Original Research Paper

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Pediatrics

TUBEROUS SCLEROSIS WITH RHABDOMYOMA: A CASE REPORT

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KEYWORDS :

INTRODUCTION-

Tuberous sclerosis, a neurocutaneous syndrome is inherited in an autosomal dominant manner with variable expression and a prevalence of 1 in 6,000 newborns.It has a wide clinical spectrum which varies from severe intellectual disability with intractable epilepsy to normal intelligence and lack of seizures. The disease affects many organs other than skin and brain including heart ,kidney, eyes ,lungs and bone. Approximately 50% of children with tuberous sclerosis have cardiac rhabdomyoma which may be detected by an echocardiogram[1]. We are reporting a case of a 9 months male child who presented with tuberous sclerosis with rhabdomyoma.

Case report:

A 9 months old male child presented with complaints of myoclonic jerks since last 1 month. 8-10 episodes per day. His birth history was normal. Development history revealed that he developed neck support at 6 months of age, neck support at 7 months and at 9 months of age he is able to creep and not able to sit without support. He was not able to speak monosyllables yet. Stranger anxiety was not developed yet. Family history revealed ashleaf macules in elder brother, mother, maternal grandmother and all of them are symptom free. On examination patient had multiple ash leaf macules all over the body. No other neurocutaneous stigmata was present. Patient was active, alert and playful and his vitals were normal. CNS examination brisk knee jerk. Rest of the examination was within normal limit. 2D echocardiography revealed mass measuring 25mm X 30 mm in left ventricular apex of heart which was single,round, homogeneous, hyperechogenic, intracavitary mass. CECT brain revealed multiple subependymal calcified nodules in bilateral lateral ventricle with a large calcified nodule at foramen of munro of right lateral ventricle measuring upto 12x9 mm. EEG done which was reported normal. USG KUB was also normal. Patient was started on vigabatrin and patient became seizure free on 3rd day of treatment. As the patient was not having any cardiac symptoms ,we have not done any intervention. We could not follow up the patient as they were from another state and did not visited our hospital later.

DISCUSSION:

Cardiac tumours are very rare in children (0.027 to 0.17%) [2].The vast majority of primary cardiac tumours in children are benign, whilst approximately 10% are malignant [3] .Rhabdomyomas are the most common cardiac tumours in children (45%)[4] Approximately 50% of children with tuberous sclerosis have cardiac rhabdomyoma [1]. Echocardiography is the estabilished primary diagnostic tool for the evaluation of cardiac tumors in children [5]Rhabdomyomas appear on echocardiography as round, homogeneous, hyperechogenic, intramural or intracavitary masses, sometimes multiple predominantly localized within the ventricles but can be observed in the atria or caval veins and may lead to obstruction of cardiac valves or inflow/outflow tracts[6].Most of them are asymptomatic but may also cause atrial or ventricular arrhythmias, sinus node dysfunction, heart block and pre-excitation [7] .Surgical resection is not usually considered unless they cause severe intractable arrhythmias, valvular obstruction, or congestive heart failure[8].

The TSC mutation results in hamartomatous lesions that affect the brain, kidney, heart, and lungs. mTOR (mammalian target of rapamycin) is a protein kinase that regulates the abnormal cellular proliferation and differentiation. mTOR inhibitors are studied to treat the subependymal giant-cell astrocytomas and renal angiomyolipomas that are usually seen with TSC. Tibero etal had described a case of a patient with significant regression of a cardiac rhabdomyoma after receiving Everolimus, an mTOR inhibitor. This finding is suggestive of a possible novel therapy for patients with clinically significant cardiac rhabdomyomas. [9,10,11]

Etuwewe B etal ,Smythe JF etal reported cases of Tuberous sclerosis with cardiac rhabdomyoma which needed surgical intervention . But in other asymptomatic cases, surgical intervention is debatable as neonatal cardiac rhabdomyomas can spontaneously regress.[12,13]

CONCLUSION-

Cardiac rhabdomyoma , a rare pediatric cardiac tumour is found in Tuberous sclerosis. Most of them don't require intervention until unless it causes complications. Everolimus and surgical correction can be considered.

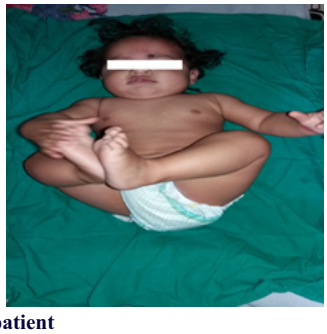

Figure 1-The patient

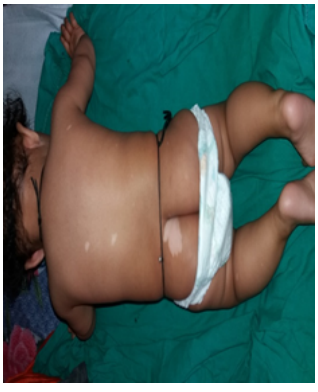

Figure 2-Ashleaf macules in back

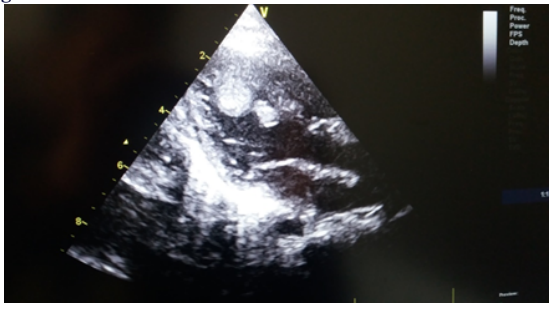

Figure 2-2D echo showing rhabdomyoma

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