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Research Article

A RARE CASE OF GLANZMANN THROMBASTHENIA

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A R T I C L E I N F O

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A B S T R A C T

Glanzmann's thrombasthenia is a rare congenital bleeding disorder, in which there is impaired or absent clot retraction with defective platelet aggregation. Patients usually present with mucocutaneous bleeding and excessive bleeding associated with trauma and/or surgery. Mostly, the disease presents with mild hemorrhage but If bleeding is severe enough, it will be life-threatening. Thrombasthenic Platelets are normal in number and morphology. Patients have aprolonged bleeding time, with deficient platelet aggregation. Basic biochemical basis for GT is quantitative or qualitative defect of GPIIb-IIIa also known as integrin αIIbβ3. Platelet Transfusion may shorten the bleeding time for a brief period. In few GT patients hemorrhagic symptoms have been earnest for the necessity of bone marrow transplantation. With supportive care hemorrhage can be managed but the course tends to be highly variable.

INTRODUCTION

Glanzmann's thrombasthenia was first described in 1918 by Dr. Eduard Glanzmannin heterogenous group of patients with bleeding disorder associated with reduced clot retraction. It is an autosomal recessive disorder where normal platelet count, lengthen bleeding time, and defective invitro platelet aggregation.[1] Because of this inadequacy of platelet function, it manifests as a bleeding disorder characterized by mucocutaneous hemorrhage of varying severity. The genesfor GPIIb-IIIa are unremarkable and placed on long arm of chromosome no 17(17q21-22). The incidence is about 1 in 1,000,000 with an equal sex predilection and association of consanguity has been reported.[2]

CASE REPORT

A 14 year old female patient present with recurrent gum bleeding on and off since 5 years of age and a long clinical history of easy and spontaneous bruising. There was positive history of repeated episodes of epistaxis and purpura. Other clinical manifestations, like hematuriawas absent in our patient.Her treatment history states that, she has been taking iron supplements and tranaxemic acid for the past 3 years, and three units of blood has been transfused. She was born to third degree consanguineous parents and there was no definite family history of similar illness. She was ectomorphic and poorly nourished. On examination, She was conscious, cooperative, and well-oriented. Pallor was noted in eyes and nailbeds of hands.

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During Inspection, ecchymotic patches on both arms and wrist region present. Examination of liver and spleen showed no abnormality. On oral examination, her gingival status showed normal appearance of gingiva with areas of bleeding along the marginal region. Generalized stains were seen on the surfaces of the teeth. gingivitis with normal periodontal status was considered. Routine laboratory investigations revealed hemoglobin (5 gm/dl), platelet count (75,000/mm3) (Mild platelet anisocytosis few giant platelets &anisogranularity on Peripherl smear), BT: >15 min, Platelet factor: 18.6% (reference 1.1-6.1%), Platelet function assay: 200, Collagen/ADP: >285 sec(reference range 62-100 sec), Collagen/Epinephrine: >300 sec(ref. range: 82-500 sec), Clot retraction: Absent. Plasma Coagulation Profile was under normal limits. Factor Assays include Factor VIII: 138.6% (ref. range 50-150%),Fibrinogen:247.1mg/dl(ref range 150-450 mg/dl), Ristocetin Cofactor Assay (Vwf:RCo): 381.0%(ref range 50-175%), Platelet aggregometry include Ristocetin: (1.5mg/ml): Aggregation followed by disaggregation, ADP(10.0mu.M): Absent Response, Collagen (2.0mu.g): Absent response, Expression of CD41 on flow cytometry: Platelets gated on FSC vs SSC using CD42b shows absent expression on CD41. Based on the history, clinical findings, and investigations, final diagnosis of Glanzmann's thrombasthenia with microcytic hypochromic anemia was made. The differential diagnosis in patients with mucocutaneous bleeding, prolonged bleeding times includes von Willebrands disease and
Bernard-Soulier Syndrome.

DISCUSSION

Glanzmann's thrombasthenia is an inherited autosomal recessive bleeding disorder characterized by a defect in the

platelet integrin $\alpha_{IIb}\beta_3$ (previously known as GP IIb/IIIa). Genetic Error in Integrin $\alpha_{IIb}\beta_3$ can repress the synthesis of functional receptor. This cause scarcity of fibrinogen receptor and defective fibrinogen binding after platelet activation.[8]As this protein is mandatory for platelet aggregation, it is found to be defective or absent. George *et al.,*[3] divided Glanzmann's thrombasthenia in-to three groups as:

Type- I: patients with less than 5% of GpIIb-IIIa
Type- II: patients with 5%-20% of GpIIb-IIIa
Type- III (variants): normal amounts of GpIIb-IIIa, but functionally inactive.

Hemorrhagic symptoms occur only in homozygous patients for GT mutations, while the heterozygous is asymptomatic. [9]The common bleeding sites are purpura, epistaxis, gingival haemorrhage, and menorrhagia, whereas gastrointestinal bleeding and hematuria are less common but with dangerous complication. Bruising typically occurs following minor trauma.[3] The laboratory tests of this patient showed prolonged bleeding time, decreased or absence of clot retraction, and abnormal platelet aggregation responses to physiologic stimuli; all stood positive for this patient.[3] The spontaneous bleeding episodes from the gingiva in the oral cavity[4] and ecchymotic patches on the skin further confirmed the medical condition in this patient.The differential diagnosis includes von Willebrand's disease, Bernard- Soulier syndrome, and platelet secretory defects.[5],[6] Normal ristocetin-induced platelet agglutination and normal platelet size clearly rule out Bernard-Soulier syndrome, a disorder of platelet adhesion. Inherited thrombocytopenias are eliminated by a normal platelet count.[7] Normal coagulation parameters rule out clotting disorders that can also affect platelet function such as congenital afibrinogenemia and von Willebrand disease. Carrier detection in GT is important to control the disease in family members. Acquired thrombasthenia must be eliminated in the absence of a family history of the disease.[7]

The use of recombinant factor VIIa and other hemostatic agents is an alternative approach for cessation of bleed, especially who have developed antibodies or having history of transfusion refractoriness. According to literature, a cure for the disease does not exist; the only effective therapy consists of transfusions of fresh platelets or platelet concentrates.[5]Localized bleeding can be managed by fibrin sealents and antifibrinolytic agents. Epistaxis and gingival bleeding can be controlled by nasal packing or gel foamed soaked in topical thrombin.

Dental hygiene lessens the gingival bleeding. Avoidance of antiplatelet agents. High doses of progesterone followed by oral contraceptive pills for severe menorrhagia. Repeated platelet transfusion produce HLA specific alloantibodies or GPIIb-IIIa specific isoantibodies, which complicate the transfusion therapy and limit the future treatment. Antibodies can be successfully removed by immunoabsorption with the use of protein-A sepharose columns which may transiently restore platelet efficacy.[10]This method requires several hours, so not for active bleeding. Allogenic marrow transplant has been reported in two patients successfully with Glanzmann's thrombasthenia.

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