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CASE REPORT

29. Aggressive Gallbladder Carcinoma in Down Syndrome Patient: A Rare Presentation

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https://www.youtube.com/watch?v=hJIcIJ1w8oM&list=PLhqNq3xJClbafO0Y5bvBcgMmXpgzJxd44&index=5&t=4485s

Background: Gallbladder carcinoma in individuals with Down syndrome (DS) is exceedingly rare, literature reports only a handful of cases. Risk factors include cholelithiasis, chronic inflammation, biliary helicobacter pylori colonization, chromosomal instability and cytogenetic abnormalities of chromosome 21.

The Case : A 45-year-old male with a history of Down syndrome, presented to the emergency department complaining of moderate epigastric pain for 3 days, 5 days of yellowish skin discoloration, and 2-weeks of postprandial nausea and vomiting of undigested food. There was no history of dysphagia, anorexia, bowel habit changes, or gastrointestinal bleeding. Family and drug history is unremarkable. On examination, the patient had a distended abdomen, jaundice, mild epigastric tenderness, and a palpable bulge in the right upper quadrant.

Laboratory findings over repeated evaluations revealed persistently elevated inflammatory markers, direct hyperbilirubinemia, hypocalcemia, mild anemia with decreased hematocrit level, and hypoalbuminemia. Liver and pancreatic enzymes were unremarkable. On ultrasound, moderate-to-severe ascites were noted. MRI abdomen showed a large exophytic hepatic mass (9 × 7.5 × 7.3 cm) in segment VI encasing and compressing the gallbladder, with multiple smaller hepatic lesions (0.2–2.3 cm) and enlarged multilobular speculated periportal/mesenteric lymph nodes measuring up to 4.3 cm in diameter, 3.3 cm right adrenal lesion and asymmetric thickening of the ascending colon. Findings were suspicious for metastatic malignancy with gallbladder involvement. Colonoscopy was performed and revealed grossly normal mucosa; histopathology was unremarkable.

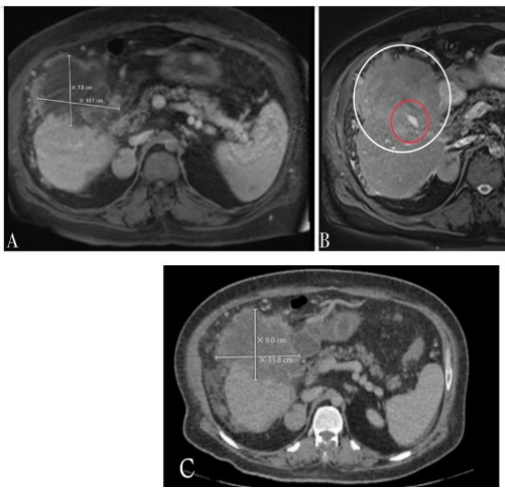
A triphasic CT scan confirmed metastatic disease; and hyperbilirubinemia was explained by hepatocellular invasion rather than obstructive cholestasis.

Echocardiography was significant for moderate pulmonary hypertension. Histopathological exam of a core biopsy of the liver lesion revealed moderately differentiated adenocarcinoma, consistent with gallbladder carcinoma with hepatic metastases. The patient was admitted for supportive and symptomatic management. He was kept nil per os (NPO) and started on intravenous fluids, proton pump inhibitors, broad-spectrum antibiotics, analgesics, anticoagulation, and diuretics. Ursodeoxycholic acid and hydrocortisone were added later in the course. Despite escalation of therapy, the patient developed clinical deterioration with ascites and worsening jaundice, requiring transfer to the intensive care unit.

During the ICU stay, the patient received supportive medications including ranitidine, famotidine, potassium chloride, calcium gluconate, and phytomenadione (vitamin K) as part of correction of electrolyte disturbances, gastric protection, and coagulopathy management, however, after few months, the patient died.

Conclusion: Our report adds to the scarce evidence by documenting an older age at presentation, establishing histopathological confirmation, and demonstrating even more extensive systemic spread at diagnosis. This case highlights the tendency for late presentation with disseminated disease and limited therapeutic options, underscoring the aggressive course of gallbladder carcinoma in DS. Collectively, these observations reinforce the importance of maintaining a high index of suspicion in DS patients presenting with hepatobiliary symptoms and support the consideration of earlier imaging and potentially screening approaches in this high-risk population. The low number of reported cases may reflect underdiagnosis or early mortality from other conditions. Larger prospective studies are needed to clarify the true incidence, evaluate screening strategies, and improve outcomes.

Figure 1. Abdominal Imaging Panel: MRI and CT of Hepatic Mass.



Legend: (A) MRI showing the large exophytic hepatic mass measuring: (10.1 × 7.8 cm). (B) T2 Weighted MRI Hepatic mass in segment VI (big circle) encasing and compressing the gallbladder (small circle). (C) Triphasic CT scan showing a large hepatic mass (9 × 11.8 cm) invading surrounding structures.

ISSN 2076-6327

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