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Jobert syndrome " a rare cause of liver dysfunction"

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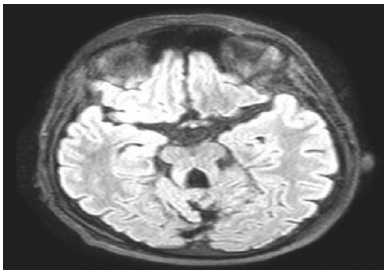
INTRODUCTION: Joubert syndrome is an autosomal recessive inherited disease with no specific gene mutation. Diagnosis is made by the evaluation of clinical and radiological findings. Coexistence of Joubert syndrome with congenital hepatic fibrosis is called COACH syndrome (Cerebellar vermis hypoplasia,Oligophrenia,Ataxia,Coloboma,Hepatic fibrosis) which is considered a subtype of Joubert syndrome. In this article, we report a case of COACH syndrome presenting with choledocholithiasis to our clinic.

CASE: A 40-year-old male patient was admitted to our clinic with complaints of right upper quadrant pain, itching and jaundice. The patient had mental retardation and ataxic movements. Laboratory findings were as follows: AST:77, ALT: 189, Serum Total / Direct Bilirubin: 8 / 7.1 mg/dl, WBC: 5360 mm3, HgB: 11.1 mg/dl, Plt:78000, Prothrombin Time: 45.6 sec, INR: 3.6, Sedimentation: 93. The Portal Doppler examination was found to be normal. MR-MRCP showed hepatomegaly, splenomegaly, an increase in the diameter of the portal vein (14 mm), intrahepatic and choledochal dilatation, and multiple stones in the gallbladder. In the ERCP of the patient multiple stones with a diameter of 20 mm were observed in the common bile duct. Ceruloplasmin and 24-hour urine copper level were found to be normal. Cranial MRI showed molar tooth appearance in the superior peduncle, superior cerebellum atrophy which leads to dilatation of the follicles. These findings were consistent with Joubert syndrome. Esophageal varices were present in the upper GI endoscopy. ANA, AMA, Anti-LKM1 and ASMA tests were found to be normal. Liver biopsy performed as the liver function tests were persistently high. In histopathological examination of liver biopsy material, Histological activity index 6 and Stage 3 Fibrosis were found which was considered compatible with congenital hepatic fibrosis. In the follow-up, the patient's bilirubin levels and coagulation parameters regressed to normal. The patient was discharged from hospital after recommending for outpatient clinic follow-up.

DISCUSSION: Joubert Syndrome is reported to be 1 in 80 000 - 100 000 live births, and COACH syndrome is a more rare condition. Due to this malformation in the liver, the expansions of the primitive bile ducts and the fibrotic expansion of the portal pathways are observed. In the literature, it is stated that liver enzymes (AST,ALT,GGT) may increase more than two times than the normal values, hepatosplenomegaly in the early stages and more severely portal hypertension and esophageal varices due to liver cirrhosis can be seen in COACH syndrome with Joubert syndrome. In our case, hepatic fibrosis, the most severe form, was present. In conclusion, though Wilson's disease is more commonly seen in patients with mental retardation, ataxia, and hepatic impairment, COACH syndrome should be considered as a differential diagnosis in addition to Wilson's disease.

Keywords: joubert, hepatic fibrosis, mental retardation



MRI of patient with joubert syndrome

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