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New-Generation Ektacytometry Study of Red Blood Cells in Different Hemoglobinopathies and Thalassemia

Abstract: Next-generation ektacytometry, as provided by the osmoscan module of the Laser Optical Rotational Red Cell Analyser (LoRRca) MaxSis, is currently one of the most advanced complementary diagnostic tools for congenital rare anemias associated with red blood cell (RBC) defects. Osmotic gradient ektacytometry (OGE) is considered the gold standard for diagnosing RBC membrane disorders, particularly hereditary spherocytosis (HS). A hallmark of hereditary hemolytic anemias is impaired RBC deformability, which leads to reduced cell survival and is generally attributed to abnormal cell shape, increased rigidity, or dehydration.

To date, next-generation ektacytometry has been primarily employed for the differential diagnosis of RBC membranopathies, while its application in structural hemoglobinopathies and thalassemia remains limited. However, with the recent development of novel therapeutic strategies for hemoglobinopathies, particularly sickle cell disease and β-thalassemia, there is growing clinical interest in ektacytometry, warranting further exploration.

In this study, we evaluated the OGE profiles obtained using the osmoscan module of the LoRRca ektacytometer in 96 patients with different hemoglobinopathies, including both structural variants and thalassemia. Our objective was to assess the utility of OGE for the early diagnosis of these disorders, either in isolation or in co-inheritance with other hereditary RBC defects. Furthermore, we aimed to enhance our understanding of the contributions of RBC deformability, osmotic fragility, and intracellular viscosity to the pathophysiology of hemolysis, particularly in the context of rare anemias.

Our findings indicate that the osmoscan profile provides valuable complementary insights into RBC deformability and hydration homeostasis, which may improve our understanding of the mechanisms underlying reduced RBC survival and hemolysis in affected patients.

Biography: Prof. Joan Lluís Vives Corrons is Professor Emeritus at the University of Barcelona and an honorary researcher at the Josep Carreras Institute for Leukaemia Research (IJC). He specializes in rare anemias, focusing on the molecular and genetic mechanisms of red blood cell (RBC) disorders, including enzymopathies, membranopathies, and hemoglobinopathies.

For over 30 years, he led the Haematology Laboratory Department (1976-1997) and the Red Blood Cell Pathology Unit (1998-2016). As principal investigator of 35+ projects, his research has centered on erythroenzymopathies (e.g., G6PD and PK deficiencies), hemoglobinopathies (e.g., sickle-cell disease and thalassemia), and hereditary hemolytic anemias.

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