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Case Report

Congenital Antithrombin Deficiency in Siblings

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Introduction

Congenital antithrombin III deficiency is one of the most severe forms of thrombophilia. It results from a genetic defect in one or more proteins involved in hemostatic processes. Inherited thrombophilia increases the risk of venous thrombosis, usually during the first years of life; however, it is generally not associated with a predisposition to arterial thrombosis. The risk of thromboembolic events increases with age. Thrombophilia caused by antithrombin deficiency is classified as severe thrombophilia (high risk of thrombosis).Congenital antithrombin III deficiency is inherited in an autosomal dominant manner. It is caused by a mutation in the SERPINC1 gene, which encodes antithrombin. The SERPINC1 gene is located on the long arm of chromosome 1 and consists of seven exons and six introns. To date, approximately 500 mutations of this gene have been identified. Homozygous antithrombin deficiency is considered a lethal variant.The prevalence of heterozygous antithrombin deficiency in the European population is estimated at 0.02–0.1%. Two types of antithrombin III deficiency are distinguished:

Type I - characterized by reduced antithrombin concentration and activity. This form is associated with a high risk of thrombosis (early onset and recurrent) and israreinthegeneralpopulation(prevalence<0.02%).

Type II - a qualitative deficiency, in which antithrombin activity is reduced while its concentration remains withinthenormalrange. Diagnosis primarily involves measuring antithrombin III activity (reference range: 80–120% of normal). In most heterozygous patients with congenital antithrombin deficiency, activity ranges from 40% to 60%. A further step in the diagnostic process is the determination of plasma antithrombin antigen levels, with reference values of 0.19- 0.31 g/L.

Clinical Case

A 13-year-old boy was admitted to the hospital due to fever, swelling, and pain in the right groin area and along the inner surface of the right thigh, which had persisted for three days prior to hospitalization. During this time, he had been treated symptomatically with Movalis, Bi-Profenid, and topical ketoprofen gel, without improvement [1]. On the day of admission, increasing swelling of the right lower limb was additionally observed. Traumatic causes of the reported symptoms were excluded. The patient had previously been healthy and was developing normally. On physical examination upon admission, the following findings were noted: swelling of the right lower limb, mainly in the thigh region; the medial surface of the right thigh was warmer, erythematous, and tender on palpation [2-4]. There was also limited mobility of the right knee joint. The pulse in the dorsalis pedis and popliteal arteries of the right lower limb was weakly palpable, and the right lower limb, particularly the foot was cooler compared to the left. The circumference difference between the thighs and calves was approximately 10 cm and 6 cm, respectively.A Doppler ultrasound of the lower limb vessels revealed thrombotic changes in the right femoral vein, external and common iliac veins, extending up to the inferior vena cava, as well as in the popliteal and tibial veins [5].

Echocardiography and chest radiographs excluded pulmonary embolism.Laboratory tests revealed the following abnormalities: elevated levels of C-reactive protein, D-dimers, and fibrinogen;

decreased prothrombin index and reduced antithrombin III activity; and positive anti-SARS-CoV-2 antibodies (the patient had not been vaccinated against COVID-19). Based on the overall clinical picture, deep vein thrombosis of the pelvic veins and the right lower limb was diagnosed. Therapeutic doses of low-molecular-weight heparin were administered, resulting in clinical improvement, including a reduction in the circumference of the right lower limb, resolution of pain, and a decrease in laboratory markers of active thrombosis. On the 8th day of hospitalization, the patient was discharged home in good general condition with recommendations to continue low-molecular-weight heparin therapy, begin early mobilization, and attend follow-up in the Hematology Outpatient Clinic [6-7].

After three months, the treatment outcome was assessed. A follow-up ultrasound examination revealed partial recanalization of the vessels affected by the thrombotic process, as well as a decrease in D-dimer levels and inflammatory markers. Reduced antithrombin III activity persisted and was confirmed in three consecutive tests. A suspicion of congenital antithrombin III deficiency was raised and subsequently confirmed by genetic testing.

Other definite causes of inherited thrombophilia (protein C and protein S deficiency, factor V Leiden mutation, and prothrombin gene mutation) as well as acquired thrombophilia (antiphospholipid syndrome) were excluded.

Treatment with low-molecular-weight heparin was continued, initially at a therapeutic dose and later at a prophylactic dose. Due to the need for ongoing prophylaxis, a decision was made to switch to an oral anticoagulant-dabigatran.Due to the identified SERPINC1 gene mutation, diagnostic testing for congenital antithrombin III deficiency was performed in the patient's brother. Genetic testing confirmed the diagnosis, and his antithrombin III activity was 57% (Table 1) [8].

Parameter	At diagnosis	Low molecular weight heparin (therapeutic dose)	Low molecular weight heparin (maintenance dose)
Antithrombin III Activity	61%	52%	64%
C-Reactive Protein	160.95 mg/l	0.63 mg/l	-
Fibrinogen	5.99 g/l	2.47 g/l	2.33 g/l
D-Dimers	30027.0 ng/ml	653.0 ng/ml	579.0 ng/ml

Table 1: Comparison of Antithrombin III Activity, CRP, Fibrinogen, and D-Dimer Levels Depending on the Treatment Used

Summary

Congenital antithrombin III deficiency is a rare condition that most commonly manifests as venous thrombosis, which was observed in our patient. A potential trigger for the thrombotic event was SARS-CoV-2 infection. The unvaccinated patient tested positive for antibodies, and a detailed medical history revealed upper respiratory tract symptoms approximately three weeks prior to the event.In cases of congenital antithrombin III deficiency, lifelong thromboprophylaxis is recommended, both pharmacological and lifestyle-related (maintaining a healthy body weight, adhering to a balanced diet according to the healthy eating pyramid, engaging in age-appropriate physical activity, and avoiding additional risk factors such as stimulants).The patient remains under regular hematology follow-up, with dabigatran doses adjusted in accordance with his natural weight gain. After three years of treatment, ultrasound examination of the venous vessels showed complete recanalization. The patient has no symptoms of post-thrombotic syndrome, though a slight asymmetry in lower limb circumference of approximately one centimeter persists.

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