Wilson Disease with Situs Inversus Totalis

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ABSTRACT

Situs inversus is a positional anomaly that rotates the abdominal internal visceria and when it is associated with transposition of the thoracic organs it is called situs inversus totalis. Wilson disease (or hepatolenticular degeneration) is an autosomal recessive hereditary disease of human copper metabolism, which causes hepatic and neuropsychiatric symptoms. We describe a case report of situs inversus totalis with Wilson disease, which is the first case to our knowledge.

Key words: *Situs inversus, dextrocardia, Wilson disease, Kayser-Fleischer ring*

Wilson Hastalığı ile Situs İnversus Totalis Birlikteliği

ÖZET

Situs inversus abdominal organların rotasyonuyla giden pozisyonel bir anomali olup torasik organ transpozisyonu da eklendiğinde hastalık situs inversus totalis olarak adlandırılmaktadır. Wilson hastalığı (hepatolentiküler dejenerasyon) hepatik ve nöropsikiatrik semptomlara neden olan otozomal resesif kalıtılan bakır metabolizması hastalığıdır. Bu olgu sunumunda Wilson hastalığı ile situs inversus totalis birlikteliği gösteren ve literatüre göre ilk vaka olan bir olgu tanımladık.

Anahtar kelimeler: *Situs inversus, dekstrokardi, Wilson disease, Kayser-Fleischer halkası*

INTRODUCTION

Situs inversus is a congenital anomaly characterized by the transposition of the abdominal visceria. It may be associated with dextrocardia, also known as situs inversus totalis (1,2). This rare genetic anomaly is usually discovered incidentally during thoracic and abdominal imaging and affects approximately 0.005 % of all live birth (3). The cause of situs inversus is unknown. This rare anomaly is associated with various gastrointestinal anomalies. In the current literature, the associations of acute appendicitis, liver transplantation which caused by biliary atresia and intestinal ischemia due to intestinal malrotation were reported. However there is no data for association with Wilson disease (or hepatolenticular degeneration). Wilson disease is an autosomal resessive disorder of hepatic copper metabolism (4). Defective copper excretion leads to systemic accumulation of copper that gives rise to typical phenoytypes that include progressive liver damage, neurological deficitis, psychiatric illness, renal tubuler disorders, presence of Kayser-Fleischer ring, hypoparathyroidism and cardiomyopathy.

CASE

A 42 years old man, who had no personal or family history of disease, referred to us with abdominal distension, jaundice, progressive malaise, liver impairment and dysartria. On physical examination, his blood pressure was 100/60 mmHg, temperature 36.7 °C, the pulse rate 92 beats/min and he was icteric. On abdominal examination, a fluid wave was noted, without any tenderness or guarding. On neurological examination the patient displayed slurred speech and ataxia. Laboratory inves-

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tigations revealed a deranged liver function profile; alanine transamianase 50 U/L; aspartate transamianase 64 U/L; alkaline phosphotase 125 U/L; total bilirubin 6,5 mg/dL direct bilirubin 3,4 mg/dL; serum albumin level 1.9 gr/dL; INR level 2,1 and viral hepatitis serology were normal. ANA, AMA and Anti-LKM1 was negative. Alpha-1 antitrypsin level was in normal range. Serum copper was 35,9 µg/dL (reference range 50-155 µg/dL) serum ceruloplasmine 14,3 mg/dL (reference range 22-58 mg/dL) and 24 hour urinary copper excretion 1654 µg/24h (reference range 3-35 µg/24h). The patient was examined by an ophtalmologist who noted bilateral Kayser-Fleischer rings. Esophagogastroduodenoscopy (EGD) disclosed evidence of portal hypertension such as esophageal varices and erosive gastritis. In light of this information Wilson disease was diagnosed. Abdominal ultrasonography showed; left-sided liver with coarsened hepatic echo texture and right-sided lien with splenomegaly, and massive ascites with no anatomical biliary tree obstruction. Also posteroanterior radiography of the chest showed dextrocardia and right sided gastric buble (Figure 1). A computed abdominal tomography showed situs inversus totalis and ascites (Figure 2). Therefore we accepted our case as Wilson disease with situs inversus totalis. D-Penicillamine therapy was started and the examination of his whole family was recommended.

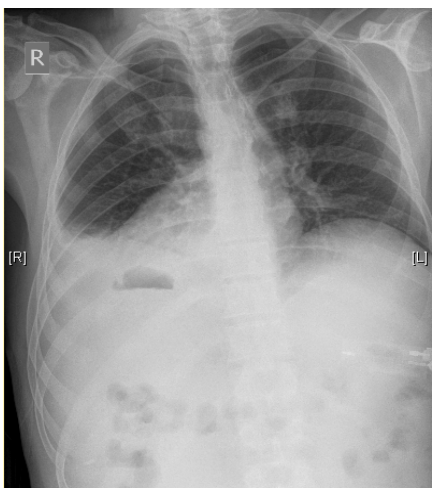

***Figure 1**. Posteroanterior radiography of the chest shows dextrocardia and right sided gastric buble*

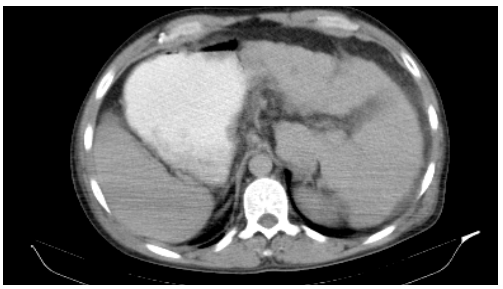

***Figure 2**. Left sided liver and right sided spleen, with ascites*

DISCUSSION

Situs inversus is a positional anomaly that rotates the abdominal internal visceria and when it is associated with transposition of the thoracic organs it is called situs inversus totalis. It affects 0.001 % to 0.01 % people with a male to female ratio of 3:2 (5). Situs inversus is generally an autosomal recessive genetic condition, although it can be X-linked or found in identical "mirror" twins (6). Situs inversus totalis is accompanied with different diseases; Toros and friends showed that salivary gland choristoma of the middle ear in a child with situs inversus totalis (7). Brown et al. reported a case about duodenal atresia with situs inversus totalis (8). Jimenen et al. reported a case who had a renal adenocarsinoma with situs inversus totalis which was associated bronchiectasis and chronic sinusitis (Kartagener's syndrome) (9). Wilson disease or hepatolenticular degeneration is a rare autosomal recessive hereditary disorder of human copper metabolism, which primarily manifests hepatic and/or neuropsychiatric symptoms. However, other organ system complications can be seen, including cataracts, renal abnormalities, hypoparathyroidism, severe hemolysis, arthritis and cardiac abnormalities. ATP7B gene located on chromosome 13 (13q143-q21.1), coding the protein for hepatic copper transport is affected in Wilson disease (4). To our knowledge we have reported a first case of situs inversus totalis accompanied with Wilson disease.

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