

ANALYTICAL DISCREPANCY IN PLATELET COUNTING REVEALING MYH9-RD: A CASE REPORT

M. De Girolamo, A. Marrocchi, F. Vitone, M. Scapaticci, M. Bovara, T. Papillo, R. Mancini

Laboratorio Unico Metropolitano, Bologna, Italy

Background. MYH9-related disease (MYH9-RD) is a rare autosomal dominant disorder caused by pathogenic variants in the MYH9 gene, encoding non-muscle myosin IIA. It is characterized by congenital macrothrombocytopenia, with giant platelets and a variable bleeding tendency, and may be associated with extra-hematological manifestations such as sensorineural hearing loss, nephropathy, cataracts, and liver enzyme abnormalities. Although thrombocytopenia is often stable over time, the condition is frequently underdiagnosed and may be misinterpreted as immune thrombocytopenia, leading to inappropriate management. Morphological hallmarks include giant platelets and Döhlelike inclusions in the neutrophil cytoplasm. Accurate diagnosis relies on integration of clinical, laboratory, and genetic findings, and is essential for appropriate patient management and follow-up.
Case Report. A 31-year-old pregnant woman underwent routine first-trimester blood testing, revealing severe thrombocytopenia (5,000/μL) measured by impedance methodology, with otherwise normal hematological parameters. Platelet histogram and leukocyte scattergrams suggested the presence of giant platelets. Subsequent platelet counting using fluorescence methodology showed a higher value (19,000/μL), highlighting a significant discrepancy between the two methods. Peripheral blood smear examination confirmed true thrombocytopenia, excluded platelet clumping, and demonstrated numerous giant platelets, some comparable in size to erythrocytes. Additionally, basophilic cytoplasmic inclusions were observed in neutrophils. Further clinical history revealed a known diagnosis of MYH9-related disease, and the inclusions were interpreted as Döhle-like bodies, a pathognomonic feature of the condition. Longitudinal data showed persistently low but stable platelet counts (mean ~30,000/μL). No evidence of renal involvement was found, with no proteinuria or hematuria, while mild elevation of liver enzymes suggested possible hepatic involvement. The discrepancy between counting methods was attributed to the presence of macroplatelets, which may be misclassified as red blood cells inmpedance-based systems, whereas fluorescence methods provide more accurate platelet identification.
Conclusions. This case highlights the importance of a comprehensive and integrated approach to thrombocytopenia, particularly during pregnancy, when common benign or gestational causes may obscure rare inherited conditions. Morphological evaluation of peripheral blood smear remains essential for identifying key diagnostic features such as giant platelets and neutrophil inclusions. The observed discrepancy between platelet counting methods underscores the limitations of impedance-based systems in the presence of macrothrombocytes and supports the use of fluorescence-based techniques for accurate quantification. Awareness of MYH9-related disease laboratory features is crucial to avoid misdiagnosis and inappropriate treatment, ensuring proper clinical management and follow-up.

