Protein-Losing Enteropathy Associated with SHOC2 Gene Mutation in Noonan-Like Syndrome

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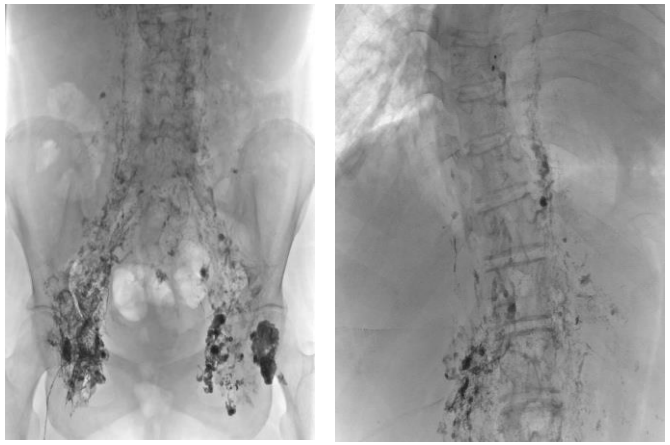
Introduction

- Protein-losing enteropathy (PLE) is the abnormal loss of protein through the gastrointestinal tract.
- The group of disorders including Noonan syndrome and Noonan-like syndromes (NS) consists of various dysmorphic and congenital abnormalities.
- Gene mutations paired with diverse phenotypic presentations have made identifying relationships between genotype and phenotype challenging.
- In rare cases, PLE has been reported with NS, but few genetic associations have been made.

Imaging



Radiograph confirming abnormal enhancement of lymphatics of the mesentery and periportal space noted on bilateral inguinal ultrasound guided transnodal lymphangiography which demonstrated abdominal collateral lymphatics and bilateral iliac lymphangiectasia.

Case Description

21-year-old female with history of NS presented to outpatient clinic with lower extremity edema and diarrhea with 4 watery bowel movements per day for 3 months.

Labs revealed albumin of 2.4 g/dL, creatinine of 0.35 mg/dL, and potassium of 3.1 mEq/L. Acute hepatitis panel and anti-tissue transglutaminase were negative. Stool studies were positive for *Clostridioides difficile* and she was treated with oral Vancomycin.

Esophagogastroduodenoscopy and colonoscopy were unremarkable.

CT Enterography suggested altered lymphatic drainage, and a lymphangiogram showed abdominal collateral lymphatics and iliac lymphangiectasia.

Overall, findings were consistent with PLE.

Serum albumin level normalized after initiation of high protein diet and medium-chain triglyceride supplementation.

Genetic analysis showed SHOC2 gene mutation.

Discussion

PLE is the loss of serum proteins through the digestive tract, which may initially present as hypoalbuminemia in the absence of liver disease.

It is important to recognize because low albumin is associated with increased mortality risk.

Lymphatic disease presenting with lymphedema, chylothorax, and lymphangiectasia, is well known in NS and may play a role in PLE development.

PLE has not been reported with SHOC2 mutation.

There is no standard treatment for PLE in NS. Supplementation with medium-chain triglycerides and periodic albumin infusions may be effective.

This case identifies SHOC2 mutation with PLE and encourages consideration of PLE in NS.

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