

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.

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Additional Questions

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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

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Initial Differential Diagnosis

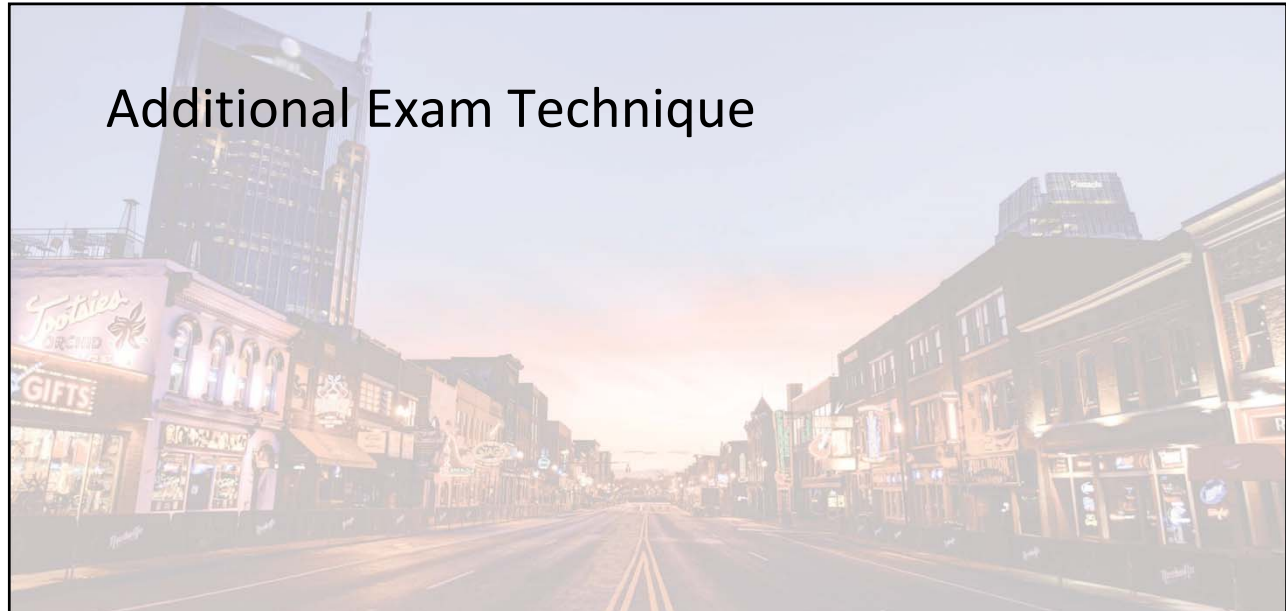
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Additional Exam Technique



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Initial Differential Diagnosis

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



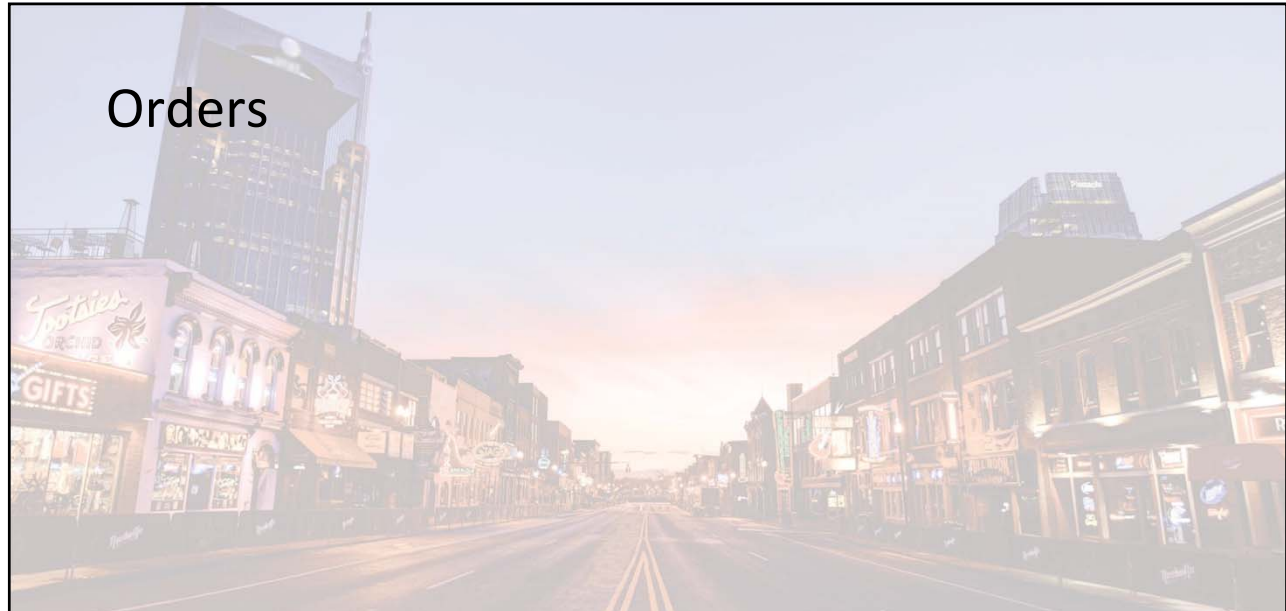
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Orders



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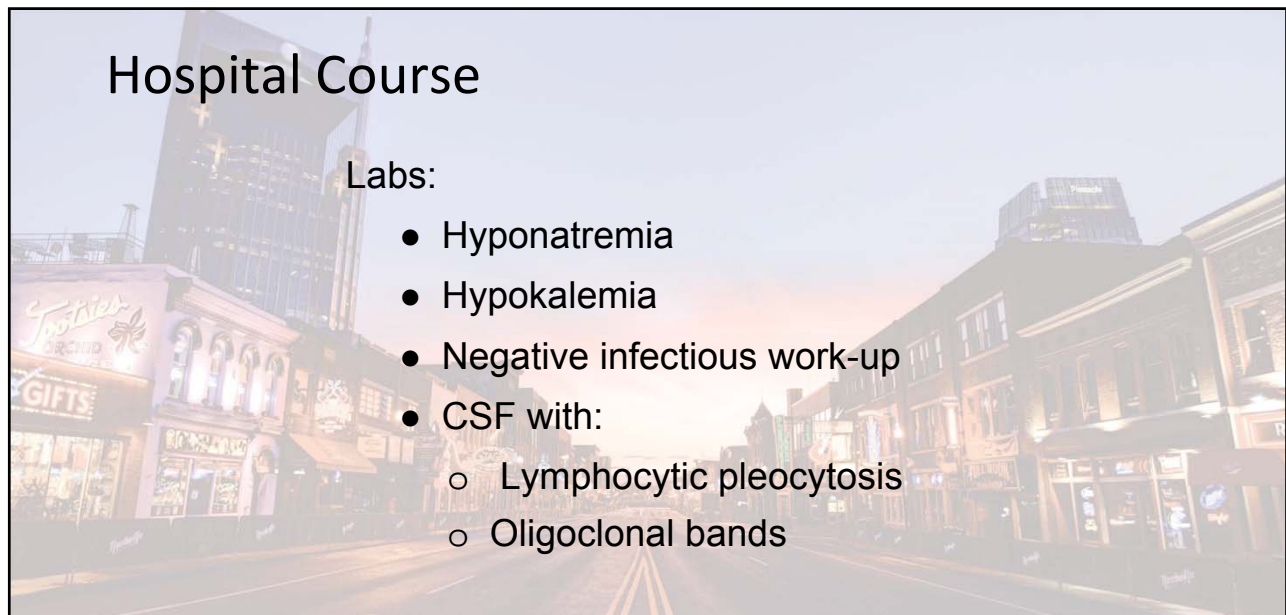
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Hospital Course

Labs:

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



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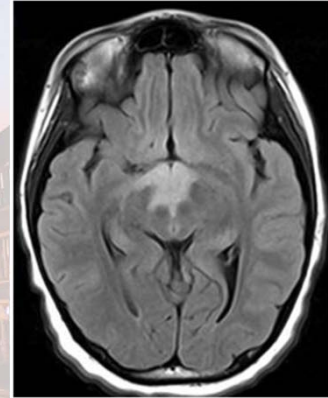
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Hospital Course

- 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



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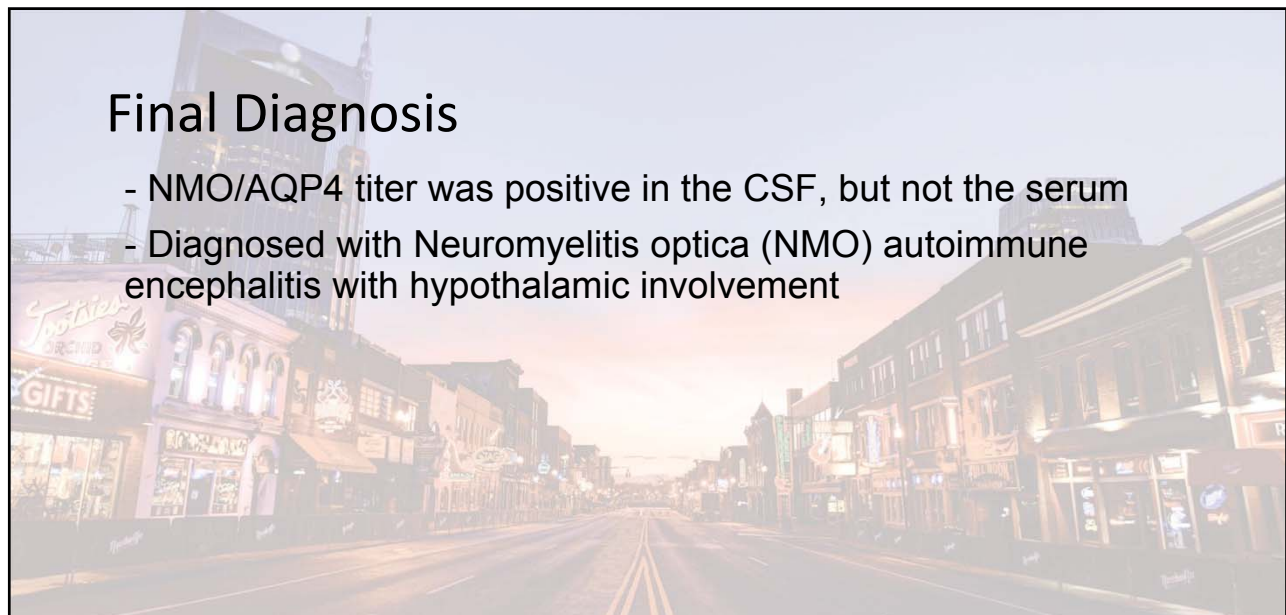
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Final Diagnosis

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



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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

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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

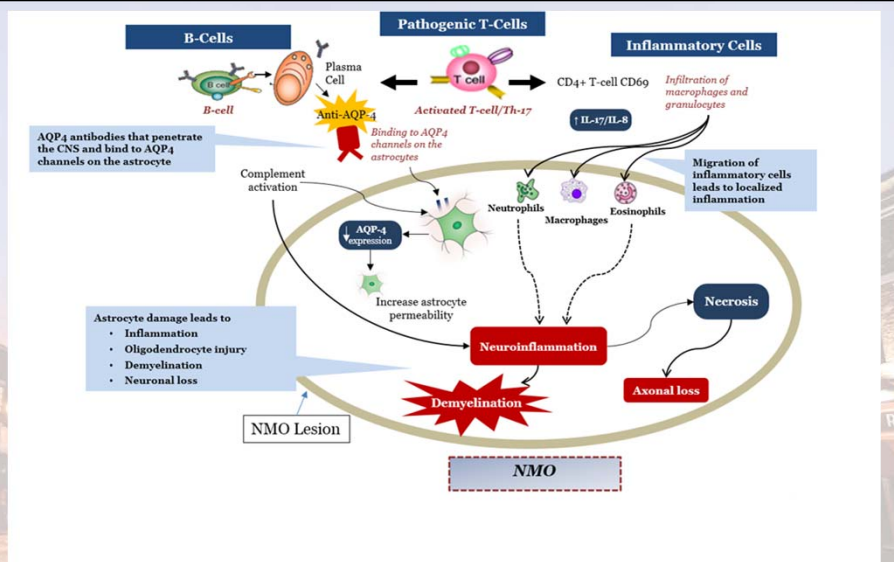
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Wrap Up



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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

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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

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