

AI and Genetics

How Computers Help Read, Interpret, and Use DNA

Artificial intelligence (AI) is transforming the way scientists and healthcare providers make sense of the vast amount of information in our DNA. By recognizing patterns that may be difficult for humans to spot, AI holds promise to accelerate discoveries about what causes disease, make genetic tests more precise, and support care tailored to everyone. These tools are still evolving, and many AI applications have not yet been as thoroughly tested or widely adopted as those in other fields, making careful evaluation and responsible use essential. Looking ahead, AI holds exciting potential to help researchers, clinicians, and patients move toward faster answers, more informed decisions, and better health outcomes.

AI IN RESEARCH VS. EVERYDAY LIFE

The AI used in research and healthcare is not the same as the AI many people use in everyday life. AI tools that people use in everyday life are designed to be broadly helpful—answering questions, writing text, or generating images—and they learn from very large, general collections of information. In contrast, clinical and research AI is built for specific purposes, such as helping to interpret genetic test results or finding patterns in laboratory and health data. These systems are typically not general-purpose “chatbots,” but specialized tools that perform focused analytical tasks behind the scenes. They are typically tested more rigorously, used within secure systems, and guided by clear rules to protect privacy and reduce errors. These tools are in their early days and still need to be perfected.

KEY TAKEAWAYS

- ✓ **Not all AI is the same:** The AI used by researchers and clinicians is very different from the AI used in everyday life. It is designed for highly specific scientific tasks, rigorously tested, and used within secure systems to protect patient privacy and ensure reliable, accurate results.
- ✓ **AI looks for patterns humans might miss:** By combining data such as symptoms, test results, and physical traits, AI can identify patterns associated with genetic conditions, sometimes suggesting possible diagnoses.
- ✓ **AI can make repeatable tasks more efficient—not automatic:** AI tools can help organize reports, summarize complex findings, and handle routine communication tasks, improving efficiency in busy clinical settings.
- ✓ **AI findings require verification:** AI used to help interpret data and streamline tasks in healthcare tends to be more trustworthy than AI available to the general public. However, it can still sometimes generate false, misleading, or entirely made-up information. Because of this, human oversight on AI is very important.
- ✓ **There are important guardrails and considerations:** Genomic data is deeply personal, so protecting privacy is essential. Strong safeguards, shared standards, and ongoing oversight are necessary to ensure AI is used safely, fairly, and responsibly.

HOW AI IS USED IN GENETICS AND GENOMICS

Predicting Disease-Causing Variants. AI tools can help predict which DNA changes are more likely to be disease-causing (often called “pathogenic” variants). Some tools estimate how genetic variants alter protein structures, while other tools look for DNA changes that may interfere with “splicing,” the cell’s process of editing RNA instructions before a protein is made. These AI-based predictions can help clinicians and researchers quickly prioritize variants that deserve closer review or follow-up testing—alongside family history, symptoms, laboratory standards, and expert judgment. These are predictions and should always be interpreted alongside other evidence, such as clinical and family history.

Phenotype Evaluations. Some AI tools are designed to help clinicians integrate many pieces of information—such as medical history, physical features, and test results—to identify meaningful patterns. In certain cases, and only with appropriate consent and privacy protections, these systems can also analyze facial features as one part of the overall clinical picture. By comparing a person’s features and health information with patterns seen across many known genetic conditions, AI tools can suggest possibilities for clinicians to consider that may involve recognizable facial traits, along with other medical findings. Clinicians carefully review the individual’s full medical history and physical exam and may order genetic testing to confirm or rule out a condition.

Clinical Care. In clinical settings, AI can support care by reducing the time healthcare providers spend on routine documentation and high-volume communication. For example, AI can help draft and organize genetic reports into clear, structured summaries—such as variant lists, links to supporting evidence, how well findings match a patient’s features, and a ranked set of candidate explanations—so laboratory professionals and clinicians can focus on

interpreting and confirming results. Human experts still review, edit, and approve every final report. AI-powered tools can also help answer common questions or prepare patients for visits, especially when clinics are busy.

Treatment and Discovery. AI is helping speed up the discovery of new treatments by giving researchers a clearer picture of how the body’s building blocks work. Genes provide instructions for making proteins, which help cells do their jobs. When a genetic change affects a protein, it can lead to disease. AI tools can estimate what a protein looks like in 3D, which can offer clues about whether a genetic change might cause problems—and where a future medicine might help. However, sometimes AI can hallucinate, or generate false, misleading, or entirely made-up information that it claims is true. This is why careful researcher oversight is still needed to interpret and verify these results.

KEY CONSIDERATIONS FOR RESPONSIBLE USE OF AI

As AI becomes a powerful tool in genetics research, using it responsibly is essential to ensure it benefits both science and society. Protecting privacy is especially important because genomic data is deeply personal. Our understanding of the genome—particularly for rare conditions—is still growing, and the data used to develop AI tools is not yet complete or standardized. Researchers are also working to establish shared standards, such as common data labels and reporting guidelines, so that results can be compared more easily and trusted across studies and institutions. With strong safeguards, ongoing research, and careful oversight, AI has the potential to advance genetic discovery while respecting privacy, promoting fairness, and strengthening trust.