From Biomarkers to Better Outcomes: Precision Medicine in Modern Internal Medicine

Laurentius Aswin Pramono[1,2]

[1]Department of Public Health and Nutrition, School of Medicine and Health Sciences, Atma Jaya Catholic University of Indonesia, Jakarta, Indonesia
[2]Department of Internal Medicine, Saint Carolus Hospital, Jakarta, Indonesia

Corresponding Author:
Laurentius Aswin Pramono, MD, PhD. Department of Public Health and Nutrition, School of Medicine and Health Sciences, Atma Jaya Catholic University of Indonesia, Jakarta, Indonesia; Department of Internal Medicine, Saint Carolus Hospital, Jakarta, Indonesia. Email: aswin.pramono@atmajaya.co.id, l.aswin.pramono@gmail.com.

Internal medicine has addressed illness from a population viewpoint for the majority of its existence. The diagnosis algorithm, first-line drugs, including the starting dose, and a guideline threshold built for the "average" patient have long defined how internists diagnose and treat. Although this strategy has been beneficial in saving lives, it has also made it possible for a treatment that has shown promise in a large clinical trial to still fail. Precision medicine can overcome this problem. Instead of asking what works for most patients with a given diagnosis, it asks what will work for this patient, based on their own molecular, genetic, and clinical profile.

Precision medicine, which was previously thought to be primarily an oncology concept, is now highly relevant to general internal medicine due to the growing biomarker toolset, multi-omics profiling, and artificial intelligence (AI) required to interpret the biological data.[1,2] These days, we describe the benefits of precision medicine for patient outcomes. It moves from the biomarkers and omics platforms that generate a molecular signal, through real examples of some diseases, to the AI and pharmacogenomic tools needed to act on that signal, and concludes by looking at the gap between what precision medicine can offer today and what is still needed for it to become part of routine clinical practice.

BUILDING THE FOUNDATION: TRANSLATING PRECISION MEDICINE INTO CLINICAL PRACTICE

Advances in biomarker-based diagnostics have improved precision medicine. Genomic, transcriptomic, proteomic, and metabolomic technologies, together as well as single-cell and spatial omics data, allow clinicians to characterize disease at a molecular level.[1] The molecular, cellular, and blood-based biomarkers have become usual clinical tools used to diagnose diseases, determine prognosis, and monitor treatment in almost all organ systems.[3] However, a biomarker test acquires clinical utility only when it alters clinical management, for instance, by starting therapy earlier, avoiding a drug unlikely to help, or escalating surveillance in a higher-risk patient. Furthermore, single biomarkers are being combined with multi-marker panels that can assess the biological heterogeneity of disease more accurately.[4]

This principle is illustrated by two of the most prevalent conditions in internal medicine practice, diabetes mellitus (DM) and chronic kidney disease (CKD). In type 2 DM, the glycated haemoglobin biomarker has been supplemented by other emerging biomarkers, including adiponectin, adropin, netrin-1, and lipoprotein(a), which can highlight metabolic imbalance before glucose measures rise and may help distinguish which patients will respond to a particular glucose-lowering agent.[4]

Similarly, in CKD, assessment based primarily on estimated glomerular filtration rate and albuminuria is being supported by other biomarkers such as kidney injury molecule-1, growth differentiation factor-15, and soluble urokinase-type plasminogen activator receptor, which enable earlier detection and strengthen the risk stratification in CKD.[5] These developments highlight that innovation in medicine is not limited to discovering new drugs. It also lies in making better decisions about when treatment should begin, which patients are most likely to benefit, and when treatment may not be necessary or appropriate.

ARTIFICIAL INTELLIGENCE IN PRECISION MEDICINE

None of this diagnostic workup can be totally translated into clinical decisions without computational support. The complexity of multi-omics and biomarker data surpasses what any clinician can integrate themselves. This is precisely where artificial intelligence (AI) has become an essential part of precision medicine, helping transform complex biological data into clinically meaningful insights.

In August 2024, the U.S. Food and Drug Administration approved nearly 950 medical devices with AI and machine learning capabilities, and almost two-thirds of clinicians surveyed by the American Medical Association said they used AI in clinical practice for several tasks, such as assessing chest radiographs and interpreting electrocardiograms.[6] AI can bring large amounts of biomarker and molecular data, helping identify patterns that may suggest a patient is unlikely to respond to standard therapy and allowing clinicians to reconsider treatment before failure becomes clinically apparent.

PHARMACOGENOMICS AND CHALLENGES IN PRECISION MEDICINE

Precision medicine is more than just using biomarkers to diagnose conditions and determine disease risk. In addition, it seeks to identify the appropriate drug and the right dose for the right patient, which is the main goal of pharmacogenomics, the study of how a person's genetic influences their response to medications. The same prescription may be beneficial for one patient but harmful for another due to genetic variations in drug-metabolizing enzymes.

These differences are often influenced by rare variants and population-specific genetic patterns that remain underrepresented in reference genomic databases. In internal medicine, where polypharmacy is prevalent among patients managing several chronic illnesses concurrently, the problem is made much more difficult.[7] International initiatives, including the Clinical Pharmacogenetics Implementation Consortium and the Dutch Pharmacogenomics Working Group, have begun to translate this genetic variability into practical, actionable prescribing guidance. In the future, pharmacogenomics may help improve therapeutic outcomes while reducing adverse drug events. Despite its advantages, pharmacogenomics' wider application is still constrained by a number of issues that must be resolved.[7]

The advancement of precision medicine still faces several challenges, particularly in its implementation. Multi-omics testing remains expensive, biomarker assays are not consistently standardized across laboratories, and few practicing internists receive well-structured training in interpreting genomic or multi-marker reports.[1,7] If these problems remain unsolved, precision medicine risks becoming a benefit available only to the patients who have access to specialized academic centres rather than an integrated part of everyday clinical care. This raises an important issue of equity, which deserves as much attention as the science itself. As a result, the health system will need to spend more on interpretable AI tools, accessible biomarkers and multi-omics technology for clinical laboratories, and training programs for genomic and multi-omics literacy in general internal medicine. Only then will "from biomarkers to better outcomes" become a reality rather than just an idea in internal medical practice.

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