumors that arise from nerve head and hamomases which are retinal hamartomas of astrocytic origin. 1 Our patient did not have these findings on fundus examination.

Approximately 50% of children with TS have rhabdomyomas of the heart. These may be numerous or located at the apex of the left ventricle, and although they can cause congestive heart failure and arrhythmias, they tend to slowly resolve spontaneously. The echocardiogram done in our case was normal.

The lungs and the kidneys are also affected in TS. Pulmonary lesions in tuberous sclerosis usually affect women, suggesting that a hormonal component is involved in the development of the pulmonary sequelae. 10 Lymphangioleiomyomatosis of the lung is characterized by cystic distortion of the pulmonary architecture by hyperplastic smooth muscle cells. Renal involvement includes angiomyolipoma, renal cyst, renal cell carcinoma, oncocytoma, perirenal cysts and polycystic kidney. 11 In our case ultrasongraphy of kidneys was within normal limits.

Management was mainly symptomatic and multidisciplinary. Anticonvulsant in form of carbamazepine was prescribed. He has remained seizure-free for two years.

In conclusion, any child who presents with seizures should be carefully examined for skin lesions which can give some clue towards the causation of seizures. Tuberous sclerosis though a rare condition, once diagnosed needs multidisciplinary management due to the varied clinical manifestations. This is to improve the quality of life of the patient and ensure maximal survival.

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