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Tuberous Sclerosis Complex

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Abstract: ***Tuberous sclerosis complex (TSC) is a rare neurocutaneous disorder that is defined by the presence of benign hamartomas. Renal angiomyolipomas (AML) are a known entity associated with TSC and are usually asymptomatic but may cause hemorrhage, the risk of which has been associated with tumor size, tumor growth, and presence of aneurysms >0.5 cm. We present the case of a patient with tuberous sclerosis complex suspected from finding bilateral renal angiomyolipomas on ultrasound. The patient was admitted for hematuria and worsening renal function. Abdominal magnetic resonance confirmed bilateral renal angiomylipomas and head computed tomography demonstrated numerous calcified subendymal nodules. Conclusion: Finding renal angiomyplipoma should may you thinking for further research if AML is associated or not with TSC.***

Keywords: Tuberous sclerosis complex(TSC), renal angiomyolipoma, subependymal calcified nodules, abdominal magnetic resonance

1. Introduction

Tuberous sclerosis complex, also known as **Bourneville disease** is a neurocutaneous disorder (phakomatosis) characterized by development of multiple benign tumors of the embryonic ectoderm (e.g. skin, eyes, and nervous system). It is a genetic disorder that causes non-malignant tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs[1,2]. It can be inherited from one parent with TSC or can result from spontaneous genetic mutation. Children have 50% chance of inheriting TSC if one of their parents has this condition [3]. Two genes have been indentified that can cause TSC[4]. Only one of the genes needs to be affected for TSC to be present. The TSC 1 gene is located on chromosome 9 and is called the hamartin gene. The other gene TSC2 is located on chromosome 16 and is called the tuberin gene. Both the TSC 1 and TSC 2 genes suppress tumor growth in the body by carefully regulating cell growth through inhibition of a protein calles mammalian target of rapamycin or mTOR for short. When either the TSC 1 or TSC 2 gene is defective cell growth is not adequately suppressed and tuberous sclerosis complex results. Hamartin, tuberin and mTOR are expressed in many different organs throughout the body, which explain why so many organs can be affected by TSC.[5]

2. Clinical Presentation

A 27 year old caucasian woman was presented to our department to perform an abdominal ultrasound. The main concern of physician was macroscopic hematuria. She had minor difficulties in communication which made us asking for her medical history. It was referred by her relatives that she was treated during her childhood for mild seizures with antiepileptic drugs.

3. Imaging Findings

First an abdominal ultrasound was performed. It demonstrated multiple hyper echoic lesions in both kidneys, located in the cortex and hypoanechoic lesions with posterior acoustic shadowing suggesting renal cysts. No other organ lesions were observed. This changes made us asking an abdominal MRI and previous examinations.

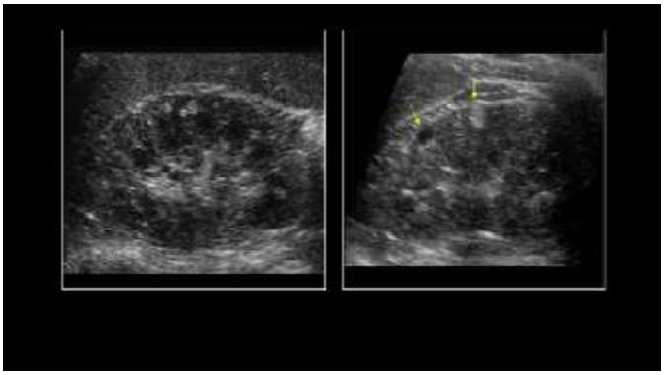

Figure 1: Multiple hyperechoic lesions suggesting angiomyolipomas in both kidneys.

On previous examination (CT-scan of the head) we noticed multiple calcified subependimal nodules.

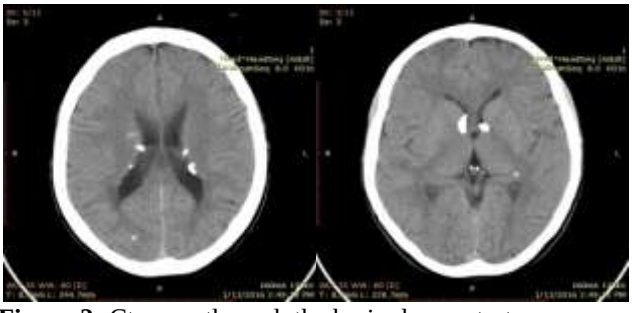

Figure 2: Ct scans through the brain demonstrate numerous calcified subependimal nodules. Calcified sub cortical tuber in the right occipital region

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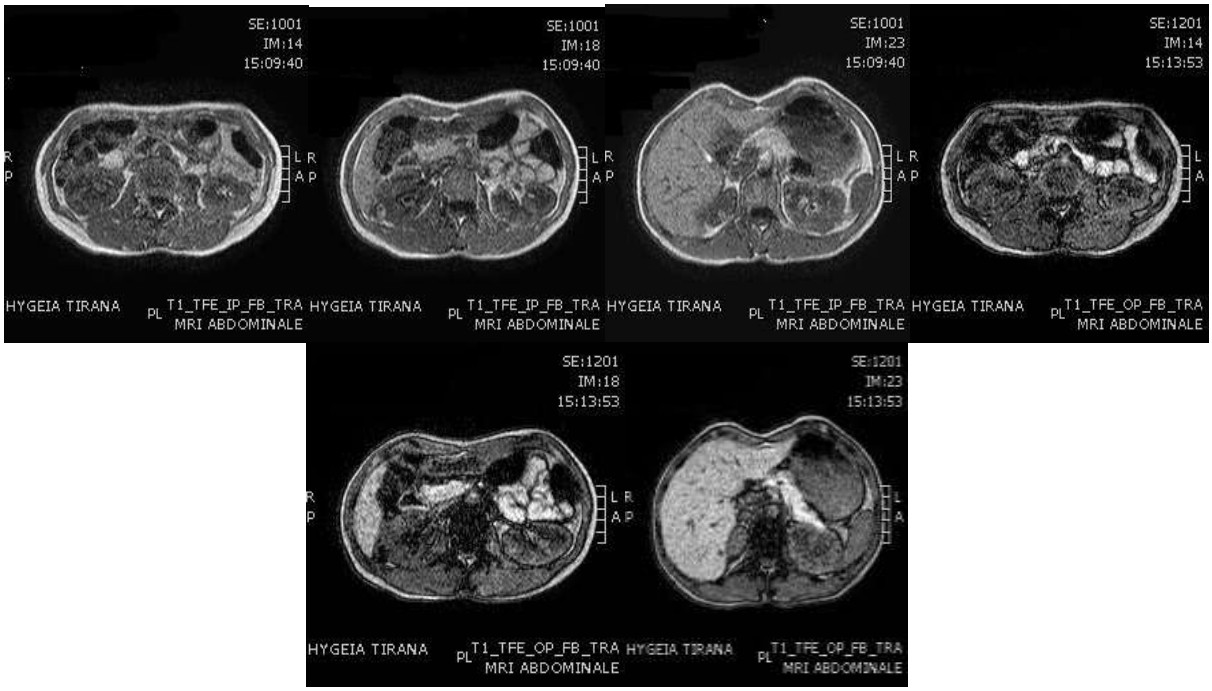


Figure 3: Abdominal MRI demonstrates: High signal intensity tumors on the in-phase images that become signal void on the out-of-phase image

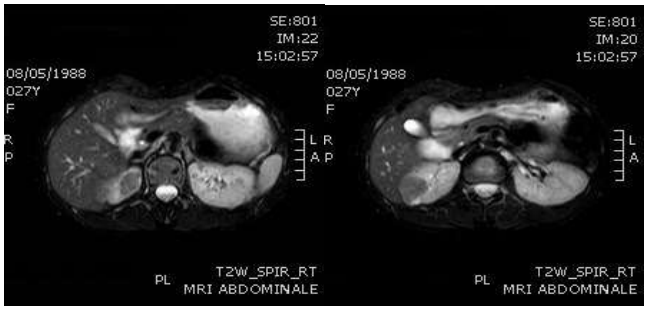


Figure 4: Fat suppressions sequences decrease the signal intensity of these lesions

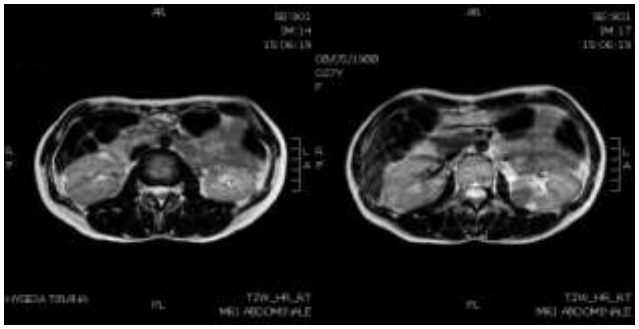
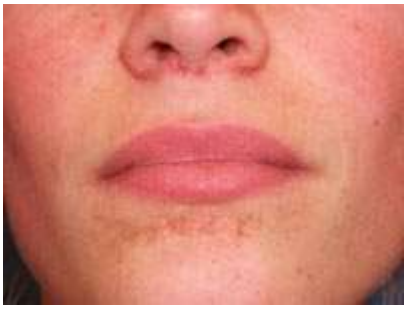


Figure 5: Multiple cysts are also present in both kidneys

On the **clinical examination** were found skin lesions angiofibromas which are facial rash that appears as a spread of small pink or red spots across the cheeks and nose in a butterfly distribution.



4. Disscussion

Angiomyolipomas are benign tumors composed of variable amounts of three elements:
1) Thick - walled blood vessels
2) Smooth muscle, and
3) Mature fat

The fat component is usually substantial, permitting characterization on combined T1 - weighted regular (in phase) and fat - suppressed images or combined T1 - weighted in – phase and out - of - phase SGE . Although benign, these tumors may increase in size over time, with larger tumors having greater propensity to bleed. Angiomyolipomas have a greater tendency to increase in size when they are multiple than when they are solitary. Because TSC can manifest in so many different ways diagnosis is generally made when two major features are identified in one individual. Major feature include; specific non malignant tumor or growths such as subependymal nodules or subependymal giant cell astrocytomas in the brain, angiomyolipomas in the kidneys, lymphangioleiomyomatosis (LAM) in the lungs and tubers in the brain or hamartomas in the eye, cardiac rhabdomyoma, abnormal skin growths or skin pigmentation.

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5. Conclusion

Nearly 1 million people worldwide are estimated to have TSC,more often in women (4:1) which suggests hormonal activity influence on the tumor growth[6]. The risk of bleeding increases with the size of a tumor and is estimated for 13% in smaller tumors (up to 4 cm) and 51% in case of tumors of the diameter exceeding 4 cm. In the majority of patients with lesions up to 4 cm clinical symptoms do not occur. These patients remain in observation and have CT examination performed once yearly. Asymptomatic AML greater than 4 cm must be regularly – every 6 months – assessed in CT.

Many cases may remain undiagnosed for many years or decades due to the relative obscurity of the disease and the mild form symptoms. People with TSC will live a normal life span. There can be complications in some organs such as the kidneys and brain that can lead to severe difficulties an even death if left untreated. To reduce these dangers, people with TSC should be monitored throughout their life by their physicians for potential complications.

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