Gaucher Disease Complications: A Case of Gaucheroma and Protein-Losing Enteropathy

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Fac TJ [illegible] analyzed for the presence of Gaucher cells, conrming the diagnosis. Based on the ndings, the treatment regimen was adjusted to optimize enzyme replacement therapy dosage. Nutritional support was initiated, including a high-protein diet and possible enteral feeding, to manage protein loss. Symptomatic treatments for diarrhea were also implemented. e patient was scheduled for regular follow-up visits to monitor response to therapy, assess gastrointestinal symptoms, and perform repeat imaging studies as necessary. is structured approach allowed for comprehensive management of the patient's condition, facilitating timely interventions and optimizing clinical outcomes.

Results and Discussion

e patient, an age was diagnosed with Gaucher disease prior to presentation. Initially managed with enzyme replacement therapy, the patient presented to our clinic with progressive abdominal distension, signi cant weight loss, and persistent diarrhea lasting over [9]. Upon physical examination, notable ndings included abdominal tenderness and signs of dehydration. Laboratory tests revealed hypoproteinemia and low serum albumin levels, indicative of protein-losing enteropathy. Imaging studies, including abdominal ultrasound and CT scan, con rmed the presence of a gaucheroma located in the which was compressing the adjacent intestinal structures. e diagnosis of gaucheroma was con rmed through a biopsy, showing characteristic Gaucher cells. e management plan included adjusting the enzyme replacement therapy dosage and initiating nutritional support to

address the protein loss. e patient was also started on symptomatic treatment for diarrhea and monitoring for any signs of obstruction or further complications. e coexistence of gaucheroma and protein-losing enteropathy in this patient underscores the complexity of Gaucher disease and its potential for unusual presentations. While gaucheroma is a rare complication, it can signi cantly a ect gastrointestinal function and contribute to malnutrition, which may further exacerbate the underlying condition [10]. is case emphasizes the necessity for clinicians to be vigilant in recognizing atypical complications in patients with Gaucher disease. Early diagnosis and comprehensive management can improve outcomes and enhance the quality of life for a ected individuals. Continued research into the mechanisms behind these complications may lead to better therapeutic strategies and improved patient care.

Conclusion

is case highlights the importance of recognizing rare complications associated with Gaucher disease, speci cally gaucheroma and protein-losing enteropathy. Despite the well-established treatment protocols for managing Gaucher disease, the emergence of atypical manifestations can signi cantly complicate patient care. Early diagnosis and a multidisciplinary approach are essential for e ectively addressing these complications, optimizing treatment strategies, and improving patient outcomes. Continued awareness and research into the varied presentations of Gaucher disease will enhance our understanding and management of this complex disorder, ultimately leading to better support for a ected individuals.

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Interest of Con ict

None

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