




# Hollow Visceral Myopathy, a Rare Gastrointestinal Disorder: A Case Report and Short Review

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## Abstract

Hollow visceral myopathy (HVM) is described as impaired intestinal function and motility in the absence of mechanical obstruction. In this case report, we describe a unique case of an 18-year-old female who presented to the hospital with complaints of persistent nausea, vomiting, inability to tolerate oral feeds, and substantial weight loss for 2 months. After appropriate investigations, a diagnosis of gastroparesis was established. The patient was started on metoclopramide, which led to significant symptomatic improvement, and she was eventually discharged home. One month after discharge, she presented to the hospital with symptoms similar to her initial presentation. After further laboratory and radiological investigation, she was diagnosed with severe gastroparesis and chronic intestinal pseudo-obstruction. Over the next month, the patient was given an extensive trial of multiple prokinetic agents such as mirtazapine, ondansetron, pyridostigmine, octreotide, and promethazine, but she failed to show clinical improvement. Due to failure of medical therapy, a nasojejunum feeding tube was placed for enteral nutrition. However, the patient reported worsening of her symptoms despite slow feeding rates; hence, a decision was made to start the patient on total parenteral nutrition and transfer her to a larger tertiary center for higher level of care. At the tertiary hospital, the patient was continued on total parenteral nutrition and underwent extensive evaluation. Ultimately, she was diagnosed with HVM after a laparoscopic full-thickness intestinal biopsy showed histopathological evidence of the disease. She underwent isolated small intestine transplant, which led to significant improvement of her symptoms and was eventually discharged home. The patient continues to be symptom-free and follows up with Gastroenterology and Transplant Surgery regularly. This case report highlights a rare clinical condition, HVM, as a potential diagnosis in patients with clinical features of intestinal obstruction without mechanical obstruction.

## Keywords

hollow visceral myopathy, familial myopathy, chronic intestinal pseudo-obstruction, intestinal transplant, gastroenterology

## Introduction

Visceral myopathies (VMs) consist of a heterogeneous group of genetic disorders characterized by dysfunctional smooth muscle cells.<sup>1</sup> It can be subdivided into different groups based on the area of smooth muscle involvement and clinical features. Enteric visceral myopathy (EVM), a subtype of VM, has predominantly gastrointestinal (GI) involvement with characteristic symptoms such as abdominal pain, distention, features of malabsorption, and, in some cases, even death.<sup>2</sup> EVM shows significant variance in clinical presentation with severe cases often diagnosed in the prenatal or neonatal period while milder forms of the disease may remain undiagnosed until adulthood.<sup>3</sup> Hollow VM (HVM), a milder form of EVM, is a rare and often underdiagnosed GI disorder with only a handful of cases described in current literature. It

is characterized by the presence of impaired intestinal function or motility in the absence of an obvious mechanical occlusion.<sup>4</sup> It may involve the smooth muscles of the whole or specific sections of the GI tract and occasionally the smooth muscles of the urinary tract.<sup>5</sup> In the literature, patients

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with HVM usually have a chronic duration of symptoms coupled with nausea, vomiting, abdominal pain, and distention, such as that seen in our patient.<sup>6</sup> Furthermore, to establish a diagnosis, mechanical obstruction should be ruled out via radiological investigations, and these patients often require histological examination of the bowel mucosa, both of which were performed for our patient.<sup>7</sup> Our case report is unique as it not only describes a rare clinical condition but also discusses the diagnostic challenges associated with the disease entity due to varied clinical symptoms and its similarity with other more common GI disorders. Additionally, we also describe the management strategies for these patients. This case report is presented in accordance with the CARE reporting checklist.

## Case Presentation

An 18-year-old female with a past medical history of migraine headaches presented to the hospital with chief complaints of persistent nausea, vomiting, inability to tolerate oral feeds, and significant weight loss for the past 2 months. The patient reported that she vomited after every meal, and vomitus mainly consisted of undigested food particles without blood. The patient also had intermittent constipation; however, she had no diarrhea. She also observed substantial weight loss despite good appetite but was unable to quantify the weight loss. The patient did not take any medications for her migraine headaches and her last flare-up was about 1 year ago. She reported multiple outpatient visits for similar complaints in the last 2 months; however, on this visit with her primary care provider, she was referred to the hospital. On clinical evaluation, she had stable vital signs. Physical examination revealed dry mucous membranes and abdominal examination was positive for mild epigastric tenderness along with decreased bowel sounds. The patient was started on intravenous (IV) fluids. Laboratory investigations such as complete blood count and comprehensive metabolic panel were ordered, which revealed a low hemoglobin level of 10.2 g/dL, hematocrit 30.2%, serum sodium 132 mEq/L, and serum potassium 3.3 mEq/L. An abdominal X-ray did not reveal mechanical obstruction. Due to concerns for gastroparesis (GP) as the underlying etiology, a Gastric Emptying Study was ordered, which revealed markedly delayed gastric emptying time as the tracer was distributed in the stomach on initial views with activity not appearing in the duodenum until 45 minutes and emptying half time was not reached with 77% of initial gastric contents remaining at the end of the examination. Hence, a diagnosis of GP was established. The patient was started on metoclopramide, which led to improvement of her presenting symptoms, and she was eventually discharged home with recommendations to taper the doses of metoclopramide as needed.

One month after discharge, the patient presented to the hospital with similar complaints as her initial presentation despite metoclopramide therapy. Laboratory investigations

were similar to initial hospitalization and revealed hemoglobin level of 10.7 g/dL, hematocrit 31.9%, serum sodium 133 mEq/L, and serum potassium 3.2 mEq/L. An abdominal X-ray was ordered, which revealed dilated bowel segments with high burden of stool in the colon without obvious mechanical obstruction. A diagnosis of severe GP and chronic intestinal pseudo-obstruction (CIPO) was established. Over the next month, the patient was given an extensive trial of numerous prokinetic agents such as mirtazapine, ondansetron, pyridostigmine, octreotide, and promethazine, but she failed to show clinical improvement. With failure of medical therapy, a nasojejunum feeding tube was placed for nutrition; however, the patient noted worsening nausea despite the slow feeding rate of 10 mL/h. Hence, after detailed discussions with the patient, a decision was made to start her on total parenteral nutrition (TPN) after which she was transferred to a tertiary center for higher level of care. At the tertiary care center, she was continued on TPN and underwent extensive evaluation. Laboratory investigations were within normal limits except mild elevation of the liver enzymes. Additionally, antinuclear antibody, extractable nuclear antigens panel, paraneoplastic antibody panel, and acetylcholine receptor binding antibodies were found to be negative, thereby ruling out an autoimmune etiology. A right upper quadrant ultrasound revealed the presence of gallbladder sludge and normal liver architecture. An esophagogastroduodenoscopy (EGD) and colonoscopy were found to be negative. A small bowel follow-through ruled out mechanical obstruction. Magnetic resonance enterography and computed tomography angiography of the abdomen were noted to be unremarkable. Furthermore, a deep rectal biopsy was performed, which was negative for amyloidosis. However, antroduodenal manometry, a diagnostic tool for GI motility disorders, showed paucity of contractile activity with very low amplitudes highly suggestive of a myopathic etiology. This was followed up with a laparoscopic full-thickness intestinal biopsy, which revealed decreased actin staining in the circular muscles of the muscularis propria of small intestine consistent with a diagnosis of HVM. During the course of the hospital stay, the patient developed recurrent bacteremia, the source of which was identified to be the central venous catheter used for TPN. She was treated with appropriate antibiotics, which led to resolution of the bacteremia. Eventually, she underwent isolated small intestine transplant and was started on mycophenolate 500 mg daily, everolimus 5 mg daily, and prednisone 10 mg daily. This led to eventual resolution of her presenting symptoms, and she was discharged home. The patient continues to be asymptomatic and follows up with Gastroenterology and Transplant Surgery regularly.

## Discussion

VM may be classified as inherited, primary idiopathic, or secondary to other etiologies (Table 1).<sup>8</sup> Intestinal involvement may range from malrotations, microcolon (neonates),

**Table 1.** Classification of Visceral Myopathy.

Inherited visceral myopathy	Primary idiopathic visceral myopathy	Visceral myopathy secondary to other disorders
Familial visceral myopathy (eg, hollow visceral myopathy)	Nonfamilial visceral myopathy	Scleroderma Systemic lupus erythematosus Amyloidosis Ehler-Danlos syndrome Stroke Encephalitis

and CIPO (all ages) to a more severe form, namely, megacystis-microcolon-intestinal hypoperistalsis syndrome. The pathogenic mechanism of VM includes missense mutations of genes such as ACTG2 and MYH11 leading to the synthesis of abnormal proteins, which are hypothesized to affect the polymerization of actin filaments thereby affecting smooth muscle contraction.<sup>9,10</sup>

HVM, a milder form of VM, is a pathological condition involving the smooth muscles of the GI tract resulting in impaired intestinal function and motility in the absence of a mechanical obstruction.<sup>4</sup> It is a rare cause of CIPO, which results from failure of propulsive peristalsis secondary to myogenic-mediated disturbances of the GI tract.<sup>11</sup> The affected smooth muscles undergo progressive degeneration and fibrous replacement resulting in the loss of GI motility.<sup>12</sup>

In patients with HVM, there is significant variance in the involvement of the GI tract, which may either be diffuse or limited to specific focal segments of the bowel.<sup>12</sup> This leads to a wide spectrum of clinical features. These patients may be completely asymptomatic or may present to the emergency department with symptoms of acute intestinal pseudo-obstruction (Ogilvie syndrome), while others, such as that seen in our case, may have symptoms of CIPO. Additionally, a rare case of intestinal perforation secondary to VM has also been reported.<sup>6</sup> As per current literature, the predominant symptoms include nausea, vomiting, chronic constipation, abdominal pain, abdominal distension, and features of malnutrition. A few cases were also reported to present with diarrhea and steatorrhea secondary to small intestinal bacterial overgrowth due to intestinal stasis.<sup>13</sup> Furthermore, extraintestinal manifestations reported in literature include ophthalmoplegia, megacystis, dementia, and seizures.<sup>14,15</sup> On examination, these patients may have abdominal distention and abdominal tenderness of varying degree, such as that seen in our patient.

Establishing a diagnosis of HVM is challenging and it is often delayed due to significant overlap in the clinical features with other more common GI disorders. In these patients, mechanical obstruction must be ruled out, and the diagnosis is usually established via histological examination of the layers of the intestinal mucosa.<sup>9</sup> An abdominal X-ray is usually the first investigation and may reveal bowel distension and air fluid levels without signs of mechanical obstruction. In

our case, the abdominal X-ray showed dilated bowel loops with evidence of high stool burden in the colon without obvious mechanical obstruction. An EGD may be performed to not only rule out obstruction but to also obtain duodenal biopsies to help rule out celiac disease or eosinophilic gastroenteropathy. In our case, EGD and colonoscopy were both negative. Furthermore, a small bowel follow-through was also performed to rule out distal intestinal obstruction. Other advanced imaging techniques such as magnetic resonance enterography allows simultaneous internal and external views of the gut wall to investigate possible causes of gut compression, while computed tomography angiography may be used to diagnose vascular abnormalities. When a neuromuscular motility disorder is suspected, intestinal manometry is an important and often diagnostic test. On manometry, uncoordinated contractions are seen in patients with neuropathic disorders, while those with myopathies have low amplitude coordinated contractions.<sup>16</sup> Moreover, along with imaging studies, these patients need extensive laboratory investigations to rule out secondary causes of CIPO such as diabetes mellitus (HbA1C), celiac disease (anti-tissue transglutaminase and antigliadin antibodies), connective tissue, and skeletal muscle disorders (antinuclear antibody, anti-double strand DNA antibody, anti-Scl-70 antibody, creatinine kinase, and aldolase). Additionally, thyroid-stimulating hormone for hypothyroidism, urine catecholamines for pheochromocytoma, and enteric neuronal antibodies (anti-hu antibodies) in patients with suspected paraneoplastic antibodies should also be investigated. The gold standard test to establish a diagnosis of HVM is the full-thickness intestinal biopsy, which can be done via a laparoscopic or endoscopic approach.<sup>17</sup> On histopathological analysis, there is decreased muscle actin staining, vacuolar degeneration of smooth muscle, or fibrous replacement of the smooth muscle in the muscularis propria.<sup>9</sup> Additionally, it may also rule out other disorders such as amyloidosis and systemic sclerosis.

The treatment for HVM, like the diagnosis, is often challenging and involves a multidisciplinary approach. The focus of management is symptomatic improvement, improvement of the nutritional status, and avoiding complications related to nutrition. For symptomatic improvement, patients are encouraged to have small, frequent meals with low fat and fiber content. Prokinetic agents, which increase intestinal transit times such as

metoclopramide, erythromycin, domperidone, somatostatin analogues (octreotide), cholinesterase inhibitors (neostigmine and pyridostigmine), and serotonergic agents (prucalopride), may also be used.<sup>2,18</sup> However, no current evidence exists which supports their use.<sup>18</sup> Gastric electrical stimulation may be considered a therapeutic option in patients with intractable nausea and vomiting. Additionally, these patients may require IV fluids and electrolyte replacement along with gastric and colonic decompression with nasogastric or rectal tubes.<sup>6</sup> From a nutritional perspective, enteral feeding may be initiated in patients with persistent symptoms and intolerance to oral feeds. In severe cases, TPN may be required; however, TPN is associated with numerous complications such as catheter-related sepsis. Our patient developed recurrent bacteremia secondary to the central venous catheter used for TPN. She was treated with appropriate antibiotics, which led to resolution of her bacteremia. As per literature, in patients with severe complications of TPN such as  $\geq 3$  catheter-related sepsis, intestinal transplant (isolated or multivisceral) is the treatment of choice.<sup>19</sup> Hence, our patient underwent an isolated small intestine transplant which included proximal segment end-to-side anastomosis of the proximal donor intestine to the recipient jejunum, distal segment end-to-side anastomosis of the donor bowel to the recipient sigmoid colon, and creation of an ileostomy from a cadaveric donor. She was started on a combination of mycophenolate, everolimus, and prednisone after the transplant, which led to marked clinical improvement.

## Conclusion

HVM is a rare GI disorder and may often be underdiagnosed due to an overlap of symptoms with other more common GI disorders. It should be considered as a differential diagnosis in patients presenting with clinical features of intestinal obstruction without radiological evidence. The first imaging investigation of choice for these patients is an abdominal X-ray. Intestinal manometry, which demonstrates low-amplitude coordinated contractions in myopathies, may also be useful. The gold standard diagnostic test for HVM is a full-thickness intestinal biopsy, either laparoscopic or endoscopic, with histopathological analysis. The management for these patients is focused on symptomatic improvement with dietary modifications and use of prokinetic agents, improvement in nutritional status via enteral nutrition and TPN in severe cases if necessary, and the avoidance of complications. Patients who develop complications on TPN benefit from intestinal transplant.

## Author Contributions

Conception and design: Dushyant Singh Dahiya  
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 Review of literature and data collection: All authors  
 Drafting the manuscript: All authors  
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Final approval of manuscript: All authors

Agreement to be accountable for all aspects of the work: All authors

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## Ethics Approval

Our institution does not require ethical approval for reporting individual cases or case series.


## Informed Consent

Verbal informed consent was obtained from the patient(s) for their anonymized information to be published.

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