GLANZMANN THROMBASTHENIA - A CASE REPORT OF RARE BLEEDING DISORDER

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Introduction: Glanzmann thrombasthenia (GT) is a rare autosomal recessive bleeding disorder associated with severe platelet dysfunction. GT is due to quantitative or qualitative defects of the platelet membrane integrins αIIb and β3, resulting from mutations in ITGA2B and/or ITGB3 genes. These integrins form platelet glycoprotein GP IIb/IIIa, which acts as the platelet fibrinogen receptor, and is thus essential molecule to platelet aggregation and hemostasis. Patients tend to present in early childhood with easy bruising and severe mucocutaneous bleeding.

Case report: A 2-year-old girl presented with recurrent massive epistaxis from infancy often associated with consecutive severe anemia. Additionally, she had a history of easy and spontaneous bruising in her extremities and prolonged bleeding following immunization. Family history was unremarkable and there were no known bleeding disorders in the family. Investigations revealed normal platelet count and normal morphology on peripheral blood smear. Screening test of the hemostatic system (PT, APTT and TT) was normal. Von Willebrand disease was ruled out as well. Platelet aggregation studies showed normal platelet aggregation with ristocetin and very reduced platelet aggregation with ADP and collagen. The GPIIb-IIIa expression rate`s by flow cytometry were markedly decreased confirming the diagnosis of GT. Epistaxis were successfully managed with supportive care including a nasal pack insertion and antifibrinolytic therapy. Platelet transfusion was given only in one excessive episode.

Conclusion: GT is a rare inherited bleeding disorder but should always be considered as differential diagnosis while evaluating any case of bleeding disorder. With careful early diagnosis and proper supportive care, GT has a very good prognosis.