CASE REPORT / OLGU SUNUMU

DOI: 10.4274/atfm.galenos.2021.87587
Ankara Üniversitesi Tıp Fakültesi Mecmuası 2021;74(Suppl 1):112-115

SURGICAL SCIENCES / CERRAHİ TIP BİLİMLERİ

# Congenital Absence of Portal Vein Presenting with Macroscopic Hematuria: A Case Report with a Brief Review of the Literature

## Makroskopik Hematüri ile Bulgu Veren Konjenital Portal Ven Agenezisi: Olgu Sunumu ve Literatür İncelemesi

**Cahit Hüseyin Yılmaz, Can Avni Karaca**

*İzmir Ekonomi University Faculty of Medicine, Department of General Surgery, İzmir, Turkey*

## Abstract

Congenital absence of the portal vein is a rare clinical entity in which portal blood flow is partially or totally diverted to the systemic circulation. This condition causes a myriad of clinical symptoms and is associated with other congenital abnormalities mostly of cardiac and vascular origin. A 14-year-old male patient with type IB Abernethy malformation who presented with macroscopic hematuria was treated with live donor liver transplantation. The medical recordings of the patient were retrospectively reviewed, and the case was presented with a brief review of the literature. The patient had associating nut-cracker syndrome and liver masses that were revealed to be focal nodular hyperplasia. The patient is doing well in the postoperative course. Liver transplantation offers a safe and radical treatment for this very rare anatomic malformation. It not only corrects aberrant portal flow but also prevents other life-threatening complications such as hepatopulmonary syndrome and possible liver malignancies.

**Key Words:** Abernethy Syndrome, Congenital Absence of Portal Vein, Nutcracker Syndrome, Portosystemic Shunt

## Öz

Portal venin konjenital yokluğu, portal kan akışının kısmen veya tamamen sistemik dolaşıma yönlendirildiği nadir bir klinik tablodur. Bu durum, sayısız klinik semptomlara neden olur ve çoğunlukla kardiyak ve vasküler kaynaklı diğer konjenital anormalliklerle ilişkilidir. Burada makroskopik hematüri ile başvuran tip IB Abernethy malformasyonlu 14 yaşında erkek hasta sunulmuş olup, olgu canlı donör karaciğer transplantasyonu ile tedavi edilmiştir. Hastanın tıbbi kayıtları geriye dönük olarak gözden geçirilerek olgu, literatürün kısa bir gözden geçirmesi ile birlikte sunulmuştur. Hastada fokal nodüler hiperplaziye ait karaciğer kitleleri ile birlikte nutcracker sendromu da mevcuttu. Hasta postoperatif dönemde sorunsuz seyretmiştir. Karaciğer nakli, bu çok nadir görülen anatomik malformasyon için güvenli ve radikal bir tedavi sunar. Sadece anormal portal akışı düzeltmekle kalmaz, aynı zamanda hepatopulmoner sendrom ve diğer olası karaciğer maligniteleri gibi diğer yaşamı tehdit eden komplikasyonları da önler.

**Anahtar Kelimeler:** Abernethy Sendromu, Portal Venin Konjenital Agenezisi, Nutcracker Sendromu, Portosistemik Şant

## Introduction

Congenital absence of portal vein (CAPV) is a rare clinical entity which was first described by John Abernethy in 1793 based on a postmortem study (1). To the best of our knowledge, there are 132 cases reported. The malformation diverts portal blood from the liver either totally or partially causing a myriad of clinical symptoms and is associated with other congenital abnormalities mostly of cardiac and vascular origin such as atrial septal defect, patent foramen ovale, ventricular septal defect and patent ductus arteriosus (2). Among these clinical manifestations hepatic nodule formation due to abnormal hepatic vascular inflow is remarkable (2,3).

**Address for Correspondence/Yazışma Adresi:** Can Avni Karaca
İzmir Ekonomi University Faculty of Medicine, Department of General Surgery, İzmir, Turkey
Phone: +90 533 483 49 58 E-mail: can.karaca@ieu.edu.tr **ORCID ID:** orcid.org/0000-0003-4930-6222
**Received/Geliş Tarihi:** 18.01.2021 **Accepted/Kabul Tarihi:** 30.03.2021

The condition is divided into two types where type I represents a total portosystemic shunt. In contrast, there is a certain amount of portal perfusion of liver is preserved in type II and the shunt is partial (2,4).

Liver transplantation (LT) is indicated for the patients with type I malformation, more than 60% shunt ratio, hyperammonemia, encephalopathy, hepatopulmonary syndrome (HPS) or hepatic tumors (5-7).

This is a representation of the case of a 14-year-old male patient with type IB Abernethy malformation presented with macroscopic hematuria who was treated with LT; with a brief review of the literature.

## Case Report

### The Patient

A-14-years old male patient sought medical help due to diarrhea persisting for 4 days and accompanying macroscopic hematuria for the last 24 hours. On physical examination, the patient showed phenotypical stature suggestive of Marphan's syndrome. Spleen was palpable 2 cm below the rib cage and his vitals were within normal range. No localized or systemic edema was present.

The patient had a history of acute rheumatic fever for which he was regularly followed up. There was no prior history of any other chronic condition or surgical intervention. Family history was also insignificant.

The patient was studied for possible causes of hematuria and blood tests for several auto immune antibodies were negative. There was only a slight decrease in complement 3 and anti streptolysin O was above 166 Todd units. Rheumatoid factor and immune globulins were all within normal range.

An abdominal ultrasound to evaluate the kidneys revealed a total portosystemic shunt between the confluence of portal vein and left renal vein (LRV). The LRV crossing between superior mesenteric artery and aorta, narrowed down to 2.5 mm in diameter resulting in nutcracker syndrome. Further evaluation with ultrasound also revealed the absence of portal flow into the liver.

With these additional findings, the patient was further studied with abdominal computed tomography (CT) and magnetic resonance imaging (MRI) (Figure 1). Absence of intrahepatic branches of portal vein was discovered with CT while MRI revealed multiple nodular lesions within the liver parenchyma suggestive of focal nodular hyperplasia (FNH) or hepatic adenoma (HA). Core needle biopsy from one of the nodules was consistent with HA however FNH could not be ruled out due to inadequate tissue sampling.

The patient was referred to our center and he was planned for LT owing to vascular complications of Abernethy syndrome and hepatic tumors.

Informed consent of the patient and his legal guardians was obtained both for the LT surgery and later for this case report.

### Surgery

The patient underwent a liver transplant from a deceased donor. Upon dissection, there was no portal vein within the hepatic hilum (Figure 2). Superior mesenteric vein (SMV) followed a normal course and joined splenic vein (SV) right under the pancreas. SMV was prepared for an anastomosis and an iliac vessel conduit was used for portal vascular inflow between the SMV and the donor liver's main portal vein. SMV was preferred for the anastomosis as this vessel was easily accessible and for its potential to drain the renal vein as well as the portal system thus resolving the nutcracker syndrome. The liver was re-perfused without any problems and the rest of the procedure was carried out in standard fashion.

Postoperative course was uneventful and the patient is alive and well without any complications after 22 months of follow-up. Recent CT demonstrated patent portal flow through the liver.

### Histologic Examination

The explanted liver weighed 930 grams. Histologic examination revealed multiple FNH nodules of which, largest measured 3 cm in diameter (Figure 3). Portal venules were absent and bile ducts showed signs of atrophy 70% of the examined portal areas (Figure 4). There was also intracellular cholestasis in the hepatocytes surrounding central veins.

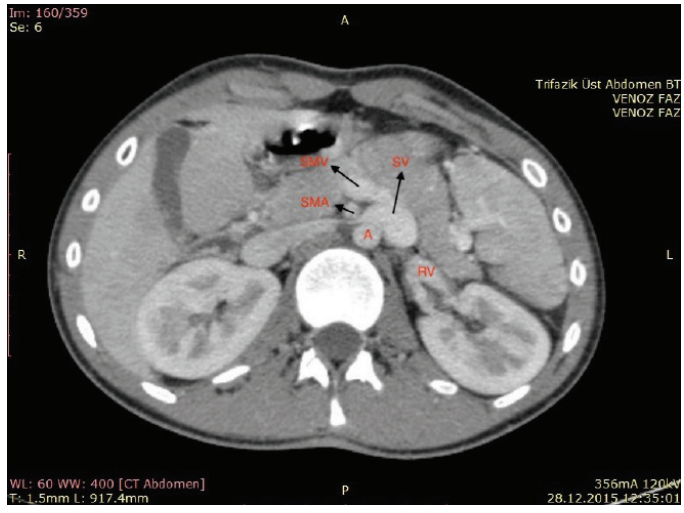


**Figure 1:** Preoperative CT imaging of the shunt vessel. Note that the SMA crosses LRV causing nutcracker syndrome

A: Aorta, RV: Renal vein, CT: Computed tomography, SMA: Superior mesenteric artery, LRV: Left renal vein

## Discussion

Centuries after the first report by Abernethy, Morgan and Superina (4) further defined the condition and suggested a classification that divides the entity into two groups (1). Type I involves a complete portosystemic shunting where portal flow is diverted outside the liver. This type is further divided into two subgroups; where separate drainage of SV and SMV to a systemic vein is defined as type Ia and their convergence just before draining as type Ib. However, in type II there is limited portal flow into the liver and portosystemic shunting is partial. There are reported female predominance for type I malformations (4,8). This case is among the rare male patients with type I malformation.

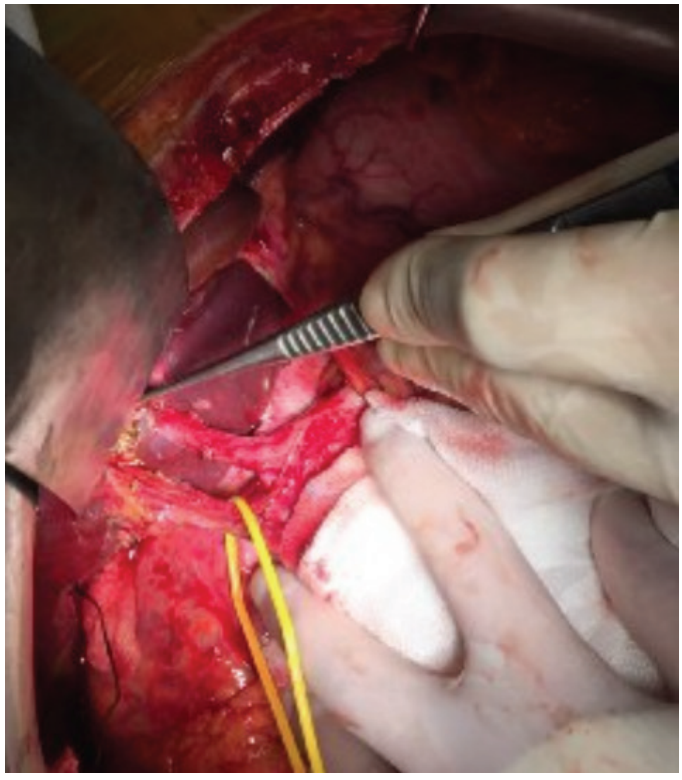

**Figure 2:** Perioperative picture of hepatic hilum. Portal vein is absent. Common bile duct is hanged with a yellow tape and hepatic artery is held with a forceps

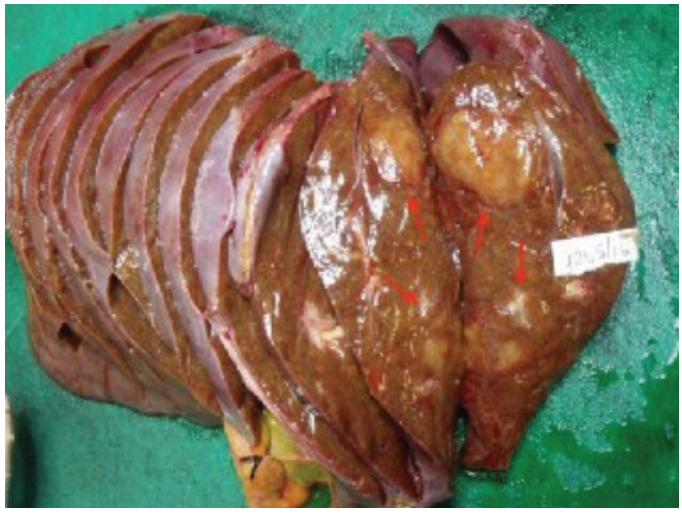

**Figure 3:** Macroscopic image of the explanted liver. Histologic examinaton of the whiteish masses (arrows) was consistent with FNH

FNH: Focal nodular hyperplasia

Typically, Abernethy malformation is characterized by the absence of the venules in the portal area (2,9). Accordingly, complete absence of venules and bile duct atrophy in the portal areas was observed in histological examination of the explanted liver in this case.

There is a variety of clinical manifestations and accompanying congenital defects reported for CAPV as mentioned in the introduction part (2). In addition to type Ib malformation, the patient suffered from hematuria due to the nutcracker syndrome of LRV which was not previously reported.

In this case the portal inflow was reconstructed via a vascular conduit between the SMV and the donor liver's portal vein. Diversion of the splanchnic flow through this new route towards the liver also relieved the venous renal hypertension caused by the nutcracker syndrome.

As the portal flow is totally or partially diverted from the liver; CAPV genuinely affects liver. Expectedly, liver volumes of these individuals are usually low with preserved regeneration capacity (2,10). Hepatic arterial flow is usually increased in compensation for the absence or lack of portal flow 8/16/21 5:53:00 PM.

Due to the absence or lack of portal hepatotrophic factors, patients with CAPV suffer from abnormal hepatic development and regeneration that results in liver nodules such as FNH,

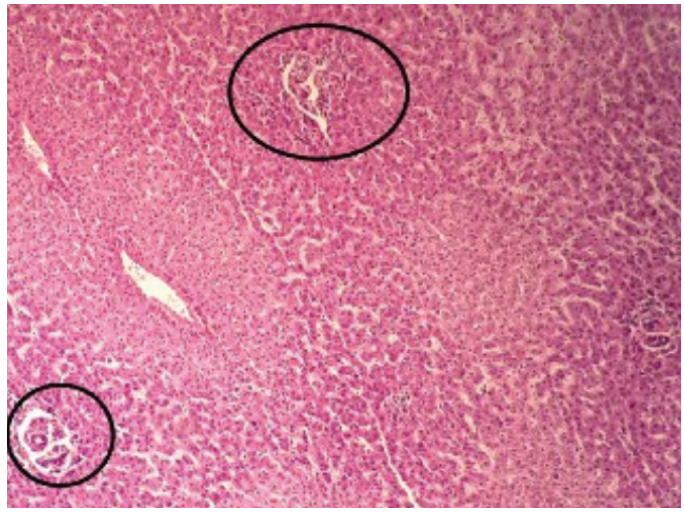

**Figure 4:** Atrophy in the portal areas. Note that the portal venules are absent

nodular regenerative hyperplasia, HA and even hepatocellular carcinoma (2,11-13). However, circulatory problems cannot be blamed alone as selective occlusion of portal vein has been proven insufficient for nodule formation (2,3). An analysis of 101 patients with Abernethy malformation, showed that nearly half (48.5%) of the patients had nodular liver lesions (11). Most frequent of these lesions was reported as FNH (36.7%). However; FNH was relatively rare (8.2%) among male patients (11). Although being male; our patient had multiple FNH nodules of which largest measuring 3 cm in diameter.

Hepatic encephalopathy is another concern regarding portosystemic shunts, however it is rarely seen among patients with CAPV (2). Contrarily, significantly low levels of ammonia are measured in the SMV of these patients suggesting a compensatory mechanism (2,9,14).

HPS is a serious complication reported to be associated with Abernethy malformation (2,5). LT successfully treats this life-threatening complication along with liver disease (5).

Treatment of Abernethy malformation depends both on the type of the shunt and clinical findings. Especially for the patients with type II, occlusion of the shunt may be considered (2) however, LT remains the sole option for the patients with type I malformation, HPS or hepatic tumors (2,5-7,15). Auxiliary partial orthotropic LT is also reported to be successful in patients with Abernethy syndrome (16,17). However, it lacks the ability to eliminate present or future tumors in the native liver. Our case was treated with LT as he had multiple liver tumors associated with type Ib malformation. There are 20 reported cases of LT in the setting of Abernethy and it offers a safe and feasible treatment option for this rare condition. This is the first reported case of Abernethy type I malformation associated with nutcracker syndrome treated successfully with LT.

In conclusion, LT offers a safe and radical treatment for this very rare anatomic malformation. It not only corrects aberrant portal flow but also prevents other life-threatening complications such as HPS and possible liver malignancies.

### Ethics

**Informed Consent:** Informed consent of the patient and his legal guardians was obtained both for the LT surgery and later for this case report.

**Peer-reviewed:** Externally peer-reviewed.

### Authorship Contributions

Surgical and Medical Practices: C.H.Y., C.A.K., Concept: C.H.Y., Design: C.H.Y., Data Collection or Processing: C.H.Y., C.A.K., Analysis or Interpretation: C.A.K., Literature Search: C.A.K., Writing: C.H.Y., C.A.K.

**Conflict of Interest:** No conflict of interest was declared by the authors.

**Financial Disclosure:** The authors declared that this study received no financial support.

## References

1. Abernethy J, Banks J. IX. Account of two instances of uncommon formation, in the viscera of the human body. Philos. Trans. R. Soc. 1793;83:59.
2. Hu GH, Shen LG, Yang J, et al. Insight into congenital absence of the portal vein: is it rare? World J Gastroenterol. 2008;14:5969-5979.
3. Kudo M. Hepatic nodular lesions caused by abnormal hepatic circulation: etiological and clinical aspects. J Gastroenterol. 2003;38:308-310.
4. Morgan G, Superina R. Congenital absence of the portal vein: two cases and a proposed classification system for portasystemic vascular anomalies. J Pediatr Surg. 1994;29:1239-1241.
5. Yilmaz C, Onen Z, Farajov R, et al. Live donor liver transplantation for a child presented with severe hepatopulmonary syndrome and nodular liver lesions due to Abernethy malformation. Pediatr Transplant. 2017;21.
6. Sanada Y, Mizuta K, Kawano Y, et al. Living donor liver transplantation for congenital absence of the portal vein. Transplant Proc. 2009;41:4214-4219.
7. Ghuman SS, Gupta S, Buxi TB, et al. The Abernethy malformation-myriad imaging manifestations of a single entity. Indian J Radiol Imaging. 2016;26:364-372.
8. Kwapisz L, Wells MM, AlJudaibi B. Abernethy malformation: congenital absence of the portal vein. Can J Gastroenterol Hepatol. 2014;28:587-588.
9. Komatsu S, Nagino M, Hayakawa N, et al. Congenital absence of portal venous system associated with a large inferior mesenteric-caval shunt: a case report. Hepatogastroenterology. 1995;42:286-290.
10. Shinkai M, Ohhama Y, Nishi T, et al. Congenital absence of the portal vein and role of liver transplantation in children. J Pediatr Surg. 2001;36:1026-1031.
11. Hao Y, Hong X, Zhao X. Congenital absence of the portal vein associated with focal nodular hyperplasia of the liver and congenital heart disease (Abernethy malformation): A case report and literature review. Oncol Lett. 2015;9:695-700.
12. Happaerts S, Foucault A, Billiard JS, et al. Combined hepatocellular-cholangiocarcinoma in a patient with Abernethy malformation and tetralogy of Fallot: A case report. Hepatology. 2016;64:1800-1802.
13. Beal EW, Foley K, Washburn K, et al. Congenital Absence of the Portal Vein. Am Surg. 2020:3134820960064.
14. Kamiya S, Taniguchi I, Yamamoto T, et al. Analysis of intestinal flora of a patient with congenital absence of the portal vein. FEMS Immunol Med Microbiol. 1993;7:73-80.
15. Li P, Hidaka M, Hamada T, et al. Living donor liver transplantation for congenital absence of portal vein in portal venous reconstruction with a great saphenous vein graft. Surg Case Rep. 2020;6:153.
16. Emre S, Arnon R, Cohen E, et al. Resolution of hepatopulmonary syndrome after auxiliary partial orthotopic liver transplantation in Abernethy malformation. A case report. Liver Transpl. 2007;13:1662-1668.
17. Matsuura T, Soejima Y, Taguchi T. Auxiliary partial orthotopic living donor liver transplantation with a small-for-size graft for congenital absence of the portal vein. Liver Transpl. 2010;16:1437-1439.