EUROPEAN ACADEMIC RESEARCH
Vol. IX, Issue 7/ October 2021

Impact Factor: 3.4546 (UIF)
DRJI Value: 5.9 (B+)

ISSN 2286-4822
www.euacademic.org

Tuberous Sclerosis with Intestinal Obstruction: Case Report and Literature Review

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Abstract

Tuberous sclerosis (TS) Epiloia or Bournervlle's Disease is one of the important neurocutaneous syndrome characterised by abnormalities and involvement of other organs, including heart, kidney, lungs, bone and gastrointestinal tract. The frequency is (1/6000-1-12000) (1-2). Molecular studies have identified 2 foci for TSC; TSC 1 gene and TSC 2 gene, the loss of either TSC 1 gene or TSC 2 gene result Information of numerous benign tumors (hamartomas). Here we represent the first presentation of 9 years old child with exceptional Presentation of TSC with intestinal obstruction which is mainly caused by intestinal band and meckle's diverticulum.

Keywords: TS Tuberous sclerosis, meckle's diverticulum, (hamartomas)

INTRODUCTION:

Von Recklinghausen first described tuberous sclerosis in 1862. , later Desire-Magloire Bourneville (a French physician) Suggest the term sclerose tubereuse . (3,4). Sherlock describe the term EPILOIA the clinical triad of tuberous sclerosis (Epi: epilepsy, Loi: low intelligence,

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A: adenoma sebaceum) which present in 29% of cases. (5,6) Molecular genetic studies have identified 2 foci for TSC; TSC 1 gene located on chromosome 9q34, and the TSC 2 is on chromosome 16p13 .Within all these 2 proteins bind to one another and work together acting as tumor suppressor genes. The loss of either TSC 1 gene or TSC 2 gene are result in the formation of benign tumors (hamartoms) (7). Three types of brain tumors are associated with TSC, giant cell (strocytoma) , cortical tuberous and sub ependymal nodules .Between 60 -80% of TSC patients have benign tumors of the kidneys called Angiomyolipomas which is commonly multiple and bilateral, in the heart called rhabdomyomas. The dermatological sign is present in 96 % of individuals and include; hypomelanic macules (Ash leaf spot) , facial angiofibromas , unequal fibromas , fibrous cephalic plaques and shagreen patches. Clinical diagnosis is easy when the patient presents with classical triad of seizures, mental retardation and adenoma sebaceum. However, in a patient presenting with an incomplete form of tuberous sclerosis, mistakes in the diagnosis are possible.

CASE REPORT:

A 9 years old child was admitted to the Department of pediatrics at Omdurman Military hospital on June 2021 with abdominal pain for 8 hours, around umbilicus, started mildly then become severe associated with vomiting , abdominal distension and dermatological lesions. There is past history of a febrile convulsion over the last 14 days. On clinical examination lesions were hyperpigmented and raised from the thick leathery skin on the forehead (1) , and hypomelanic macules in the back (2) , and facial angiofibroma (3) , then patient diagnosed as TSC based on dermatological manifestation plus Sub ependymal nodules in the brain MRI (4) . Patient looks ill, pale, not dysmorphic, abdomen tender and distended, no palpable organ. Investigations show normal CBC and U/S Abdomen, Abdominal x-ray show feature of Intestinal obstruction. The patient sent for an emergency laparotomy, abdominal exploration revealed perforated meckle's diverticulum with congenital band.

DISCUSSION

In the TSC Consensus Conference in Annapolis, Maryland 1998, (8,9) revised the criteria of diagnosis (The revised criteria (Table 1)), reflecting an improvement of understanding, the clinical manifestations of TSC and its genetic and molecular mechanisms. A diagnosis of TS is definite when two major features or one major and two minor features exist, probable if one major and one minor feature are present, and possible when more than two minor features or only one major feature is present. (10).

TS is thought to result from sporadic mutation in the majority of patients, since most patients have no family history of the disease (11).

The triad of symptoms of TS, as described by Vogt 13 , consists of seizure, adenoma sebaceum (facial angiofibroma), and mental retardation. Not all patients have this classic triad, however,and half of all patients are of normal intellect and a quarter do not have seizures 14 . Although facial angiofibromas are commonly described as the hamartomatous lesions of TS, hamartomas may involve virtually any organ.

The presented case is interesting because of simultaneous of 2 very uncommon condition; TSC and perforated meckle's diverticulum which is diagnosed mainly in this case intraoperatively, and unfortunately our patients did not sent the biopsy to histopathology. This case has not a similar existence in the literature.

Meckle's diverticulum is congenital anomaly of the gastrointestinal tract and is caused by incomplete obliteration of the omphalomesentric duct Between the 5th and 7th weeks of gestation, the duct attenuate and separates from the intestine. meckles' diverticulum has been conveniently referred to by the "rule of 2's , which is 2% of general population, usually located 2 feet proximal to the iliocecal value and 2 inches in length. Can contain 2 types of ectopic tissue, generally present before the age of 2 years, and are found twice as commonly in females. (12).

Symptom of meckle's diverticulum usually arise in the 1st or 2nd year of life, but intestinal symptom can occur in the 1st decade. The majorty of symptomatic meckles' diverticulum cause painless rectal bleeding, the stool is typically brick colored or currant jelly

colored .Less often it is associated with partial or complete obstruction , the mechanism of obstruction result from intra peritoneal bands Connecting residual omphalomesentric duct remnant to the ileum and umbilicus. This band causes obstruction by internal herniation or volvulus of the small bowel around the band. A meckle's diverticulum occasionally become inflamed and manifest similarly to acute appendicitis. These children are older with a mean of 8 years of age leading to perforation and peritonitis, as in case presentation.

Confirmation of meckle's diverticulum can difficult, the most sensitive study is meckle's Radionuclide- 99m pertechnetate, or exploratory laparoscopy.

The treatment of asymptomatic meckles' diverticulum is surgical excision, there is significant debate regarding the proper management of an asymptomatic meckle's diverticulum and weather excision as observation is appropriate.

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FIGURE 1: Multiple angiofibromas on the Face

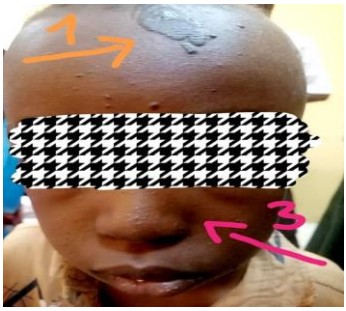


FIGURE-2-Hypomelanic macules in the back

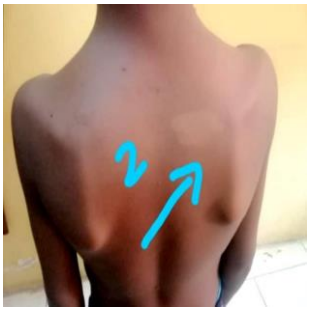


FIGURE -3-Multiple fluid level (Intestinal Obstruction)

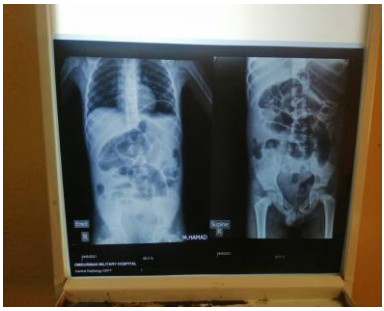


FIGURE 4: MRI Brain images showing the right & left Sub ependymal (T1,Low on T2)

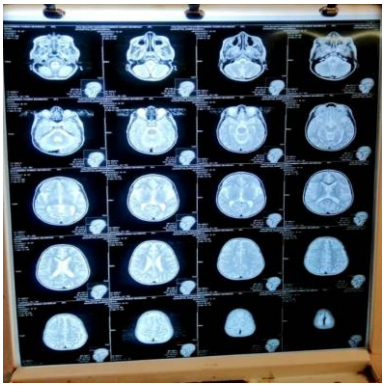
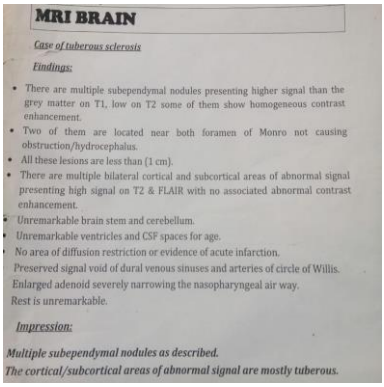


FIGURE-5-MRI Report



MRI BRAIN

Case of tuberous sclerosis

Findings:

- There are multiple subependymal nodules presenting higher signal than the grey matter on T1, low on T2 some of them show homogeneous contrast enhancement.
- Two of them are located near both foramen of Monro not causing obstruction/hydrocephalus.
- All these lesions are less than (1 cm).
- There are multiple bilateral cortical and subcortical areas of abnormal signal presenting high signal on T2 & FLAIR with no associated abnormal contrast enhancement.
- Unremarkable brain stem and cerebellum.
- Unremarkable ventricles and CSF spaces for age.
- No area of diffusion restriction or evidence of acute infarction.

Preserved signal void of dural venous sinuses and arteries of circle of Willis.
Enlarged adenoid severely narrowing the nasopharyngeal air way.
Rest is unremarkable.

Impression:

Multiple subependymal nodules as described.
The cortical/subcortical areas of abnormal signal are mostly tuberous.

Table 1. Diagnostic criteria for TSC:

Major criteria	Minor criteria
1 Facial angiofibroma	1 Non calcified subependymal nodules
2 Multiple ungula fibroma (Koenen tumours)	2 Hamartomatous, Rectal polyps
3 Cortical tuber	3 Gingival fibroma
4 Subependymal nodules	4 Non renal hamartomas
5 Multiple astrocytomas	5 Multiple renal cysts
6 Renal angiomyolipomas	6 Retinal achromic patch
7 Lymphangioliomyomatosis	7 -Enamel hypoplasia
8 Hypomelanotic macules (3 or more	8- Bone cysts
9 Cardiorhabdomyoma	9- Confetti's skin lesion
10 Forehead plaque	
11 Shagreen patches	
12 Retinal macular hamartoma	

Definitive diagnosis: 2 major features or one major + 2 minor features
Probable diagnosis: 1 major + 1 minor features