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A Case of Cortical Vein Thrombosis and Deep Vein Thrombosis Due to Factor V Leiden Mutation,Lupus Like Anticoagulant and Protein S Deficiency

KEYWORDS: Deep Vein Thrombosis,Cortical Vein Thrombosis,Factor V Leiden Mutation.

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ABSTRACT *A patient who presented with recurrent venous thrombosis is reported following an episode of cortical venous thrombosis {CVT}.she was started on oral anticoagulant therapy,which she discontinued.she presented with deep vein thrombosis of both lower limbs and improved with oral anticoagulant therapy.Evaluation for hypercoagulable state revealed factor V leiden dutation, protein-S deficiency,lupus anticoagulant deficiency by polymerised chain method,.Long term anticoagulation has been planned.*

Introduction :
Thrombus formation requires an alteration in either the vessel wall or flow of blood or in the coagulation of blood. Alteration in cogulability resulting in hypercoagulable state is an important contributing factor for thrombosis. The significance of several primary hypercoagulable states as predisposing factors to hyperthrombotic states are now recognised as the single most common cause of heriditary thrombophilia.we report a patient who developed both deep vein thrombosis of lower limb and cerebral venous thrombosis,was evaluated for a hypercoagulable state and was detected to have Factor V leiden mutation.

Case Report:
A 23 yr old female presented to hospital with swelling both lowerlimbs with associated pain for the past 1 week. No history of fever,abdominal pain,jointpains,rash,breathle ssness,chestpain,bleeding per vaginum,weight loss or loss of apetite.patient is not a known alcoholic or smoker.History of cortical vein thrombosis present 8 months back for which patient was started on acitrom 3mg/day.But patient stopped 2 months back.No history of drug abuse.

On examination diffuse swelling of both lower limbs present with tenderness on palpation.No discharge or blebs formation.Abdominal,per vaginal and per rectal examination was normal.No obvious neurological deficit at present.

On investigating complete blood picture,blood urea,serum creatinine,serum electrolytes, liver function tests,USG abdomen were normal.Venous doppler of both lower limbs showed thrombus in external iliac veins in both limbs.Coagulation profile of the patient revealed Factor V leiden mutation,Protein S deficiency and lupus anticoagulant.patient CT Venogram done,CVT revealed.Straight sinus,Right transverse,Sigmoid sinus thrombosed extending in to right internal jugular vein.Patient was stented on anticoagulation with low molecular weight heparin 40 mg BD for 5 days and Tab.Acitrom 3mg OD daily.patient improved partially and discharged.

Discussion:[1]
This is a well-documented case of factor V Leiden mutation.This patient who had a peripheral venous system thrombosis as the first manifestation, was treated with anticoagulant therapy initially although the etiology of the thrombosis was not established. Venous thrombo embolism associated with factor V Leiden mutation can be protean and may range from a simple lower limb deep venous thrombosis to the Budd-Chiari syndrome[2].Cerebral venous sinus thrombosis is well known to occur in patients with factor V Leiden mutation[3].

In a case control study of 55 patients with cerebral venous thrombosis evaluated for factor V

Leiden mutation, eight (14.5%) were detected to have the same compared with 17 of 272 controls (6.25%).[4] It was also noted in this study that the recurrences of venous thromboembolic events were more frequent in patients with the mutation (5 of 8 patients, 62.5%) than in those without (8 of 47 patients, 17%; $p < 0.005$).[5]Other neurological syndromes known to be associated with factor V Leiden mutation include childhood ischemic stroke[5] and central retinal vein occlusion[6] .

The mechanism by which factor V Leiden mutation confers the risk of thrombosis deserves mention.Activated partial thromboplastin time of normal plasma is prolonged by addition of activated protein C (APC) that inhibits factors Va and VIIIa thus preventing the efficient generation of thrombin. Dahlback and colleagues showed that in patients with recurrent thromboembolism, addition of APC produced a much shorter prolongation of the clotting time. Thus these individuals were supposed to have resistance to the action of APC or APC resistance. This abnormal response to APC could be corrected in vitro by the addition of normal plasma and this capacity of the normal plasma to correct the APC resistance phenotype was identified to be a property of factor V thus suggesting that this factor was resistant to the APC cleavage[7].The molecular basis for APC resistance arises as a result of single G A mutation at nucleotide 1765 (CGA CAA) within the factor V gene. This mutation

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results in the replacement of the normal arginine at position 506 by a glutamine (Arg506Gln).[8] This mutation confers resistance on factor Va for cleavage by APC. This APC resistant, dysfunctional factor V molecule is known as factor V Leiden.

In conclusion, a patient with recurrent thrombotic events related to factor V Leiden mutation is reported.Deep venous thrombosis in the lower limb was followed by cerebral venous thrombosis resulting in quadriplegia. Awareness of the condition and a high index of suspicion may be required to detect the complications arising from the same and thus avert life-threatening

Complications[9].It is recommended that all patients less than 50 years of age presenting with spontaneous lower limb deep venous thrombosis are screened for factor V Leiden mutation and appropriately counseled and managed when the same is detected.

REFERENCE

1.JAPI july 2005;www.japi.org/july/2005/CR-642.pdf | 2. Denninger MH, Beldjord K, Durand F, Denie C, Valla D,Guillin MC. Budd-Chiari syndrome and Factor V Leiden mutation. Lancet1995;345:525–26 | 3. Dulli DA, Luzzio CC, Williams EC, Schutta HS. Cerebral venous thrombosis and activated protein C resistance. Stroke 1996;27:1731–33. | 4. Ludemann P, Nabavi DG, Junker R, et al.Factor V Leiden mutation is a risk factor for cerebral venous thrombosis: a case-control study of 55 patients. Stroke1998;29:2507–10 | 5. Ganesan V, Kelsey H, Cookson J, Osborn A, Kirkham FJ.Activated protein C resistance in childhood stroke. Lancet 1996;347:260. | 6. Scat Y, Morin Y, Morel C, Haut J. Retinal vein occlusion and resistance to activated protein C.J Fr Ophthalmol 1995;18:758–62. | 7. Dahlback B, Carlsson M, Svensson PJ. Familial thrombophilia due to a previously unrecognised mechanism characterized by a poor anticoagulant response to activated protein C:Prediction of a cofactor to activated protein C. Proc Natl Acad Sci USA1993;90:1004–08. | 8. Bertina RM, Koeleman BP, Koster T, et al.Mutation in blood coagulation factor V associated with resistance to activated protein C. Nature1994;369:64–67. | 9. Schulman S. Care of patients receiving long-term anticoagulant therapy. N Eng J Med2003;349:675–83. |