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Glanzmann's Thrombasthenia with Involuntary Upper and Lower GI Bleeding: A Case Report

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Abstract

A rarely seen bleeding illness called Glanzmann's thrombasthenia (GT) is inherited via the autosomal recessive gene. Platelet membrane glycoprotein IIb/IIIa (integrin aIIbb3) structural abnormalities and malfunction play a major role in the development of GT. The most prevalent signs of GT are various types of bleeding, including menorrhagia in females, recurrent nasal bleeding, mucocutaneous bleeding, and continuous bleeding after injury or surgery. Some patients may not survive after undergoing such a hemorrhage. It is unusual for GT to experience spontaneous upper and lower gastrointestinal hemorrhages. In this case study, a young boy with accidental bleeding in vomitus and fresh blood in his stool was hospitalized. His primary GT is an uncommon autosomal recessive genetic condition characterized by dysfunctional platelet aggregation symptoms that were sporadic and recurring chronic fresh blood in his stool. Blood dripped from the stomach body wall during the gastroscopy. However, except for petechiae no visible lesions were discovered, like erosion or ulceration. Further investigations revealed that the patient's BT and CT were prolonged and platelet aggregation was poor. After receiving aggressive treatment, the patient showed clinical improvement and was eventually diagnosed with GT.

Keywords: glycoprotein gpIIb-IIIa; platelet; purpura; endoscopy; menorrhagia; thrombasthenia

Introduction

Glanzmann first identified Glanzmann's thrombasthenia (GT) in 1918. Platelets with GT, a severe congenital platelet condition, lack glycoprotein IIb/IIIa (integrin aIIbb3), which results in a significant defect in platelet aggregation [1]. Although uncommon, when present it is more prevalent in families with consanguineous marriages. Regarding the incidence of GT, there was no appreciable difference between the patients who were male and female. Visceral hemorrhage is a rare clinical symptom of GT; mucocutaneous hemorrhage is the most common clinical symptom [2]. A general wellness checkup may not always detect anomalies linked to the GT; as a result, the GT is frequently ignored or incorrectly diagnosed. For an evaluation of GT, laboratory tests are necessary. In individuals with this disease, platelet structure and count are usually within normal limits, but the bleeding time is much greater [3]. Consequently, the confirmation of GT is based on the recognition of abnormalities in platelet aggregation, and it is crucial to use more precise techniques, such as flow cytometry or gene detection, to make a definitive diagnosis [4]. There is currently no known full treatment for GT; instead, symptomatic care is the focus [5]. Here, we present a difficult case of GT and examine its clinical characteristics, diagnostic criteria, therapeutic options, and prognosis.

Case Presentation

A 17-year-old boy was brought into our hospital with severe anemia, vomiting, and epigastric pain. Before admission, he had sporadic and recurring gum bleeding and pallor, but these symptoms were thought to be due to nutritional deficiency and worm infestations. However, as time progressed, his symptoms worsened, becoming more regular and severe with brand-new symptoms, including nausea, dizziness, and epigastric pain. The pain typically started along with bleeding and subsided once the blood in stools and melena ended. Before the

commencement of hematochezia, melena, or epigastric discomfort, the individual had no history of taking any specific medications or illnesses. We meticulously reviewed his medical background and performed a thorough physical examination. When he was younger, he frequently had significant volumes of accidental bleeding from the gingiva and nose, which occurred throughout the year. He had previously been treated with blood transfusions for severe anemia. His parents had non-consanguineous marriages and were both in good health. At the time of admission, a physical examination revealed pale skin and epigastric soreness. Some laboratory investigations are shown in Table 1.

Table 1: Hematological investigation at presentation.

Test	Reference	Result
Haemoglobin (gm/dl)	11.5-17.5	2.5
Platelets	150000-350000	312000
PT (secs)	12	12
aPTT (secs)	28	28
BT (mins)	2-11	>15
CT (mins)	5-10	10

Abbreviations; PT: Prothrombin Time, APTT: Activated Partial Thromboplastin Time, BT: Bleeding Time, CT: Clotting Time

When the patient underwent gastroscopy following an initial diagnosis of upper gastrointestinal bleeding, it was discovered that blood flowed through the gastric antrum wall and petechiae were also noted. Colonoscopy revealed non-significant as well. Without any complications, he received endoscopic hemostasis therapy. Following the surgery, proton pump inhibitor (PPI), and somatostatin a were administered. Additionally, blood was also transfused, which helped to gradually stabilize his condition. Gastroscopy revealed no evident lesions such as abrasions or ulcerations. We speculated that the patient may have had several blood illnesses given his history of bleeding, and no aggregation was observed with adenosine diphosphate or epinephrine in platelet aggregation experiments; however, a normal response to ristocetin was observed. Unfortunately, flow cytometry could not be performed owing to limited resources, and the diagnosis was established based on the ristocetin test as shown in Table 2.

Table 2: Platelet function study of patient.

Parameter	Reference	Result
Platelets	150000-400000	312000
Bleeding time (mins)	2-11	>15
Platelet aggregation	Collagen	No response
	Adenosine Diphosphate	No response
	Epinephrine	No response
	Ristocetin	Normal Response
Hess test		Negative

Once the diagnosis had been established, the patient received platelet transfusion therapy to stop surgical bleeding. He gradually returned to his usual diet as the hematochezia and melena subsided. He quickly recovered and left the hospital after being released. Following discharge, the patient came for a follow-up appointment on a biannual basis. He underwent numerous endoscopies, none of which showed a return to upper gastrointestinal hemorrhage. The patient permitted dissemination of this report.

Discussion

Glanzmann's thrombasthenia is an uncommon genetically inherited disorder with an autosomal recessive pattern, characterized by dysfunctional platelet clumping due to a congenital deficiency in the platelet fibrinogen binding integrin aIIbb3 [6]. Only 9% of deficits in platelet functionality are caused by this disease, which has a very low incidence. In recent years, an increasing number of new cases have been reported. Bleeding from mucosal surfaces, purpura, and menorrhagia are the common clinical

presentations of the disease [7]. The severity of hemorrhagic symptoms may vary greatly. Most hemorrhagic symptoms are brought on by surgery, childbirth, or injury; idiopathic bleeding is rare. Young people may pass away from major hemorrhages, whereas others may experience minor symptoms. In patients with GT, bleeding occasionally improves with age [8]. Because the clinical signs of GT, which involve bleeding from the digestive tract, resemble those of bleeding caused by common gastrointestinal disorders, GT is frequently undiagnosed [9]. According to earlier research, upper and lower gastrointestinal bleeding can be enormous and challenging to manage, as it occurred intermittently in our case. Additionally, many GT patients who experience spontaneous gastrointestinal hemorrhage also have gastrointestinal conditions, such as gastrointestinal tract polyps, gastroduodenal ulcers, or Helicobacter pylori infection [1,6] In the current investigation, the patient exhibited upper abdominal discomfort; however, gastroscopy revealed no organic disorders, which may have been related to gastrointestinal spasms caused by bleeding. Detailed medical and family history of the patient usually provides helpful clues in making the diagnosis. Patients who have experienced frequent bleeding during infancy or childhood, as was the case with our patients, should be given special consideration for GT [3]. Furthermore, a thorough physical examination of patients with GT frequently reveals ancient skin ecchymosis. Laboratory testing is clinically important for confirming GT diagnosis. Patients with GT typically exhibit longer bleeding times, normal platelet counts, poor blood clot retraction, and impaired platelet aggregation in their test results [10]. Ristocetin-induced platelet aggregation was typically normal, as observed in our case. To rule out alternative sources of hemorrhage, such as gastric ulcers, gut malignancies or polyps, inflammatory bowel disease, and, vascular illnesses. To rule out any primary GI disease endoscopic investigations are usually done [11]. Treatments for symptoms and supportive measures are the main therapeutic approaches for GT [12]. In individuals with GT, gastrointestinal bleeding poses a life-threatening risk. Our therapy was successful because the patient's bleeding was halted by endoscopic intervention, medications, hemostasis, and other proactive measures.

Additionally, refraining from using nonsteroidal anti-inflammatory medicines (NSAIDs) and eliminating H. pylori can prevent some episodes of gastrointestinal hemorrhaging [6]. GT can be effectively treated with platelet transfusion. To avoid heavy bleeding following trauma, surgery, or childbirth, patients also require platelet infusions. However, frequent platelet transfusions are not advised. An increase in platelet antibody levels and the likelihood of hematogenous transfer of infection are both possible side effects of this therapy, which may result in platelet refractoriness [13]. In recent years, recombinant factor VIIa is effective in treating hemorrhagic symptoms of GT. Thus, recombinant factor VIIa may offer a different therapeutic option for the management of GT [14]. The only treatment option is transplantation of hematopoietic stem cells [15].

Conclusion

Glanzmann's thrombasthenia is an uncommon inherited bleeding condition. It is mostly found in a small number of populations where consanguineous marriage is prevalent. Patients typically have easy bruising and bleeding from epistaxis and teeth extractions when they first arrive. Rarely it can also present as upper and lower GI bleeding. GT has an excellent prognosis if given the right supportive care. When assessing any case of a bleeding condition, GT should always be taken into consideration as a differential diagnosis.

Consent: Written informed consent was taken from the father of the patient.

Declarations

Human subjects: Consent was obtained or waived by all participants in this study. NA issued approval. Permission from Patient taken, no IRB required.

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References

1. Marlu R, Barthelon J, Durand A, et al. (2015). Long-term therapy with bevacizumab in a patient with Glanzmann's thrombasthenia and recurrent digestive bleeding due to gastrointestinal angio dysplastic lesions. Am J Gastroenterol, 110:352-353.
2. Nurden AT. (2006). Glanzmann thrombasthenia. Orphanet J Rare Dis. 1:10.
3. Qiao Z, Chen Y, Shi W, Yang J, et al. (2020). Glanzmann's thrombasthenia with spontaneous upper gastrointestinal bleeding: a case report. Journal of International Medical Research. 48:10.
4. Calabrese C, Di Febo G, Areni A, et al. (2000). Severe and relapsing upper gastrointestinal bleeding in a patient with Glanzmann's thrombasthenia. Dig Dis Sci. 45:633-636.
5. Solh T, Botsford A and Solh M. (2015). Glanzmann's thrombasthenia: pathogenesis, diagnosis, and current and emerging treatment options. J Blood Med. 6:219-227.
6. Bakdash S, Lyons JM, Bastacky SI, et al. (2008). Management of persistent gastric bleeding in a patient with Glanzmann's thrombasthenia. Am J Hematol. 83:411-415.
7. Ganapule A, Jain P, Abubacker FN, et al. (2017). Surgical procedures in patients with Glanzmann's thrombasthenia: case series and literature review. Blood Coagul Fibrinolysis. 28:171-175.
8. Binder A and Aledort L. (2015). Glanzmann's thrombasthenia: meeting the anticoagulation challenge. Haemophilia. 21:322-323.
9. Khosravi A, Rahimi H and Mansouritorghabeh H. (2015). Coincidence of Glanzmann's thrombasthenia with hereditary haemorrhagic telangiectasia in a man with gastrointestinal bleeding. Blood Coagul Fibrinolysis. 26:98-100.
10. Kurdi M, Frere C, Amour J, et al. (2018). Perioperative management of a patient with Glanzmann thrombasthenia undergoing a coronary artery bypass graft surgery: a case report. Blood Coagul Fibrinolysis. 29:327-329.
11. Mesquita R, Santos I and Monteiro H. (2018). Severe intestinal bleeding in a woman with Glanzmann thrombasthenia. Eur J Case Rep Intern Med. 5:000796.
12. Buckley F, Norris A and Kerr R. (2018). Management of abdominoperineal excision of the rectum in a patient with Glanzmann thrombasthenia. Acta Haematol. 139:243-246.
13. Poon MC, Di Minno G, d'Oiron R, et al. (2016). New insights into the treatment of Glanzmann thrombasthenia. Transfus Med Rev. 30:92-99.
14. Naderi M, Habibpour M, Alizadeh S, et al. (2018). Study of the relationship between HPA-1 and HPA-5 gene polymorphisms and refractory to platelet therapy and recombinant factor VII in Glanzmann thrombasthenia patients in southeast of Iran. Int J Hematol Oncol Stem Cell Res. 12:43-48.
15. Cid AR, Montesinos P, Sa´nchez-Guiu I, et al. (2017). Allogeneic hematopoietic cell transplantation in an adult patient with Glanzmann thrombasthenia. Clin Case Rep. 5:1887-1890.

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