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Pulmonary Embolism Revealing Protein S Deficiency

Nezha Reguig, Rajae El Kilali, Rachida Zahraoui, Jamal Eddine Bourkadi, Mouna Soualhi

Abstract:
Pulmonary embolism (PE) is a serious condition that can be associated with inherited thrombophilia, including protein S deficiency—a genetic disorder impairing the anticoagulant pathway. We present a case of a 46-year-old patient with no significant medical history, admitted with acute-onset chest pain and hemoptysis. Imaging confirmed a distal pulmonary embolism, and subsequent thrombophilia screening revealed protein S deficiency. This case underscores the importance of screening for thrombophilic disorders in unexplained venous thromboembolism cases.

Keywords: *thromboembolism, Protein S deficiency, protein S activity testing, thrombophilia*

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I. Introduction:

Pulmonary embolism (PE) is a critical medical condition characterized by the obstruction of the pulmonary arteries. Among the myriad of risk factors contributing to the development of PE, inherited thrombophilia plays a significant role. Protein S deficiency, a rare but significant genetic disorder, impairs the natural anticoagulant pathway, thereby predisposing individuals to venous thromboembolism (VTE).

II. Case Presentation:

A 46-year-old patient, a 20 pack-year smoker with no other known medical conditions and no family medical history, was admitted to the pneumology department for right-sided chest pain evolving over 20 days. The pain was worsened by breathing and associated with minor hemoptysis, while the patient's general condition remained preserved. Clinical examination on admission was unremarkable, with SpO2 at 98% and a heart rate of 78 beats per minute. Chest X-ray revealed a heterogeneous opacity in the right lower base with blunting of the right pleural cul-de-sac (figure 1). The admission workup was unremarkable. Given the appearance of pneumonia, the patient was started on antibiotic therapy with amoxicillin-clavulanic acid and hemostatic treatment. After 7 days of antibiotic therapy, there was no clinical improvement, with persistent chest pain and hemoptysis. Due to this atypical evolution, a contrast-enhanced thoracic CT scan was performed, showing a distal right postero-basal pulmonary embolism (figure 2) with a necrotic pulmonary infarct focus in the posterior basal segment of the right lower lobe (figures 3 and 4).

Given the pulmonary embolism in a young patient with no risk factors, an etiological workup was requested. The transthoracic echocardiogram was unremarkable. The immunological panel, including antinuclear antibodies, anti-dsDNA, anti-Jo1, anticardiolipin antibodies, lupus anticoagulant, and anti Beta-2-glycoprotein antibodies, returned negative results. Cancer screening did not reveal any suspicious lesions, and bronchoscopic examination was also unremarkable. However, thrombophilia screening identified a protein S deficiency with an activity of 47% (reference value: 65%-130%). The patient was started on oral anticoagulants (Rivaroxaban). The clinical course was favorable, with a regression of symptoms and radiological resolution (Figure 5) .

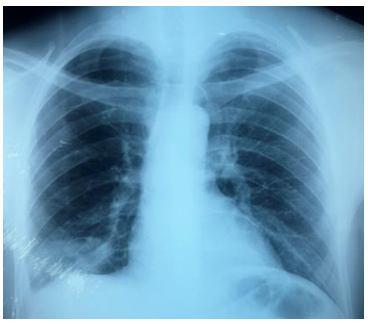

Figure 1: Chest X-Ray Revealed A Heterogeneous Opacity In The Right Lower Base With Blunting Of The Right Pleural Cul-De-Sac

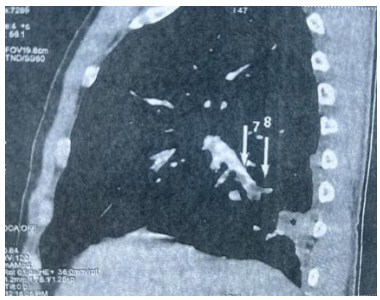

Figure 2: Sagittal Section Of A Contrast-Enhanced Chest CT Scan Showing Distal Right Postero-Basal Pulmonary Embolism

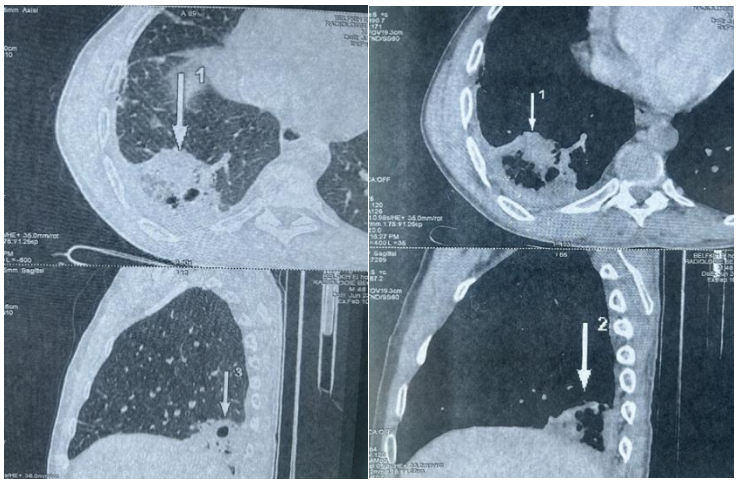

Figure 3,4 : Parenchymal And Mediastinal Transverse Sections Showing Necrotic Pulmonary Infarct Focus In The Posterior Basal Segment Of The Right Lower Lobe

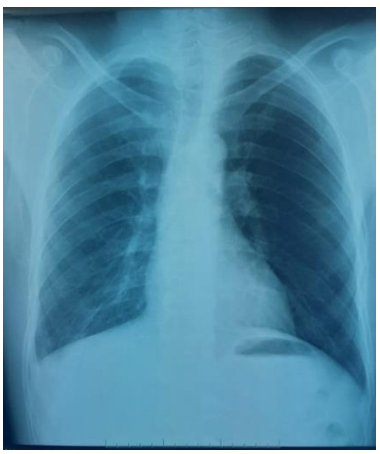

Figure 5 : Chest X-Ray One Month After Treatment Showing Radiological Clearing.

III. Discussion

Pulmonary embolism (PE) is a serious condition associated with a high risk of mortality that can be associated with inherited thrombophilia, including protein S deficiency.

Protein S (PS) is a vitamin-K-dependent plasma glycoprotein that acts as a cofactor for activated protein C (APC) in the degradation of factor Va and factor VIIIa. Additionally, PS directly inhibits factors Va and Xa [1,2]. Due to its anticoagulant properties, a deficiency in PS leads to a hypercoagulable state. As a result, hereditary PS deficiencies are often associated with recurrent venous thromboembolic events. This deficiency is implicated in 3-6% of unexplained thromboses in young adults, with a prevalence of 0.03% to 0.13% in the general population.

Protein S deficiency is caused by mutations in the PROS1 gene (3q11-q11.2) [3]

However, PS deficiency manifests variably in heterozygotes, with related individuals carrying the same molecular anomaly exhibiting a range of clinical presentations, from no thrombotic symptoms to recurrent thromboembolic events. The presence of associated risk factors can influence the expression of these anomalies. Homozygous PS deficiencies are rare and are linked to severe clinical manifestations, such as purpura fulminans, occurring from birth.[4]

Testing for Protein S (PS) deficiency is part of the etiological workup for venous thromboembolism. Current evidence indicates that screening for inherited thrombophilia is appropriate in cases of VTE without obvious cause < 45 to 50 years; VTE in patients with a family history of thrombosis; recurrent VTE; thrombosis at an unusual location; and developing VTE during pregnancy, use of oral contraceptives, or hormone replacement therapy [5]

The diagnosis relies on three biological tests: measuring the total plasma concentration of PS, the concentration of free PS, and its cofactor activity with activated protein C (APC). The PS activity assay has >90% sensitivity but only a moderate specificity of 40% to 70% [6,7], 2 PS activity assays can have interference (causing erroneous results) from a variety of substances or conditions that must be ruled out prior to testing.

In our case, thrombophilia screening was conducted as part of the etiological investigation for pulmonary embolism, given the age of our patient was 46 years old and that the etiological workup was unremarkable. The diagnosis of protein S deficiency in our case was confirmed through protein S activity testing.

IV. Conclusion

Protein S deficiency, is recognized as a risk factor for venous thrombosis and pulmonary embolism. Diagnosis involves assessing PS activity, crucial in thrombophilia workup. Screening is vital for early management of recurrent thromboembolic events