CASE REPORT

Unusual presentation of adult Marfan syndrome as a complex diaphragmatic hiatus hernia

Shruti Thakur a,*, Anupam Jhobta a, Brij Sharma b, Arun Chauhan c, Charu S. Thakur a

a Department of Diagnostic Radiology, Indira Gandhi Medical College and Hospital, Shimla, Himachal Pradesh, India
b Department of Gastroenterology, Indira Gandhi Medical College and Hospital, Shimla, Himachal Pradesh, India
c Department of General Surgery, Indira Gandhi Medical College and Hospital, Shimla, Himachal Pradesh, India

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Summary Marfan syndrome is multisystem connective tissue disorder that primarily involves the skeletal, cardiovascular, and ocular systems. The gastrointestinal complications in Marfan syndrome are rare, with only a few case reports described in the literature. We present a 25-year-old woman who presented with acute abdominal pain for 1 day. The imaging features revealed complex diaphragmatic hiatus hernia with organoaxial gastric volvulus. This is a unique case report about an adult patient with Marfan syndrome who presented with symptomatic paraesophageal hernia and organoaxial gastric volvulus.

1. Introduction

Marfan syndrome is a connective tissue disorder caused by mutation in the fibrillin-1 gene located on chromosome 15. It is an autosomal dominant disorder with a high penetrance rate. Although the syndrome is not uncommon, with a prevalence of 2–3 individuals per 10,000, gastrointestinal involvement is rare, with only a few case reports.

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2. Case Report

A 25-year-old woman presented in the emergency department with severe abdominal pain for 1 day. On chest X-ray, multiple air–fluid levels were seen on both sides of the lower chest. The Ryle’s tube could not be passed, so upper gastrointestinal endoscopy was planned. On endoscopy (Olympus; 180 series), abnormal alignment and malposition of the stomach was found. The endoscope could not be navigated further into the antpyloric region (Fig. 1A and B). Although there was no history of severe abdominal trauma, the intrathoracic herniation of gut loops through the ruptured diaphragm remained a possibility. Contrast-enhanced computed tomography was performed in the portal venous phase with 100 mL nonionic iodinated contrast agent on 64-slice GE light speed Xte (It is the name of CT machine. The company is GE and it is a 64 slice machine) for diagnostic confirmation. The imaging showed the stomach in the right thoracic cavity with organoaxial rotation and herniation of part of the transverse colon, and splenic flexure in the left hemithorax (Fig. 2A). These herniated bowel loops showed normal wall enhancement with no ischemic changes. Of significant interest was the intrathoracic herniation of omental fat and vessels in between anteriorly pushed inferior vena cava (IVC) and posteriorly placed aorta (Fig. 2B), indicating that the port of herniation was the lax esophageal hiatus and not the ruptured diaphragm. This type of diaphragmatic hiatus hernia is considered to be type IV. The patient underwent emergency surgery. Explorative laparotomy was done. The operative findings were an apparent empty abdominal cavity with herniation of the stomach, part of the ascending colon and transverse colon, splenic flexure and omentum on both sides of the thoracic cavity, through lax and dilated esophageal hiatus. The hernial contents were returned into the abdominal cavity after assessing the viability. The hiatus was approximated and fundoplication was done with interrupted nonabsorbable sutures. The postoperative recovery was uneventful.

The coincidental findings upon imaging in this case were lumbosacral dural ectasia along with lateral meningoceles (Fig. 2C) and dilatation of the aortic root at the level of the sinus of valsalva (Fig. 2D). The proximal pulmonary arterial trunk was normal in caliber. There was thoracolumbar scoliosis of 30°. The chest wall was normal with no pectus excavatum or carinatum. No bullae or pneumothorax was seen. The patient had striae atrophicae in the bilateral lumbar regions without prior pregnancy. She was tall and thin and had no visual complaints. The family history and her past history were unremarkable. The unusually lax esophageal hiatus that led to massive herniation, along with other imaging findings and marfanoid body habitus of the patient, resulted in clinical diagnosis of Marfan syndrome, in accordance with Ghent1 nosology, which is an internationally accepted diagnostic criterion for this syndrome. Ghent1 nosology involves major and minor criteria for the diagnosis of Marfan syndrome and includes seven fields: skeletal, cardiovascular, ocular, pulmonary, skin and integument, dura matter, and genetic analysis. The gastrointestinal involvement has not been mentioned because of its rare association. The patient was started on β-blockers and was maintained under close follow-up for early detection of possible complications.

3. Discussion

Marfan syndrome is a multisystem connective tissue disorder with distinct physical characteristics. It shows an autosomal dominant pattern of inheritance and 25–30% of cases show sporadic mutation. The disorder is caused by mutation in the fibrillin-1 gene located on chromosome 15. This gene encodes fibrillin-1, a glycoprotein that is the main constituent of the microfibrils of the extracellular matrix. Reduced or abnormal fibrillin-1 leads to tissue weakness. The disorder shows clinical variability with predominant involvement of skeletal, cardiovascular and ocular systems. A variety of skeletal manifestations is seen owing to bone overgrowth and laxity of joints. The cardiovascular manifestations include aortic root dilatation, aortic dissection, mitral valve prolapse, and enlarged proximal pulmonary artery. The cardiovascular involvement is the major source

![Figure 1](image-url) (A) Gastroesophageal junction (small white arrow) and fundus (large white arrow). (B) Pylorus (white arrow) beyond which the endoscope was not navigable.
of morbidity and mortality in these patients. Myopia is the most common ocular feature. The ocular hallmark is ectopia lentis (lens subluxation) that is usually bilateral. The other systems that are involved are the pulmonary (bullae and spontaneous pneumothorax) and integumentary (striae atrophicae and recurrent/incisional hernias) systems.1 Dural ectasia is also an important feature.

Genetic testing may also be done. However, closely related mutations are also seen in phenotypes similar to Marfan syndrome which make this syndrome a diagnostic challenge. The closest differential is Loeys–Dietz syndrome, which shows overlapping features and may be considered as Marfan type 2.4 It is also an autosomal dominant condition with heterozygous mutations in the genes encoding transforming growth factor beta receptors (TGFBR) 1 and 2. It affects the aortic arch and mitral valve. The affected patient shows bifid uvula, cleft palate, and hypertelorism. Aortic aneurysm and dissection are seen at an earlier age.7 Other syndromes that show mutations in fibrillin genes or TGFBR1 or TGFBR2, along with absence of full phenotypic features of Marfan syndrome or Loeys–Dietz syndrome are congenital contractual arachnodactyly, ectopia lentis syndrome, and familial mitral valve prolapse syndrome (MASS phenotype: mitral, aortic, skeletal, skin). Given the variable and wide clinical spectrum of the disease, diagnosis of Marfan syndrome depends on the assessment of major and minor criteria given in Ghent nosology.4

The gastrointestinal involvement is rare and is described as paraesophageal hiatus hernias mainly in neonates and the pediatric age group1,9 and gastroesophageal reflux disease (GERD), small hiatus hernia, diverticulitis coli, and Zenker’s diverticula in adults as case reports only.2,9 The most common type of hernia in Marfan syndrome is inguinal.9 However, there is a predisposition to paraesophageal hernias also.2,10 This is due to weak and defective collagen formation seen in this syndrome that leads to abnormally lax esophageal hiatus and lax gastric ligamentous attachments. The non-fixation of the stomach...
and other abdominal organs leads to intrathoracic herniation. Four types of hiatus hernias are described in the surgical literature: Type I is sliding hiatal hernia; Type II is paraesophageal hiatal hernia; Type III is combined sliding and paraesophageal hiatal hernias with features of both Type I and Type II hiatal hernias; and Type IV is complex paraesophageal hiatal hernia characterized by intrathoracic herniation of other abdominal organs such as the small bowel, large bowel, omentum, spleen, and liver. It may have a sliding component. This type of hernia may be associated with massive herniations with unusual complications.

The unique presentation of Marfan syndrome led to its diagnosis in the present case. Appropriate therapy was started to prevent any adverse outcome. The life span in these patients is generally reduced, mainly due to progressive dilatation and dissection of the aorta. The medical treatment is β-blockers, which decreases the systolic aortic pulse pressure, thereby delaying aortic dilatation. These patients are also advised not to indulge in strenuous exercise. A multidisciplinary approach that requires a cardiothoracic surgeon, ophthalmologist, and orthopedician is needed for comprehensive care of such patients.

In conclusion, diaphragmatic hernia as a presentation of Marfan syndrome is unusual. Clinicians should rule out the possibility of Marfan syndrome in these patients as prompt diagnosis is life saving in those with asymptomatic cardiovascular disease.

References