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Artificial intelligence in prenatal genetic: Current advances and future directions

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Abstract

To review the main advances in the application of artificial intelligence (AI) to prenatal genetic data analysis, as well as its clinical and ethical implications. Methods: A narrative review of recent scientific literature was conducted in international databases, including studies on machine learning and deep learning applied to prenatal diagnosis. Results: AI has shown to improve diagnostic accuracy in detecting chromosomal abnormalities, optimize the interpretation of non-invasive genomic tests, and support personalized medicine during pregnancy. Nevertheless, limitations persist regarding clinical validation, algorithmic bias, and bioethical regulation. Conclusions: AI represents a promising tool in prenatal genetics, but its implementation requires a strong regulatory framework, data protection, and interdisciplinary training.

Keywords: Artificial intelligence, Prenatal genetics, Machine learning, Prenatal diagnosis, Personalized medicine

Introduction

For a variety of uses, especially in the medical field, artificial intelligence (AI) has become a more practical and dependable instrument. Prenatal diagnosis constitutes one of the most sensitive and significant areas of modern medicine, as it allows for the early detection of chromosomal, genetic, and structural alterations in the fetus [1,2]. Advances in next-generation sequencing (NGS) and other molecular techniques have exponentially increased the quantity and complexity of the data obtained, posing a challenge for its analysis and clinical interpretation [3,4]. In this context, artificial intelligence (AI) is emerging as a fundamental tool for optimizing diagnostic processes, reducing human error, and providing a more precise and personalized approach to large volumes of genetic information [5]. AI, supported by machine learning and deep learning prediction algorithms, has shown significant progress in identifying hidden patterns in large volumes of biomedical data. ML makes decisions exclusively through information obtained from data rather than direct user input [6,7]. In the field of prenatal genetics, these technologies can be applied to the analysis of genomic sequences, microRNAs, copy number variants (CNVs), as well as the detection of aneuploidies and monogenic diseases.

In addition to its diagnostic value, AI has potential in obstetric risk prediction and preventive medicine, facilitating data-driven clinical decision-making. However, its implementation presents challenges related to clinical validity, interpretation of results, protection of sensitive data, and the ethical dilemmas associated with prenatal genetic information [8,9]. Figure 1 shows a concentric circle graphic that gradually dives into more specialized aspects of the discipline while describing the hierarchical relationships within AI. [10,11].
Therefore, it is essential to review the current status of the application of AI in the analysis of prenatal genetic data, identifying its advances, limitations, and future development prospects, as well as discussing its clinical, ethical implications, and future perspectives [12,13, 34, 35]. As illustrated in Figure 2.

The importance of contextualizing this overall review lies in understanding the transformative potential of AI in prenatal genetics, while recognizing its limitations and ethical implications. Our approach

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goes beyond merely technical developments to include a thorough analysis of how AI affects medical outcomes, prenatal genetic diagnosis, and societal ramifications [14].

Methodology

Artificial intelligence (AI) has demonstrated a growing role in prenatal genetics, being applied in different phases of the diagnostic and prognostic process.

First, in next-generation sequencing (NGS), machine learning algorithms allow for more precise identification of copy number variants (CNVs) and single nucleotide polymorphisms (SNPs), which are highly relevant for prenatal diagnosis.

Likewise, in noninvasive prenatal testing (NIPT), based on the analysis of circulating cell-free fetal DNA, AI has optimized the detection of common aneuploidies such as Down syndrome, Edwards syndrome, and Patau syndrome, increasing sensitivity and specificity compared to traditional methods.

Another significant contribution lies in genetic risk prediction, where advanced AI models integrate genomic and clinical data to estimate the probability of developing monogenic and multifactorial diseases.

Finally, in the field of personalized medicine, AI contributes to the identification of prenatal biomarkers that can guide individualized preventive and therapeutic strategies during pregnancy [15,16].

Study Selection Process (Simplified PRISMA)

The PRISMA methodology was applied for this systematic review. The study selection process was as follows:

- Records identified in databases: 50 articles.
- Records after eliminating duplicates: 40 articles.
- Records reviewed by title and abstract: 40 articles.
- Records excluded due to lack of relevance: 10 articles.
- Full-text articles evaluated: 30 (corresponding to the included references).
- Articles excluded after full evaluation: 0 articles.
- Studies included in the final synthesis: 30 articles.

This selection flow is summarized in Figure 2 (simplified PRISMA diagram), which shows the process of filtering and selecting the scientific literature used in this review.

Advantages over traditional methods

The application of AI in prenatal genetic offers substantial benefits over conventional methodologies, including:

- Greater capacity for processing and analyzing large volumes of data [17].
- Reduction of human error in the interpretation of genetic results [18].
- Increased sensitivity and specificity of prenatal testing [19,20].
- Possibility of integrating clinical, genomic, and environmental information into more robust predictive models[21].

Limitations and challenges

However, despite its advantages, the use of AI in prenatal genetic presents important limitations that must be considered:

- Clinical validation of several algorithms in diverse populations is still insufficient [22].
- Algorithmic biases that limit its universal applicability [23, 36].
- Ethical and legal challenges related to data privacy and informed consent [24,25].
- The need for professionals trained in bioinformatics and medical genetics to properly interpret the results [26, 37].

Discussion

The potential to improve by encouraging more structure and efficiency in doctors' practices, which enhances patient care and results.AI makes care more accessible, which enhances patient satisfaction and follow-up. . The integration of AI into the analysis of prenatal genetic data constitutes a step toward precision medicine in pregnancy. Compared with conventional methods, AI demonstrates significant advantages in terms of efficiency, diagnostic

accuracy, and reduction of false positives. However, its implementation in clinical practice must be done with caution, considering the need for regulation, transparency in algorithms, and robust clinical validation [27,28,29].

The first AI-related query was raised in the 1950s by the "Project on Duty." Could an artificial machine function and make decisions that are identical to those of humans? Alan Turing asked this seemingly straightforward question [30] Recent studies show that machine learning algorithms can outperform conventional screening and diagnostic methods in accuracy, reducing the rate of false positives and false negatives in tests such as noninvasive screening (NIPT). This increased reliability can reduce the need for invasive procedures, such as amniocentesis, reducing risks for both mother and fetus. According to scientists, these investigations show that artificial intelligence (AI) is feasible and, with careful planning, might equal or even surpass human intelligence [31][32].

The future points toward the development of hybrid systems that combine medical expertise with the predictive capacity of AI, ensuring more accurate and ethically responsible diagnoses [33]

The integration of artificial intelligence into prenatal genetic represents a paradigm shift in the way healthcare professionals interpret and use genomic information. However, despite its potential benefits, the clinical implementation of AI still faces significant barriers. One of the main limitations lies in the variability and heterogeneity of genomic data, which requires the creation of more robust and representative databases. Furthermore, algorithms often function as "black boxes," hampering clinical interpretation and limiting healthcare professionals' confidence in AI-based decision-making.

Another crucial aspect is the ethical and legal debate surrounding the use of AI in prenatal genetics. The information derived from analyses can have implications not only for the fetus, but also for the family and future generations. This poses challenges regarding confidentiality, informed consent, equity in access to the technology, and the risk of genetic discrimination.

Looking ahead, it will be necessary to establish clear regulatory frameworks that guarantee quality, safety, and equity in the application of AI in prenatal medicine. Likewise, collaboration between bioinformaticians, geneticists, obstetricians, and ethicists are needed to maximize the benefits and minimize the associated risks.

Conclusions

Artificial intelligence is emerging as a transformative tool in the field of prenatal genetics, with the potential to revolutionize early diagnosis and personalized medicine during pregnancy. However, its impact will depend on its responsible integration into clinical practice, the strengthening of bioethics, and the ongoing training of healthcare professionals.

Although AI is emerging as a revolutionary tool in prenatal genetics, its success will depend on a balanced integration of technological innovation, clinical validation, and ethical responsibility.

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APPENDIX

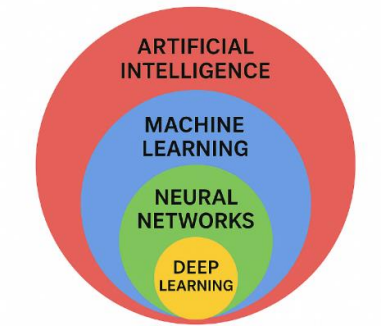

Figure 1. The image shows a diagram of concentric circles representing the hierarchical relationship between different fields of artificial intelligence: Red circle (largest): Artificial Intelligence → this is the broadest field, encompassing all techniques and methods for building machines that can carry out operations that normally call for human intelligence. Machine learning is a subset of artificial intelligence that focuses on techniques that let machines learn from data and enhance their performance without explicit programming. This is the blue circle (inside AI). Green circle (machine learning): Neural networks are a specific method that draws inspiration from how the human brain functions. . Yellow circle (innermost): Deep Learning → is a type of neural networks with multiple layers, which allows for more complex learning and the processing of large volumes of data, such as images, voice, or text.(Hirani R, 2024).

Fig. 2 Pregnant woman in profile, holding her belly. A digital, luminous representation of the fetus is observed. Surrounding the figure are multiple technological and medical icons (such as DNA, locks, graphs, connections, and hexagons), symbolizing the application of artificial intelligence, biotechnology, and data analysis in the field of pregnancy and prenatal genetics.

Figure 3 Simplified PRISMA diagram in Spanish, used to show the flow of study selection in a systematic review.

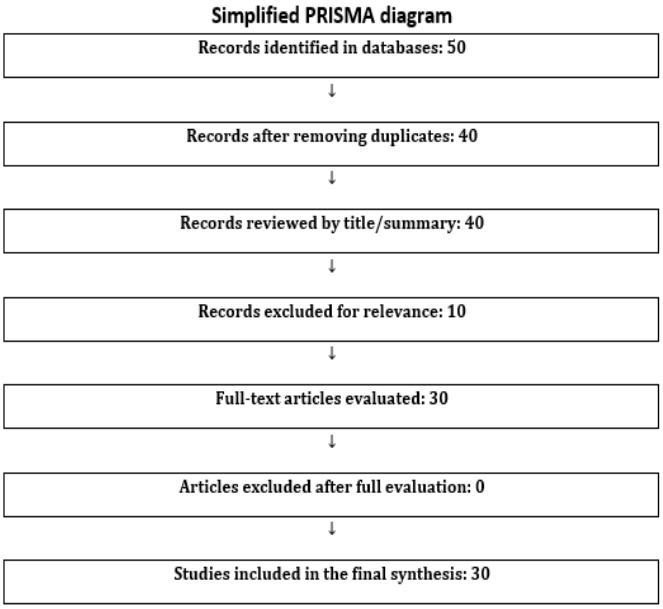

Figure 3. Simplified PRISMA diagram summarizing the study selection process: 50 articles identified, 40 screened after removing duplicates, 30 included in the final synthesis.