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Case Report

Extrahepatic Portal Vein Obstruction Associated with Turner Syndrome

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Abstract

Turner syndrome (TS) is a sex chromosomal disorder. It is due to complete or partial absence of the second sex chromosome in females. Liver functions test abnormalities is common in Turner patients, but vascular hepatic involvement in Turner syndrome patients has been scarcely reported. 18-year-old girl presented with complaints of discomfort/dragging sensation in left upper abdomen for last one year and primary amenorrhea. She had short stature, short neck and low hairline with multiple naevi over face. Her TSH was 2.29 uIU/ml. FSH was 100.40 mIU/ml and LH was 18.40 mIU/ml. HBsAg and Anti HCV antibody was negative. Ultrasonography abdomen with Doppler revealed splenomegaly of 20cm with collaterals at hilum and lateral surface with formation of portal cavernoma. Agenesis of gonads with hypoplastic uterus was noted. Endoscopy revealed Grade I x III varices in esophagus. Echocardiography revealed bicuspid Aortic valve with mild Aortic stenosis. Karyotyping revealed 45, XO. Patient was started on beta-blockers and conjugated estrogen. *Conclusion:* This case report highlights that vascular hepatic involvement in Turner syndrome patients is not uncommon and it could be part of a more generalized vascular disorder.

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Introduction

Turner syndrome (TS) is a sex chromosomal condition that describes girls and women with common features that are caused by complete or partial absence of the second sex chromosome. TS occur in approximately 1 of every 2,000 live female births and approximately 10% of all miscarriages. It can cause a variety of medical and developmental problem including "short stature", "failure to begin puberty", "infertility", "heart defects and certain learning difficulties, skeletal abnormalities and kidney problems" [1]. Liver functions test abnormalities appears to be common in TS patients. The cause of liver test abnormalities in TS patients has been debated [2, 3]. Non cirrhotic Portal Fibrosis (NCPF) is a disorder of unknown etiology, clinically characterized by features of portal hypertension (PHT), moderate to massive splenomegaly, with or without hypersplenism, preserved liver functions, and patent hepatic and portal veins. Extra hepatic portal vein obstruction (EHPVO) is one of the causes of non cirrhotic portal hypertension. Thromboembolic complications occur more frequently in TS patients. These might contribute to the obliterative portal venopathy and to increased risk of deep venous and portal vein thrombosis inTS patients [4]. But to our knowledge, EHPVO with TS has not been reported in literature till date. Here we described a case of TS with EHPVO.

Case Series

A 18 -year- old girl who was admitted to the

Department of Medicine with complaints of discomfort/dragging sensation along with lump involving the left upper abdomen, noted for last one year. Detailed history revealed that she had primary amenorrhea. General physical examination revealed short stature (145 cm), with "short neck and low hairline with "multiple naevi over face with wide carrying angle" (Figure 1). Chest was broad shield like with widely spaced nipples. She had stage 1 for both breast and pubic hair development on Tanner staging. Abdominal examination revealed massive splenomegaly.

Investigations were ordered to evaluate for spleenomegaly as well as to confirm Turner syndrome. "Blood glucose fasting", "renal function test", "liver functions test and electrolytes were normal". Her hemoglobin was 7.2 gm/dl with TLC of 9000 cells/cumm and DLC of P72L25M21. PBF revealed microcytic picture with mild to moderate hypochromia. Her TSH was 2.29 uIU/ml. FSH was 100.40 mIU/ml and LH was 18.40 mIU/ml. Serum albumin and total proteins were 4.2g/dl and 7.4 g/dl respectively with prothrombin time of 14 seconds (control of 12 seconds). HBsAg and Anti HCV antibody was negative. The immunologic workup for autoimmune hepatitis (anti-smooth muscle anti-bodies; antinuclear factor, antimitochondrial antibodies) were negative, and immunoglobulin levels were normal. Serum ceruloplasmin was within normal limits. Kayser- Fleischer ring was not seen on slit lamp biomicroscopy.

Fig. 1: Picture of patient showing low set hair and multiple naevi over face

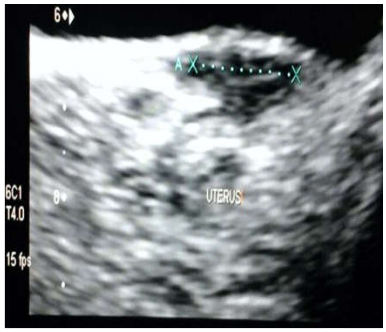

Fig. 2: USG image showing hypoplastic uterus

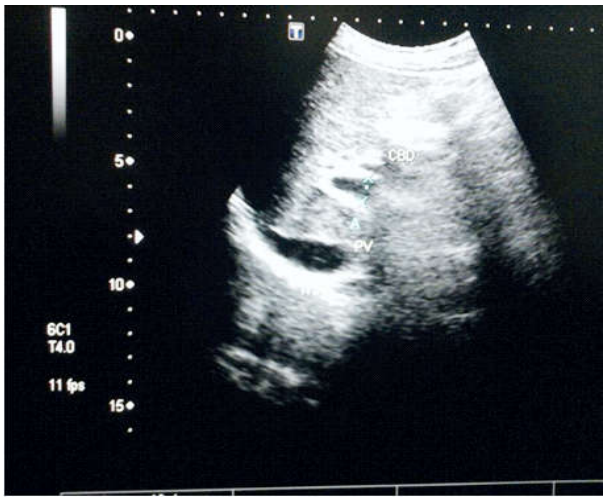

Fig. 3: Doppler image showing portal cavernoma

USG abdomen with Doppler showed splenomegaly of 20cm (grade 3) with collaterals at hilum and lateral surface. Intrahepatic branch of portal vein have been replaced by small tortuous vessels suggestive of portal cavernoma (Figure 4). Periportal sclerosis was also seen(Figure 3). Kidney ureters and bladder were normal. Agenesis of gonads with hypoplastic uterus was noted (Figure 2). Endoscopy revealed Grade I x III varices in esophagus, but stomach and duodenum were normal. Echocardio-graphy revealed bicuspid Aortic valve with mild Aortic stenosis. Karyotyping revealed 45, XO. A final diagnosis of Turner Syndrome with Extrahepatic portal vein obstruction was made in view of lack of features of hepatic dysfunction (both clinically and biochemically) in the presence of esophageal varices and portal cavernoma on Doppler ultrasonography. Patient was started on propranolol and conjugated estrogen and she is on regular follow up.

Discussion

Liver involvement appears to be frequent in TS patients. The prevalence of liver test abnormalities ranges from 20% to 80% depending on the patient's age, with the highest values in the oldest patients. The cause of liver test abnormalities in TS patients is still not clear [2, 3].

NCPF is a disorder of unidentified etiology,

variously called as Idiopathic PHT (IPH), hepatoportal sclerosis and obliterative venopathy [5,6]. Infections and prothrombotic states are commonly incriminated etiological factors. EHPVO) is an important cause of NCPF, especially in Third World countries. The etiology and clinical presentation are different in children and adults. Although the liver may appear normal, functional compromise develops in the long term. There is cavernomatous transformation of portal vein.

Liver biopsy is not essential for the diagnosis of EHPVO unless the underlying chronic liver disease is suspected, but it is indicated in NCPF/IPH to exclude cirrhosis and other etiologies of PHT [7].

Venous malformations, such as agenesia or hypoplasia of the portal venous system, may occur in TS women [8,9]. A case of presinusoidal portal hypertension was reported which was caused by congenital hypoplasia of the intrahepatic portal system [10].

Thromboembolic complications occur more frequently in TS patients. In one study a considerable proportion of individual TS have high levels of vWF, factor VIII, fibrinogen and CRP. There is also an increased frequency of the Leiden mutation, with important associations with carotid intimal medial thickness (CIMT) and blood pressure, suggesting that a subset of TS patients may have an unfavorable haemostatic balance, which may contribute to the increased risk of premature ischemic heart disease and possibly increase the risk of deep venous and portal vein thrombosis [11-13]. In addition, vascular abnormalities e.g. aortic coarctation, bicuspid aortic valve, cerebral vessel aneurysm, and gastrointestinal telangiectasia are common in TS [14].

Conclusion

There are convincing evidences that vascular hepatic involvement in Turner syndrome patients could be part of a more general vascular disorder, likely of congenital origin, involving vessels of different sizes, types and locations

Conflict of Interest: None

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