



Embryonal Rhabdomyosarcoma of Biliary Tract a Diagnostic Dilemma

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I. INTRODUCTION

Hepatobiliary rhabdomyosarcoma is a rare lesion accounting for about 0.04% of all childhood cancers ¹. Diagnosis of Hepatobiliary tract Rhabdomyosarcoma (RMS) is usually difficult because of its rare prevalence, non-specific clinical and radiological findings and failure of demonstration of biliary origin and intra ductal growth of the tumor. This uncommon tumor was originally reported by Wilks and Moxon in 1875.

II. CASE REPORT

A 4 years old male child got admitted to our hospital with complaints of persistent abdominal pain and distension, yellowish discoloration of eyes, reduced appetite, mild drowsiness and swelling of both legs for 4 months.

Clinical examination of the child revealed that he was pale, icteric and appeared drowsy. He had bilateral pitting pedal edema with evidence of wasting of muscles in hands and leg. Per abdomen examination showed presence of shifting dullness and liver was palpable 8 cm below the right costal margin, firm in consistency and caput medusa noted.

Routine blood investigations were done. Liver function tests showed, Total bilirubin: 2.7 mg/dL; Conjugated bilirubin: 2.5 mg/dl Unconjugated bilirubin: 0.2 mg/dL; Globulin: 5.9 g/dL; GGT: 165 U/L; SGPT: 27 U/L; SGOT: 34 U/L, ALP: 299 U/L total protein : 7.9g/dl Albumin:3g/dl Blood smear showed microcytic hypochromic to normocytic normochromic picture. Hemoglobin variant analysis showed normal

hemoglobin pattern. HBsAG, Total anti HBC, Anti HCV were negative. Hb electrophoresis pattern, ANA (Antinuclear Antibody) was negative, Serum ceruloplasmin was normal (46.4 mg/d). Direct Coomb's Test was positive; Free T4 was 1.37 ng/dL; Free T3 was 2.85 pg/m. TSH was 7.31 mIU/L; LDH: 195 U/L; 24 hours urinary copper was normal (18.58 ug/day; 24 hours total urine volume: 120 mL/24hrs PT: 34.8 Seconds; INR (International Normalized Ratio): 3.3.

3TMRI abdomen with MRCP finding was suggestive of Type 4 Choledochal cyst with multiple small cholangitic abscess in both lobes of liver communicating with the bile duct (more in the right lobe). Loculated fluid collection with internal septae within the right hepatic duct and the entire length of the common bile duct - sequelae of cholangitis, moderate ascites and moderate right pleural effusion.

Portal vein doppler showed enlarged liver with dilatation of common bile duct, right and left intra hepatic biliary radicles, compression of main portal vein just below the level of porta by the dilated hepatic ducts. Portal vein was normal in caliber with normal color flow and velocity. Chest x-ray PA view was normal and UGI Endoscopy showed Antral Gastritis with mild extraneous impression in D1. No esophageal varices were noted. Whole Exome sequencing-THERMOFISHER: Variant of uncertain significance related to Obesity phenotype was detected.

SNV(s)/INDELS: Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	classification \$
MC4R (-) (ENST00000299766.5)	Exon 1	c.457T>A (p.Tyr153Asn)	Heterozygous*	Obesity (OMIM#618406)	Autosomal dominant; Autosomal recessive*	Uncertain Significance (PM2, PP3)



He underwent Laparotomy and Choledochal Cyst Excision + Hepaticojejunostomy and Liver Biopsy under General anesthesia.

HPE reports showed tumor cells of variable morphology with subepithelial area showing undifferentiated round cells and deeper area showed skeletal muscle differentiation with moderate to marked pleomorphism showing hyperchromatic nucleus and abundant eosinophilic cytoplasm. Some of the cells showed elongated tadpole like cytoplasm with striations resembling strap cells. Occasional

multinucleated tumor cells are also seen. Features are consistent with Embryonal Rhabdomyosarcoma of the biliary tract. The liver biopsy showed chronic active hepatitis. Immunohistochemistry showed Desmin - Clone : GM007, Myogenin - Clone : F5D, MyoD1 - Clone : EP212 positive in tumor cells. He was started on chemotherapy (VAC regimen- Vincristine, Actinomycin and Cyclophosphamide). He symptomatically improved and was followed up on OPD basis.

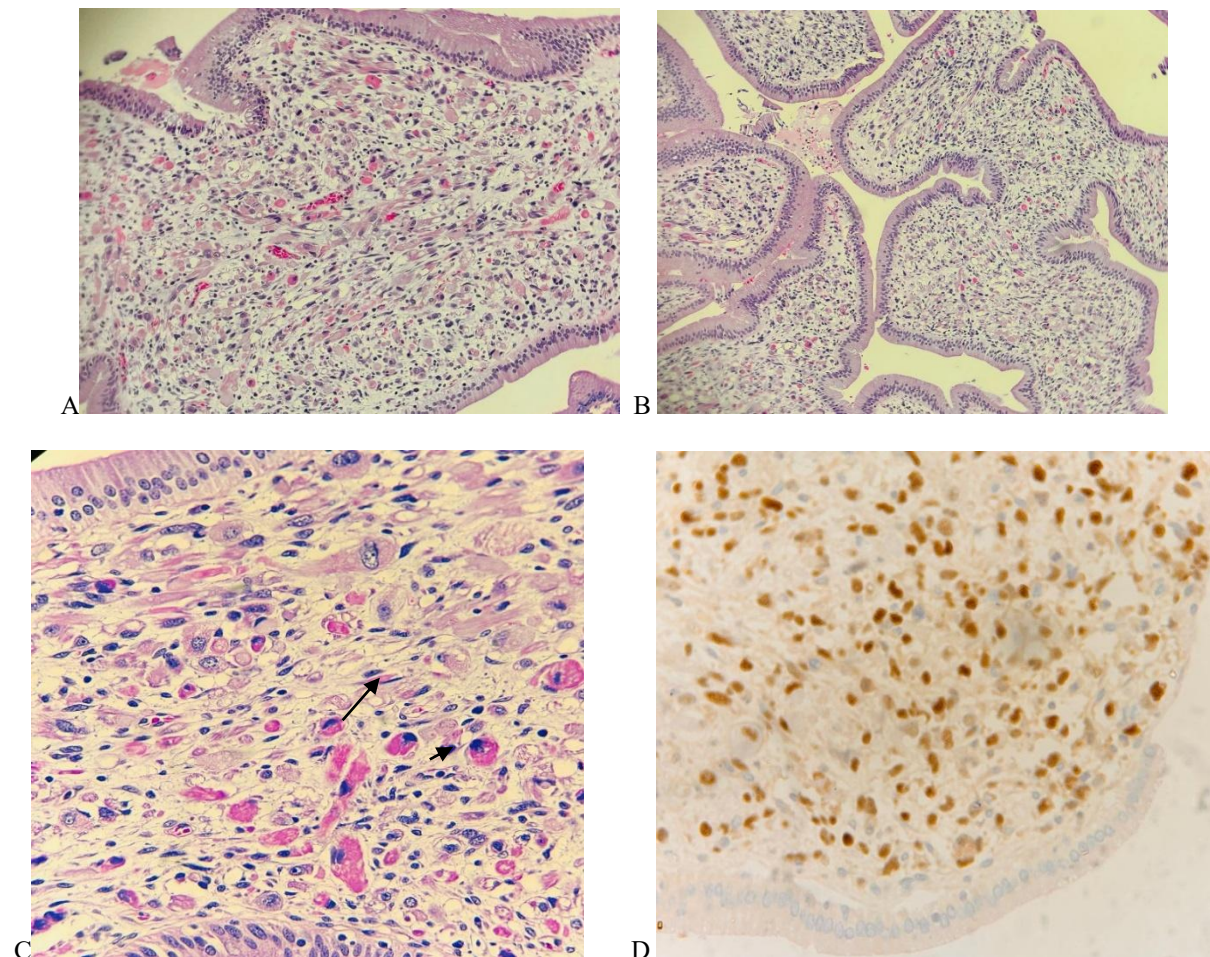


Figure 1: A & B showing rounded blue cells under low power.
C & D showing pleomorphic, strap cells (arrows) with Desmin positivity.

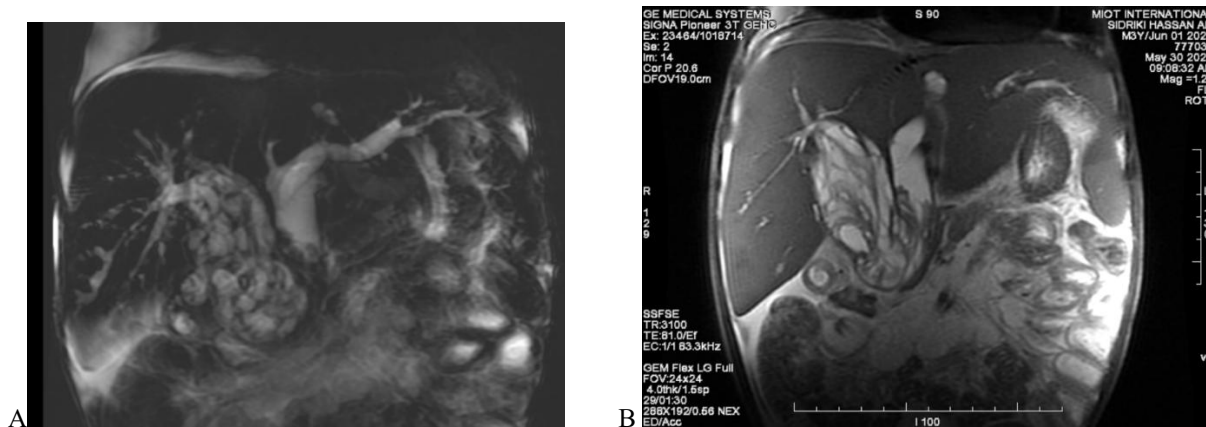


Figure 2: A & B showing Type IV choledochocyst with multiple septations/ abscesses later on turned out to be Rhabdomyosarcoma



Figure 3: Intra operative gross gel like appearance of mass (RMS)

III. REVIEW OF LITERATURE

Tumors that arise from the biliary tree in children are generally very rare.² Hepato Biliary tract RMS represents about 0.5% of pediatric rhabdomyosarcoma and about 0.04% of all pediatric malignancy.³ Wilks and Moxon reported the first case of Hepatobiliary RMS on 1889 and till now about 100 cases have been reported in the literature.^{2, 4, 5} Common presentation of Hepatobiliary RMS is jaundice associated with abdominal pain, vomiting and fever. This non-specific presentation makes the diagnosis of RMS difficult, since more common differential diagnosis like infectious hepatitis and choledochal cyst are considered first.

Most of the patients reported in the literature were initially treated as infectious hepatitis, especially when they showed initial improvement, until they were referred to specialized higher centers when the condition was refractory to treatment. This is the reason for the usual long duration between the onset of symptoms and definitive diagnosis and treatment of Hepato Biliary tract RMS in most of the reported cases.³ An

abdominal ultrasound and findings of mild transaminitis will help in exclusion of infectious hepatitis which is usually the initial suspect.⁶

Most of the reported cases in recent literature were misdiagnosed as choledochal cysts until tissue diagnosis is obtained.^{7, 8, 9} Differentiation between Hepato Biliary tract RMS and choledochal cyst may be difficult even during pathological examination of the surgical specimen. Clinical and laboratory findings in both diseases are the same with no significant difference. Differentiation by diagnostic radiology is difficult when there is no visible mass and detection of intraductal growth cannot be achieved.¹⁰ Since embryonal RMS has both solid and cystic components, preoperative imaging may not be very accurate to define the lesion precisely.¹³ Botryoid RMS, the histological variant of RMS together with embryonal type, may initially be misdiagnosed as benign lesion as developmental anomaly or chronic inflammation.¹¹

Differentiation of Hepatobiliary RMS from choledochal cyst is very important as it may help avoiding unwarranted surgery in case of Hepato



Biliary tract RMS.¹² Hepato Biliary tract RMS should be considered in the differential diagnosis in any child with obstructive jaundice especially when a mass, intraductal growth or distant metastasis are detected.³ Finally, the detection of solid masses during surgical exploration for choledochal cyst can be sent for frozen section and when rhabdomyosarcoma is suspected, immunohistochemical staining using monoclonal antibody to the myogenic regulatory protein "MyoD1" can be used on frozen tissue.¹⁰ Nevertheless, our case is an example of how differentiation between HRMS and choledochal cyst can be difficult even after surgical exploration and intraoperative cholangiography were performed and no intraductal growth or hepatic masses were detected.

Chemotherapy using vincristine, dactinomycin, and cyclophosphamide can be used in case of extensive disease or R1 resection followed by second look surgery to evaluate resectability. Radiotherapy is indicated when chemotherapy fails or when there is postoperative residual tumor. Palliative treatment of obstructive jaundice is done by ERCP and biliary stenting.⁶

The concept of operability in Hepato Biliary tract RMS is different from other hepatic tumors. Some authors find it unnecessary to perform major surgical resection with available treatment alternatives especially when the long term survival is not affected by R0 resection.¹² Large tumors involving both liver lobes is not a contraindication for surgery because the tumor is usually localized to the bile ducts and complete excision is not mandatory for long term survival. Also, second look surgery can be performed after chemotherapy for extensive and residual tumors.¹⁰

To conclude, Embryonal rhabdomyosarcoma must be considered in the differential diagnosis of a child with obstructive jaundice. Having a strong suspicion, broad differential diagnosis and multi modal diagnostic modalities are needed to differentiate the tumor from choledochal cyst.

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