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Glanzmann's thrombasthenia: updated. Nair S, Ghosh K, Kulkarni B, Shetty S, Mohanty D.

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Glanzmann's thrombasthenia is an autosomal recessive disorder, rare in a global context, but a relatively more common platelet function defect in communities where consanguineous marriages are more frequent. On clinical grounds alone, it cannot be distinguished from other congenital platelet function defects. Epistaxis, gum bleeding, menorrhagia are the common clinical manifestations, whereas large muscle hematoma or hemarthrosis seldom occur in these patients. Essential diagnostic features are a normal platelet count and morphology, a greatly prolonged bleeding time, absence of platelet aggregation in response to ADP, collagen, epinephrine, thrombin and to all aggregating agents which ultimately depend on fibrinogen binding to platelets for this effect, flow cytometry, studies of GPIIb-IIIa receptors on the platelet membrane surface using monoclonal antibodies. The present review describes some of the uncommon features of the disorders and the currently available options which the treating physicians should be aware of during the management of these patients. Although by definition all patients with Glanzmann's thrombasthenia have a virtually complete failure of platelet aggregation, a number of variant forms of GT have been described in which the glycoproteins are present in normal or near normal amounts but are functionally defective. Understanding the pathophysiology of the disorder by the treating physicians is of utmost importance. Presence of high affinity platelet receptors resulting in thrombasthenia-like phenotype may require an antagonistic treatment atypical of classical GT management. It has now been established that different genetic mutations of either GPIIb or IIIa genes results in such a heterogeneity of thrombasthenia phenotype. Glanzmann's thrombasthenia is a paradigm for treating coronary artery disease patients with GPIIb-IIIa antibody and inhibitors. By using these medicines we create a temporary GT-like situation. Hence, understanding this disease is of utmost importance to the practicing cardiologist. As mutations for different variant forms of GT become known, our understanding of how GPIIb-IIIa molecules can be activated to act as a receptor for fibrinogen molecules will be increased. Such understanding undoubtedly will help us to devise better drugs with GPIIb-IIIa inhibitors. Molecular biology techniques have enabled us to equivocally detect heterozygote carriers who are clinically asymptomatic. However, there may be several laboratories in the developing world, which have no access to molecular biology techniques. Development of more robust techniques of quantitation of platelet receptors has enabled an accurate diagnosis of heterozygote carriers or an unborn fetus in the second trimester. The importance of the GPIIb-IIIa polymorphisms in carrier and prenatal diagnosis has not been properly studied. Nowadays, the less direct method of PLAI typing (determination of the levels of platelet antigen) of the foetal platelets as early as 16 weeks of intrauterine life can be used for prenatal diagnosis of GT.