

Thinking of Rare Bleeding Disorders in the Emergency Room: A Diagnosis of Glanzmann Thrombasthenia in a Pakistani Child

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Abstract

Introduction: Glanzmann thrombasthenia is a rare inherited bleeding disorder, with a global prevalence of about one in one million. It is an autosomal recessive condition characterised by episodes of mucocutaneous bleeding. A defect in the membrane protein results in a defective platelet aggregation.

Case presentation: A fifteen-month-old female child presented to the emergency department with nosebleed for the last two days. The patient had two similar episodes of epistaxis in previous few months which were managed at home. There was an accompanying complaint of easy bruising since birth. After emergency management and anterior nasal packing, extensive blood tests were ordered to determine the cause for recurrent haemorrhages. Platelet aggregation study concluded a diagnosis of Glanzmann thrombasthenia and the patient was treated accordingly. Family members were thoroughly counselled, and safety netting was vigilantly done.

Conclusion: Many public tertiary cares set ups in Pakistan lack the adequate diagnostic and therapeutic facilities for bleeding disorders. This does not only result in delaying appropriate care but also causes patients to remain undetected. Through this report, we aim to encourage emergency physicians to have a high index of suspicion for haematological disorders in patients presenting early in life with recurrent bleeding episodes.

Keywords: Epistaxis • Easy bruising • Glanzmann thrombasthenia • Platelet functional disorder • Inherited bleeding disorder • Pakistani • Emergency department • Paediatric department

Introduction

Glanzmann thrombasthenia is an autosomal recessive condition, characterised by mucocutaneous bleeding [1]. There is variability in the way the disease presents, with mild nosebleeds, gum bleed and bruises, to episodes of menorrhagia and large gastrointestinal haemorrhages, albeit rare. Glanzmann thrombasthenia is a functional platelet disorder, where a deficiency or a qualitative defect of glycoprotein IIb/IIIa receptor protein is seen, resulting in defective platelet aggregation. Platelet aggregation is one of the essential steps in clot formation. The condition is more prominently seen to occur in ethnic groups where consanguinity is a common practice [2]. It is estimated that the prevalence is about 1:1,000,000 in the general population [3]. One of the common symptoms seen by emergency physicians in the paediatric department includes epistaxis. It is approximated that around 9 percent of the children are affected by nosebleeds [4]. In some cases, the bleeding may be severe and not managed by applying pressure over the nose, hence intervention is pertinent as a more definitive management.

Case Report

A fifteen-month-old female child presented with recurrent episodes of mucocutaneous bleeding. The patient was accompanied by her mother to the emergency room, where she came with epistaxis lasting for the last two days. Bleeding was spontaneous, intermittent and manageable at home by pinching manually; however, it recurred after a few hours. Initially the patient was taken

to a local clinic where anterior nasal packing was done with cotton wool, but adequate control over the bleeding was not achieved. The guardian quantified the blood as being two to three tablespoons full each time, and dull red in colour. A secondary complaint of numerous skin bruises throughout the body, since birth, was also present. The bruises were reported to resolve spontaneously but were noticed to reappear within after some days. They were atraumatic and the possibility of domestic child abuse was eliminated as well. No medical attention was sought previously for easy bruising. Past medical history revealed two similar episodes of epistaxis at six and ten months of age. Both times the patient was managed at home by applying pressure over the nose. There was no history of bleeding from other sites on the body, gum bleeding, and hematoma formations after intramuscular injections, haematuria or melena. The patient was a second product of a non-consanguineous marriage. No significant family history was discovered. Antenatal, birth and post-natal history were unremarkable. Developmental milestones were achieved on time, and immunisation status was up to date. Examination revealed an ill-looking child, with significant pallor, lying in bed, actively oozing blood from both nostrils. Multiple bruises of various ages were observed on both lower limbs and trunk, with the largest one measuring 2 cm × 1.5 cm on the left lower limb anteriorly. Patient's vitals were as shown in Table 1. Anthropometric measurements were found to be appropriate for the patient's age and gender. Rest of the systemic examination was unremarkable. In the emergency room, the patient was resuscitated and monitored vigilantly. Intravenous tranexamic acid was administered, anterior nasal packing was done, group and save was arranged and other relevant blood tests were ordered. Investigations are shown in Table 2. Patient was investigated for possible bleeding disorders, and a platelet aggregation study was arranged, the report is shown in Table 3.

Interpretation

- Absent response to collagen, Adenosine Di-Phosphate (ADP) and epinephrine
- Normal response to ristocetin

These findings were suggestive of Glanzmann thrombasthenia. A decision

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Table 1. Vitals.

Blood pressure	100/60 mmHg
Heart rate	110 beats per minute
Pulse	Regular and low volume
Respiratory rate	28 breaths per minute
Temperature	99-degree Fahrenheit
Oxygen saturation	99% on room air

Table 2. Blood investigations.

Full blood count	
Haemoglobin	10.8 mg/dL
Mean corpuscular volume	76.5 fL
Total leucocyte count	8600/microlitres
Neutrophils	22%
Lymphocytes	77%
Platelets	274 000/microlitre
Peripheral film revealed anisocytosis, microcytosis and hypochromia with normal platelets	
Clotting profile	
Prothrombin time	11 seconds
Activated partial thromboplastin time	25 seconds
International National Ratio	0.9
Bleeding time	Greater than 7 minutes

Table 3. Platelet aggregation study.

Test	Result	Normal Ranges
Bleeding time	Greater than 7 minutes	1-7 minutes
Ristocetin (0.5 mg/mL)	0%	0-8
Epinephrine (10uM)	0%	48-103
Collagen (2 ug/mL)	0%	46-112
ADP (10 uM)	3%	50-110
Ristocetin (1.25 mg/mL)	43%	53-103
Platelet count	$332 \times 10^9/L$	150-400

to admit and observe the patient was made. Nasal packing was removed after two days, and vitals were closely monitored throughout the hospital admission. A discussion on diagnosis, the nature of the bleeding disorder, and a thorough counselling for future care was provided to the family members. Patient's caretakers were advised on avoiding blood-thinning medications, trauma and promptly returning to the hospital in the event of significant bleeding. It was also advised that family members undergo screening for hereditary bleeding disorders.

Discussion

Glanzmann thrombasthenia was first described by a Swiss paediatrician, Eduard Glanzmann, in the year 1918. It is an inherited bleeding disorder caused by mutations in chromosome 17q21 [1]. The condition is characterized by a quantitative or a qualitative defect in platelet protein alpha IIb beta-3. An acquired form of the disease also exists when antibody to platelet fibrinogen receptor is formed. Such is seen to occur in conditions with autoantibody formation like in a case of systemic lupus erythematosus [5]. Through this case report, we aim to highlight that rare bleeding disorders are a possible diagnosis in a patient with recurrent bleeding symptoms. Nonetheless, due to the busy nature of an emergency department and a lack of adequate resources in a public set up, especially in struggling economies, often important steps towards future care are overlooked. The usual course of action after resuscitating patients involves discharging them home. The responsibility of referral or admission for further investigation falls upon the emergency physicians, particularly in cases where the underlying cause for the symptoms remains undiagnosed. This is especially true in cases of severe bleeding, where the emergency doctors are usually the first point of medical contact. It is vital to identify the need for haematological and clotting studies, advance consultations and family screening, in order to extend the best possible long-

term care to the patients and their relatives. Such conditions tend to have lifelong consequences; therefore, timely diagnosis and management can result in better prognosis and quality of life for the patients. Recurrent bleeding episodes, especially in girls, grossly impact the overall life. For instance, girls may experience heavy and prolonged menstrual bleeding, and it is likely to worsen the existing iron-deficiency anaemia and associated symptoms as in the reported case of a twelve-year-old Nigeran girl, soon after menarche [3]. Up to 90% of the cases diagnosed in India and Pakistan are observed among families of consanguineous marriages, in contrast to the parents of this patient, who were unrelated prior to their marriage [3]. Therefore, intermarriage setting is an important but not an essential link for the diagnosis.

Conclusion

Rare bleeding disorders are unlikely to be thought of as a potential differential diagnosis in the emergency department. Nevertheless, bleeding episodes early in life call for a high index of suspicion and we encourage emergency doctors to redirect suspected patients either to outpatient or inpatient care, where appropriate investigations can aid in reaching a definitive diagnosis. The need for awareness, diagnostic and therapeutic advances for such rare conditions in public hospitals is required in our healthcare facilities for prompt management. The outcome of Glanzmann thrombasthenia is generally good and timely diagnosis prevents future life-threatening complications.

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How to cite this article: Salma Mainosh, Ayesha Akbar Khalid, Mushtaq Ahmed, and Syeda Shaheera Hashmi Riaz. "Thinking of Rare Bleeding Disorders in the Emergency Room: A Diagnosis of Glanzmann Thrombasthenia in a Pakistani Child." *Clin Case Rep* 11 (2021): 1431.