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EDITORIAL

Understanding Molecular and Genomic Pathophysiology: Key to Effective Molecular Diagnosis

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Abstract

In this issue, we present three original studies that address various rare genetic conditions. The first focuses on West syndrome and provides a detailed analysis of its diagnosis along with a proposed therapeutic approach to this complex condition. The study emphasizes the importance of early diagnosis and timely, appropriate intervention. Researchers from the Universidad de La Sabana in Chía, Colombia, have done an outstanding job highlighting key aspects of clinical management and listing the genes associated with this disorder.

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The second original study was conducted by the team at the Genomics Institute of the Social Security Fund in Panama. The report includes 78 cases and describes the variants found in the *SERPINA1* gene, which is associated with alpha-1 antitrypsin deficiency due to proteolytic degradation. This work is of great importance as it represents one of the first studies of its kind in Panama and the region.

The third original contribution, conducted by medical students, presents an analysis of the evidence listed in OMIM regarding the *MTHFR* gene, which has been linked to multiple conditions such as neural tube defects, schizophrenia, thrombosis, and abnormalities in homocysteine levels. This review highlights the most compelling evidence while also identifying gaps in our understanding of this gene and its clinical impact. The study was carried out by an enthusiastic group of students from the School of Medicine at the Interamerican University of Panama.

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Finally, a research group from the Chromomed Center in the Dominican Republic presents a case study and review of a rare instance of diploid/triploid mosaicism. An exhaustive investigation was conducted using multiple molecular, cytogenetic, and imaging methods. This study stands out for the complexity of the case and the richness of the descriptive evidence, making a significant contribution to the understanding of clinical genetics and genomics in our region.

Undoubtedly, these contributions are pioneering and offer significant academic value for training and advancing our understanding of molecular diagnostic technologies. Although such technologies have supported genetic diagnosis in Latin America for decades, ongoing training and the development of a new generation of professionals focused on the molecular mechanisms of disease are still needed. Medicine today is personalized and will continue to focus on the molecular details of each condition, ultimately leading to better healthcare for our population.

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