A 77-year-old woman with osteogenesis imperfecta was admitted with symptomatic choledocholithiasis. Endoscopic retrograde cholangiopancreatography (ERCP) was carried out and mild resistance was encountered in the distal esophagus with “red out” during intubation. The duodenoscope was withdrawn and gastroscopy revealed a tear in the distal esophagus (Fig. 1). As a perforation was suspected, a nasogastric tube was deployed, intravenous antibiotics were started, and surgery was consulted. An esophagogram confirmed the esophageal leak (Fig. 2) and thoraco-abdominal computed tomography (CT) showed a 0.7-cm esophageal perforation and air–fluid collection (3 cm × 10 cm) in the posterior mediastinum (Fig. 3). Conservative management was decided on due to clinical stability and poor operative candidacy. Within 48 hours the liver function tests normalized and parenteral nutrition was started. The day 7 esophagogram showed partial healing. Enteral feeding was started with a nasojejunal tube placed fluoroscopically. An esophagogram on day 27 showed complete healing (Fig. 4). The patient started an oral diet and was discharged with plan for elective cholecystectomy.

Osteogenesis imperfecta is an autosomal dominant disorder of collagen synthesis (1 in 10000 births). Gastrointestinal manifestations in osteogenesis imperfecta are rare although spontaneous colon perforation has been reported, where histopathological examination showed absence of muscle layers in the perforated segment [1]. Murine models of osteogenesis imperfecta showed thinner esophageal submucosa and muscularis compared with wild mice [2]. ERCP-related perforations are infrequent (0.4 %) and are commonly related to improper guide-wire insertion or sphincterotomy [3]. Esophageal perforation in ERCP is rare and is attributed to use of sharp accessories [3, 4]. This is the first reported case of esophageal perforation in osteogenesis imperfecta. We believe that perforation in our patient occurred due to abnormal esophageal submucosa and muscularis and limited view with duodenoscope. Esophageal perforations are traditionally managed surgically; however, good outcomes are reported with conservative management in smaller perforations and contained leaks [5]. The present case highlights the propensity for esophageal perforation in osteogenesis imperfecta. We recommend caution and low threshold to suspect perforation during gastrointestinal endoscopy in osteogenesis imperfecta.

Fig. 1 Gastroscopy showing linear mucosal tear in the distal esophagus (black arrow) following endoscopic retrograde cholangiopancreatography (ERCP) in a 77-year-old woman with osteogenesis imperfecta and symptomatic choledocholithiasis.

Fig. 2 Esophagogram with water-soluble contrast demonstrating the distal esophageal leak (black arrow).
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Corresponding author
Rajan Kochar
Division of Gastroenterology and Hepatology
The University of Texas Medical Branch
301 University Boulevard RT 0764
Galveston
Texas 77555
USA
Fax: 409-772-4789
rakochar@utmb.edu

Fig. 3 Thoraco-abdominal computed tomography (CT) scan showing paraesophageal pooling of the oral contrast (arrows on transverse section, a) tracking down towards the retroperitoneum (arrow on coronal section, b).

Fig. 4 Esophagogram with water-soluble contrast on day 27 showing complete healing.