CASE REPORT

A RARE CASE OF GLANZMANN THROMBASTHENIA

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ABSTRACT
Recurrent Epistaxis often is due to thrombocytopenia. Child with recurrent Epistaxis had abnormal bleeding time, coupled with normal platelet count congenital Glanzmann Thrombasthenia (GT) though rare is a possible condition. Severe bleeding in GT required platelet transfusions, and whole blood (as replacement for gross blood loss). Suspect Glanzmann Thrombasthenia, when recurrent Epistaxis with prolonged bleeding time is associated with normal platelet count. Mild bleeding is best treated with local and IV Tranexamic acid (30 mg / kg).
Key Words: Epistaxis; Glanzmann's Thrombasthenia; Recurrent Epistaxis

Introduction

Glanzmann Thrombasthenia is rare inherited autosomal recessive disorder. This disorder was first described by Dr. Eduard Glanzmann in 1918. He aptly described the abnormalities as, abnormally prolonged bleeding time, with normal platelet count, and poor aggregation of platelets and peripheral smear showing platelets seen in singly.[1]

Glanzmann Thrombasthenia is a genetic platelet disorder in which platelet glycoprotein αIIb/β3 the major integrin complex (fibrinogen receptor) is either deficient or present but dysfunctional.[2] Platelet glycoprotein αIIb/β3 complex levels of <5 % leads to severe bleeding tendencies, called as Type 1, and 10-20 % levels of the platelet glycoprotein αIIb/β3 complex lead to mild to moderate bleeding episodes (Type 2). The genes of both these platelet glycoprotein αIIb/β3 complex are on chromosome 17. 50 % of activity of each protein (seen in heterozygote state) is enough to support normal platelet aggregation.[3] Defect in the GP IIb /IIIa complex leads to defective platelet aggregation and subsequent bleeding.

Case Report

A nine year old male child born out of a consanguineous marriage was admitted for evaluation of recurrent episodes of bleeding from nose from four years. The bleeding episodes used to occur once in two months mostly from left nostril. It was accompanied with black vomitus (digested blood) and melena during bleeding episodes. The child was evaluated for bleeding and the further specific tests were done outside.

Investigations showed the following results: Total Count – 8200 cells/cumm (N-67% L-28% M-2% E-3%); Haemoglobin – 11.0 g%; Red Cell Indices – MCV: 81 fl, MCH: 27.3 Pg; MCHC: 33.6%; Platelet Count – 3.34 lakhs /cumm; PCV – 32 7%; Bleeding Time – 4.45 min (Normal 1-3 min); Clotting Time – normal 5.45 min (3- 8 min); APTT – 28 Sec (Control- 26-36 Sec); PT – 15 Sec (Control - 12-14 Sec); Fibrinogen Assay – 238 mg% (Normal value- 200-440 mg %). Whole blood platelet aggregation (PAGS) studies: (1) ADP activation – 2 U (Normal value: 53-122); (2) Arachidonic acid activation – 0 U (Normal value: 74-136); (3) Ristocetin – low sensitive for VWD type ii b: 1 IU (Normal value 1-34 IU); (4) Fibrinogen assay – 238 mg% (Normal value: 200-440); (5) Factor xiii – clot solubility: patient: stable clot; (6) Factor VIIIc assay – 200 % (normal 70-180). Ristocetin aggregation is normal indicating Glanzmann Thrombasthenia. Ristocetin aggregation will be abnormal in Bernard Souvelier Syndrome (Giant Platelet Syndrome).

Treatment

On admission the patient was bleeding profusely and immediate Adrenaline, and Tranexamic acid nasal packing(anterior and posterior) was done on admission and one unit of platelet concentrate was given , this was followed by intravenous fluid replacement for volume loss, routine antibiotic cover and tranexamic acid intravenous , following which bleeding was controlled .

On day two 15ml/kg of whole blood was given in view of prior acute blood loss, and was repeated on day three. Nasal packing was removed and new pack introduced on day four three hours after removal of nasal packing patient again had second episode of bleeding since

admission which was controlled on platelet concentrate administration and tranexamic acid both iv and local application along with anterior and posterior nasal packing. The patient did not have any further bleeding episodes in next two days and was discharged with advice that the patient has to avoid any strenuous and physical activity which may lead to bleeding including sports, he was also advised not to take any NSAIDs - Aspirin etc .Prognosis was explained that as age advances the disease severity will decrease. As Comparative to a case of Von-Willebrand Disease the prognosis of Glanzmann Thrombasthenia improves with age.

Discussion

Glanzmann's Thrombasthenia is a hereditary platelet dysfunction due to quantitative or qualitative defect in the platelet glycoprotein αIIb/β3 the major integrin complex which leads to inability of plate aggregation by attachment to fibrinogen, leading to non -formation of platelet plug, and thus excessive, apparently spontaneous bleeding. Usually the presentation is as Epistaxis, bleeding gum, or petechiae. Haematuria and GI bleedings are uncommon. Adolescent and adult females may present first time as menorrhagia. Good oral hygiene can prevent bleeding gums, and combination contraceptive pills may reduce or prevent menorrhagia. Local and systemic tranexamic acid helps to stop mild Epistaxis.

Recombinant Human- activated factor VII (r FVIIa, novo seven, novo nordiskis) used effectively in some Glanzmann's Thrombasthenia cases with severe bleeding, not responding to many units of platelet transfusion, at a dose of 120 mg/kg infusion.[4] Allogenic bone marrow transplant done in some severe cases of GT has led to normalization of bleeding time, and normal platelet glycoprotein α IIb/β3 complex levels.[5] Ankaferd Blood Stopper, a new nanotechnology local application formulation based on herbal product, is found to be effective in Warfarin treated animals to stop bleeding from injury. Its efficacy in control of local bleeding due other causes like GT is yet to be proved.[6]

Mild Bleeding Epistaxis best Treated with Tranexamic Acid, oral / injectable (30 mg/kg) with local packing. Avoid platelet transfusion as it may lead to platelet antibody development and make it difficult to treat later. Severe Bleeding required platelet transfusion and Recombinant Human- activated factor VII administration.

Conclusion

A nine year old male child admitted with history of recurrent bleeding episodes with prolonged bleeding time. The Prothrombin time, thromboplastin time and clotting time were normal. Platelets were adequate in numbers and were seen singly. On further investigations such as platelet aggregation study, ADP, Arachidonic acid tests which turned out to be abnormal. The patient also had normal platelet morphology ruling out Bernard Soulier Syndrome and normal Ristocetin low assay confirmed Glanzmann Thrombasthenia (ruling out Von Wille-Brand Disease,). As there was severe bleeding it required multiple platelet concentrates, along with nasal packing and IV Tranexamic acid.

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