

Hereditary Coagulation Factor X Deficiency

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Stuart Prower factor (Factor X) deficiency is a rare hereditary autosomal recessive coagulation disorder. We have come across three cases in the course of last 20 years at our institute. These patients presented with prolonged bleeding after minor trauma, epistaxis, subcutaneous bluish black nodules and two of them presented with history of consanguinity in parents. Hematological findings in correlation with clinical manifestations revealed severe factor-X deficiency.

Keywords: *Coagulation disorder, Factor X deficiency, Stuart factor*

The rare disorder of factor X deficiency was described independently by Telfer, *et al.* (1) and Hougie, *et al.* (2), in Prower and Stuart Kinships respectively (hence the nomenclature Stuart Prower Factor).

Incidence in general population is approximately one in two million (3). In heterozygous state it remains asymptomatic, but in autosomal recessive condition it presents symptomatically (4) with bleeding manifestations, resembling moderate haemophilia (easy bruising, subcutaneous bleeding from mucous membranes and menorrhagia in

females). Marked deficiency may present as severe post-traumatic bleeding or even hemarthrosis. Cases with intracranial bleed have been reported (5). The optimum level of factor X required for hemostasis is 10% (6).

The laboratory diagnosis is based upon correction studies of PT and APTT with various plasma/serum reagents and specific factor X assays. Till now 50 cases have been reported in the world literature inclusive of Indian references (3,6-8).

Case Reports

Case 1

A 15-year-old Hindu boy presented with repeated episodes of epistaxis, gum bleeds and profuse bleeding after tooth extraction (the presenting complaint). There was history of prolonged bleeding after cuts and formation of bluish black nodules since age of 3 years. There was no history of similar complaint in maternal or paternal side.

Case 2

A 2-year-old Muslim child presented with severe epistaxis and bleeding from tongue bite. There was history of subcutaneous nodules and prolonged bleeding after minor trauma. Apart from bleeding manifestation there was no other clinical finding except for mild anemia. Family history revealed consanguinity (1st cousin). Earlier, a female sibling had died with similar bleeding manifestations.

Case 3

An 8-month-muslim male child born of consanguinous marriage (1st cousin) presented with severe bleeding manifestations in the form of subcutaneous nodules and spontaneous epistaxis. One of the female cousins of the patient died due to CNS bleed in early childhood.

The results of screening test, diagnostic test

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and factor assay of all the 3 cases are given in *Table-I*. The screening and diagnostic tests of coagulation suggested defect in common pathway; factor X deficiency. Factor assays revealed level to be 10% in case 1 and <1% in cases 2 & 3 (*Table-I*). The factor assay was performed using commercial factor X deficient plasma.

Collection, processing and analysis of the samples was done according to the standard method as described by Dacie & Lewis(9). All the samples were assessed in duplicate. Control values in comparison to the tests have been shown in *Table I*.

Discussion

Factor X deficiency is an extremely rare hereditary bleeding disorder with prevalence

of approximately 1 in 2 million in general population(6). The disease is more common in communities with social acceptability of consanguinity. Some of the large series have been reported from countries like Iran which form number of patients registered at National Hemophilia Centre London upto 1998(6).

Majority of these patients (56%) were with severe deficiency (<1%), 28% presented with moderate (1-5%) and only 15% of them were suffering from mild (6-10%) deficiency of factor X. In India there has been sporadic reports(7,8). These cases in Indian series presented with severe manifestation correlating with factor level <1%. In the present group of patients the clinical and hematological findings are identical with the previous case reports and add to the numbers of

TABLE I—Screening and Diagnostic Tests of 3 Cases

Screening Test	Diagnostic test correction study								Factor X assay*	
			Mixture of normal pool plasma and test plasma (1:1)		Mixture of aged serum and test plasma (1:1)		Mixture of adsorbed plasma and test plasma (1:1)			
	PT (secs)	APTT (secs)	PT (secs)	APTT (secs)	PT (secs)	APTT (secs)	PT (secs)	APTT (secs)		
Case 1	C:15 T:42	C:38 T:63	15	42	21	43	44	63	10%	
Case 2	C:15 T:140	C:38 T:126	16	50	20	60	145	125	<1%	
Case 3	C:13 T:130	C:40 T:185	14	45	13	60	142	185	<1%	
Remarks	Raised APTT along with raised PT: indicates defect in common pathway		Correction with aged serum indicates factor X deficiency and no correction with adsorbed plasma confirms the same.							

* The method used for factor X assay, was manual technique using commercial factor X deficient plasma (from Diagnostic Stago) as described by Dacie & Lewis(9) (Semi quantitative assay).

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cases. Also, clinically presenting bleeds with resemblance to mild/moderate hemophilias and with the background of consanguinity a possibility of factor X should always be kept in mind and analysed for.

Given the higher incidence of consanguinity in minority community, it shall be worth while to fully investigate for factor X in cases not other wise turning out to be Hemophilia A or Hemophilia B. In some cases where there is difficulty in diagnosis, correction of coagulation abnormality following vitamin K administration may also help in establishing the diagnosis of its deficiency. Not only it helps in establishing the diagnosis but also prevents erroneous labeling of vitamin K deficiency to a congenital factor deficiency state(10).

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REFERENCES

1. Telfer TP, Denson KW, Wright DR. A 'new' coagulation defect. *Br J of Hematol.* 1956. 2: 308-316.
2. Hougie C, Barrow HM, Graham JB Stuart clotting defect. I. Segregation of an hereditary hemorrhagic state from the heterogenous group heretofore called "stable factor" (SPCA, Proconvertin, factor VII) deficiency. *J Clin Invest* 1957; 36: 485-496.
3. Barik S, Budhraj A, Bhalla M, Diwan S. Inherited F X deficiency in two brothers. *Indian J Pathol Microbiol.* 1997; 40: 63-65.
4. Uprichard J, Perry DJ. Factor X deficiency. *Blood Rev* 2002; 16: 97-110.
5. Citak A, Uesel R, Karabocuoglu M, Unuvar A, Uzel N. A rare case of intracranial hemorrhage: factor X deficiency. *Pediatr Emerg Care.* 2001; 17: 349-350,
6. Peyvandi F, Mannucci PM, Lak M, Abdoullah M, Zeinali S, Sharifian R, *et al.* Congenital factor X deficiency: Spectrum of bleeding symptoms in 32 Iranian patients. *Br J of Hemat* 1998; 102: 626-628.
7. Mehta BC Agarwal MB. Inherited coagulation disorders in India. *Indian. J Pediatr* 1981; 48: 525-531.
8. Kashyap R, Saxena R, Choudhry VP. Rare inherited coagulation disorders in India: *Hematologia (Budap)* 1996 28: 13-19.
9. Dacie JV, Lewis SM. *Practical Hematology*, 6th edn. Edinburgh. Churchill Livingstone, 1984; p. 230-232.
10. Parmar RC, Bavdekar SB, Sahu DR, Kamat JR. Factor X deficiency and hemophilia A: Occurrence of two different coagulation defects in a family. *Indian Pediatr* 2001; 38: 183-186.