



PERSPECTIVES

Advances and Prospects for Genetic Diagnosis in Panama

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Abstract

Genetic diagnosis in the field of medical genetics, which dates back to the pioneering efforts of Dr. John R. Ward, has experienced remarkable progress in Panama in recent years due to increased capacity, the growing number of specialized Panamanian professionals, and the development of all aspects related to this field within institutions such as the Social Security Fund, the Children's Hospital, the Gorgas Institute, and the INDICASAT Institute, among others. There has also been an increase in private offerings and industry support. The prospects for growth go hand in hand with the growing need for medical evaluation by geneticists, genetic diagnosis as part of the daily medical management of all specialties, and the short-term advent of personalized or precision medicine.

When I was proposed this topic, I said to myself, "complex, broad, diverse topic with many actors and, therefore, with a high risk of unsatisfied opinions." So, from now on, before starting, I apologize in advance to anyone who, due to an involuntary omission, I do not mention in this brief text, which is about diagnosis in medical genetics.

However, without fear of being wrong, I believe that most will agree that when we talk about references in clinical genetics diagnosis in Panama, we must all turn our gaze and unavoidably recognize the work of the lone pioneer and considered Father of Genetics

in Panama, Dr. John Robert Ward S., whom we can glimpse, imagining almost poetically, at the beginnings of this process of advancement for Panamanian medical science, both in the Social Security Fund and the Children's Hospital, manually making karyotypes, in a small room obtained reluctantly, where the light of this specialty was being born, which since then, and still for many, is misunderstood. I wish he could see that, in current times, at the National Specialized Center for Medical Genetics and Genomics that bears his name, through the great efforts of Dr. Luis Sotillo Bent and his team in the City of Health, these same karyotypes are achieved with the assistance of robots and a great specialized team, some of whom Dr. Ward himself helped to train.

The analytical capabilities of this center are innovative for our country and the region and cover many other areas to offer the highest quality of a complete genetic diagnosis, including neonatal screening, classic maternal screening, and non-invasive prenatal testing (NIPT), prenatal diagnosis through amniocentesis, metabolic, molecular, and next-generation genomics studies, with a highly trained team of more than 50 professionals, also including techniques such as FISH, CGH (microarray), QF-PCR, gas-mass spectrometry, and sequencing, among others.

We must emphasize that all these techniques are only ordered and are preceded and framed by a thorough and strict genetic evaluation by clinical geneticists, or a relevant request by another specialty

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as the case may be, and their utility is closely linked to genetic counseling and its interpretation in a particular clinical context. It is not about ordering a genetic test just because, but the selection of these and their interpretation must be contextualized and carefully performed, and the progressive awareness that genetic tests are specialized tests that must be administered carefully is also something that has been improving in recent years.

Important advances have also been reported in genetic diagnostic capabilities at the Children's Hospital with the memorable work of Dr. Gladys Cossio, who, along with Dr. Celia Cantón and Dr. Sotillo Bent, was one of the great promoters of the initial version of the current Neonatal Screening Law, of vital importance for the well-being of the population and an important milestone in the history of genetic diagnosis in Panama.

In the last 30 or 40 years, the offer of genetic diagnosis has increased notably, thanks to other actors such as the Gorgas Memorial Institute (ICGES), the INDICASAT Institute, the Institute of Medical Sciences in central provinces, as well as through the contribution of the private sector and industry, thanks to which, considering doing a genetic test is no longer an almost heroic act, as it was a few years ago.

It is important to note that the advances in genetic diagnosis in Panama, apart from the infrastructure capabilities and specialized personnel that must continue to grow to improve national coverage, begin to take shape from medical suspicion, right from that moment when all medical and health team personnel, when facing a patient, the great question arises: "Is what the patient has genetic?" since from the moment of considering the differential diagnosis, the diagnostic doubt arises, whether what this patient is experiencing will have a genetic etiology.

In this way, we are definitely approaching clarifying the condition of this patient, since, with great certainty, from the ultimate diagnosis, their response to therapy, and even the way of expressing a multifactorial condition, will be nuanced by their genetic reality.

With satisfaction, we observe firsthand how, in all these years of medical practice, the demand for genetic evaluation and diagnosis has increased notably, and more and more colleagues, both specialists and primary care providers, regularly consider the diagnosis of genetic entities within their differential diagnoses.

Therefore, the future and perspectives of this type of diagnosis involve improving training and awareness in medical genetics, both for health personnel, mainly doctors, as well as for the general population, especially with the certainty that everything genetic will become increasingly relevant, as we approach giant steps (and we are already in it), to the era of molecular medicine, the massive use of new therapies resulting from genetic engineering, and the era of precision medicine or personalized medicine [1], which involves pharmacogenomics (that is, using individual genetic data to personalize therapies and make them more effective) and other aspects of the idiosyncrasy or unique constitution of the person, which influence the type of manifestations and course of the disease.

It is increasingly easy to perceive, and Panama will not escape it, how fiction becomes reality, making genetic diagnosis an indispensable tool for everyday medical management.

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