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Isolated Exocrine Pancreatic Insufficiency Presenting as Chronic Refractory Diarrhea in a Young Adult: A Case Report.

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Abstract

Background: Exocrine pancreatic insufficiency (EPI) is characterized by inadequate secretion of pancreatic digestive enzymes resulting in maldigestion and malabsorption. It is commonly associated with chronic pancreatitis, pancreatic surgery, cystic fibrosis, or diabetes mellitus. Isolated EPI in young adults without structural pancreatic disease is uncommon and often remains undiagnosed for prolonged periods. ***Case Presentation:*** A 21-year-old male presented to a private gastroenterology clinic in Begusarai, Bihar, with chronic diarrhea of one year's duration. The patient reported 4–6 loose stools daily, associated with abdominal discomfort, weight loss, and generalized weakness. Multiple empirical treatments including antibiotics, probiotics, antispasmodics, and antidiarrheal medications had been prescribed without significant improvement. Extensive investigations for infectious, inflammatory, and metabolic causes were unremarkable. Stool culture showed no pathogenic organisms, and fecal calprotectin was within the normal range (15.10 µg/g), excluding inflammatory bowel disease. Glycemic parameters were normal with HbA1c of 5.1%, and there was no history of diabetes mellitus, alcohol use, or pancreatitis. Further evaluation revealed a markedly reduced fecal pancreatic elastase level of 10 µg/g, confirming severe exocrine pancreatic insufficiency. Pancreatic enzyme replacement therapy was initiated, resulting in dramatic symptomatic improvement within weeks, with normalization of bowel habits and weight gain during follow-up. ***Conclusion:*** This case highlights isolated exocrine pancreatic insufficiency as an uncommon but treatable cause of chronic diarrhea in young adults. Measurement of fecal elastase should be considered in patients with persistent unexplained diarrhea when routine investigations are inconclusive.

Keywords: Exocrine pancreatic insufficiency, chronic diarrhea, fecal elastase, pancreatic enzyme replacement therapy, malabsorption.

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INTRODUCTION

Chronic diarrhea is a common clinical problem encountered in gastroenterology practice and may arise from infectious, inflammatory, malabsorptive, endocrine, or functional gastrointestinal disorders. While conditions such as inflammatory bowel disease, celiac disease, irritable bowel syndrome, and chronic infections are frequently investigated, exocrine pancreatic insufficiency (EPI) is often overlooked, particularly in young patients without apparent pancreatic disease. Exocrine pancreatic insufficiency results from inadequate production or delivery of pancreatic enzymes required for digestion of fats, proteins, and carbohydrates. Deficiency of these enzymes leads to maldigestion, nutrient malabsorption, steatorrhea, weight loss, and various nutritional deficiencies(1). The condition is classically associated with chronic pancreatitis, pancreatic carcinoma, pancreatic resection, cystic fibrosis, and advanced diabetes mellitus. However, isolated EPI without structural pancreatic abnormalities is increasingly recognized but remains underdiagnosed. Fecal pancreatic elastase measurement has emerged as a reliable, non-invasive, and highly sensitive test for assessing pancreatic exocrine function. Values below 200 µg/g suggest pancreatic insufficiency, whereas levels below 100 µg/g indicate severe disease. Because symptoms overlap with more common gastrointestinal disorders, diagnosis is frequently delayed, resulting in prolonged patient morbidity(2). The present case describes a young adult with chronic refractory diarrhea who underwent extensive evaluation over a period of one year. Routine investigations failed to identify an etiology until fecal elastase testing revealed severe exocrine pancreatic insufficiency. The absence of diabetes mellitus, chronic pancreatitis, pancreatic surgery, and inflammatory bowel disease makes this case unusual and clinically significant. Early recognition and treatment with pancreatic enzyme replacement therapy led to complete symptomatic resolution and improved quality of life(3)..

CASE PRESENTATION

A 21-year-old male presented with complaints of chronic loose stools for approximately one year. The patient experienced 4–6 episodes of non-bloody loose stools daily associated with abdominal bloating, excessive flatulence, intermittent abdominal discomfort, generalized weakness, and gradual weight loss.

Several consultations had been sought previously, and multiple therapeutic trials including antibiotics, probiotics, proton pump inhibitors, antispasmodics, and antidiarrheal agents were administered without sustained benefit.
There was no history of:

- Diabetes mellitus
- Chronic alcohol consumption
- Recurrent abdominal pain suggestive of pancreatitis
- Pancreatic surgery
- Tuberculosis
- Celiac disease
- Family history of pancreatic disorders

Physical examination revealed a thin-built young male with mild nutritional compromise. Vital parameters were stable.

Investigations

Table 1. Summary of Relevant Laboratory Findings

Investigation	Result	Reference Range
Hemoglobin	14.6 g/dL	13–17
HbA1c	5.1%	Non-diabetic
Serum Amylase	50 U/L	28–100
Serum Lipase	13.84 U/L	13–60
Total Bilirubin	0.27 mg/dL	<1.2
Fecal Calprotectin	15.10 µg/g	≤50
Stool Culture	Negative	No pathogen
Fecal Elastase	10 µg/g	>200 Normal

The patient's stool calprotectin was normal, making inflammatory bowel disease unlikely. Stool culture did not reveal pathogenic bacterial growth. HbA1c was normal, excluding diabetes mellitus. Serum pancreatic enzymes were within reference limits. Most importantly, fecal pancreatic elastase was only 10 µg/g, strongly indicating severe exocrine pancreatic insufficiency.

Diagnosis
Based on:

1. Chronic refractory diarrhea
2. Exclusion of infectious causes
3. Normal inflammatory markers
4. Absence of diabetes mellitus
5. Markedly reduced fecal elastase (10 µg/g)

A diagnosis of **isolated severe exocrine pancreatic insufficiency** was established.

Management
The patient was started on pancreatic enzyme replacement therapy (PERT) containing pancreatic lipase, amylase, and protease with meals.
Additional recommendations included:

- Nutritional counseling
- High-protein diet
- Fat-soluble vitamin supplementation
- Regular follow-up

Outcome and Follow-Up
Within four weeks of initiating pancreatic enzyme supplementation:

- Stool frequency reduced from 5–6 episodes/day to 1–2 formed stools/day.
- Abdominal bloating improved significantly.
- Appetite improved.
- Weight gain of approximately 3 kg was documented over two months.
- General well-being and daily functioning improved markedly.

No adverse effects were reported.

DISCUSSION

Exocrine pancreatic insufficiency is frequently underrecognized in patients presenting with chronic diarrhea. Most clinicians initially investigate infectious, inflammatory, and functional gastrointestinal disorders. Consequently, diagnosis may be delayed for months or years, especially in younger patients lacking classical risk factors(4). The present case is noteworthy because the patient had severe pancreatic exocrine dysfunction despite having no history of chronic pancreatitis, diabetes mellitus, pancreatic surgery, alcohol abuse, or cystic fibrosis. The diagnosis became evident only after measurement of fecal pancreatic elastase, which was profoundly reduced at 10 µg/g, consistent with severe EPI(5).

Fecal elastase has become the preferred screening test for pancreatic insufficiency because it is non-invasive, inexpensive, and unaffected by pancreatic enzyme supplementation. Values below 100 µg/g are highly suggestive of severe exocrine dysfunction. An important aspect of this case was the extensive negative workup. Stool culture was negative, calprotectin levels were normal, and microscopic stool examination revealed no evidence of parasitic infection or inflammatory disease. These findings effectively excluded common causes of chronic diarrhea and directed attention toward pancreatic function testing(6).

The normal HbA1c value and absence of endocrine dysfunction indicate preserved pancreatic islet-cell function despite marked exocrine impairment. This phenomenon has occasionally been reported in isolated pancreatic acinar dysfunction and supports the concept that endocrine and exocrine pancreatic functions may be affected independently. The dramatic clinical response following pancreatic enzyme replacement therapy further confirmed the diagnosis. Symptomatic improvement after PERT is considered an important therapeutic and diagnostic indicator in suspected EPI(7).

This case emphasizes the need for heightened clinical awareness of pancreatic exocrine disorders in patients with chronic unexplained diarrhea. Earlier diagnosis can prevent unnecessary investigations, prolonged symptoms, nutritional deficiencies, and reduced quality of life(8).

CONCLUSION

Isolated exocrine pancreatic insufficiency is an uncommon but important differential diagnosis in patients presenting with chronic refractory diarrhea. Young adults without diabetes, pancreatitis, or structural pancreatic disease may still develop severe pancreatic exocrine dysfunction. Routine investigations may be unrevealing, leading to diagnostic delays. Fecal pancreatic elastase testing provides a simple, reliable, and non-invasive method for identifying exocrine pancreatic insufficiency. Early recognition and initiation of pancreatic enzyme replacement therapy can result in remarkable clinical improvement and restoration of nutritional status. Clinicians should maintain a high index of suspicion for EPI in patients with persistent unexplained diarrhea and malabsorption, particularly when conventional investigations are negative.

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