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Case Reports

Refractory Diarrhoea in Hyaline Fibromatosis Syndrome: A 5-Month-Old Infant Stabilized Through Precision Gut-Directed Nutritional Therapy

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ABSTRACT

Hyaline Fibromatosis Syndrome (HFS) is an uncommon autosomal recessive disorder characterized by progressive deposition of hyaline material across multiple organ systems. In infants, the presentation may be subtle initially, but progression often leads to significant gastrointestinal complications. This case highlights a 5-month-old infant presenting with severe refractory diarrhoea, profound feeding difficulty, failure to thrive, and E. coli urosepsis. Conventional management failed, prompting the introduction of a structured gut-directed nutritional therapy involving semi-elemental feeds, osmotic load reduction, and carefully titrated fluid-electrolyte management. The infant demonstrated marked improvement, emphasizing the importance of aligning nutritional strategies with the underlying mucosal pathology in HFS.

Keywords: Hyaline Fibromatosis Syndrome; Refractory Diarrhoea; Gut-Directed Nutritional Therapy; Semi-Elemental Feeding; Infant Nutrition; Gastrointestinal Manifestations

INTRODUCTION

Hyaline Fibromatosis Syndrome (HFS) encompasses a spectrum of disorders caused by pathogenic variants in ANTXR2, resulting in the deposition of amorphous eosinophilic hyaline material in the skin, gastrointestinal tract, and musculoskeletal system. Although dermatological and joint-related manifestations often dominate the clinical picture, gastrointestinal symptoms may lead to the most severe complications, including dehydration, electrolyte imbalance, malnutrition, and recurrent infections. The pathophysiological basis involves disruption of the lamina propria, reduced absorptive surface, mucosal stiffness, and impaired intestinal motility. Despite these significant implications, formal guidelines for the management of diarrhoea in HFS remain limited. This case sheds light on a physiology-driven nutritional intervention that may improve outcomes in similar clinical scenarios.

CASE PRESENTATION

A 5-month-old male infant, born at term with no perinatal complications, demonstrated progressive symptoms beginning at approximately two months of age. The earliest findings included irritability during handling, firm plaques over the scalp and neck, and the appearance of small papulonodular lesions near the perianal and auricular areas. These manifestations gradually worsened and were accompanied by feeding difficulty and faltering growth, raising concerns for an underlying systemic disorder. At 3½ months of age, dermatology evaluation noted prominent indurated plaques and rubbery nodules. A biopsy of these lesions revealed deposition of eosinophilic hyaline material, confirming the diagnosis of Hyaline Fibromatosis. Syndrome. At the time of acute admission, the child was severely dehydrated, lethargic, and markedly underweight. He had experienced persistent watery diarrhoea for ten days, leading to oliguria and worsening irritability. Laboratory investigations demonstrated leukocytosis, elevated inflammatory markers, metabolic acidosis, hyponatremia, and urine culture positive for E. coli. Standard management was initiated, including intravenous antibiotics, fluid correction using balanced crystalloids, zinc supplementation, probiotics, and lactose-free feeding. However, the diarrhoea did not improve and feed tolerance remained poor. Repeat assessments revealed progressive electrolyte derangements, raising concern for gastrointestinal mucosal dysfunction due to hyaline deposition. Given this pathophysiological background, a tailored gut-directed nutritional strategy was implemented. This included a transition to semi-elemental formula to reduce digestive burden, small-volume frequent feeds to optimize absorption, strict

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osmotic reduction, and precise electrolyte replacement. Within 48–72 hours, the child demonstrated substantial clinical improvement, including decreased stool frequency, improved hydration, better feed tolerance, and normalization of urine output. He was subsequently discharged with a structured feeding plan and regular outpatient follow-up.

DISCUSSION

The gastrointestinal system is particularly vulnerable in HFS due to the accumulation of hyaline material within the mucosal layers. This disrupts nutrient absorption, weakens mucosal integrity, and impairs peristaltic movement. These structural changes explain the persistent and refractory nature of diarrhoea seen in affected infants, often rendering conventional paediatric diarrhoea protocols ineffective. In this case, standard management modalities showed minimal impact, emphasizing the unique nature of diarrhoea in HFS. The notable improvement following the introduction of semi-elemental feeds supports the hypothesis that reducing the digestive and antigenic load may substantially improve gastrointestinal handling. Additionally, meticulous fluid and electrolyte management proved essential, given the susceptibility of these patients to rapid deterioration. This case underscores the need for heightened clinical suspicion of HFS in infants presenting with early cutaneous lesions, joint stiffness, and unexplained diarrhoea. It also demonstrates that targeted nutritional strategies may significantly alter the trajectory of disease progression and reduce the risk of severe complications such as sepsis.

CONCLUSION

This case reinforces the importance of individualized nutritional strategies in managing gastrointestinal manifestations of Hyaline Fibromatosis Syndrome. Precision gut-directed therapy, involving semi-elemental feeding and electrolyte-optimized hydration, may offer a practical and effective intervention for infants with refractory diarrhoea. Early recognition and prompt management can mitigate complications and improve clinical outcomes in this vulnerable population.

ETHICAL APPROVAL AND PATIENT CONSENT

Written informed consent was obtained from the patient's parent/legal guardian for publication of this case report and accompanying clinical information/images. Patient identity has been adequately anonymized in accordance with ethical publishing standards and GlobalMeetx journal guidelines.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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