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PERSPECTIVES

Towards clinical genomics in Mexico: History and proposals

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Abstract

Clinical genomics is an essential tool for the diagnosis and management of genetic disorders however, its development in Mexico has been limited and uneven. Despite recent progress, such as the establishment of new medical genetics residency programs and specialized clinical units, services remain highly centralized and diagnostic infrastructure is scarce. Most hospitals lack in-house genetic laboratories, forcing reliance on private or foreign services that are often prohibitively expensive, creating significant barriers for patients. Furthermore, there is a shortage of clinical geneticists and available positions in the public health system, contributing to a considerable lag in care. Initiatives such as the National Rare Disease Registry and the efforts of patient advocacy organizations (e.g., FEMEXER, AMAER, ANER) have increased visibility and support for individuals with genetic and rare diseases, but they cannot fully address the wide spectrum of conditions, particularly ultra-rare disorders. To overcome these challenges, five key priorities are proposed: strengthening public genomic diagnostic infrastructure, decentralizing clinical services, expanding training programs and job opportunities for clinical geneticists, consolidating a standardized national rare disease registry, and reinforcing collaboration between institutions and civil society. These actions are fundamental to ensuring the constitutional right to health and advancing equitable access to genomic medicine in Mexico.

In the year 2016, at the beginning of July, I received my certification as a specialist in Medical Genetics from the Mexican Council of Genetics A.C., completing my studies to dedicate myself to the medical specialty I had chosen four years earlier.

Since I was halfway through the medical specialty, I noticed that the training in medical genetics was insufficient for the overwhelming technological development that was occurring since the infinitely cited works of Mendel (van der Waerden, 1968) and the proposal of the DNA structure by Watson and Crick (WATSON & CRICK, 1953), with the well-known failure of the academy to recognize Franklin for her contributions to it. So I decided to continue with two postgraduate degrees but with the aim of learning about genomic technology and its clinical applications.

No geneticist in training can feel fully prepared to interpret the vastness of biological variation, including genetic variants, gene function, inheritance patterns, and other biological characteristics that distinguish such a dynamic knowledge to ask the simple question: Is this disease affecting the patient who comes to my consultation hereditary?

Clinical genomics is a powerful tool to answer it, address the rest of the family (if required), explain its origin, and on rare occasions offer a better treatment.

Since 2004, in its description by Dr. Zenteno and Dr. Kofman of the genetics services in Mexico City (Kofman-Alfaro & Zenteno, 2004) dissecting the recent population change, enumerating the available Medical Genetics services and evidencing the clear need for economic, structural, and academic support of genetics in that decade, there is no major analysis

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or dissemination on the matter, except for the highly recommended analysis by Daiana Bucio and her collaborators on the need for genetic counseling in Mexico (Bucio et. al., 2019), through a structured survey and information gathering on medical genetics services in Mexico, where I highlight their comments on the need to improve this practice, both its clinical application, through professional academic courses, the development of community (non-centralized) genetics services, and the need for increasing job positions for geneticists, similar recommendations were later made by Jara-Ettinger and collaborators in 2021 (Jara-Ettinger et al., 2021).

New genetics services have been implemented in the State of Mexico (Ixtapaluca) and the Teletón network, as well as the opening of medical specialty courses in Puebla and Guadalajara have increased the offer for the diagnosis of genetic diseases in the country. However, most do not have specialized laboratories for the diagnosis of any genetic disease that requires a confirmatory genetic study.

Genomic diagnostic services such as the Rare Diseases Unit of UNAM, and the Genomic Diagnostic Laboratory at the National Institute of Genomic Medicine in the public area, Genos Médica in the private area, and other private laboratories intermediaries of foreign laboratories, which are presented as an alternative to the necessary confirmatory genomic diagnosis in patients. However, each of these laboratories offers studies at a cost to the patient's economy, which represents a significant barrier in many cases. Without misinterpreting this idea, these laboratories are the available and reliable option for the diagnosis of these patients and my sincere recognition for their work, but the high cost represents discrimination by the Mexican health system by not guaranteeing health protection for Mexicans with genetic diseases. This right is based on Article 4 of the current Mexican political constitution.

Other initiatives such as the rare diseases registry led by Dr. Gonzaga (Calvo Aspiros & Gonzaga-Jauregui, Claudia, 2024) which recently presented its advances, have contributed to the discussion and recognition of the application of clinical genomics, but have also sparked controversies related to the misunderstanding of its methodology for registration and the complexities that arise in the scenario where patients register their condition themselves.

Foundations in Mexico for rare diseases constitute a very important piece for the diagnosis and treatment of diseases of genetic origin, FEMEXER (Mexican Federation of Rare Diseases), AMAER (Mexican Association for the Care of Rare Diseases), ANER (National Alliance for Rare Diseases) among others, focus their attention and efforts on a multitude of needs such as economic support, dissemination, training, access to diagnosis, treatment, among various areas, but they cannot cover each and every condition termed as rare as a patient with a diagnosis of an ultra-rare disease recently told me, of which no foundation has knowledge or available support according to herself.

The incredible acceleration of biological knowledge about diseases and their hereditary component, the poor implementation and dispersion of genetics and genomics services in Mexico maintain a considerable lag in clinical care.

Identifying these strengths and weaknesses in Clinical Genomics allows me to list a couple of proposals that I consider timely:

1. Strengthen the public infrastructure of genomic diagnosis
Create and fund public genetic and genomic diagnostic laboratories in different regions of the country, avoiding dependence on private and foreign laboratories. This would ensure universal access and reduce economic discrimination in the care of patients with genetic diseases.

2. Decentralize clinical genomics services
Promote the creation of community genetics and genomics services in regional and state hospitals, with telemedicine programs, so that families do not depend solely on reference centers in the country's capitals.

3. Expand training and positions for clinical geneticists
Increase the number of specialty programs in medical genetics and open more job positions in public hospitals, ensuring that each region has specialists trained in genetic counseling and clinical genomics.

4. Integrate a standardized national registry of rare diseases
Consolidate a unique and validated national registry that centralizes information on patients with rare diseases, with clear methodological protocols, interoperability with hospitals, and ethical supervision, avoiding fragmentation and biases derived from self-registration.

5. Strengthen collaboration with foundations and civil society
Establish a formal collaboration mechanism between health institutions, universities, and foundations (such as FEMEXER, AMAER, ANER), with public and private funding, to cover areas that remain unattended today: diagnosis of ultra-rare diseases, family support, and access to innovative therapies.

All these proposals require regional scrutiny and validation, as well as the effort of all involved participants.

However, the results would justify any necessary sacrifice and effort, because they are results difficult to quantify. They are results that contribute to the human dignity of the fundamental principle of medical practice, of helping others.

He who does not live to serve, does not serve to live (Mother Teresa of Calcutta).

REFERENCES

[1] Calvo Aspiros, C. E., & Gonzaga-Jauregui, Claudia. (2024). First year results and insights from the Mexican Rare Disease Patient Registry,. Rare, Volume 2(100046). https://doi.org/10.1016/j.rare.2024.100046.

[2] Jara-Ettinger, A. C., Cárdenas-Conejo, A., Huicochea-Montie, J. C., & Araujo-Solís, M. A. de J. (2021). [The lag of genetic counseling in Mexico]. Revista medica del Instituto Mexicano del Seguro Social, 59(1), 101–105. https://doi.org/10.24875/RMIMSS.M21000058

[3] Kofman-Alfaro, S., & Zenteno, J. C. (2004). Genetic services in Mexico City. Community Genetics, 7(2–3), 142–145. https://doi.org/10.1159/000080785

[4] van der Waerden, B. L. (1968). Mendel's experiments. Centaurus; International Magazine of the History of Science and Medicine, 12(4), 275–288. https://doi.org/10.1111/j.1600-0498.1968.tb00098.x

[5] WATSON, J. D., & CRICK, F. H. (1953). Molecular structure of nucleic acids; a structure for deoxyribose nucleic acid. Nature, 171(4356), 737–738. https://doi.org/10.1038/171737a0

[6] Bucio D, Ormond KE, Hernandez D, Bustamante CD, Lopez Pineda A. A genetic counseling needs assessment of Mexico. Mol Genet Genomic Med. 2019 May;7(5):e668. doi: 10.1002/mgg3.668. Epub 2019 Apr 1. PMID: 30938092; PMCID: PMC6503023.