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How AI and Genomics are merging to transform precision medicine?

22 May 2025 | Views | By Dr Ravi Gupta, Vice President – Bioinformatics, MedGenome

The convergence of AI and Genomics is not only making medicine more efficient but also more compassionate

Precision medicine has enabled us to deliver customized healthcare by leveraging the interplay between molecules, environment, and lifestyle across populations. Today, with techniques such as next generation sequencing, we can analyze genomic data in a much faster and cost-effective manner. Add to this the integration of multiomics technologies, including transcriptomics, proteomics, epigenomics, metabolomics, and microbiomics, which has further improved our understanding of complex biological interactions and their application in providing better health outcomes. All this has been made possible with advancements in data science and artificial intelligence.

From Risk to Readiness: How AI is Enabling Disease Prediction

Traditionally, disease prediction relied on a patient's family history, lifestyle risks, and the general population patterns. While these are useful metrics, they discount the unique individual biological makeup and its impact on the individual. Genomics offers this precision, and AI makes it actionable.

Genomics decodes a person’s genetic code to identify specific genetic variants that may increase their risk for certain diseases, allowing for a far more accurate and individualized understanding of health conditions. For example, knowing that both parents are carriers of a rare condition and understanding the available treatment options helps couples make informed decisions while planning a pregnancy.

However, genomic data is vast and complex. This is where AI steps in. From analyzing genetic data to interpreting health records, today, AI algorithms can identify data patterns, reducing manual intervention, improving accuracy, allowing for faster intervention, and improving patient outcomes.

AI tools help make the assessment of hundreds of thousands of genetic variants, necessary to determine an individual's predisposition to certain conditions, faster and more precise. For instance, Genessense Polygenic Risk Scores (PRS) tool uses AI & ML algorithms to analyze and predict an individual's genetic risk for a variety of diseases, including cancers, cardiovascular diseases, diabetes, neurological disorders, autoimmune conditions, and other genetic diseases, offering

valuable insights for early intervention and personalized treatment strategies.

Additionally, AI algorithms also enhance the analysis process in non-invasive prenatal testing (NIPT), improving the detection of chromosomal abnormalities like Trisomy 21 (Down syndrome), assisting in early risk assessment. In newborns, screening powered by AI can improve the process of early detection of genetic conditions like congenital hypothyroidism or phenylketonuria (PKU), which require timely intervention. AI can enable readiness over reaction, often before a condition manifests clinically.

AI-Powered Diagnostics: From Symptoms to Sequencing

Diagnosis often starts with symptoms, routine diagnostics, imaging, or histopathology, and moves towards requiring molecular and genome-level analysis in complex or rare conditions. While genomic data equips clinicians with actionable insights, making sense of this vast data quickly and accurately is what is driven by AI tools today.

A single human genome contains over 3 billion base pairs. Sequencing data can highlight millions of variants, among which also lie the disease-causing variants. AI tools help to:

- Differentiate between benign and pathogenic variants
- Predict the likelihood of disease manifestation
- Drastically reduce turnaround times for genetic reports

AI's diagnostic power is especially transformative in rare diseases, where diagnosis often takes a very long time and delays the treatment process. A case in point is MedGenome’s VarMiner, an AI-driven gene variant mining tool.

As the company expanded, growing business meant increasing sample volumes. This led to not just higher analysis costs but also a rise in manual errors. Addressing this challenge, MedGenome developed a proprietary AI algorithm that enabled its variant mining software to prioritize rare and clinically significant variants from genomic datasets.

This AI-enabled software, called VarMiner, significantly improved accuracy and reduced the time required for manual analysis, particularly for rare genetic diseases. The timely adoption of AI helped MedGenome reduce test reporting times, accelerating clinical interventions. In some cases, the duration, which previously took hours, has also been cut to under 30 minutes.

VarMiner also helps in understanding inheritance patterns and designing clinical trials tailored to specific genetic profiles. Consequently, it not only makes operations more efficient by enabling clinicians to save time consumed in analysis but also improves the accuracy and speed of the diagnosis process, helping doctors quickly identify the genetic cause of a rare disease and choose the most effective treatment options for their patients.

Incorporating AI into its process, MedGenome has improved its throughput, enhanced reporting accuracy, and reduced operational costs. Across India, such real-world examples continue to demonstrate how AI and genomics are changing the diagnostic journey, especially in pediatric and neonatal settings, even in low-resource environments.

Precision Management: Personalizing Care with AI and Genomics

Diagnosis is just the starting point. The combined power of AI and genomics extends into the ongoing management of disease, enabling care that is highly individualized and responsive over time.

In oncology, for instance, tumour profiling through targeted gene panels or whole-exome sequencing allows oncologists to understand the specific mutations driving cancer. AI helps interpret these mutations to guide the use of targeted therapies and immunotherapies, greatly improving treatment precision.

Liquid biopsy, an emerging technology that analyses circulating tumour DNA (ctDNA), provides a non-invasive method for detecting residual disease or early recurrence. With AI capabilities layered in, clinicians can:

- Monitor treatment response in real time
- Predict relapse with higher accuracy
- Select or adjust treatment strategies based on current tumour biology

Personalised oncology care, powered by genomics and AI, is now becoming standard in research institutions and many tertiary care hospitals across India. Beyond cancer, AI is also making chronic disease management smarter. Genetic data, combined with AI, helps predict how a patient might respond to a particular drug, guiding therapy choices and minimizing trial-and-error prescribing. It can enable clinicians to make more timely, informed decisions and adjust disease management plans dynamically based on real-time inputs.

The Path Forward: A Smarter, More Humane Healthcare Ecosystem

The intersection of AI and genomics is redefining healthcare. By making possible predictions, quicker diagnosis, and highly personalised care, this convergence is not only making medicine more efficient but also more compassionate.

To achieve its full potential, healthcare systems will have to:

- Embed genomic technologies and AI into routine clinical workflows
- Develop digital infrastructure to facilitate data processing and sharing
- Enlarge clinician education and public health literacy
- Invest in population genomics programmes to serve diverse populations

Precision medicine has the potential to improve the health and productivity of the population while building patient trust in the healthcare system. This shift from reactive to a more proactive healthcare can also provide significant health cost-benefits both at an individual and population level. Integration of AI with Genomics in precision medicine has proven that it has the power to transform diagnosis as well as prognosis, resulting in better disease management in the coming years.

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