

**TUBEROUS SCLEROSIS IN PREGNANCY**

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**Abstract**

Tuberous sclerosis is an autosomal dominant neurocutaneous disorder or neuroectodermatosis affecting multiple organ systems with variable clinical manifestations. We are reporting a case of a 26 years old female with history of epilepsy with mental retardation presented with fever and convulsions following an episode of stillbirth on first time with normal fetal outcome on second time. She had facial angiofibromas, shagreen patch, ash-leaf macules, periungual and subungual fibromas. She also had bilateral renal angiomyolipomas with haemorrhages and subependymal cortical tubers in brain. We report such a unique case having all clinically diagnostic physical signs of tuberous sclerosis with complicated obstetric history.

**Key words:** tuberous sclerosis; multisystem involvement; stillbirth; renal angiomyolipomas; cortical tubers

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**Introduction**

Tuberous sclerosis or Bourneville's disease is an autosomal dominant neurocutaneous disorder affecting multiple organ systems with various skin manifestations. It is characterised by presence of potato like tumours (Tuberous) in multiple organs. 30 percent of cases have Vogt's triad comprising of epilepsy, mental retardation and adenoma sebaceum. Many cases have been reported, but our case is unique in presence of nearly all major signs in a single patient, with association of a stillbirth [1].

We report such a unique case having all clinically diagnostic physical signs of tuberous sclerosis with complicated obstetric history.

**Case Report**

A 26 years old female presented with history of epilepsy since the age of 2 years with mental retardation. The patient had multiple asymptomatic skin lesions over face and back since the age of 7 years (Fig. 1A - C). No other family member had similar complaints or lesions. Patient presented with fever and generalised tonic clonic convulsions following an episode of stillbirth. The baby was stillborn with a large head and distended abdomen at 27 weeks of gestation. After two years she had normal healthy male child without any maternal complications. On examination, patient had angiofibromas (adenoma sebaceum) on face, shagreen patch over lumbosacral area, subungual and

periungual fibromas on right forefinger and middle finger. She had ash leaf macules on the back.

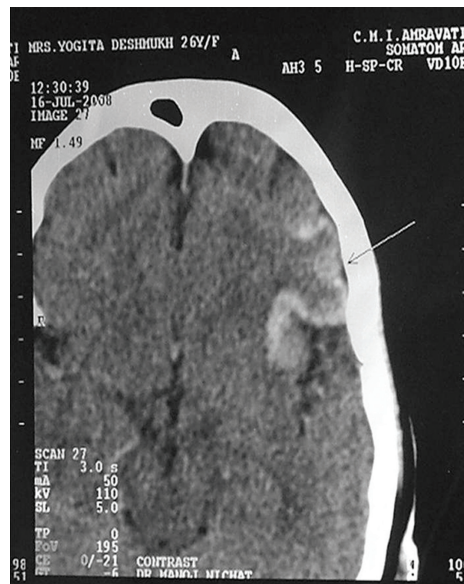
Patient's encephalogram was normal. Patient's Intelligent Quotient was 60, suggestive of mild mental retardation. Chest radiograph and radiograph of skull were normal. Ultrasonography of abdomen and pelvis showed large angiomyolipoma (with hemorrhages within) in upper pole of right kidney with multiple small sized angiomyolipoma in rest of both kidney with mild splenomegaly. Computed tomogram (CT) of brain showed calcified and enhancing subependymal cortical tubers along both lateral ventricles and Enhancing cortical tubers noted in left frontal lobe with gyriform enhancement of left frontal lobe suggestive of hamartoma (Fig. 2).

**Discussion**

Tuberous sclerosis is an autosomal dominant disorder. Two genetic loci have been identified in Tuberous Sclerosis Complex. The first gene, tuberous sclerosis complex-1 (TSC-1), maps to chromosome 9, specifically 9q34, and encodes the protein hamartin, which is a tumour suppressor gene. The second gene (TSC -2) maps to chromosome 16, specifically 16p13, and codes for tuberlin. Hamartin and tuberlin act synergistically to regulate cellular growth and differentiation. The desregulation in organogenesis results in hamartomas, which may affect any organ in the body [2].



**Figure 1A - C.** A. Facial angiofibroma, previously termed adenoma sebaceum, in a patient with tuberous sclerosis complex (TSC); B. A shagreen patch is a connective tissue hamartoma with a leathery texture and is found most commonly in the lower back region; C. Periungual fibroma on the thumb of a patient with tuberous sclerosis complex (TSC).



**Figure 2.** Frontal lobe hamartoma.

The characteristic Vogt's triad comprising of Epilepsy, Mental retardation and Adenoma sebaceum is present in 30% of patients. Gomez, in 1979 gave criteria for diagnosis of tuberous sclerosis. Gomez criteria include [3]:

**Primary Criteria: (one of the following)**

Adenoma sebaceum, Periungual fibroma,  
Cortical tubers, Retinal hamartomas.

**Secondary Criteria: (two of the following)**

Infantile spasms, Ash leaf macules,  
Shagreen patch, Bilateral renal AML,  
Cardiac Rhabdomyomas, single retinal hamartomas.

The Vogt's triad is present in our patient and Gomez criteria are also fulfilled confirming the diagnosis of tuberous sclerosis. Our case had all the major criteria of Gomez (Fig. 1A - C).

Our patient had a stillbirth at 27 weeks of gestation with large head and distended abdomen, most probably due to nonimmune hydrops fetalis or renal involvement. Tuberous sclerosis can lead to intrauterine fetal death. So it is important that the patient's genetic counseling has to be done and regular antenatal follow up is required if the patient conceives.

The renal angiomyolipomas may enlarge, rupture and bleed which can cause retroperitoneal haemorrhage and death. Renal angiomyolipomas, though major criteria, is uncommonly looked for. Ultrasonography should be done in all patients of tuberous sclerosis, to rule out renal angiomyolipomas (Fig. 3, 4).

There is no specific treatment for this disease. The seizures can be treated with carbamazepine, lamotrigine and vigabatrin. Surgical decompression can be tried for renal angiomyolipomas and ungual fibromas can be excised. Laser, cautery, diathermy and dermabrasion can be used for adenoma sebaceum and shagreen patch.

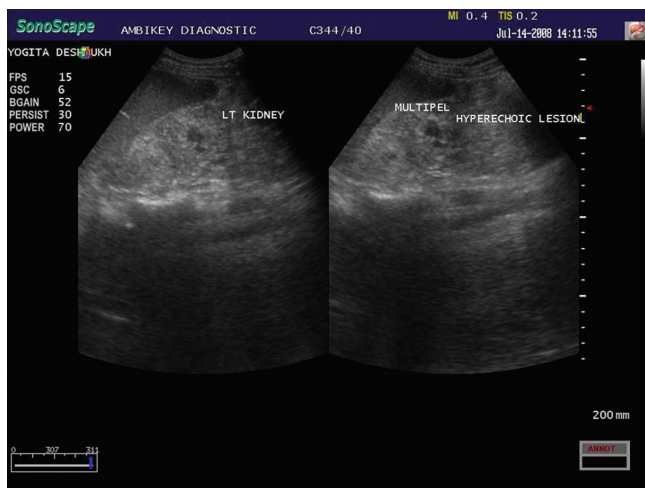


Figure 3. Renal angiomyo lipoma.

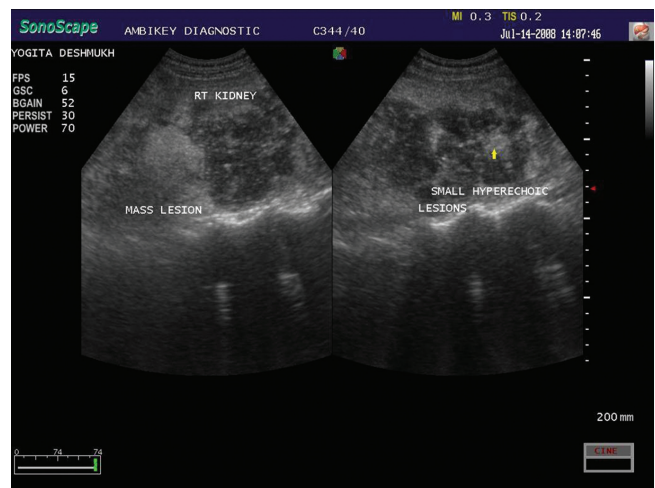


Figure 4. Haemorrhage in Right sided Renal angiomyo lipoma.

There are only four cases of tuberous sclerosis in pregnancy in the literature. Two of these had favourable maternal and fetal outcomes and the remaining two presented with serious maternal and fetal complications. These included acute intra-abdominal bleeding due to a ruptured renal tumour, which led to renal failure requiring haemodialysis, and severe preeclampsia with pathologically enlarged kidneys noted at the time of caesarean section.

We present a case of tuberous sclerosis in pregnancy with renal involvement with bleeding into a renal cyst, renal failure, preeclampsia, and severe intrauterine growth retardation during first conception. Similar case on second time delivered healthy male child with no maternal complication. Renal involvement appears to be the single most important prognostic factor in pregnancies with tuberous sclerosis. Renal evaluation should be performed in any patient who presents for pre-conceptional counselling [4].

We report this case due to its rare association between pregnancy and tuberous sclerosis.

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