



Coach syndrome: The first case from Turkey

To the Editor,

A 21-year-old mentally retarded male was admitted to our department with abdominal swelling. A physical examination revealed mental retardation and nystagmus. The liver and spleen were 7 and 15 cm palpable, respectively. Ascites and edema of the leg were not detected. There was marked ataxia. Other findings of the physical examination were normal. Blood test results included hemoglobin: 12.2 gr/dL, White blood cell WBC: 1520/mm³, platelet count: 27.000/mm³, SGOT: 154 IU/L (normal range: 5-35), SGPT: 109 IU/L (normal range: 5-40), ALP: 1190 IU/L (normal range: 90-260), GGT: 324 IU/L (normal range: 7-32), total bilirubin: 1,42 mg/dL (normal range: 0,1-1). Other laboratory investigations were found to be within normal ranges. Chronic liver disease and portal hypertension were found on Doppler ultrasonography. Viral hepatitis (including hepatitis A, B, C, EBV, CMV, and HIV), Wilson's disease, and

autoimmune hepatitis were ruled out. Propranolol was given because an upper gastrointestinal endoscopy revealed grade 2 esophageal varices. A liver biopsy revealed all of the criteria for congenital hepatic fibrosis (CHF) with ductal plate malformation (Figure 1). Magnetic resonance imaging of the brain revealed superior vermis atrophy (Figure 2). The patient was diagnosed with COACH syndrome due to the congenital hepatic fibrosis, superior vermis atrophy, ataxia, nystagmus and mental retardation (1). Coloboma was not detected on ophthalmoscopic examination, and abdominal computed tomography revealed no renal abnormalities. The patient was discharged with planned outpatient follow-up visits to the neurology and gastroenterology departments. Informed written consent was obtained from the patient.

The term CHF, which is encountered mainly in children and young adults, was first introduced by Kerr et al. (2).

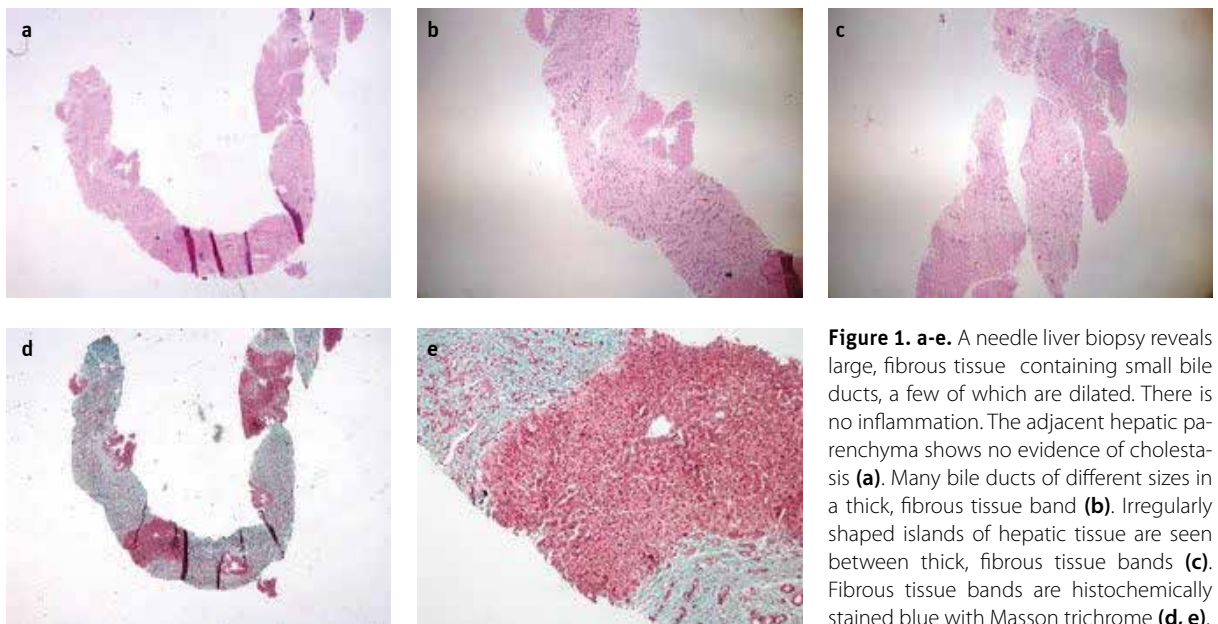


Figure 1. a-e. A needle liver biopsy reveals large, fibrous tissue containing small bile ducts, a few of which are dilated. There is no inflammation. The adjacent hepatic parenchyma shows no evidence of cholestasis (a). Many bile ducts of different sizes in a thick, fibrous tissue band (b). Irregularly shaped islands of hepatic tissue are seen between thick, fibrous tissue bands (c). Fibrous tissue bands are histochemically stained blue with Masson trichrome (d, e).

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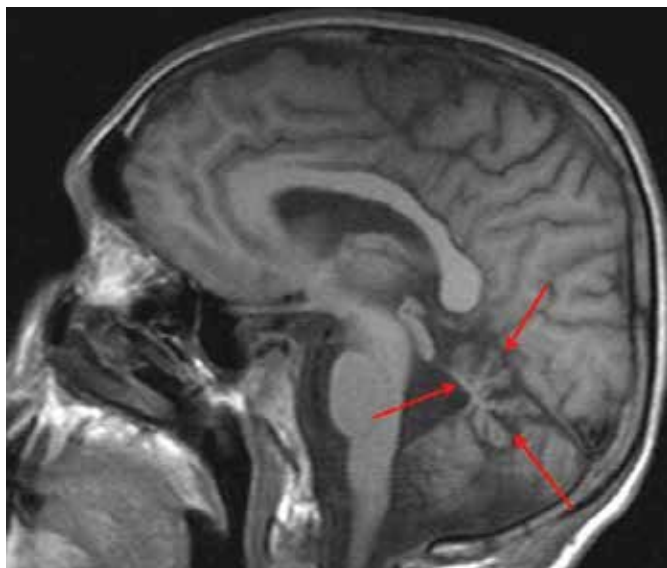


Figure 2. Magnetic resonance imaging of the brain indicating superior vermian atrophy (arrows).

CHF may be associated with other congenital abnormalities such as ocular abnormalities, renal disease, cerebellar malformations, and mental retardation (3). However, the term oculo-encephalo-hepato-renal syndrome does not represent a single disorder, but rather a group of disorders including Meckel syndrome (4), Joubert syndrome (5), Arima syndrome (6), and COACH syndrome (1). COACH syndrome was first described by Verloes in 1989, and is characterized by hypoplasia of the cerebellar vermis, oligophrenia, congenital ataxia, coloboma and hepatic fibrosis (1). The present case did not present with oligophrenia and coloboma, which was also seen in other cases of COACH syndrome in the literature. This syndrome is extremely rare, and to our knowledge, we are presenting the first COACH syndrome case from Turkey. We believe that possible oculo-encephalo-renal disorders should be investigated in young

patients with mentally retardation, ataxia and CHF.

Ethics Committee Approval: N/A.

Informed Consent: Written informed consent was obtained from patient who participated in this study.

Peer-review: Externally peer-reviewed.

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