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CASE REPORT

Neonatal Deep Vein Thrombosis Reveals an Inferior Vena Cava Agenesis associated with Antithrombin Deficiency: A Case Report

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Abstract

Inferior vena cava (IVC) agenesis is a rare condition. It can be either congenital or acquired due to thrombotic events. It is a rare cause of deep vein thrombosis. We describe the case of a female newborn who presented with edema and cyanosis of the right lower limb. Lower extremity Doppler US showed thrombosis of the external iliac vein, common and superficial femoral right veins. Abdominal Doppler US suspected the absence of the infrarenal segment of IVC. CT abdominal angiography confirmed the agenesis of the infrarenal segment of IVC. Blood tests found an antithrombin level of 11% confirming the diagnosis of congenital antithrombin deficiency. Pharmacological measures were implemented to treat thrombosis since a surgical approach was not feasible.

Keywords

Deep vein thrombosis, Inferior vena cava agenesis, Thrombophilic disorder, Antithrombin deficiency, Case report

Introduction

Inferior vena cava (IVC) agenesis is a rare condition that occurs in less than 0.0005% to 1% of the population [1]. Often asymptomatic and fortuitous discovery. However, it may be discovered following deep vein thrombosis at a young age [1]. Other vascular, gastrointestinal, cardiac, and pulmonary malformations are commonly present [2]. The main cause of the condition is either embryogenesis defects "true agenesis" or acquired because of intrauterine or perinatal IVC thrombosis often caused by hereditary thrombophilia [3]. The purpose of this case report is to provide a comprehensive overview of the diagnosis and management of deep venous thrombosis in newborn infants caused by agenesis of the IVC associated with antithrombin deficiency, as well as to analyze the current literature.

Case Report

A female newborn, conceived from non-conscientious parents and a 37-year-old mother, G_{IV}, P_{II}, A_{II}, has hypothyroidism, asymmetric kidneys, and congenital antithrombin deficiency at a rate of 46%. Her brother died at the age of 5 days due to a very high prematurity. Pregnancy was conducted under Acetylsalicylic acid and Enoxaparin with a normal course and normal prenatal ultrasound. Delivery was done at 37 SA and 4 days by cesarean section for a scared uterus with good adaptation to the extra-uterine life, a birth weight of 2780 g, a height of 46 cm, and a cranial perimeter of 34 cm.

At the age of 16 days, she underwent hospitalization due to edema and cyanosis of her right lower limb. The clinical examination found hepatomegaly, an edematous, cyanotic, painful right lower limb (Figure 1), warm extremities, and symmetrical pulses.

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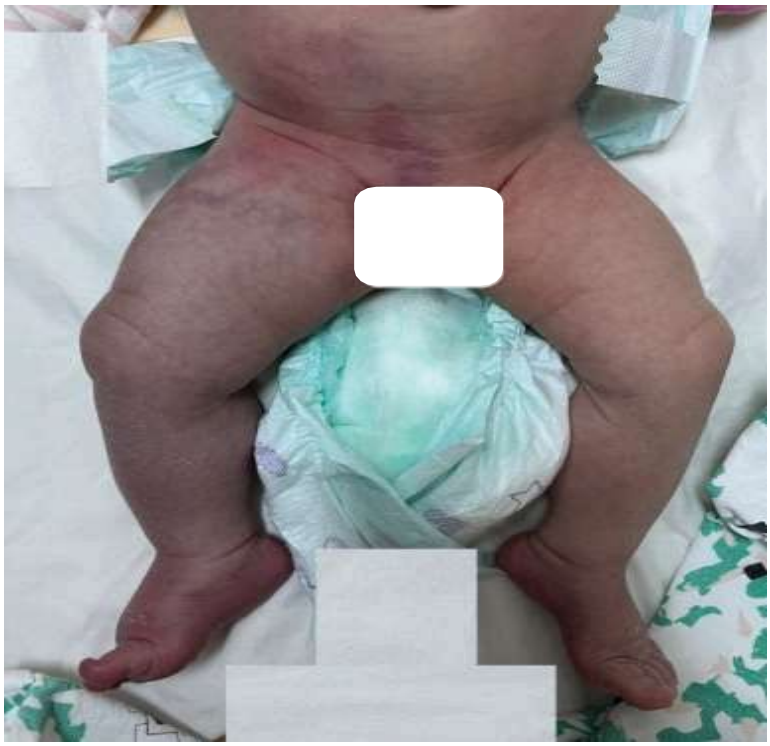

Figure 1: Clinical examination at the admission: Edema and cyanosis of the right lower limb.

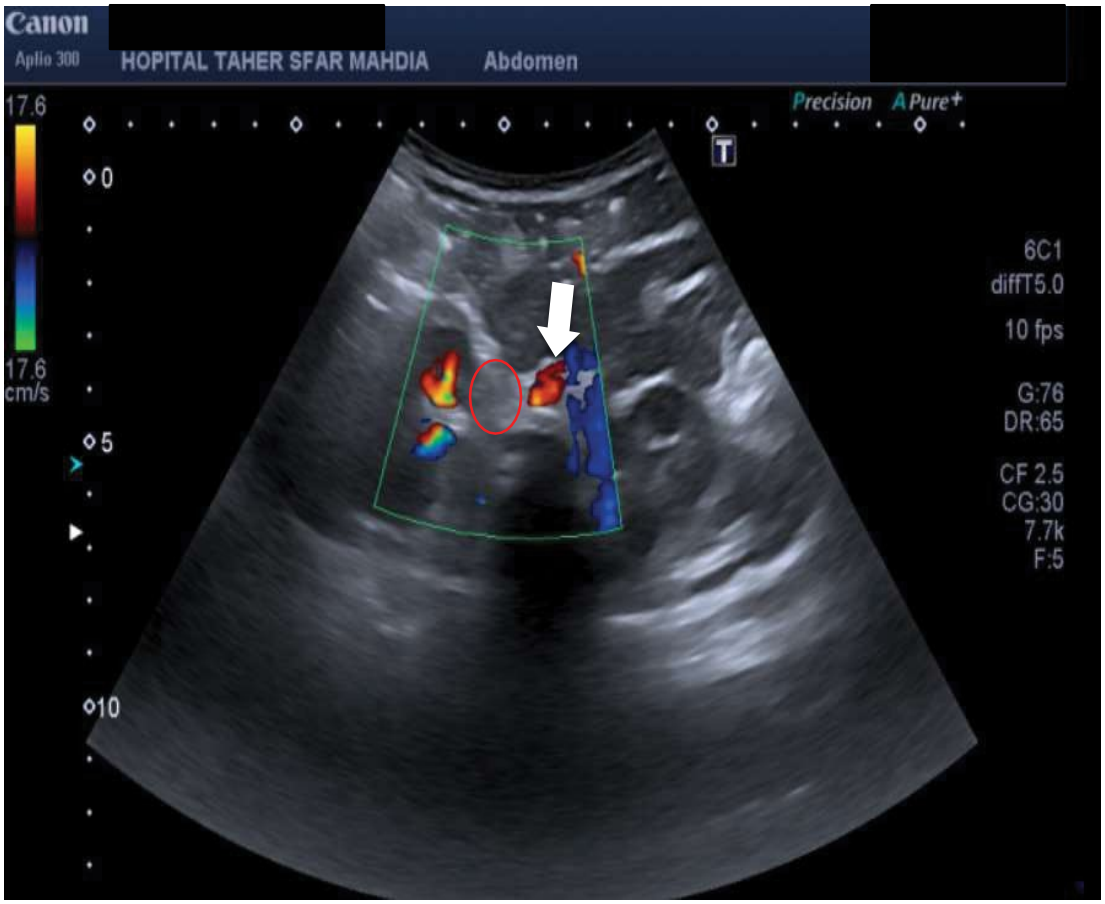

Figure 2: Axial ultrasound image of the abdomen shows the absence of the infrarenal segment of IVC (Red cercle). Abdominal aorta (Arrow).

Lower extremity Doppler US showed thrombosis of the external iliac vein, common and superficial femoral right veins. Abdominal Doppler US showed the absence of the infrarenal segment of IVC (Figure 2) and a hepatic hemangioma of segments VII and VI measuring 45 × 22 mm. The CT abdominal angiography revealed a calcified

thrombosis of the suprarenal segment of IVC (Figure 3), agenesis of the infrarenal segment of IVC (Figure 4 and Figure 5), enlarged azygos vein (Figure 6), the common left iliac veins drain into the ascending lumbar veins (Figure 7), a giant hepatic hemangioma and a supernumerary subhepatic vein (right inferior subhepatic vein) (Figure 4).

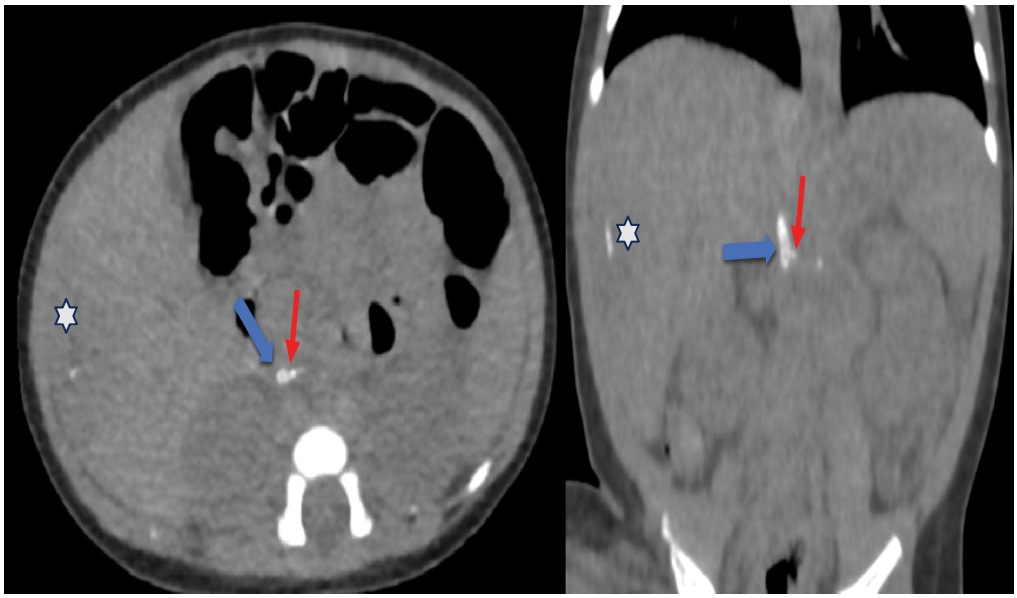

Figure 3: Non-contrast axial (a) and coronal (b) CT images of the abdomen: Calcified thrombosis of the suprarenal segment of IVC (Blue arrow) extended to the initial portion of the left renal vein (Red arrow). Hypodense subcapsular hepatic lesion of segments VII and VI, containing calcification (Star).

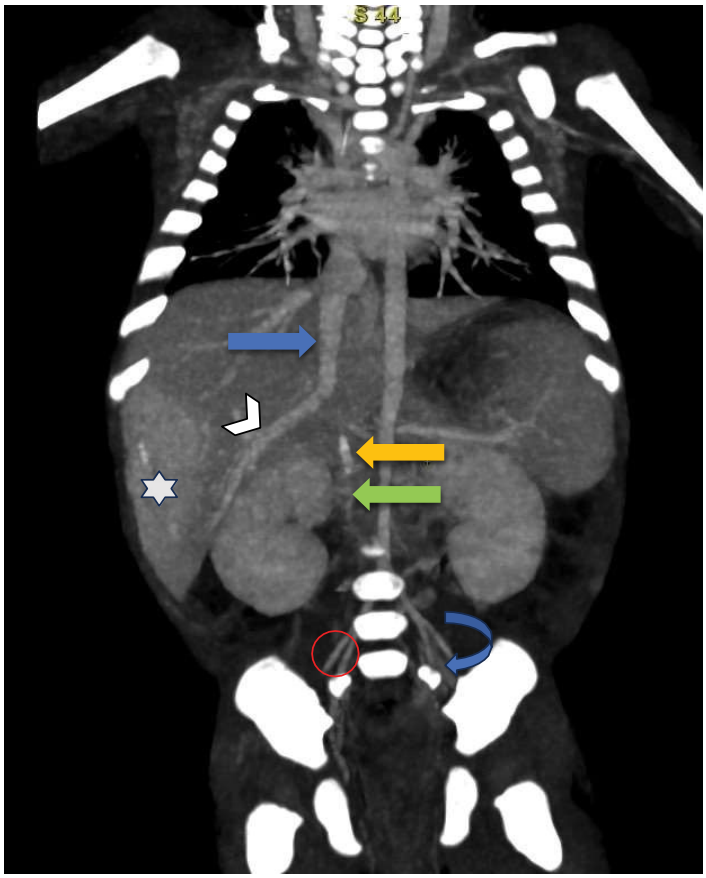

Figure 4: Postcontrast axial maximum intensity projections (MIP) CT image of the chest and abdomen shows normal retrohepatic segment of IVC (Blue arrow), calcified thrombosis of the suprarenal segment of IVC (Yellow arrow), absence of the infrarenal segment of IVC (Green arrow), Supernumerary subhepatic vein: Right inferior subhepatic vein (Head of arrow), common right iliac vein thrombosis (Cercle), normal left iliac vein (Incurved arrow. Subcapsular hepatic lesion of segments VII and VI, containing calcification (Star) referring to a hepatic angioma.

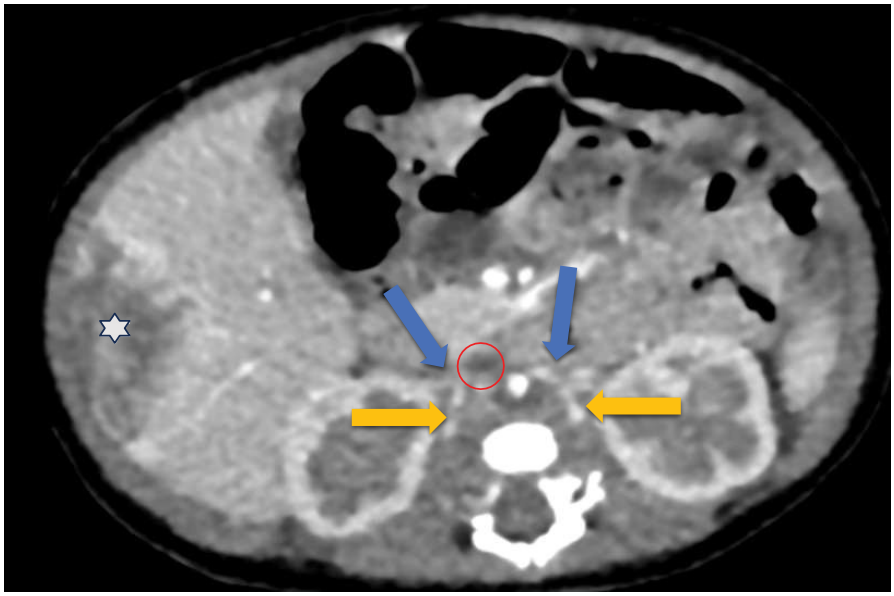

Figure 5: Postcontrast axial CT image of the abdomen shows the absence of the infrarenal segment of IVC (Red Cercle), renal veins (Blue arrows) drain into the ascending lumbar veins (Yellow arrows). Sub-capsular hepatic lesion of segments VII and VI having progressive peripheral enhancement with more centripetal fill-in referring to a hepatic angioma (Star).

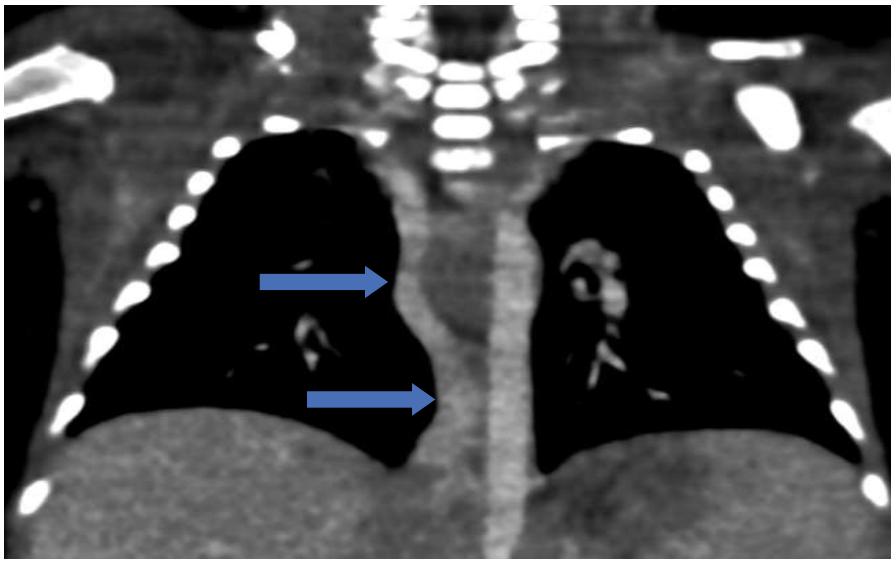

Figure 6: Postcontrast coronal CT image of the chest shows an enlarged azygos vein (Blue arrow).

The brain MRI was normal. Normal CBC, CRP, TSH, PR, aPTT, and fibrinogen were found in laboratory investigations. The level of antithrombin was 11% confirming the diagnosis of congenital antithrombin deficiency with normal S and C proteins. The patient was treated with Enoxaparin 200 IU/kg/12h, Propranolol 2 mg/kg/day, antithrombin 50 IU/kg/day for 15 days, and a transfusion of fresh frozen plasma (FFP) for 4 days. The evolution was marked by a healthcare-associated infection without isolated germ treated with Imipenem and Gentamicin, stability of the hepatic hemangioma, the edema in the right lower limb completely reversed, an improvement in antithrombin levels to 54%, anemia with Hb at 7 g/dl justifying a PRBC transfusion and the persistence of partial venous thrombosis in the right external iliac vein after 10 weeks of treatment with Enoxaparin. According to the opinion of the cardiovascular surgeons, there was no surgical indication for agenesis of IVC and no indication for thrombectomy. At the age of 3 months, she was released with a normal clinical examination (Figure 8), a weight of 5670 g, on Warfarin 0.3 mg/kg/day, or 2 mg/day to have an INR of 2.5, and Propranolol 2 mg/kg/day. Doppler US of the lower right limb at age 4 months showed a total regression of thrombosis of the external iliac vein and common and superficial femoral right veins. Doppler abdominal US showed hepatic hemangioma of segments VII and VI stability measuring 45 × 25 mm.

Discussion

IVC development has a complex embryological pathway. It is formed by the merge of three veins (supra cardinal, sub cardinal, and posterior cardinal)

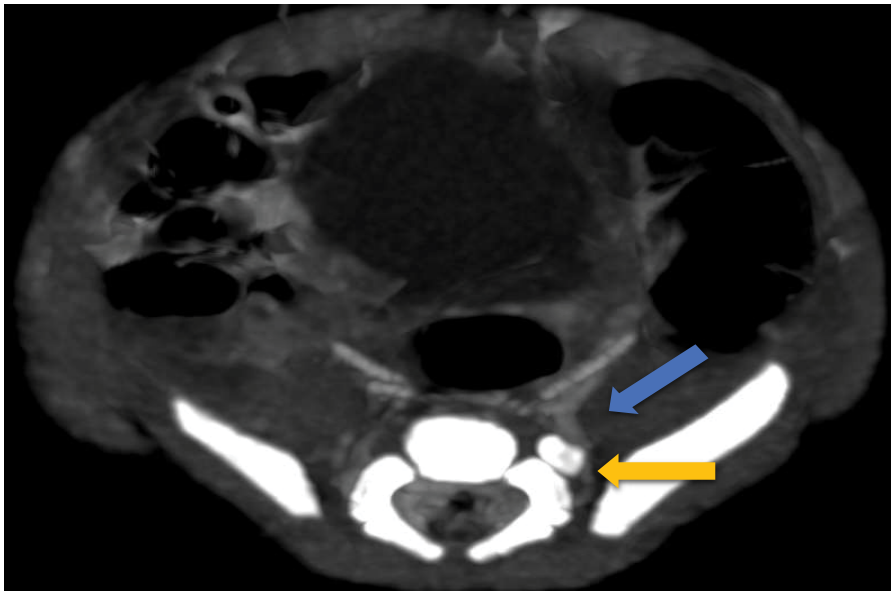

Figure 7: Postcontrast axial maximum intensity projections (MIP) CT images of the pelvis: The common left iliac veins (Blue arrow) drain into the ascending lumbar veins (Yellow arrow).

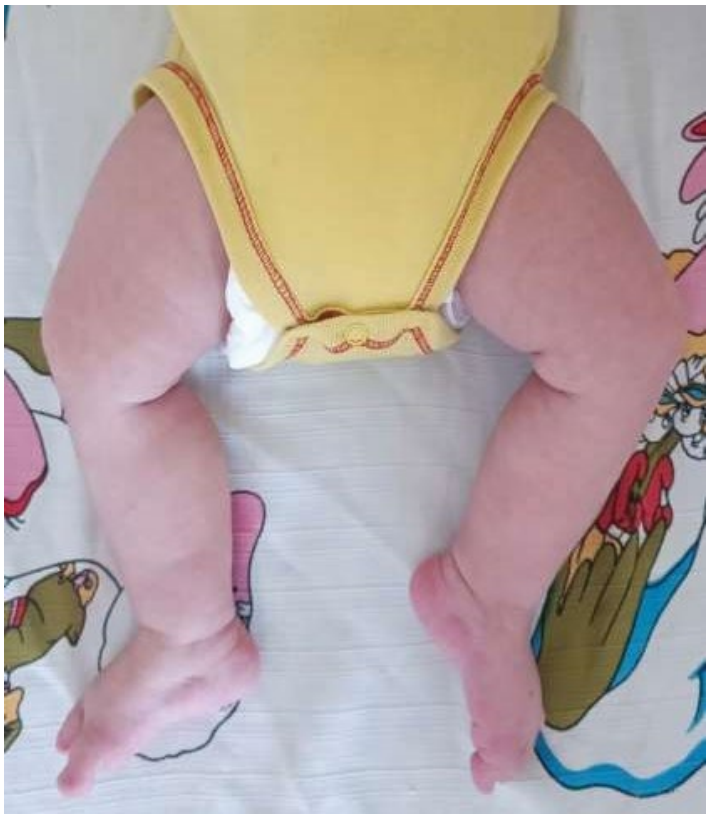

Figure 8: Clinical examination at age 4 months: Total regression of edema and cyanosis of the right lower limb.

during the fifth and seventh weeks of pregnancy [1]. The development failure of supra cardinal or posterior cardinal veins can lead to complete or segmental IVC agenesis. Segmental agenesis is divided based on the level into intrahepatic, suprarenal, renal, and infrarenal segment agenesis [1]. The true congenital absence of the IVC has an unknown incidence and unclear cause [3]. However, thrombophilic disorders have also been suggested to be the cause of secondary IVC agenesis.

Hereditary thrombophilia, including antithrombin deficiency, protein C or S deficiency, antiphospholipid antibodies, hyperhomocysteinemia, elevated factor VIII, dysfibrinogenemia, activated protein C resistance, prothrombin G20210A gene mutations, and factor V Leiden mutations, induced thrombotic events during the perinatal period or early life can lead to thrombotic occlusion of the IVC and secondary IVC agenesis [2]. The incidence of IVC agenesis was found to be high in

patients with antithrombin deficiency [4].

IVC agenesis leads to the expansion of the collateral veins, which allows venous drainage of the lower extremities to the superior vena cava by the azygos and hemiazygos veins, resulting in the absence of symptoms in most patients. However, the alternative pathways may predispose to venous hypertension and stasis, which can cause deep vein thrombosis [5].

Diagnosis is usually made with a Doppler color US that demonstrates the extent of deep vein thrombosis, and this could suggest IVC agenesis, particularly in newborn infants, but confirmation via CT or MRI is required. CT or MRI angiography confirms the absence of IVC and precisely identifies the preserved segment in cases of segmental agenesis. Other imaging findings can be detected on CT or MRI angiography, such as the absence of common iliac veins, the presence of collateral paraspinal circulation, and the drainage of the external and internal iliac veins into the ascending lumbar veins, which drain into the azygos-hemiazygos system. A calcified thrombosis at the level of the IVC interruption indicates that IVC agenesis is acquired.

No clear scientific evidence exists to determine the most effective therapeutic management, optimal duration, or anticoagulant therapy in newborns with deep vein thrombosis associated with antithrombin deficiency and IVC agenesis [2]. Unfractionated heparin and LMWH are the drugs that are commonly used. Although there is no definitive consensus on the optimal duration of treatment, it should generally last between six weeks and three months, depending on the clinical condition [6]. Enoxaparin is the most common LMWH used in newborn infants. To manage thrombosis, it is recommended to give an injection every 12 hours, with 170 UI/kg for term infants and 200 UI/kg for preterm infants. It is suggested to use half of the therapeutic dose for prophylaxis [6]. In the case of antithrombin deficiency, an adequate amount of antithrombin is required for Heparin's function. A 50 IU/kg/day dose for 15 days is recommended.

Due to the invasive nature of maintaining anticoagulation with LMWH for several weeks, oral anticoagulation may be recommended for long-term management. Warfarin and Acenocumarol are both drugs that can be used. To achieve an INR between 2 and 3,40, the recommended starting dose is 0.2 mg/kg/day, but it has been found that the average useful dose for infants is around 0.33 mg/kg/day [6].

Conclusion

Hereditary thrombophilia, particularly antithrombin deficiency, may lead to IVC agenesis, as shown in our case. Patients with IVC agenesis can be significantly burdened by deep vein thrombosis. Imaging has made monitoring easier, emphasizing its importance for thrombus diagnosis and management. Anticoagulation in newborns has not been standardized, so the therapeutic approaches used are derived from studies conducted in a small number of cases and guidelines adapted to treatments for adults.

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