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CAROLI'S DISEASE – A CASE REPORT

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ABSTRACT: Caroli's disease, also known as congenital communicating cavernous ectasia of the biliary tree, is a rare autosomal recessive developmental abnormality characterized by saccular dilatation of the intrahepatic bile ducts⁽¹⁾. Caroli's disease is a rare congenital disorder, and occasional cases have been reported from Japan and other parts of Asia ⁽²⁾. We present our experience with an unusual case of Caroli's disease.

INTRODUCTION: Jacques Carole in 1958 first described a rare disease with multifocal, segmental, saccular dilatation of the large intrahepatic bile ducts, which causes stagnation of bile formation of biliary sludge and stones⁽³⁾ Etiology- unknown, Occurrence- sporadic, Caroli's disease results in recurrent cholangitis, abdominal pain and hepatic abscesses. Diagnosis- confirmed by ERCP and percutaneous transhepatic cholangiography. Purpose of treatment is to restore normal bile flow. Prognosis is poor despite drainage of bile and patients die from sepsis, hepatic abscess, hepatic failure or portal hypertension. Mortality is indirect and caused by complications. After cholangitis occurs, patients typically die within approximately 5-10 yrs⁽⁴⁾ We report a case of 30 yr female who was diagnosed as Caroli's disease. Most of the reported cases in literature are in pediatric age group.

HISTORY: A 30 year old female presented with chief complaints of intermittent fever since 2 months, decreased appetite and vomiting also of same duration. She gave no history of hematemesis, melena or abdominal distension. She had not received any blood transfusion in the preceding months. Patient was diagnosed with Rheumatoid arthritis 11 years ago, but she was asymptomatic at presentation. There was no past history of diabetes mellitus, IHD or bronchial asthma. No one else in the family had similar illness

PHYSICAL EXAMINATION: Patient was afebrile. Her pulse was 70 b/min and blood pressure 100/70 mm Hg. On general examination patient had pallor, deep icterus, swan neck deformity in the middle finger of right hand, boutonniere deformity in right little finger. There was no cyanosis/clubbing/edema feet/lymphadenopathy. Patient did not have any signs of acute or chronic liver failure.

On cardiovascular system examination - Heart sounds were Normal and there was no murmur. On Respiratory system examination- Air entry was equal bilaterally and there were no adventitious sounds. On per abdomen examination- Tender, soft, mild Hepatomegaly with firm and rounded margin. There was no splenomegaly/ascitis/bruit/hum. On central nervous system examination patient was conscious and oriented. There were no flaps and no other neuro deficit.

LABORATORY ASSESSMENT: On Complete blood count examination results were Hb-6.8 gm%, TLC- 6400 cu.mm, DLC-N68% L26% E04% M02% platelets- 3.34 lakhs/ mm. Peripheral Smear showed Normocytic normochromic, No abnormal cells seen. Erythrocyte sedimentation rate was

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34mm/1hour. Kidney function test showed blood urea- 20.8mg%, serum creatinine-0.6mg % sodium-140 meq/lit potassium 3.5 meq/lit. Her random blood sugar was 96 mg per dl. HIV; HBs Ag, Anti HCV, HEV IgM tests were negative. Sickling test was negative

Date	04.06.2013	13.06.2012	18.06.2012
S. Bilirubin (mg %)	20.79	18.4	21.2
Direct (mg %)	12.67	9.8	10.0
Indirect (mg %)	8.12	8.6	11.20
SGPT(U/litre)	105	96.9	114.7
SGOT(U/litre)	127.3	150.8	153.2
Alkaline Phosphatase (U/litre)	699.5	935.1	772.4
Total Proteins (gm %)	6.4	6.1	5.7

IMAGING: Chest X- ray: Normal. USG Abdomen: Coarse echotexture of liver with scattered IHBR showing focal dilation with normal common bile duct, possibility of cholangitis. CECT abdomen: Mild hepatomegaly with multiple cystic areas in hepatic parenchyma seen communicating with the intrahepatic ducts, giving beaded appearance. These represent saccular dilated intrahepatic ducts s/o Caroli's Disease with secondary cholangitis.

DIAGNOSIS - CAROLI'S DISEASE

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Treatment: Antibiotics for cholangitis-Tab ofloxacin 200 mg twice daily for 28 days and Ursodeoxycholic acid for cholestasis- Tab. Ursodeoxycholic acid 300mg twice a day for 28 days.

DISCUSSION: Originally described by Jacques Caroli in 1958. A rare congenital disorder characterised by multiple segmental or saccular dilatation of the large intrahepatic bile ducts. The cause appears to be genetic; the simple form is an autosomal dominant trait while the complex form is an autosomal recessive trait. [5] Females are more prone to Caroli disease than males. [6] Family history may include kidney and liver disease due to the link between Caroli Disease and ARPKD. [7] PKHD1, the gene linked to ARPKD, has been found mutated in patients with Caroli syndrome. PKHD1 is expressed primarily in the kidneys with lower levels in the liver, pancreas, and lungs, a pattern consistent with phenotype of the disease, which primarily affects the liver and kidneys. [5][7] The genetic basis for the difference between Caroli disease and Caroli syndrome has not been defined. Disease affects about 1 in 1,000,000 people, with more reported cases of Caroli syndrome than of Caroli disease. [4] when associated with congenital hepatic fibrosis- Caroli's syndrome. Caroli's syndrome which has autosomal recessive or dominant inheritance occurs more frequently than in Caroli's disease. Caroli disease is also associated with polycystic kidney disease and with liver failure. Pathogenesis – Congenital malformation theory; Genetics poorly defined. Pathology- cystic dilatation occurs throughout the intrahepatic biliary tract. Saccular dilatation predisposes to bile stagnation, biliary sludge and intraductal lithiasis, which may result in chronic. Abdominal pain and pancreatitis. Bacterial cholangitis occurs frequently, complicated by sepsis and hepatic abscess. Signs and symptoms-The first symptom typically include fever, intermittent abdominal pain, and hepatomegaly. Occasionally jaundice occurs. Caroli disease usually occurs in the presence of other diseases such as autosomal recessive polycystic kidney disease, cholangitis, gallstones, biliary abscess, septicemia, liver cirrhosis, renal failure and cholangiocarcinoma. People with Caroli disease are 100 times more at risk for cholangiocarcinoma than the general population. [6] Complications- Cholangitis, sepsis, Choledocholithiasis, Hepatic abscess. Lab Assessment –Raised bilirubin, SGOT, SGPT, ALP, Leucocytosis may indicate cholangitis. Raised urea creatinine suggests underlying polycystic renal disease. Transabdominal imaging- Doppler sonogram and CT scan Abdomen. ERCP-remains the gold standard for diagnosis. Liver biopsy not required. Treatment- Antibiotics to treat Cholangitis and sepsis [8]; Hepatectomy for focal disease ; [9] Liver transplantation for diffuse disease. [10] Prognosis-Poor despite drainage of bile. 46% patient dies from complications. In our case Caroli's disease was suspected after USG report, CECT abdomen supported the diagnosis.

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