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CASE REPORT:- A RARE CASE OF RECURRENT PULMONARY THROMBOEMBOLISM DUE TO FACTOR V LEIDEN MUTATION

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ABSTRACT
Pulmonary thromboembolism is one of the most common life threatening conditions encountered in cardiorespiratory emergencies. Many risk factors predisposes the individual for the development of pulmonary thromboembolism. These risk factor can be broadly grouped under two broad categories i.e. Acquired and Inherited risk factors. In this case report, a case of Recurrent pulmonary thromboembolism due to Factor V leiden mutation which is an Inherited risk factor has been discussed. Factor V leiden mutation is most common inherited risk factor among the general population. Diagnosis of factor V leiden mutation requires the activated Protein C resistance assay and/or DNA analysis of theF5gene, which encodes the Factor V protein. Pulmonary thromboembolism due to factor V lieden mutation is treated with long term anticoagulants but Decisions regarding the optimal duration of anticoagulation are based on an individualized assessment of the risks for venous thromboembolism recurrence and anticoagulant related bleeding.

KEYWORDS: Many risk factors predisposes the individual for the development pulmonary thromboembolism.

INTRODUCTION
Pulmonary Thromboembolism is one of the most common life threatening conditions encountered in cardiorespiratory emergencies. From the clinical standpoint, DVT and pulmonary embolism (PE) can be considered a continuum of the same disease, and the two terms are often collectively referred to as venous thromboembolism.[1] Pulmonary embolism (PE) is an obstructive disease of the pulmonary arterial system commonly occurring due to the embolization of thrombus originating from the deep veins of the lower extremities.[2,3] It is one of the leading causes of mortality globally .The various factors, such as environmental, clinical and genetic, play a significant role for the pathogenesis of VTE/PE. So risk factor for venous thromboembolism are grouped into acquired and inherited risk factor. In this article our team is going to report a case of Recurrent Pulmonary thromboembolism due to an inherited risk factor i.e. Factor V leiden mutation.

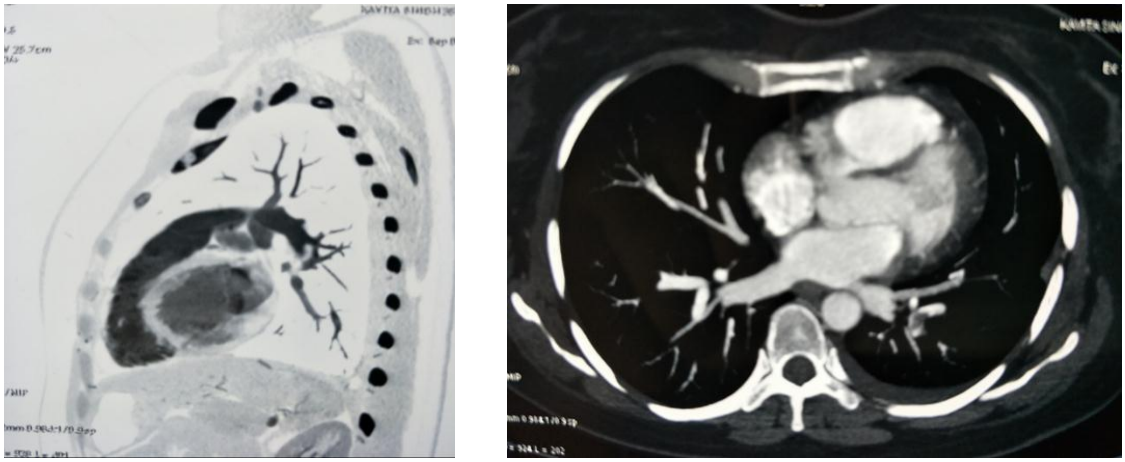
CASE REPORT
A 35 year old married lady presented with chief complain of sudden onset breathlessness, chest pain, and three episodes of hemoptysis in emergency department. She was Caucasian and from north India Gangetic plane. There was a past history of left side pleural effusion but etiology was in dilemma between the TB and Pulmonary embolism but ATT was started on trial basis on other center probably due to north India Gangetic plane is an endemic zone for TB and there is very high prevalence of TB in general population. But she doesn't respond to ATT and continued complaining pain in left lower coastal region on deep inspiration. There was no history of TB in any family member.

She did not have any acquired VTE risks, such as obesity, cancer, surgery, congestive heart failure/cor pulmonale, prior venous thrombosis, estrogen therapy, immobilization or recent long travel etc. There is no history of VTE in family as well. She has two children. There is no history of any abortion and both the delivery were spontaneous vaginal delivery. She was non diabetic, non hypertensive, non smoker, vegetarian. On admission, her blood pressure was 130/72 mmHg, HR= 116 bpm, SpO2 = 98%, Respiratory rate = 24/min. On examination there was decreased air entry on left side with crepts in upper part. Routine blood investigation (CBC, RFT, LFT, RBS, ABG) was within normal limit except for slightly reduced Pco2 level with mild alkalosis. Digital x ray chest shows left side mild pleural effusion. ECG shows right axis deviation. pleural fluid tapping was done and after doing routine microscopy fliud comes to be heamorragic, exudative, with low ADA level.

D-Dimer was significantly raised (>5000ìg/L). Echocardiography shows slightly dilated right ventricle and bilateral lower limb colour Doppler shows no sign of deep vein thrombosis. CT pulmonary angiography shows eccentric luminal filling defect in left inferior lobar, posterior basal and lateral artery s/o pulmonary thromboembolism with mild left pleural effusion and basal subsegmental consolidation.

An ordinary investigation of most factors related to thrombus formation (including protein C (88%), protein S(77%), antithrombin (112%), factor VIII activity (82%), cardiolipin IGG(11.84GPL/ml), cardiolipin IGM (4.42MPL/ml), anti B2 glycoprotein 1 IGG (<1.80RU/ml), anti B2 glycoprotein 1 IGM (15.22RU/ml), lupus Anticoagulant (absent), serum homocysteine (13.44ìmol/L) revealed that the blood concentrations of all such factors were within the normal reference range. Only Factor V Leiden mutation was detected.

Since she was hemodynamicaly stable, thrombolytic therapy was not recommended; therefore, anticoagulation therapy using low molecular weight heparin(LMWH) was initiated. LMWH was switched to oral anticoagulant; 5 mg daily administration of warfarin was initiated. At present she was on 5mg warfarin with INR between 2.5 to 3 with no symptoms.



CT pulmonary angiography showing eccentric luminal filling defect in left inferior lobar, posterior basal and lateral artery.

DISCUSSION

In clinical practice, factor V Leiden mutation and prothrombin gene mutation are the most common inherited conditions and account for more than half of the cases of inherited thrombophilia related venous thromboembolism; three other conditions (deficiencies in antithrombin III, protein C, or protein S) account for most of the remainder.[1] Factor V Leiden mutation was first described in 1993 by Dahlback and designated as a factor V Leiden mutation.[4] Factor V Leiden is the most common genetic risk factor for VTE, found in 20–25% of patients with VTE and 50% of patients with familial thrombophilia.[5]

Factor V Leiden thrombophilia is characterized by a poor anticoagulant response to activated protein C (APC) and an increased risk for venous thromboembolism (VTE).[5] It is the consequence of a single point mutation on the factor V gene (adenine for guanine) resulting in factor Va with diminished sensitivity to the natural anticoagulant effect of activated protein C.[1]

Deep venous thrombosis and pulmonary thromboembolism are two most common manifestion of factor V Leiden. Manten et al[6] conducted a study comparing the correlation of FVL with the different clinical presentations of venous thromboembolic disease. The result showed that the prevalence of the FVL mutation was lowest in patients with Symptoms of PE only and highest in patients with DVT only, whereas the prevalence was in between in patients with Symptoms of both mutation. In a patient without acquired risk factors, FVL was present in 22.7% of the patients presenting with DVT only, and in 11.5% of the patients with PE only.[7] Overall, the FVL increased the risk of PE by three-fold, and DVT by seven-fold in the carriers of FVL. Factor V Leiden is also associated with a 2- to 3-fold increased relative risk for pregnancy loss and possibly other obstetric complications such as preeclampsia, intrauterine growth restriction etc, although the probability of a successful pregnancy outcome is high.

In this case patient presented with Pulmonary Thromboembolism and there is no evidence of deep venous thrombosis. Patient also has two children without any obstetric complication in past. Diagnosis of Factor V leiden mutation requires the activated Protein C resistance assay (a coagulation screening test) or DNA analysis of the F5 gene, which encodes the Factor V protein. Patient of Factor V leiden mutation with Pulmonary Thromboembolism has good prognosis when treated with oral anticoagulant. Decisions regarding the optimal duration of anticoagulation are based on an

individualized assessment of the risks for venous thromboembolism recurrence and anticoagulant related bleeding. In the absence of a history of thrombosis, long term anticoagulation is not routinely recommended for asymptomatic Factor V Leiden heterozygotes, although prophylactic anticoagulation may be considered in high-risk clinical settings.[5]

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