ORIGINAL RESEARCH PAPER

Volume-7 | Issue-2 | February-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A CLASSICAL CASE OF BOURNVILLES DISEASE IN YOUNG FEMALE

Radiodiagnosis

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ABSTRACT

Tuberous sclerosis also known as Bournville disease is a phakomatosis affecting multiple organ systems. Our manuscript focusess on clinical, radiological findings present in 29 year female presented with altered consciousness as the sole symptom

KEYWORDS

Tuberous sclerosis, Angiomyolipoma, Adenoma sebaceum, Koenen tumor, Giant cell astrocytoma, Rhabdomyoma.

Case report

A 29 year old female presented to the emergency department with complaint of altered consciousness. Her vitals were stable and physical examination revealed rashes on both sides of cheeks. Detailed clinical history taking revealed breathlessness occasionally.

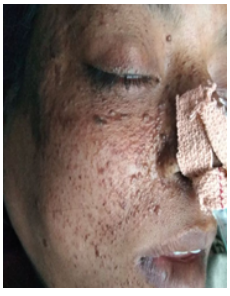


Figure 1.1 depicting the characteristic adenoma sebaceum

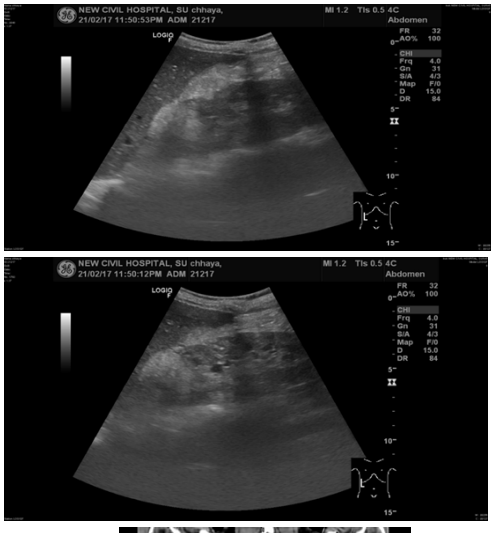
Figure 1.2 depicting the characteristic adenoma sebaceum

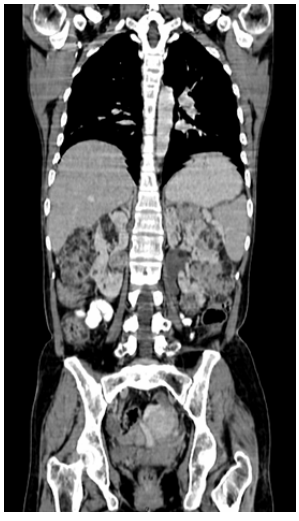
Detailed clinical examination revealed characteristic butterfly pattern of distribution of reddish spots on nose and cheeks (adenoma sebaceum) & fleshy swellings under finger nails (Periungual fibromas).

Figure 1.3 Depicts the characteristic Periungual fibroma

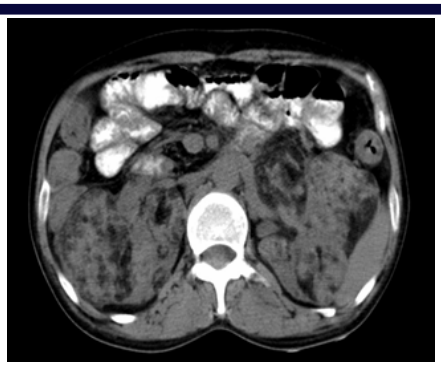
Biochemical parameters of the patient proved uneventful. On radiological evaluation,Usg and CT revealed Renal angiomyolipomas

Figure 1.4 & 1.5 Depicting the presence of echogenic lesions at the upper pole of right kidney - Suggestive of angiomyolipomas

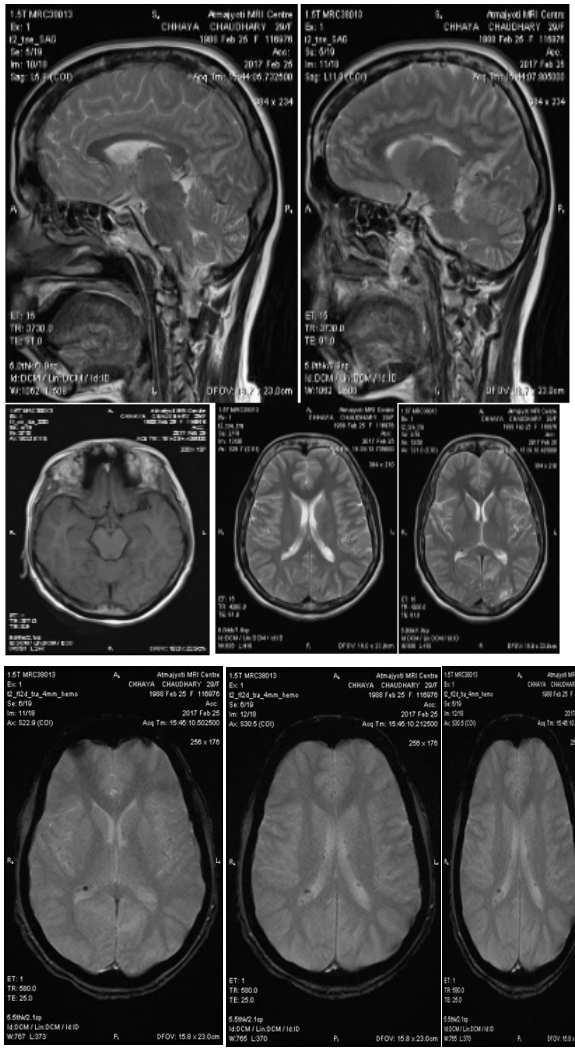




CT findings Multiple ill defined mixed density leisons with few areas of fat density with minimal post contrast enhancement noted involving both kidneys with few of them appears exophytic with largest measuring 75x68 mm2 in size noted involving mid and lower pole of right kidney and measuring 61x54 mm2 in size noted involving mid and lower pole of left kidney



MRI Brain was performed which revealed the Presence of Interventricular nodules



Discussion

Tuberous sclerosis also known as Bournvilles disease is a rare multisystem disorder affecting brain, heart, kidneys, lungs, eyes and skin.The basic defect in tuberous sclerosis is formation of hamartomas.

The Central nervous system lesions are one of the most consistent and are responsible for symptoms most often. Various CNS lesions in tuberous sclerosis include Giant cell astrocytoma, Cortical tuber formation, Subependymal nodules

The kidneys are the second most consistently involved organs in Tuberous sclerosis. Renal lesions are usually angiomyolipomas. major complications of renal angiomyolipomas are catastrophic hemorrhage.

The skin features are the markers suggesting the diagnosis of Tuberous sclerosis. Various skin features of tuberous sclerosis include Facial angiofibromas (called Adenoma sebaceum), periungual fibromas (Koenen tumors), Shagreen patches on back.

Other features of Tuberous sclerosis are lymphangioleiomyomatosis (LAM), Rhabdomyomas of the heart, phakomas of eye may be seen.

Diagnostic criteria for Tuberous sclerosis

Table. 1: Major and Minor Criteria of tuberous sclerosis complex

Major Criteria	Minor Criteria
1. Cortical tuber	1. Cerebral white matter migration lines
2. Subependymal nodule	2. Multiple dental pits
3. Facial angiofibroma or forehead plaque	3. Gingival fibromas
4. Ungual or periungual fibroma (nontraumatic)	4. Bone cysts
5. Hypomelanotic macules (>3)	5. Retinal achromatic patch
6. Shagreen patch	6. Confetti skin lesions
7. Multiple rentinal hamartomas	7. Nonrenal hamartomas
8. Cardiac rhabdomyoma	8. Multiple renal cysts
9. Renal angiomyolipoma	9. Hamaromatous rectal polyps
10. Pulmonary lymphangioleimyomatosis	

Results

Tuberous sclerosis is a disease involving multiple organs which is asymptomatic in most of the cases. The role of radiology is immense in diagnosing these conditions as it may procure a early diagnosis and proper diagnosis and necessary treatment can be done before complications. Radiologic assessment is useful not only in diagnosis but also in determining treatment. The presence of common manifestations, including cortical or subependymal tubers, white matter abnormalities, cardiac rhabdomyoma, and renal AML, allows us to confirm the diagnosis in cases with characteristic symptoms or skin lesions and to suspect Tuberous sclerosis in new cases without any clinical signs. The lungs, digestive system, retroperitoneum, and bone, which can be less frequently involved, should also be evaluated in patients with TS. Although recent advances in treatment have improved morbidity, the prognosis is still quite poor and nearly 40% of patients die by the age of 35 years. The cause of death varies depending on the patient's age.

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