Portal Ven Anevrizması Saptanan Klinefelter Sendromu Olgusu

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ABSTRACT

Klinefelter syndrome (KS) is the most common chromosomal disorder in men characterized by clinical features of hypogonadism and infertility. About 90% of cases have classically 47,XXY karyotype and the remaining have additional X or Y chromosomes, high grade aneuploidies or X chromosome structural abnormalities (1).

Venous aneurysms (VAs) have been reported to occur in most major veins frequently in popliteal, jugular, or saphenous veins. Visceral VAs have been increasingly described in recent years probably due to increase in imagings.

Portal vein aneurysms (PVA) are the most common site of visceral VA, representing fewer than 3% of all VAs (2-4). Less than 200 cases of PVAs have been reported since first discovered in 1956 by Barzilai and Kleckner (5,6). PVA has been frequently detected incidentally on imaging studies. Patients are frequently asymptomatic or have unrelated gastrointestinal complaints of abdominal pain, nausea and vomiting. Further investigations revealed 23 mm anechoic, saccular expansion in the left branch of the portal vein. It is well known that KS is associated with venous thromboembolic diseases including portal venous thrombosis, but association with portal vein aneurysm has not been previously reported.

Key Words: Portal vein aneurysm, klinefelter syndrome

INTRODUCTION

Klinefelter syndrome (KS) is the most common chromosomal disorder with an incidence of 1:500 male live births. It is characterized by clinical features of hypogonadism and infertility. About 90% of cases have classically 47,XXY karyotype and the remaining have additional X or Y chromosomes, high grade aneuploidies or X chromosome structural abnormalities (1).

Venous aneurysms (VAs) have been reported to occur in most major veins frequently in popliteal, jugular, or saphenous veins. Visceral VAs have been increasingly described in recent years probably due to increase in imagings. Portal vein aneurysms (PVA) is the most common site of visceral VA, representing fewer than 3% of all VAs (2-4). Less than 200 cases of PVAs have been reported since first discovered in 1956 by Barzilai and Kleckner (5,6). PVA is usually detected incidentally on imaging studies. Patients are frequently asymptomatic or have unrelated gastrointestinal complaints of abdominal pain or nausea that prompt abdominal imaging. Doppler ultrasound is the diagnostic standard for intrahepatic ones (7). Monitoring for expansion. Maximum portal vein diameter of 20 mm is the diagnostic standard for extrahepatic portal vein aneurysms and 9 mm is the diagnostic standard for intrathoracic ones (7).

In this report we describe a 19 year-old man with KS and incidentally detected PVA.

CASE REPORT

A 19-year-old man was admitted with the complaints of abdominal pain, nausea, and vomiting. He had recently been diagnosed with KS by chromosomal analysis, while being investigated for eunucoid appearance and gynecomastia. He had not yet received testosterone treatment. On physical examination, no abdominal tenderness was found, and liver and spleen was nonpalpable. Normal bowel sounds were present. The laboratory studies revealed: white blood cell counts, 6,600/mm³; hemoglobin level, 14.6 g/dL; AST, 18 U/L (0-35); ALT, 9 U/L (0-45); GGT 20 U/L (2-22); ALP 111 U/L (30-120); amylase 40 U/L (28-100); total bilirubin 1,47 mg/dL (0,3-2); direct bilirubin 0,39 mg/dL (0-0,2); glucose 97 mg/dL (74-106); creatinine 0,64 mg/dL (0,39-0,7); albumine 4,72 g/dL (3,5-5,2); CRP 1 mg/L(0,01-5); FSH 52,62 mIU/mL(1,5-12,4); LH 23,11 mIU/mL (1,7-8,6); total testosterone 3,28 ng/ml(1,75-7,81); estradiol 21,59 pg/ml (7,6-43); TSH 0,69 µU/mL(0,51-4,3). Chest and abdominal radiographs were normal. An abdominal ultrasound scan showed 23 mm anechoic, saccular expansion in the left branch of the portal vein (Figure. 1). Monophasic, turbulent venous flow was detected in aneurysmal dilatation by doppler ultrasonography (Figure. 2). There were no features of thrombosis, portal hypertension, chronic liver disease, pancreatic mass or pancreatitis. No aneurysmal change was noted in the arterial tree and there were no pathological findings in other intraabdominal organs.
He was referred to perform oesophagogastroduodenoscopy and revealed pangastritis. His abdominal pain was attributed to gastritis.

DISCUSSION

We have reported coexistence of KS and PVA in this paper. Patients with KS are at risk for a number of certain health problems including autoimmune disorders, venous thromboembolic disease, psychiatric disorders and connective tissue diseases (8,9). Intracranial aneurysms have been reported in KS (10,11). However; an association between KS and PVA was not previously reported. García González JP et al., previously reported a case of KS with atrial septal aneurysm. In this report; the aneurysm was thought to be related with connective tissue abnormality (12). Kasten R et al. and Ishihara K et al. previously reported cases of KS with mixed connective tissue disease and stated that low androgen level is the cause of connective tissue abnormality in KS (9,13).

CONCLUSION

The association of KS with aneurysms is not clearly determined however connective tissue abnormalities secondary to androgen deficiency are accused. PVAs are usually asymptomatic. However in KS; it is important to early diagnosis of PVA to follow up the patient for the risk of portal vein thrombosis and the other complications including aneurysmal rupture, compression to adjacent organs and portal systemic shunts. More detailed studies are needed to determine the relationship between PVA and CS.

Conflict of interest
No conflict of interest was declared by the authors.

REFERENCES