Scholars Journal of Medical Case Reports **ISSN 2347-6559 (Online)**
Sch J Med Case Rep 2016; 4(2):102-104 **ISSN 2347-9507 (Print)**

(An International Publisher for Academic and Scientific Resources) DOI: 10.36347/sjmcr.2016.v04i02.014

A Lady with Tuberous Sclerosis with Abdominal Distension

Abhinav Gupta[1], Radhika Govil[1], Atiq Ur Rehman[2], Preshant Choubey[3]
[1]Post Graduate student in Medicine, [2]MD Medicine, Assistant Professor, [3]MD Medicine, Associate Professor,
Department of Medicine, Peoples College of Medical Sciences And Research Centre, Bhopal (M.P), India

***Corresponding author**
Dr. Abhinav Gupta
Email: abhinavguptasmims@gmail.com

Abstract: Tuberous sclerosis also called tuberous sclerosis complex (TSC), is a rare genetic disease that causes benign tumors to grow in body, resulting in a variety of hamartomatous lesions that may affect virtually every system of the body. Tuberous sclerosis is often detected during infancy or childhood. Some people with tuberous sclerosis have such mild symptoms that it couldn't diagnosed until adulthood, while others experience serious disabilities. The signs and symptoms of tuberous sclerosis widely vary, depending on where the tumors develop and how severely a person is affected. The best-known cutaneous manifestation of TSC is adenoma sebaceum, which often does not appear until late childhood or early adolescence.
Keywords: tuberous sclerosis, Renal Angiomyolipoma,adenoma sebaceum

INTRODUCTION

First described in the 1880s, "tuberous sclerosis complex" also called as "Bourneville's disease" named after the French physician Desire-Magloire Bourneville. Tuberous sclerosis is a neurocutaneous autosomal dominant disorder with an estimated prevalence of 9/100000 population and a varied clinical presentation which affects multiple organs like skin, brain, kidneys, eyes, lungs, teeth, etc. However, some organs are involved more than other [1]. Neurological presentation of tuberous sclerosis occurs typically in children with seizures and intellectual impairment .However approximately 50% of patients who fulfil the diagnostic criteria have normal intellect and 15% remain free from seizures [2]. Genetic studies have shown that two thirds of cases do not have affected parents, and the disease results from a new dominant mutation either in the TSC1 gene on chromosome 9q343 or the TSC2 gene on chromosome 16p13with the latter accounting for an estimated 78% of cases[3, 4].

Here we report a case of 48 yr old female came to our hospital with complains of abdominal distension.

CASE REPORT

A 48 year old female came to the hospital with complain of abdominal distension from the past 10 years. Patient was able to feel two distinct masses in abdomen which started from lower abdomen and was gradually progressive to reach the upper abdomen. This was associated with nausea, vomiting, pain abdomen, reduced appetite and weight loss.

In the past, patient had history of seizures till the age of 7 yrs after which it got subsided on its own without any medication. She had 6 children out of which 3 children had history of seizures, in which 2 are dead and one is still alive. Parents and siblings have no such history.

On examination patient had multiple hyperpigmented papules over nose and cheeks(FIGURE 1),hyperkeratosis in nail beds of fingers and toes(FIGURE 2). Two distinct lump in either half of the abdomen was palpated which are present in right and left lumbar, iliac, and hypogastria region for which MRI was done which showed large heterogeneous masses from both kidneys occupying almost entire abdomen(FIGURE 3), mixed fat and soft tissue intensity was noted in the masses which most likely represent Renal Angiomyolipoma. For hyperpigmented papules on face biopsy was done which was suggestive of adenoma sebaceum. In due course routine biochemical investigations were done which came out to normal except for hypoproteinemia. With all the findings the final diagnosis of tuberous sclerosis with renal Angiomyolipoma was made. As the patient had financial constrains so the patient couldn't continue the treatment in our hospital and was referred to government hospital.

Fig.1:hyperpigmented papules over nose and cheeks (adenoma sebaceum)

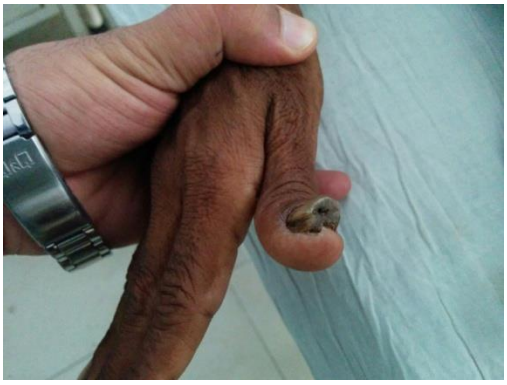

Fig.2: hyperkeratosis in nail beds

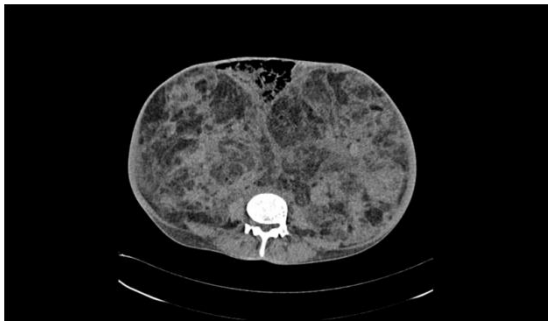

Fig.3: MRI Abdomen showing renal angiomyolipoma

DISCUSSION

TS is a rare multisystem autosomalinherited disorder that causes non-malignant tumours to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. TS arecaused by a mutation of either of two genes, TSC landTSC2, which code for the proteins hamartin and tuberin respectively. These protein sactin a complex as growth suppressors by inhibiting the activation of a master, evolutionarily conserved kinase called mTOR. Loss of regulation of mTOR occurs incells lacking either hamartin or tuberin, and this leads to abnormal differentiation and development, and to the generation of enlarged cells [5].

The benign, non-invasive lesions of tuberous sclerosis canappear in any organ like the brain, heart, skin, eyes, kidney, lung, and liver. Therefore, TS has a wide clinical spectrum. The diagnosis of definitive TS is based on specificclinical features and requires the presence of two majorcriteria, or one major and two minor [6]. Pulmonary lymphangioleiomyomatosis, renal Angiomyolipoma andfacial angiofibroma are some of the major clinical features. The most frequent cause of death in patients with TS isrenal complication [6, 8]. Multifocal, bilateral Angiomyolipoma are found in about 70-90% of adult patients [6],and the prevalence increases with age, being less frequentin children [3,4]. These lesions are more often prevalentin women, suggesting a hormonal component to thetumour growth [9]. The Angiomyolipoma are composed ofvarying amounts of mature adipose tissue, smooth muscle, and abnormal blood vessels [6,9]. The demonstrationof intratumoral fat with negative attenuation values at CTis virtually pathognomonic of Angiomyolipoma. Thin-section unenhanced CT is essential to visualize the fat content of Angiomyolipoma [10]. Progressive enlargement oftumours and haemorrhage into the lesion can result in flankpain, a palpable tender mass and gross or microscopichematuria, and interfere with renal function [9]. Tumourslarger than 4 cm in diameter have a greater risk of spontaneous or traumatic rupture resulting in hemorrhagic complications [9], which is the most common cause of deathin patients with TS [11]. Some patients with TS carry a contiguous germline deletion that affects both the TSC2 geneand the adjacent gene, polycystic kidney disease type 1(PKD1), resulting in a polycystic kidney phenotype thatleads to early renal insufficiency [6,7]. In our patient, as there is no family history which indicates that she might have acquired mutation in the TSC2 gene. Renal cell carcinoma canoccur in approximately 2-3% of adults with TS [6].

The principal goal of AML management is preservation of renal tissue and prevention or treatment of symptoms, particularly haemorrhage, which can be life threatening. Treatment methodology is decided basedon the size of the tumour, symptoms,and rate of growth, complications, andthe degree of diagnosis certainty onradiologic results. Selective arterialembolization has shown itself effective either in the treatment of haemorrhage or as the initial treatment of AML. However, the disadvantage of this method is its inability to provide tissue for histological examination; therefore, it should not be undertaken as the only treatment unless there is a very high degree of diagnostic certainty. Regarding surgical treatment, tumorectomy, partialnephrectomy, ortotal nephrectomymay be performed [12]. Total nephrectomy should be performed sparingly—in cases

of uncontrollable bleeding, central tumours, massive tumours, presence of extensive necrosis, or when there is a diagnosis of RCC in the same kidney [12]. Recently, cryotherapy and radiofrequency ablation have been suggested as therapeutic options.

CONCLUSION

Tuberous sclerosis complex (TSC) is an autosomal dominant disorder characterized by hamartomatous tumours that involve multiple organ systems. Approximately 80% of patients with TSC develop renal angiomyolipoma (AML).

The principal goal of AML management is preservation of renal tissue and prevention or treatment of symptoms, particularly hemorrhage.

Cryotherapy and radiofrequency ablation have recently been suggested as therapeutic options.

REFERENCES

1. Osborne JP, Fryer A; Epidemiology of Tuberous Sclerosis. NY Acad Sci., 1991; 615: 125-7.
2. Roach ES, Gomez MR, Northrup H; Tuberous sclerosis complex consensus conference: revised clinical diagnostic criteria. JChild Neurol., 1998; 13:624–8.
3. Van Slegtenhorst M, de Hoogt R, Hermans C, etal; (theTSC1 Consortium). Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. Science, 1997; 277:805–8.
4. The European Chromosome 16 Tuberous Sclerosis Consortium; Identification and characterization of the tuberous sclerosis gene on chromosome16. Cell, 1993; 75:1305–15.
5. http://ninds.nih.gov/disorder/tuberous_sclerosis. Retrieved on November 2013.
6. Curatolo P, Bombardieri R, Jozwiak S; Tuberous sclerosis. Lancet, 2008; 372(9639):657-68.
7. Crino PB, Nathanson KL, Henske EP; The tuberous sclerosis complex. N Engl J Med., 2006; 355(13):1345-56.
8. Tuzel E, Kirkali Z, Mungan U, Culer C, Sade M; Giant Angiomyolipoma associated with marked pulmonary lesions suggesting lymphangioleiomyomatosis in a patient with tuberous sclerosis. IntUrolNephrol., 2000; 32(2):219-22.
9. Wright T, Sooriakumaran P; Renal Angiomyolipoma presenting with massive retroperitoneal haemorrhage due to deranged clotting factors: a case report. Cases J., 2008; 1(1):213.
10. Prasad SR, Surabhi VR, Menias CO, Raut AA, Chintapalli KN; Benign renal neoplasm's in adults: cross-sectional imaging findings.AJR Am J Roentgenol., 2008; 190(1):158-64.
11. Yoshida S, Hayashi T, Ishii N, Yoshinaga A, Ohno R, Terao T, Watanabe T, Yamada T, Osada H; Bilateral renal Angiomyolipoma coexistent with pulmonary lymphangioleiomyomatosis and tuberous sclerosis. Int Urol Nephrol., 2006; 38(3-4):413-5.
12. Schneider-Monteiro ED, Lucon AM, de Figueiredo AA, et al; Bilateral giant renal Angiomyolipoma associated with hepatic lipoma. Rev HospClinFac Med Sao Paulo, 2003; 58:103-108.