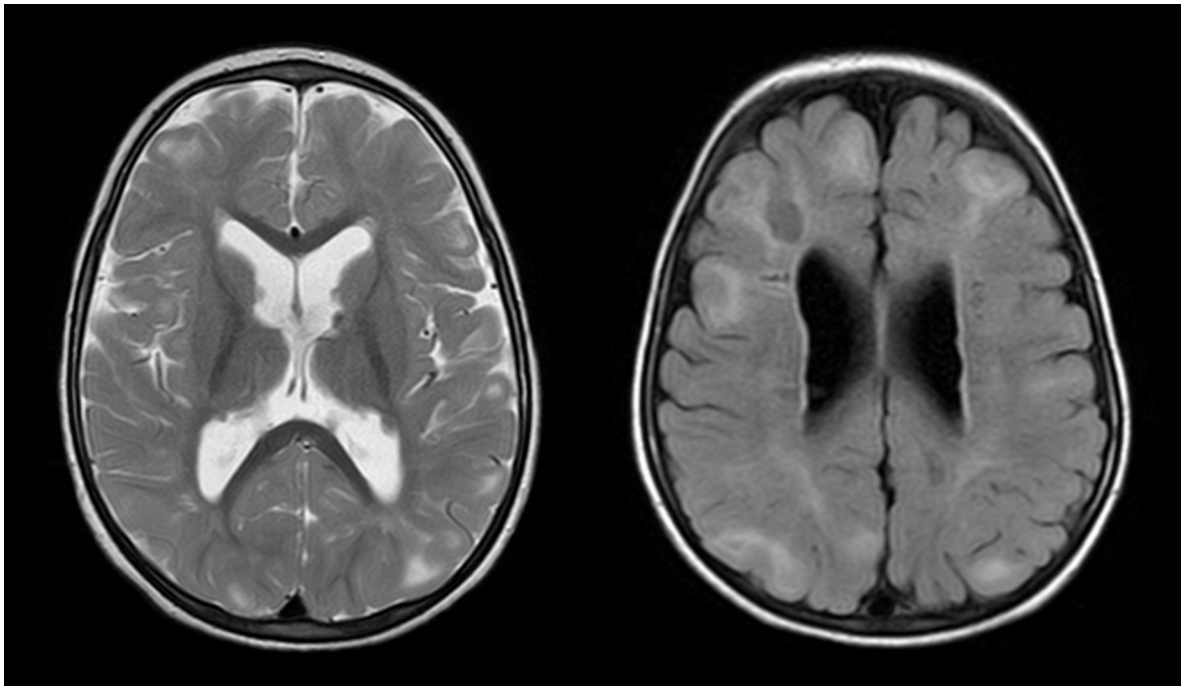
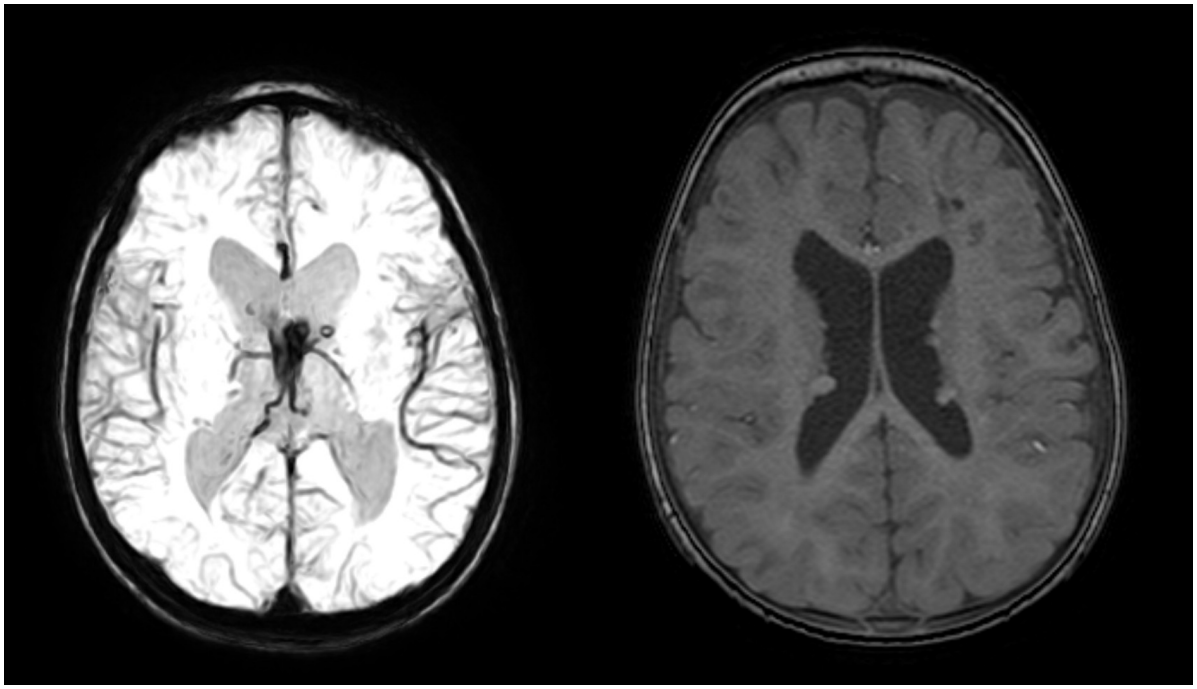
Clinical History

A 1-year-old child presented with a history of multiple episodes of seizures. The seizures started when the child was 6 months old and have been increasing in frequency. The child has also had developmental delays and has not been achieving milestones as per age.

Imaging Findings



AXIAL T2 AXIAL FLAIR



SWI POST CONTAST T1 FS

Multiple areas of altered signal intensities, showing expanded gyri are noted in bilateral cerebral hemispheres showing thickened and hyperintense cortex with bands extending through underlying white matter upto bilateral lateral ventricles-s/o Cortical tubers with radial bands.

Multiple subependymal nodular lesions which are hypointense on T2WI and hyperintense on T1- showing post contrast enhancement, few of which showing blooming on SWI are noted in body of bilateral lateral ventricles ~ subependymal nodules.

- Cortical tubers
- Radial bands
- Enhancing subependymal nodules

Probable Imaging Differentials

The imaging findings are most consistent with tuberous sclerosis complex (TSC).

Other possible differential diagnoses include:

- Neurofibromatosis type 1
- Subependymal Giant cell astrocytoma.
- subependymal grey matter heterotopia (Smooth and ovoid- long axis parallel to the ventricular surface)

Discussion on Other Probable Intra and Extra Cranial Manifestations of Tuberous Sclerosis

In addition to the brain lesions, TSC can also involve other organs, including the skin, kidneys, heart, and lungs. Some of the other possible intra and extracranial manifestations of TSC include:

- Renal lesions: Angiomyolipomas and renal cysts
- Cardiac lesions: Rhabdomyomas and cardiac arrhythmias
- Skin lesions: Hypomelanotic macules, angiofibromas, shagreen patches, and periungual fibromas
- Pulmonary lesions: Lymphangioleiomyomatosis

Our case was further evaluated with Echocardiogram which revealed Cardiac rhabdomyoma.

Conclusion

Tuberous sclerosis is characterized by the development of multiple benign tumors of the embryonic ectoderm affecting a spectrum of organs (e.g. skin, eyes, and nervous system, Lungs, Heart…).

The child is being followed regularly by a multi-speciality panel of doctors comprising of Neurologist (For Seizure management), cardiologist to evaluate for cardiac rhabdomyomas, dermatologist to evaluate for skin lesions.