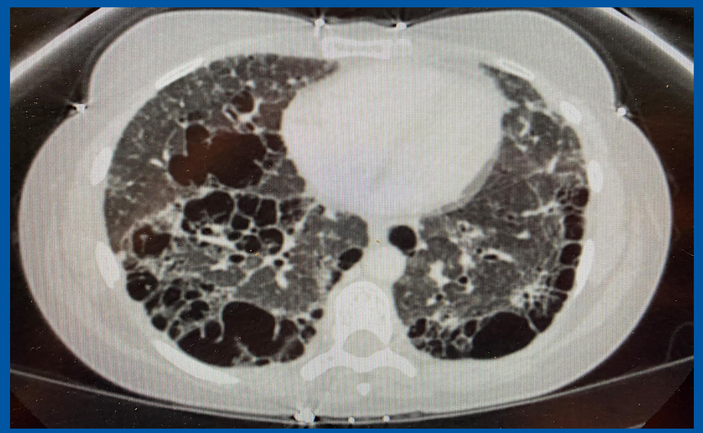
UF College of Medicine
Department of Surgery
UNIVERSITY of FLORIDA

The role and timing of anti-reflux procedures in patients with Hermansky-Pudlak Syndrome

Kelly M. Herremans, MD, J. Christian Brown, MD, Alexander L. Ayzengart, MD, MPH, FACS

Introduction

Hermansky-Pudlak Syndrome (HPS) is an extremely rare, hereditary disorder with a prevalence of 1 to 2 in 1,000,000 individuals worldwide. It is characterized by oculocutaneous albinism, bleeding diathesis, granulomatous colitis, gastroesophageal reflux disease (GERD) and frequently pulmonary fibrosis. Pulmonary fibrosis in HPS often manifests at 30-40 years of age and lung transplantation remains the only treatment to prolong survival. GERD may exacerbate pulmonary fibrosis and has the potential to lead to transplantation rejection.[1]



Objectives

1. To highlight the pathophysiology of Hermansky-Pudlak Syndrome, particularly in relation to pulmonary fibrosis and GERD.
2. To explore the role and optimal timing of anti-reflux procedures in two patients with HPS.

A Case Series

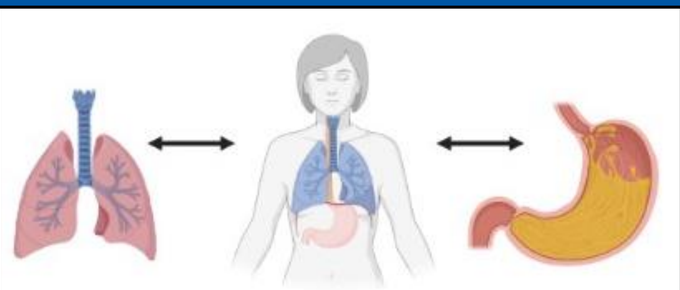
Case 1: A 48-year old female with a history of HPS and associated platelet dysfunction and pulmonary fibrosis presented with symptomatic GERD one-year following bilateral lung transplantation. Endoscopy showed evidence of hiatal hernia and LA Grade A reflux esophagitis and pH probe analysis revealed an elevated Demeester score (66.5). Fluoroscopic examination, manometry and gastric emptying study were unremarkable. Patient underwent robotic-assisted modified Toupet (partial, posterior) fundoplication and hiatal hernia repair. Her postoperative course was unremarkable and she continues to follow with lung transplantation with no evidence of rejection.

Case 2: A 49- year old female with a history of HPS, obesity (BMI 38), type 2 diabetes, and pulmonary fibrosis presented with symptomatic GERD six-months following bilateral lung transplantation. Gastric emptying study showed evidence of gastroparesis and pH probe analysis revealed an elevated Demeester score (50). CT chest was concerning for findings of aspiration. Fluoroscopic evaluation and manometry were unremarkable. In the setting of her associated comorbidities, patient was recommended to undergo laparoscopic Roux-en-Y gastric bypass. She is currently undergoing preoperative bariatric work-up and will be scheduled for surgery following the completion of this evaluation.

Conclusion

Patients with HPS pose a unique challenge in the surgical treatment of GERD. We propose that:

1. Patients with HPS undergo screening for GERD in order to delay or potentially prevent the progression of pulmonary fibrosis.
2. Anti-reflux operations should be considered before, possibly simultaneously with, or soon after lung transplantation in order to avoid rejection. Earlier intervention has the potential to improve outcomes in patients with HPS.



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References

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