CASE REPORT

A Case of Glanzmann Thrombasthenia

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ABSTRACT

Glanzmann thrombasthenia (GT) is a rare congenital autosomal recessive platelet function disorder characterized by defective platelet aggregation due to abnormalities in the glycoprotein IIb/IIIa complex. Clinically, it presents with mucocutaneous bleeding, including epistaxis, gingival bleeding, and easy bruising.

Key words: Glanzmann thrombasthenia, epistaxis and gingival

INTRODUCTION

Glanzmann thrombasthenia is a rare congenital autosomal recessive disorder.

Clinical features include bleeding manifestations such as epistaxis, gingival, and mucocutaneous bleeds.

CASE REPORT

A 1-year-old male citizen of Pune who is the second kid of a non-consanguineous marriage brought by parents with recurrent bouts of spontaneous nasal bleeding when he was quite small. There is a previous history of receiving numerous blood transfusions. The bleeding symptoms did not correlate with any prior history of bruises, hematemesis, gingival bleeding, trauma, or purpura. There is no significant family history or birth history. The patient is immunized for age and is developmentally normal.

Anthropometrically, weight for height was −1 to −2 standard deviation, which was normal.

At each episode of nasal bleeding, anterior nasal packing was done, and blood transfusion was given.

On examination, vitals were stable and pallor was present.

On per abdomen, palpation of hepatomegaly was seen.

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Laboratory Investigations

A peripheral blood smear revealed anisopoikilocytosis, primarily normocytic normochromic red bold cells (RBCs), a small number of microcytic hypochromic RBCs, a few teardrop and pencil cells, and an appropriate platelet count and size, along with a hemoglobin of 7.4 g%.

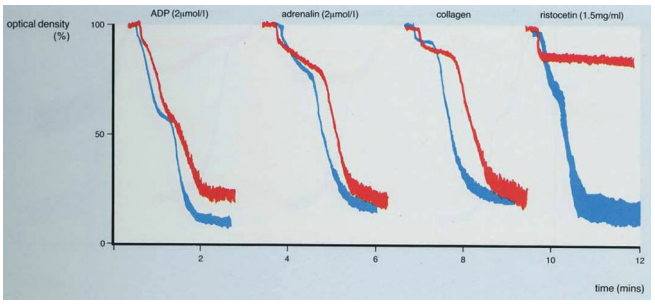
ENT opinion was taken. And an X-ray (paranasal sinus) was done, which was normal.

Prothrombin time showed 11.8 s (10–13), and

aPTT = 28.4 (31.1), which was normal.

Prolonged bleeding time was seen at 6 min 20 s (2–5 s).

Subsequently, platelet function disorder was suspected, and a platelet function test revealed aggregation with ristocetin only.



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Differential Diagnosis

Bernard Soulier

Von Willebrand Disease

Treatment

Patient education and immunization

- Platelet transfusions
- Symptomatic treatment.

Available treatment options

- Anti-fibrolytic therapy such as tranexamic acid and epsilon aminocaproic acid
- Recombinant factor VIIa
- Rituximab (antiCD20), bevacizumab – use of systemic corticosteroids, cyclophosphamide, azathioprine, plasmapheresis, IVIG
- Hematopoietic stem cell transplant.

DISCUSSION

With normal-sized platelets and morphologic characteristics on peripheral blood smears, Glansmann thrombasthenia is a platelet condition characterized by significantly abnormal bleeding time or PfA-100 closure times. All agonists except ristocetin, which does not require metabolically active platelets, elicit aberrant or absent aggregation in aggregation tests. In this syndrome, there is a deficiency in the platelet fibrinogen receptor 2b3a, the major integrin complex on the platelet surface.

The main integrin complex changes form in response to inside-out signaling that occurs during platelet stimulation. When a platelet is stimulated, fibrinogen binds to this complex, causing the platelets to coalesce. Gene mutations that can be identified and that are inherited autosomally recessively produce Glansmann thrombasthenia. Stem cell transplants have been used in curative therapy, according to reports.[1,2]

CONCLUSION

Glanzmann thrombasthenia should be considered as a rare possibility in patients presenting with recurrent epistaxis.

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