Biliary Rhabdomyosarcoma Mimicking Choledochal Cyst

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Abstract

A 3-year old male presented with complaints of pruritus, abdominal pain for 3 weeks and jaundice. Stools were acholic. There was jaundice, liver palpable 3 cm below right costal margin, no ascites or palpable masses. Serology revealed albumin 2.9 g/dl; ammonia of 31 mmol/l; elevated conjugated bilirubin, GGT, ALT, AST and alkaline phosphatase; alpha fetoprotein 1.3 nmol/ml; hCG 9.1 IU/l; PT 12.3 secs, INR 0.9; negative hepatitis A,B,C serology. CT scan showed a non-calcified heterogeneously enhancing mass centered at the liver hilum. MRCP showed a large heterogeneously enhancing, partially solid mass in the region of the porta hepatic. Liver biopsy revealed patternless proliferation of polymorphic oval to spindled shaped neoplastic cells. There was bile ducts distortion. Immunohistochemistry revealed positivity for vimentin, desmin. These findings were diagnostic for biliary rhabdomyosarcoma. There was no evidence of metastasis. Chemotherapy was initiated. Repeat imaging 6 months after initiation of treatment showed improvement in the degree of intrahepatic ductal dilatation and decrease in tumor bulk size. Rhabdomyosarcoma is the most common malignant tumor of the biliary tree in childhood. It is difficult to diagnose and delayed diagnosis influences the prognosis.

Keywords


Introduction

Rhabdomyosarcoma (RMS), a soft tissue malignant musculoskeletal tumor, accounts for approximately 1% of the cases of cancers among children aged 0-14 years and 2% of the cases among adolescents and young adults aged 15 to 18 years. It is one of the rare causes of biliary tract obstruction. Nevertheless, it is the most common cause of obstructive jaundice due to neoplastic biliary obstruction in children.

The first case was reported in 1875 [1]. Rhabdomyosarcoma involving the liver accounts for 0.8% of all RMS and 1.3% of all liver tumors [2]. It almost exclusively occurs in children; 75% of patients are under 5 years of age with clear predominance of male patients. About 2% of tumors are present at birth [1-3]. The tumors are most commonly found in the hilum of the liver. Intermittent jaundice with or without abdominal distention, fever and anorexia is the typical presentation. The radiological appearance of the lesion is similar to that of a congenital choledochal cyst [4].

We report the case of a child with obstructive jaundice secondary to RMS of the biliary tree.

Case report

A 3-year old male presented with complaints of pruritus, epigastric pain for 3 weeks and jaundice for one day with stools appearing acholic. There was no history of fever, weight loss, or rashes. There was no contributory family history and no history of travel, trauma, use of herbal remedies / supplements or other medications by the patient. On physical examination the patient was found to have jaundice, no oral lesions, no cervical adenopathy, normal bowel sounds, a liver palpable 3cm below the right costal margin, no ascites or palpable masses and no splenomegaly. Laboratory studies showed conjugated bilirubin of 7.9 mg/dl; albumin 2.9 g/dl; ammonia of 31 mmol/l; GGT 468 U/l; ALT 149 U/L, AST 155 U/l; AP 1,351 U/L; white cell count 9,800/mm3; hemoglobin 11.2 g/dl; platelet count of 520,000/mm3; alpha fetoprotein 1.3 ng/ml; hCG 9.1 IU/l; prothrombin time 12.3 secs, INR 0.9; negative hepatitis A,B,C serologies.

Abdominal ultrasound showed a liver mass. CT scan of the abdomen (Fig. 1) showed a non-calcified heterogeneously enhancing mass centered at the liver hilum.
This mass had marked intrahepatic ductal dilatation and dilatation of the common bile duct. Magnetic resonance cholangiopancreatography (MRCP) (Fig. 2) showed a large (3.2 cm x 3.6 cm) heterogeneously enhancing, partially solid mass in the region of the porta hepatitis. Microscopic sections from fluoroscopic-guided liver biopsy revealed a pleomorphic spindle shaped neoplasm with irregular and occasionally lobulated hyperchromatic nuclei with abnormal mitoses. There was distortion of bile ducts consistent with an infiltrative lesion. Immunohistochemistry revealed cytoplasmic positivity for vimentin, desmin and focal nuclear myogenin reactivity within the neoplastic cells (Fig. 3). These findings were diagnostic for biliary embryonal RMS. The RMS was classified to be in stage I.

There was no evidence of metastasis. Surgery was not an option in this case, due to the tumor site. Chemotherapy consisting of vincristine, actinomycin-D and cyclophosphamide with mesna was initiated (COG ARST0331), and was later switched to POG 9803 which has a higher dose of cyclophosphamide.

Repeat imaging 6 months after initiation of treatment showed improvement in the degree of intrahepatic ductal dilatation and decrease in tumor bulk size. The patient recently received his last dose of chemotherapy. A recent MRI study showed further regression of the tumor with no evidence of metastasis.

**Discussion**

The diagnosis of biliary RMS can be difficult. It is a rare, aggressive malignant tumor that typically occurs in children. It often produces biliary tract obstruction with a hilar mass leading to signs and symptoms of obstructive jaundice. Jaundice is seen as a presenting symptom in 60-80% of cases and may be accompanied by acholic stools and
hepatomegaly. In patients with jaundice, moderate elevation of conjugated bilirubin and alkaline phosphatase can occur with elevation of transaminases.

The Intergroup Rhabdomyosarcoma Study Group (IRSG) has developed new protocols for children with sarcoma. For protocol purposes, patients are classified as low, intermediate or high risk [5]. Staging of RMS is relatively complex [6]. It includes assigning a local tumor group, staging and assigning a risk group. The estimated 5 year survival is 66%.

Undetectable distant metastases at diagnosis, grossly complete surgical removal of localized tumor, botryoid histology, tumor size <5cm and patient’s age <10 years at the time of diagnosis are favorable prognostic factors. Treatment can include surgery (usually biopsy only or subtotal resection) followed by chemotherapy and radiotherapy [7]. The extent of surgical resection currently recommended depends on the primary tumor site, and initial complete resection is generally recommended if it does not involve loss of organ function.

Immunohistochemical analysis is very useful in the diagnosis of embryonal RMS. More than 95% of these tumors are positive for desmin. In addition, nuclear staining with myogenin, a gene product that induces skeletal muscle differentiation, is useful in the diagnosis because of its specificity for rhabdomyosarcoma [6]. It is important to know that this very rare childhood tumor can simulate a congenital choledochal cyst. The presence of a soft tissue mass in porta hepatis rules out a choledochal cyst as a potential cause [4].

To sum up, rhabdomyosarcoma should be considered in the differential diagnosis of obstructive jaundice in children. With advances in the combined treatment of surgery, radiotherapy and chemotherapy, the prognosis has become much better and the chances of long term survival have improved.

References