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Diagnostic approach of Hereditaryred blood cell membranopathies: from osmotic gradient ektacytometry (oge) to next generation sequencing (ngs)

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Hereditary red blood cell (RBC) membranopathies are characterized by mutations in genes encoding skeletal proteins that alter the membrane complex structure. Hereditary spherocytosis (HS) is the most common inherited RBC membranopathy leading to hereditary hemolytic anemia with a worldwide distribution and an estimated prevalence, in Europe, of about 1:2000 individuals. The recent availability of targeted next generation sequencing (t-NGS) and its combination with RBC deformability measured with a laser-assisted optical rotational ektacytometer (LoRRca) has demonstrated to be the most powerful contribution to lower the percentage of hereditary hemolytic anemia undiagnosed cases. In order to know the kind and frequency of RBC membrane mutations in our geographical area (Catalonia) and to better understand their pathophysiology, 42 unrelated, non-transfusion- dependent (NTD) patients with hereditary hemolytic anemia have been studied by combining t-NGS and LoRRca. The osmoscan module of LoRRca provides three rheological profiles that reflect the maximal deformability (Elmax), osmotic fragility (Omin), and hydration state (Ohyper) of RBCs and contribute to a better understanding of the contribution RBC rheology to the severity of anemia. From the 42 patients studied, 37 were suspected to be a RBC membrane defect due to phenotypic characteristics and abnormal RBC morphology and, from these, in 31 patients (83.8%of cases) the mutation was identified by t- NGS. No definite diagnosis was achieved in 11 patients (26.2% of cases), including 6 out of 37 cases, with suspected membranopathy, and 5 with unclassifiable HHA. In all these undiagnosed patients, the existence of hemoglobinopathy and/or enzymopathy was ruled out by conventional methods

Biography

Joan-LLuis Vives Corrons is Emeritus Professor of the University of Barcelona and Group Leader of the Rare Anemias Unit at the Institute for Leukaemia Research Josep Carreras in Catalonia. He has been the Head of the Haematology Department at the Hospital Clinic of Barcelona from 1976 to 1996, and the founder of the Red Blood Cell Pathology Unit from 1997 to 2016. This Unit is the largest referral center for the diagnosis of anemias , congenital and acquired. Research activity has been focused on the hereditary red blood cell defects (enzymopathies , hemoglobinopathies , membranopathies and CDA). He developed a fellowship in Paris,France (Haematology Department of Hôpital Beaujon) and in la Jolla California, USA (Scripps Research Institute, Department of Molecular and Experimental Medicine). Prof. Vives Corrons has published more than 600 professional papers and reviews and has edited three books on hematology and rare anemias

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