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Genetics and Clinical Genomics

EDITORIAL

A Genomic Revolution: Advancing Health and Knowledge in Latin America

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As we enter the second year of the Genetics and Clinical Genomics Journal, there is much anticipation for the future of this field in the region. It's remarkable to see how far we've come in such a short time. This journal was born from a vision to bring together the best of genetics and genomics research and make a meaningful impact on healthcare and education in our region. That vision is becoming a reality, thanks to the incredible contributions of our authors and the support of our readers.

This issue features an impressive selection of articles that highlight the power of genomic research to transform lives.

Our Case Reports section includes stories of rare and complex conditions where genetics has played a critical role:

In **Hypotonic syndrome** as a manifestation of an ultra-rare disease caused by a novel de novo variant in the PLA2G6 gene, we see the intricate process of solving a diagnostic mystery and its profound implications for patient care.

De novo variant in the COL1A1 gene associated with an orphan genetic disease: **Type I Osteogenesis Imperfecta** sheds light on a rare condition that challenges both families and clinicians but also presents opportunities for targeted treatments.

De novo genetic variant in **Epileptic Encephalopathy**: The importance of specific diagnosis highlights how pinpointing a genetic cause can lead to more effective therapeutic strategies.

Finally, Detection of a genetic variant in Apert syndrome examines a condition that, while rare, has significant developmental and therapeutic implications.

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Our Review Articles, **Advances and perspectives on genetic pathologies in the 21st century**, provide valuable overviews of current trends and emerging possibilities, serving as essential guides for anyone engaged in genetic research or clinical practice.

Why This Matters?

Genetics and genomics are no longer rare disciplines they're reshaping how we understand health and disease. In our region, this knowledge has the potential to address long-standing healthcare inequities, ensuring that advances in precision medicine benefit all communities, not just a select few.

Equally exciting is the role genetics can play in education. Training the next generation of healthcare providers and researchers to harness the power of genomics is essential. By investing in education, we're laying the groundwork for a future where genomic medicine is fully integrated into everyday practice.

This journal is a testament to what's possible when we work together—scientists, clinicians, educators, and policymakers. It's an invitation to keep pushing boundaries, asking tough questions, and finding answers that matter.

Thank you for joining us on this journey. I hope you find this issue inspiring, thought-provoking, and a reminder of the incredible potential within genetics and genomics to create a healthier, more equitable world.

Jorge Méndez

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