A RARE CASE OF CONGENITAL PORTOSYSTEMIC SHUNT (ABERNETHY MALFORMATION TYPE II) IN A CHILD: DIAGNOSIS, ENDOVASCULAR INTERVENTIONAL STRATEGY, AND CLINICAL FOLLOW-UP

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Abstract

Congenital portosystemic shunt, also known as Abernethy malformation, is a rare vascular anomaly in which portal venous blood partially or completely bypasses the liver and enters the systemic circulation. This condition can lead to severe complications such as hepatopulmonary syndrome, hepatic encephalopathy, hypoxemia, and hyperammonemia. This article presents the first clinically confirmed and successfully treated case of Abernethy type II malformation in a child in Kazakhstan, who developed severe hepatopulmonary syndrome. A detailed description is provided of the diagnostic algorithm, angiographic evaluation, the endovascular intervention technique, and the patient's clinical course over a five-year follow-up period. Shunt closure resulted in marked clinical improvement, including normalization of oxygen saturation (from 60% to 98%), reduction in blood ammonia levels, increased exercise tolerance, and enhanced quality of life.The findings support the effectiveness of an individualized, stepwise approach in the management of type II CPSS. The importance of early recognition of this condition is emphasized, particularly in pediatric patients presenting with unexplained cyanosis, hypoxemia, or signs of hepatic dysfunction. Endovascular techniques, due to their minimally invasive nature and high clinical efficacy, represent an optimal therapeutic strategy in pediatric practice.

Introduction

Abernethy malformation, or congenital portosystemic shunt (CPSS), is a rare vascular anomaly in which blood from the portal circulation partially or completely bypasses the liver and drains directly into the systemic venous system. This condition disrupts hepatic metabolic function and may result in severe complications, including hepatopulmonary syndrome (HPS), hepatic encephalopathy, hypoxemia, hyperinsulinemia, and, in some cases, sudden death.[1,2]

The formation of the portal venous system occurs early in embryogenesis—between the 4th and 10th weeks of gestation. Aberrant vascular development during this period may lead to the forma-

tion of extra- or intrahepatic portocaval shunts. The congenital absence of the portal vein was first described by John Abernethy in 1793 during the autopsy of a child. A modern classification system for CPSS was proposed by *Morgan and Superina* in 1994, dividing the condition into two types: Type I — complete absence of the intrahepatic portal venous system; and Type II — presence of hypoplastic intrahepatic portal branches with partial shunting.[3]

Later, *Lautz et al.* refined the classification of Type II shunts into subtypes IIa, IIb, and IIc based on the anatomical origin of the shunt. *Kanazawa et al.* further introduced a classification based on the degree of hypoplasia of intrahepatic branches (mild, moderate, or severe), which is crucial for determining the optimal treatment strategy.[4,5]

The estimated prevalence of CPSS is approximately 1 in 30,000–50,000 live births.[6] However, due to the nonspecific nature of its clinical manifestations, diagnosis is often delayed. The most characteristic features include arterial hypoxemia, cyanosis, exertional dyspnea, platypnea, mucocutaneoustelangiectasias, and digital clubbing. These symptoms are typical of hepatopulmonary syndrome, which develops in more than 50% of patients with CPSS and is associated with severe oxygenation disorders.[7,8]

Other commonly observed findings include nodular regenerative hyperplasia of the liver (up to 50%), hyperammonemia, hyperinsulinemia with hypoglycemic episodes, and neurological disturbances related to manganese deposition in the basal ganglia. Hepatic encephalopathy often manifests at later stages, particularly in preschool-aged children.[4,6]

Despite the availability of modern imaging modalities such as ultrasonography, CT, MRI, and angiography, CPSS is frequently diagnosed late. This delay is due in part to the lack of awareness among clinicians to whom such patients typically present (e.g., gastroenterologists, neurologists, endocrinologists, and surgeons).[9] A diagnostically relevant laboratory marker is elevated blood ammonia; in one study, the mean ammonia level in children with CPSS was 123 ± 37 μg/dL, compared to a normal upper limit of 66 μg/dL.[5]

The cornerstone of treatment is shunt closure—either surgically (ligation) or via endovascular techniques (e.g., occluder placement). The choice of intervention depends on the results of an occlusion test: if portal pressure remains below 25 mmHg during temporary shunt closure, one-stage closure is feasible; otherwise, a staged approach is recommended to prevent portal hypertension.[4,7]

Endovascular techniques such as Amplatzer device placement offer high clinical efficacy with lower invasiveness compared to open surgery. However, complex vascular anatomy may necessitate surgical ligation.[7,10] Following intervention, ammonia levels typically normalize, and imaging (ultrasound, MRI, CT, angiography) is used for post-treatment monitoring.

Thus, early identification of CPSS and an individualized treatment strategy can prevent severe complications and significantly improve outcomes in pediatric patients. The present study reports the first documented case in the Republic of Kazakhstan of successful treatment of Abernethy type II malformation in a child with severe hepatopulmonary syndrome.

To assess the effectiveness of endovascular treatment for congenital portosystemic shunt (Abernethy malformation type II) in a child with hepatopulmonary syndrome, based on clinical outcomes and long-term follow-up.

Clinical case presentation

For the first time in clinical practice in the Republic of Kazakhstan, we identified and successfully treated a case of a congenital portosystemic shunt (CPSS) type II, also known as Abernethy malformation. The patient, S., a 7-year-old boy, was urgently transported by air ambulance and admitted to the Department of Cardiac and Interventional Pediatric Surgery at the National Center for Pediatric Pathology and Surgery. His presenting complaints included progressive dyspnea, marked fatigue, reduced tolerance to even minimal physical and emotional

exertion, intermittent leg pain, numbness in the fingers, and central cyanosis in the perioral area.

The medical history was notable for multiple hospitalizations dating back to 2018. The patient had been diagnosed twice with brain abscesses (in 2018 and 2019), which were complicated by right-sided hemiparesis, facial nerve neuritis, and toxic myocarditis. Additionally, neuroimaging revealed cerebral and arachnoid cysts, subcortical abnormalities, and hydrocephalus. Abdominal ultrasound (2020) identified hypoechoic liver lesions in the right lobe, and in 2021, an abnormality of the portal venous system was first suspected. Despite extensive evaluations, the diagnosis of CPSS was not established until March 2021 (Table 1).

Table 1.
Prior Medical History and Interventions

Date	Prior Medical History and Interventions
July 2018	Inpatient treatment. Diagnosis: Right cerebral hemisphere abscess. Conservative therapy administered. Discharged in improved condition.
April 2019	Inpatient treatment. Diagnosis: Left cerebral hemisphere abscess. Complicated by right-sided hemiparesis and right-sided facial nerve neuritis. Mild iron-deficiency anemia. Toxic myocarditis. Conservative treatment. Discharged in improved condition.
December 2020 – January 2021 – February 2021 March 2021	Chest CT (29.12.2020): No organic pathology. Abdominal ultrasound (29.12.2020): Diffuse parenchymal liver changes, gallbladder deformation, hypoechoic lesions in segments 7–8. Brain MRI (04.01.2021): Encephalopathy, cerebral cysts. Neurology consultation (08.01.2021): Multiple cerebral cysts and sequelae of brain abscesses. Referred to neurology department. Echocardiography (04.02.2021): Normal cardiac anatomy, LVEF 64%. Abdominal CT (26.02.2021): No significant pathology.
	Hospitalization at the National Center for Pediatric Pathology and Surgery for angiopulmonography and cardiac catheterization to confirm diagnosis and determine treatment strategy.
June 2021	Rehospitalization at the same center. A one-stage endovascular occlusion of the abnormal portosystemic shunt was successfully performed.

Patient Status and Interventions At the time of admission to the National Center for Pediatric Pathology and Surgery, the patient's condition was assessed as severe, with marked respiratory insufficiency (SpO_2 60–70%), cachexia (body weight 21 kg, height 126 cm), and a forced squatting posture with knees drawn to the chest. Pulmonary angiography, right and left heart catheterization, aortography, and retrograde mesenterico-splenoportography confirmed the diagnosis of congenital portosystemic shunt (CPSS) type II. The measured mean portal pressure was 22 mmHg, which allowed for a one-stage endovascular closure of the shunt.

On July 14, 2021, the patient underwent percutaneous transcatheter occlusion of the portosystemic shunt. The early postoperative course was complicated by persistent hypoxemia. However, after 12 days, follow-up angiopulmonography and embolization of pathological pulmonary arteriovenous communications were performed, resulting in significant clinical improvement.

Follow-up assessments over the subsequent three years demonstrated sustained recovery: weight gain, normalization of oxygen saturation (up to 98%), improved laboratory parameters (a decrease in blood ammonia from 95.1 to 72.3 μmol/L), and a marked increase in exercise tolerance—walking distance extended to 1500–2000 meters (Table 2).

Table 2.
Timeline of Interventions and Clinical Outcomes in a Patient with Abernethy Syndrome Type II

Date	ClinicalPresentation	DiagnosticEvaluation	Interventions
March 2021	Tachypnea, cyanosis, forced posture, SpO_2 60–70%, weight 21 kg, height 126 cm	Right/left heart catheterization, aortography, mesenterico-splenoportography, occlusion test. Portal pressure: 22 mmHg. Diagnosis: CPSS type II (Abernethy syndrome)	Diagnostic catheterization with angiography and occlusion testing
July 2021	Minimal weight gain, persistent cyanosis, SpO_2 60–65%	Abdominal ultrasound: portocaval anastomosis; Brain CT: hydrocephalus, cystic lesions; ALT: 786 U/L, AST: >913 U/L	Transcatheter closure of the portosystemic shunt (July 14, 2021)
July 26, 2021	Severe condition, respiratory failure, SpO_2 with oxygen: 60–65%, weight 18 kg	ALT: 7.0 U/L, AST: 14.0 U/L; BP: 106/72 mmHg; RV pressure: 115/8 mmHg; PA pressure: 110/54 mmHg. Conclusion: Abernethy syndrome, postshuntclosure status	Endovascular occlusion of distal pulmonary arteries bilaterally
August 6, 2021	Clinical improvement, SpO_2 85–90%, walking tolerance up to 100 m	—	—
September 2021	Weight gain, SpO_2 92–95%, walking distance up to 500 m	—	—
October 2021	+5 kg weight gain, SpO_2 95%, walking distance up to 1000 m	—	—
January 2022	Weight: 25.7 kg, height: 131 cm, SpO_2 98%, walking distance up to 1500 m; blood ammonia decreased from 95.1 to 72.3 μmol/L	—	—
October 2023	Further weight gain, SpO_2 92–95%, walking distance up to 1500 m; planned hospitalization	—	—
January 2024	Weight gain, SpO_2 92–96%, walking distance up to 1500–2000 m; planned hospitalization	—	—

This report presents a rare clinical case of Abernethy malformation type II successfully diagnosed and treated in Kazakhstan. Timely intervention during childhood prevented the progression of irreversible hepatic encephalopathy and severe hypoxemia, resulting in excellent long-term clinical outcomes and preserved quality of life (Figures 1 and 2).

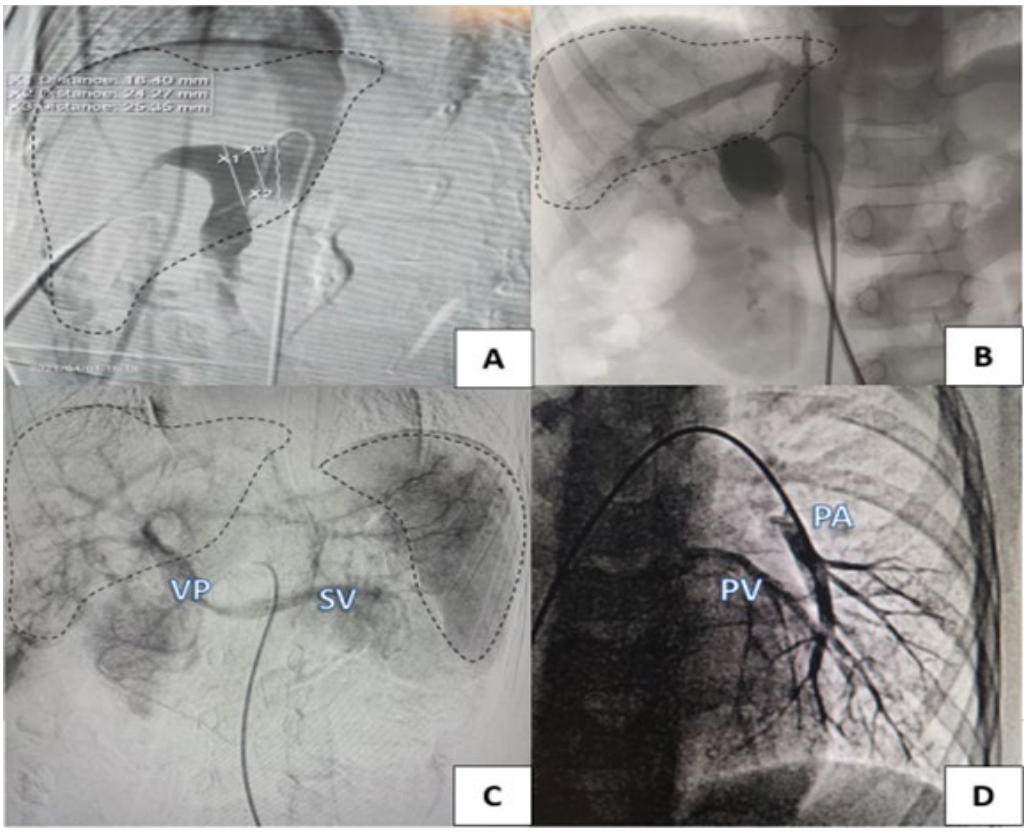

Figure 1
A – Direct cavaportography demonstrating a hypoplastic intrahepatic segment of the portal vein;B – Occlusion test with simultaneous portal venous manometry. The mean portal pressure measured at 22 mmHg;C – Retrograde celiaco-splenoportography showing angiographic visualization of the intrahepatic portal venous system following surgical disconnection of the congenital portosystemic shunt (CPSS);D – Selective segmental pulmonary angiography revealing simultaneous opacification of segmental pulmonary arteries and veins without parenchymal phase, indicative of pathological arteriovenous shunting.

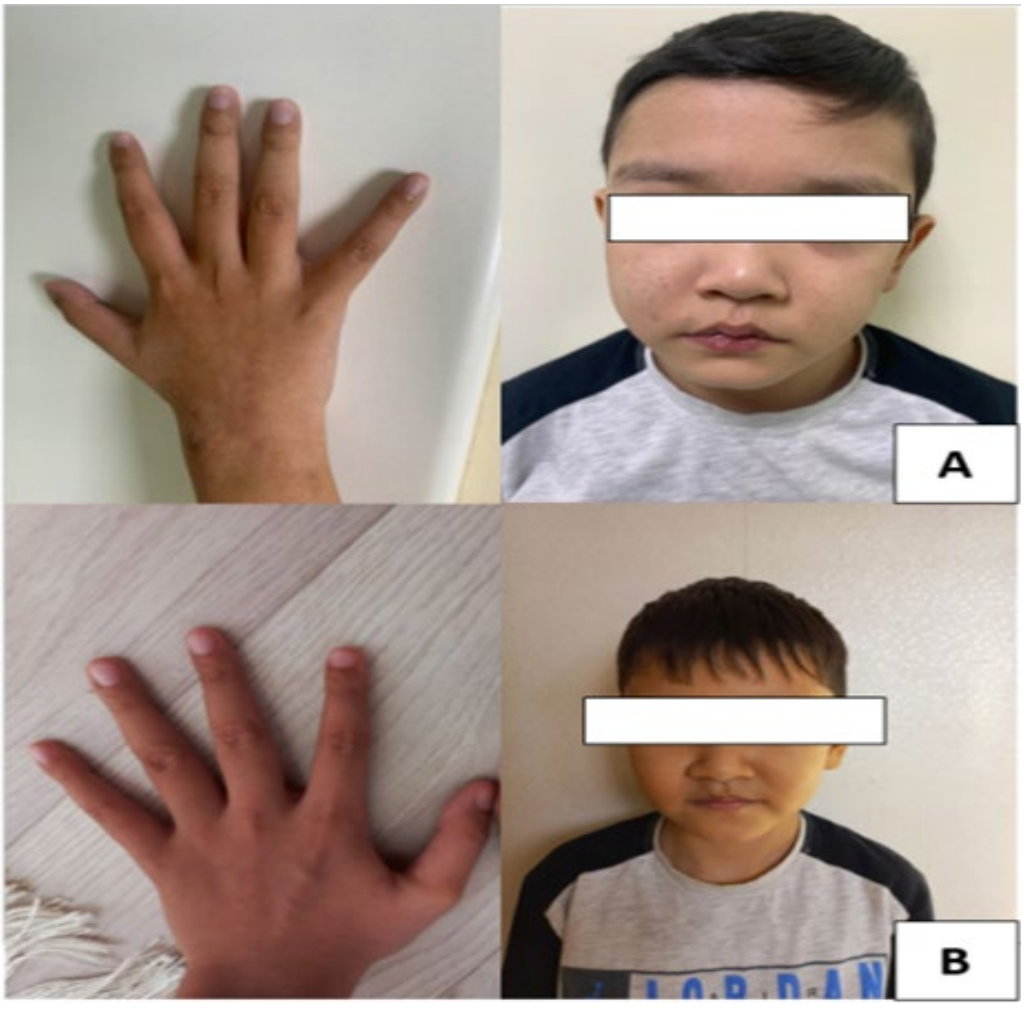

Figure 2.
A – External appearance of the patient with congenital portosystemic shunt (CPSS);B – External appearance of the patient 5 months after surgical disconnection of the CPSS.

Discussion

Congenital portosystemic shunts (CPSS), including Abernethy malformations, are rare vascular anomalies in which blood from the portal venous system partially or completely bypasses the liver and drains directly into the systemic circulation.[1,2] The Morgan and Superina classification categorizes CPSS into two types: type I is characterized by complete absence of intrahepatic portal venous branches, whereas type II retains partial intrahepatic portal perfusion.[3]

Abernethy malformation type II, as presented in this case, is a potentially correctable condition. According to current guidelines and clinical practice, the most informative preoperative assessment includes catheter-based evaluation with an occlusion test and direct measurement of portal venous pressure to assess the risk of portal hypertension following shunt closure.[4] In our case, the mean portal pressure was 22 mmHg, which was considered acceptable for endovascular intervention.

Modern treatment strategies for CPSS include both open surgical correction and minimally invasive transcatheter embolization, with the latter being particularly suitable for patients with type II anomalies.[6,7] In our patient, a staged hybrid approach was employed: initial shunt occlusion followed by selective pulmonary artery intervention due to severe hepatopulmonary syndrome. This strategy enabled the restoration of physiological portal flow and regression of clinical manifestations including hypoxemia and developmental delay.[8]

Early diagnosis of CPSS remains challenging due to the heterogeneity of clinical presentations. Cyanosis, hypoxemia, growth retardation, neurocognitive symptoms, and hyperammonemia may be the initial manifestations.[9] Therefore, CPSS should be included in the differential diagnosis of pediatric patients with unexplained oxygen desaturation and neurologic symptoms.

High-resolution imaging modalities such as multiphase CT, MRI, Doppler ultrasonography, and angiography, combined with interventional diagnostic procedures, provide accurate visualization of the shunt anatomy and aid in treatment planning.[10] In our case, the patient demonstrated sustained clinical improvement over a 5-year follow-up, including normalized oxygen saturation (95–98%), weight gain, reduction in neurological symptoms, and stabilization of hepatic biochemical markers.

This case underscores the importance of early detection and a staged interventional approach in the management of Abernethy type II malformations. It represents the first documented case in the Republic of Kazakhstan of successful endovascular and surgical treatment for this rare condition.

Limitations. This study is limited by its single-case design, reflecting the rarity of CPSS, particularly type Ia and Ib, which are frequently underdiagnosed or associated with early mortality before surgical intervention is feasible. The statistical power is therefore restricted, limiting extrapolation to a broader population. Furthermore, the study was conducted in a single specialized center, introducing potential selection bias. The absence of a control group precludes direct comparison with conservative or alternative therapeutic modalities.

What's known? CPSS is a rare congenital vascular anomaly with a broad clinical spectrum, ranging from asymptomatic forms to severe complications such as hepatic encephalopathy and hepatopulmonary syndrome. The Morgan and Superina classification, supplemented by Kanazawa's grading of intrahepatic portal hypoplasia, facilitates individualized therapeutic planning. Shunt closure, when indicated, can result in reversal of clinical symptoms and improvement in laboratory indices. Delayed diagnosis remains common due to the non-specific nature of symptoms and limited awareness among clinicians.

What's new? This is the first reported and documented case in Kazakhstan of successful treatment of Abernethy malformation type II in a child with severe hepatopulmonary syndrome. The efficacy of a staged approach—occlusion testing and portal pressure monitoring

using Kanazawa's protocol, followed by transcatheter shunt closure—was confirmed. Long-term remission was achieved, including normalization of oxygen saturation, weight gain, reduced ammonia levels, and resolution of respiratory symptoms. This case highlights the importance of including CPSS in the differential diagnosis of children with unexplained neurocognitive and hepatogastrointestinal symptoms, as well as nodular hepatic hyperplasia.

Conclusion

Congenital portosystemic shunting is a rare but potentially curable vascular anomaly requiring high clinical vigilance and a multidisciplinary approach. This case illustrates a successful diagnostic and therapeutic pathway for type II CPSS in a child with advanced hepatopulmonary syndrome. The use of an occlusion test with portal pressure measurement enabled a safe, staged treatment plan. Transcatheter intervention was effective and minimally invasive, making it highly suitable in pediatric settings. Restoration of physiological portal venous flow led to significant improvement in metabolic parameters and overall clinical status. Early diagnosis and timely intervention are crucial to improving outcomes and preventing irreversible complications.

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References

1. Zhang JS. Congenital intrahepatic portosystemic shunt in 27 children: an experience and treatment strategy of a single centre in China. Front Pediatr. 2024;12:1428270. doi:10.3389/fped.2024.1428270
2. Bahadori A, Kuhlmann B, Debray D, et al. Presentation of Congenital Portosystemic Shunts in Children. Children (Basel). Feb 11 2022;9(2) doi:10.3390/children9020243
3. Goncalves I, Barros D, Araujo M, Machado AI, Oliveira C, Pinto L. Type II Abernethy Malformation: A Rare Cause of Hepatic Encephalopathy in Adulthood. Eur J Case Rep Intern Med. 2022;9(1):003145. doi:10.12890/2022_003145
4. Kanazawa H, Nosaka S, Miyazaki O, et al. The classification based on intrahepatic portal system for congenital portosystemic shunts. J Pediatr Surg. Apr 2015;50(4):688-95. doi:10.1016/j.jpedsurg.2015.01.009
5. Rajeswaran S, Johnston A, Green J, et al. Abernethy Malformations: Evaluation and Management of Congenital Portosystemic Shunts. J Vasc Interv Radiol. May 2020;31(5):788-794. doi:10.1016/j.jvir.2019.08.007
6. Ponce-Dorrego MD, Hernandez-Cabrero T, Garzon-Moll G. Endovascular Treatment of Congenital Portosystemic Shunt: A Single-Center Prospective Study. Pediatr Gastroenterol Hepatol Nutr. Mar 2022;25(2):147-162. doi:10.5223/pghn.2022.25.2.147
7. Bhatte S, Cahill AM, Dunn M, Foran A, Perez A, Acord MR. Endovascular closure of a congenital extrahepatic portosystemic shunt for the treatment of hepatopulmonary syndrome in an infant. Pediatr Radiol. Feb 2024;54(2):357-361. doi:10.1007/s00247-023-05837-w

8. Warner S, McKiernan PJ, Hartley J, et al. Hepatopulmonary Syndrome in Children: A 20-Year Review of Presenting Symptoms, Clinical Progression, and Transplant Outcome. Liver Transpl. Sep 2018;24(9):1271-1279. doi:10.1002/lt.25296
9. Abdullah L, Hussein L, Al Houri HN, Khouri L. Abernethy malformation (Type 1B) presenting in a 6-year-old boy with hematochezia and hematuria: A case report. Radiol Case Rep. Sep 2022;17(9):3318-3320. doi:10.1016/j.radcr.2022.06.045
10. El-Medany AY, Rego G, Williams M, Lyen S, Turner M. Multimodality imaging of Abernethy malformation. Echocardiography. Mar 2022;39(3):524-527. doi:10.1111/echo.15324