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AI-augmented Precision Healthcare

Pushing the Realm of Stratified Medicine

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Why you should care

Healthcare delivery is shifting from a one-size-fits-all approach to a more targeted and stratified model. Complex chronic diseases, rising healthcare costs, and poor healthcare outcomes drive this change. Precision Medicine (PM) approaches, which can reduce costs and improve therapeutic efficiencies, lead this transformation. PM benefits span the care continuum, from disease screening and risk assessment to diagnosis, treatment, and dose selection. Currently, PM tools primarily benefit cancer and infectious disease management, where over- and undertreatment can cause drug resistance, serious side effects, and treatment failure. Other focus areas include cardiovascular and neurological diseases.

Overburdened healthcare systems and a lack of skilled lab technicians necessitate streamlined, accurate precision diagnostics. High prices for advanced therapeutics like cell and gene therapies drive the need for novel biomarker-driven strategies in therapy development and selection. Despite progress, clinical PM translation remains sporadic, and improved adoption hinges on developing more reliable tools.

AI is emerging as a disruptive force in precision medicine, leveraging exponentially growing data from multiple sources into clinically actionable insights. While advances in imaging, molecular analyses, and pathology form PM's core, integrating AI tools into molecular profiling and identifying novel biomarkers can unlock PM's potential and build a robust framework. AI adoption in PM strategies has accelerated notably – a recent analysis shows scientific literature on AI in PM has nearly doubled in the last four years (992 publications in 2023, up from 567 in 2020).¹

What you need to know

Omics technologies boom has generated an exponentially growing amount of multidimensional data (genomics, proteomics, transcriptomics, microbiome, metabolomics, lipidomics, etc.). Advances in high-throughput biological analysis approaches add data layers that require holistic and accurate analysis. AI tools sieve through large amounts of data, accelerate processes, reduce Turnaround Time (TAT), and identify unknown linkages and “cryptic” patterns in biological processes that human eyes might miss to identify.²

As interest in the precision health industry intensifies, there is a notable surge in funding for AI platforms and a concomitant increase in the number of companies, product launches, and R&D efforts.

¹ <https://doi.org/10.1016/j.imu.2024.101475>

² <https://intlbn.com/2023/04/10/shift-towards-precision-medicine-to-improve-healthcare-outcomes/>

However, limited clinical validation of AI-powered diagnostics and reimbursement challenges have deterred widespread adoption.

- **Diagnostics and screening.** AI adoption in diagnostics is the highest, with an influx in radiology, imaging, and pathology followed by molecular diagnostics. Using AI to analyze genomic tests, microbiome consumer tests, and wearables data has gained momentum, evolving for accurate screening, prediction, detection, and disease monitoring. For instance, Cardio Diagnostics Holdings, Inc.³ is developing an AI-augmented multiomic test to detect Congenital Heart Disease (CHD), while Preventio⁴ is developing a Coronary Artery Disease (CAD) diagnosis test. Molecular tests use multidimensional biomarkers to screen for hereditary cancers, cardiovascular diseases, and genetic conditions. Zio Health's iRhythm exemplifies cardiac wearables that detect arrhythmias like atrial fibrillation
- **Clinical decision support.** AI-powered clinical tests support screening, risk assessment, patient cohort selection, therapy response monitoring, and prognosis and disease recurrence assessment. AI enhances radio genomics and molecular diagnostics analysis, improving disease understanding, prognosis, treatment optimization, and surveillance. Companies like Myriad Genetics, Fabric Genomics, and Nalagenetics have developed precision clinical genomics tests for cancer screening and risk assessment
- **Drug discovery and development.** AI in drug discovery and development enables targeted therapeutics development, precision clinical trials, and improved understanding of disease pathogenesis. AI-powered tools enhance the data intensive clinical trial workflows for patient recruitment, real-world data analyses, and precision trial design. Tempus AI, Exscientia, and InSilico Medicine lead AI-powered precision drug discovery with novel biomarker discovery pipelines and AI-driven clinical trial platforms
- **Health insurance.** Payers can use AI platforms to design personalized services based on individual clinical and biological profiles. These services can predict individual risks and plan coverage accordingly

Challenges

Key challenges in adopting AI for PM include:

Security and privacy. AI models require large-scale data sharing, storage, and collaboration to become more robust. Current data storage and privacy guidelines hinder ecosystem connectivity. Federated learning (used by Owkin and Zephyr AI) and blockchain platforms could improve privacy-protected federation of real-world data.

Accuracy and reliability. Data complexity and size overwhelm many AI algorithms, leading to reliability and bias challenges. Continuous training must improve model reliability, reduce bias, and enhance predictive capabilities for clinical practice. Robust clinical validation is required to ensure platform accuracy

³ <https://doi.org/10.1016/j.imu.2024.101475>

⁴ <https://cdio.ai/>

and reliability. Improving data models with Real-world Evidence (RWE) or synthetic data enhances their productive capabilities.

Costs and mindset. Adopting AI into clinical workflows requires a major mindset shift and significant infrastructure investments. Despite potential cost benefits from improved efficiencies, consumers and physicians remain skeptical about using AI in routine practice.

Regulatory challenges. AI tools and solutions evolve constantly, requiring dynamic regulations to match developments. This proves difficult to achieve. In addition, many AI models operate as “black boxes,” and regulatory bodies often scrutinize solutions that lack transparency, especially for high-risk healthcare applications.

Examples in motion

AI-augmented PM tools are developing exponentially, often outpacing our ability to keep up. Current developments primarily focus on cancer screening, diagnosis, and patient stratification. It will be interesting to see how innovations emerge for different disease indications and applications.

Several global players are investing in solutions around:

- Precision oncology / Early detection and disease management.** Diagnostics developers and academia are developing more precise liquid biopsies for early detection and disease management, enabling tumor recurrence prediction and prognosis. Companies like Veracyte (C2iGenomics), Sophia Genetics, and Syantra use AI platforms to provide novel insights and pattern recognition. Beyond oncology, infectious disease diagnosis, surveillance, and cardiovascular disease management show promise for AI applications. AI-powered molecular diagnostics can predict and diagnose myopathies and ventricular dysfunction, and even predict myocardial infarction
- Novel biomarker discovery and targeted therapy development.** AI-augmented workflows draw linkages between previously unknown biological patterns and accurately model diseases. In Silico Medicine, Recursion Bio and others actively use AI in drug discovery workflows to uncover novel biomarkers driving targeted drug development
- Triaging.** AI-based tools facilitate rapid, precise clinical decision-making in emergency and patient triaging. Precision clinical tests can handle large patient groups during potential pandemic outbreaks. AI can predict the severity and length of hospital stays and determine optimal drug combinations
- Preventive healthcare and screening.** There are several developments in wearables, AI-driven clinical genomics, and AI-based retinal imaging. Technology giants like Google are exploring AI's potential in retinal screening for Diabetic Retinopathy (DR) and other chronic diseases. Meanwhile, innovative companies like Eyenuk, Inc., AEYE Health, Aireen, and Optomed

lead in leveraging retinal testing for chronic disease and DR screening with FDA-approved products.

Where is the pulse on AI-augmented precision healthcare heading?

AI technologies' evolution is vital for accurate screening, risk assessment, early diagnosis, patient stratification, and prognosis. While developments have been rapid, clinical workflow adoption lags. A rigorous framework to make AI tools more reliable, scalable, platform-agnostic, and predictive will accelerate adoption. Several stakeholders are focusing on improving AI tools' precision and predictability. Generative AI in drug discovery workflows and clinical diagnostics will grow steadily, though developments remain nascent. Companies like Qure.ai, Niramai Health, Owkin, and InSilico Medicine are advancing generative AI to create synthetic data for de novo drug design, novel drug target discovery, precision clinical trials, and drug response prediction, potentially revolutionizing the field.

Precision oncology, with many diagnostic companies developing AI-based analysis for pathology, radiology, and liquid biopsies to analyze tumors, select therapies, and predict prognosis. This trend expands to other diseases, particularly infectious and cardiovascular disease prediction and management, aligning with their growing burden.

Increased awareness and training can help AI tools provide clinically actionable insights to clinicians for appropriate therapeutic interventions and dose management. Overburdened healthcare systems can benefit from AI tools for patient referrals, triaging, and remote patient monitoring.

Several countries, including the US, UK, Japan, and China, have launched large-scale PM efforts. Adopting widespread digitalization and supporting technology-enabled companies via incubators and accelerators will foster innovation. Despite such technologies' promise and potential applications, investors should approach this crowded space cautiously, understanding the platform's nuances before investing.

Final word

AI adoption in PM is not a passing trend. As nations promote widespread PM, demand for AI tools will rise. Harmonizing AI integration into clinical diagnostics

(omics, imaging, pathology) with clinical trial data and RWE will support healthcare systems in designing strategies and population-level initiatives.

Integrating AI into PM will remain a sustainable trend for chronic disease diagnostics, precision public health strategies, screening and preventive tests, theranostics, companion diagnostics, and therapy selection. Biopharma and diagnostics companies should partner with multiple stakeholders, including patients, hospitals, and consumers, to improve the scalability and clinical utility of these solutions. Rapid innovation in AI platforms will enable PM to achieve tangible, more predictive, personalized, and preventive healthcare results.

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