ABERNETHY SYNDROME; REPORT OF A CASE

Case Report

ABERNETHY SENDROMU; BİR OLGU SUNUMU

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ABSTRACT

Congenital extrahepatic portosystemic shunt known as Abernethy malformation is a rarely seen disorder. It is classified into two types based on the pattern of anastomosis between the portal vein(PV) and inferior vena cava(IVC), and the presence or absence of an intrahepatic portal venous supply. Patients with Abernethy malformations have cardiac and hepatic abnormalities. It rarely can lead to hepatocellular carcinoma in the long term. Here we report a 16- years- old girl who is diagnosed as Abernethy malformation as a result of investigations of early puberty.

Key words: Abernethy syndrome, portosystemic shunt, child

ÖZET

Konjenital ekstrahepatik portosistemik şant (Abernethy malformasyonu) çok nadir görülen bir bozukluktur. It is classified into two types based on the pattern of anastomosis between the portal vein(PV) and inferior vena cava(IVC), and the presence or absence of an intrahepatic portal venous supply. Patients with Abernethy malformations have cardiac and hepatic abnormalities. It rarely can lead to hepatocellular carcinoma in the long term. Here we report a 16- years- old girl who is diagnosed as Abernethy malformation as a result of investigations of early puberty

Anahtar kelimeler: Abernethy sendromu, portosistemik şant , çocuk

INTRODUCTION

Congenital extrahepatic portosystemic shunt known as Abernethy malformation was first defined in 1793 after 10 months-old baby's autopsy by John Abernethy (1). Congenital extrahepatic portosystemic shunt is classified into two types based on the pattern of anastomosis between the portal vein(PV) and inferior vena cava(IVC), and the presence or absence of an intrahepatic portal venous supply (2). Patients with Abernethy malformations have cardiac and hepatic abnormalities (3-6) There is evidence that the disease may cause hepatocellular carcinoma in the long term (3,7,8) Therefore, early recognition of the disease is important, early treatment of complications that may occur in the follow-up the disease. In this case report we present a 16-years- old girl who is diagnosed as Abernethy malformation as a result of investigations of early puberty.

CASE REPORT

16 years old girl was examined by a physician for accelerated growth one year ago. Her detailed history revealed early puberty Laboratory tests were done and HOMA-IR was high and indirect bilirubin 1,5mg/dl with normal transaminases, so metformin and UDCA were started. Abdominal ultrasound revealed liver and biliary tract abnormalities with suspect of ademoas therefore the patient was directed to us. The patient had no complaint. On physical examination her heigt was 186cm (>97.percentil), weight was 93kg(>97.percentil). Abdominal examination was normal, there was no jaundice, hepatosplenomegaly and evidence of encephalopathy. She was Tanner grade 5 and having normal menstrual cycle. Laboratory tests were ALT: 30 U/L, AST: 33 U/L, total bilirubin: 0.77 mg/dL, direct bilirubin:0.38 mg/Dl, ft4: 0.859 ng/dl, testosteron:0.805 ng/dl (Tanner grade 5), free testosteron: 3.39pg/mL. Abdominal USG revealed congenital portal vein agenesis, aberrant dilated venous structures associated with vena cava and multiple nodules in liver.

Upper abdominal contrast-enhanced MR revealed the absence of intrahepatic portal vein; Abernethy malformation type 1b and multiple mass lesions in liver (Figure 1-2). The patient was diagnosed with Type1b Abernethy Malformation and the parents and the patient herself were informed about the disease. The patient is being followed every 6 months for transaminases and alpha fetoprotein levels and once a year abdominal ultrasound for possible malignant condition

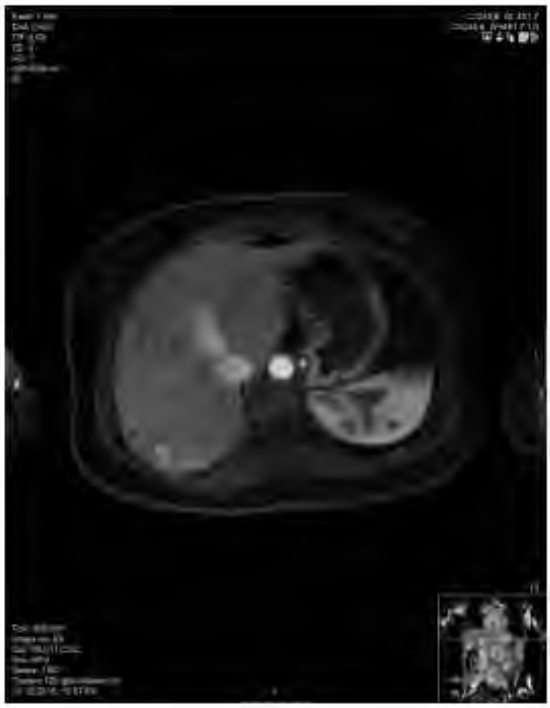

Fig1

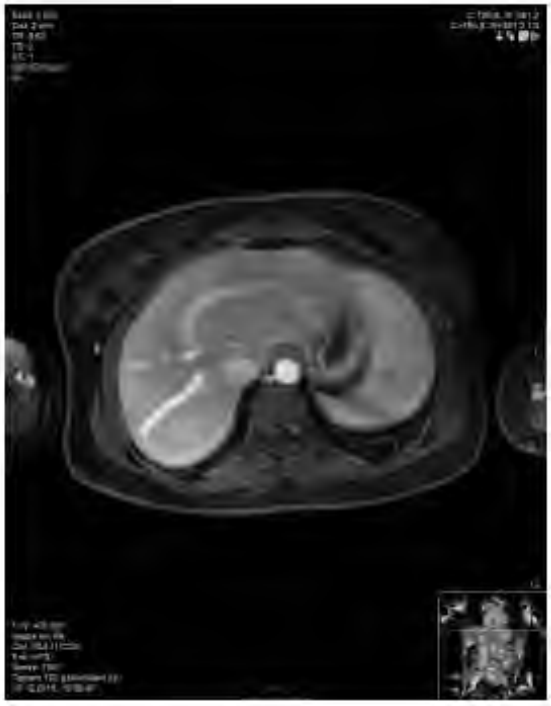

Fig2

DISCUSSION

Congenital extrahepatic portosystemic shunt known as Abernethy Malformation is a very rare splanchnic venous system anomaly. Nowadays, with the development of imaging techniques patients are diagnosed earlier.

Portal vein system develops in the 4th-10th weeks of the embryonic life as a result of the selective apoptosis in one part of vitelline veins. IVC development also corresponds to that period. The result of the insufficiency in the anastomosis, porto-systemic and porto-portal shunts happens olacak galiba in this period.

There are two types of anomalies of porto-systemic shunts. Type 1 malformation all venous blood drains to inferior vena cava with absence of the intrahepatic portal vein. . Type 1 malformation is divided into two subgroups that based on the absence or precense anastomosis between the the splenic vein and superior mesenteric vein. In the Type 1a shunt superior mesenteric and splenic vein drains to inferior vena cava, iliac veins and renal veins separately; in type 1b the superior mesenteric vein and splenic vein combines and inferior drains in to the vena cava. Type 2 malformation portal venous blood partially drains into the IVC through side-to- side anastomosis. Our patient was finally diagnosed with type 1b malformation.

Congenital intra-hepatic portosystemic shunts can present in the early neonatal period with growth restriction (9), galactosemia (9-12), neonatal cholestasis (13), and hepatic encephalopathy (4). Some cases can be diagnosed by neonatal screening tests due to presence of galactosemia in some countries having neonatal screening programmes (9-12). Patients with Abernity malformations almost have cardiac and hepatic abnormalities.

At the same time polysplenia, biliary atresia, annular pancreas, situs inversus, urinary tract anomalies and skeletal abnormalities may be associated with. Symptoms of secondary complications like hypoglycemia, hyperammonemia, encephalopathy, and heart failure secondary complications can be seen as temporary and resolve spontaneusly (4). Subclinical course is more common and some patients might not have any symptoms throughout life (4,5,14) Any other abnormality has not detected in the examinations of our patient.

There are 2 cases reported with early pubarche, with inappropriately low DHEAS levels, hyperandrogenemia and advanced bone age, both had hyperinsulinemia. The presented case is similar with those girls who are reported from Turkey.

We suggest patients with suspected liver or portal system abnormalities should be evaluated at spesific centers. There are novel differences in portosystemic shunts so one should be careful when dealing with such patients and all systems should be evaluated as that abnornality may effect many systems except from gastrointestinal system.

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