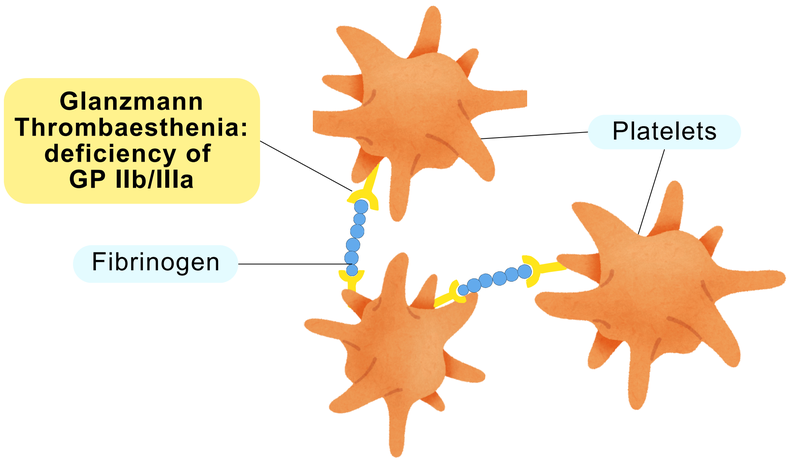
Glanzmann Thrombaesthenia: A Rare but Potentially Life Threatening Bleeding Disorder

• Ishita Agarwal • Dr. Emine A. Rahiman • Dr. Archana M.V. • Dr. Vasudeva Bhat K • Dr. Swathi P.M. •
Department of Paediatric Haematology & Oncology, KMC Manipal

INTRODUCTION

Glanzmann's thrombasthenia (GT) is an autosomal recessive disorder caused by defective integrins αIIb and β3. These integrins form the platelet glycoprotein (GP) IIb/IIIa.
The prevalence of GT is estimated to be one in a million and is said to be more common in communities with high levels of consanguinity.



CASE REPORT

We present the case of a 5-year-old girl who was born preterm at 7 months of gestation through assisted reproductive technology. She presented to the hospital with history of recurrent episodes of epistaxis and skin bleeding since the age of 3. Her family history was positive for third-degree consanguineous marriage. There was no history of bleeding gums, bone pain, or recurrent fevers.

On examination, she had pallor with multiple petechiae, and ecchymosis scattered across her body. Her vitals were stable. Systemic examination was unremarkable.

Recurrent moderate-severe mucocutaneous bleeding

CBC	Normal Platelet Count (2.26 lakhs/μL) with Low Haemoglobin (8.9 g/dL).
Bleeding Time	Prolonged (>15 seconds)
Coagulation screening tests	aPTT and PT-INR were normal. Serum Fibrinogen was 186.2 mg/dl (ref. 200-400 mg/dl).
Others	Thrombin time, clot retraction time, factor XIII levels and vWF levels normal

Platelet aggregation tests

Decreased/Absent aggregation with all agonists except ristocetin in 2 different samples.

Normal → Consider other platelet disorders

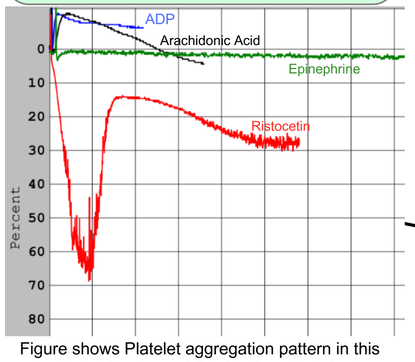


Figure shows Platelet aggregation pattern in this patient. (Courtesy of Dr. Sindhura Lakshmi, Department of Pathology, KMC Manipal).

Can be confirmed via platelet glycoprotein analysis to look for decreased/absent GP IIb/IIIa. ITGA2B and ITGB3 molecular analysis can be done for further confirmation

TREATMENT

Platelet transfusion and injection tranexamic acid to manage her bleeding. → Packed red blood cell transfusion since her Hb was low. → Discharged with advice to follow up.

On the subsequent visit with complaints of epistaxis, a diagnostic nasal endoscopy was done, which revealed dilated nasal vessels. These were cauterized with aluminum hydroxide, and saline nasal drops were prescribed for one week.

DISCUSSION

Although there is no known cure for GT, the prognosis is generally good.

Management

- **Control Bleeding**
  - Compression, Nasal Packing , and Cauterization
  - Antifibinolytic Agents
  - Platelet Transfusions
- **Prevention & advice**
  - Avoid NSAIDs and Aspirin
  - Avoid high risk sports
  - Regular follow up

Repeated transfusions can lead to development of anti-HLA antibodies. Hence, they should be done only in cases of severe bleeding and before any invasive procedure.

CONCLUSION

This case highlights the importance of keeping GT in the differential diagnosis for any case of recurrent bleeding, especially in a child with normal platelet counts.