

Artificial intelligence (AI) has become important in interpreting data from genetic tests, from sorting through large amounts of data to identifying sequencing errors. While AI's role in genetic testing is expected to increase in the coming years, involvement of humans and having the "human touch" remains essential. From double-checking AI's findings to explaining results to patients, providers like clinical geneticists and genetic counselors will always have the last word in assessing genetic data.

**Question 1:** What genetic tests are typically used in a clinical setting, and how does AI make the tests more effective?

### Karyotyping

**What It Is:** A test that uses cells to create an image of chromosomes, which are the structures that contain the genes in our cells. It is used to detect chromosomal abnormalities that can lead to genetic conditions and disease. The test is inexpensive and used throughout the world. Learn more about [karyotypes](#)<sup>1</sup>.

**Role of AI:** AI allows many chromosomal images to be analyzed quickly and accurately and can spot chromosomal changes that are otherwise hard to see with the naked eye.

### Chromosomal Microarray

**What It Is:** A technology that can detect chromosomal abnormalities too small to be seen by a regular microscope, including [copy number variations](#)<sup>3</sup> such as [small deletions](#)<sup>4</sup> and [duplications](#)<sup>2</sup>. It has been particularly effective in detecting the genetic cause of neurological and neurodevelopmental disorders, including epilepsy, autism, and intellectual disabilities.

**Role of AI:** With AI's assistance, subtle changes can be rapidly detected, accelerating the pace of analysis, diagnosis, and the development of customized treatments.

### Targeted Panels

**What It Is:** A type of next-generation sequencing that analyzes for mutations in a set of genes that lead to different categories of diseases and disorders. It is fast, accessible, and offers high resolution. Targeted panels for cancer have been particularly effective in matching patients to potential personalized treatments, a transformative change in cancer care.

**Role of AI:** AI plays a key role in identifying the function of mutations and their potential relationship to disease.



## Whole Exome Sequencing (WES)

**What It Is:** The analysis of only the protein-coding areas of the genome where many of the disease-causing variants are found. It can more efficiently identify both common and rare variants and lead to earlier diagnosis of genetic disorders.

**Role of AI:** AI sheds light on the interpretation of the variants that are uncovered to determine which ones could impact gene function and are more suspicious of being disease-causing. This provides a more complete picture about how they can impact health.

## Whole Genome Sequencing (WGS)

**What It Is:** The analysis of the entire DNA sequence of a person's genome, generating vast amounts of data.

**Role of AI:** AI is crucial for managing and interpreting this data efficiently, identifying and helping to classify both common and rare variants, that then facilitates earlier diagnoses of genetic disorders.

## Question 2: What is the promise of AI? What are its limitations?

AI has the potential to improve speed, accuracy, and efficiency of data analysis for most genetic tests. But AI is only as good as the data on which it is trained. Right now, very little data is available for underrepresented populations (such as those from Africa, Asia, or Latin America) and in the developing world, so any medical and genetic guidance that is provided comes from information that is overall less complete. Additionally, AI isn't experienced with predicting environmental risks, which contribute to disease progression. Finally, AI's potential for WES and WGS is not yet fully known.

## Question 3: What's next for AI?

AI is here to stay—and will continue to evolve and improve. Human experts, however, remain essential for reviewing AI's interpretations and, most importantly, for communicating results to patients with empathy and context.

As knowledge about AI and genetics continues to advance, it's essential to combine technology with ethical, inclusive, and patient-centered approaches. Furthermore, AI's known limitations need to be addressed to make the technology as trustworthy as possible.

**Want to dive deeper?** Explore the history of AI: [Artificial Intelligence, Explained | Carnegie Mellon University's Heinz College](https://www.heinz.cmu.edu/media/2023/July/artificial-intelligence-explained)<sup>5</sup>.

### References

1. Karyotypes: <https://www.genome.gov/genetics-glossary/Karyotype>
2. Copy Number Variations: <https://www.genome.gov/genetics-glossary/Copy-Number-Variation-CNV>
3. Small Deletions: <https://www.cancer.gov/publications/dictionaries/cancer-terms/def/gene-deletion>
4. Duplications: <https://www.genome.gov/genetics-glossary/Duplication>
5. Artificial Intelligence, Explained | Carnegie Mellon University's Heinz College: <https://www.heinz.cmu.edu/media/2023/July/artificial-intelligence-explained>