2014 Diagnostic Slide Session
Case 1

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Disclosures

No relevant financial relationships or conflicts of interest
Clinical History

- 72 yo F with depression, possible wide based-gait and chronic RA
- Admitted for septic shock
- Died 10 days after admission
- Perforated gastric ulcer on autopsy
Imaging

SWI

T2
Additional stains?

Diagnosis?
FINAL DIAGNOSIS

Neurodegeneration with Brain Iron Accumulation (NBIA)
• Clinically and genetically heterogeneous

• Pathologic Hallmarks
  • Iron deposition
  • Neuroaxonal spheroids
  • Lewy bodies +/- Tau pathology
NBIA

- Extrapyramidal symptoms, gait abnormalities, dementia
- Poor prognosis
- No definitive therapy
“Eye of the Tiger” Sign

http://rad.usuhs.edu/medpix/kiosk_image.html?imageid=55922&quiz=#pic
Imaging

SWI

T2
# NBIA Subtypes

<table>
<thead>
<tr>
<th>Type</th>
<th>Associated Gene</th>
<th>Pathologic Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type I / PKAN</td>
<td><em>PANK2</em></td>
<td>NFTs, spheroids</td>
</tr>
<tr>
<td>Type II / PLAN</td>
<td><em>PLA2G6</em></td>
<td>Lewy bodies, NFTs, spheroids</td>
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<tr>
<td>MPAN</td>
<td><em>c19orf12</em></td>
<td>Lewy bodies, NFTs, spheroids</td>
</tr>
<tr>
<td>BPAN</td>
<td><em>WDR45</em></td>
<td>Lewy bodies, NFTs, spheroids</td>
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<tr>
<td>ACP</td>
<td><em>CP</em></td>
<td>Foamy spheroid bodies, bizarre astrocytes</td>
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<tr>
<td>NFT</td>
<td><em>FTL</em></td>
<td>Distorted, iron-laden nuclei</td>
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<tr>
<td>FAHN</td>
<td><em>FA2H</em></td>
<td>???</td>
</tr>
<tr>
<td>KRS</td>
<td><em>ATP13A2</em></td>
<td>???</td>
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</tbody>
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Differential Diagnosis

- Huntington disease
- Wilson disease
- Other neuroaxonal dystrophy
- Parkinson’s disease
- Vitamin E deficiency


THANK YOU!