Case Report:
Coagulation Factor V Deficiency: Two Case Reports With a review of Literature

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Factor V deficiency is a very rare autosomal recessive trait. Commonest mode of presentation is bleeding from mucous membrane in the form of epistaxis or menorrhagia, later may be so severe as to be life threatening. We received two cases of factor V deficiency at the Pathology Department. of King Edward Medical College, Lahore. One child from paeditric surgery department. and other from Lady Willingdon Hospital, Lahore. Both were investigated, all the tests were normal but prothrombin time and activated partial thromboplastin time was prolonged which was corrected with adsorbed plasma. The diagnosis was confirmed by factor V assay.
Key Words: Factor V, Coagulation defects, clotting factors.

Factor V deficiency was discovered in 1944 by Owren[1]. It is extremely rare autosomal recessive trait, factor deficiency is demonstrated by homozygotes and is mild to silent in heterozygotes.[2] Easy bruising, spontaneous epistaxis and menorrhagia are the most common symptoms[3,4,5]. Menorrhagia may be so severe as to be life threatening, post partum haemorrhage may be frequent.[6] Severe bleeding is also reported after surgical procedures or dental extractions. Pt and APTT are prolonged, but thrombin time is normal. Bleeding time is prolonged in one third of cases.[7] Diagnosis of this disorder is confirmed by factor V assay.[8]

Case Reports:

Case I:

A male child, 3 years of age presented with post-circumcision bleeding which lasted for 3 days and required two pints of fresh blood to be transfused. On interrogation, there was a history of prolonged bleeding from minor cuts and wounds.

Investigation:

Hb - level was 8 g/dl., Total leucocyte count 8.6×10^9/l, DLC was normal. Reticulocyte count was 4%. Bleeding time was 4 minutes 15 seconds (Ivy's method) He had a platelet count of 160×10^9/l. Coagulation time was prolonged and was 12 mins. and 45 sec. (Lee and White method). His prothrombin time was 30 seconds with a control of 15 seconds. APTT was 65 seconds with a normal control of 30 sec. Thrombin time was normal Mixing experiment was done with adsorbed plasma. Both PT and APTT were corrected (Dacie and Lewis). Factor V assay was done and factor V level was 5U/dl.

Case II:

The second patient was a female, 17 years of age, who presented with menorrhagia for last 2 years and frequent expistaxis. For menorrhagia she had to be transfused thrice. History of haematoma formation was there since childhood.

Investigations:

On complete blood examination, her Hb level was 6g/dl, T.L.C. was 4.8×10^9/l, with a normal differential count. Reticulocyte was 3%.

She had a platelet count of 150×10^9/l, and a bleeding time of 5 mins. (Ivy's method). Coagulation time was 13 min and 30 seconds (Lee and White method). Her prothrombin time was 26 seconds as compared to normal control of 14 seconds. APTT was also prolonged and it was 55 sec. with normal control value of 25 seconds.

Both PT and A.P.T.T. were corrected with 0.1 vol of adsorbed plasma. (Dacie and Lewis) Coagulation Factor V assay was done using factor V deficient plasma (Dade). Factor V level was 7 u/dl.

Discussion And Conclusion

Factor V deficiency is an extremly rare autosomal recessive trait[2]. In our study as well over two years period, there were only 2 (2%) cases of factor V deficiency out of a total of 100 bleeding disorders with congenital coagulation defect. One was three years old male child, whereas the other was a girl of 17 years. The male child presented with post-circumcision bleeding for last 3 days. It was also accompanied with history of profuse bleeding from minor cuts and wounds since birth, and that was the reason for delay in circumcision. The female patient had a history of menorrhagia for the last 2 years. She also complained of recurrnt epistaxis.

Hougie[5] (1977); and Wintrobe et al[9] also observed the similar findings and conclude that bleeding following surgery of any type may be the first sign of disease, whereas epistaxis is usually severe.

Rizza[4] (1976) found a prolonged bleeding after minor trauma as significant feature in his cases. In few studies,[3,4,5] menorrhagia is reported to be the most common symptom and life threatening also.[6]

Our female patient also presented with severe menorrhagia for which she required frequent transfusions.

On investigations all the other tests were normal except PT and APTT which were moderately prolonged, however the bleeding time was normal in our cases. Prolongation of bleeding time has been reported in 1/3rd of patients of one study[2] Factor V deficiency was confirmed by doing factor assay which in our cases was mild to moderate in severity. (Factor V level, 5 u/dl and 7 u/dl). Factor V level correlated with the severity of the symptoms in our cases.

Conclusions

In a female patient with menorrhagia possibility of factor V deficiency should be considered. It is recommended that every male child undergoing surgery including circumcision should undergo PT, APTT and Platelets count as coagulation screening instead of bleeding and clotting time

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