Pictorial CME

A Cause of Recurrent Seizure — A Neuro Cutaneous Syndrome

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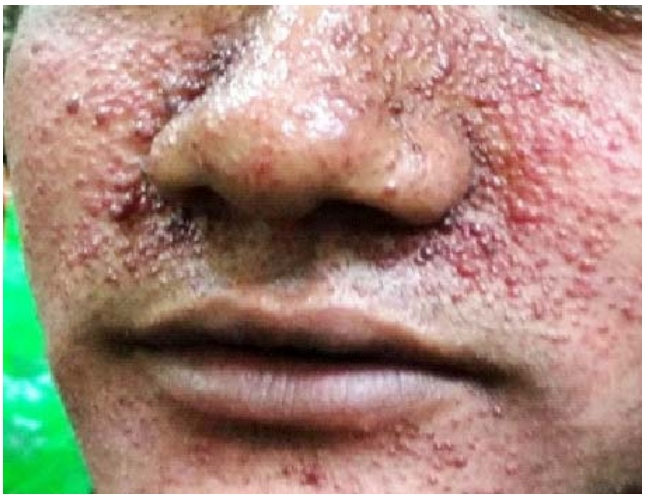
A 14 year old male from a village of Purulia presented to Medicine OPD with history of recurrent seizure for last 1 year without any h/o fever or altered sensorium. His seizure was recurrent and according to the description of the eye witness it was generalized. He gained consciousness following the events and continued his daily work. His birth history was normal and no developmental delay was there. He had poor academic performance and could not continue his school after class 4. He also developed some behavioral abnormality like occasional agitation and compulsiveness. He had a low socio economic background and he took alternative medicines for his ailment. But the seizure activity persisted and he attended a tertiary hospital in Kolkata.



On examination he hadpapular skin lesions on his face which was present since his childhood, gradually increasing in number but they did not bother him as they were painless and non pruritic.

Based on typical skin lesion of facial angiofibromas (adenoma sebaceum) and h/o recurrent seizures , it was clinically suspected to be a case of TUBEROUS SCLEROSIS.

Q1. What are the other sites to examine?

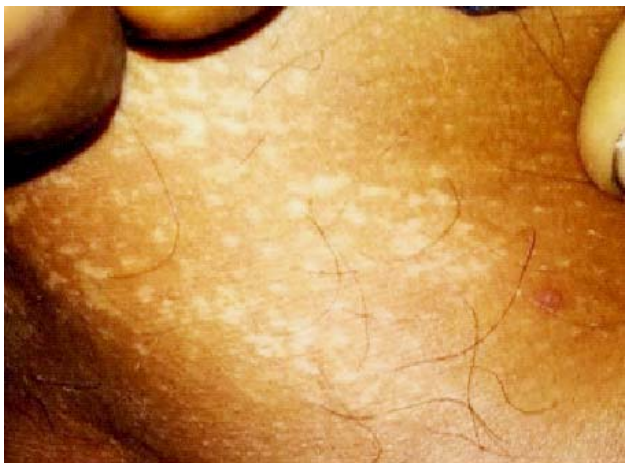
Q 2. What are the MRI features of Tuberous sclerosis?

MRI BRAIN

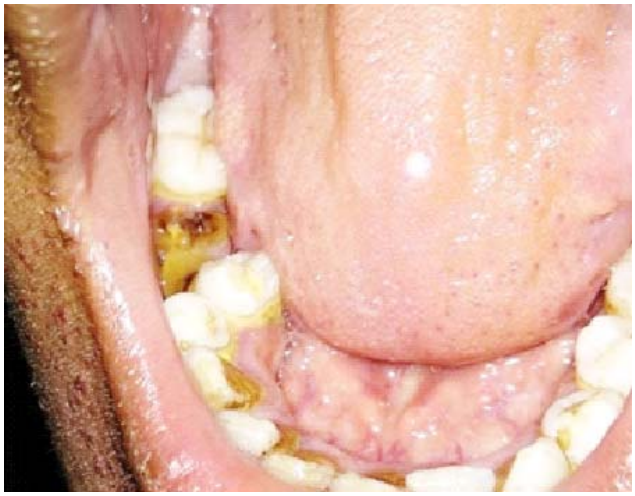
MRI brain reveals classical cortical tubers in both T1 and T2 weighted images.

Q3. What is the genetic basis of the syndrome?

It is an autosomal dominant disorder with an incidence of ~1 in5000–10,000 live births. It is caused by mutations in either the *TSC1*gene, which maps to chromosome 9q34 and encodes a protein termedhamartin, or the *TSC2* gene, which maps to chromosome 16p13.3and encodes the protein tuberin. Hamartin forms a complex withtuberin,



Generalised skin condition: Ash-leaf (Hypomelanic) macules in the back



Dental lesions: Dental enamel pits

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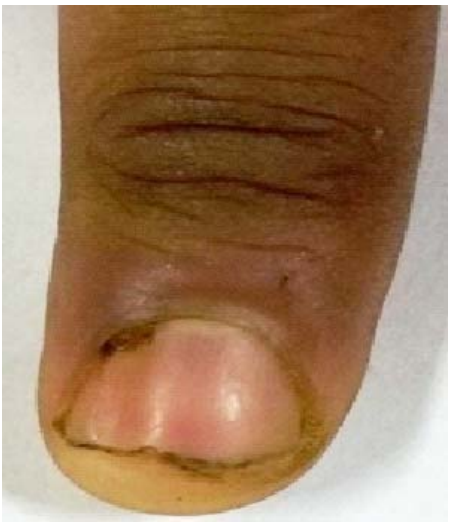
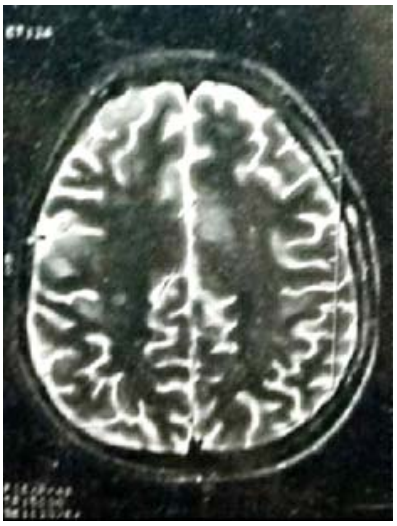
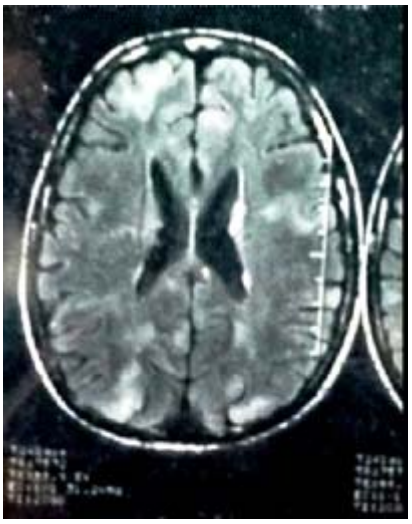


Nail condition : Periungual fibromas/ Koenen'stumors



T1W : Subependymal nodules: form in the walls of ventricles



T2W : cortical and subcortical tubers

which inhibits cellular signaling through mTOR, and acts asa negative regulator of the cell cycle.

Q4. What are the cutaneous lesions associated with this condition?

i. A**denoma sebaceum**becomes manifest usu-ally between 5 and 10 years of age and typically consists of reddened nodules on the face (cheeks, nasolabial folds, sides of the nose, and chin) and sometimes on the forehead and neck.

ii. Subun-gual fibromas,

iii. Shagreen patches (leathery plaques of sub-epidermal fibrosis, situated usually on the trunk)

iv. Leaf-shaped hypo-pigmented spots.

Q5. What are other associated conditions with Tuberous Sclerosis?

Patients with tuberous sclerosismay have seizures, mental retardation, periungualfibromas, renal angiomyolipomas, and benign cardiac rhabdomyomas. Thesepatients have an increased incidence of subependymal nodules, corticaltubers, and subependymal giant-cell astrocytomas (SEGAs).

6. What are the possible management modalities?

Patientsfrequently require anticonvulsants for seizures. SEGAs do not alwaysrequire therapeutic intervention, but the most effective therapy is withthe mTOR inhibitors sirolimus or Everolimus, which often decreaseseizures as well as SEGA size.

References

1 Cardis MA, DeKlotz C — Cutaneous manifestations of tuberous sclerosis complex and the paediatrician's role. Archives of disease in childhood 2017; **102(9):** 858-63. https://doi.org/10.1136/archdischild-2016-312001

2 Portocarrero LKL, Quental KN, Samorano LP, Oliveira ZNP, Rivitti-Machado MCDM — Tuberous sclerosis complex: review based on new diagnostic criteria. *An Bras Dermatol* 2018; **93(3):** 323-331. doi:10.1590/abd1806-4841.20186972 (Diagnostic criteria journal)