



ARTTYPE

From Diagnostic Complexity to Precision Medicine: Integrating Cytogenetics, Molecular Biology, and Targeted Therapies

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Received: August 31, 2026 / Accepted: August 31, 2026 / Published: August 31, 2026

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The continuing evolution of molecular medicine is transforming not only how diseases are diagnosed, but also how their mechanisms are understood and how patients are treated. Increasingly, complex clinical presentations require an integrated approach in which traditional clinical evaluation, pathology, cytogenetics, molecular diagnostics, computational biology, and targeted therapeutics complement one another. The articles presented in this issue illustrate this transition across a remarkably diverse spectrum of genetic, neoplastic, cardiovascular, and infectious diseases.

The first article, **Chromosomal Triple Mosaicism in a Male with Turner Syndrome, 8p21.1-pter Deletion, and Yq Deletion: A First Case Report**, demonstrates the complexity that can underlie chromosomal disorders and the importance of combining complementary diagnostic methodologies. The identification of three chromosomally abnormal cell lines, including numerical and structural alterations involving the X, Y, and chromosome 8, highlights both the power and the limitations of individual genetic techniques. Importantly, this case reminds us that newer molecular approaches do not necessarily replace conventional cytogenetics. Rather, their greatest diagnostic value may emerge when they are interpreted together, particularly in patients with complex phenotypes that cannot be adequately explained by a single genomic finding.

The second contribution, **Clinical Management and Therapeutic Advances in Pediatric Patients with Neurofibromatosis Type 1 Associated Plexiform Neurofibromas**, moves from molecular diagnosis to molecularly guided treatment. Plexiform neurofibromas remain among the most challenging manifestations of neurofibromatosis type 1, particularly when their infiltrative growth limits the effectiveness of surgical intervention. The experience presented in this pediatric case series illustrates the changing therapeutic landscape created by MEK inhibition. The clinical and radiological responses observed with selumetinib exemplify how understanding disease-associated signaling pathways can be translated into targeted interventions with the potential to improve function, reduce symptoms, and alter the clinical course of previously difficult-to-treat lesions.

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Keywords: editorial, precision medicine, genomics, targeted therapies**Licence and Distribution:** Published by Infomedic International under the Creative Commons Attribution 4.0 International License (CC-BY-NC-ND).**Data access:** Raw data will be provided by the corresponding author upon request.**DOI:** 10.37980/im.journal.ggcl.en.20262779

The third article, **In Silico Structural Characterization of the Novel TTN p.Arg3936Ter Variant in Dilated Cardiomyopathy**, explores another dimension of contemporary precision medicine: the interpretation of genetic variation at the transcript, protein-domain, and structural levels. The study emphasizes an increasingly important principle in genomic medicine—that the biological consequence of a variant cannot always be understood from its genomic position alone. Through integration of transcript annotation, ACMG/AMP interpretation, protein-domain analysis, nonsense-mediated decay prediction, and AlphaFold2-based structural modeling, the authors demonstrate an isoform-specific consequence of a truncating TTN variant. Such approaches illustrate the growing contribution of bioinformatics and structural prediction to the interpretation of clinically relevant genomic findings.

Finally, **Cutaneous Leishmaniasis Mimicking Low-Grade Myofibroblastic Sarcoma: A Case Report from Resistencia, Chaco, Argentina** reminds us that precision diagnostics are equally important beyond inherited disease. The clinical and histopathological resemblance between cutaneous leishmaniasis and a soft tissue neoplasm created a significant diagnostic challenge, ultimately resolved through molecular detection of *Leishmania (Viannia) braziliensis*. This case underscores the importance of maintaining a broad differential diagnosis and integrating epidemiological context with pathology and molecular testing, particularly in regions where infectious diseases may present with unusual or misleading morphological features.

Although these four contributions address very different diseases, they converge on a common message. Diagnostic accuracy increasingly depends on the ability to integrate information generated at multiple levels—from clinical phenotype and histopathology to chromosomes, DNA sequences, transcripts, protein structures, and molecular pathways. At the same time, the expanding availability of targeted therapies demonstrates that increasingly precise diagnosis has consequences that extend well beyond classification: it can directly influence clinical management.

This issue therefore reflects a broader transformation taking place across medicine. Conventional diagnostic methods remain indispensable, but they are increasingly strengthened by molecular assays, genomic analysis, computational modeling, and mechanism-based therapeutics. The challenge ahead is not simply to adopt increasingly sophisticated technologies, but to determine how they can best complement established clinical and laboratory practices and how their benefits can be made accessible to the patients and populations who need them.

Taken together, the studies presented in this issue demonstrate the value of interdisciplinary investigation and reinforce a fundamental principle of modern medicine: complex clinical problems are often best resolved not by a single technique, but by the thoughtful integration of complementary evidence.

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