Case Report

A Rare Case of Bleeding Disorder: Glanzmann's Thrombasthenia

Dr. Sanjay Chavan[1], Dr. Karthik S. Kolkur[2], Dr. Darshita Shukla[2], Dr. Sharad Agarkhedkar[3]

[1]Professor, [2]Resident, [3]Professor & Head, Department of Paediatrics, Dr. DY Patil Medical College, Hospital & Research Center, Pimpri, Pune- 411018, Maharashtra, India

Abstract:

Glanzmann's thrombasthenia is an extremely rare autosomal recessive inherited bleeding disorder characterized by defective platelet aggregation leading to prolonged bleeding time. Patients may present with easy bruising, purpura, epistaxis, menorrhagia and gingival bleeding. Though the disease is rare, the prognosis is usually excellent with supportive care. Here, we report the case of Glanzmann's thrombasthenia in a young female who presented with complaints of epistaxis and a history of easy bruising. The patient improved with symptomatic and supportive care. The patient got discharged and is doing well under regular follow-up.

Key words: Platelet, Bleeding, Hemorrhage, Epistaxis

Introduction:

Glanzmann's thrombasthenia (GT) is a rare bleeding disorder, which is characterized by a lack of platelet aggregation. It is characterized by qualitative abnormalities of the platelet membrane glycoprotein IIb/IIIa.[1] It is an extremely rare autosomal recessive disorder with an incidence of about 1 in 1,000,000 and an equal sex predilection. It is found more frequently in areas of the world where consanguineous marriage is common. Approximately, till now 500 cases have been reported in the literature.[2]

The symptoms of Glanzmann's thrombasthenia usually begin at birth or shortly thereafter. Patients may present with easy bruising, purpura, epistaxis, gingival bleeding, and intermittent gastrointestinal bleeding. Sometimes they bleed profusely, especially after surgical procedures. The severity of the symptoms varies greatly depending on the type of the disease. Mortality due to hemorrhage in diagnosed patients is rare unless associated with trauma or other diseases. The prognosis is excellent with careful supportive care. Here, we report the case of Glanzmann's thrombasthenia in a young female who presented with complaints of epistaxis and a history of easy bruising, she improved with symptomatic & supportive care and discharged. she is doing well and is under regular follow up

Case report:

4 years old female child born out of 3rd degree consanguineous marriage came with complaints of nasal bleeding for 4 days. Past history of multiple ecchymotic patches present. No similar past history. No h/o hospital admission. Nasal packing was done still bleeding persisted, endoscopic cauterization of bleeding arteries done still bleeding persisted. On examination PR- 100/min, RR 30/min, peripheral pulses well felt+, BP 102/62mmhg, Spo2 100%. Systemic examination is normal. No organomegaly

Hb- 7.70, platlet-4,40,000. TLC 14300, LFT and RFT -normal

Clotting time-12mins, prothrombin time-13secs, aptt-31secs, thrombin time -11secs, factor 13- normal, Iron studies, and vit B12- normal, Platelet aggregation test with prp: ADP(6uM) –nil,

Corresponding Author: *Dr. Karthik S. Kolkur*

Email ID: *kolkurkarthik@gmail.com*

Address: *Department of Paediatrics, Dr. D. Y. Patil Medical College, Hospital & Research Center, Pimpri, Pune- 411018, Maharashtra, India*

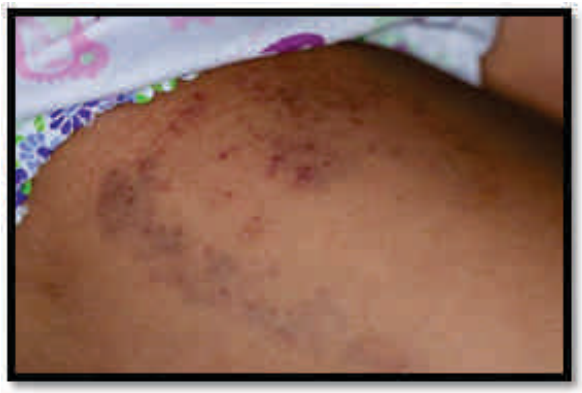
ISSN No. *: (p) 2348-523X, (o) 2454-1982*

DOI: 10.46858/vimshsj.8306

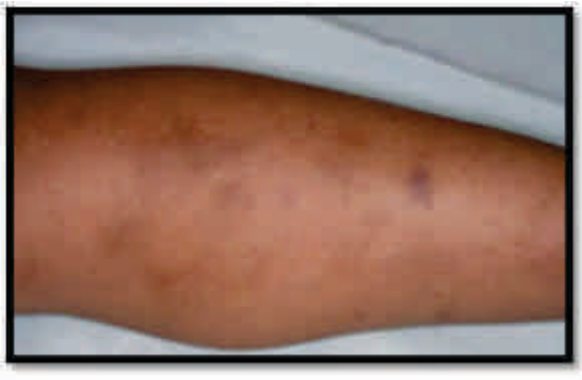
Date of Published : *25th September 2021*

Collagen –nil, Arachidonic acid –nil, Glycoprotein receptor by flow cytometry Gp1b- 99.6%,Gp11b-111a- nil, Gp IX- 99.2% Fibrogen-77.2 There is no specific treatment. Blood transfusions are usually suggested. Platelet transfusions to replace damaged platelets. Medications such as ibuprofen, aspirin, blood thinners such as warfarin and anti-inflammatory drugs are avoided. Most people with platelet disorders only need treatment during surgical procedures and after any bleeding manifestations. The patient was treated with platelet transfusion and other supportive care, improved and discharged. she is under close follow-up and is doing well with supportive care. The patient was advised for transfusion with group-specific screened fresh blood/ FFP (Fresh frozen plasma)/ Cryoprecipitate and platelet-rich plasma or concentrate in the event of bleeding, prior to surgery or dental extraction. she was also advised to avoid intramuscular injection, aspirin and other drugs affecting platelet function. Importance of maintaining oral hygiene was explained to the patient and his parents

Ecchymotic patches over Thigh (A) & Forearm (B)



(A)



(B)

Discussion:

Glanzmann's thrombasthenia was first documented in 1918 by Dr. Eduard Glanzmann, who described a functional abnormality of platelets with defective clot retraction and abnormal appearance on stained film.[1] It is an extremely rare autosomal recessive disorder with an incidence of about 1 in 1,000,000 with an equal sex predilection.[3] It is caused by an abnormality in the genes for glycoproteins IIb/IIIa which code for a group of linked proteins normally found on the surface of platelets. The gene responsible is carried on the long arm of chromosome 17. George et al[4] divided Glanzmann's thrombasthenia into three groups as: Type- I: patients with less than 5% of GpIIb-IIIa, Type- II: patients with 5%-20% of GpIIb- IIIa and Type- III (variants) with normal amounts of GpIIb- IIIa, but functionally inactive. Our patient belongs to the type I category. Though this condition is more common in populations where intermarriage is common[5], our case is an exception.

This disease is characterized by normal or subnormal platelet count, prolonged bleeding time and a deficiency or absence of platelet aggregation.[6,7] This functional platelet deficiency manifests as a bleeding disorder characterized by mucocutaneous hemorrhage of varying severity like easy bruising, epistaxis and gingival hemorrhage, which was there in our patient. Bruising typically occurs following minor trauma.[8] It is characterized by prolonged bleeding time, decreased or absence of clot retraction, and abnormal platelet aggregation responses to physiologic stimuli which were positive for this patient. Glanzmann's thrombasthenia is definitively diagnosed by tests that determine the deficiency of the a IIbβ3 (GPIIb/ GPIIIa) receptor. These tests usually involve monoclonal antibodies and flow cytometry.[9] Genetic tests can identify the DNA mutations responsible for the disorder.

Spontaneous mucocutaneous bleeding is common and can lead to fatal bleeding episodes which happened in the siblings of our case.

Control and prevention of bleeding among these patients pose a challenge to the treating physician. Treatment is primarily supportive mainly through local therapy, antifibrinolytics, and transfusions of fresh platelets or platelet concentrate.[10,11] Several reports suggest that platelet transfusion should be avoided except in case of severe bleeding as it may lead to platelet antibody development.[12] Tranexamic acid has also been used successfully in the form of a mouthwash or tablets after minor surgical procedures to inhibit postoperative bleeding episodes.[13] Prescribing medications that interfere in the normal function of platelets, such as aspirin and other nonsteroidal anti- inflammatory drugs (NSAIDs) should be avoided. Venkat V et al also reported a case of Glanzmann's thrombasthenia who presented with complains of spontaneous gingival bleeding and improved with supportive care.[14]

Gelatin sponge or gauze, antifibrinolytic agents such as topical thrombin, and YAG laser can be used to control minor bleeding. Desmopressin (DDAVP) has been tried in some patients with Glanzmann's thrombasthenia and may shorten the bleeding time in patients with Type 2 only. Bone marrow transplantation can be considered in patients with severe cases unresponsive to conventional therapies. Gene therapy and stem cell transplantation offer a potential cure for this disease, but both are costly and remain experimental at this point. Carrier detection by protein analysis and direct gene analysis is important to control the disorder in family members.

Conclusion:

Spontaneous mucocutaneous bleeding is common and can lead to fatal bleeding episodes in patients with Glanzmann's Thrombasthenia. Although Glanzmann's thrombasthenia is one of the rare bleeding disorders, awareness of this condition is important. Early diagnosis and prompt treatment carry a good prognosis. People should be counselled to avoid consanguineous marriage. The patient and their caregivers should be educated to avoid bleeding episodes by avoidance of anti-platelet drugs, and trauma. Control and prevention of bleeding among these patients pose a challenge to the treating doctor.

References:

1. Rao, A.K., Coller, B.S. Hereditary qualitative platelet disorders. In Williams Hematology, 9th Edition, Kaushansky, K., Lichtman, M.A., Prchal, J.T., Levi, M.M., Press, O.W., Burns, L.J., Caligiuri, M.A., eds. McGraw-Hill, Inc., New York, 2015, pps2039-2071.
2. Kamruzzaman M, Hossain MI, Rahman MA, Sharifunnesa -. Glanzmann's Thrombasthenia: A rare platelet functional disorder. Faridpur Med Coll J. 2021;15(2):103-5.
3. Glanzmann's thrombasthenia- A rare case report of spontaneous gingival bleeding.J Indian Acad Oral Med Radiol.2018;30:88-91
4. George JN, Caen JP, and Nurden AT. Glanzmann's Thrombasthenia: The Spectrum of Clinical Disease Blood 1990;75:1383-95.
5. Patel N, Modi M, Asari.D. Glanzmann thrombasthenia: A rare autosomal recessive bleeding disorder. Int J Sci Res. 2015;4.
6. Sebastiano C, Bromberg M, Breen K, Hurford MT. Glanzmann's thrombasthenia Report of case and review of the literature. Int J Clin Exp Pathol. 2010;3:443-7.
7. Gopalakrishnan A, Veeraraghavan R, Panicker P. Hematological and surgical management in Glanzmann's thrombasthenia: A case report. J Indian SocPedodPrev Dent. 2014;32(2):181-4.
8. Bisch FC, Bowen KJ, Hanson BS, Kudryk VL, Billman MA. Dental considerations for a Glanzmann's thrombasthenia patient: case report. J Periodontol. 1996;67:536-40.
9. Solh T, Botsford A, Solh M. Glanzmann's thrombasthenia: Pathogenesis, diagnosis, and current and emerging treatment options. J Blood Med. 2015;6:219-7.

10. Mehta DN, Bhatia R. Dental considerations in the management of Glanzmann's thrombasthenia. Int J Clin Pediatr Dent. 2010;3:51-6.
11. Nurden AT. Glanzmann thrombasthenia. Orphanet J Rare Dis. 2006; 1:10.
12. Varkey I, Rai K, Hegde AM, Vijaya MS, Oommen VI. Clinical Management of glanzmann'sthrombasthenia: A Case Report. J Dent (Tehran). 2014;11:242-7.
13. Sindet-Pedersen S, Ramstrom G, Bernvil S, Blomback M. Hemostatic effect of tranexamic acid mouthwash in anticoagulant-treated patients undergoing oral surgery. N Engl J Med. 1989;320:840-3.
14. Venkat V, Kalluri S, Hanumantha SRB. Glanzmann's thrombasthenia – A rare case report of spontaneous gingival bleeding. J Indian Acad Oral Med Radiol 2018;30:88-91