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Genetics and Clinical Genomics

EDITORIAL

Editorial - Case reports as regional references

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Dear colleagues and readers,

We are pleased to present this new issue of the journal, which brings together original contributions that reflect both the scientific depth and the growing regional leadership of biomedical research in Latin America. The articles included in this edition span conceptual reflection, molecular genetics, and clinically grounded case reports, offering readers a diverse and meaningful perspective on contemporary challenges in medicine and genomics.

A distinctive feature of this issue is its engagement with fundamental questions about life and disease. The review article “Genetics and Entropy: Turning our gaze toward the thermodynamics of biological systems” proposes an integrative view that connects genetics, biology, and physics through the concept of entropy. By revisiting classical and modern scientific thought, the authors invite readers to consider biological organization, evolution, aging, and cancer as expressions of underlying thermodynamic principles. This discussion is further enriched by a Letter to the Editor from Panama, which reflects on entropy as a conceptual tool in medical practice and highlights its potential to deepen our understanding of human health and disease.

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The issue also places strong emphasis on rare genetic disorders through carefully documented case reports that illustrate real diagnostic and therapeutic challenges. From Panama, the report on vitamin D–dependent rickets type 1A caused by a pathogenic CYP27B1 variant underscores the importance of early genetic diagnosis and demonstrates how effective treatment strategies can be implemented even in settings where access to standard therapies is limited.

Two contributions from Colombia address the clinical complexity of chromosomal duplication syndromes. The case describing 22q11.2 duplication syndrome highlights the wide phenotypic variability associated with this condition and reinforces the value of chromosomal microarray analysis in

achieving accurate diagnosis and guiding management. In a similar vein, the report on 15q11–q13 duplication syndrome (Dup15q) illustrates how advanced genomic testing can clarify long-standing neurodevelopmental phenotypes, inform prognosis, and support appropriate genetic counseling for patients and families.

Completing this issue is a review from Mexican authors examining miransertib as an emerging therapeutic option for Proteus syndrome, a rare disorder driven by abnormal activation of the PI3K/AKT pathway. This article provides a concise and clinically relevant overview of targeted therapy in a condition for which treatment options remain limited, emphasizing the growing role of precision medicine in rare diseases.

Collectively, the articles in this issue highlight the strength of regional clinical experience, the increasing integration of genomic technologies into medical practice, and the importance of interdisciplinary thinking. They reflect a shared commitment to advancing knowledge while addressing the practical realities faced by clinicians and researchers in our region.

We extend our sincere thanks to the authors and reviewers for their valuable contributions and invite our readers to engage with the insights and cases presented in this new issue.

Sincerely,

Dr. Jorge D. Méndez-Rios
Editor-in-Chief
Genetics and Clinical Genomics