Intracerebral Tuberous 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 within left ventricle
MRI Axial T2WI

- Isointense to hyperintense signal
- Hypointense foci

BIDMC 2002
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

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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.

Kaiser V., Tarr R. University Hospital of Cleveland NeuroImaging Teaching Files. 2002.
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)

Kaiser V., Tarr R. University Hospital of Cleveland NeuroImaging Teaching Files. 2002.
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

Kaiser V., Tarr R. University Hospital of Cleveland NeuroImaging Teaching Files. 2002
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
References

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