Case 7

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Financial Disclosures

• None
Clinical

- 55-year-old man
  - 3 months of progressive gait unsteadiness; numbness and tingling in feet and fingertips

- Neuro Exam:
  - Ataxic gait, positive Romberg, loss of vibration, proprioception, pinprick in feet, reflexes brisk in UEs absent in LEs

- Microcytic anemia

- Biopsy confirmation of celiac disease

- Negative laboratory tests:
  - vitamin B12, thiamin, vitamin E, SPEP, glucose, Hb A1C, LFTs, TSH, ANA, paraneoplastic, PET-CT, serologies (Anaplasma, Erlichia, Babesiosis, B. burgdorferi)
MR Image

Very subtle T2 signal hyperintensity in the dorsal columns
Clinical History

• Additional laboratory testing established a diagnosis

• Five months later, he collapsed suddenly and died

• Autopsy disclosed a ruptured basilar tip aneurysm with marked subarachnoid hemorrhage
Autopsy Findings
Spinal Cord Histology

H&E  Luxol Fast Blue  Neurofilament
Discussion

Diagnosis?
Diagnosis

Copper Deficiency Myelopathy

- Serum copper markedly decreased at 0.27 µg/mL (normal range 0.75-1.45)
- Ceruloplasmin decreased at 9.8 mg/dL (normal range 15-30)
- Serum zinc normal
Copper Deficiency Myelopathy

• Long recognized to occur in ruminant animals, termed “swayback” or “enzoonotic ataxia”

• Identical presentation and pathology to subacute combined degeneration due to B12 deficiency
  • Gait disturbances and paresthesias
  • Vacuolating myelin loss of the dorsal columns
  • Cervical spinal cord typically most severely affected

• Hematologic manifestations often also present (anemia and neutropenia)
Most Common Etiologies

- Upper GI surgery (including bariatric surgery)
- Celiac Disease
- Zinc overload, often from denture cream

Treatment and Patient Outcome

- Copper supplementation
  - Resolution of copper deficiency and hematologic abnormalities
  - Stabilization or minimal improvement of neurologic symptoms

Questions & Discussion