Case Report

A Rare Case of Suprahepatic Gall Bladder with Phocomelia and Pancytopenia: Detected by Tc-99m Mebrofenin Scintigraphy

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Abstract

The possibility of an ectopic gallbladder should always be considered whenever there is a failure to localize it in its normal anatomical position on routine imaging. Although a very rare entity, the anomalous position of gallbladder can result in misinterpretation of imaging findings and create clinical confusion. Awareness of such an anomaly facilitates proper diagnosis and subsequent management. The authors report a very rare case of suprahepatic gallbladder associated with phocomelia, pancytopenia, and splenomegaly in a young 25-year-old female. The suprahepatic gallbladder was initially visualized on Technetium-99m (Tc-99m) Mebrofenin radionuclide hepatobiliary scintigraphy. Subsequent magnetic resonance cholecystopancreatography (MRCP) was also done to confirm the diagnosis.

Keywords: Gallbladder, magnetic resonance cholecystopancreatography, Mebrofenin, suprahepatic

Introduction

Various anomalies of gallbladder in terms of shape, size, and location have been described in surgical literature.[1] Anomalies of location include intrahepatic gallbladder, retrohepatic gallbladder, transverse gallbladder, left-sided gallbladder, and floating gallbladder.[2] Suprahepatic gallbladder is the rarest form of the locational anomaly of the gallbladder, and till date, very few cases of such rare anomaly have been reported.[3] In case the gallbladder is not visualized in its normal anatomical location, the possibility of its ectopic location should be considered because the failure to do so will result in misinterpretation of imaging findings and subsequent mismanagement. Patients with ectopic gallbladder may either remain asymptomatic or may present with the complications arising out of its ectopic location like acute cholecystitis, torsion, gangrene, or herniation.[4] Sometimes the typical symptoms and signs of acute cholecystitis may be absent if the gallbladder is situated away from the peritoneum. Ultrasonography, computed axial tomography (CAT), and radionuclide imaging are the common modalities that pick up an ectopically placed gallbladder. Radionuclide imaging using iminodiacetic acid derivatives like Technetium-99m (Tc-99m) Mebrofenin, in addition to localizing an ectopic gallbladder, provides information about its functional parameters. Magnetic resonance cholecystopancreatography (MRCP) confirms the diagnosis as well as characterizes the anomaly; however, the demonstration of an ectopic location on hepatobiliary scintigraphy rarely requires further confirmation by other imaging modalities.[2]

Case Report

A 25-year-old female presented to the Department of Nuclear Medicine for hepatobiliary scintigraphy with complaints of recurrent epigastric pain, abdominal distention, and recurrent episodes of jaundice in the past and with the suspicion of a functional gallbladder pathology. The patient gave history of bleeding gums,
Rather et al.: Suprahepatic gall bladder skin rashes, menorrhagia, and epistaxis of 1 year duration. She was being treated as a case of pancytopenia with hypersplenism. The patient was born out of a non-consanguineous marriage, normal delivery, and was second in birth order. Developmental history did not reveal any delay in achieving milestones except for an absent right hand below elbow since birth. There was no family history suggestive of any congenital disorders like absent limbs, bleeding disorders, etc. On physical examination, there was no jaundice, pallor, loss of hair, or any muscle wasting. There were multiple purple spots which did not fade on pressure on both upper limbs. The right hand was absent below elbow. Abdominal examination revealed splenomegaly with the spleen palpable to 6 cm below the costal margin. Laboratory investigations showed decreased hemoglobin (7.3 gm/dl), Total Leukocyte Count (3.01 × 10³/uL), platelets (23 × 10³/uL), and mean cell volume (67). Kidney function tests were normal. Liver function tests showed elevated serum bilirubin (1.50 mg/dl), alanine aminotransferase (123 IU/l), and normal alkaline phosphatase (300 U/l). Serum amylase was normal. Hepatitis profile (Hepatitis B surface Antigen, Hepatitis B core Antigen, HepatitisC virus) was negative. Upper gastrointestinal endoscopy showed two small chronic pyloric ulcers. An USG abdomen done 2 years back had revealed contracted gallbladder with splenomegaly and left ovarian cyst; a repeat abdominal USG done 2 months prior to the hepatobiliary scan could not locate the gallbladder in its normal anatomical location.

A 90-min dynamic Tc-99m Methrofenin hepatobiliary study was performed using an intravenous dose of 7 mCi (259 MBq) Tc-99m Methrofenin. The patient was placed in a supine position under a large field-of-view dual-head gamma camera using low-energy all-purpose parallel-hole collimator and 20% energy window centered at 140 keV. Ninety dynamic frames of 1 min duration each for the dynamic study (matrix size 128 × 128) followed by two static frames of 3 min duration each at 90 min and 3 h (matrix size 256 × 256) were acquired using a single detector (anterior).

The scan showed an intense focus of tracer uptake in relation to the upper border of the right lobe of the liver [Figure 1a], which became visible within 15 min of Tc-99m Methrofenin injection. At 15 min post injection, the patient was given standard fatty meal (half and half milk 8 oz/70 kg) orally. The intensity of the focal uptake decreased gradually, and by the completion of 90 min, uptake in the suprahepatic region had markedly decreased. The liver showed uniform tracer uptake with small-sized right lobe. Liver to bowel transit time was normal (45 min). Gallbladder emptying (ejection fraction) was also calculated. Emptying was 90% at 90 min and 100% at 3 h. A diagnosis of an ectopic (suprahepatic) gallbladder with normal ejection fraction, and right hepatic lobe hypoplasia was made. A Tc99-m sulfur colloid SPECT transverse section showed gallbladder in suprahepatic location [Figure 1b]. A subsequent MRCP [Figures 2a and b] confirmed the scintigraphic diagnosis of suprahepatic gallbladder. On MRCP, the

![Figure 1a: Tc-99m Methrofenin hepatobiliary radionuclide scan showing gallbladder located in the suprahepatic region](image-url)
axial HASTE [Figure 2a] and coronal TRUFI [Figure 2b] images demonstrated a suprahepatic gallbladder located superior to segment VIII, hypoplasia of the right hepatic lobe with compensatory hypertrophy of the left hepatic lobe, and gross splenomegaly. A 3.5 M Hz routine abdominal USG was done which revealed an ectopic gallbladder (right subphrenic) with small right hepatic lobe and hypertrophied left hepatic lobe [Figure 3a], which was previously reported as normal [Figure 3b].

Discussion

Three types of gallbladder congenital anomalies have been identified: Anomalies of form, anomalies of location, and congenital absence. Anomalies of location or ectopic location of gallbladder is very rare, its incidence being 0.1-0.7%. [4] Among the various ectopic locations, suprahepatic region is the rarest site with only a few reports appearing in either surgical or radiological literature. [5-10] Failure to identify an ectopic gallbladder can result in misinterpretation of image finding. In the present study, USG was unable to localize the gallbladder in its normal anatomical position; however, the same was diagnosed on Tc-99m hepatobiliary scan. Awareness of the possibility of such anomaly facilitates proper diagnosis and helps the treating physician to plan a proper strategy for its future management. Failure to identify the ectopic gallbladder can cause confusion resulting in dangerous complications like torsion and subsequent gangrene, herniation through foramen of Winslow into the lesser sac, or acute cholecystitis with or without peritoneal signs. [4]

A suspected focal defect on a liver image with Tc-99m labeled hepatobiliary agents in locations other than the normal anatomical location of the gallbladder may be due to ectopic gallbladder and needs to be differentiated from choledochal cysts, biliary diverticulum, or Caroli’s disease, all of which show focal hepatic accumulation of bile as well as of hepatobiliary pharmaceuticals. [2] The demonstration of gallbladder contraction and emptying in response to intravenous cholecystokinin (CCK) or standard fatty meal can be diagnostic of ectopic gallbladder and excludes other bile containing lesions. Computed axial tomography (CAT) scan and MRCP may be used to confirm the ectopic location. However, the demonstration of an ectopic location on hepatobiliary scintigraphy rarely requires further confirmation by other imaging modalities. [2]

In 1965, Regen et al. reported a case of suprahepatic gallbladder, which was followed by a report in 1968 by Dever et al. where the ectopic gallbladder had undergone torsion and subsequent gangrene. [5,6] In 1980, Faintuch et al. reported three cases of such ectopia seen in a 10-year
Rather, et al.: Suprahepatic gall bladder period, all of which had associated hypoplasia of the right lobe of the liver and upward displacement of the hepatic flexure of the colon, which overlapped the liver border.[7] All patients complained of recurrent pain in the right upper quadrant of the abdomen, suggestive of biliary disease, but only one case had calculi in the gallbladder that was acute cholecystitis. Two patients underwent cholecystectomy, and operative findings confirmed the preoperative diagnosis. It was speculated that the primary defect in this modality of suprahepatic gallbladder might be hypoplasia or atrophy of the right lobe of the liver of a congenital nature, with subsequent vicious orientation of the gallbladder and upward displacement of the colon. Similar cases of suprahepatic gallbladder with hypoplasia of the right lobe of the liver were reported in males by Youngworth in 1983[8] and Hopper in 1994.[9] In 2003, Kabourdis reported a case of suprahepatic gallbladder in a 65-year-old male who had right colonic cancer.[9] The last case report in this series was by Vishal Aggarwal in 2011 of a 70-year-old male patient who had history of chronic liver disease and suspected hepatocellular carcinoma and was being evaluated for liver transplantation.[10] The patient was subjected to fluorine-18 fluorodeoxyglucose (F-18 FDG) positron emission tomography-computed tomography (PET-CT) which revealed a suprahepatic gallbladder that was confirmed on magnetic resonance imaging (MRI).

In the review of literature, it was found that majority of the patients were above 40 years of age with underlying hepatic abnormalities, hypoplasia of the right hepatic lobe being the commonest. Our case report is the first of its kind where the patient was a young 25-year-old female who, in addition to a suprahepatic gallbladder and right hepatic lobe hypoplasia, had splenomegaly, thrombocytopenia, and congenital absence of right hand. The presence of phocomelia with ectopic gallbladder and pancytopenia may suggest some sort of common developmental abnormalities. Though phocomelia is known to have associated malformations like those of biliary tract and gastrointestinal tract, no such evidence exists for congenital ectopic gallbladder being associated with other congenital malformations. This case suggests that patients with ectopic gallbladder pathology may harbor a genetic abnormality predisposing them to other associated congenital malformations. In future, such patients may need a relevant genetic profiling for proper management and advice.

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How to cite this article: Ah Rather T, Khan SH, Singh M, Choh NA. A Rare Case of Suprahepatic Gall Bladder with Phocomelia and Pancytopenia: Detected by Tc-99m Mebrofenin Scintigraphy. World J Nucl Med 2013;12:41-4.

Source of Support: Nil. Conflict of Interest: None declared.