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## Tuberous sclerosis: Seizure in infant – A case study

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### Abstract

Tuberous sclerosis is a genetic abnormality having multisystem manifestations. In this case report, we discussed the MRI imaging findings of an infant presenting with seizures and found to have neurological manifestation of tuberous sclerosis with subcortical tubers and subependymal nodules. MRI of brain provides a conclusive diagnosis and evaluation of seizures in infant and can exclude ischemia, hypoxia, developmental morphological abnormalities, dysplasias, infective and manifestations of tuberous sclerosis with high degree of sensitivity and specificity.

**Keywords:** Tuberous sclerosis, infantile seizures, magnetic resonance imaging

### Introduction

A 7 month old male child presented with history of seizures since the age of 3 months. Developmental milestones were normal. No evidence of maternal diabetes, birth asphyxia or injury. Infant could sit with support. Developmental milestones normal. No cutaneous pigmentation or nodules. No cosmetic deformity. No random eye movements. Plantar response normal.

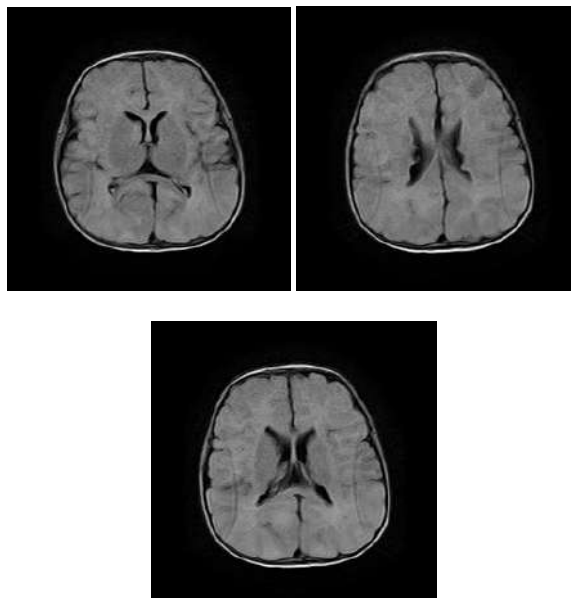
Infants in seizure are caused by multiple etiologies including hypoxic-ischemic encephalopathy, encephalitis, meningitis, hypoglycemia, traumatic brain injury, or developmental morphological disorders and dysplasia.

### Materials and Method

#### Investigations and Interventions

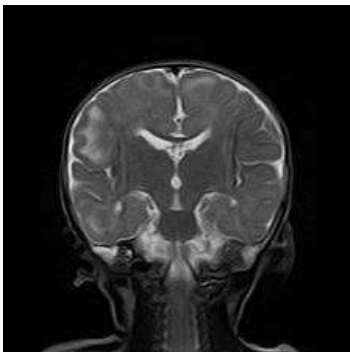
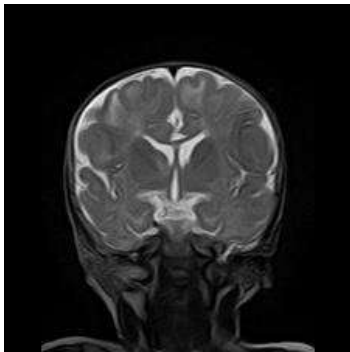
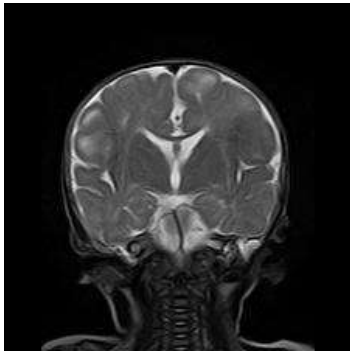
MRI of brain was performed including axial DWI, ADC, FLAIR, T2, T1, GRE, Coronal T2 and sagittal T2 and T1 sequences.

#### a. FLAIR – AXIAL

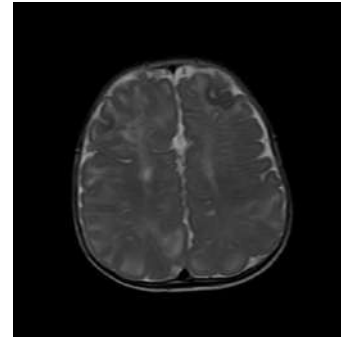
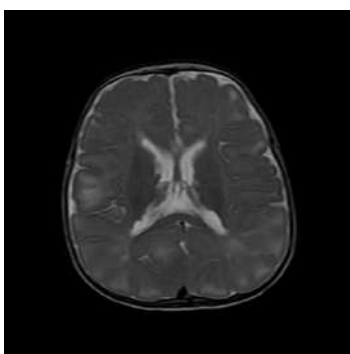
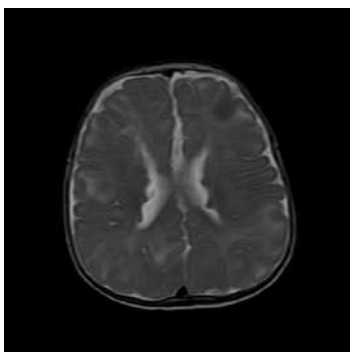


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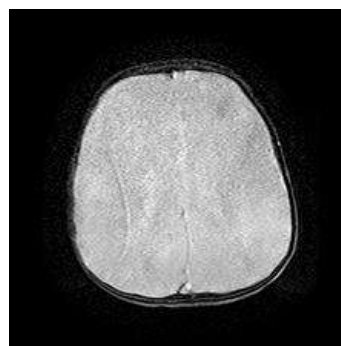
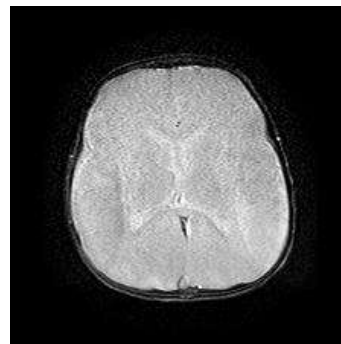
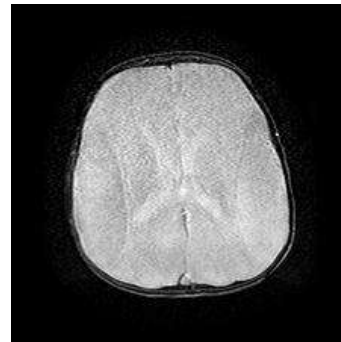
**b. T2W-CORONAL**



**c. T2W AXIAL**



**d. GRE AXIAL**



**Figures a, b, c and d:** MRI of brain reveals triangular shaped T2 hyperintense and T1 hypointense lesions at bilateral subcortical white matter consistent with cortical/subcortical tubers. Tiny subependymal nodules of 4-6 mm sizes are noted along bilateral lateral ventricles which appear isointense on the T2 images consistent with subependymal hamartomas. No evidence of blooming on GRE sequence suggests absence of calcification. Findings are consistent with tuberous sclerosis.

Once radiologic and clinical diagnosis of tuberous sclerosis complex is made, further genetic studies are advised.

**Treatment**

Treatment of seizures is the main consideration, varying supportive care is recommended, depending on the degree

of intellectual disability. Treatment will be dictated by individual manifestations (e.g., organ involvement, types of hamartoma, subependymal giant cell astrocytoma, or renal angiomyolipoma and their complications present at the time of diagnosis).

Approximately 40% of patients do not survive beyond age 35 -40 due to complications.

### Discussion

Tuberous sclerosis, a neurocutaneous disorder is characterized by the development of multiple benign tumors of the embryonic ectoderm (e.g., skin, eyes, and nervous system). Tuberous sclerosis has an incidence of 1:6000-12,000, with most being sporadic. Désiré-Magloire Bourneville was a French neurologist who gave the initial description of tuberous sclerosis, hence the name Bourneville disease. It is sometimes also referred as "Bourneville-Pringle disease". Heinrich Vogt was a German neurologist that is notable by establishing the three pathognomonic clinical signs for tuberous sclerosis that became known as "Vogt triad" consisting of seizures, mental retardation, and adenoma sebaceum.

Spontaneous mutations account for majority of case, with the remainder as an autosomal dominant condition. The mutation involve two tumor suppressor genes, both part of the mTOR pathway 3, 13:

**TSC1:** Encoding hamartin, on chromosome 9q32-34

**TSC2:** Encoding tuberin, on chromosome 16p13.3

The tuberous sclerosis diagnostic criteria have been developed to make a definitive diagnosis of tuberous sclerosis.

### Genetic criteria

The identification of genetic mutation leads to a definite diagnosis of tuberous sclerosis complex.

### Clinical criteria

Definitive TS complex: either 2 major features or 1 major and 2 or more minor

**Possible TS complex:** either 1 major or  $\geq 2$  minor

### Major features include

1. Angiofibromas (3 or more) or fibrous cephalic plaque,
2. Non-Traumatic unguar or periunguar fibroma (2 or more),
3. Hypomelanotic macules (3 or more, at least 5mm diameter),
4. Shagreen patch,
5. Multiple retinal nodular hamartomas,
6. Cortical dysplasias (include tubers and cerebral white matter migration lines),
7. Subependymal nodule,
8. Subependymal giant cell astrocytoma,
9. Cardiac rhabdomyoma
10. Lymphangiomyomatosis (LAM) and
11. Angiomyolipomas (2 or more)

### Minor features include

1. Dental enamel pits (3 or more),
2. Intraoral fibromas (2 or more),
3. Non-renal hamartomas,

4. Retinal achromic patch,
5. 'confetti' skin lesions and
6. Multiple renal cysts.

MRI of brain can detect with fair certainty neurological components like cortical/subcortical tubers, subependymal hamartomas, often associated with calcification, although calcification absent in early childhood, subependymal giant cell astrocytomas, cerebellar atrophy, infarcts (due to occlusive vascular disorders), cerebral aneurysms, dysgenesis of the corpus callosum, Chiari malformations, arachnoid cysts and chordoma

### Conclusion

Seizures in infants are a common occurrence with a majority associated with sequela of hypoxic ischemic encephalopathy associated with perinatal asphyxia and prolonged labor. Tuberous sclerosis is a rare autosomal dominant neurocutaneous syndrome characterized by benign congenital tumors in multiple organs. The diagnosis is arrived on the basis of diagnostic criteria applied to clinical or radiologic findings. Radiologic examinations play an important role in the diagnosis of tuberous sclerosis and in excluding other abnormalities that can cause infantile seizures. Knowledge of clinical and radiologic findings in various organs is crucial in diagnosis and treatment. MRI of brain provides a conclusive diagnosis and evaluation of seizures in infant and can exclude ischemia, hypoxia, developmental morphological abnormalities, dysplasias, infective and manifestations of tuberous sclerosis with high degree of sensitivity and specificity. MRI of the brain can detect with fair certainty neurological components like cortical/subcortical tubers, subependymal hamartomas, often associated with calcification, although calcification absent in early childhood, subependymal giant cell astrocytomas, cerebellar atrophy, infarcts (due to occlusive vascular disorders), cerebral aneurysms, dysgenesis of the corpus callosum, Chiari malformations, arachnoid cysts and chordoma.

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