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Editorial

Awareness of rare genetic diseases in daily clinical practice

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In this issue, we are pleased to present 4 multidisciplinary papers that include disciplines such as Pediatric Genetics, Hematology, and Nephrology. These high quality papers represent the efforts of months of specialist colleagues in the area of multidisciplinary clinical genetics. Each contribution represents a tool for education and teaching at multiple levels of medical learning.

The work published by **De La Rosa, Servalle and Bigay from the Dominican Republic** emphasizes and demonstrates the main comorbidities in the pediatric population of the Dominican Republic suffering from Down Syndrome. The impact of these findings has the potential to have an impact on public health in this sister country and the region, allowing health systems to increase screening for comorbidities at the primary care level. One of their findings is the confirmation that the increase in comorbidities occurs with maternal ages between 40 and 44 years, revealing that up to 22% of patients may present one or more comorbidities.

The case report presented by **Sanchez-Vargas and Moreno Giraldo from Colombia** presents a rare case called Familial Hemophagocytic Lymphohistiocytosis. Despite having an extremely low incidence (0. 12-1. 0 per 100,000 inhabitants), in many countries it has not yet been incorporated into the official list of rare and neglected diseases. In a way, the authors present the problem of recognition of this disease by means of a case report. This publication serves as an educational tool for better recognition of this clinical entity, its reporting, and therefore, increased early management of these patients.

The second case report by **Austin and Villegas from the Republic of Panama** presents a case of aggrecanopathies, identifying a variant by complete genomic sequencing. Here they describe the phenotypic variability that these patients may present in terms of stature, dysmorphias, and the need to differentiate them from other skeletal dysplasias and collagenopathies whose manifestations may be similar.

Finally, the research group in nephrology **of Courville and Bustamante from Panama**, make a detailed bibliographic review of membranous nephropathy, its genetic origins, associated triggering factors, and make a review of the antigens that have been associated to date with this renal entity. This work allows visualizing the importance of genetic variants in genes such as PLA2R1, THSD7A, and the EXT1 and EXT2 genes, which are involved in most of the membranous nephropathies of genetic origin.

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It is hoped that this issue can serve as a reference for specialists in Genetics, Nephrology, Hematology, and Pediatrics for a new vision of these pathologies and in benefit of the population that suffers them. We also hope that this knowledge could be transmitted and implemented in primary care levels for the better recognition and early diagnosis of these rare conditions and comorbilities.

We hope that this reading will be plesant, and we also invite you to participate as an author with us in this multidisciplinary clinical genetics regional effort.

Best regards,

Jorge Mendez Ríos

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