

Applying AI-assisted genetic risk analysis to precision medicine can transform healthcare



Source: KAUST Photolibrary, Comparative Genomics and Genetics Lab 2026.

Note: Precision medicine uses artificial intelligence to integrate and analyze patient biomedical information for developing personalized care.

Unlocking the potential of AI tools in precision medicine

Precision medicine is an innovative approach that uses biomedical data, including genetic information, to tailor healthcare strategies to individual patient needs. Recent advances in artificial intelligence have greatly increased the potential for leveraging data on genetic risk factors (GRFs) in personalized medical care to improve disease prediction, diagnosis, and treatment. AI tools can decode and interpret large amounts of complex genetic information, thereby deepening understanding of individuals' genetic risks and the roles that GRFs can play in customization, but these tools have not yet been well integrated into healthcare, and GRFs are not yet reaching their potential.

To fill that gap, a KAUST-led team of researchers investigated the potential of implementing AI workflows to improve the use of GRFs to support more personalized care and better health outcomes. This policy brief presents their findings on the potential for using AI to better integrate GRFs into personalized patient care, including a proposed roadmap, and it highlights promising applications of AI augmentation at each stage of healthcare.

The bottom line

- A KAUST²-led research team identifies opportunities for integrating AI-driven analysis of individuals' GRFs into precision medicine and improving health outcomes.
- Opportunities exist along the entire workflow of precision medicine, starting with early screening and extending to diagnosis, treatment, and personalized health management.
- The team proposes a roadmap for AI and GRF integration with pointers to specific opportunities for AI augmentation.
- Real-world experience demonstrates the value of incorporating AI and GRFs in patient healthcare.



AI-augmented genetic risk analysis can support every stage of precision medicine

Figure 1 depicts AI augmentation in a workflow that divides precision medicine into four distinct stages, each of which can be informed by biomedical data and genetic risk analysis. AI tools based on foundation models, such as large language models, can use GRF data from genetic databases and other biomedical sources to support early screening, precision diagnosis, precise clinical treatment, and AI-augmented personalized health management. The approach integrates data from rich and varied sources in disparate forms, such as medical records, imaging, and novel biomarkers, to enable early detection of disease risks, accurate disease classification, tailored treatment design, and ongoing health management.

AI connects data and knowledge to a wide range of medical applications

AI uses different types of biomedical data and learning to assess GRFs and create tailored patient care.

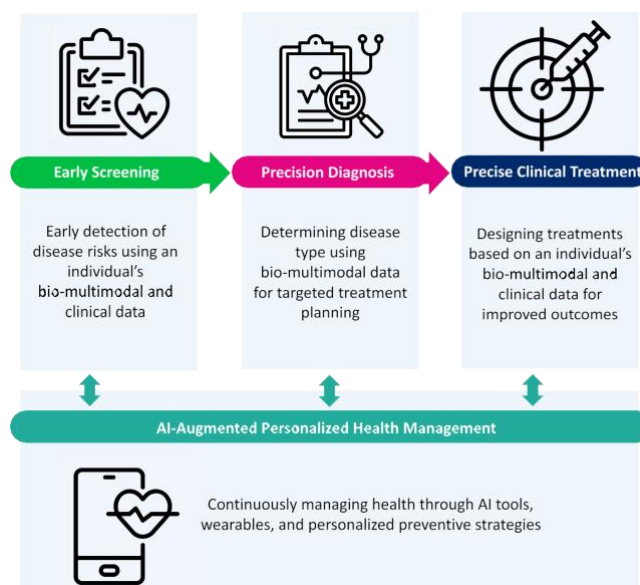
AI can play a central part in data integration and knowledge representation, which convert complex data into usable, structured frameworks for biomedical research. Potential uses of AI in this domain range from literature searches and data visualizations to risk ranking. For example, AI-supported data mining and retrieval techniques can locate information from disparate sources and platforms and in repositories such as PubMed and Med-Plus.

The main computational tasks for AI involve using biomedical data for GRFs analysis and prediction. These tasks include statistically identifying continuous relationships between input features, such as genetic characteristics, and outputs, such as disease progression; evaluating associations in structured datasets; graphically representing relationships as structured networks; clustering to recognize inherent patterns in data; and classification into predefined categories, such as pathogenetic or benign genetic variants.³ The tasks differ by methods, some of which AI can leverage to better effect. For example, AI can employ machine learning and statistical methods for deeper insight into the functionality and pathogenicity of genetic variants, which can shed light on genetic regulation and on how disease itself unfolds.

Biomedical data resources underpin all the proposed tasks and applications. High-quality datasets can provide fertile training grounds for AI, foster collaboration among researchers, improve models, and accelerate progress. However, sharing sensitive data poses privacy risks. Federated, or decentralized, learning can provide a layer of

protection without blocking research. It synthesizes knowledge reports across sources, originating in public and private domains. The information is combined in a way that helps to safeguard data and individual privacy.

Figure 1. Precision medicine workflow using data that has been integrated and analyzed by AI



Source: Alsaedi et al. 2025.⁴

Note: This figure illustrates the framework of precision medicine, categorized into four stages: early screening, precision diagnosis, precise clinical treatment, and AI-augmented personalized health management.

AI-augmentation roadmap can advance integration

The study proposes a roadmap for AI-augmented genetic risk analysis and precision medicine applications. The roadmap emphasizes robustness and accuracy, as well as predictive strength and interpretative quality, along three workflows that, together, would provide deeper insight into disease and its treatment for customization, as shown in Figure 1. The roadmap provides specific recommendations for AI tools at each stage of the workflows and anticipates computational resource needs from lowest to highest.

Figure 2 offers a much-simplified representation of the roadmap, indicating where the tools would play their parts without citing them. The workflow involves various types of annotation, which involves identifying and describing the structure and function of genetic material.

Experience demonstrates the value of integration

Integrating AI-enhanced GRFs into clinical practice has tangible benefits evident in practical applications and real-world experience.



Integrative analysis of biometric data. At its core, AI-assisted precision medicine combines different types of health data, including genomic profiles, electronic health records of past diagnoses and treatments, and real-time data from wearable sensors. AI analyzes these large datasets to detect patterns, improving diagnostics and creating tailored treatment. By connecting these different types of data, AI helps pinpoint the exact cause of illnesses and matches treatments to a patient's genetic profile to improve their success rates.

More precise disease prevention and detection. GRFs help identify people at high risk for diseases before symptoms appear. People with higher genetic risk can receive more frequent, intensive checkups to help detect illnesses early and use medical resources efficiently. For example, in brain diseases such as Alzheimer's, checking genetic markers allows for earlier identification.

More accurate diagnoses and better patient outcomes. AI combined with GRFs improves medical care by enabling faster, more accurate diagnoses and better patient results. For example, in cancer research, AI identifies genetic markers that respond to customized treatments, helping doctors predict disease behavior and choose the best therapy.

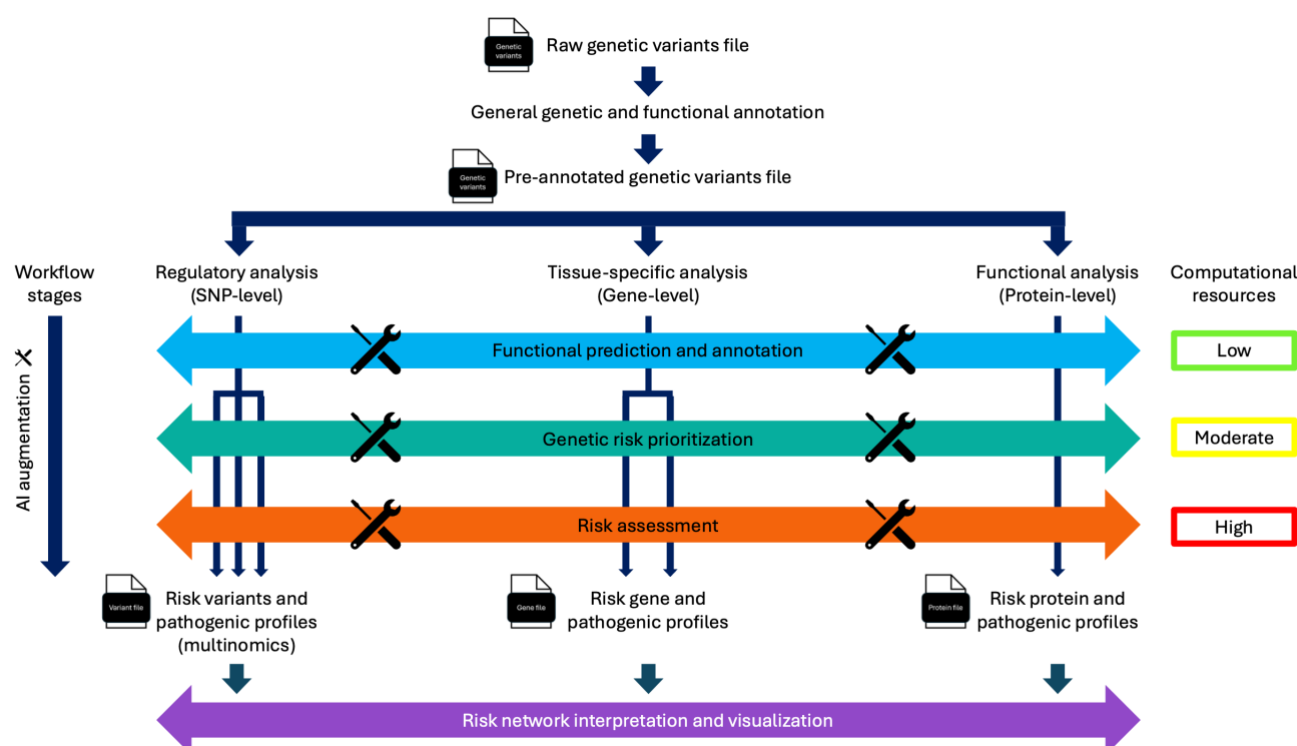
Targeted treatments and drug effects. AI tools have significantly improved how various diseases are treated. For instance, one tool helps identify compounds that interact with drug targets for metabolic disorders and screens for high blood pressure treatments.

Understanding disease mechanisms and molecular-level risk networks. Combining AI with genetic risk data can improve understanding of how diseases work at a molecular level. AI-enhanced tools are crucial for studying disease causes and complex biological networks. Advances enabled by these tools also shape research goals and shed light on how diseases develop.

Summing up

Precision medicine is using individual patient's genetic information to customize their treatment and improve their health. AI can support customization at every stage of precision medicine by analyzing massive, diverse biomedical datasets to identify GRFs and enable applications that meet individuals' healthcare needs. Recent real-world experience demonstrates the benefits.

Figure 2. Roadmap and workflow for implementing GRF analysis



Source: Alsaedi et al. 2025, figure 5, with substantial simplification.

Notes: ✂ represents multiple AI tools suggested for each stage of each workflow; 📁 represents inputs and output to and from the workflow stages; SNP = single nucleotide polymorphism, which is the genetic building block.



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King Abdullah University of
Science and Technology

About the KAUST Researchers



Sakhaa Alsaedi

Ph.D. Graduate, Computer Science
King Abdullah University of Science and Technology



Takashi Gojobori

Distinguished Professor, Bioscience

[View full profile on kaust.edu.sa](https://www.kaust.edu.sa/en/about/policy-institute)

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King Abdullah University of Science and Technology
Building 16 Level 3, Room 3612
Thuwal 23955-6900, Saudi Arabia

1. This policy brief summarizes findings from Alsaedi, Sakhaa, Michihiro Ogasawara, Mohammed Alarawi, Xin Gao, and Takashi Gojobori. 2025. "AI-Powered Precision Medicine Utilizing Genetic Risk Factor Optimization to Revolutionize Healthcare." *NAR Genomics and Bioinformatics* (7). <https://doi.org/10.1093/nargab/lqaf038>. All rights and any third-party permissions remain with the respective rightsholders.
2. King Abdullah University of Science and Technology (KAUST).
3. A genetic variant is a difference in a person's DNA sequence that may or may not, if benign, affect their health or risk of developing a disease.
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