You Spin Me Right Round, Baby
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Disclosure of Financial Relationships

• No financial relationships to disclose relating to this presentation
History of Present Illness

• 16 year old male with APECED (autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy)
• Presents with intermittent episodes of vertigo, dizziness, headaches, blurry vision, that have increased in frequency over the last 5 months
• Episodes range from 1 day to 2 weeks and are self-resolving
• Mom has to help him ambulate during episodes due to unsteady gait
• Have presented symptoms to many specialists over the last few months with no answers

Past Medical History

• As a result of his APECED, he has:
  • Chronic autoimmune hepatitis
  • Chronic pancreatitis
  • Enteritis: has a g-tube to maximize his nutrition
  • Failure to thrive
  • Chronic kidney disease - stage II
  • Nephrolithiasis
  • Cytopenias of unknown etiology
  • Prolonged QTc
  • JRA
• On multiple medications including tacrolimus, ursodiol, and prednisone
• Previously on growth hormone
Physical Exam

• T: 98.1, BP 110/80, HR 71, RR 16, O2 97%, Wt 22.5kg
• No orthostatic hypotension
• General: small for age, chronically malnourished, otherwise comfortable
• Neuro:
  • Mentation intact
  • Dizziness reported with standing
  • Unsteady, shuffling, wide based gait
  • Cranial nerves were intact
  • No nystagmus or papilledema
  • Normal strength and reflexes in extremities

Growth Chart
Initial Differential Diagnosis?

A) Vestibular
B) Cardiac arrhythmia
C) Medication side effect
D) Infection: ADEM, encephalitis
E) Intracranial process

Initial Differential Diagnosis

- Vestibular etiology
- Vasovagal
- Arrhythmia
- Medication side effects
- Intracranial mass
- Sinus venous thrombosis
- Encephalitis
- Acute disseminated encephalomyelitis
Hospital Course

- Admitted to the acute care floor
- Remained stable, at his baseline
- Symptoms improved by morning, but parental concerns persisted due to chronic, intermittent nature of symptoms
- Consult – Ophthalmology, Neurology, ENT
- MRI brain

New information/Clinical Course

![Brain MRI images with highlighted areas]
New information/Clinical Course

Diagnostic Pause

- Differential based on imaging alone:
  - Vascular etiology (vasculitis, PRES)
  - Infection (viral > bacterial)
  - Inflammatory (ADEM)
  - Drugs/toxins (lead, cyanide, carbon monoxide)
  - Non-inherited metabolic (Wernicke’s encephalopathy, hepatic or renal failure)
  - Mitochondrial disorders
Final diagnosis?

A) Vascular etiology (vasculitis, PRES)
B) Infection (viral > bacterial)
C) Inflammatory (ADEM)
D) Drugs/toxins (lead, cyanide, carbon monoxide)
E) Non-inherited metabolic (Wernicke’s encephalopathy, hepatic or renal failure)

Final Diagnosis – Wernicke’s Encephalopathy

• Rare disorder in the pediatric population
• Classic triad – confusion, ophthalmoplegia, ataxia
• Majority of WE cases in pediatrics is in oncology patients, followed by those who receive prolonged parenteral nutrition, bariatric surgery, or those with eating disorders
Hospital Course

• Thiamine level was drawn (level was 60 nmol/L – normal is 70-180 nmol/L)
• Patient was empirically started on IV thiamine immediately
• Symptoms were likely due to his long standing history of autoimmune enteritis and malabsorption
• Drastic improvement in symptoms

Wrap Up – Pathophysiology

• Thiamine is absorbed in the duodenum and proximal jejunum
• Majority of patients do not present with class triad
  • Ataxia or mental status changes
  • Ocular findings
• Important to check for other vitamin level deficiencies
• Glucose metabolism relies on thiamine
Wrap Up – Final Thoughts

• Challenges in diagnosis
  • Complex medical history
  • Non-specific complaints
  • Intermittent, self-resolving nature of symptoms
• Nutrition status is often overshadowed by complex medical conditions
  • Integral to clinical status
  • Lack of exposure to nutritional deficiencies results in failed recognition

References