Genet Clin Genom 2025 3(2):33-34

Genetics and Clinical Genomics

Letter to the Editor

Advances in genetic diagnosis in Colombia

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Received: August 29th 2025 / **Accepted**: August 30th 2025 / **Published**: August 31th 2025

Dear Mr. Editor,

In the last 20 years, genetic diagnosis has positioned itself as a tool of utmost importance for prevention, early diagnosis, and personalized treatment of various conditions, both of genetic and multifactorial causes. Colombia is a paradigmatic example of how the global discussion on the subject has had a considerable influence on legislation and public policies, which is why it is imperative to have a critical view of the achievements and difficulties that this entails. One of the most distinctive features of the national scenario is that, by virtue of the Statutory Health Law, genetic tests must be covered by the Health Benefits Plan (PBS), an instrument based on the reach of health as a fundamental right. Therefore, access to all genetic diagnostic tests is guaranteed to any Colombian citizen who requires them. Likewise, Resolution 5267 of 2017, from the Ministry of Health and Social Protection, establishes the health services and technologies covered by the PBS, which is why molecular biology tests and genetic studies are part of them. On the other hand, Law 1392, which regulates the comprehensive care of orphan diseases, is essential to allow patients to access genetic tests without administrative difficulties. The same spirit is reflected in Decree 1954 of 2012, which regulates the surveillance of clinical laboratories involved in molecular tests. This set of regulations takes up the global discussion and positions Colombia as a regional example of equitable access to genomic medicine.

In the country, the development of massive sequencing platforms, which include studies of multigenic panels, clinical exomes, and complete genome analyses, has been possible. Academic institutions such as the University of the Andes, the National University, and high-complexity healthcare centers have strengthened their diagnostic capabilities, allowing them to identify genetic variants in hereditary cancer, cardiopathies, neuromuscular disorders, among others. The implementation of genetic telemedicine has allowed the expansion of services to distant regions, made possible by state-funded projects and the support of medical and scientific associations. The universality of access to genetic tests has direct repercussions on public health. In oncology, for example, the search for mutations in the BRCA1 and BRCA2 genes led to the identification of carriers with increased risk, allowing for the definition of intensified surveillance strategies and targeted therapies. In pediatrics, the early identification of hereditary metabolic diseases has allowed for timely interventions that modify the natural history of the disease. In infectious diseases, Colombia was a pioneer in the incorporation of molecular biology techniques for the diagnosis of the Zika virus and, subsequently, SARS-CoV-2.

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Keywords: genomic medicine, genetic diagnosis, Colombia

Bioethical aspects: The authors declare that institutional ethical standards were followed.

Funding: The authors declare that there was no external funding for this work. No potential conflict of interest were declared.

DOI: 10.37980/im.journal.ggcl.en.20252688

Despite the progress, there are considerable challenges. Firstly, the infrastructure remains unevenly distributed: while most molecular biology and clinical genetics laboratories are located in major cities, rural areas rely on sample referrals, delaying response times. The training of specialized human resources is still insufficient: given the growing demand, there are not enough medical geneticists in the country. On the other hand, the mechanisms for accessing tests are free, but various administrative and bureaucratic barriers often delay authorization. Lastly, the ethical framework and protection of genomic data are still not fully developed. Although Law 1581 of 2012 regulates the protection of personal data, genetic data, due to its sensitive nature, requires specific guidelines that have not yet been developed. The greatest challenge is the consolidation of a national genomic medicine system, within the framework of integrating population databases that allow for the characterization of the genetic diversity of Colombians. This would be crucial for translational research and the development of personalized medicine. In turn, international cooperation, the inclusion of expanded neonatal screening programs, and regulation around direct-to-consumer tests are priority topics in the coming years.

In conclusion, Colombia has managed to take important steps in the field of genetic diagnosis, thanks to the robust legal framework that guarantees universal access to these technologies through the health system. However, problems still persist in terms of infrastructure, human resources, and ethical regulation, and the path to consolidating a true national genomic medicine policy is still long. That said, Colombia has all the possibilities to become the leader in the region if it manages to combine scientific research with equity in access and the protection of patients' rights.

About the Author

Dr. Rivera is a geneticist at the Pediatric Hospital of the Fundación CardioInfantil-LaCardio in Bogotá, Colombia.