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Case Report

Congenital Absence of the Inferior Vena Cava and Genetic Coagulation Abnormalities: A Rare Associated Risk Factor for Recurrent Idiopathic Deep Vein Thrombosis

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Summary: Congenital absence of the inferior vena cava (AIVC) has been reported as a risk factor of deep vein thrombosis (DVT) in young people. DVT is caused by an interaction between congenital coagulation abnormalities and acquired risk factors. We observed an 18-year-old patient with AIVC who developed recurrent deep vein thrombosis at the left leg. Molecular studies showed an heterozygosity for FV Leiden gene (*G1691A*) and a homozygosity

for methylenetetrahydrofolate reductase gene (*C677T*) in absence of folate and vitamin B₁₂ deficiency. After the second DVT episode, the patient has been treated with heparin and oral anticoagulant without discontinuation.

Key Words: Thrombosis—Absence of inferior vena cava—Factor V Leiden.

The incidence of deep vein thrombosis (DVT) in Western population is estimated at 1 in 1,000 individuals per annum (1). DVT is caused by an interaction between congenital coagulation abnormalities and acquired risk factors (2). Congenital absence of the inferior vena cava (AIVC) has been reported as a risk factor of DVT in young people (3–6). However, the percentage of AIVC in patients with DVT could have been underestimated because the standard diagnostic tool (compression B-mode scanning ultrasonography) does not allow accurate examination of abdominal veins because they cannot be ade-

quately compressed (7). Nevertheless in few cases, recently reported, we cannot observe a congenital thrombophilic state. We report a patient with recurrent DVT caused from acquired and congenital risk factors.

CASE REPORT

In 1990, we observed an 18-year-old patient in whom a deep vein thrombosis at the left leg developed. Inferior vena cava congenital absence was identified on abdominal contrast-enhanced computed tomography. Coagulation study showed normal plasma levels of antithrombin III, protein C, and protein S. LAC and anticardiolipin autoantibodies were not present.

The patient was treated early using heparin and oral anticoagulation for 12 months; during this period, complete resolution of thrombosis occurred. Eight years later, a new deep vein

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thrombosis developed in the right leg; molecular studies showed an heterozygosity for FV Leiden gene (*G1691A*) and a homozygosity for methyl-entetrahydrofolate reductase gene (*C677T*) in the absence of folate and vitamin B₁₂ deficiency.

The patient has been treated with heparin and oral anticoagulants without discontinuation.

DISCUSSION

The prevalence of congenital venous malformations has been estimated at 1% of the general population (8) and IVC abnormalities are present in approximately half of these cases. All previously described congenital conditions predispose to development of DVT by inducing a hypercoagulable state.

In recently reported cases (4,5), no patients present with congenital coagulation abnormalities in association with IVC anomalies; on the contrary in our patient, presenting with an IVC anomaly, we showed the concomitant presence of factor V Leiden and MTHFR mutations.

In a patient series with IVC anomalies, there were no recurrences of DVT in the subsequent 24 months of oral anticoagulant (OA) therapy but it is doubtful that a long-term follow-up would be necessary. In our patient, a new DVT event occurred 8 years after OA discharge, perhaps because two congenital coagulation abnormalities were present. When observed for a long time, IVC abnormalities in young people are presumably associated with a constant lifelong thrombosis risk; of course, in our patient, who presents

with two congenital risk factors for DVT, it would be mandatory to continue OA drug just after the first DVT event.

In conclusion, further studies are required to evaluate the long-term outcome of patients with AIVC, and the optimal duration of warfarin therapy in the patients with AIVC and an associated thrombophilia state.

REFERENCES

1. Anderson FA Jr, Wheeler HB, Goldberg RJ, et al. A population based perspective of the hospital incidence and case-fatality rates of deep vein thrombosis and pulmonary embolism. The Worcester DVT study. *Arch Intern Med* 1991;151:933.
2. Rosendaal FR. Venous thrombosis: A multicausal disease. *Lancet* 1999;353:1167.
3. Hamoud S, Nitecky S, Engel A, et al. Hypoplasia of the inferior vena cava with azygous continuation presenting as recurrent leg deep vein thrombosis. *Am J Med Sci* 2000;319:414.
4. Ruggeri M, Tosetto A, Castaman G, et al. Congenital absence of the inferior vena cava: A rare risk factor for idiopathic deep-vein thrombosis. *Lancet* 2001;357:441.
5. Chee YL, Culligan DJ, Watson HG. Inferior vena cava malformation as a risk factor for deep venous thrombosis in the young. *Br J Haematol* 2001;114:878.
6. Tiesenhausen K, Amann W, Thalhammer M, et al. Aplasia of the vena cava inferior as a cause for recurring thrombosis of the lower extremities and pelvic veins. *Vasa* 1999;28:289.
7. Lensing AW, Prandoni P, Brandjes D, et al. Detection of deep-vein thrombosis by real-time B-mode ultrasonography. *N Engl J Med* 1989;320:342.
8. Eifert S, Villavicencio JL, Kao TC, et al. Prevalence of deep venous anomalies in congenital vascular malformations of venous predominance. *J Vasc Surg* 2000;31:462.