

ABSTRACT

Glanzmann's thrombasthenia (GT) is an autosomal recessive inherited bleeding disorder due to a defect in platelet function. The hallmark of this disease is severely reduced/absent platelet aggregation in response to multiple physiological agonists. Bleeding signs in GT include epistaxis, bruising, gingival hemorrhage, gastrointestinal hemorrhage, hematuria, menorrhagia, and hemarthrosis. Homozygous or compound heterozygous mutations in the genes of GPIIb and GPIIIa lead to GT. A patient with GT, with no possible causative mutations in GPIIb and GPIIIa genes, may harbor defects in a regulatory element affecting the transcription of these 2 genes. GT occurs in high frequency in certain ethnic populations with an increased incidence of consanguinity such as in Indians, Iranians, Iraqi Jews, Palestinian and Jordanian Arabs, and French Gypsies. Carrier detection in GT is important to control the disorder in family members. Carrier detection can be done both by protein analysis and direct gene analysis.