Received: 21 June 2020 | Revised: 2 September 2020 | Accepted: 29 January 2021

DOI: 10.1002/ccr3.3920

CASE REPORT

Clinical Case Reports Open Access WILEY

Nephronophthisis and central veins abnormalities: A case report

Mohamed Amine Rahil[1] | **Messaoud Hadjmhamed**[2]

[1]Interventional Nephrology Unit, Bachir Ben Nacer Hospital, Biskra, Algeria

[2]Interventional Nephrology Unit, Gardaia Central Hospital, Ghardaia, Algeria

Correspondence
Mohamed Amine Rahil, Interventional Nephrology Unit, Bachir Ben Nacer Hospital, El alia Township, 70001 Biskra, Algeria.
Email: mohamedaminerahil@gmail.com

Abstract

Patients with genetic disorders are potentially more susceptible to present vascular abnormalities compared to the general population. For these patients, unusual difficulties could appear during a CVC placement procedure that could lead to major complications if venous abnormalities are undiagnosed. Ultrasound and fluoroscopy guidance should be used routinely for all patients in order to avoid complications and catheter misplacement.

KEYWORDS

arteriovenous fistula, azygos vein, dextrocardia, left superior vena cava, nephronophthisis, tunneled hemodialysis catheter

1 | INTRODUCTION

Nephronophthisis (NPHP) is a genetic disease affecting pediatric populations leading to end-stage renal diseases. Many syndromes could be associated with NPHP. We reported a child affected by NPHP with dextrocardia, LSVC, and LAV in which diagnosis was determined by fluoroscopy after having encountered difficulties in central venous catheter insertion.

Nephronophthisis (NPHP) is an autosomal recessive disease that affects pediatric populations during the first decade of life. The effect of NPHP on the kidneys is characterized by a chronic tubulointerstitial nephritis (CTN) leading to end-stage renal disease (ESRD). Many syndromes could be associated with NPHP including cerebellar ataxia (Joubert syndrome), retinitis pigmentosa (Senior–Løken syndrome), mental retardation, cardiac malformation, situs invertus, and many others.[12] For these patients, congenital vascular abnormalities are diagnosed fortuitously during a central vein catheter placement. The procedure could present unusual difficulties, such as guide wire misplacement, vein perforation, or catheter dysfunction.

We report a case of a 13-year-old child in ESRD affected by NPHP with dextrocardia, left superior vena cava (LSVC), and left azygos vein (LAV). These anatomical abnormalities were diagnosed by fluoroscopy with contrast injection after encountering unusual difficulties to insert a hemodialysis catheter.

2 | CASE

A 13-year-old child with end-stage renal disease (ESRD) was brought to our facility to undergo a tunneled hemodialysis catheter placement. The patient was transferred to hemodialysis treatment after peritoneal dialysis failure. Several unsuccessful attempts to create a native arteriovenous fistula were made, and the patient required several short-term catheters to undergo hemodialysis.

The patient has been followed for his NPHP for seven years by the pediatricians. Diagnosis was reached by evaluating clinical and radiological data. The detection of a compound heterozygous or homozygous mutations in a gene contributing to NPHP is not available in our country. Clinical assessment found cerebral ataxia and developmental delay, and dextrocardia was confirmed by chest X-ray. Echocardiography showed classic mirror dextrocardia without associated malformations.

The procedure was completed in an operating room and under general anesthesia guided by fluoroscopy and contrast injection. The central vein puncture was done by ultrasound guidance. After puncturing the left internal jugular vein, the guide wire mislocated to the left subclavian vein and failed to reach the cardiac atrium (Figure 1).

Angiography of the chest's left side revealed a thrombosed jugular vein, a thrombosed subclavian vein, and a thrombosed brachiocephalic vein, followed by a free LSVC and a left azygos vein (Figure 2). Angiography of the chest's right side revealed a free jugular vein and a free brachiocephalic vein (Figure 3). Guided by fluoroscopy, a hydrophilic guide wire was placed in the left cardiac atrium through the right internal jugular vein. A 12 French double-lumen tunneled hemodialysis catheter was placed in the cardiac atrium with an accurate tip catheter position (Figure 4).

3 | DISCUSSION

Three types of NPHPs were described: juvenile (or type 1) NPHP, in which symptoms emerge between ages 4 and 6 and lead to ESRD at age 13 or later; infantile (or type 2) NPHP, which leads to ESRD before age 2; and adolescent (or type 3) NPHP, where ESRD developed at a mean age of 19 years.[13] Extrarenal syndromes are associated with NPHP in 10%-20% of cases, including cerebellar ataxia (Joubert syndrome), retinitis pigmentosa (Senior–Løken syndrome), mental retardation, cardiac malformation, situs invertus, and many others.[1] NPHP with situs invertus or congenital heart abnormalities occur predominantly in infants; the most common congenital heart defect in this setting is ventricular septal defect.[12]

Patients with genetic disorders are potentially more susceptible to organ or vascular abnormalities compared to the general population.[11] LSVC occurs in 0.3-0.5% of the general

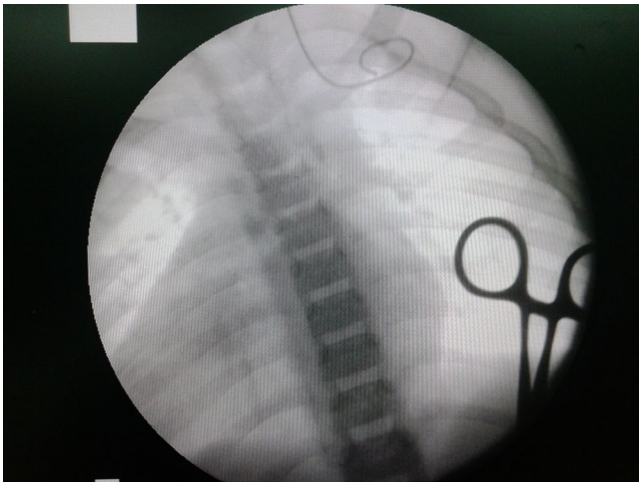

FIGURE 1 The guide wire cannot reach the cardiac atrium

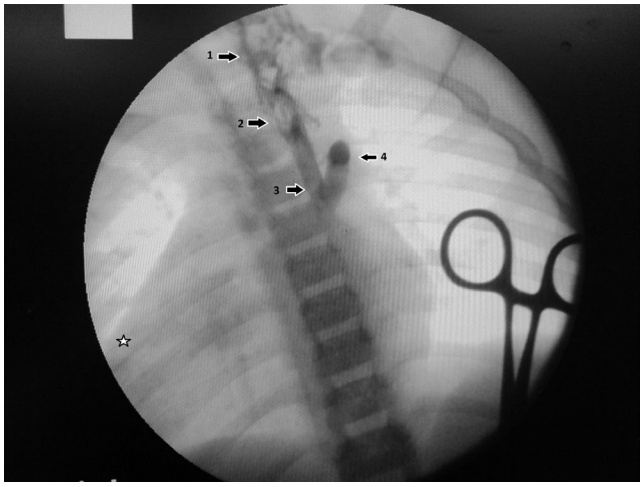

FIGURE 2 Angiography of the left side of the chest showing a dextrocardia (*), a thrombosed left internal jugular vein,[1] a thrombosed left brachiocephalic vein,[2] a free LSVC,[3] and a left azygos vein [4]

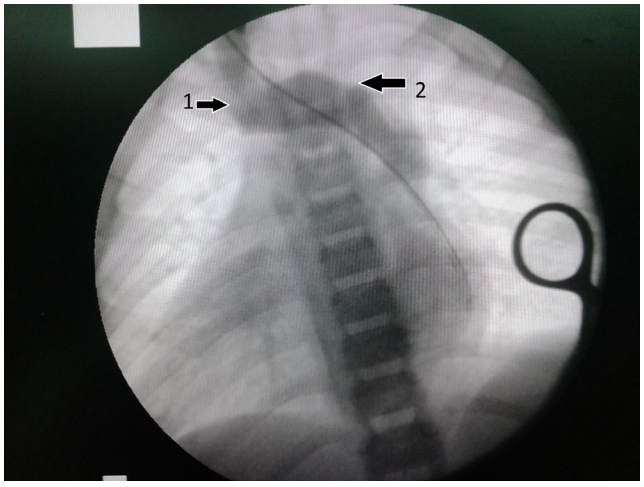

FIGURE 3 Angiography of the right side of the chest showing a free right jugular vein,[5] and a free right brachiocephalic vein [6] connected to the LSVC

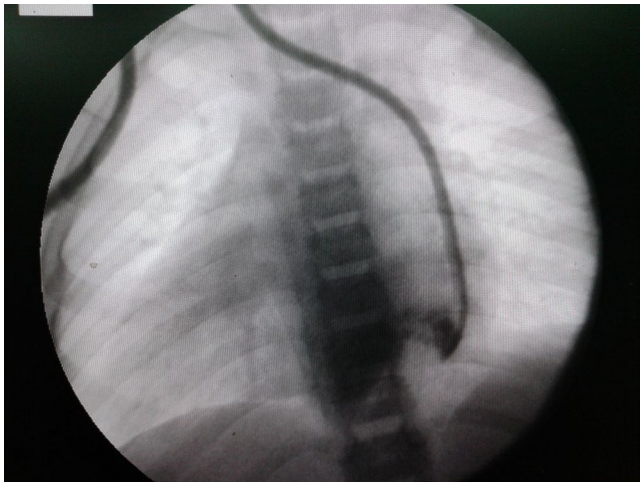

FIGURE 4 A 12 French double-lumen tunneled catheter placed through the right internal jugular

population and in 3%-5% of patients with congenital heart disease.[6,7] The development of the left anterior cardinal vein occurs a complete regression of the right SVC.[7-11] Usually, the LSVC allows blood to reach the right atrium through the coronary sinus. LSVC is typically asymptomatic and does not require treatment unless accompanied by other cardiac anomalies.[6-8]

If central vein abnormalities are not ruled out, central vein procedures could lead to serious complications.[2] Ultrasonography and fluoroscopy have shown fewer complications and fewer catheter tip malpositions relative to procedures performed in a blinded fashion.[3-5] Chait et al described a surgically placed left-sided subclavian central vein catheter (CVC) in a patient with a left-sided superior vena cava that caused a hemothorax; subsequently, an interventional radiologist placed a CVC in the left internal jugular vein under fluoroscopy.[2]

Our patient presented two major difficulties: central vein abnormality (LSVC) and a thrombosed left jugular vein with thrombosed left brachiocephalic vein secondary to repeated short-term catheter placement. For these patients, ultrasound guidance for vein puncture and fluoroscopic guidance for accurate catheter positioning should be mandatory to prevent major complications or CVC misplacement.[9-11]

4 | CONCLUSION

NPHP is one of many genetic disorders that can lead to ESRD. One or several organs could be affected, including the eyes, brain, bone, liver, or heart. Patients with genetic disorders are potentially more susceptible to present vascular abnormalities compared to the general population. For these patients, unusual difficulties could appear during a CVC placement procedure that could lead to major complications if venous abnormalities are undiagnosed. Ultrasound and fluoroscopy guidance should be used routinely for all patients in order to avoid complications and catheter misplacement.

ACKNOWLEDGMENTS

Published with written consent of the patient.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

AUTHOR CONTRIBUTIONS

Mohamed Amine Rahil performed the angiography and the CVC placement. Hadjmhamed Messaoud performed the catheter insertion. Both authors contributed to writing the manuscript and approved the manuscript in its current version.

ETHICAL APPROVAL

Ethical approval was not required, as no experiments were performed on human participants or animals.

INFORMED CONSENT

Informed consent was obtained from all individual participants included in the study.

ORCID

Mohamed Amine Rahil https://orcid.org/0000-0002-1468-5413

REFERENCES

1. Salomon R, Saunier S, Niaudet P. Nephronophthisis. *Pediatr Nephrol*. 2009;24:2333-2344.
2. Chait PG, Temple M, Connolly B, John P, Restrepo R, Amaral JG. Pediatric interventional venous access. *Tech Vasc Interv Radiol*. 2002;5(2):95-102.
3. Ash SR. Advances in tunneled central venous catheters for dialysis: Design and performance. *Semin Dial*. 2008;21:504-515.
4. Weber E, Liberek T, Wołyniec W, Rutkowski B. Catheter tip malposition after percutaneous placement of tunneled hemodialysis catheters. *Hemodial Int*. 2015;19(4):509-513.
5. Foley MJ. Radiologic placement of long-term central venous peripheral access system ports (PAS Port): Results in 150 patients. *J Vasc Interv Radiol*. 1995;6:255-262.
6. Park Myubng K. *Pediatric Cardiology for Practitioners*, 4th edn. Mosby; 2002:14-263.
7. Minniti S, Visentini S, Procacci C. Congenital anomalies of the venae cavae: embryological origin, imaging features and report of three new variants. *Eur Radiol*. 2002;12:2040-2055.
8. Gerber TC, Kuzo RS. Images in cardiovascular medicine. Persistent left superior vena cava demonstrated with multislice spiral computed tomography. *Circulation*. 2002;105:e79.
9. Lamperti M, Bodenham AR, Pittiruti M, et al. International evidence-based recommendations on ultrasound-guided vascular access. *Intensive Care Med*. 2012;38(7):1105-1117.
10. Khoo SW, Han DC. The use of ultrasound in vascular procedures. *Surg Clin North Am*. 2011;91(1):173-184.
11. Rossi UG, Rigamonti P, Torcia P, et al. Congenital anomalies of superior vena cava and their implications in central venous catheterization. *J Vasc Access*. 2015;16(4):265-268.
12. Otto EA, Schermer B, Obara T, et al. Mutations in INVS encoding inversin cause nephronophthisis type 2, linking renal cystic disease to the function of primary cilia and left-right axis determination. *Nat Genet*. 2003;34:413-420.
13. Wolf MTF. Nephronophthisis and related syndromes. *Curr Opin Pediatr*. 2015;27(2):201-211.

How to cite this article: Rahil MA, Hadjmhamed M. Nephronophthisis and central veins abnormalities: A case report. *Clin Case Rep*. 2021;9:1977–1979. https://doi.org/10.1002/ccr3.3920