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Case Report
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Undiagnosed Achalasia Progressing to Wernicke's Encephalopathy: A Need for Early Detection and Treatment

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Abstract

A woman in her thirties was admitted for altered mental status and progressive ophthalmoplegia with a five-month history of weight loss and dysphagia. On admission, she was suspected to have Wernicke's encephalopathy and thiamine replacement therapy was initiated. Her neurological status and functional capacity gradually improved. Manometry was performed for dysphagia, and she was diagnosed with type II achalasia. Our case report demonstrates the importance of early detection and prevention of Wernicke's encephalopathy in patients with achalasia. Physicians should have a high suspicion of achalasia in patients presenting with unexpected weight loss, emesis, dysphagia, and regurgitation. Wernicke's encephalopathy should be considered when patients present with ataxia, confusion, and/or ophthalmoplegia even when thiamine levels are within reference range. Once a patient is suspected to have achalasia, initiating nutritional supplementation may be needed to prevent detrimental effects of malnutrition.

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Background

Esophageal achalasia is caused by impaired esophageal smooth muscle resulting in aperistalsis and failure of the lower esophageal sphincter to relax with swallowing. Annual achalasia incidence rate is believed to be 0.5-1.2 per 100000, however annual incidence has risen to 1.6 per 100000 [1]. Diagnosis is done by endoscopy, barium esophagram, and manometry [2]. Presenting symptoms include progressive dysphagia to solids and liquids, regurgitation, and respiratory complaints. Treatment is pneumatic balloon dilation or laparoscopic myotomy [1].

Detrimental long-term effects of achalasia include malnutrition, which likely occurs due to reduced food intake from difficulty swallowing, chest discomfort and regurgitation. According to a study, over 70% of patients were at moderate to high risk of malnutrition [3].

Thiamine deficiency is classically linked to chronic alcoholism which progresses to beriberi and Wernicke-Korsakoff syndrome [1]. However, in a postmortem series of 29 cases, in which chronic alcoholism was excluded, gastrointestinal causes of WE included peptic ulcers, acute pancreatitis, esophageal metastasis and stomach and esophageal carcinoma [4]. WE commonly present as ataxia, confusion, ophthalmoplegia, and MRI may demonstrate hyperintense signals in the dorsal medial thalamic nuclei, mammillary bodies, periaqueductal gray area and the third or fourth ventricles [5]. Treatment involves immediate administration of thiamine.

Case Presentation

A woman in her thirties with a history of dysphagia, opiate use disorder, and bipolar I disorder previously treated with lithium presented to the emergency department for altered mental status and acute bilateral, progressive "blurry" vision. Neurology had evaluated the patient at an outside hospital and appreciated mild horizontal nystagmus and bilateral central vision loss with limited peripheral vision. The patient was empirically treated with oral prednisone and vitamin B12.

Prior to the patient's visual deficits, she was having immediate regurgitation of foods and liquids and difficulty swallowing for the past five months resulting in 30 kg weight loss. She underwent an esophagogastroduodenoscopy that was unremarkable; her dysphagia was attributed to opiate-induced dysmotility. She was placed on pantoprazole 20 mg two times a day without symptomatic

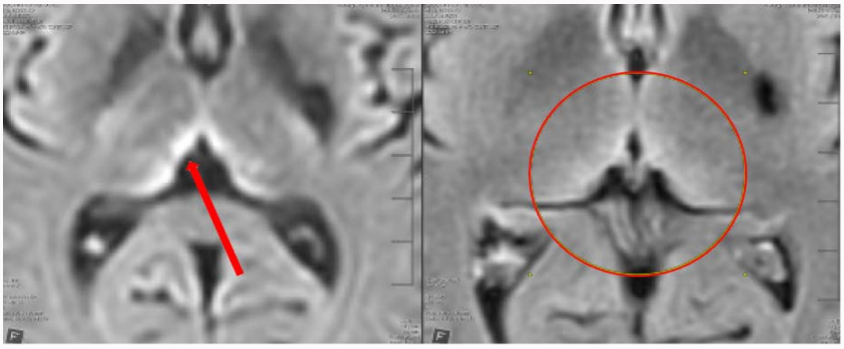

Figure 1: Diffusion-Weighted Imaging (DWI) and Fluid-Attenuated Inversion Recovery (FLAIR) hyperintensities of the walls of the third ventricle.

Table 1: Summary of case reports from PubMed search for "achalasia" AND "Wernicke's encephalopathy."

Demographics	Presentation	Neurological Symptoms	Neuroimaging	Treatment and Outcome
34-year-old female	- History of triple A syndrome - Fatigue and dizziness for 5 days	- Upbeat nystagmus - Horizontal gaze palsy - Gait ataxia - Mild confusion	- Signal elevation in the medial thalami and periaqueductal gray	- IV therapy of 300 mg of thiamine per day - Ocular abnormalities and gait ataxia improved
28-year-old male	- Dysphagia - Vomiting - Regurgitation - He was diagnosed with achalasia and underwent surgical therapy - On day 3, he became confused	- Pupils were sluggishly reactive - Bilateral vertical nystagmus - Truncal ataxia	Signal change in the posteromedial thalamus bilaterally	- Oral thiamine replacement - Discharged with minimal residual neurologic impairment
23-year-old female	- 15-year history of persistent postprandial vomiting - Dysphagia to solids and liquids - Heartburn - Weight loss	- Sixth nerve palsy - Confusion - Psychosis	N/A	- High dose thiamine given for three days
28-year-old female	- History of achalasia surgical therapy - Presented 45 days after childbirth, with recurrent vomiting during pregnancy	- Restricted extraocular motility to lateral gaze - Vertical nystagmus - Catatonia	Increased signal involving the mammillary bodies, periaqueductal area, and periventricular region of the third ventricle	- Parenteral thiamine - Significant clinical improvement
60-year-old man	- Dysphagia - Postprandial vomiting - Weight loss of 10 kg over 5 months. - Was diagnosed with achalasia and underwent surgical treatment	Two days post-surgery the patient developed: psychomotor agitation, confusion, delirium with frank visual hallucinations, retrograde, anterograde, and semantic memory were severely impaired, truncal and limb ataxia, incontinent of urine and feces	Symmetrical aqueductal, thalamic, mamillary body and quadrigeminal hyperintensity	- Parenteral thiamine for 5 days followed by enteral thiamine for 3 months - After 1 week of treatment: he was still disorientated, confabulating and incontinent. Severe retrograde and anterograde memory loss remained as did his visual hallucinations. Ataxia was still present. After 4 months of intensive neurorehabilitation, persistent anterograde memory loss and ataxia were still present
26-year-old female	- Acute worsening of chronic dyspepsia - Recurrent vomiting - Severe dehydration - Poor nutrition status - Diagnosed with achalasia and underwent surgical treatment	Five days post-surgery: diplopia, nystagmus, dysmetria, ataxia, and short-term memory loss	Hyperintensity of both thalamic system and mamillary bodies	- Intramuscular thiamine - Significant clinical improvement

improvement.

At our facility, physical examination was notable for impaired mental status with an inability to follow commands. Her eyes were asymmetrically responsive in blink to threat and exhibited intermittent horizontal nystagmus. Bilateral upper extremities were strongly resistant to movement; however, lower extremities exhibited decreased strength and areflexia with an inability to flex to noxious stimulation. MRI revealed hyperintensities of the walls of the third ventricle (Figure 1) and mammillary bodies.

Labs were significant for thiamine levels at 68 nmol/L (normal: 70 nmol/L to 180 nmol/L). A barium esophagram showed a distally tapered esophagus that was concerning for achalasia. The patient was started on aggressive thiamine replacement (beginning with 500 mg IV every hour) due to high suspicion of WE.

The patient's neurological status and functional capacity gradually improved. However, she continued to endorse severe dysphagia. The patient was discharged with an outpatient manometry, which demonstrated abnormal relaxation with aperistalsis consistent with type II achalasia. She underwent a laparoscopic Heller myotomy with no complications. One month after surgical intervention she reported marked improvement in tolerating oral intake.

Discussion

Our case brings light to the importance of early detection of achalasia. Physicians should have a high suspicion of achalasia in patients presenting with unexpected weight loss, emesis, dysphagia, and regurgitation. Initiating nutritional supplementation, even prior to endoscopic or radiographic confirmation of achalasia, may prevent detrimental effects of malnutrition.

There is a lack of literature examining the relationship between achalasia and malnutrition. A PubMed search for "achalasia" AND "Wernicke's encephalopathy" yielded 6 reports. The characteristics of the included case reports are summarized in Table 1 [6-11].

One way to assist physicians in having a high suspicion of achalasia on initial presentation of a patient is by utilizing a scoring system. Currently, no tool exists to categorize patients at low, moderate, or high-risk of having achalasia based on presenting symptoms. However, scoring tools for evaluation of severity of symptoms, stages, and efficacy of treatment exist [3]. The Eckardt Symptom Score (ESS) is used to assess severity of achalasia by examining symptoms of dysphagia, chest pain, regurgitation, and weight loss [2]. Patients determined as high-risk of achalasia may need to be initiated on a similar dose of thiamine supplementation.

Another benefit of utilizing a scoring tool to assess risk of achalasia is preventing physicians from misdiagnosing a patient. The patient was misdiagnosed with opiate-induced dysmotility due to her presenting symptoms of dysphagia and history of opioid use disorder. Our case demonstrates the negative impact of cognitive biases, specifically anchoring bias, which is an individual's tendency to depend on the first piece information offered [12].

All in all, the case demonstrates the importance of early detection and prevention of WE in patients with achalasia. The patient's thiamine level was just below the normal range, however, her symptoms improved with thiamine repletion. Providers should consider WE in their differential even when thiamine levels are within reference range because it is a clinical diagnosis. Future studies should investigate scoring tools to determine patients at high-risk of having achalasia to allow immediate thiamine monitoring and supplementation.

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