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EDITORIAL

From Variants to Vision: Reframing Precision Medicine in Diverse Populations

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This issue of Genetics and Clinical Genomics brings together a collection of studies that, although diverse in scope, from thrombotic risk to ultra-rare syndromes and oncogenomics, converge on a central theme: the urgent need to contextualize genomic medicine within real populations, with real limitations and concrete clinical dilemmas.

At the core of this issue lies a question that continues to challenge our discipline: how rapidly are we implementing precision medicine, and when will therapies become available for our patients within their specific genetic context?

The Myth of the Universality of Genetic Risk

The study on thrombophilic variants, specifically Factor V Leiden, prothrombin G20210A, and the C677T variant of the MTHFR gene, offers a critical reminder: genetic risk is not universally transferable across populations.

While Factor V Leiden and the prothrombin variant showed an association with increased thrombotic risk in the Panamanian cohort, the absence of association with MTHFR C677T highlights a persistent issue in clinical genomics: the excessive extrapolation of findings derived primarily from populations in other regions.

This is not merely an academic concern; it has direct clinical consequences. The data presented in this issue reinforce the need to validate genetic markers in regional contexts before integrating them into diagnostic algorithms.

In this sense, precision medicine must progressively evolve into a medicine tailored to specific populations within their genetic context.

The Diagnostic Power and Responsibility of Genomics

The clinical case of Costello syndrome, driven by a pathogenic variant in the HRAS gene, exemplifies the transformative role of genomic sequencing in modern diagnosis. When conventional cytogenetics fails to reveal the patient's true diagnosis, as occurred in this case, whole-exome sequencing ceases to be an option and becomes an ethical imperative.

However, this power carries additional responsibilities. The identification of a pathogenic variant is not the endpoint; it marks the beginning of a chain of clinical actions, prognostic considerations, and long-term follow-up strategies. In RASopathies such as Costello syndrome, where oncological risk is inherent, genomic diagnosis requires structured surveillance systems that many healthcare settings are not yet prepared to provide.

The question that then emerges is:

How long will it take for new therapeutic capabilities to match the rapidly expanding diagnostic capacity?

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Oncogenomics and the Fragmented Landscape of Risk
The study on ovarian cancer in Mexican women highlights the significant impact of BRCA1 variants, particularly in the region known as the ovarian cancer cluster region (OCCR). The high prevalence of pathogenic variants in a relatively small cohort is notable, but equally important are the nuances: young patients without family history, variants of uncertain significance, and structural rearrangements.

These findings challenge traditional paradigms of hereditary risk assessment, which rely heavily on family history. They also underscore the importance of integrating molecular data into early detection strategies, especially in malignancies such as high-grade serous carcinoma, where clinical presentation is often late and nonspecific.

In this context, genomics is not only diagnostic, it is predictive, preventive, and profoundly disruptive to traditional clinical workflows.

Structural Variation: An Unfinished Story
Reports on craniofacial microsomia, Cat-eye syndrome, and 22q11.2 deletion remind us that not all genomic variation is captured by sequencing. Structural chromosomal alterations, duplications, deletions, and complex rearrangements, continue to represent a diagnostic frontier, requiring multiple technologies that are not always available within our healthcare systems.

Phenotypic overlap among different syndromes and the reliance on cytogenetic confirmation highlight a critical gap: access. In many regions, the tools necessary to distinguish between clinically similar but genetically distinct conditions are simply not available.

This raises a broader issue of equity in genomic medicine that is gradually being addressed. The implementation of molecular technologies is already established in developed countries, while new advances are increasingly evident in emerging regions such as Latin America. Continued implementation will ensure that precision medicine does not remain a privilege, but rather becomes a standard of care.

Toward a More Coherent Genomic Future
Taken together, the studies in this issue illustrate a field in transition. We are moving from gene discovery to gene interpretation, from isolated findings to integrated systems, and from theoretical precision to practical implementation.

However, several challenges remain:

- **Population specificity vs. global generalization**
- **Diagnostic capacity vs. clinical preparedness**
- **Technological advancement vs. equitable access**
- **Variant detection vs. variant interpretation**

Addressing these tensions will define the next decade of clinical genomics.

Final Reflection
If there is a unifying message in this issue, it is this: our region is advancing in molecular diagnostics, alongside the continuous training of our professionals. Contributions such as those presented in this issue demonstrate our growing capabilities and our commitment to democratizing precision medicine so that it becomes accessible to our population. We remain optimistic that we will also be able to implement new therapies, allowing us not only to diagnose but also to reduce the suffering of our patients.

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