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Glanzmann's Thrombasthenia: A Rare Cause Of Abnormal Uterine Bleeding

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ABSTRACT
Glanzmann thrombasthenia is a rare inherited autosomal recessive bleeding disorder characterized by abnormal platelet aggregation and clot retraction. Women with GT may experience prolonged, heavy, or irregular menstrual bleeding, especially at menarche. This bleeding can be difficult to manage and may not respond to treatments like packed red blood cells, platelet concentrates, or recombinant activated factor VII (rFVIIa). We are presenting a case of a 15year old girl presenting with Glanzmann thrombasthenia with complaints of heavy menstrual bleeding. As such, the management of these individuals continues to be complicated. The only effective treatment for GT is still bone marrow transplantation, however gene therapy is being investigated more and more as a potential future option.

INTRODUCTION

Glanzmann thrombasthenia is a rare inherited autosomal recessive bleeding disorder characterized by abnormal platelet aggregation and clot retraction. Main etiology is due to defect in platelet dysfunction that is defect in platelet membrane glycoprotein receptors IIb-IIIa (integrin alpha 2b beta 3) preventing platelet activation in response to ADP agonists, collagen or thrombin[2]. It is associated with homozygous and heterozygous mutation in ITG-A2B and ITGB3 which encodes GPIIB-GPIIIA. The clinical presentation includes hemorrhagic symptoms, mainly purpura, epitaxis, gingival hemorrhage and menorrhagia

The blood loss pattern in Glanzmann's thrombasthenia is usually one of excessive menstrual bleeding or major bleeding secondary only to minor trauma. Spontaneous bleeding is rare. The diagnosis of this disorder include criteria like normal platelet count, normal platelet morphology, prolonged bleeding time, absent or severely diminished platelet aggregation in response to adenosine diphosphate and other agonists, normal platelet agglutination by ristocetin, and normal plasma coagulation studies.

The severity of bleeding in thrombasthenia is variable and not predictable. some patients may require multiple red blood cell transfusions, while others may not manifest evidence of bleeding[3].

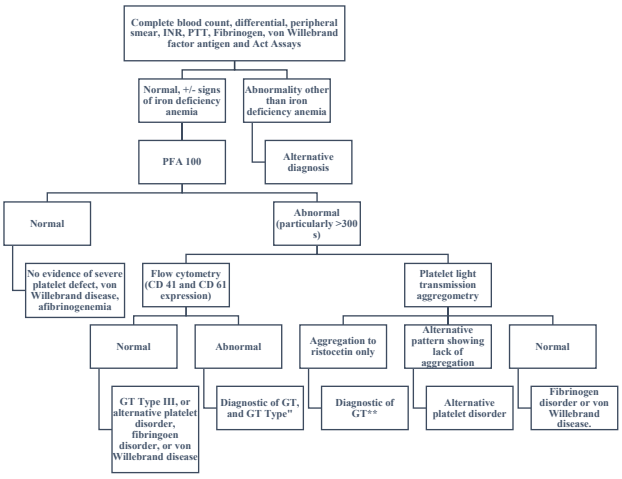

Fig. 1: Diagnostic algorithm for GT. *Consider proceeding to platelet light transmission aggregometry if suspicion for platelet defect remains high. **Consider genetic testing to identify specific mutation of ITGA2B and ITGB3 and/or flow cytometry to differentiate GT type. †Consider clot retraction assay (if available) and platelet light transmission aggregometry or genetic testing of ITGA2B and ITGB3 to make the diagnosis of GT Type III. ††Consider genetic testing to identify specific mutation of ITGA2B or ITGB3.

Fig 2 :Clinical manifestation

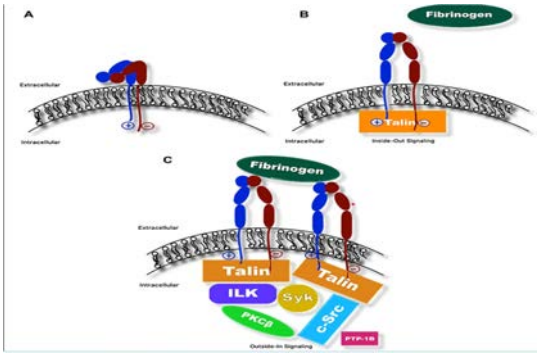

Fig 3 :Pathogenesis

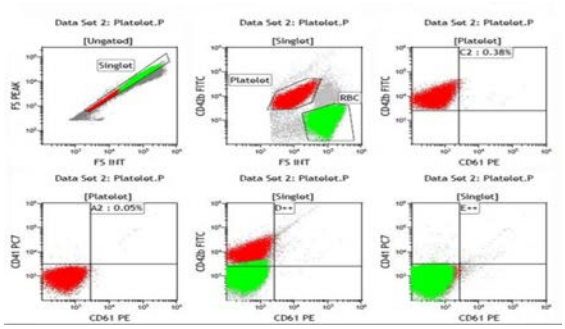

Fig 4 : Flowcytometry

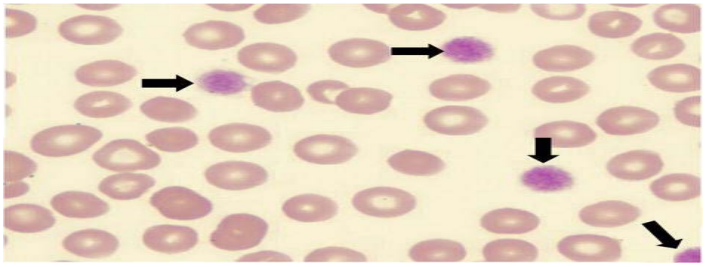

Fig 5 : Peripheral Smear

Clinical Details: A 15year old girl presented to the OBG out patient department with known history of Glanzmann thrombasthenia with complaints of heavy menstrual bleeding since 9-10 days. On taking detailed history it was learned that patient attained her menarche for the first time 9 days back and since then she had heavy menstrual bleeding. Patient was hospitalized earlier for epistaxis and gave history of multiple blood and platelet transfusion since the age of 2 years. 1st episode of epistaxis was noted at 1 and half yrs of age. Patient also gave positive family history of similar complaints in her younger sister who is 12 years of age and younger brother of 9 years of age having a mental disorder (details unknown). Priorly this patient was investigated by haemotologist and flow cytometry was done. On flow cytometry there was absence of CD41 and CD61 which encodes for GPIIB-PIIIa receptors . Her laboratory findings which included showed Hb 9.7g/dl Platelet 2.07L TLC 5.56 cells/mm^3, eripheral smear-imorphic anemia LFT within normal limits. Supportive management was given.

RESULTS AND DISCUSSIONS

Patient can be treated for acute blood loss by giving anti fibrinolytic agents, platelet and blood transfusions. Its a rare disorder with incidence of <1 in 1 million. It is seen in populations such as French, Roman, South Indian Hindus and Jews in all of whom
consanguinity is common. It starts from early childhood and is a lifelong illness. In 1918, Dr Glanzmann, a Swiss Paediatrician, coined the term thrombasthenia or weak platelets when describing the patient exhibiting purpura despite having platelets of normal quantity and appearance on peripheral smear[2].

In the year 1998 a series 21 women with same disorder were studied by Ofer Markovitch *et al,* out of which there was only one case of severe vaginal bleeding that required multiple transfusion. Patient who delivered by cesarean section had fewer hemorrhagic complications[3]

Since recombinant FVIIa has been effectively utilized to treat hemorrhagic problems related to pregnancy and gynecology, it may be a viable alternative hemostatic drug for women who suffer from this bleeding disorder[4]. Two cases of rFVIIa being successfully used in gynecologic oncology patients were documented by Sajdak[5]. Erikci[6] used rFVIIa to treat a lady with acute myeloid leukemia who was experiencing significant uterine hemorrhage. Additionally, recombinant factor VIIa was utilized to treat severe bleeding linked to uterine atony and life-threatening postpartum hemorrhage[4].

Steps to be Taken in Future: There are numerous difficulties in diagnosing and treating GT patients. Testing in specialized labs is necessary for the diagnosis and additional genetic testing does not, as of now, assist forecast severity. But as the Glanzmann Thrombasthenia Registry grows, also will our knowledge of the many mutations that cause GT. The cornerstone of treatment at the moment is still supportive. But if anti-platelet antibody detection increases, there will undoubtedly be a greater need for therapeutic solutions. Bone marrow transplant regimens will need to become more standardized for this population and acknowledged as a therapy option at an early age in order to meet this demand. Additionally, the advancement of gene therapy technology for GT patients will provide a different approach to curing an otherwise incurable condition[1].

CONCLUSION

It is one of the rare causes of menorrhagia in obstetric practice. Appropriate counselling and symptomatic treatment is the main stay of treatment. Hematopoietic stem cell transplantation has shown some results however it is very costly and is not affordable . Recombinant factor VIIa transfusion has been advised in some cases. rFVIIa is a well-known and secure alternative therapy option that has been effectively applied to stop bleeding during obstetric and gynecological operations. It appears that rFVIIa could be a useful therapeutic adjunct as well with intense, protracted menstrual bleeding in GT individuals who refused to react to a massive transfusion of platelets. While it might provide insight for more research, to determine a specific therapy protocol for the patient, bigger case series and interaction with alternative treatment methods are required for control of menorrhagia in GT[7].

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