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EDITORIAL

Editorial - New horizons in genomics for Latin America

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Sharing knowledge is necessary for a society to advance. This is the conclusion I have come to after getting to know academic infrastructures in developed and undeveloped countries. In our field, genetics and genomics are new disciplines, both in the field of biology, and even more so in their clinical application. Their high capacity to identify genomic changes and the ability to associate them with diseases has allowed us to expand our knowledge of the delicate balance between health and disease, and the understanding of the function of the 3 trillion base pairs that make up our genome. In the face of such technology, Latin America has not been left behind.

In the last 10 years, public health systems and private hospitals have begun to incorporate molecular diagnostics. This was even more accelerated with the arrival of the epidemic caused by SARS-CoV-2 in 2019. Methodologies such as qPCR, CGH, WES, WGS, and NIPT were implemented to achieve diagnosis of rare diseases affecting our population, and to equip us with the tools that were being implemented in other latitudes. However, the rapid implementation has created new challenges in terms of quality control, human resource training, laboratory certification and validation, sensitivity and reproducibility of molecular tests, new ethical dilemmas, and the delicate balance between cost-benefit for the patient. These aspects are not new to these technologies; however, they are vital for their correct implementation.

Our journal, in both its English and Spanish versions, has as its main objective to create a platform at the service of health professionals in our Spanish-speaking countries to strengthen the knowledge of genetics and clinical genomics in a multidisciplinary way. The vision is that each medical specialty can be represented to address the core and particular aspects of each specialty. We have started this first issue with representation of experts from Spain, Mexico, Dominican Republic, and Panama. Our vision is that each medical specialty can be represented to address the challenges presented in genetics from each discipline.

In this first issue, we present selected articles relevant to multiple medical specialties including Genetics, Pediatrics, Hematology, Maternal Fetal Medicine and Psychiatry. The pediatric articles describe the difficulties in the process of implementing genetic screening in pediatrics in Latin America. They also discuss the different technologies available for prenatal diagnosis, which depends on gestational age, and the type of genetic change suspected (See Figure 1). Two other articles demonstrate the associations between genomic variants with specific diseases such as epilepsy and suicide risk. Finally, we present a brief review of the literature reviewing the current knowledge about hemophilia and the assessment of the quality of life of these patients.

The knowledge shared here will serve to improve our laboratory processes, workflows, interpretation of molecular results, emphasize genetic counseling, and share our technical and clinical expertise for the benefit of our patients and continuous professional improvement. Our collective knowledge and collaboration has been and is the engine of change in our countries, not only for the use and implementation of new diagnostic technologies, but also for the acquisition of judgment criteria for their correct use and application. We welcome you, our reader, and we hope to exceed your expectations with this journal.

Best regards,

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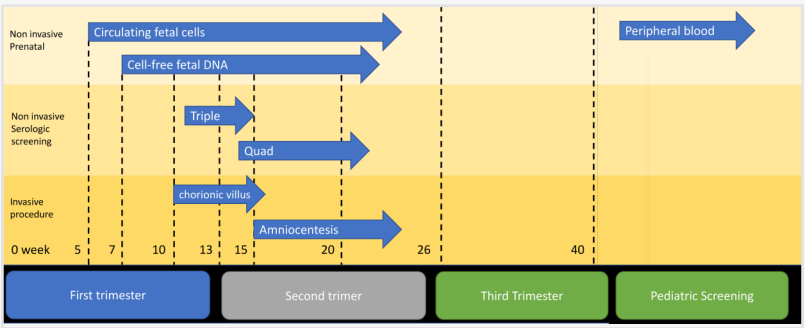

Diagram depicting the different strategies of prenatal and neonatal/pediatric diagnosis over time and the type of samples regularly used for molecular diagnosis. Modified from Keller NA, Rijshinghani A. Clin Case Rep. 2016 Jan 18;4(3):244-6. doi: 10.1002/ccr3.493.