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Don't Rely on the First Glanz: Glanzmann Thrombasthenia Hidden in the Shadows of Transfusion-Dependent Anemia: Case Report

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INTRODUCTION

Glanzmann thrombasthenia is a rare autosomal recessive disorder with normal or sub-normal platelet count, prolonged bleeding time, and deficiency or absence of platelet aggregation. A general physical examination does not readily reveal abnormalities associated with GT; hence, misdiagnosis of GT readily occurs. Laboratory tests are essential for a diagnosis of GT.Detection of platelet aggregation defects is the basis of the diagnosis of GT, and it is important to perform more specific methods such as flow cytometry to achieve a definitive diagnosis. A complete cure of GT has not been established; instead, symptomatic treatment is mainly performed. We report a challenging case of GT and explore the features, diagnostics, treatment practices, and prognosis of this disease.

HISTORY

A 5 year 8 months old male child product of non consanguineous marriage with normal growth and development with complaints of spontaneous bruises and mucocutaneous bleeding since early infancy and intermittent episodes of black colored stool and epistaxis, diagnosed with factor 12 deficiency at 1& ½ year of Age. Family history of death due to undiagnosed bleeding disorder and had progressive symptoms for last 6 months in form of black stool ,pallor and generalized weakness requiring blood transfusion, diagnosed as CDA II outside

Presented with chief complaints of

- Spontaneous bruises since infancy
- Epistaxis off and on since 2 years of age
- Black colored stool for 6 months
- Progressive pallor for 6 month

EXAMINATION

On general physical examination, multiple petechiae were noticed on both arms and over the abdomen. Severe pallor was also seen without any lymphadenopathy. Vitals were stable. On anthropometric examination, the child had a normal height, weight, and BMI. On systemic examination, the abdomen was soft and non-tender without any hepatosplenomegaly, and the rest of the systems were essentially normal as well.

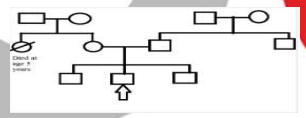
DIFFERENTIAL DIAGNOSIS

Provisional diagnosis was taken as congenital dyserythropoietic anemia with Factor XII deficiency, but due to the points in history and examination contrary to the above, new differential diagnoses were made as follows:

- Clotting disorders which affect platelet function
- Von Willebrand disease
- Congenital afibrinogenemia
- Platelet aggregation defects
- Constitutional bone marrow failure syndromes

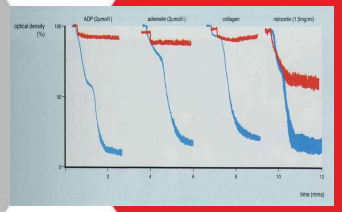
LABORATORY INVESTIGATIONS

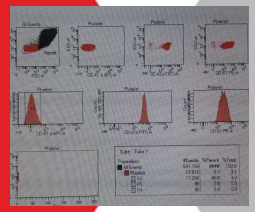
- CBC: 8.1/14390/2.48
- Reticulocyte count: 8%
- Peripheral smear: Normal
- Stool for occult blood: Positive
- Hypercellular marrow with marked erythroid hyperplasia with features of dyserythropoiesis and nuclear changes
- G6PD: Normal
- OF: Normal
- DCT: Negative
- HPLC: Normal for the patient; father has a beta-thalassemia trait
- Flow cytometry: Suggestive of Glanzmann's
- Platelet aggregation studies: Absent or markedly impaired aggregation to all agonists



- Bleeding time 11 minute 5 seconds
- Clotting time 7 minute 30 seconds
- Prothrombin time/INR—14/1.09
- Thrombin time—18/18©
- APTT—34/34©
- CRT 48 %

	Heading	Headig	Heading
CD41a	100 %	100 %	5 %
CD42a	100 %	100 %	100 %
CD100	100 %	100 %	84.5 %





MANAGEMENT

Treatment:

- Packed cell transfusion was administered in view of severe pallor.
- Fibrinolytics were given.

Family Counseling:

- The family was counseled about the prognosis and the risk of bleeding.
- Danger signs were explained.
- They were also counseled on activities to be avoided.

Further Course:

- Child is under follow up
- Gut bleeding stopped after use of fibrinolytics
- RBC scan to identify the source of bleeding is planned if recurrence
- No further transfusion required
- Siblings' work up done showing normal platelet function and CD41a levels

CONCLUSION

Diagnosing Glanzmann Thrombasthenia (GT) is challenging as it mimics common bleeding disorders. GT should be considered in patients with severe bleeding from minor trauma, recurrent GI bleeds, or epistaxis, especially with a family history of consanguinity or affected members. Symptoms range from mild to life-threatening and may improve with age. GT is often misdiagnosed due to its similarity to common gastrointestinal bleeding disorders.