LABORATORY DIAGNOSIS OF CLINICALLY RELEVANT PLATELET FUNCTION DISORDERS

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ABSTRACT
Inherited platelet function disorders (IPFDs) represent a significant fraction of congenital hemorrhagic disorders, and may be associated with bleeding of considerable severity. IPFDs may be difficult to diagnose and a preliminary accurate clinical examination and an objective evaluation of the severity of the bleeding history are mandatory. The laboratory investigation of IPFDs should follow a rational algorithm based on a streamlined panel of laboratory tests with subsequent steps of increasing levels of complexity. First screening tests include platelet count, peripheral blood smear, light transmission aggregometry, measurement of platelet granule content and release, and the expression of glycoproteins by flow cytometry. Several of these tests have been largely employed, and a few validated, for the diagnosis of IPFDs and some recent developments are discussed. Point-of-care tests may provide the advantage of rapidity and the possibility to study platelet function in whole blood, but further studies are required to clarify their potential diagnostic application. Genotyping is recommended for some conditions (genotype/phenotype correlations, forms associated with a high risk of developing hematologic malignancies) but, especially when carried out by next generation sequencing (NGS) techniques, needs to be critically evaluated taking into account clinical and laboratory phenotypes.