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CASE REPORT

A rare case report on Glanzmann thrombasthenia

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ABSTRACT

Glanzmann thrombasthenia (GT) is a rare inherited blood clotting disorder characterized by the impaired function of platelets that are essential for proper blood clotting and can lead to prolonged bleeding time. Patient with GT may experience menorrhagia, easy bruising, purpura, epistaxis, and gingival bleeding. Here, we report a case of a young male, who was presented with complaints of malena, epigastric pain, and generalized weakness. Computed tomography and magnetic resonance imaging showed hemoperitoneum and perihepatic hematoma, respectively. Later, he also developed black-colored stools and occasional cough. Coagulation profile was suggestive of Glanzmann thrombasthenia. The patient was treated symptomatically, and as the conditions improved, the patient was discharged in a stable state of health. Only up to 500 cases were reported regarding GT till this date. As GT is a rare disorder, it needs to be reported in the current clinical setting.

KEY WORDS: Epistaxis; Hemoperitoneum; Glanzmann Thrombasthenia; Perihepatic Hematoma; Purpura

INTRODUCTION

Glanzmann thrombasthenia (GT) is a rare inherited blood clotting disorder characterized by the impaired function of platelets that are essential for proper blood clotting and can lead to prolonged bleeding time.[1] It is believed to be caused by a defect in chromosome 17, which codes for the platelet aIIbb3 (GPIIb/IIIa) integrin family receptor, thereby preventing platelets from performing its functions when bleeding occurs. Till now, over 500 cases have been reported most of which are from certain ethnic groups.[2] GT affects males and females in equal numbers although some studies reported greater prevalence in females. Patient with GT may experience menorrhagia, easy bruising, purpura, epistaxis, and gingival bleeding. Diagnosis should be suspected in patients with bleeding symptoms, and proper medical history, light transmission aggregometry, platelet function analyser, and flow cytometry are remained as useful tools.[3] Treatment mainly includes local therapy, antifibrinolytics, and platelet transfusion.[3,4] Recombinant factor VII is an acceptable alternative with excellent response rates. Bone marrow transplantation can be considered in patients with severe GT who are unresponsive to conventional therapies. Undetected GT can be life threatening so early diagnosis is a must to improve the prognosis.

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CASE REPORT

A 28-year-old gentleman presented with complaints of malena, epigastric pain, and generalized weakness for which he was evaluated and was found to have anemia and transfused with packed red blood cell. Ultrasonography showed increased size of mass, and computed tomography (CT) showed hemoperitoneum. Magnetic resonance imaging showed perihepatic hematoma. Later, he also developed

black-colored stools and occasional cough. Endoscopy was normal. Colonoscopy showed sigmoid ulcer. Sigmoid ulcer biopsy was performed to denote nature and shows multiple fragments of large intestinal mucosa with muscularis mucosa included in one of them. The lamina propria is mildly edematous and contains few congested blood vessels and focal lymphoid aggregates. The normal crypt architecture is preserved. There was no cryptitis or crypt abscess, and no granuloma or parasites were seen. There were no features to suggest inflammatory bowel disease, and there was no dysplasia or malignancy. Coagulation profile was suggestive of Glanzmann thrombasthenia. The patient was treated accordingly with stool softeners, antibiotics, packed red blood cell (PRBC) transfusion, tranexamic acid, and iron supplements. His serial hemoglobin levels were monitored and there was no significant blood loss, the patient improved symptomatically and is being discharged in a stable state of health.

DISCUSSION

GT is an autosomal recessive, inherited bleeding disorder characterized by normal platelet levels but is dysfunctional and prolonged prothrombin time, impaired aggregation of platelets, and absent or decreased clot formation.[5] It is found more common within populations that have a prevalence of consanguineous marriages although frequency of this is disorder is one in 1,000,000 individuals. Various reports have revealed greater predominance of GT in females compared to males (58% vs. 42%), consistent with its mode of inheritance.[3] Our patient was a young male who had no reports of consanguineous marriages or significant history in his family. The main symptoms of GT patients mainly manifested as bruising, epistaxis, bleeding from gums, menorrhagia, bleeding during or after surgery, etc.,[4] all of which stood false for this patient. He was presented with the complaints of epigastric pain and malena and had no other bleeding manifestations. Proper medical history, light transmission aggregometry, platelet function analyser, and flow cytometry were remained as the gold standard for the diagnosis of GT.[3] Studies have showed that antifibrinolytic therapy such as tranexamic acid, aminocaproic acid, recombinant factor VII, and platelet transfusions is the beneficial therapies for a patient with GT.[6] Allogeneic bone marrow transplantation performed in several severe cases of GT had led to the potential cure of this disease.[7] This patient was managed with PRBC transfusion, tranexamic acid, and iron supplements. However, several reports suggest that platelet transfusion should be avoided except in case of severe bleeding as it may lead to platelet antibody development.[6] As there were only a few reports regarding the GT, it needs to be reported which will give more attention to bleeding symptoms which may be misdiagnosed as some other disorders that may cause treatment delay and poor prognosis.

CONCLUSION

Only up to 500 cases were reported regarding GT till this date. As GT is a rare disorder, it needs to be reported in the current clinical setting.

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