CASE REPORT

Tuberous Sclerosis: An Incidental Finding in A Woman with Long Standing Abdominal Distension and Heartburn

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ABSTRACT

A 38-year old female patient was admitted into the Department of Medicine of Shaheed Suhrawardy Medical College Hospital, Dhaka, Bangladesh, with the complaints of abdominal distension and retrosternal burning sensation for last eight months. She also gave history of previous hospitalization for the same reason. She also had complaints of an episode of seizure two years back. However, she was treated as a case of peptic ulcer disease in her previous admission in the district hospital. On clinical examination, adenoma sebaceum were observed on her face; hepatomegaly, and bilateral mid- and lower-abdominal masses were also found. She was sent for radiological investigations in the Department of Radiology & Imaging of the same hospital. The ultrasonogram of the abdomen showed multiple echogenic space occupying lesions in the liver along with bilateral echogenic kidneys and mild ascites. CT scan of the abdomen revealed hepatic angiomyolipomas, bilateral grossly enlarged and distorted kidneys having multiple angiomyolipomas, and sclerotic foci in the scanned part of bones. Moreover, CT scan of brain revealed multiple subependymal calcified nodules along with multiple sclerotic foci in the scanned part of bones. Those features were consistent with tuberous sclerosis. The patient was kept admitted and treated accordingly. After obtaining a written informed consent, she was presented as a special case in clinical education.

Keywords: *Tuberous sclerosis, radiological investigation*

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INTRODUCTION

Tuberous sclerosis (TS), also known as tuberous sclerosis complex (TSC) or Bourneville disease, is a phakomatosis (neurocutaneous disorder) characterized by the development of multiple benign tumours in multiple organs.[1-4] Tuberous sclerosis has an incidence of 1:6000-12,000, with most being sporadic.[1,2] Tuberous sclerosis was classically described as presenting in childhood with a pathognomonic triad (Vogt triad) of seizures, intellectual disability and adenoma sebaceum.[1-4] However, the full triad is only seen in a minority of patients (~30%). Therefore, diagnostic criteria have been developed to aid the diagnosis of definite tuberous sclerosis complex based on either 2 major features or 1 major and 2 or more minor criteria.[5] Tuberous sclerosis is usually diagnosed in infancy

or early childhood because a child presents with seizures, developmental delay, or hypomelanotic macules. However, the diagnosis of tuberous sclerosis can be made earlier or later on the basis of other features that manifest themselves at other ages. For instance, cortical/subcortical tubers and cardiac rhabdomyomas are detected prenatally and in infancy, while renal, pulmonary, and osseous lesions are identified more commonly in adulthood.[1,4] Here we have presented an incidental finding of tuberous sclerosis in a woman in her 40s with long standing abdominal distension and heartburn.

CASE SUMMARY

A 38-year old female patient was admitted into the Department of Medicine of Shaheed Suhrawardy Medical College Hospital, Dhaka, Bangladesh, with the complaints of abdominal distension and retrosternal burning sensation for last eight months. She also gave history of previous hospitalization for the same reason. She also had complaints of an episode of seizure two years back. However, she was treated as a case of peptic ulcer disease in her previous admission in the district hospital. On clinical examination, adenoma sebaceum (fibrous papules) were observed on her face (Fig. 1); hepatomegaly, and bilateral mid- and lower-abdominal mass were also found. Then requisition was given for her radiological investigations in the Department of Radiology & Imaging of the same hospital. Ultrasonogram and CT scan of the whole abdomen as well as CT scan of brain were done. The ultrasonogram report showed multiple echogenic space occupying lesions (SOLs) in the liver along with bilateral echogenic kidneys and mild ascites. CT scan of the abdomen revealed hepatic angiomyolipomas, bilateral grossly enlarged and distorted kidneys having multiple angiomyolipomas, and sclerotic foci in the scanned part of bones (Fig. 2). Moreover, CT scan of brain revealed multiple subependymal calcified nodules along with multiple sclerotic foci in the scanned part of bones (Fig. 3A & 3B). Those features were consistent with tuberous sclerosis. The patient was kept admitted and treated accordingly. After taking her written informed consent, she was presented as a special case in clinical education.

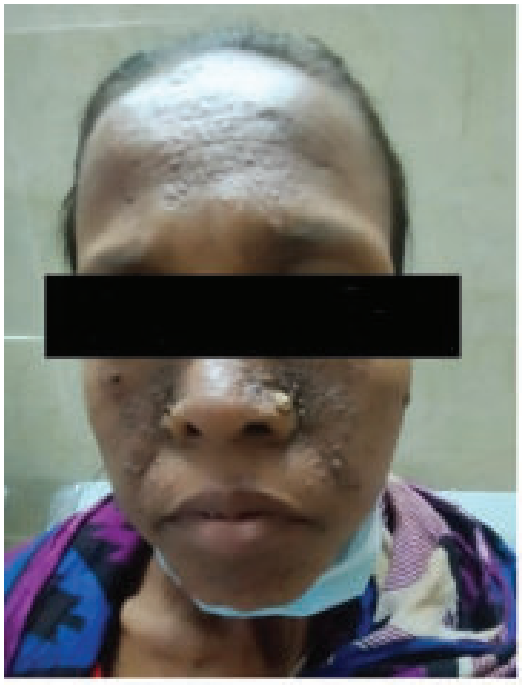

Fig. 1: *Adenoma sebaceum as observed on the face of the patient (facial angiofibromas).*

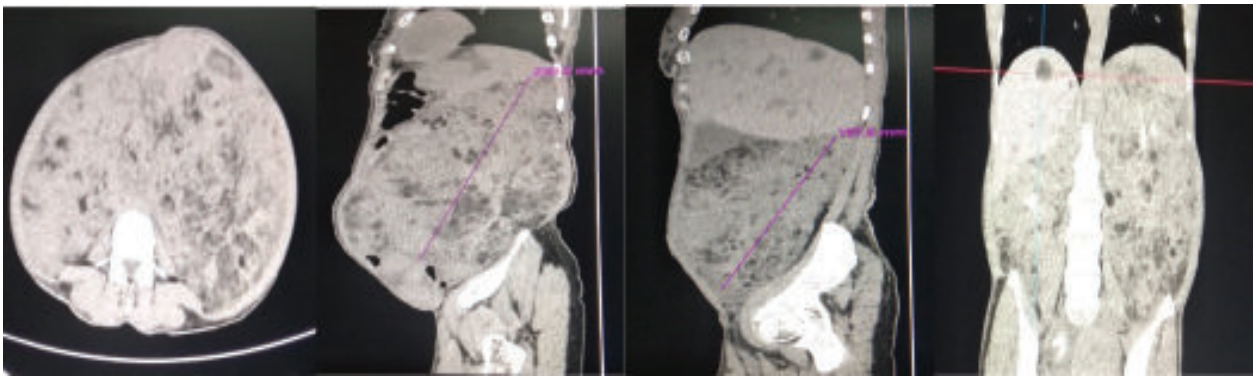

Fig. 2: *CT scan of the abdomen showing hepatic angiomyolipomas, bilateral grossly enlarged distorted kidneys having multiple angiomyolipomas, mild ascites, and sclerotic foci in the scanned part of bones.*

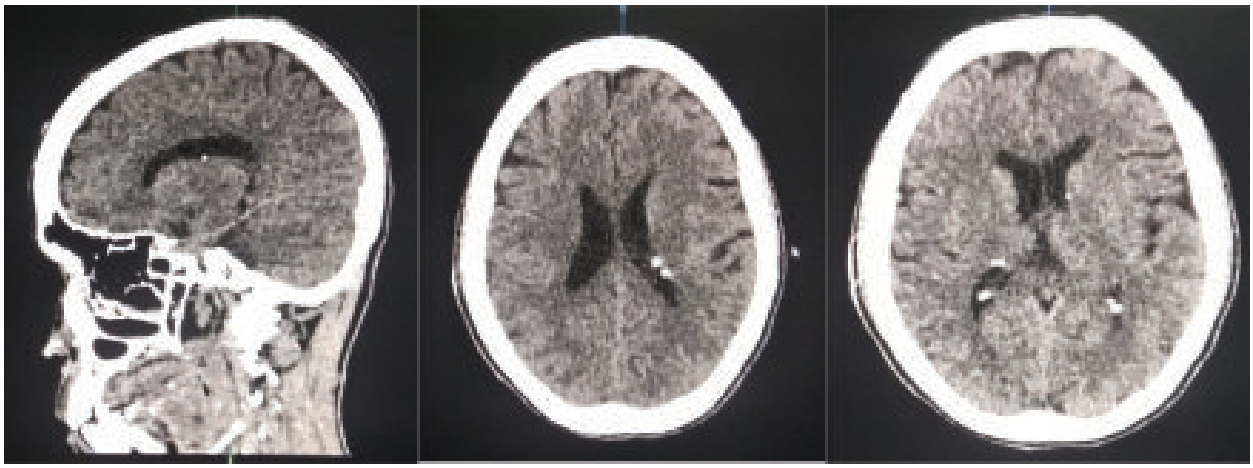

Fig. 3A: *CT scan of brain showing multiple subependymal calcified nodules.*

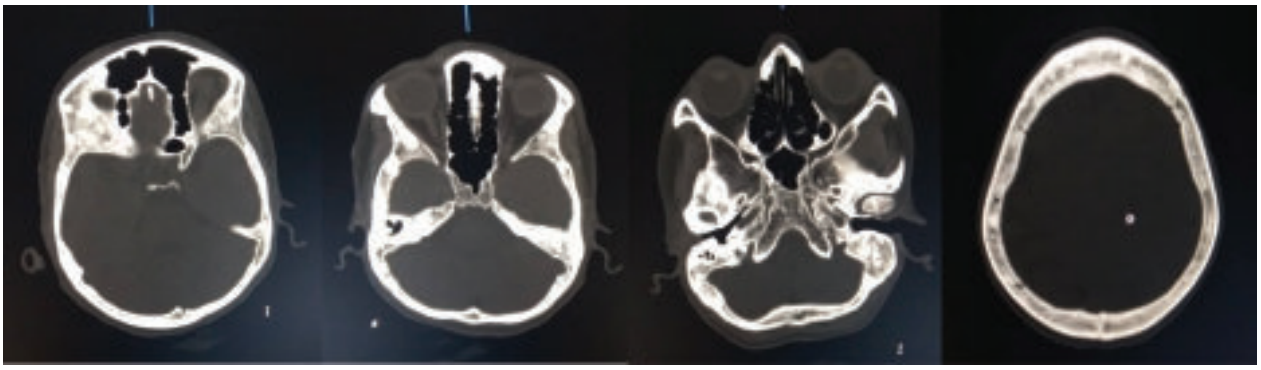

Fig. 3B: *CT scan of brain showing multiple sclerotic foci in the scanned parts of bones.*

DISCUSSION

Tuberous sclerosis is a rare autosomal dominant neurocutaneous syndrome characterized by the presence of benign congenital tumours in multiple organs.[1-4] The diagnosis is usually established based diagnostic criteria applied to physical or radiologic findings.[2,5] Because the classical triad of epilepsy, mental retardation, and adenoma sebaceum is uncommonly found at clinical examination, radiologic examinations usually play an important role in the diagnosis of tuberous sclerosis and in its treatment.[1,2] In our case, we observed adenoma sebaceum (fibrous papules) on patient's face. Adenoma sebaceum are solitary, dome-shaped, skin-colored to red papules located on the central face, usually around the nose and on the malar eminences. In tuberous sclerosis, angiofibromas typically arise symmetrically on the cheeks, nasolabial folds, nose, and chin. They can start off as erythematous macules that form into the red to red-brown papules that can coalesce into plaques.[1-4] Hepatic and renal angiomyolipomas were found in our case. Angiomyolipomas are the most common benign mesenchymal neoplasm and are composed of varying amounts of fat, smooth muscle, and blood vessels. Angiomyolipomas are present in 80% of patients with tuberous sclerosis.[1,6-8] Our patient also had cortical tubers and subependymal nodules. Cortical/ subcortical tubers are one of the most common causes of epilepsy, which is sometimes medically intractable.[1,3,9,10]

Although there is no cure for tuberous sclerosis, treatment can help manage specific symptoms. For example, anti-seizure medications may be prescribed to control seizures. Other medications may help manage heart arrhythmias (if present), behaviour problems or other signs and symptoms. If a growth affects the ability of a specific organ (e.g., liver, kidney or heart) to function, the growth may be surgically removed. Surgical procedures such as dermabrasion or laser treatment may improve the appearance of

skin growths. A mental health provider can also help address behavioural, social or emotional issues and recommend treatment as well as resources.[9,10]

CONCLUSION

Tuberous sclerosis has a significant number of manifestations, involving many organ systems. The clinical course of the disease and patient prognosis depend on the sites of manifestations. Familiarity with the diverse clinical and radiological features facilitates diagnosis and helps in treatment planning and monitoring response to treatment of this multisystem disorder. Tuberous sclerosis is a lifelong condition that requires careful monitoring and follow-up because many signs and symptoms may take years to develop. Early identification of problems can help prevent complications.

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