Pediatric Gorham–Stout Syndrome with Chylothorax, Ascites, and Osteolysis: A Case Report

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Abstract

Gorham–Stout syndrome (GSS) is a rare lymphatic vascular disorder characterized by progressive osteolysis, and pediatric cases with multicompartment involvement are exceedingly uncommon. We describe a 12-year-old boy who presented with progressive dyspnea, weight loss, and abdominal distension. Imaging revealed large bilateral pleural effusions, diffuse ascites, minimal pericardial effusion, and extensive osteolytic lesions involving the ribs, thoracic vertebrae, pelvis, and scapula. Pleural fluid analysis confirmed chylothorax with triglyceride levels exceeding 1,159 mg/dL. Lymphoscintigraphy demonstrated abnormal lymphatic flow with partial obstruction. Rib biopsy showed non-neoplastic lymphatic proliferation, consistent with GSS. The patient was managed with thoracic drainage, total parenteral nutrition, a peptide-based formula, and a medium-chain-triglyceride diet. Sirolimus therapy (0.8 mg/m² twice daily) resulted in a marked reduction in chylous drainage within 15 days, allowing removal of one chest tube and stabilization of osteolytic lesions on follow-up imaging. This case highlights one of the rare pediatric presentations of GSS with simultaneous chylothorax, ascites, and widespread bone involvement, and demonstrates that early mTOR inhibition may be effective in controlling lymphatic leakage and stabilizing disease progression.

Keywords: Gorham–Stout syndrome; Chylothorax; Pediatric; Lymphatic Anomaly; Osteolysis; Sirolimus.

Introduction

Gorham–Stout syndrome (GSS) is a rare lymphatic vascular anomaly, first described by Jackson in 1838, characterized by progressive bone resorption due to intraosseous proliferation of non-malignant lymphatic and/or blood vessels.[1,2] It is also known as "vanishing bone disease" or "massive osteolysis," with the eponym established after Gorham and Stout's 1955 histopathological report.[3]

Although most cases occur in young adults, pediatric presentations are reported.[4] The pathogenesis remains unclear; lymphangiogenic factors such as VEGF-A, VEGF-C, VEGF-D, bFGF, and PDGF may contribute. Clinical features vary by site and may include pathological fractures, chylothorax, pericardial effusion, and functional loss.[5] Thoracic involvement is critical because of its association with lymphatic leakage and potentially fatal chylothorax.[1]

Case Report

A 12-year-old boy had two months of progressive dyspnea, cough, wheezing, 10-kg weight loss, and abdominal distension. He had poor appetite for a year, no fever, and was under the 10th percentile for weight and height. Examination revealed tachypnea, bilateral decreased breath sounds (right > left), jugular venous distension, and ascites; neurological status was normal.

Chest radiography demonstrated bilateral pleural effusions, more prominent on the right side [Figure 1]. Laboratory findings were as follows: leukocyte count 5,700/µL (79.5% neutrophils, 13% lymphocytes), absolute lymphocyte count (ALC) 700/µL, hemoglobin 13.2 g/dL, platelet 360,000/µL, C-reactive protein 0.2 mg/L, erythrocyte sedimentation rate 3 mm/h, calcium 9.6 mg/dL, phosphorus 4.8 mg/dL, alkaline phosphatase 134

U/L, 25-hydroxy vitamin D 17.3 ng/mL, triglycerides 36 mg/dL, total protein 7.6 g/dL, total cholesterol 131 mg/dL, lactate dehydrogenase (LDH) 197 IU/L, and albumin 4.6 g/dL. Bone mineral density assessment revealed an L1–L4 vertebral Z-score of –2.2.

Thoracentesis was performed, and the aspirated fluid had a chylous appearance [Figure 1]. Pleural fluid analysis revealed a leukocyte count of 4,200/μL, ALC 2,400/μL, triglycerides >1,159 mg/dL, protein 5.2 g/dL, LDH 124 IU/L, and albumin 3.6 g/dL. No atypical cells were observed on cytologic examination, Gram staining was unremarkable, and cultures were sterile. The pleural fluid cholesterol-to-serum cholesterol ratio was below 1 (0.86). The effusion was interpreted as exudative. TORCH serologies and respiratory viral PCR assays were negative. Adenosine deaminase activity was 13.4 U/L, and the interferon-gamma release assay was normal. Serum immunoglobulin levels at presentation were within normal. Echocardiography showed minimal pericardial effusion. Chest radiography demonstrated bilateral pleural effusions, more prominent on the right side (Figure 1A). Thoracentesis revealed chylous pleural fluid (Figure 1B). Follow-up chest radiography on day 20 showed marked reduction in pleural effusion (Figure 1C). Thoracic and abdominal CT revealed extensive osteolytic lesions involving the ribs, thoracic vertebrae, pelvis, and scapula (Figure 1D–E). Lymphoscintigraphy demonstrated partial obstructive lymphatic flow. Based on clinical and imaging findings, GSS was diagnosed. Bone biopsy from an affected rib confirmed non-neoplastic vascular/lymphatic proliferation without malignancy. Treatment included total parenteral nutrition, peptide-based enteral feeding, and a fat-free medium-chain triglyceride diet. Sirolimus 0.8 mg/m² twice daily was initiated; therapeutic levels were achieved within one week. During sirolimus therapy, the patient was closely monitored for potential adverse effects. No clinically significant side effects, including cytopenia, mucositis, hyperlipidemia, hepatotoxicity, or infectious complications, were observed during treatment or follow-up. By day 15, chylous drainage had decreased markedly, allowing removal of the left chest tube; the right continued to drain smaller volumes. Parenteral nutrition was discontinued, and the oral fat-free diet was continued. Radiological follow-up over a 6-month period showed stabilization of the osteolytic lesions, with no further bone loss or signs of progression.

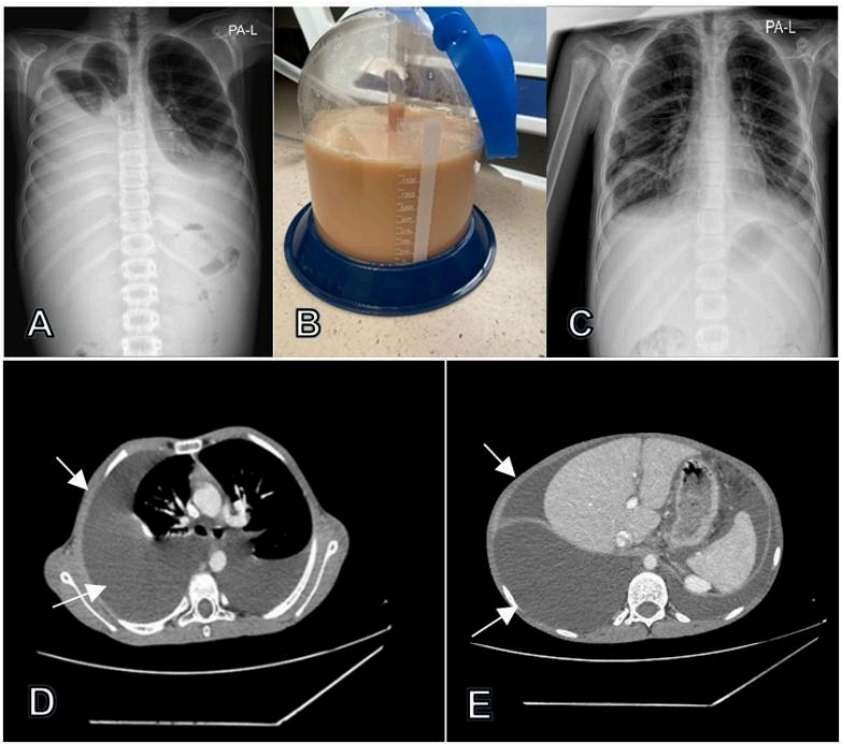

Figure 1. Chest radiograph (A: at presentation, C: day 20) and pleural fluid appearance (B: at presentation). Thoracic (D) and abdominal (E) CT scans obtained at presentation.

Discussion

GSS is an uncommon disorder, and pediatric cases are particularly rare. The hallmark of the disease is progressive osteolysis resulting from uncontrolled lymphatic or vascular proliferation within bone [2, 3]. Although the etiology is unclear, dysregulated lymphangiogenesis involving VEGF-A, VEGF-C, and VEGF-D has been

implicated. Thoracic involvement may manifest as chylothorax, which is associated with significant morbidity due to respiratory distress, electrolyte imbalance, and nutritional compromise.[1]

Multicompartment lymphatic leakage, such as the coexistence of chylothorax and ascites, has been described only in a limited number of pediatric reports.[4,5] This combination suggests widespread disruption of lymphatic integrity and may indicate more severe disease. In our case, lymphoscintigraphy demonstrated abnormal lymphatic flow and leakage, supporting the pathophysiological mechanism of extensive lymphatic involvement.

Management of GSS is challenging because no standardized treatment exists. Options include dietary modification, pleural drainage, thoracic duct ligation, radiotherapy, interferon-α, bisphosphonates, and recently, mTOR inhibitors. Sirolimus has emerged as a promising therapy because it inhibits lymphangiogenesis by suppressing the mTOR pathway. Several reports have noted reductions in chylous effusions and stabilization of bone lesions in patients with complex lymphatic anomalies treated with sirolimus.[1]

In our patient, the rapid decline in chylous drainage within two weeks was consistent with previous observations and highlights the therapeutic potential of sirolimus in severe pediatric GSS.[1] Stabilization of osteolytic lesions on follow-up imaging further supports its role in halting disease progression.

This case contributes to the limited pediatric literature by describing a rare combination of chylothorax, ascites, and widespread osteolysis, and demonstrates the beneficial effect of early sirolimus therapy.[1]

Conclusion

This case highlights an exceptionally rare pediatric presentation of Gorham–Stout syndrome with simultaneous chylothorax and ascites. Early diagnosis and prompt initiation of sirolimus resulted in rapid improvement in lymphatic leakage and stabilization of bone disease, supporting its therapeutic value in severe GSS.

Disclosure

The authors have no financial or non-financial conflicts of interest to declare. Written informed consent was obtained from the patient's parents for publication of the clinical data and accompanying images.

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