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Case Study

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UNEXPLAINED BLEEDING - MANIFESTATION OF GLANZMANN'S THROMBOSTHENIA

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ABSTRACT

Glanzmann's thrombosthenia(GT) is a rare inherited platelet disorder characterized by deficiency of platelet fibrinogen receptor alpha IIb β3.[1] Here we report two cases of GT from single family born to parents with 3 degree consanguineous marriage. Among the two cases, older sibling i.e 3.5years old female child presented with ecchymosis and epistaxis and second patient, her younger brother aged 1 year had history of gum bleeding and ecchymosis. GT is rare bleeding disorder. Still, in a case of unexplained bleeding in a child, one should suspect platelet function disorder. With proper supportive care Glanzmann's thrombasthenia has a very good prognosis.[2]

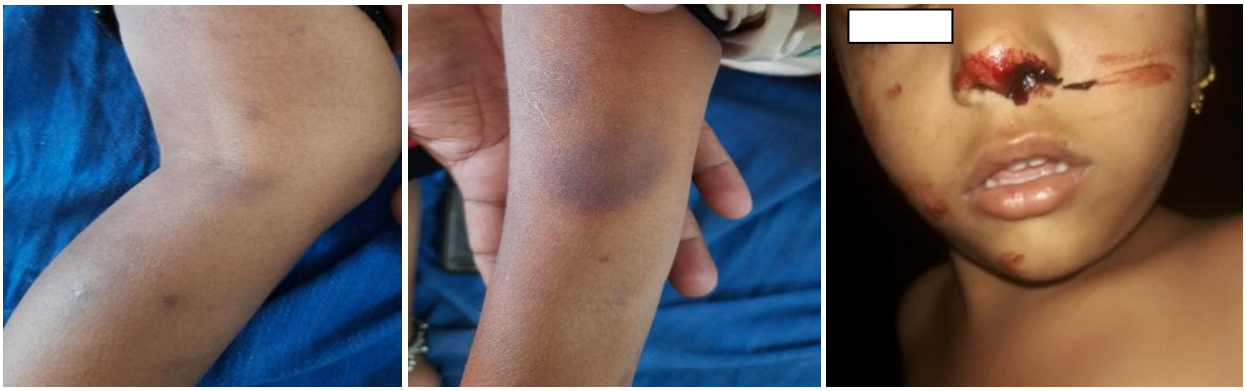
INTRODUCTION

Glanzmann's thrombosthenia ia a rare congenital bleeding disorder characterised by impaired platelet function, caused by deficiency of platelet fibrinogen receptor alpha IIb β3.[1] It is an Autosomal recessive disorder. Child with GT generally presents with mucocutaneous bleeding including epistaxis, gingival bleeding, GI bleeding & increased susceptibility to easy bruising. It occurs 1 in 1,00,000 with equal sex predilection.[1] One should suspect GT when there is unexplained bleeding with normal platelet count with no clumps of platelets on peripheral smear examination (on finger prick sample) with normal bleeding profile. Similar history in the family strongly points towards a bleeding disorder. We report two siblings born through consanguineous marriage who presented with bleeding manifestations, who were found to have Glanzmann's disease.

CASE 1: 3.5 years old female child born was brought by parents with c/o petechiae on body on and off since birth, h/o ecchymosis on and off since 1 year, h/o epistaxis, spontaneous in onset 2months back. h/o blood transfusion at 3 years of age for anemia (CBC s/o Hb 3g/dl, TLC 5800/cumm, platelet count 2,77,000/cumm, peripheral smear examination s/o microcytic, hypochromic with anisopoikilicytosis). No further investigations were done at that time. Again after 2 months child had epistaxis from one nostril following trivial trauma, which lasted for 2 days and resolved spontaneously. Family history s/o ecchymosis in younger brother on and off since birth. Birth history: born through LSCS, no significant neonatal history. Child was referred to our hospital for further investigations. On admission child had ecchymosis all over body. No active bleeding from any other site.

o/e: Afebrile, HR 88/min, RR 20/min, BP 86/50 mm of hg. Ecchymosis present on upper limbs, lower limbs, back. No pallor, icterus, lymphaenopathy, edema. No hepatosplenomegaly. Rest systemic examination was normal.

So our differential diagnosis was Platelet Disorder or Coagulation Disorder. Investigations like CBC with PS, PT, APTT, TTwere sent. CBC showed hb 10.6g/dl, TLC 7400/cumm, platelets 3, 08, 000/cumm PS: Normocytic, Normochromic, platelets adequate. PT 14.4 (P) / 15.1(C), APTT 28.6(P)/ 27.7(C), TT 13.3(P) / 15.1(C). Peripheral smear(finger prick sample) was reviewed by haematologist where platelets were adequate in number but **they were not in clumps**. So we suspected functional platelet disorder. We subjected patient for platelet aggregation studies. Aggregation studies showed absent aggregation with all agonists (ADP,ATP,Collagen) except with Ristocetin. This was confirmed by flow cytometry. Flow cytometry was suggestive of absent GP IIbIIIa, GP IIb receptor. This confirmed Glanzmann's Thrombosthenia.



Case 2: 1.5 years male child, younger sibling of above patients had h/o ecchymosis on and off since birth and episodes of spontaneous gum bleeding since last 6 months. No hepatosplenomegaly, lymphadenopathy, pallor or joint swelling.

CBC - Hb 10g/dl, TLC 9700 /cumm, Platelet count 3.32/cumm, Platelets on peripheral smear examination (finger prick sample) were adequate in number but no platelet clumps were seen. PT 14.5/13.5, APTT 27.9/28.7, TT 13.5/15.5. Flowcytometry was suggestive of absent GP IIbIIIa, GP IIb receptor. This confirmed Glanzmann's Thromboasthenia.

DISCUSSION

Glanzmann's thrombasthenia was first documented in 1918 by Dr. Eduard Glanzmann, who described a functional abnormality of platelets with defective clot retraction.[2] Glanzmann Thrombasthenia is a rare congenital disorder, which is Autosomal recessive. It is caused by deficiency of platelet surface fibrinogen receptor αⅡb-β3 (GPⅡb-Ⅲa), the major integrin complex on the platelet surface that undergoes conformational changes by inside-out signaling when the platelets are activated.[1] The exact incidence has been difficult to calculate, but is estimated to be 1 in 1,000,000.[3] This equally affects females and males.

One should suspect platelet function disorders if child presents with h/o easy bruising and bleeding manifestations. Also family history of bleeding strongly points towards bleeding disorder. Epistaxis is particularly prevalent in children,[4] and other common manifestations are menorrhagia and gingival bleeds. Gastrointestinal bleeding is less frequently reported, and some cases of GT are undiagnosed until an invasive procedure is performed 5. Most patients with GT are diagnosed at an early age, with symptoms often appearing after birth or shortly thereafter. Severity of symptoms varies greatly. Some children have only mild bruising but some may have severe bleeding manifestations leading to life threatening events.

On evaluation these patients have prolonged bleeding time, Normal platelet count with normal size and morphology on peripheral smear they are not seen in clumps as seen in normal patients. Bleeding profile like PT, APTT will be normal. Aggregation studies show abnormal or absent aggregation with all agonists (ADP, ATP, Calciumions, Collagen) except with Ristocetin because Ristocetin agglutinates platelets and doesn't require metabolically active platelets. Diagnosis is confirmed by flow cytometry analysis of patient's platelet glycoprotein. Genetic tests can identify the DNA mutations responsible for the disorder.

Treatment

Initial local measures like cold compression, pressure application, topical thrombin, nasal packing to stop bleeding (especially epistaxis)[5]; Antifibrinolytic agents like tranexamic acid and epsilon amino caproic acid resolves mild to moderate mucocutaneous bleeding.[3] Treatment with rFVIIa have shown promising results.[3,4] Platelet transfusion is required when above methods fails, PCV transfusion is needed if child has anemia as a result of profuse bleeding. Hematopoietic stem cell transplantation is the curative treatment for severe recurrent bleeding episodes or in patients who develope platelet antibodies.[2]

CONCLUSION

GT is one of the rare bleeding disorder. Any unexplained bleeding in a child should be investigated for platelet function disorders. With proper supportive care child can be managed well. Those with severe frequent bleeding with life threatening events will need bone marrow transplantation as a curative therapy.

Declaration of patient consent

The authors certify that they have taken informed and written consent of relatives. Relatives had given consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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