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Intracerebral Tuberos Sclerosis

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Agenda

- Patient Presentation
- Overview of Tuberous Sclerosis
- MR imaging
 - Three CNS manifestations: (1) tubers (2) subependymal nodules (3) white matter changes
 - Pediatric vs. Adult presentation
- Patient Follow-Up



Our Patient

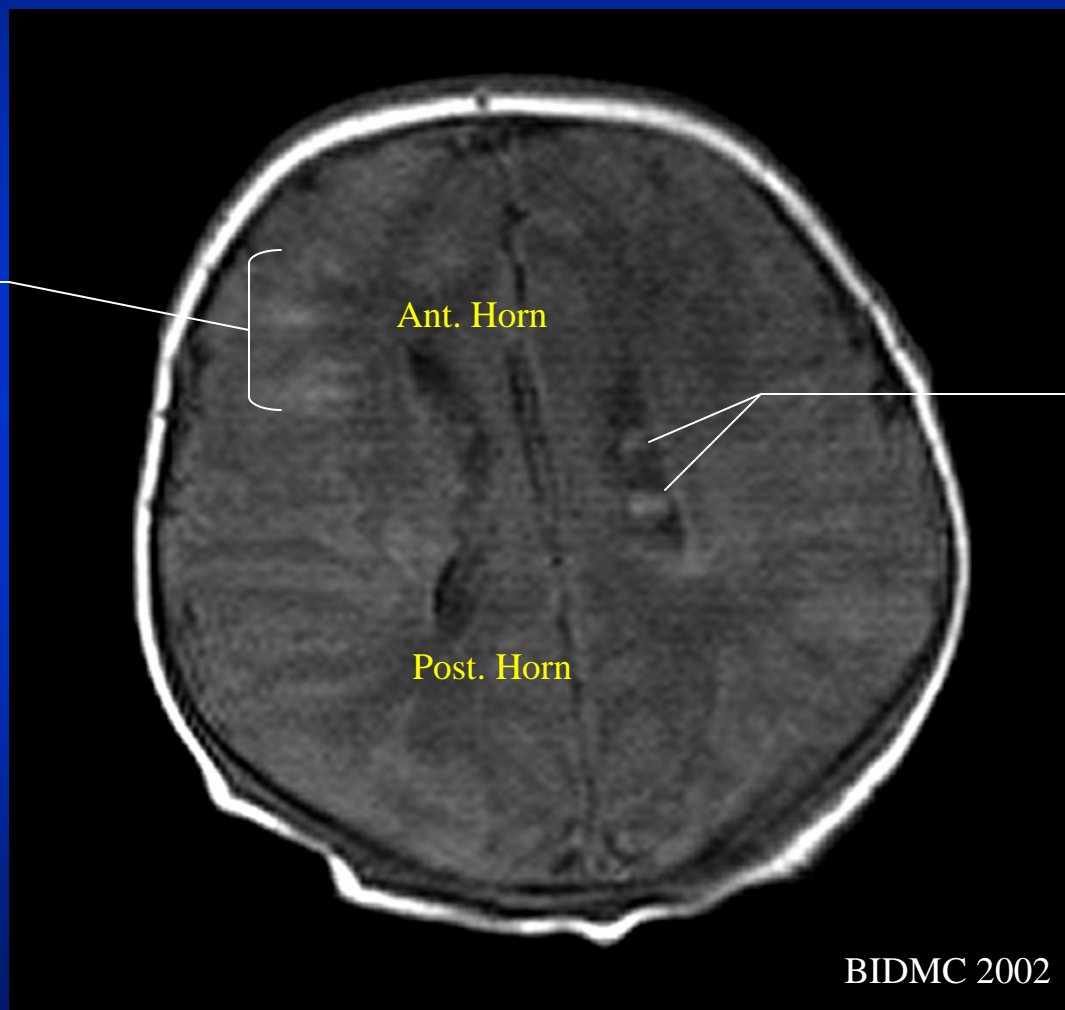
- **CC/ID:** 3.755 kg male baby born to 32 y.o. G2P1 mother by Caesarean section
- **ROS:**
 - Resp: weaned to room air off CPAP
 - CV: ECG w/ intermittent sinus arrhythmias
 - FEN: IV fluids
 - Neuro: no seizure activity
 - Heme/ID: CBC WNL, Blood cx (-)

Our Patient: Imaging

- **Echocardiogram**
 - Cardiac rhabdomyomas, no outflow obstruction, good ventricular function
- **Abdominal ultrasound**
 - L pelvic kidney, mild R hydronephrosis
- **MRI brain**
 - Axial, sagittal T1WI, axial gradient echo, T2WI.
Findings?

MRI Axial T1WI

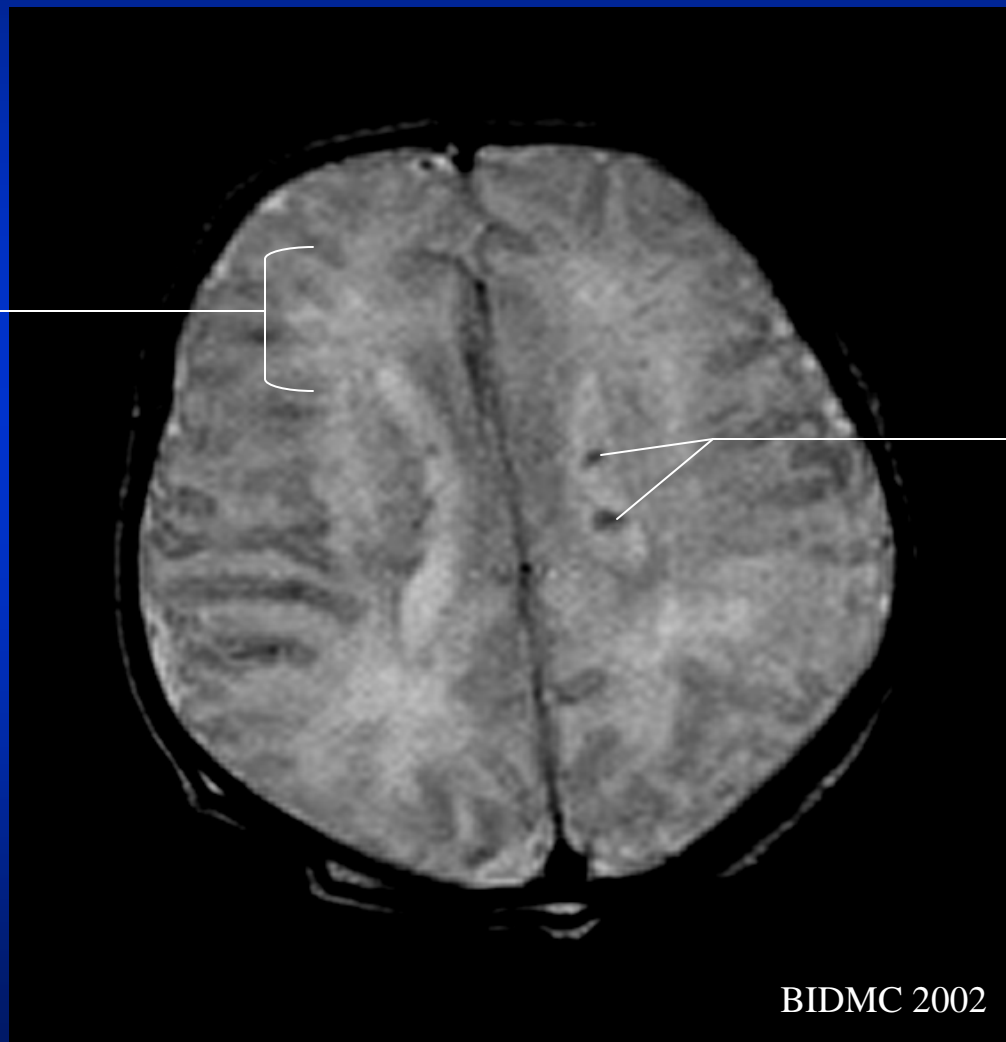
Increased signal in frontal lobe subcortical white matter



Foci of increased signal w/in left ventricle

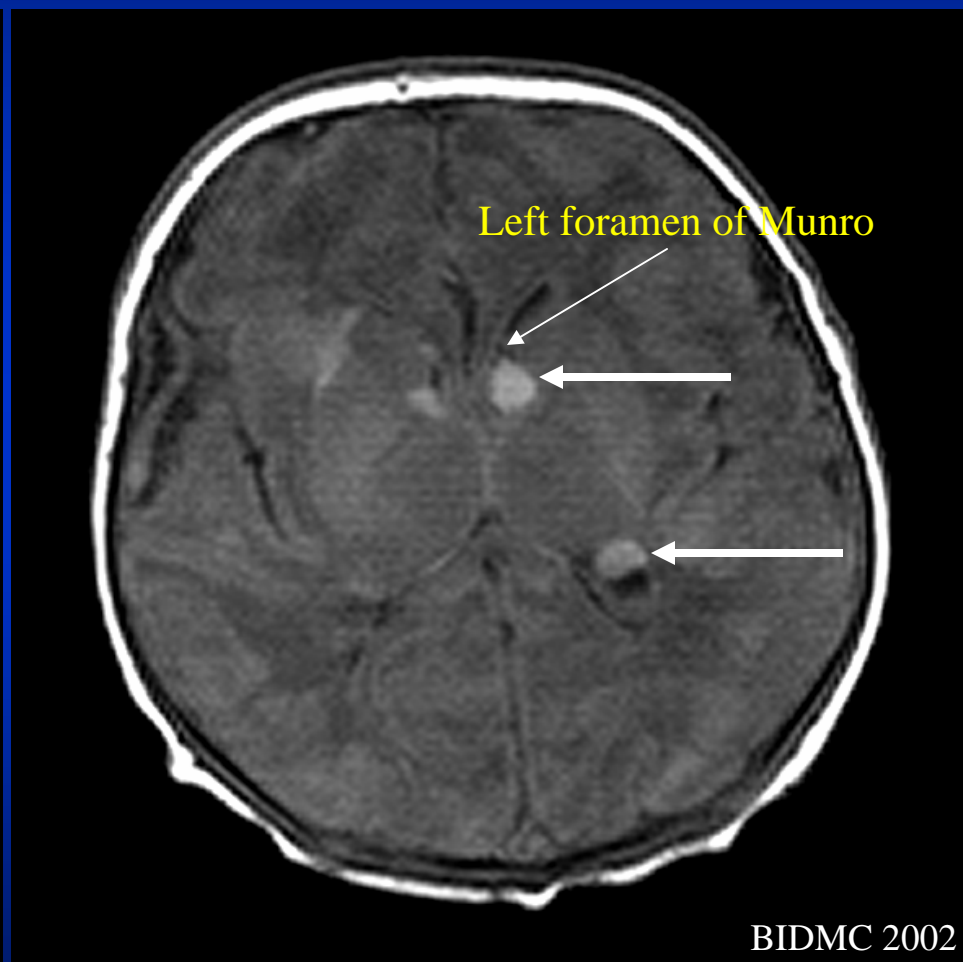
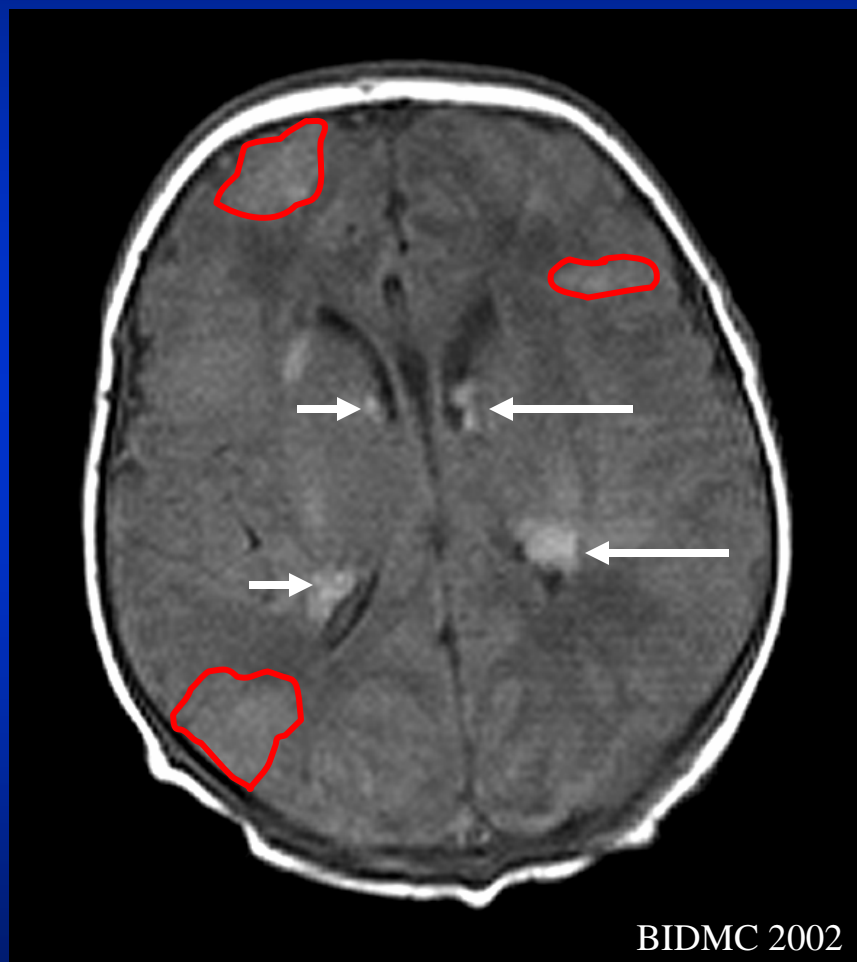
MRI Axial T2WI

Isointense to
hyperintense
signal



Hypointense
foci

MRI Axial T1WI



Periventricular, subependymal and subcortical lesions

Tuberous Sclerosis 1

- Heredofamilial neurocutaneous syndrome (phakomatosis) first described in 1880
- Multisystem hamartomatous involvement (brain, kidney, skin, retina, heart, lung)
- Vogt's classic \triangle , 1908: seizure, retardation, adenoma sebaceum

Pringle's disease: skin only

Bonneville disease: nervous system only

Tuberous Sclerosis 2

- Epidemiology

-  → 40,000 Americans
-  → 2,000,000 worldwide

- No race or sex predilection

- 1/6800 in children age 11-15 yrs.; 1/12,900 in individuals age 0-20 yrs.

Genetic Basis

TSC 1 (1997)

TSC 2 (1993)

Chromosome
location

9q34

16p13

Mutations (#
abnormalities)

139

250

Protein name

hamartin

tuberin

Protein
Function

unknown

negative growth
regulator

Cortical Tubers (parenchymal hamartomas)

- Several millimeters to centimeters in size
- Rounded protrusions of single gyri
- Expanded gyri can blur white/gray margins
- **Inner core** typically hypointense on T1WI , hyperintense on T2WI vs. gray matter
- **Peripheral** component isointense to mildly hyperintense to gray matter on T2, T1WI

Cortical Tuber in Adult



a region of decreased signal intensity located within the left frontal cortex c/w a cortical tuber. Subependymal nodules are also present within the lateral ventricles.

Cortical Tuber Imaging

Note: Presence of (1) gyral deformity (2) abnormal thickening of cortical gray matter, and/or (3) blurring of gray-white junction

+

Lack of high signal on T2WI



Consider cortical dysplasia in ddx

Subependymal Nodules

- Originate from basal ganglia, from surface of caudate adjacent to foramina of Munro, or from 3rd, 4th ventricles
- Firm, hard secondary to calcification
- Isointense to hyperintense on T1WI, isointense to hypointense on T2WI (compared to gray matter)
- Signal void on T2WI. Utilize CT imaging

Subependymal Nodule in Adult



Right lateral ventricle
subependymal nodule,
near foramen of Munro



‘Candle gutterings’
(multiple, adjacent
nodules)

White Matter Lesions

- Oriented in radial pattern from ventricle to cortical surface
- Similar signal intensity to cortical tubers
- May represent areas of demyelination or hypomyelination
- Clusters of giant cells identical to those in tubers

White Matter Lesions in Adult



Nonspecific conglomerate, hypointense foci. Other patterns seen on MR:

- (1) straight/curvilinear bands
- (2) wedge-shaped lesions
- (3) cerebellar radial bands

Pediatric Tuber Imaging

- In infants <1 yr. old, appearance of cortical tubers differs from that in patients > 2 years, when myelination pattern = to adult
- Multiple case studies of neonates w/ inverse contrast behavior (Stricker *et al.* 1991; Altman *et al.* 1988)
- ➔ **hyperintense** to premyelinated white matter on T1WI, **hypointense** to premyelinated white matter on T2WI

Pediatric Tuber Imaging

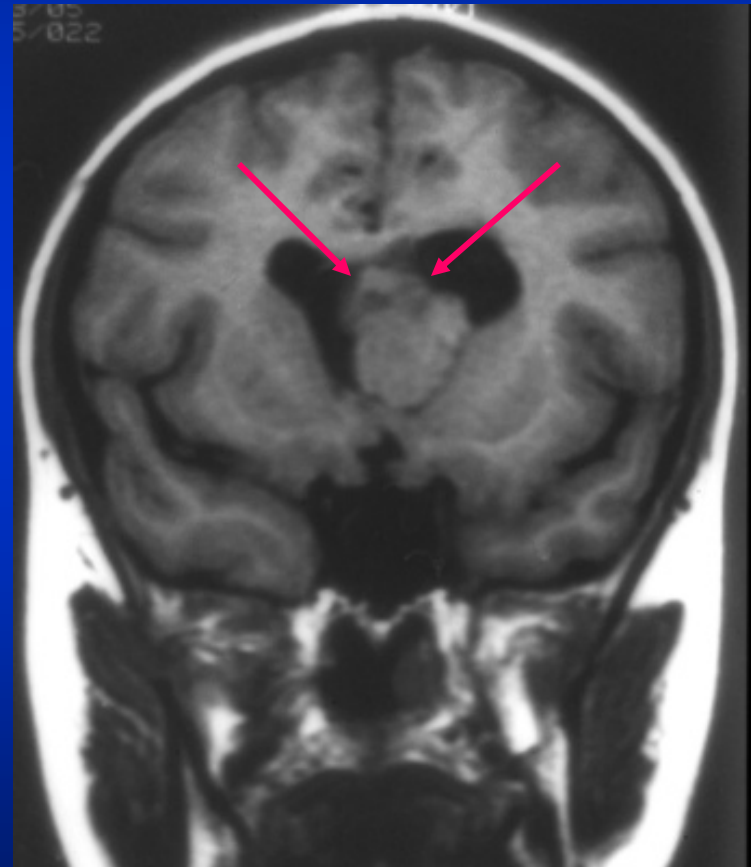
- Baron Y, Barkovich AJ. AJNR 1999.
- Examined MR characteristics of tuberous sclerosis in neonates and infants (N =7)
- **Results:** nodular subependymal and linear parenchymal lesions in infants < 3 yrs. are hyperintense on T1WI and hypointense on T2WI.
- Lack of myelination aids in ID of white matter anomalies; the latter are less visible as myelination occurs.

Our Patient: Follow-up

- Cardiology
- Neurology (6-9 weeks for brain imaging)
- Ophthalmology
- Renal U/S in 1-2 months
- Genetic testing

Why Follow-up?

- Malignant degeneration can occur!
- Subependymal tubers can become **giant cell astrocytomas**, usually at foramen of Munro
- Can result in secondary obstructive hydrocephalus





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