The Mystery Case of Chronic Thrombocytopenia in a Firefighter

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INTRODUCTION

We present a case of chronic unexplained thrombocytopenia and incidental thromboembolism in a patient with newly diagnosed COPD. He was admitted to the hospital for assessment of anticoagulant tolerance and workup for his thrombocytopenia.

CASE PRESENTATION

57 year-old non-smoker male recently diagnosed with COPD was undergoing outpatient workup for chronic thrombocytopenia. CT chest revealed bilateral pulmonary emboli, mild hepatic steatosis, gastric varices, and splenomegaly. He was admitted to the hospital for initiation of anticoagulation under medical supervision to assess tolerance given his chronic thrombocytopenia.

Hospital Course:

- Labs significant for chronic thrombocytopenia (50k) and elevated ALP and GGT.
- Lower extremity duplex: Extensive thrombus within the entire left femoral/popliteal veins
- Abdominal ultrasound: Negative for a portal vein thrombosis.
- Hepatitis panel, ceruloplasmin, lupus anticoagulant, and JAK-2 were normal.
- Anti-neutrophilic, anti-smooth muscle, anti-mitochondrial, and antiphospholipid antibodies were negative.
- AAT level was low at 33mg/dL and genotype ZZ with liver biopsy further confirmed a diagnosis of AAT deficiency.

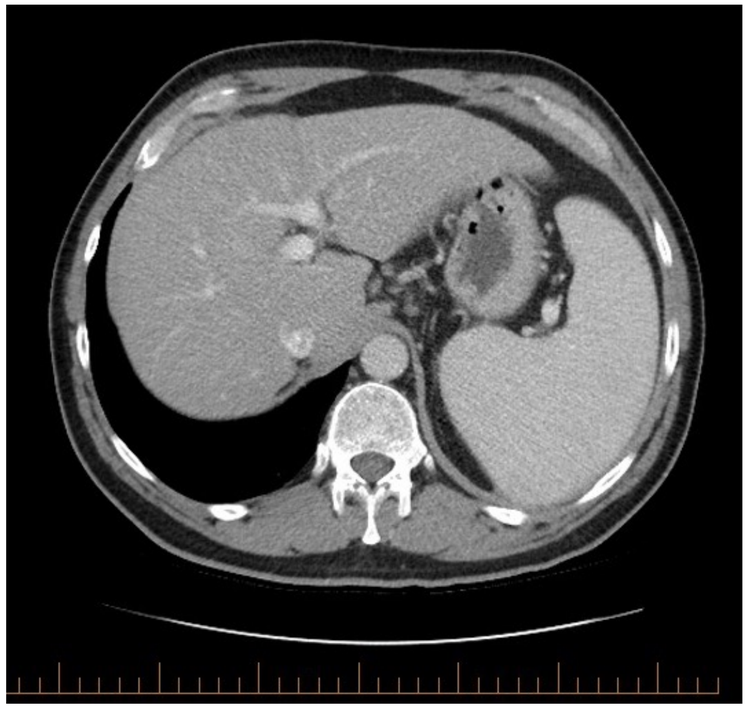

Figure 1. CT chest/abdomen showing splenomegaly in this image. It also revealed bilateral pulmonary emboli, emphysema, mild hepatic steatosis, and gastric varices.

CONCLUSION

The diagnosis of AAT deficiency is generally made after the identification of COPD or liver disease in a young adult or after the deficiency has been diagnosed in a family member. However, some patients may present with a secondary diagnosis as their presenting symptom which should prompt the identification of a unifying disease state. Our patient likely had thrombocytopenia secondary to his undiagnosed cirrhosis.

CONCLUSION cont.

Severe AAT deficiency in those homozygous for ZZ allele has been shown to cause an increased risk of thromboembolism compared to the general population. In the appropriate cohort of patients with the constellation of liver and/or lung disease and unprovoked thromboembolism, AAT deficiency should be part of the workup as the morbidity and prognosis are greatly changed with this diagnosis.

Our patient's case is an example of a diagnosis of alpha-1 antitrypsin deficiency. The case was further complicated by thrombocytopenia which makes treatment of his thromboembolism more difficult.

There have been no reported cases of AAT deficiency presenting with thrombocytopenia and concomitant thromboembolism. We propose the inclusion of AAT deficiency as part of the workup for such patients as the outcomes of the diagnosis are vast.

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