Non-cirrhotic Portal Hypertension in Turner's Syndrome: A Case Report

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Introduction

- ✓ Turner's syndrome is one the most common chromosomal aneuploidies in humans.
- ✓ Although it can affect multiple organs, involvement of gastrointestinal (GI) organs is rare in this condition.
- ✓ Herein, we report a case of non-cirrhotic portal hypertension in a young adult with Turner's syndrome.

Case Presentation

History of Present Illness

A 20-year-old female with history of Turner's syndrome presented to a well-being visit, complaining of fatigue without any other symptom.

Medical History

Turner's syndrome confirmed by karyotype shortly after birth, premature ovarian failure, and mild mitral regurgitation

Surgical History

No history of prior surgery

Medication History

Estradiol and medroxyprogesterone

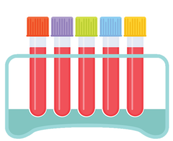
Social History

No smoking, no consumption of alcoholic beverages, and no creational drug use.

Physical Examination

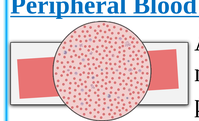
Stable vital signs, pale conjunctiva, widespread chest, cardiac 3/6 systolic murmur, and few healing ecchymosis.

Laboratory Findings



Pancytopenia with leukocyte count of 1.6 × 1000 cells/μL, hemoglobin of 7.1 g/dL, and platelet count of 74.0 × 1000 cells/μL with iron studies suggestive of iron deficiency anemia. Her AST, ALT, and bilirubin were in normal range with slightly elevated alkaline phosphatase (115 IU/L). Tests for viral hepatitis, auto-immune hepatitis, primary biliary cholangitis, Wilson's disease, Alpha 1 antitrypsin deficiency and hemochromatosis were negative.

Peripheral Blood smear



A variety of red cell shape abnormalities, including anisocytosis, microcytosis, ovalocytosis, tear drop cells, hypochromasia and poikilocytosis.

Bone Marrow Biopsy

Normal cellular marrow with minor dyserythropoiesis and decreased iron storage.

Case Presentation

Genetic Studies

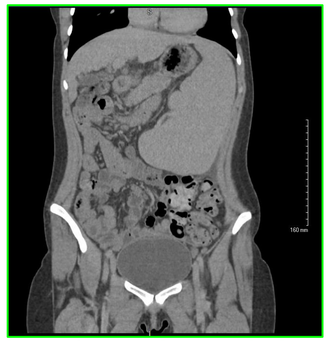
NeoGenomics and extended FISH analysis were negative for myelodysplastic syndrome

Imaging

Computed tomography of the chest, abdomen and pelvis with and without contrast shows severe splenomegaly, tiny varices in the anterior mediastinum along the gastrohepatic ligament, no portal vein thrombosis.

Abdominal ultrasound revealed severe diffuse heterogeneity of the liver parenchyma with a slightly nodular hepatic contour.

Fibroscan showed controlled attenuation parameter of 245 consistent with S1 steatosis and 11.8 kilopascals consistent with advanced fibrosis F3.



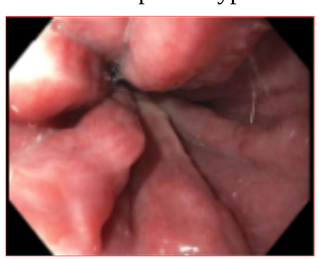
Coronal view of computed tomography demonstrates enlarged spleen measuring 20.2 ×11.6 × 20.4 cm

Liver Biopsy

Focal mature fibrous expansion of some portal tracts, with delicate bridging fibrosis with subtle lobular parenchymal changes, suggestive of portosinusoidal vascular disease with no evidence of cirrhosis

Upper GI Endoscopy

Grade II esophageal varices and portal hypertensive gastropathy.



Grade II esophageal varices

Treatment and Follow up

➢She received iron supplementation which improved her anemia.

➢She was started on propranolol for gastroesophageal varices and her heart rate is currently well-controlled. Follow-up abdominal ultrasound is negative for further changes in the liver size, morphology, or masses.

Discussion

- In individuals with Turner's syndrome, hepatic involvement has been reported at both biochemical and structural levels.
- Despite the common involvement of the liver in Turner's syndrome, portal hypertension is rare in this condition. Absence of liver cirrhosis in cases with portal hypertension in individuals with Turner's syndrome is even more rare.
- In a case-series on individuals with Turner's syndrome who underwent liver biopsy, four out of 27 cases, had evidence of portal hypertension (including two cases with cirrhosis) [1].
- To the best of our knowledge, there are only two case reports on the presence of portal hypertension due to portosinusoidal vascular disease in individuals with Turner's syndrome:
  - A three-year-old patient with Turner's syndrome who presented with upper GI bleeding due to portal hypertension with liver biopsy revealing intra-hepatic venous and arterial derangements with obliterated portal veins [2]
  - A nine-year-old girl with Turner's syndrome who presented with a massive GI bleeding due to esophageal varices with liver biopsy showing mild dilation of sinusoids with normal liver architecture [3]
- Vascular anomalies including hemangioma, vascular ectasia, and telangiectasia have been reported throughout the GI tract in Turner's syndrome and may share similar underlying mechanisms with portal vein irregularities in this condition.

Conclusion

- ❖ Turner's syndrome might be associated with non-cirrhotic portal hypertension.
- ❖ High index of clinical suspicion and serial assessments of the liver function followed by a thorough investigation in cases with functional or biochemical derangement, can lead to early diagnosis of portal hypertension, reducing the burden of complications of portal hypertension.

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References:

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