Case Report

Factor X Deficiency: A Rare Case of Inherited Coagulation Disorder Presenting as Hemarthrosis

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Abstract

Hemophilia due to factor VIII deficiency is most common inherited bleeding disorder in paediatric population. Hemarthrosis is initial manifestation in large number of patients. We are presenting a case of factor X deficiency who presented with knee hemarthrosis.

Key words: Hemarthosis, Factor X deficiency, Bleeding disorder.

Introduction

Hemarthrosis is classical presentation in Hemophilia A which is a genetic disorder of factor VIII deficiency.

However other coagulation disorders ususally do not present with hemarthrosis. Factor X deficiency is a very rare disorder of coagulation. Two patients at different places with the names of Ms.

Prower and Mr. Stuart were first to be diagnosed with factor X deficiency in the year 1956 and 1957 [1]. The incidence of severe factor X deficiency is 1 in 20,00,000 [2]. It is a vitamin K dependent serine protease that circulates in plasma.

Plasma levels are 8-10 µg/ml. Half-life in plasma is 34-40 hours. It plays a crucial role in the coagulation cascade. Factor X is activated either by factor VIIa/TF (tissue factor) complex via extrinsic pathway or by IXa/VIIIa complex via intrinsic pathway.

It is also activated by Russell Viper venom (RVV). Factor Xa subsequently forms a macromolecular complex with its cofactors Va, a phospholipid surface and calcium ions to convert prothrombin into thrombin [3-5]. One muslim boy who presented with hemartrosis was diagnosed with severe deficiency of factor X. Hemarthrosis becuase of severe factor X deficiency is rare presentation [6] which compelled us to report .

Case report

A 2 year old muslim boy was brought in paediatric OPD for complaints of swelling in right knee.

On further evaluation history of similar illness was confirmed in the past. Clinical examination confirmed hemarthrosis.

Systemic examination did not reveal any other significant findings. A provisional diagnosis of Hemophilia A was made.

The child was refered to department of hematology for confirmation of factor VIII deficiency.

Routine hematological tests were done which were found within normal limits. Platelet count, bleeding time, coagulation time, Prothrombin Time (PT) and Activated Partial Prothrombin Time (APTT) were done .

First stage analysis confirmed disorder of common pathway as PT and APTT both were prolonged.

The mixing experiments, Russell viper venom test and factor deficient plasmas were used to confirm the specefic factor deficiency.

Factor X deficiency was diagnosed. Activity of factor X was found to be less than 1% . The results are shown in Table no 1 and 2.

Case Report

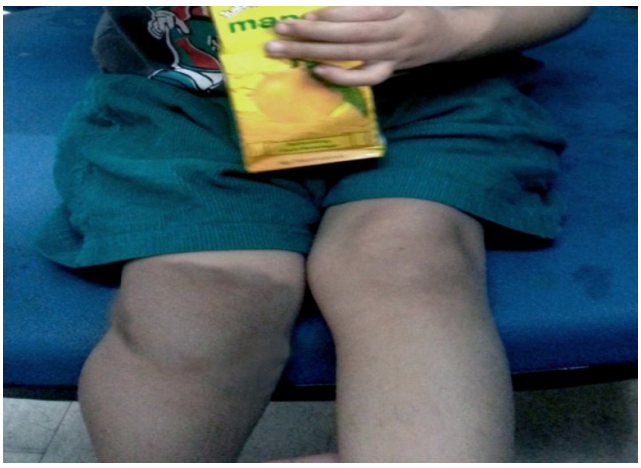

Fig 1: Hemarthosis in Right knee

Table 1: Results of hematological tests and coagulation profile

S No.	Name of Test	Control/Ref. range	Test results	Interpretation
1.	Bleeding Time (Ivy method)	3 to 10 minutes	6 minutes	
2.	Clotting time	2 to 6 minutes	15 minutes	prolonged
3.	Platelet count	100000 to 300000 /cmm	2.20 lacs/cmm	
4.	Prothrombin Time	11 to 16 seconds	80 seconds	prolonged
5	Activated Partial Prothrombin Time	32 to 40 seconds	150 seconds	prolonged
6.	Russell' s viper venom test	11 to 13 seconds	50 seconds	prolonged
7.	Fibrinogen	150 to 400 mg %	250 mg %	

Table 2: Mixing experiments and assay

S No	Mixing experiment done with	Result	Interpretation
1.	Adsorbed plasma	PT, APTT not corrected	Disorder of common pathway
2.	Fresh serum	PT , APTT corrected	Fibrinogen deficiency ruled out
3.	Normal plasma	PT , APTT corrected	Factor X deficiency
4.	Factor VIII deficient plasma	PT , APTT corrected	Factor VIII deficiency ruled out
5.	Factor IX deficient plasma	PT , APTT corrected	Factor IX deficiency ruled out
6.	Factor X deficient plasma	PT , APTT not corrected	Factor X deficiecy
7.	Factor X activity	Less than 1 %	Factor X deficiency

Discussion

Factor X deficiciency is commonly an inherited disease however it has occasionally developed in patients with liver diseases, vitamin K deficiency, amyloidosis, multiple myeloma, mycoplasma pneumoniae infection, leprosy and methyl bromide exposure [6-8]. The gene for factor X is located on chromosome 13q32 .The inherited deficiency is transmitted in incomplete autosomal recessive pattern. Clinically significant bleeding usually do not appear if activity of factor X is more than 10%. The homozygosity of transmission is usually seen in groups where consanguinity of marriages is common occurrence like musllims, jews. Severe deficiency of factor X has been seen in Pakistan where such marriages are common [9]. A variety of mutations result in defects involving either reduction in antigen or defect in one or more activation pathways [4]. Two types are described. Type-I, in which reduction in factor X activity parallels reduction in factor X antigen,

and type-II in which activity is less than antigen [10]. The factor X is required to convert prothrombin into thrombin which subsequently converts fibrinogen into fibrin. Factor X deficiency presenting as hemarthrosis is seen rarely.

Conclusion

Hemarthrosis is an usual presentation in Hemophilia A and B. Factor X deficiency should also be made as provisional diagnosis when consanguinity of marriage and particular ethnic group is noted. Prolonged PT and APTT should always be investigated further by mixing experiments to confirm factor X deficiency. The patient had factor X deficiency of severe variety presenting as hemarthrosis who is either Prower kindred (presence of antigen) or Stuart kindred (absence or reduced antigen).

Conflict of Interests

We certify that there is no conflict of interests for publication of this paper.

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