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Case Report *Volume 6 Issue 1*

Glanzmann Thrombasthenia as a Rare Cause of Menorrhagia, A Case Report with Review of Literature

Manjari BH*, Naimisha M, Kavitha B and Sneha G

Department of Obstetrics & Gynecology, Mamata Medical College, India

***Corresponding author:** Dr. Basanta manjari Hota, Department of Obstetrics & Gynecology, Mamata Medical College, Khammam, Telangana State, India, Pin: 507002, Tel: +91 7893305290; Email: drmanjarihota@gmail.com

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Abstract

Glanzmann Thrombasthenia is a rare autosomal recessive disorder characterized by recurrent episodes of spontaneous or traumatic bleeding starting from the first decade of life. Affection of the gene located in long arm of chromosome-17 prevents platelet agglutination but the morphology and count remains normal. It is common in female, certain races and offspring in consanguineous marriage. As it may be life threatening in certain cases, should be considered in bleeding disorder including menorrhagia with normal platelet and coagulation profile but high bleeding time and defective clot retraction. Anti fibrinolytics (Tranexamic acid), blood and platelet transfusion are the treatment in bleeding episodes. Combined oral contraceptive pills help in menorrhagia. We report a case of Glanzmann Thrombasthenia variant type with history of recurrent bleeding from the age of six, reported to the gynecology outpatient department of Mamata Medical College, Khammam, Telangana state, India at the age of 18 years with menorrhagia and severe anemia. She was diagnosed at the age of nine years by platelet Aggregometry and flow cytometry in addition to tests for other causes of abnormal bleeding. As reported with severe anemia she was treated with blood transfusion, iron and folic acid, Vitamin C and combined oral contraceptive pills. Though a rare and life threatening bleeding disorder, prognosis is good with early diagnosis and adequate management.

Keywords: Thrombasthenia; Platelet; Hereditary; Bleeding; Menorrhagia

Abbreviations

CNS: Central Nervous System; CVS: Cardiovascular System; RS: Respiratory System; BT: Bleeding Time; CT: Clotting Time; MCH: Mean Corpuscular Hemoglobin; MCHC: Mean Corpuscular Hemoglobin Concentration; PT: Prothrombin Time; APTT: Activated Partial Thromboplastin Time; TT: Thrombin Time; RicoF: Ristocetin Co Factor; vWFAg: Von Willebrand Factor Antigen; FSC: Forward Scatter; SSC: Side Scatter; µm:Micromole.

Introduction

Glanzmann Thrombasthenia (GT), a rare autosomal recessive genetic bleeding disorder was first reported by Dr. Eduwad Glanzmann in 1918 [1,2]. Prolong bleeding time and abnormal clot retraction with normal or low platelet count but defective platelet aggregation is the reason behind the hemorrhage [3]. Incident is one in one million with female (60%) predominance [4]. Consanguineous marriage contributes to the incidence [5]. It is common in ethnic

groups like South Indian Hindus, Iraqi Jews, French Gypsies and Jordanian nomadic tribes [6]. The clinical presentation varies from minimal bruising to fatal hemorrhage. Though bleeding gum, purpura, epistaxis and Menorrhagia are usual hemorrhagic presentations, visceral bleeding, hematuria, hemarthrosis and spontaneous intracranial hemorrhage are also reported. In spite of being a rare disease it is significant for life threatening bleeding in both spontaneous, following trauma and surgical procedures. We present a case of menorrhagia with severe anemia in a known case of GT for awareness and statistical record.

Case Report

A 18 year old unmarried girl, known case of Glanzmann Thrombasthenia reported to the gynecology Out Patient Department of the institution with menorrhagia, generalized weakness, palpitation and giddiness for 10 days. She started taking combined oral contraceptive pills two days back and bleeding stopped. Her menstrual cycles were 10-12 days over 30-60 days with heavy flow for about 10 days since menarche which she attended at the age of 12 years and last menstrual period was 10 days back. She developed recurrent episodes of epistaxis, gum bleeding from the age of six years and was investigated in Christian Medical College Vellore at the age of 9 years. Her report showed normal platelet count and morphology with prolonged bleeding time and normal PT, APTT, TT and Fibrinogen. Factor VIII, IX, XI were normal. RiCoF and vWFAg was normal for her blood group. Platelet Aggregometry was Ristocetin (0.5mg/ml) : Absent Response, ADP (10.0 µm): Absent Response, Epinephrine (10.0 µm): Absent Response, Collagen (2.0 µg): Absent Response, Arachidonic Acid(10.0 µm): Absent Response, PFA-200: no closure, Collagen/ADP: >223sec (normal), Collagen/Epinephrine: >239 sec (ref 82-150 sec), Expression of CD41 on flow cytometry: control- platelets gated on FSC vs SSC using CD42b showed 100 % expression of CD41 and Patient: platelets gated on FSC vs SSC using CD42b showed 100% expression of CD41. The impression was: Platelet function Disorder. Aggregometry is suggestive of Glanzmann Thrombasthenia- variant. She was advised to avoid intramuscular injection, non-steroid anti-inflammatory drugs and consult hematologist before any surgery. Local or systemic Tranexamic acid in bleeding episodes (systemic use is contraindicated in hematuria) in addition to pressure to stop bleeding and consult physician in persistent cases. The patient was counseled to report to the hospital on attending menarche to prevent and treat menorrhagia.

Her parents were not consanguineous, neither her brother nor any of her family members had this bleeding disorder. She is a student in Higher Sec Education, not allergic to any medication, no history of any other major illness and taken eight units of blood transfusion till date.

On examination patient was thin built with BMI- 16.4 Kg/M^2, severe pallor, no icterus / lymphadenopathy / thyromegaly/ pedal edema and vital were normal. CNS, CVS, RS and Abdomen were clinically normal. Investigation showed hemoglobin of 04.6 gm%, TRBC-02.64mil/ cmm, MCH-17.0 pg, MCHC-26.4%, Platelet-01.6 La/ cmm, peripheral smear was microcytic hypochromic with anisopoikilocytosis and normal WBC. Her BT, CT, PT, APTT were within normal limit. Abdominopelvic ultrasonography was within normal limit. She was treated with two unit s of packed RBC transfusion, oral iron and folic acid, Vitamin C, combined oral contraceptive pills and advised for whole blood transfusion but got discharged on request to report latter.

Discussion

GT is characterized by prolonged bleeding time, abnormal platelet aggregation and defect in clot retraction. Pathophysiology of this rare bleeding disorder due to platelet malfunction is mutation of gene located in long arm of chromosome 17 that codes for Glycoprotein (GP) II β/ III α membrane receptor on platelet. This receptor binds with fibrinogen, vitronectin and fibronectin, for platelet aggregation which is the first step in clot formation [5,7]. Affection in either or both GP II β and III α results in same level of platelet dysfunction. Type -1 (Severe) has <5% of normal , type – II (moderate) has 10-20% of normal and type III(mild or variant type) with 50-100% normal GP II β /III α level but malfunctioning platelets [8]. Though these subtypes are categorized by level of GP II β / III α, clinical presentation rarely correlate with this [7]. Flow cytometry and DNA analysis can detect carrier state and level GP II β / III α by monoclonal antibody and detection of platelet antigen may help in prenatal diagnosis of type -1 GT and heterozygous state [1]. Common presentation of GT is recurrent bleeding episodes, either spontaneous or following injury starting in first decade of life [9]. Our patient developed recurrent epistaxis and gum bleeding at the age of 6 years, a South Indian Hindu and female by sex.6 She does not have a family history of such bleeding disorder and her parents are non-consanguineous [4-6]. Light transmission aggregometry, though time taking is the gold standard for diagnosis.4 History, hematological investigations including bleeding and clotting time, coagulation profile, clot retraction test and Platelet function analyzer flow cytometry are done for the diagnosing the condition. Von Willebrand disease and Bernad Soulier syndrome are considered for differential diagnosis [7]. Prevention of this bleeding disorder includes avoidance of consanguineous marriage and use of Non steroid anti-inflammatory drugs. Fibrinolytic inhibiters, topical and oral/ Injectable as the

need may are helpful as treatment. Platelet transfusion prior to operative procedures and may be repeated as and when required. Though Platelet alloimmunization against HLA group and/or GP II β / III α glycoprotein is reported it is not a contraindication [1]. Recurrent blood transfusions may be needed depending on frequency and severity of hemorrhage. Our patient had 08 units of whole blood transfusion over a period of 09 years. Vaccination against Hepatitis B is mandatory to prevent the infection. Regular use of combined oral contraceptive pills help to prevent and treat menorrhagia. Our patient being diagnosed at the age of 09 years, was advised to review after menarche for this purpose. As she was not taking the drug regularly she had menorrhagia and severe anemia. Recombinant Factor VIIa has been successfully used in preventing and treating the bleeding not responding to platelet transfusion in GT [10]. There are reports of allogenic bone marrow transplantation and cure of disease in rare cases [11]. Gene therapy and stem cell transplantation as cure for this bleeding disorder are in experimental level.

Conclusion

Glanzmann Thrombasthenia is a rare but dangerous inherited bleeding disorder which may be life threatening at times. Suspicion of the condition in a patient with history of bleeding disorder with normal platelet count and morphology, abnormal clot retraction, increased bleeding time and normal coagulation profile are the key to its diagnosis by more sophisticated tests. Prognosis is good with early diagnosis and adequate management of Glanzmann Thrombasthenia.

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Conflict of Interest

Nil

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