Case Report

A Case of Glanzmann Thrombasthenia Presenting at Birth- With Petechiae and Ecchymosis- Early Presentation

Mohamed Salman J[1], Janani Sri V[2], Madhu Dharshini S. R[1], Farjana G[2], Raj Parath R[3], Archana Dinaraj[4], Ganavi Ramagopal[5]

[1]Post Graduate, Department of Paediatrics, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India, [2]Post Graduate, Department of Pathology, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India, [3]Assistant Professor, Department of Paediatrics, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India, [4]Senior Resident, Department of Paediatrics, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India,[5]Professor and HOD, Department of Paediatrics, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India.

Abstract

Platelet dysfunction caused by mutations in the glycoprotein-encoding genes gpIIb or gpIIIa is linked to Glanzmann's thrombasthenia, an uncommon congenital bleeding condition. Glanzmann's thrombasthenia is difficult to identify based on a conventional physical examination and is typically misdiagnosed since the bleeding duration is substantially greater, yet the platelet counts and morphology are normal. Here we present a case of newborn male child delivered with spontaneous multiple petechiae and ecchymosis over the trunk and mucocutaneous nasal bleeding while suctioning since birth, need admission to the newborn intensive care unit. An initial hematologic workup showed normal coagulation profile and sepsis screening was negative but-with suspicion when evaluated platelet function was slightly impaired. This case underscores the critical need for early recognition of Glanzmann's thrombasthenia in neonates within 24 hrs of life. The patient's family received counselling about the disorder's lifelong nature and the need for supportive and preventive care during bleeding episodes. With careful early diagnosis and proper supportive care Glanzmann's thrombasthenia has a very good prognosis.

Keywords: Glanzmann thrombasthenia, Platelets, Purpura, Ecchymosis, neonate.

Received: 04 February 2026 **Revised: 28 February 2026** **Accepted: 19 March 2026** **Published: 26 March 2026**

Introduction

Glanzmann thrombasthenia (GT) is a rare autosomal recessive platelet functioning disorder (1 in 1,000,000). Dr. Eduard Glanzmann initially reported Glanzmann's thrombasthenia (GT) in 1918, describing a unique platelet anomaly with aberrant appearance on stained film and faulty clot retraction. It is typified by a deficiency in the platelet receptor GPIIb/IIIa, which results in decreased or nonexistent platelet aggregation with physiological agents and clot retraction.[1-4]

Muco-cutaneous bleeding, which can occasionally be fatal, is the most common presentation for patients. Even if the symptoms start in infancy or early childhood, these children are typically diagnosed before the age of five because of recurrent episodes of gum bleeding and epistaxis.[1,2]

In patients with Glanzmann's thrombasthenia, the platelet morphology and platelet count are often within normal levels, but the bleeding duration is noticeably greater. Therefore, the diagnosis of Glanzmann's thrombasthenia is based on the detection of platelet aggregation abnormalities, therefore in order to get a definitive diagnosis, more specialised procedures like gene identification or genetic testing are essential. Early suspicion and diagnosis help parents and patients for better prognosis and follow up care.[3-7]

We present a case of Glanzmann's thrombasthenia that appeared at birth, which we think is the youngest example ever documented in the literature.

Case Report

A New born-non asphyxiated male child (birth order-2) born to non-consanguinous parents through Elective LSCS at Late - In the first few minutes of life, a preterm (36W6D) baby with a birth weight of 3.218 kg had a widespread, non-blanchable petechial rash on the front and rear of the trunk and groin. The mother did not take any anti-platelet, anti-epileptic, or anti-tuberculous medications during the uncomplicated antenatal period. His older sibling was in good health, and there was no noteworthy family background. Apgar scores were 8 and 9 at 1 and 5 minutes, respectively, and the baby weighed 3.218 kg at birth. No extended umbilical stump bleeding or resuscitation attempt was needed. Upon physical inspection, the baby's vital signs were fine and he was well-nourished. The infant was afebrile, lively, and not pale upon evaluation. A widespread petechial rash [Figure 1, 2] and a few ecchymotic areas [Figure 3] were present.

Other bleeding symptoms did not exist. She had a red reflex and

Address for correspondence: Dr. Ganavi Ramagopal, Professor and HOD, Department of Paediatrics, Dhanalakshmi Srinivasan Medical College and Hospital, Siruvachur, Perambalur, Tamil Nadu, India
E-mail: ggganavi10gmail.com

DOI:
10.21276/amit.2026.v13.i1.445

How to cite this article: Salman JM, Sri VJ, Dharshini SRM, Parath AR, Dinaraj A, Ramagopal G.A Case of Glanzmann Thrombasthenia Presenting at Birth- With Petechiae and Ecchymosis- Early Presentation. Acta Med Int. 2026;13(1):805-808.

no dysmorphic characteristics. Her tone was normal, and all four limbs were moving. The length was 46 cm, the occipito-frontal circumference was 34 cm, the Ponderal Index was 3.3, and the anterior fontanelle was normal. Hepatosplenomegaly was absent. The neonatal checkup was otherwise normal.

His platelet counts were normal (265,000/mm3) at the beginning of his work-up and remained normal throughout. 12.4 mg/dl of haemoglobin was present. A blood image revealed sufficient platelets, sporadic large platelets, and no active clump formation. A pathologist examined his initial peripheral blood smear and determined that platelet aggregates were faulty. Other cell lines were normal and there were no unusual cells. The septic test came back negative. Prothrombin time (PT) and PT-INR are within normal limits; however, the activated partial thromboplastin time (APTT) is higher (39.7). There was no evidence of intracranial haemorrhage on the brain ultrasound. Fundusexamination-Normal, No e/o Bleeding manifestation, USG Abdomen and pelvis- Normal, ECHO-Normal. There was no ABO, Rh, and other blood group incompatibility

An additional intravenous dose of vitamin K is administered as a precaution against potentially fatal haemorrhage. He did not require any platelet transfusions. The rash become non progressive (Disappearing rash [Figure4]), no new onset rash over body.

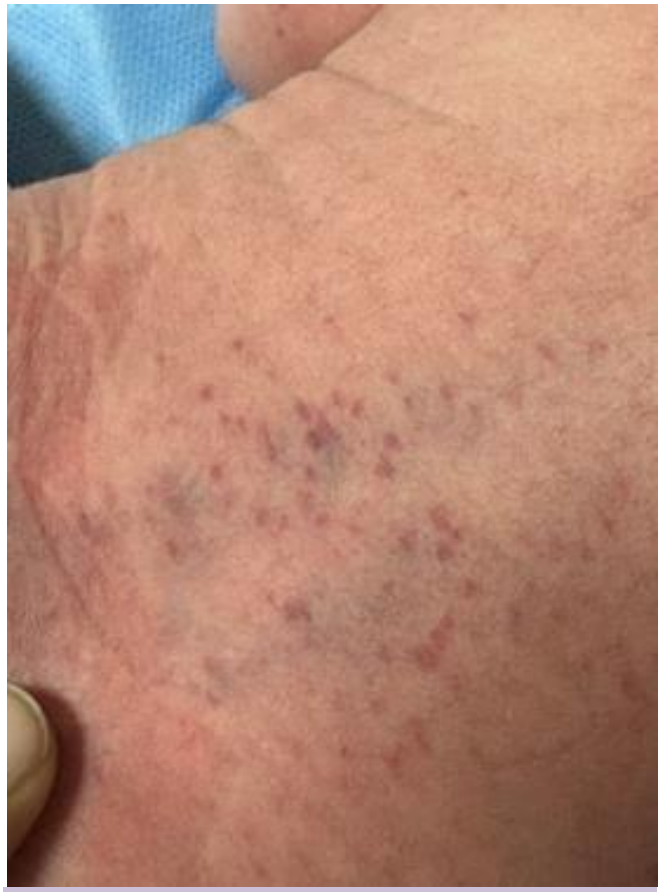

Figure 1: Petechiae seen over the back

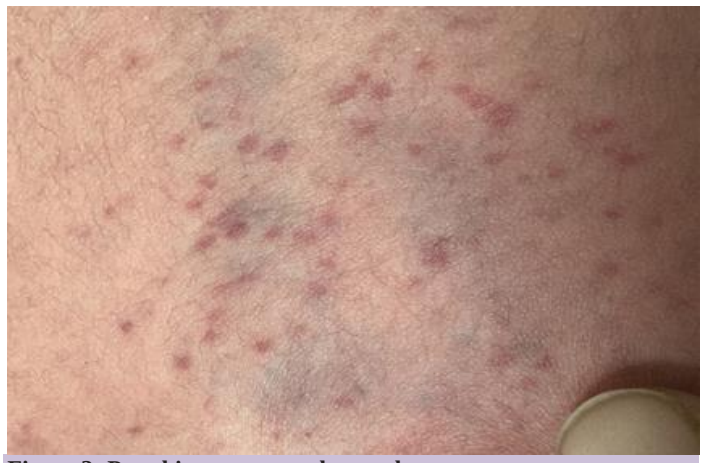

Figure 2: Petechiae seen over the trunk

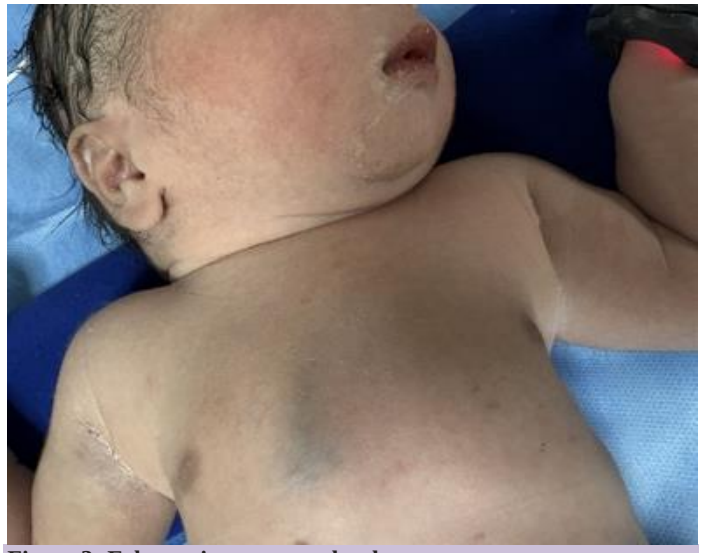

Figure 3: Echymosis seen over the chest

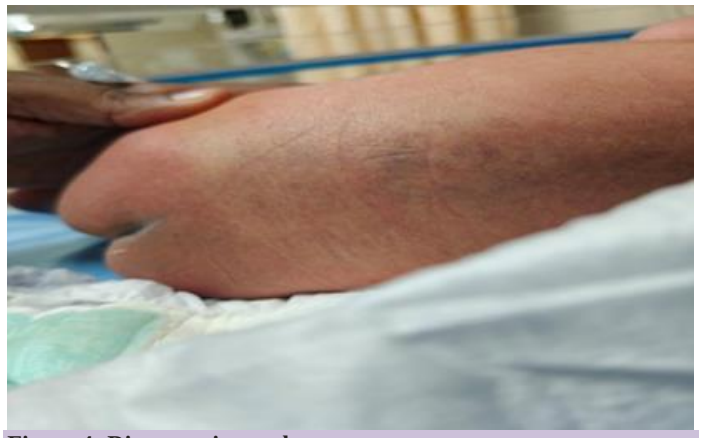

Figure 4: Disappearing rash

Sample sent for Factor assay at Day 2 of life results came as normal. On day seven, a platelet function test showed a main reaction to ristocetin (Only at high doses; response absent at low dose) and no aggregation response to collagen, arachidonic acid, or adenosine-5'-diphosphate (both high and moderate dosages). On flow cytometry absent expression of CD-41, On

Global hemostasis test Reduced maximum clot firmness on ROTEM, suggestive of diagnosis of Glanzmann thrombasthenia.

Although the peripheral blood smear showed many platelets without clumping, the platelet count was within normal limits. Baby on followup without any bleeding problem.

Discussion

First identified in 1918, Glanzmann thrombasthenia (GT) is a congenital bleeding condition brought on by an alpha IIb beta3 platelet integrin abnormality or deficit.[9] The ITGA2B or ITGB3 genes on chromosome 17q21 are mutated in GT, which is inherited as an autosomal recessive condition. Although it typically affects children and young adults, GT can afflict people at any age.[10,11]

The main pathophysiology in this disease is explained as follows, the platelet's fibrinogen receptor is called alpha IIb beta3 integrin, originally GPIIb-IIIa. When platelets are activated, the alpha IIb beta3 integrin transforms into its active form, allowing fibrinogen binding. Platelets agglomerate and offer the crucial primary haemostasis as they bind the fibrinogen. Bleeding can happen on its own or in response to an injury if there are insufficient or nonfunctioning fibrinogen receptors. Furthermore, only when the platelet block is stabilised by cross-linked fibrin—which is also affected by GT—does secondary haemostasis occur. Furthermore, GT platelets have poor thrombin production efficiency, a crucial component of converting fibrinogen to fibrin.[11,12]

History of bleeding or family history of bleeding may be recorded in some cases. Children are especially prone to epistaxis,[10] and gingival bleeding and menorrhagia in females are other typical symptoms,[11] Although it is less common, gastrointestinal bleeding can also occur, and some GT instances go misdiagnosed until an invasive surgery is performed.[11,13] The majority of GT patients have an early diagnosis, with symptoms typically manifesting during the first year of life. Our kid, who was born with bruises and petechiae, is the youngest patient documented in the literature. After circumcision, bleeding may occur and might necessitate a transfusion.[2]

For evaluation of case of GT, Preliminary laboratory tests include a complete blood count (CBC), prothrombin time, activated partial thromboplastin time (PTT), and von Willebrand disease assays include von Willebrand Factor (vWF) antigen, ristocetin cofactor activity, and factor VIII coagulant activity. Next-generation sequencing or platelet function investigations are used for additional assessment. A blood smear, platelet granule release measurement, light transmission aggregometry (LTA), and flow cytometry to examine platelet surface glycoproteins are some examples of screening methods.[14]

The gold standard test, LTA, demonstrates altered platelet aggregation in the presence of agonists other than ristocetin, as we discovered in this instance. 14 Although certain patients with deficient integrins may exhibit normal expression, flow cytometry usually reveals poor expression of alpha IIb beta 3 integrin. And similar in our case the integrin components are recognised as CD61 (beta 3) and CD41 (alpha IIb). A crucial glycoprotein for binding vWF, CD42b, ought to be expressed regularly.10Genetic studies done show Gene mutations in ITGA2B or ITGB3.[15]

Local pressure, cauterisation, sutures, or ice therapy may be the first line of treatment for minor bleeding episodes.[11,12,15]In our case the baby was under monitoring only and Direct breast feeds, no platelet transfusions also given.

Recombinant activated clotting factor VII (rFVIIa) and/or platelets may be required if the patient is having surgery or if the bleeding is unresponsive to local therapies.[11]

Female patients should be checked for iron deficiency and may need therapy of menorrhagia.

Patients with GT should receive counselling and instruction on how to spot unusual bleeding. They should also be told not to take any drugs that affect platelet function and to avoid needless trauma. It is also important to emphasise how important dental hygiene is. Certain patient subgroups, such as young female patients who experience severe monthly bleeding, may require extra care. For certain patients, family planning conversations on the danger of childbirth and impacted offspring are crucial when necessary.[12]

Vocational training, appropriate sports, and other psychological concerns should also be taken care of.[12]

The morbidity of GT can only be decreased by using such a team approach.

Conclusion

GT is one of the rare inherited bleeding disorder, Presenting usually with easy bruising and bleeding from epistaxis and dental extractions. With proper supportive care, GT has a very good prognosis.GT should always be considered as one of the differential diagnosis while evaluating any case of bleeding disorder among any age group including newborn though uncommon.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Jpau Scot, Congenital abnormalities ofplatelet function. In: Kliegman RM, Stanton BMD, St. Geme J, Schor NF,editors. Nelson Textbook of Pediatrics 20thedition. Elsevier; 2016. p.2407.
2. Sebastiano C, Bromberg M, Breen K, Hurford MT. Glanzmann'sthrombasthenia: Report of a case and review of the literature. Int J ClinExpPathol. 2010 25;3(4):443 7.
3. NurdenAT. Glanzmannthrombasthenia. Orphanet J Rare Dis 2006; 1:10.
4. Glanzmann E. HereditaireHamorrhagischethrombasthenic. EinBeitragZurPathologie der Blutplattchen.JahrbuchKinderheilkde. 1918;88:1–42.
5. Morawakkorala RN, Senarathna AMRD,de Alwis ACD, Rathnamala V. Glanzmannthrombasthenia – presenting as prolongedsevere bleeding following minor trauma. Sri Lanka Journal of Child Health 2006;35:36-7.
6. Swathi J, Gowrishankar A, JayakumarSA, Jain K. A rare case of

bleeding disorder: Glanzmann'sthrombasthenia. Annals ofAfrican Medicine 2017; 16(4):196-198.
7. Solh T, Botsford A, Sollh M. Glanzmann'sthrombasthenia: pathogenesis, diagnosisand current and emerging treatmentoptions. Journal of Blood Medicine 2015;6: 219-27.
8. Savithri, K &Ekneligoda, A &Wimalasiri, Aanjeli&Thilakarathna, H &Weerasekera, M. (2020). A case of Glanzmannthrombasthenia presenting at birth. Sri Lanka Journal of Child Health. 49. 83-84. 10.4038/sljch.v49i1.8907.
9. Doherty D, Singleton E, Byrne M, Ryan K, O'Connell NM, O'Donnell JS, Lavin M. Missed at first Glanz: Glanzmannthrombasthenia initially misdiagnosed as Von Willebrand Disease. TransfusApher Sci. 2019 Feb;58(1):58-60. [PubMed: 30551951]
10. Poon MC, Di Minno G, d'Oiron R, Zotz R. New Insights into the Treatment of GlanzmannThrombasthenia. Transfus Med Rev. 2016 Apr;30(2):92-9.
11. Humphreys I, Saraiya S, Belenky W, Dworkin J. Nasal packing with strips of cured pork as treatment for uncontrollable epistaxis in a patient with Glanzmannthrombasthenia. Ann OtolRhinolLaryngol. 2011 Nov;120(11):732-6.
12. Lee A, Poon MC. Inherited platelet functional disorders: General principles and practical aspects of management. TransfusApher Sci. 2018 Aug;57(4):494-501. [
13. Sheikh AY, Hill CC, Goodnough LT, Leung LL, Fischbein MP. Open aortic valve replacement in a patient with Glanzmann'sthrombasthenia: a multidisciplinary strategy to minimize perioperative bleeding. Transfusion. 2014 Feb;54(2):300-5
14. Solh T, Botsford A, Solh M. Glanzmann'sthrombasthenia: pathogenesis, diagnosis, and current and emerging treatment options. J Blood Med. 2015;6:219-27.
15. Gresele P., Subcommittee on Platelet Physiology of the International Society on Thrombosis and Hemostasis. Diagnosis of inherited platelet function disorders: guidance from the SSC of the ISTH. J ThrombHaemost. 2015 Feb;13(2):314-22. [PubMed: 25403439].