Case Report

Neglected right diaphragmatic hernia with transthoracic herniation of gallbladder and malrotated left liver lobe in an adult✩✩

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ABSTRACT

Diaphragmatic hernias are commonly encountered in radiological practice, especially in pediatric patients. Delayed presentation of diaphragmatic hernias can occur in children and less frequently in adult patients. We present an extremely rare case of a 15-year-old male patient with recurrent attacks of cholestatic hepatitis who has been misdiagnosed to have primary sclerosing cholangitis and was planned for liver transplantation. On imaging, the patient was found to have neglected right diaphragmatic hernia causing hypogenesis of the right hemithorax and right lower lung lobe. A transthoracic herniation of gallbladder and malrotated left liver lobe is also diagnosed on imaging. Biliary dilatation with transition point at the diaphragmatic hernia neck was identified. Major hepatic anomaly such as liver malformation is an exceedingly rare case with only 3 reported cases in the literature. In our case, the patient suffered from recurrent cholestatic hepatitis and unfortunate misdiagnosis with primary sclerosing cholangitis, which have delayed his appropriate management. In addition, the patient was found to have interrupted IVC with azygos and hemiazygos continuation without evidence of heterotaxy syndrome.

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Case report

A 15-year-old male with recurrent attacks of cholestatic hepatitis, who has been labeled as having primary sclerosing cholangitis at his local hospital, referred to our hospital for liver transplantation. The patient has reported normal growth and development. The patient has not have had significant breathing problems or chronic illnesses, other than the presumed diagnosis of primary sclerosing cholangitis. The physical examination was unremarkable. Blood tests and blood biochemistry were normal. The liver function test showed elevated Alanine transaminase (ALT) (145 U/L), Aspartate transaminase (AST) (71.6 U/L), Alkaline phosphatase (ALP) (464 U/L), and Gamma-glutamyltransferase (GGT) (264 U/L). Bile acid levels was elevated (95 μmol/L) with normal total bilirubin levels. Alpha fetoprotein, cancer embryo antigen, and car-

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bohydride antigen 19-9 concentrations were all within normal ranges.

Contrast-enhanced computed tomography and magnetic resonance studies was obtained and revealed central right diaphragmatic defect through which part of the liver has herniated into the right hemithorax (Fig. 1). The configuration of the herniated left hepatic lobe is abnormal, with the fissure of the ligamentum teres located posterolaterally in the right lower chest (Fig. 2) and gallbladder resting against the descending thoracic aorta and esophagus (Fig. 3). These indicate a 270° anticlockwise left hepatic lobe malrotation. There was moderate left hepatic biliary dilatation with the transition point at the neck of the hernia (Figs. 1 and 3). There were no biliary strictures to suggest the presumed diagnosis of sclerosing cholangitis. There was mild abnormal reticular high signal intensity on T2-WIs involving the periphery of the hepatic parenchyma in segment 4, indicating element of fibrosis (Fig. 4). There were no signs of cirrhosis. An interesting finding of complete absence of infrahepatic Inferior vena cava (IVC) with azygos and hemiazygos continuation is also present (Fig. 1A). The hepatic and portal veins are patent. The left atrium, stomach, and spleen are located in the left side. The right hemithorax and right lower lung lobe are significantly smaller than the left side. The lung fissures and

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Fig. 1 – (A,B) Axial and sagittal reformatted image of contrast-enhanced CT study of abdomen showing central interruption of the right hemidiaphragm (white arrowhead) with diaphragmatic defect (asterisk) through which part of the liver has herniated. The left portal vein is patent and seen above the level of the right diaphragm (white arrow). In addition, moderate biliary dilatation seen predominantly in the herniated left liver lobe. Note the incidental finding of interrupted IVC with azygos and hemiazygos continuation in A (black arrow).

Fig. 2 – Coronal T1-WI postgadolinium-based contrast agent administration again showing the herniated left liver lobe and the abnormal location of the fissure of the ligamentum teres which is seen posteriorly facing the lower right chest (white arrow).

Fig. 3 – Axial T2-WI of the upper abdomen showing the abnormal location of the gallbladder (white arrowheads) resting against the esophagus and descending thoracic aorta (asterisk). Biliary dilatation is also seen in the image, and predominantly in the herniated part of the liver (white arrow). Note the significantly smaller right hemithorax.
anatomy were normal. There was no aberrant pulmonary venous drainage. The heart is normal.

Percutaneous biopsy was performed and revealed changes of chronic cholangiopathy with ductular proliferation and early bridging fibrosis. There was no cirrhosis and no significant plasma cell infiltrates in the specimen.

The patient has continued with follow-up with adult hepatology transplant team and has not had surgery at this time.

Discussion

Diaphragmatic hernias are commonly encountered radiological diagnoses, more often diagnosed in pediatric and trauma patients. Presentation of diaphragmatic hernia in adult patients is relatively uncommon and usually asymptomatic. Diaphragmatic hernia is divided into congenital and acquired, based on their time of development. Congenital diaphragmatic hernias are further divided into Bochdalek and Morgagni hernias. Bochdalek hernia is more common and caused by defective formation or fusion of the pleuroperitoneal membranes, more commonly in the left side, with the rest of the embryological components of the diaphragm. Morgagni hernia is caused by failure of closure of the pars sternalis with the seventh costochondral arch. Almost any nonretroperitoneal structure can herniate through diaphragmatic defects [1]. The diagnosis of congenital diaphragmatic hernias in adults is uncommon with 0.17% incidence [2]. The diaphragmatic hernia in our case is of an unknown type. The patient was not previously known to have diaphragmatic hernia and denied prior major trauma. However, the patient’s right diaphragmatic hernia is consistent with an old neglected hernia due to the smaller size of the right lower lung lobe and hemithorax.

During third to fourth weeks of gestation, an outpouching starts to develop from the ventral surface of the caudal end of the foregut. This pouch, known as hepatic diverticulum, penetrates the septum transversum of the diaphragm. The liver bud enlarges rapidly and then differentiates into two buds, which will give rise to right and left liver lobes [3]. Minor variations in the liver lobulation are not uncommon, but congenital anomalies are rare [4]. Malrotation of the liver is an exceedingly rare entity. To the best of our knowledge, there are only 3 reported cases of liver malrotation in the literature, all of which were incidental findings without relevant symptomatology [5–7]. Raymond in 1956 described the first case of liver malrotation in an adult patient with congenital diaphragmatic hernia [5]. Then in 2013 and 2014, 2 more cases with liver malrotation has been described, however, both of them were in situ and not associated with congenital diaphragmatic hernia [6,7]. All of the 3 reported cases in the literature has come to the medical attention for reasons irrelevant to the liver malrotation. Our case is unusual in that the patient presented with long standing symptomatology which was mistakenly attributed to primary sclerosing cholangitis. The patient in our case has suffered from chronic intermittent cholestatic hepatitis which is likely caused by biliary obstruction due to a combination of biliary compression at the neck of the diaphragmatic hernia and left liver lobe malrotation.

An interesting vascular finding in our case was azygos continuation of interrupted IVC. IVC anomalies occur in <1% of patients. The reported prevalence of interrupted IVC with azygos and hemiazygos continuation is 0.6% and commonly discovered incidentally [8]. Majority of cases with azygos and hemiazygos continuation of IVC is associated with heterotaxy syndrome [9]. There is no known association with congenital diaphragmatic hernias.

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