Brainstorming the Case: An unusual presentation of autoimmune encephalitis

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Disclosure of Financial Relationships

None
History of Present Illness

-Previously healthy 12-year-old girl presents to the ED with:
  - Altered mental status
  - Hypothermia (T 33.2 C)
  - Bradycardia (HR 42)

-One week of abdominal pain, fatigue and flattened affect
-Family denied ingestion, fever, cardiorespiratory symptoms, headaches or unusual exposures.

Additional Questions
Physical Exam

- Sluggishly reactive pupils
- Disconjugate gaze
- Intermittently follows commands and localizes to pain
- Bradycardic, otherwise regular rate and rhythm and no murmurs
- Abdomen soft with no masses or hepatosplenomegaly

Initial Differential Diagnosis
Additional Exam Technique

Initial Differential Diagnosis

- Infectious
  - Meningitis
  - Encephalitis
  - Systemic illness/Sepsis
- Psychiatric
- Autoimmune
- Endocrine
- Drugs/Toxins
- Metabolic
Orders

Hospital Course

Labs:

- Hyponatremia
- Hypokalemia
- Negative infectious work-up
- CSF with:
  - Lymphocytic pleocytosis
  - Oligoclonal bands
Hospital Course

• Imaging:
  • MRI Brain:
    • Non-enhancing, ill-defined lesion involving hypothalamus diencephalon
    • Scattered grey-matter lesions

• Pathology:
  • Brain Biopsy: hypercellularity

New information/Clinical Course

• Initially admitted to the PICU due to vital sign instability with hypothermia and bradycardia
• Intubated for a brief period of time
• Predominant symptoms included optic and motor deficits in addition to global encephalitis
• High dose pulse steroids and IVIG provided minimal, if any, improvement
• One dose of rituximab given with near-resolution of symptoms
Diagnostic Pause

Final Diagnosis

- NMO/AQP4 titer was positive in the CSF, but not the serum
- Diagnosed with Neuromyelitis optica (NMO) autoimmune encephalitis with hypothalamic involvement
Wrap Up

• Autoimmune encephalitis (AE) is an increasingly recognized etiology of encephalitis

• Challenging to diagnose due to:
  • Varied presentation
  • Limited antibody testing
  • Common presentations:
    • Delirium, seizures, behavioral changes
    • Autonomic instability, respiratory failure, coma, and sepsis

Wrap Up

• Neuromyelitis Optica usually presents with:
  • Optic neuritis or transverse myelitis
  • Can present as an autoimmune encephalitis
    • And rarely as specifically related to diencephalic and brainstem pathology as in this case

• ~ 65% of children with suspicion for NMO have positive AQP4-IgG in the serum

• Seronegativity should not exclude the diagnosis, AQP4-IgG may appear as late as 4-5 years after initial symptoms
Wrap Up

PEARLS

• Hypothalamic variants of AE, while rare, do exist and should be considered
• Early recognition and treatment can lead to reduced morbidity and mortality
• Absence of antibodies should not delay treatment if AE is suspected
Pearls

• Treatment includes immunomodulatory therapies
• First line therapies: Steroids, Plasmapheresis, IVIG
• Second-line therapies: Rituximab, Cyclophosphamide
• Clinical response should guide therapy
• Treatment may take days-to-weeks to take effect