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Radiodiagnosis

Tuberous Sclerosis: Case Report of a Classical Case with Multisystem Rare Findings and Unusual Presentation

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Abstract | **Case Report**

A 45 year old female with chief complaint of abdominal distension was referred to department of radiodiagnosis for further evaluation. CT & MRI of abdomen revealed grossly enlarged kidneys with multiple fat containing lesions nearly replacing normal renal parenchyma. On examination, she also had multiple facial papules which raised suspicion for phakomatosis and she was further evaluated. Multiple system findings and stigmatas of tuberous sclerosis were found in the patient and are described with descriptive images in this case report. There was no significant family history which suggests sporadic nature of tuberous sclerosis.
Keywords: radiodiagnosis, CT, MRI, abdominal distension.

OBSERVATIONS

Central nervous system

Patient did not have history of seizures, had average level of intelligence and was cooperative in the examination.

Subependymal nodules

CT head showed multiple (>10) calcified sub-ependymal nodules (Fig 1).

Subependymal giant cell astrocytoma

A soft tissue density lesion measuring approximately 1.8x1.5 cm was seen in

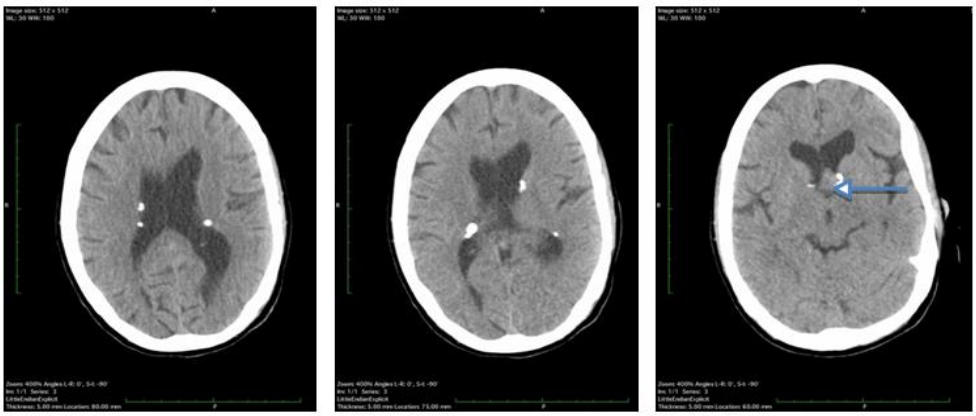

Fig-1: Left foramen of Monroe which showed internal calcification – likely sub-ependymal giant cell astrocytoma

Fig 1: NCCT head of patient revealed calcified subependymal nodules and a soft tissue density lesion in left foramen of Monroe (arrow) with mildly dilated ventricles.

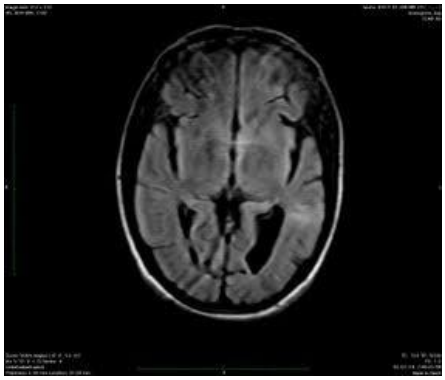

Fig-2: MRI brain showing FLAIR hyperintensities

Cortical Tubers

MRI brain showed multiple FLAIR hyper intensities (fig 2) - suggestive of cortical tubers

Chest

Lymphangioleiomyomatosis

HRCT chest of patient showed multiple small thin walled cysts of variable shapes diffusely scattered throughout the lung parenchyma-suggestive of *lymphangioleiomyomatosis* (Fig 3). There was no associated pneumothorax or pleural effusion.

Multiple micronodular pneumocyte hyperplasias

Multiple tiny randomly scattered nodules were also seen in the lungs (arrows) – likely *multiple micronodular pneumocyte hyperplasias (MMPH)* which is a rare finding in tuberous sclerosis [1] (Fig 3). Heart was normal & showed no lesions to suggest rhabdomyosarcomas.

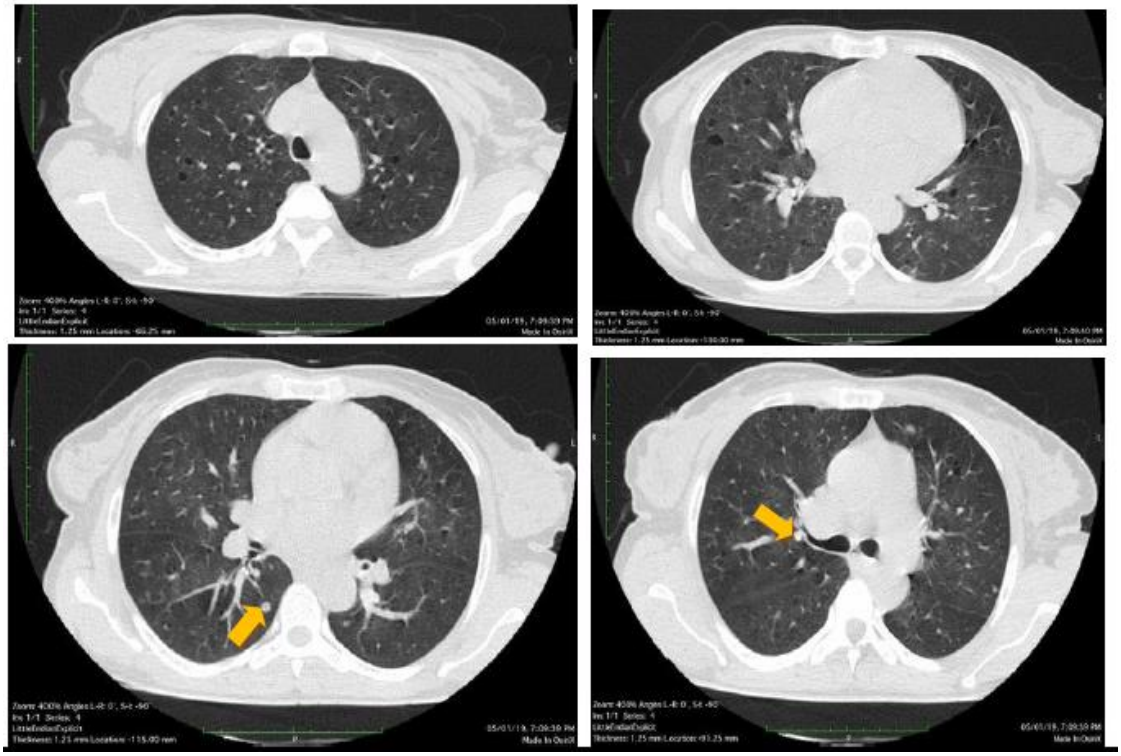

Fig-3: Multiple variable sized thin walled cystic lesions. Few randomly scattered tiny nodules are also seen (arrows). There is no pleural effusion or pneumothorax

Abdomen

Multiple renal & hepatic angiomyolipomas

Both kidneys were enlarged and showed multiple variable sized fat containing lesions virtually replacing the entire renal parenchyma.

Ultrasound of the patient revealed grossly enlarged hyper-echoic kidneys showing loss of cortico-medullary differentiation (Fig 4a). Few hyper echoic lesions were also visible in liver (Fig 4b).

CT & MRI abdomen revealed grossly enlarged both kidneys showing loss of cortico-medullary

differentiation with multiple variable sized fat containing lesions nearly replacing normal renal parenchyma. Reniform shape of right kidney was maintained, however left kidney showed distorted contour with a large exophytic lesion arising from lower pole, displacing bowel loops peripherally and reaching upto anterior abdominal wall (Fig 5 & 6).

T1-T2 Hyperintense lesions were also seen in liver which showed loss of signal on fat saturated images.

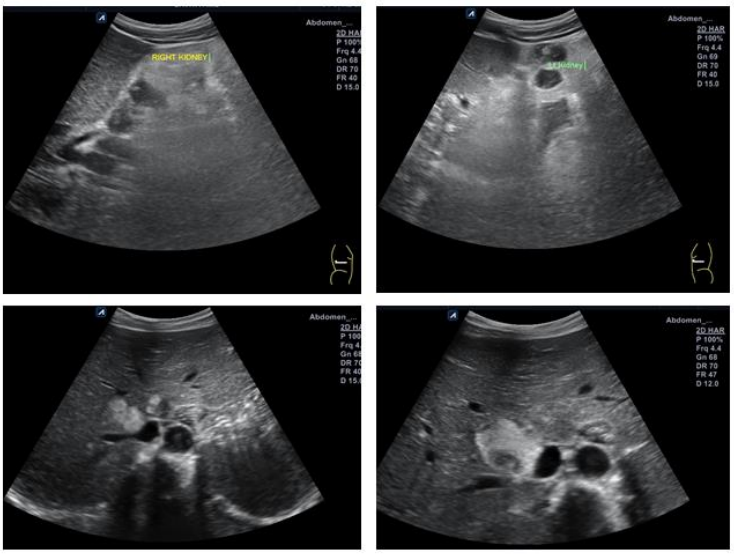

Fig-4(a): Both kidneys were enlarged, hyperechoic and showed complete loss of cortico-medullary differentiation
Fig-4(b): Hyperechoic lesions of variable sizes were also seen in liver

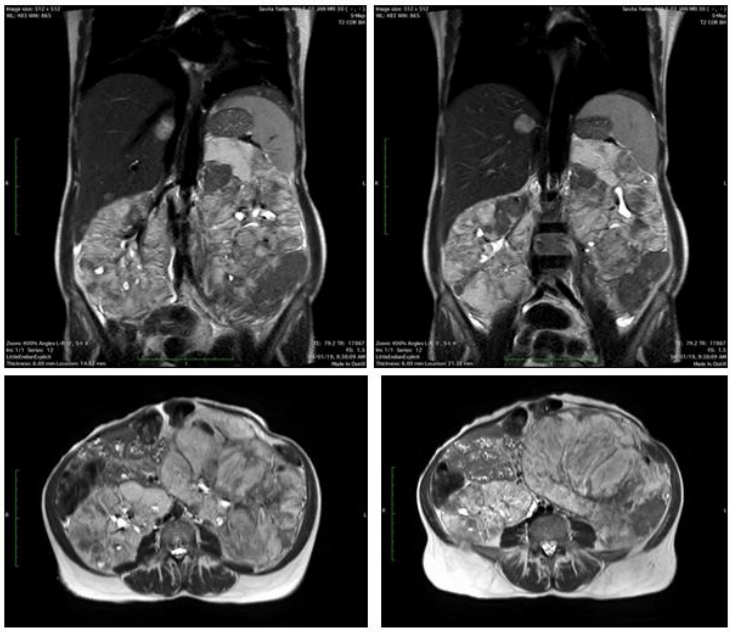

Fig-5: MRI abdomen showing grossly enlarged kidneys with multiple variable sized lesions nearly replacing renal parenchyma. Few lesions are also seen in liver

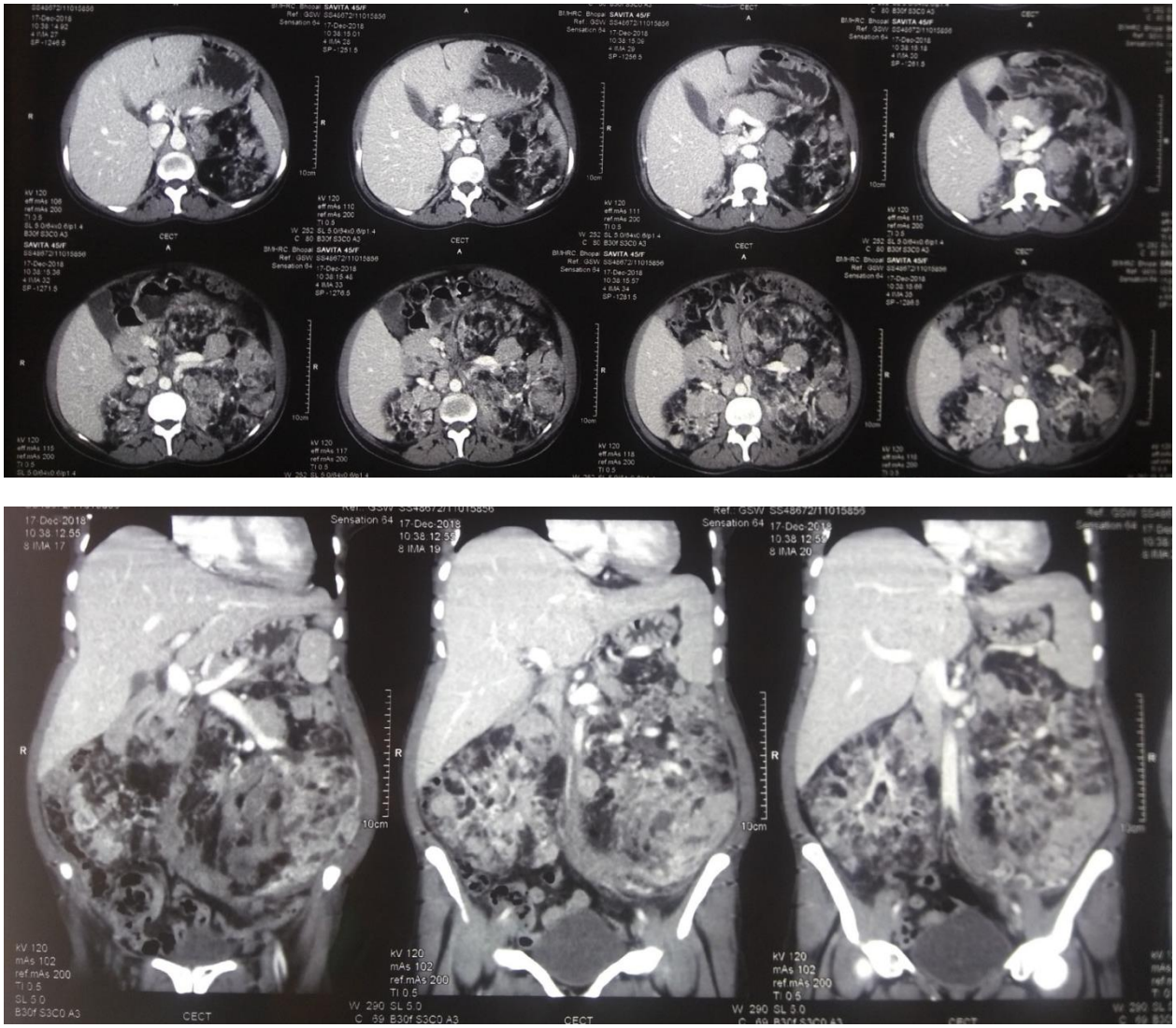

Fig-6: Axial & coronal reformatted images of CECT abdomen showing grossly enlarged kidneys with multiple variable sized fat containing lesions

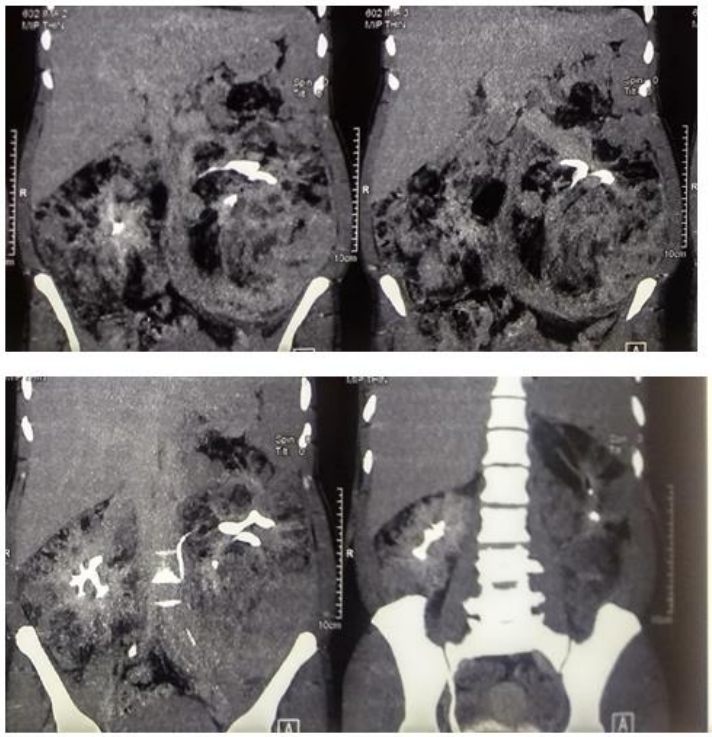

Fig-7: CT urography images depicting functional kidneys with splaying of pelvi-calyceal system

CT urography demonstrated that both kidneys were functional. The pelvi-calyceal system was distorted and splayed (Fig7).

Skeletal system

Patient had multiple foci of sclerosis in thoracic vertebrae, involving vertebral bodies and posterior elements which is one of skeletal manifestations of tuberous sclerosis [2] (Fig 8).

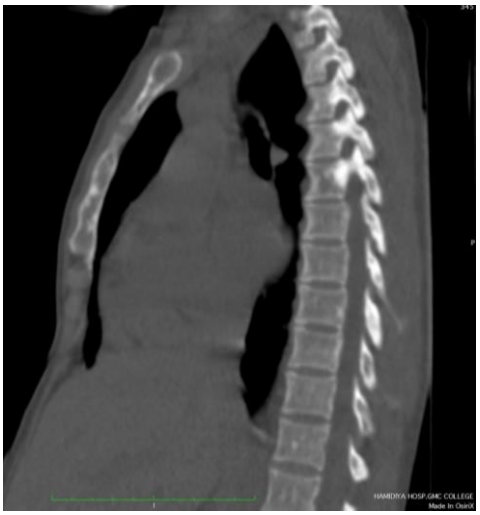

Fig-8: Few sclerotic foci are noted involving vertebral bodies and posterior elements of thoracic vertebra

Dermatological findings

Facial angiofibromas /adenoma sebaceum

These are papules distributed in butterfly pattern found in almost 75 % patients of tuberous sclerosis (fig 9). Besides these, patient also had a large papule with secondary changes.

Forehead fibrous plaque

These are found in about 25% patients with tuberous sclerosis and were paired with facial angiofibromas in diagnostic criteria in1988. There are studies to suggest association between forehead plaques and CNS involvement in tuberous sclerosis [3]. These are being considered cutaneous markers of CNS involvement. This patient also had both supporting the argument (Fig 9)

Molluscum Pendulum

Patient had brown sessile and pedunculated papules on neck since birth. (Fig 9) Such brown papules circumferentially involving the neck give the "necklace sign"which is a classical finding in tuberous sclerosis [4].

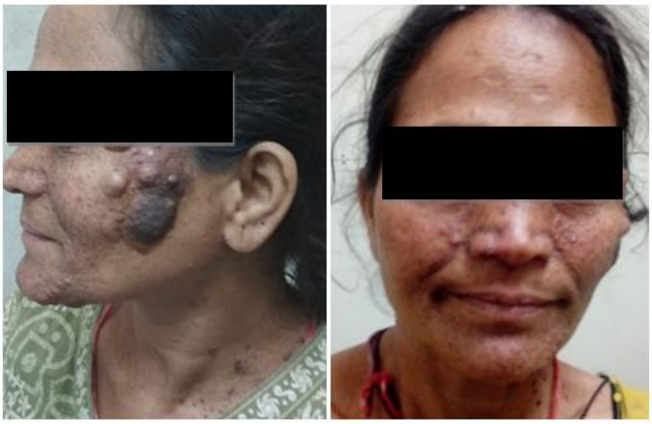

Fig-9: Bilateral symmetrical papules in butterfly distribution – suggestive of adenoma sebaceum. Also note plaques on patient's forehead and brown papules on neck

Ash leaf macules

These are hypopigmented macules found in 90% of patients with tuberous sclerosis (fig 10). They are usually more than 3 in number. These come under major diagnostic critera for diagnosing tuberous sclerosis.

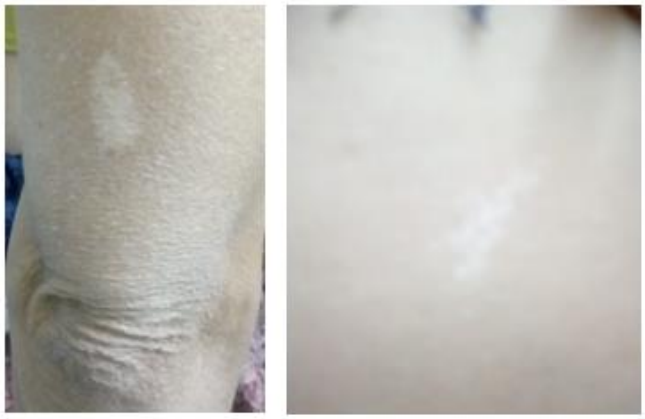

Fig-10: Hypopigmented macules (Ash leaf spots) on patient's trunk and arm.

Shagreen's patch

These are connective tissue hamartomas found in nearly half of tubersous slcerosis patients and fall under major diagnostic criteria (Fig 11). Usually these are noted in lumbo-sacral region.

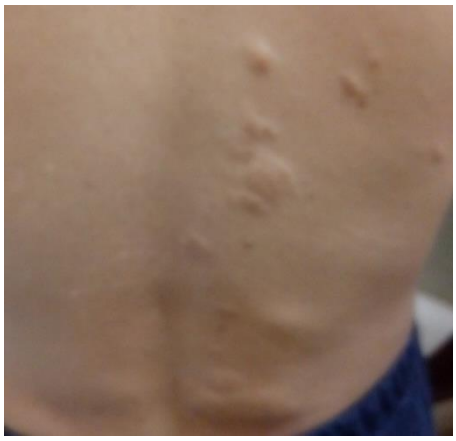

Fig-11: Shagreen's patches in lumbo-sacral region

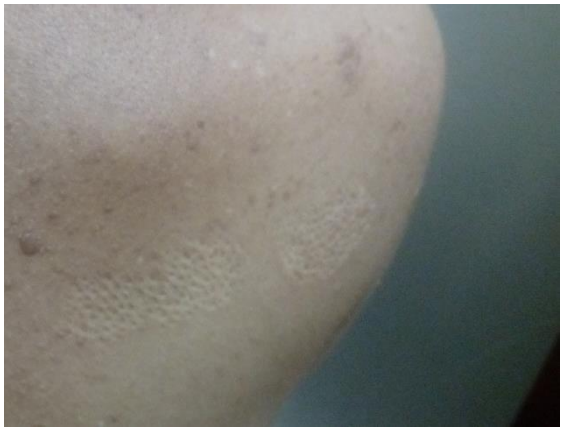

Fig-12: Hypopigmented 'pig-skin'patch in region of scapula in same patient

Pigskin lesion

These are special type of hypopigmented shagreen's patches which are very rare (Fig 12).

Koenen's tumor (Periungual fibroma)

These are skin-colored or reddish peri-ungual or sub-ungual papules noted in in patients with tuberous sclerosis which can be found in all the digits, but are mostly seen in thumb & great toe (Fig 13).

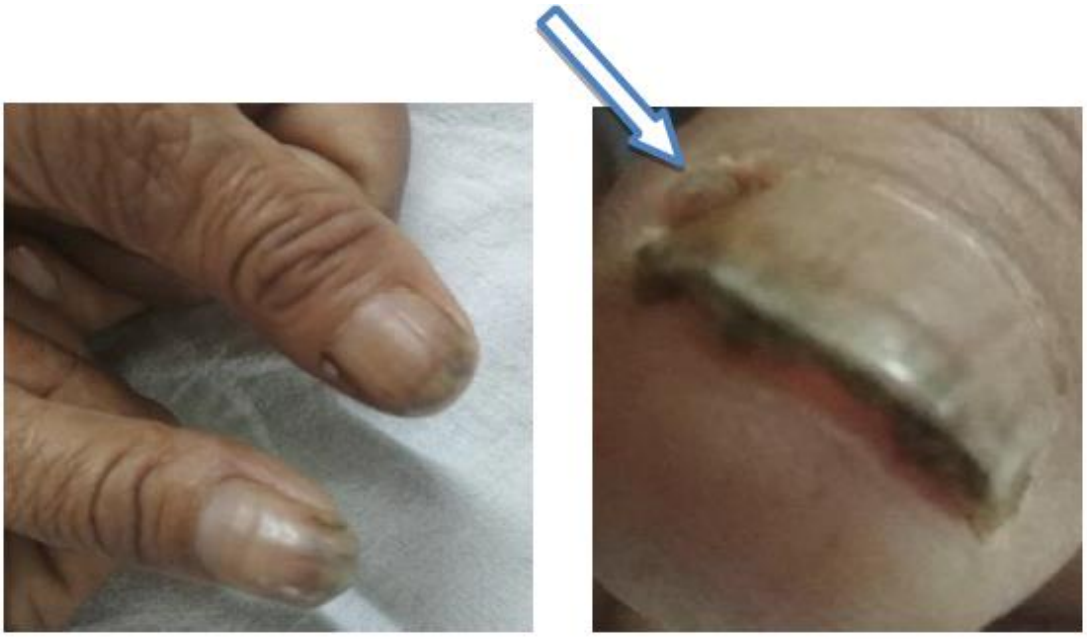

Fig-13: Peri-ungual fibroma (arrow) in patient's left thumb

"Confetti-like" macules

These are tiny (1-3mm) white macules usually found on the trunk or extremities (Fig 14). These are grouped under minor criteria for tuberous sclerosis owing to their varied frequency (ranging from 3% to 58 %) in studies.

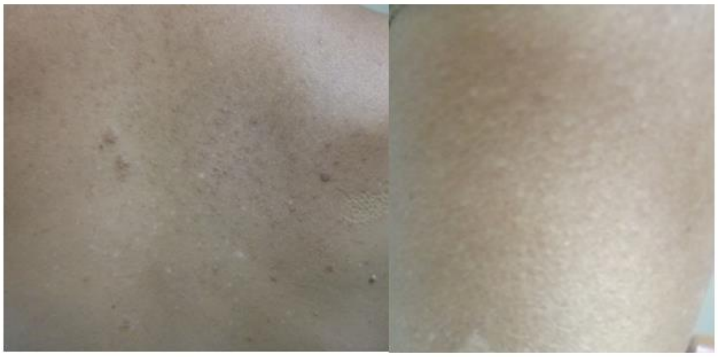

Fig-14: Randomly scattered tiny hypopigmented macules on patients elbow and trunk

Dental enamel pits

These are randomly distributed pits in dental enamel (Fig 15) which become more pronounced with food dyes. Enamel pitting is very common finding in patients with tuberous sclerosis, but is not very specific for it. They can also be found in general population and are considered minor diagnostic criteria.

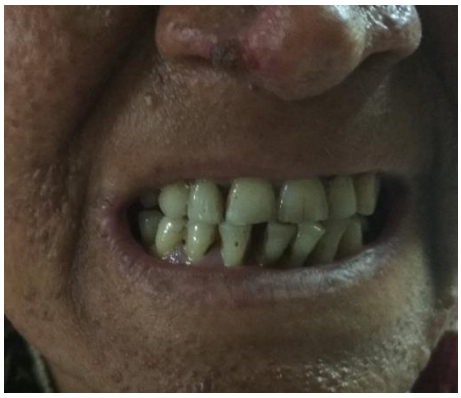

Fig-15: Few randomly distributed pits can be seen in patient's upper and lower incisors

CONCLUSIONS

- Tuberous sclerosis or Bourneville syndrome is a phakomatosis caused by mutation in tumor suppression genes "tuberin" and "hamartin" which leads to formation of hamartomas in multiple organ systems.
- Although it is an autosomal dominant disorder, about two third cases are sporadic in nature
- Vogt's triad of adenoma sebaceum, seizures and mental retardation is frequently *absent*.
- For diagnosing tuberous sclerosis 11 major & 6 minor criteria are tabulated [5], of which this patient had 9 major criteria (Ash leaf macules, adenoma sebaceum, peri-ungual fibromas, shagreen patch, cortical dysplasia, subcependymal nodules, sub-ependymal giant cell astrocytoma, LAM, renal AML) and 2 minor criteria (confetti skin lesions and dental enamel pits).
- Although the patient had multiple stigmatas of tuberous sclerosis, the chief complaint was of abdominal distension which was attributed to bulky kidneys with multiple large renal AMLs. Renal angiomyolipomas in patient with tuberous sclerosis are often bilateral, larger and multiple; as depicted in this case.
- Patient had lymphangioleiomyomatosis, but there was no pneumothorax or pleural effusion.
- For hepatic lesions in any other patient, the first differential would have been hemangioma, but in tuberous sclerosis patient, fat containing lesion goes in favor or angiomyolipoma.
- Although the patient had all the central nervous system findings – subependymal nodules, cortical tubers and even subependymal giant cell astrocytoma, there was no history of seizures and the patient's intelligence was normal.

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