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Picture Prognosis

A 14-month-old boy born to first-degree consanguineous parents presented with recurrent gingival bleeding, spontaneous epistaxis, prolonged bleeding after vaccinations, and widespread ecchymoses. Platelet count, PT, aPTT, factors VIII/IX, and von Willebrand factor levels were normal. Platelet aggregation studies showed absent aggregation to all agonists except ristocetin. What's your diagnosis?

1. Hemophilia A
2. Glanzmann Thrombasthenia
3. Factor XIII Deficiency
4. von Willebrand Disease

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Answer: Glanzmann Thrombasthenia

The infant had recurrent gingival bleeding, epistaxis, ecchymoses, normal platelet count, normal PT/aPTT, and platelet aggregation absent with all agonists except ristocetin, which confirmed Glanzmann thrombasthenia. The history of consanguinity and a family history of severe unexplained bleeding further supported this diagnosis. The patient had normal factor VIII levels (121.8%) and presented primarily with mucocutaneous bleeding, whereas hemophilia A is associated with factor VIII deficiency. von Willebrand factor activity (85.6%) and antigen levels (83.2%) were normal, excluding von Willebrand disease in this case. Although factor XIII testing was not performed, the diagnosis was established by the characteristic platelet aggregation study findings, which demonstrated a platelet function disorder consistent with Glanzmann thrombasthenia.