

Case Report

Tuberous Sclerosis Associated with Bilateral Ovarian Dermoid Cysts

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ABSTRACT Summary Tuberous sclerosis complex is an autosomal dominant multisystem disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. We present the case of a 24 years old woman, who fulfilled the diagnostic criteria for definite tuberous sclerosis, based on Revised Diagnostic Criteria for Tuberous Sclerosis Complex, with histologically confirmed subependymal giant cell astrocytoma, and multiple hypomelanotic macules shown on skin examination. The patient was also diagnosed with bilateral ovarian dermoid cysts which is a rare condition. To the best of our knowledge, there are not published cases of tuberous sclerosis associated with bilateral ovarian dermoid cysts.

KEY WORDS *Tuberous sclerosis, MRI, Dermoid cyst, Brain, Ovary*

Introduction

Tuberous sclerosis is a rare neurocutaneous autosomal dominant disorder with a recently estimated incidence of 1 in 6,000 live births. It occurs in both sexes and in all races and ethnic groups and has a varied clinical presentation [1]. We present a patient with tuberous sclerosis also diagnosed with ovarian dermoid cysts. As for the association between tuberous sclerosis and ovarian dermoid cysts, there are not cases presented in the literature.

Case report

A 24-year-old woman complaining of diffuse headache, transitory diplopia, loss of balance and vomit was admitted to our hospital. The patient was born of non-consanguineous marriage and had no history of inherited diseases. On admission, neurological examination showed that the patient was conscious with normal intelligence; she had mild muscle hypotonia without motor deficiency. General examination, in particular skin examination revealed multiple hypomelanotic maculae, on the arms and face.

On non-contrast computer tomography (CT) is seen an isodense mass with minimum mass effect and peripherally located nodular calcifications, occupying the frontal horn and partially the central part of left lateral ventricle, extending in the region of foramen of Monroe (**Figure 1a**). Enhancement is intense and homogeneous (**Figure 1b**). On magnetic resonance imaging (MRI) the tumor is heterogeneous both on T1 and T2 weighted images. On T1 weighted (T1W) the lesion is isointense (**Figure 2a**). On T2 weighted (T2W) besides hyperintense foci, hypointense areas are seen (**Figure 2b**). Intense but

inhomogeneous enhancement can be seen after contrast administration (**Figure 2c**). Moderate hydrocephaly was found, but no obvious edema. The patient received a craniotomy for a subependymal giant cell astrocytoma (SGCA), which was confirmed histological.

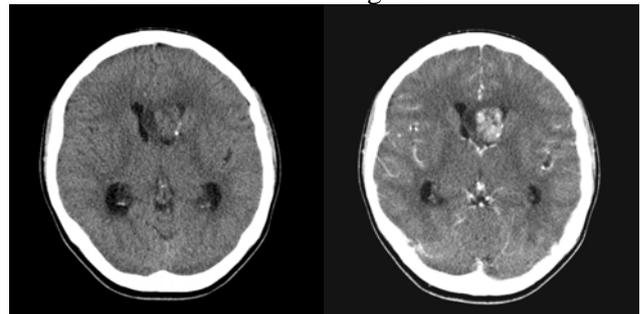


Figure 1: Multislice CT images showing an isodense intraventricular mass with peripherally calcifications (a) and intense enhancement (b).

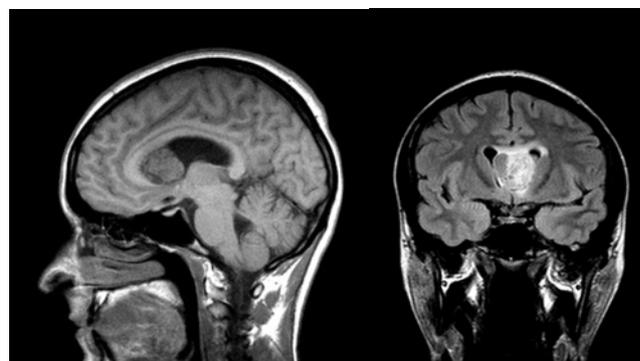


Figure 2: Non-contrast Sag T1W (a), Cor T2W FLAIR (fluid attenuation inversion recovery) (b) and contrast enhanced Ax T1W (c) MRI exam shows a mass in the left lateral ventricle, isointense on T1W, high heterogeneous signal on T2W and intense inhomogeneous enhancement.

After 6 months the patient was admitted again in our hospital for pain in the lower abdomen, a palpable mass on abdominal examination and abnormal vaginal bleeding. The MRI exam showed bilateral ovarian mass, larger on the left ovary, with cystic, fat and tissue elements on T1W, T2W fat suppression images (**Figure 3a**). The tumours had septa and well-delineated thin wall. There was regular enhancement of tumours' outlines and septa (**Figure 3b**), no enhancement of the solid tissue, no invasion in the adjacent structures or lymph nodes, all in favours of benign tumours. The diagnosis was mature bilateral ovarian dermoid cysts. The patient underwent surgical intervention and the histopathology exam confirmed the diagnosis.

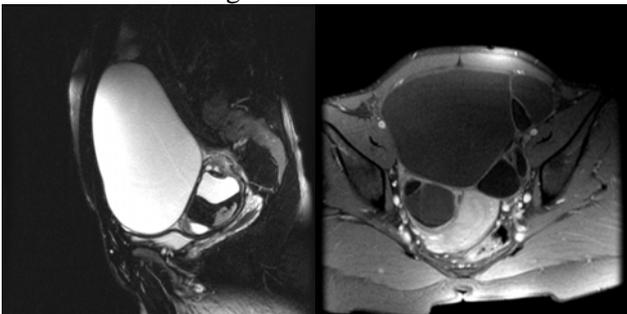


Figure 3: Noncontrast Sag T2W (a) and contrast enhanced Ax T1W (b) with fat suppression images shows bilateral ovarian mass, larger on the left ovary, with heterogeneous signal and uniform enhancement at the level of septa and tumours' wall. A marked mass effect with deviation of the bladder and uterine structures was apparent.

Discussion

We described a woman of 24 years with normal intelligence and no personal or family history of clinical manifestations of tuberous sclerosis, who presented with a neurological syndrome not typically associated with this ovarian disorder. Our patient fulfilled the diagnostic criteria for definite tuberous sclerosis [2], based on Revised Diagnostic Criteria for Tuberous Sclerosis Complex, which includes as major features the subependymal giant cell astrocytoma and hypomelanotic macules (more than three). Also the diagnosis of dermoid cysts was confirmed histopathologically.

TS is an autosomal dominant multisystem disorder and mutations in one of two genes, TSC1 on chromosome 9q34 and TSC2 on chromosome 16p13 [3], have been identified as causes for this disease. The typical manifestations of TS include skin disorders, brain, eye and heart lesions, and a classical clinical triad (seizures, mental retardation, and adenoma sebaceum) [4].

However, many patients with TSC are asymptomatic and of normal intelligence [5].

The most commonly identified brain lesions in TS are cortical tubers, subependymal nodules, SGCA, and white-matter abnormalities [6]. The SGCA can be diagnosed macropathologically, histopathologically, or immunohistochemically. In patients with SGCA, CT scan often shows a soft tissue with a clear margin protruding into the ventricles from the ependymas. Necrosis and calcification can be found inside. On MRI, the tumour is a relatively large mass which is isointense on T1W and hyperintense on T2 W. Usually, obstructive hydrocephalus can be found. By contrast-enhanced MRI, the tumour is homogeneous or heterogeneous; and subependymal nodules (31%), cortical nodules (3%), and white matter lesions (12%) may be found [3]. According to a previous report of Cuccia and colleagues [7], a SGCA can be suspected when the tumour is over 12 mm in diameter, the tumour tends to increase in size, or the patient has complicated hydrocephalus.

Mature cystic teratomas of the ovary account for 10-25% of all ovarian neoplasms and they are bilateral in 8-15% of cases [8, 9]. Teratomas most commonly are found during the reproductive years, with a peak incidence in the second and third decades [10]. Diagnosis is usually not difficult with the use of ultrasound (US), CT, and MR imaging [11]. The US diagnosis is complicated by the fact that these tumours may have a variety of appearances. At CT, fat attenuation within a cyst, with or without calcification in the wall, is diagnostic for mature cystic teratoma. At MR imaging, the sebaceous component of dermoid cysts has very high signal intensity on T1-weighted images, and variable signal intensity on T2-weighted images. MR imaging with frequency-selective fat saturation allows accurate differentiation between teratomas and hemorrhagic cysts.

In conclusion, TS is a complex multisystem disorder that may present in many ways. We report an unusual presentation of tuberous sclerosis manifesting with clinical and imaging signs suggesting a cerebral lesion associated with bilateral ovarian dermoid cysts. Our case expands the association of various tumours and TS and emphasizes the possibility that dermoid cysts may be a component of this syndrome. Radiology plays an important role in the diagnosis of new cases, confirming the diagnosis in clinically suspected cases, and also in establishing the extent of organ involvement in TS patients.

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