AMSER Case of the Month
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A pediatric patient with epileptic seizures since infancy

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Inland Imaging
Patient Presentation

- **HPI:** Pediatric patient with a history of epilepsy with medically difficult to control seizures since infancy and a significant developmental delay with behavioral issues. Breakthrough seizure
- **PMHx:** Epilepsy, myotonic muscular dystrophy and autism
- **FMHx:** Father with Tuberous Sclerosis
What Imaging Should We Order?
Select the applicable ACR Appropriateness Criteria\(^1\)

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>CT head without IV contrast</td>
<td>Usually Appropriate</td>
<td>★★★★</td>
</tr>
<tr>
<td>MRI head without IV contrast</td>
<td>Usually Appropriate</td>
<td>0</td>
</tr>
<tr>
<td>MRI head without and with IV contrast</td>
<td>May Be Appropriate</td>
<td>0</td>
</tr>
<tr>
<td>CT head with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>★★★</td>
</tr>
<tr>
<td>CT head without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>★★★</td>
</tr>
<tr>
<td>FDG-PET/CT brain</td>
<td>Usually Not Appropriate</td>
<td>★★★</td>
</tr>
<tr>
<td>MEG</td>
<td>Usually Not Appropriate</td>
<td>0</td>
</tr>
<tr>
<td>MRI functional (fMRI) head without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>0</td>
</tr>
<tr>
<td>HMPAO SPECT or SPECT/CT brain ictal and interctal</td>
<td>Usually Not Appropriate</td>
<td>★★★</td>
</tr>
</tbody>
</table>

This imaging modality was ordered first
Followed by MRI head with contrast and CT abdomen pelvis with contrast to look for additional abnormalities
Head non-contrast CT Findings (unlabeled)
Head non-contrast CT Findings (labeled)

Yellow Arrows:
Calcified subependymal nodules
Head MRI Findings: (unlabeled)

Ax T1 C+

Ax T2 FLAIR
Head MRI Findings: (labeled)

Red Arrows: Cortical tubers are triangular-shaped lesions at cortex/juxtacortical location, with apex toward the ventricles

T1: low signal
T2/FLAIR: high signal
T1 C+ (Gd): lack of enhancement
Head MRI Findings: (unlabeled)
Head MRI Findings: (labeled)

Yellow arrows:
Subependymal hamartomas at the Foramen of Monro

Ax T1 C+
Ax T2 FLAIR
Abdomen and Pelvis CT Findings: (labeled)

Yellow arrows:
Multiple fat-attenuation renal masses consistent with bilateral angiomyolipomas
Final Diagnosis:

Tuberous Sclerosis
Case Discussion (1-3 slides)

• Presentation
  • Intellectual disability, infantile spasms, seizures, brain tumors, cardiac rhabdomyoma, and renal disease. ², ⁴
  • Skin/cutaneous ², ⁴
    • Adenoma sebaceum (facial angiofibromas)
    • ash leaf spots
    • shagreen patches
    • Ungual fibromas

• Diagnostic Studies
  • EEG², ⁴
    • Seizure activity
  • Genetic testing ², ⁴
    • Autosomal dominant mutation in tumor suppressor genes TSC1 or TSC2 gene
Case Discussion

• Diagnostic Imaging findings:
  • Neurologic 3
    • Cortical/subcortical tubers (50% in frontal lobe)
    • Subependymal hamartomas (88% with calcification)
    • Giant cell astrocytoma
    • White matter abnormalities such as radial bands
  • Abdominal 3
    • Renal cysts (18-53% of patients with tuberous sclerosis)
    • Angiomyolipoma (55-75% of patients with tuberous sclerosis)
    • Renal cell carcinoma (similar prevalence as general population but occur at younger age with tuberous sclerosis).
    • Pancreatic neuroendocrine tumors
    • Hepatic angiomyolipoma
Case Discussion

• Diagnostic imaging findings continued:
  • Thoracic
    • Cardiac rhabdomyoma (seen in 50-65% of patients with tuberous sclerosis, can cause ventricular hypertrophy and arrhythmias)
  • Musculoskeletal
    • Sclerotic bone lesions (seen in 40-66% of patients with tuberous sclerosis)
    • Scoliosis
    • Bone cysts

• Management
  • Anti-seizure medications for epilepsy.
  • mTOR inhibitors for renal angiomyolipoma and inoperable astrocytoma.
  • Surgery for obstructive hydrocephalus and drug resistant seizures
  • Renal disease and epilepsy are most common cause of death
References: