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A rare cause of upper gastrointestinal bleed

"Downhill" esophageal varices is a rare condition, first reported in 1964 by Felson and Lessure.[1] These varices have been reported in association with obstruction of superior vena cava(SVC) or its tributaries, secondary to extrinsic compression or thrombosis. Very rarely the SVC obstruction has been reported in mediastinal fibrosis.[2,3] We present a case of acute upper gastrointestinal bleeding in an individual with fibrosing mediastinitis and past history ofintake of anti-tubercular therapy for pulmonary tuberculosis and history of active alcohol intake.

Case Report

A 42-year-old man presented withthe history of a single episode, single bout of hematemesis and melena. No history of postural symptoms such as dizziness, pre-syncope and syncope was reported. He denied any abdominal pain or previous episodes of gastrointestinal bleeding. He also denied painkiller intake. His medical history included past history of anti-tubercular therapyfor pulmonary tuberculosis 13 years back.

Physical examination of the patient revealed stable vitals. His abdominal examination was normal with no distension or tenderness. The remaining systemic examination was within normal limits. Initial laboratory tests revealed hemoglobin of 11 grams/dl with normal platelet count and coagulation profile. His renal and liver chemistries were within normal limits.

After adequate resuscitation, an esophago gastroduodenoscopy was performed. Endoscopy revealed grade III esophageal varices with red colour sign (RCS) **(Figure 1)** along the entire length of the esophagus. No active bleeding was noted from esophageal varices at the time of the procedure. However, altered blood was noted in the stomach. His duodenum was grossly normal and no other bleeding lesions were found either in the stomach or in the duodenum. In view of large variceswith history of recent bleeding, endoscopic band ligation (EBL)was performed and three EBL – bands were applied.

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In view of the history of alcohol abuse and finding of large esophageal varices, contrast enhanced Computed Tomography (CECT) of abdomen was performed to rule out possibility of chronic liver disease or chronic pancreatitis with splenic vein thrombosis. CECT Abdomen revealed normal study of liver, pancreas, portal and splenic veins but showed peri-esophageal collaterals. Subsequently, a CECT Chest was performed which revealed features suggestive of fibrosing mediastinitis with encasement of right brachiocephalic vein and superior vena cava with dilated azygous and hemiazygous veins with multiple collaterals in paravertebral and paraesophageal regions **(Figure 2)**. The retained diagnosis was downhill esophageal varices, secondary to compression of the superior vena cava by fibrosing mediastinitis.

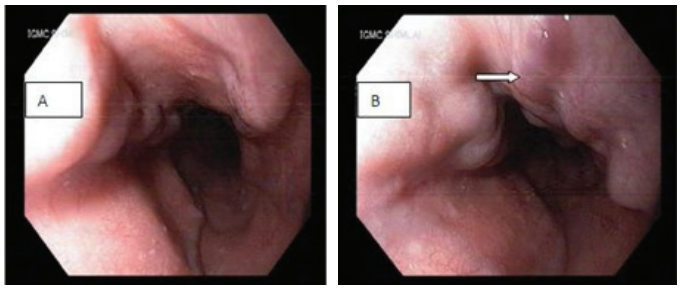

Figure 1: Esophagogastroduodenoscopy (A) - showing downhill varices in esophagus (B) - arrow showing red color sign (RCS) over varices.

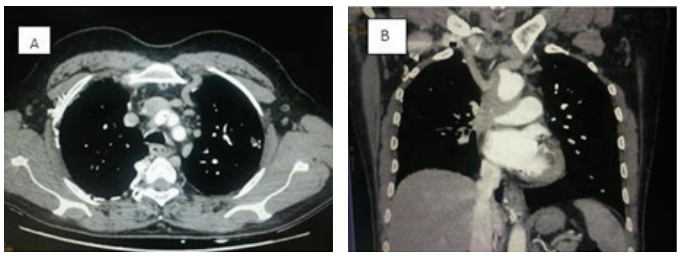

Figure 2: CECT - images (A - axial and B - coronal, mediastinal windows) showing fibrosing mediastinitis with encasement of superior vena cava.

Discussion

"Downhill" esophageal varices are dilated veins resulting from SVC obstruction whose blood flow is directed caudally towards the azygous vein[4] or the inferior vena cava (IVC). They are either located in the upper esophagus or may involve the entire esophagus depending on the level of obstruction above or below the azygous venous system, respectively.[5] If the lesion is proximal to the azygos vein, then drainage can occur through mediastinal collaterals to the patent azygos system below the level of obstruction. This downhill venous flow through proximal esophageal vessels results in formation of varices limited to the upper portion of the esophagus. If the obstruction is distal to the azygos vein, then the azygos system is unable to bypass the lesion. The resulting venous drainage via the esophageal plexus causes formation of varices along the entire length of the esophagus.

There is a long list of etiologies of downhill varices,[6] described in the literature including central venous catheterization, mediastinal fibrosis, primary and metastatic mediastinal tumors, mediastinal lymphadenopathy secondary to head and neck cancers, substernal goiters and thyroid masses, thyroid carcinoma, lung cancer, thymoma, systemic venulitis, Behcet disease, Castleman disease, and as a late complication after correction of congenital heart defects. Downhill varices may also develop without superior vena cava thrombosis, as seen in goiter, history of thyroid surgery, severe pulmonary hypertension, thyroid tumors, abnormal cricopharyngeal muscle constriction or any abnormal muscular constriction of the hypopharyngeal vein.

Clinical presentation of downhill esophageal varices is dominated by clinical symptoms of superior vena cava obstruction that were present in 91.4% of the cases described in the literature.[7]

Accidentally discovered downhill esophageal varices and upper gastrointestinal bleeding may be the first presentation.[8] Downhill varices represent only 0.1% of all esophageal variceal bleeding.[6] Lower risk of bleeding may be due to lack of coagulopathy andsubmucosal and higher location of varices in the esophagus, away from erosive gastroesophageal reflux.[9]

There are no definitive recommendations on how to screen and manage downhill varices. Treatment plan needs to be individualized. Primary treatment of downhill esophageal varices is directed toward the underlying etiology.[9]

Sclerotherapy was complicated by spinal cord infarction in some cases, caused by flow of sclerosant

from the azygos to spinal veins when injected at the level of the middle and upper esophagus.[6] Variceal band ligation is effective for controlling bleeding but site of banding is not clearly defined. The risk of bleeding or perforation seems higher because of the weakness of the proximal esophageal posterior wall and overall lack of serosa. The use of a Sengstaken-Blakemore tube can be lifesaving in case of uncontrolled bleeding.[6]

This case represents a rare but physiologically plausible cause of acute upper gastrointestinal bleeding. Though standard recommendations are currently unavailable to help guide physicians to manage acute bleeding associated with "downhill" varices, we suggest that awareness, prompt diagnosis, and management on a case by case basis using available endoscopic, radiological, and surgical interventions can be successful.

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Progressive familial intrahepatic cholestasis: clinico-molecular correlation in Indian children

Progressive familial intrahepatic cholestasis (PFIC)is an autosomal recessive disorder characterized by early onset cholestasis, pruritus, hepatomegaly and growth failure and can progress to liver failure before adolescence due to secondary cirrhosis.[1] PFIC accounts for 10-15% of cases of cholestasis in children and is the cause of 10-15% of pediatric liver transplants. PFIC I and II represent about two-third of the cases of PFIC, the remaining being PFIC III.[2] Although PFIC has been reported worldwide, only 11 cases have been described from India.[3-5] Here we present a case series of five PFIC patients who enrolled at our liver clinic and were diagnosed to be PFIC by molecular studies, and discuss their clinical phenotypes.