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EDITORIAL

Identificación de variantes e interpretación

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The exercise of variant identification by molecular technologies, such as sequencing, is a complex process that requires protocols, quality controls, and technical and clinical knowledge. The complexity of the process does not end with the identification of a genomic variant, but molecular geneticists and clinicians must have sufficient knowledge and experience to interpret the finding and suggest the impact that this variant may or may not have on the patient.Interpretation is not an isolated exercise, but requires repetitive review and systematic review of the available literature on the variant in question. It also requires knowledge of epidemiology, statistics and research methodology to be able to establish a reasonable interpretation with the greatest possible certainty, which is especially important in a clinical setting. Mastering new computer resources such as variant databases (ClinVar, ClinGen, among others) and knowing the population frequency of that variant are additional skills necessary in the exercise of clinical molecular diagnosis.

For this reason, in this issue we are pleased to present three clinical cases focused on molecular diagnostics: a case of Allan-Herndon-Dudley Syndrome, another on familial hypercholesterolemia, and finally, a third clinical case on Pitt-Hopkins Syndrome. This three case reports present different approaches, perspectives, and conclusions, allowing us to see the methodologies already implemented in our region. We also present an article on pain and its genetic basis, putting into perspective the variability of perception of each individual to pain stimuli, which may depend on genetic, epigenetic and environmental factors.

This issue shows our regional efforts to carry out molecular diagnostics and precision medicine, which are already a reality in our countries, and at the same time, we share knowledge and experiences, which will allow us to document the advances in this discipline of Genetics and Clinical Genomics.

Once again, we thank you, our reader, for appreciating this group effort, and we invite you to participate in this academic exercise in our next issues. We hope you will once again enjoy this second issue we have prepared for you.

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