Nephronophthisis and central veins abnormalities : a case report.

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Abstract

Nephronophthisis is a pediatric genetic disease. The kidney effect is characterized by a CTN leading to ESRD. Many syndromes could be associated with NPHP. We reported a child affected by NPHP with dextrocardia, LSVC and LAV where the diagnostic was done by fluoroscopy after encountering unusual difficulties to perform CVC.

Nephronophthisis and central veins abnormalities: a case report

Short Title: Catheter in left superior vena cava.

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Abstract:

Nephronophthisis is a pediatric genetic disease. The kidney effect is characterized by a CTN leading to ESRD. Many syndromes could be associated with NPHP. We reported a child affected by NPHP with dextrocardia, LSVC and LAV where the diagnostic was done by fluoroscopy after encountering unusual difficulties to perform CVC.

Keywords: Nephronophthisis, tunneled hemodialysis catheter, dextrocardia, left superior vena cava, azygos vein, arteriovenous fistula.

Key Clinical Message :

Organ abnormalities should be eliminated in all patients with genetic diseases. For these patients, invasive procedures could present unusual difficulties and lead to a major complication.

Abbreviations:

NPHP : Nephronophthisis

CVC : central vein catheter

LSVC : Left superior vena cava

SVC : superior vena cava

ESRD : end stage renal disease.

LAV : left azygos vein.

CTN : chronic tubulonephritis

Introduction:

Nephronophthisis (NPHP) is an autosomal recessive disease that affects pediatric population between the first and the second decade. The kidney effect is characterized by a chronic tubulointerstitial nephritis leading to end stage renal disease. 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 as a guide wire misplacement, vein perforation or catheter dysfunction.

We reported a case of a 13 years child in ESRD affected by NPHP with dextrocardia, left superior vena cava (LSVC) and left azygos vein where the diagnostic of theses anatomical abnormalities was done by fluoroscopy with contrast injection after encountering unusual difficulties to perform a hemodialysis catheter.

Case:

A 13 years child with end stage renal disease was brought to our facility to undergone a tunneled hemodialysis catheter. The patient was switched to hemodialysis after peritoneal dialysis failure.

Several attempts to create a native arteriovenous fistula were done without success, and the patient undergone several short term catheters to have hemodialysis sessions.

The patient has seven years follow up by the pediatricians for his NPHP. The diagnostic was done by clinical and radiological evidence. The detection of compound heterozygous or homozygous mutations in a gene contributing to NPHP is not available in the country.

Clinical assessments found: cerebral ataxia, developmental delay, and a dextrocardia at the chest x-ray. Echocardiography showed classic mirror dextrocardia without associated malformations.

The patient was placed in an operating room. The procedure has been done under general anesthesia guided by fluoroscopy and contrast injection. The central vein puncture was done by ultrasound guidance. After needling the left internal jugular vein, the guide wire could not reach the cardiac atrium and it was mislocated in the left subclavian vein (Fig 1).

The angiography of the chest left side has shown: a thrombosed jugular vein, a thrombosed subclavian vein and a thrombosed brachiocephalic vein followed by a free LSVC and a left azygos vein (Fig 2). The

angiography of the chest right side has shown: a free jugular vein and a free brachiocephalic vein (Fig 3). A hydrophilic guide wire has been placed in the left cardiac atrium guided by fluoroscopy through the right internal jugular vein.

A 12 french double lumen tunneled hemodialysis catheter was placed with an accurate tip catheter position in the cardiac atrium. (Fig 4).

Discussion:

Three type of NPHP were described : juvenile or type 1 NPHP which the symptoms start between 4 and 6 years and lead the patient to ESRD at 13 years or later, infantile NPHP or type 2 which lead the patient to ESRD before 2 years; and adolescent NPHP or type 3 where ESRD occurred at mean age of 19 years (13). Extrarenal syndromes are associated with NPHP in 10 to 20 %, 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 mostly in infantile patients. The most common congenital heart defect in this setting is ventricular septal defect (12).

Patients with genetic disorders are potentially more susceptible to present organs or vascular abnormalities compared to general population (11). LSVC occurs in 0.3% to 0.5% of the general population and in 3% to 5% of the 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). Most commonly the LSVC allows blood to reach the right atrium through the coronary sinus. LSVC is usually asymptomatic and does not require treatment unless accompanied by other cardiac anomalies (6-7-8).

If central veins abnormalities do not rule out, central vein procedures could lead to serious complications (2). Ultrasonography and fluoroscopy have shown fewer complications and fewer catheters tip malposition compared to procedures done by blinded fashion (3-4-5). Peter .G and col described surgically placed left-sided subclavian CVC in a patient with a left-sided superior vena cava which 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 which is: 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 the accurate position of the catheter should be mandatory to avoid major complications or CVC misplacement. (9-10-11).

Conclusion:

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

Authors' contributions Mohamed amine Rahil is the corresponding author, performed the angiography, the CVC placement and wrote the manuscript. Hadjmhamed messaoud performed the catheter and wrote the manuscript.

All authors read and approved the manuscript in its current version.

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Figure 1: shows the impossibility of the guide wire to reach the cardiac atrium.

Figure 2: represented the angiography of the chest left side and shows : a dextrocardia ( ), a thrombosed left internal jugular vein (1), a thrombosed left brachiocephalic vein (2), a free left superior vena cava (3), and a left azygos vein (4).

Figure 3: represented the angiography of the chest right side and shows: a free right jugular vein (5); and a free right brachiocephalic vein (6) connected to the LSVC.

Figure 4: shows a 12 french double lumen tunneled catheter placed through the right internal jugular.