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A CASE REPORT OF TUBEROUS SCLEROSIS

Paediatrics

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ABSTRACT

Neurocutaneous syndromes have cutaneous and CNS manifestation. Cutaneous manifestation can help in clinching the diagnosis of the syndrome like Tuberous sclerosis, Neurofibromatosis (1 and 2),Von hippel lindau disease, Sturge-Weber Syndrome, Ataxia –Telangiectasia, PHACE, Incontinentia pigmenti. All these can be easily diagnosed because of skin manifestations. Tuberous sclerosis complex (TSC) is a neurocutaneous syndrome. Cutaneous manifestation includes Café au lait spot, ash leaf macule, shagreen patch, facial angiofibroma, gingival fibroma, hypomelanotic macule. CNS manifestation includes cortical tubers, subependymal nodule. The hallmark of Tuberous Sclerosis Complex is the involvement of CNS.

KEYWORDS

Neurocutaneous syndromes, Café au lait spots

INTRODUCTION

Tuberous sclerosis complex (TSC) or Bourneville's disease, is a rare genetic neurocutaneous disorder of autosomal dominant inheritance with the prevalence of 1 in 6000 live birth, affecting both sexes and all ethnic groups. Neurocutaneous syndromes have cutaneous and CNS manifestation. Cutaneous manifestation can help in clinching the diagnosis of the syndrome like Tuberous sclerosis, Neurofibromatosis (1 and 2), Von hippel lindau diasease, Sturge-Weber Syndrome, Ataxia –Telangiectasia, PHACE,Incontinentia pigmenti can be easily diagnosed because of skin manifestations. Tuberous sclerosis complex (TSC) is a neurocutaneous syndrome. Cutaneous manifestation includes Café au lait spot, ash leaf macule, shagreen patch, facial angiofibroma, gingival fibroma, hypomalenotic macule. CNS manifestation includes cortical tubers, subependymal nodule. Hypomelanotic macules are present at birth and almost all lesions are evident within the first two years of life. Facial angiofibromas (adenoma sebaceum) are present during preschool years in the malar area as small pink to red dome shaped papules in a “butterfly distribution”. The shagreen patch is found in the lumbosacral region characteristically present as an irregularly roughened raised lesion with orange peel consistency. It is clinically characterized by triad of epilepsy, Intellectual disability and adenoma sebaceum. The hallmark of TSC is the involvement of the central nervous system. Brain MRI is the best way of identifying cortical tubers, which can form before birth. Subependymal nodules are lesions found along the wall of the lateral ventricles where they undergo calcification and project into the ventricular cavity, producing cradle-dripping appearance. These lesions do not cause any problems; however in 5-10% of cases these benign lesions can grow into subependymal giant cell astrocytomas.

Case Study

A 3 months old male child from Anjum colony, Juhapura, Ahmedabad presented to hospital with history of multiple episodes of generalized tonic-clonic seizure for 3 days. Which was associated with up rolling of eyeball, deviation of mouth at left side,tightning of upper limb. He was born out of non-consanguineous marriage with uneventful birth history. There was no history of seizure in family members. Development was appropriate for age (Social smile, recognize mother, cooing sound) .On general examination there were one large café au lait spot and four hypopigmented macules (Ash leaf macule) which suggested possibility of tuberous sclerosis.

Laboratory investigations were within normal limits. CNS examination was normal with no intellectual disabilities. Other systemic examination and fundus examinations revealed no abnormality. MRI brain with contrast shows cortical subcortical abnormal signal intensities in B/L fronto-temporo-parieto-occipital lobe possibility of cortical tuber.Multiple mildly enhancing subependymal nodules in both lateral ventricles possibility of subependymal hamartomas. Patient was treated according to standard protocol.

DISCUSSION:

Prevalence of tuberous sclerosis is 1 in 6000 to 10000 newborns. Tuberous sclerosis is an autosomal dominant genetic disorder and fresh mutations can be seen in 50% cases. Complications of neurological involvement are the most common causes of mortality and morbidity.

Awareness regarding cutaneous manifestations of these syndromes lead to early diagnosis. No significant family history of tuberous sclerosis in this patient suggests a new mutation. In this patient early presentation was noted.

CONCLUSION:

Cutaneous manifestation can help in diagnosing neurocutaneous syndrome. The quality of life depends on the neurological manifestation like seizures and mental retardation which is improved by multidisciplinary approach and symptomatic organ specific treatment.

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