Wits Journal of Clinical Medicine, 2025, 7(1) 33–36 http://dx.doi.org/10.18772/26180197.2025.v7n1a7

Case Report

Idiopathic Non-Cirrhotic Portal Hypertension: A case report

Munir Ageal, Abd Elrhamn Ahmed, Nieka Calabria Gordon*

Department of Internal Medicine, Klerksdorp-Tshepong Hospital Complex, University of the Witwatersrand, Klerksdorp, South Africa

Corresponding Author: *nieka.gordon@icloud.com*

ABSTRACT

Idiopathic non-cirrhotic portal hypertension is a rare diagnosis with an unknown aetiology. It is characterised by increased intrahepatic portal pressures in the absence of underlying liver disease. We present a unique case of a 32-year-old male patient who presented with a right hydrocele and an incidental finding of pancytopenia, massive splenomegaly, and features of portal hypertension. After known causes of portal hypertension were excluded, a liver biopsy was performed. Based on the liver biopsy findings, which demonstrated fibrotic portal tracts in the absence of cirrhosis and exclusion of other known causes of portal hypertension, our patient was diagnosed with idiopathic non-cirrhotic portal hypertension. Due to the persistent presence of hypersplenism and recurrent episodes of oesophageal variceal bleeding, our patient underwent a splenectomy, after which his features of portal hypertension resolved.

INTRODUCTION

Idiopathic non-cirrhotic portal hypertension (INCPH) is a rare disorder of intrahepatic portal hypertension in the absence of liver cirrhosis, other identifiable intrinsic liver diseases, splanchnic vein thrombosis, post-hepatic causes, including Budd Chiari syndrome, inferior vena cava webs, as well as cardiac causes such as constrictive pericarditis and congestive cardiac failure. It is characterized by portal hypertension, splenomegaly, hypersplenism, and pancytopenia. The underlying pathogenesis remains unclear, although many theories

Idiopathic non-cirrhotic portal hypertension is known as non-cirrhotic portal fibrosis, hepatoportal sclerosis, incomplete septal cirrhosis, obliterative portal venopathy, partial nodular transformation, and nodular regenerative hyperplasia.(1,2)

Idiopathic non-cirrhotic portal hypertension has a worldwide distribution but is more prevalent in developing countries, particularly in Asia, notably India, Nepal, and Japan. In India, INCPH prevalence rates as high as 23% have been reported, while in the Western populations, much lower rates (3%–5%) have been reported.(1–3)

The presence of portal hypertension is necessary for the diagnosis of INCPH. Most patients are diagnosed with INCPH through investigation of thrombocytopenia and splenomegaly. However, some patients may present with variceal bleeding, which is well tolerated due to preserved liver function.(1,2)

The underlying pathogenesis remains unclear, although many theories have been proposed. Coupled with the absence of a specific positive diagnostic test, the diagnosis of INCPH is challenging. Patients are frequently misdiagnosed as having liver cirrhosis. INCPH is a diagnosis of exclusion. An extensive diagnostic workup, including laboratory tests, hepatic and splenic imaging studies to rule out veno-occlusive causes, and a liver biopsy is recommended to rule out underlying intrinsic liver diseases.(1–3)

Our case demonstrates the necessity of an extensive diagnostic workup and complications associated with INCPH.

CASE PRESENTATION

A 32-year-old male was referred to our institution with pancytopenia and massive splenomegaly. This was an incidental finding after he had presented with a right hydrocele. He had no previous medical history or history of childhood illness. He had no prescription or over-the-counter drug history and no history of recreational drug use. He also had no history of alcohol use and no family history of liver disease.

On presentation, he reported a one-year history of fatigue, dizziness, headache, and shortness of breath. He also reported having had one episode of hematemesis the year before, as well as two episodes of melena stools. Physical examination revealed pallor, bilateral pitting oedema, a pan systolic murmur over the mitral area, and massive splenomegaly.

His laboratory tests revealed haemoglobin levels of 2.7 g/dl, leucocyte count of 0.49/μL, and platelet count of 36/μL (Table 1). The peripheral blood morphology showed

Table 1: Laboratory values at the time of first hospitalization

Parameter	Patient result	Normal value
White cell count (WBC)	0.49	3.92-10.40 x 10^9 / L
Haemoglobin (Hb%)	2.7	13.4-17.5 g/ dL
Mean corpuscular volume (MCV)	64.8	83.1-101.6 fL
Ferritin	3	30-400 mcg/ml
Reticulocyte production index (RPI)	0.2	1-2
Platelets	36	171-388 x 10^9 /L
Lactate dehydrogenase (LDH)	219	140-280 U/L
Total protein	71	60-78 g/L
Albumin	43	35-52 g/L
Total bilirubin	18	5-21 umol/L
Conjugated bilirubin	6	0-3 umol/L
Alanine transaminase (ALT)	28	10-40 U/L
Aspartate aminotransferase (AST)	40	15-40 U/L
Alkaline phosphatase (ALP)	108	35-128 U/L
Gamma-glutamyl transferase (GGT)	84	< 68 U/L
International normalized ratio (INR)	1.20	

anisocytosis, hypochromia, and teardrop cells. Platelet morphology was normal. His liver function, renal function, haemostatic function, and serum electrolyte were all within normal range. Hepatitis B and C serology markers were negative. Comprehensive autoimmune screening serology tests were all negative. Serum ACE was normal, and Schistosomiasis serology as well as HIV ELISA test were also all negative. A thrombophilia and JAK 2 mutation screen was found to be within normal limits.

Other tests, including Cytomegalovirus and Epstein-Barr virus immunoglobulin G (IgG), were positive, indicating past infections. Bone marrow aspirate and trephine biopsy showed no evidence of bone marrow infiltration but demonstrated variable cellular bone marrow with trilineage haematopoiesis, absent iron stores with reduced sideroblasts, indicating the presence of iron deficiency anaemia, and suggested that these features were most likely secondary to hypersplenism. Iron was administered through oral and parenteral routes.

Due to the unavailability of requisite expertise, hepatic venous pressure gradient measurement could not be performed. Abdominal ultrasound showed a dilated portal vein and a markedly enlarged spleen. An abdominal CT scan revealed a normal liver size and contour with homogenous parenchymal enhancement. The hepatic veins and inferior vena cava were patent. However, the portal vein was markedly dilated and tortuous, as was the splenic vein. The terminal splenic vein showed terminal narrowing (21.6 mm) with an enlarged calibre proximally (31.1 mm). There was no portal or splenic vein thrombosis. The spleen was grossly enlarged, spanning 30.8 cm in craniocaudal diameter extending into the left iliac fossa. There were no focal splenic lesions. Splenic, periportal, and distal oesophageal varices were noted. The cardiac echocardiogram was normal.

Gastroscopy showed grade II–III oesophageal varices, with no stigmata of acute bleeding, and the varices were banded.

As no cause of the portal hypertension could be found, a liver biopsy was performed, which was reported as "benign hepatic parenchyma with features of portal tract fibrosis as well as bands of fibrous connective tissue which extends between the portal tracts and terminal hepatic venules with focal areas of bridging fibrosis" (Figure 1).

Based on the clinical findings of portal hypertension as evidenced by splenomegaly, hypersplenism, presence of gastroesophageal varices, exclusion of known causes of chronic liver disease causing portal hypertension, presence of patent portal and hepatic veins on imaging and the absence of cirrhosis on liver biopsy, our patient was diagnosed with INCPH.

The patient continued to experience recurrent episodes of oesophageal variceal bleeding, which was initially managed using banding and a beta blocker, as these methods

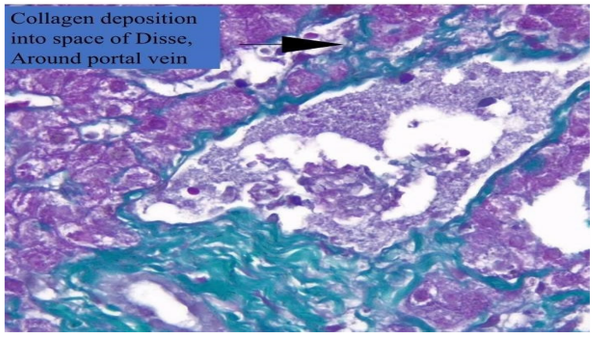

Figure 1: Liver biopsy histology (using Masson's trichrome special stain) showing a dilated portal vein displaying marked fibrous intimal thickening with extension of fibrous tissue into the space of Disse (perisinusoidal space around hepatocytes)

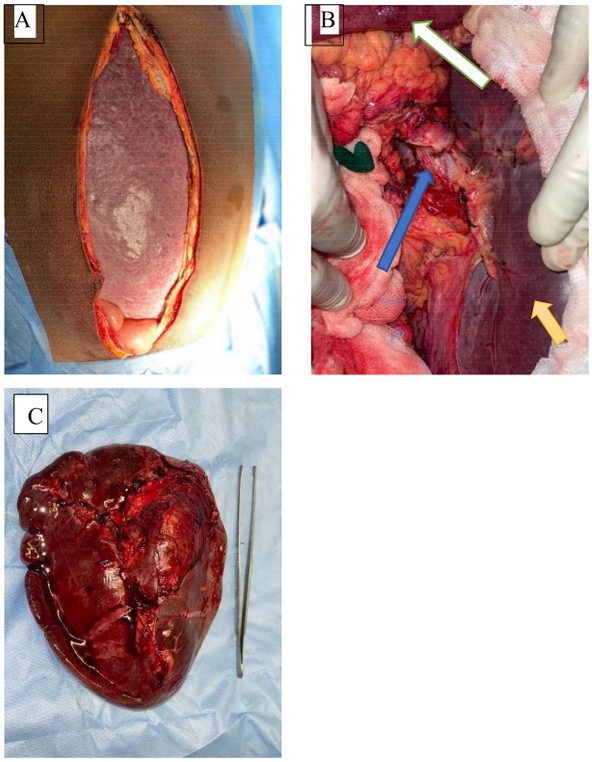

Figure 2: Intraoperative images of the spleen: (A) Massive spleen upon opening the abdomen. (B) Liver's irregular edge (white arrow), huge splenic vein (blue arrow,) and the spleen (yellow arrow). (C) showing the postoperative spleen

are less invasive and pose low risk compared to transjugular intrahepatic portosystemic shunt (TIPS). In the presence of splenomegaly and severe hypersplenism, a decision was made to perform a splenectomy. Prior to splenectomy, our patient received appropriate vaccinations.

Intraoperatively, the liver surface was irregular, with no apparent nodules (Figure 2). The splenic vein was massively dilated, and multiple collateral veins were noted around the splenic hilum and hepatoduodenal ligament. The spleen was enlarged, and a splenule, measuring a 7×5 cm, was also found. A splenectomy was performed. Biopsy of the spleen was consistent with features of dilated and congested parenchymal sinuses, veins with variable fibrosis, and Gamna-Gandy bodies.

The patient had an uneventful postoperative course and was discharged on the 7th postoperative day. Subsequently, his features of hypersplenism resolved, and a reactive thrombocytosis secondary to splenectomy was recorded.

DISCUSSION

Idiopathic non-cirrhotic portal hypertension is a rare disease, and its diagnosis is made on exclusion. For the diagnosis to be made, the following criteria should be met: presence of unequivocal signs of portal hypertension (e.g., hepatic venous pressure gradient more than 5 mmHg, gastroesophageal varices, ascites and/or splenomegaly); absence of cirrhosis; advanced fibrosis or other causes of chronic liver disease that can cause portal hypertension; absence of thrombosis of the hepatic veins or the portal vein and a patent inferior vena cava in the absence of cardiac disease and pulmonary hypertension.(2)

The aetiology and pathogenesis of INCPH remain unclear. However, many theories have been proposed. Associations with immunological disorders, acute or chronic infections, medications and toxins, genetic disorders, and thrombophilia have been postulated. The aetiology may also be multi-factorial. The difference in worldwide prevalence may be explained by the influence of genetic predisposition along with geographic associations.(2,3)

Most patients present asymptomatically with signs of portal hypertension. However, our patient was referred to us with an incidental finding of pancytopenia and splenomegaly. Later in the course of the disease, he experienced recurrent episodes of oesphageal variceal bleeding and signs of hypersplenism. Current data suggests that the mortality of variceal haemorrhage in INCPH is lower compared to those with liver cirrhosis. The 10-year mortality rate in patients with INCPH is reported to be 18%–44%.(1–3)

Currently, there is a lack of data on treatment and prophylaxis of variceal bleeding in patients with INCPH. The consensus is to follow the guidelines on prophylaxis and managing cirrhotic variceal bleeding. Patients with INCPH tend to have massive splenomegaly, which also contributes to the presence of portal hypertension. It has been shown that splenectomy and partial splenic embolization decrease portal hypertension, thus partially alleviating the problem with splenic arterial inflow but not the resistance to blood flow through the liver. Therefore, shunt procedures are preferred. Liver transplantation should be considered in patients with medically unmanageable portal hypertension, hepatopulmonary syndrome, hepatic encephalopathy, and progressive hepatic failure. However, these are unusual complications in patients with isolated INCPH.(2,4)

Our patient is currently being managed on carvedilol 6.25 mg twice daily and aspirin 75 mg daily for reactive thrombocytosis. He is being periodically reviewed at our Gastroenterology clinic. Following the splenectomy, he has not had an episode of variceal bleeding, and a follow-up endoscopy is planned.

REFERENCES

1. Fiordaliso M, Marincola G, Pala B, et al. A narrative review on non-cirrhotic portal hypertension: not all portal hypertensions mean cirrhosis. Diagnostics. 2023; 13(20):3263.
2. Sith Siramolpiwat, Seijo S. Idiopathic non-cirrhotic portal hypertension. J Rare Dis Res Treat. 2016; 1(3):10–16.
3. Schouten JNL, Garcia-Pagan JC, Valla DC, Janssen HLA. Idiopathic noncirrhotic portal hypertension. Hepatology. 2011; 54(3):1071–1081.
4. Khanna R, Sarin SK. Non-cirrhotic portal hypertension – diagnosis and management. J Hepatol. 2014; 60(2):421–441.