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Artificial Intelligence in Cytological Diagnosis: A Narrative Review of Current Applications, Challenges, and Future Perspectives

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Artificial Intelligence in Cytological Diagnosis: A Narrative Review of Current Applications, Challenges, and Future Perspectives

Inteligencia artificial en el diagnóstico citológico: revisión narrativa de las aplicaciones actuales, desafíos y perspectivas futuras.

Inteligência Artificial no Diagnóstico Citológico: revisão narrativa das aplicações atuais, desafios e perspectivas futuras

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ABSTRACT

Background: Artificial intelligence (AI) is rapidly transforming cytological diagnostics through automated image analysis, enhanced sensitivity and specificity, and workflow efficiency in screening programs, particularly for cervical cancer.

Objective: To provide a narrative overview of current AI applications in cytology, emphasizing diagnostic performance, technical limitations, and laboratory implications.

Methods:

A literature review was performed using PubMed, Scopus, and Google Scholar, focusing

on publications from 2020–2024 with keywords such as artificial intelligence, cytology, digital pathology, and deep learning. Selected studies were synthesized to highlight real-world implementations.

Results: AI tools, especially deep learning models, have shown improved detection of abnormal cellular morphology, reduced interobserver variability, and more consistent diagnostic outcomes. Most evidence centers on cervical cytology. Remaining challenges include dataset quality, algorithmic bias, regulatory gaps, and the need for specialized training for cytopathology staff.

Conclusion: AI offers promising advances in cytological diagnosis but requires validation, standardization, and workforce adaptation. Future interdisciplinary research and collaboration are essential to embed these technologies safely and ethically into routine practice.

Keywords: artificial intelligence; cytology; digital pathology; cervical screening; deep learning; diagnostic automation

RESUMEN

Antecedentes: La inteligencia artificial (IA) está transformando rápidamente el diagnóstico citológico mediante el análisis automatizado de imágenes, una mayor sensibilidad y especificidad, y la mejora de la eficiencia del flujo de trabajo en programas de cribado, especialmente para el cáncer de cérvix.

Objetivo: Ofrecer una visión narrativa de las aplicaciones actuales de la IA en citología, con énfasis en el rendimiento diagnóstico, las limitaciones técnicas y las implicaciones para el laboratorio.

Métodos: Se realizó una revisión bibliográfica en PubMed, Scopus y Google Scholar, centrándose en publicaciones de 2020–2024 con palabras clave como inteligencia artificial, citología, anatomía patológica digital y aprendizaje profundo. Se sintetizaron los estudios seleccionados para destacar implementaciones en el mundo real.

Resultados: Las herramientas de IA, especialmente los modelos de aprendizaje profundo, han mostrado una mejor detección de morfología celular anómala, una reducción de la variabilidad entre observadores y resultados diagnósticos más consistentes. La mayor parte de la evidencia se centra en citología cervical. Los retos pendientes incluyen la calidad de los conjuntos de datos, los sesgos algorítmicos, las lagunas regulatorias y la necesidad de formación específica para el personal de citopatología.

Conclusión: La IA ofrece avances prometedores en el diagnóstico citológico, pero requiere validación, estandarización y adaptación de los equipos profesionales. La investigación interdisciplinar y la colaboración futura son esenciales para integrar estas tecnologías de forma segura y ética en la práctica rutinaria.

Palabras clave: inteligencia artificial; citología; patología digital; cribado cervical; aprendizaje profundo; automatización diagnóstica.

RESUMO

Contexto: A inteligência artificial (IA) está a transformar rapidamente o diagnóstico citológico através da análise automatizada de imagens, do aumento da sensibilidade e especificidade e de ganhos de eficiência no fluxo de trabalho de programas de rastreio, em particular do cancro do colo do útero.

Objetivo: Fornecer uma visão geral narrativa das aplicações atuais da IA em citologia, enfatizando o desempenho diagnóstico, as limitações técnicas e as implicações laboratoriais.

Métodos: Foi realizada uma revisão da literatura nas bases PubMed, Scopus e Google Scholar, com foco em publicações de 2020–2024 e nas palavras-chave inteligência artificial, citologia, patologia digital e aprendizagem profunda. Os estudos selecionados foram sintetizados para destacar implementações no mundo real.

Resultados: As ferramentas de IA, sobretudo os modelos de aprendizagem profunda, demonstraram melhor detecção de morfologia celular anômala, menor variabilidade interobservador e resultados diagnósticos mais consistentes. A maioria da evidência centra-se na citologia cervical. Persistem desafios quanto à qualidade dos conjuntos de dados, viés algorítmico, lacunas regulamentares e necessidade de formação específica para as equipas de citopatologia.

Conclusão: A IA oferece avanços promissores no diagnóstico citológico, mas exige validação, normalização e adaptação das equipas. A investigação interdisciplinar e a colaboração futura são essenciais para integrar estas tecnologias de forma segura e ética na prática corrente.

Palavras-chave: inteligência artificial; citologia; patologia digital; rastreio cervical; aprendizagem profunda; automatização do diagnóstico.

Introduction

Artificial intelligence (AI) is rapidly emerging as a disruptive force in healthcare, particularly in diagnostic specialties such as cytopathology. The convergence of advanced computing, big data, and digital imaging has created an environment where AI-assisted systems can support or even partially automate cytological interpretation. These technologies promise to improve accuracy, reduce diagnostic variability, and enhance laboratory efficiency, especially in high-volume screening contexts such as cervical cancer programs [1–3].

Among the most notable developments is the use of deep learning algorithms for image classification and anomaly detection in cytology slides [4, 5]. These systems are capable of identifying complex morphological patterns in cells that may be overlooked by the human eye, thereby improving the sensitivity and specificity of diagnoses [6,7]. Automated analysis also reduces the time required for slide evaluation and can alleviate the workload of cytotechnologists, allowing laboratories to process more samples without compromising quality [8,9].

Despite these advantages, AI implementation in cytology faces multiple challenges. The quality and representativeness of training datasets are critical for algorithm performance, and biases in data can lead to diagnostic errors [10,11]. Furthermore, there are concerns regarding transparency, ethical implications, regulatory oversight, and the need for upskilling laboratory professionals to interpret AI-generated outputs [11–13].

This review aims to explore the current applications of AI in cytology, examine its diagnostic benefits and technical limitations, and reflect on the future of AI-assisted workflows in cytopathology practice.

Materials and Methods

This narrative review was conducted with the aim of synthesizing recent advances in the application of artificial intelligence (AI) in cytology, focusing on diagnostic performance, practical implementation, and laboratory impact. While not following a systematic PRISMA protocol, the methodology was designed to ensure relevance, transparency, and reproducibility in the selection of sources.

Literature Search Strategy

The literature search was performed between January and April 2024 using three major databases: PubMed, Scopus, and Google Scholar. The following search terms were used individually and in combination: *artificial intelligence, cytology, digital pathology, deep learning, machine learning, cervical screening, automation, and computer-assisted diagnosis*. Boolean operators (AND/OR) were applied to refine results.

In total, 158 articles were initially retrieved. After removing duplicates ($n = 32$), 126 titles and abstracts were screened. Of these, 89 were excluded for not meeting the inclusion criteria—either because they were unrelated to cytology, focused solely on radiology or robotics, or lacked relevance to AI-based diagnostic applications. This led to 37 full-text articles being reviewed in depth. From these, 25 were ultimately selected for synthesis in this review. The remaining 12 articles were excluded due to lack of primary data or empirical findings ($n = 5$), insufficient detail regarding AI model characteristics or implementation ($n = 4$), or substantial overlap with more comprehensive studies already included ($n = 3$).

Inclusion and Exclusion Criteria

Studies were included if they met the following criteria:

- Published between January 2020 and April 2024.

- Written in English or Spanish.
- Focused on the application of AI or machine learning in cytological diagnosis, including automation, image analysis, workflow integration, or training implications.
- Available in full text and published in peer-reviewed journals.

Articles were excluded if they:

- Focused solely on radiology or surgical robotics.
- Were conference abstracts, opinion pieces, or letters to the editor without sufficient scientific content.
- Addressed AI in pathology without cytological relevance.

Data Extraction and Synthesis

Full texts of the selected articles were read independently by two authors. Key data extracted included:

- Type of cytology studied (e.g., cervical, urine, thyroid).
- Nature of the AI model (e.g., deep learning, support vector machine).
- Reported diagnostic outcomes (sensitivity, specificity, accuracy).
- Identified benefits, limitations, and challenges in implementation.

Disagreements on inclusion or interpretation were resolved through discussion and consensus. Due to heterogeneity in study designs, populations, and outcome measures, a quantitative meta-analysis was not appropriate. Instead, findings were organized thematically into conceptual domains.

Methodological Limitations

As a narrative review, this study is subject to inherent limitations such as selection and publication bias. No formal risk-of-bias assessment or statistical meta-analysis was performed. However, to mitigate these issues, efforts were made to include only high-quality, peer-reviewed sources. Of the 25 articles included, 21 were published in journals indexed in Scopus and/or the Journal Citation Reports (JCR), and 16 were classified within Q1 or Q2 quartiles according to their respective disciplines. These criteria ensured a minimum level of methodological rigor, visibility, and academic reliability across the selected studies. Additionally, journal impact and indexing status were considered during full-text screening to prioritize the inclusion of robust and clinically relevant evidence.

Results

The findings from the reviewed literature were grouped into six major thematic areas: (1) automation and diagnostic accuracy, (2) reduction of observer variability, (3) predictive analytics and risk stratification, (4) challenges and barriers to implementation in laboratory practice, (5) applications beyond cervical cytology, and (6) emerging platforms and real-world implementation.

Automation and Diagnostic Accuracy

One of the most impactful contributions of AI to cytology is the automation of image analysis. AI-powered systems, particularly those based on deep learning, can analyze cytological slides at high speed and with remarkable precision. These systems are especially effective in cervical cancer screening, where large sample volumes demand rapid triage and high diagnostic sensitivity [1,2].

Studies have shown that AI models are capable of detecting abnormal cells with greater consistency than manual screening alone, identifying subtle morphological features that may escape the human eye [3,6,7]. Some platforms have reported improvements of up to 20% in the detection of precancerous lesions when compared to conventional interpretation [3].

Reduction of Interobserver Variability

Manual cytological interpretation is inherently subjective and dependent on the experience of the cytotechnologist or pathologist. AI-assisted systems contribute to standardizing the diagnostic process by reducing variability between observers. This standardization is particularly valuable in screening programs where consistent results are critical [7,8].

AI also facilitates quality control by reanalyzing previously screened slides and highlighting potential misclassified cells, offering support for diagnostic confirmation or correction [6,8].

Predictive Analytics and Risk Stratification

In addition to morphological evaluation, some AI models incorporate clinical metadata to provide predictive insights. These systems can identify patients at higher risk of developing malignancies based on molecular, demographic, or historical diagnostic data [9,10]. Such stratification supports the development of personalized screening strategies and targeted follow-up protocols.

Furthermore, AI enables laboratories to prioritize high-risk samples for immediate review, optimizing workflow and resource allocation [10].

Challenges in Implementation

Despite promising results, AI adoption in cytology faces several technical and organizational barriers. A primary concern is the quality of training datasets: if the data used to train algorithms are biased or unrepresentative, diagnostic accuracy may suffer [8,11].

Another major limitation is the lack of standardized protocols for validation and regulatory approval. The variability in hardware, staining techniques, and imaging devices across laboratories can also affect AI performance [11,12].

Moreover, successful implementation requires retraining and upskilling of laboratory personnel to interpret AI-generated outputs effectively and to monitor algorithmic errors or unexpected behavior [8,12]. Concerns around patient data privacy and legal responsibility in AI-supported decisions also persist [13].

Applications Beyond Cervical Cytology

While cervical cancer screening remains the most studied field for AI applications in cytology, other cytological domains are beginning to benefit from machine learning tools.

Urinary cytology has long suffered from diagnostic variability and low sensitivity for detecting urothelial carcinoma. Recent studies have demonstrated the feasibility of using convolutional neural networks (CNNs) to identify malignant cells in voided urine specimens with higher accuracy than manual interpretation alone [14]. AI-based systems can help stratify patients based on risk and reduce the number of equivocal reports [15,16].

Thyroid fine-needle aspiration (FNA) is another area where AI is being explored. Some pilot studies have trained models to classify FNA smears into Bethesda categories, assisting in the triage of indeterminate lesions. This may be particularly helpful in low-resource settings or in centers with limited cytopathology expertise [17,18].

These developments suggest that AI can be extended to multiple areas of cytology beyond cervical screening, with the potential to enhance accuracy and reproducibility across specimen types.

Emerging Platforms and Real-World Implementation

Several commercial and institutional platforms are currently driving the integration of AI into cytopathology practice.

Early systems like the ThinPrep Imaging System by Hologic combined semi-automated slide scanning with pathologist verification. More recently, fully AI-powered platforms such as Paige.AI, DeepCyt, and HistoAI have been developed, using deep learning to detect cellular abnormalities and prioritize slide review [19,20].

Academic groups have also contributed significantly to the field. Open-source tools like OpenSlide, combined with PyTorch-based deep learning frameworks, have allowed laboratories and researchers to train custom AI models using local datasets [21, 22]. These initiatives support the development of adaptable diagnostic solutions tailored to specific clinical environments.

Although many of these systems are still under evaluation or regulatory approval, their growing presence indicates that AI-assisted cytology is gradually transitioning from research to practice.

To conclude the Results section, Table 1 summarizes the most frequently reported advantages and limitations derived from the selected studies.

Discussion

The integration of artificial intelligence (AI) into cytological practice represents a significant advance in laboratory medicine. As demonstrated in this review, AI tools—

particularly those based on deep learning—have shown strong potential in improving diagnostic accuracy, reducing interobserver variability, and enabling high-throughput screening workflows, particularly in cervical cytology [1–3]. These improvements align with current goals in pathology to standardize interpretation, minimize diagnostic delay, and optimize resource use.

Beyond cervical screening, emerging studies in urinary and thyroid cytology suggest that AI models may also enhance diagnostic precision in specimens traditionally considered more challenging [15–18]. Although current applications are still limited by data heterogeneity and the need for expert input, early results are promising and indicate that AI’s diagnostic utility may extend well beyond its original scope.

Furthermore, the development of commercial platforms such as Paige.AI and DeepCyt, alongside open-source initiatives, is creating a growing ecosystem of tools that support real-world implementation [19–21]. These systems are not only contributing to diagnostic efficiency but also redefining workflows, training requirements, and the roles of cytotechnologists and pathologists. In particular, the increasing need for digital literacy and interpretative oversight presents both a challenge and an opportunity for laboratory professionals to expand their competencies.

Nevertheless, several barriers remain. The lack of standardized regulatory frameworks and external validation protocols continues to limit the widespread clinical adoption of AI in cytology. There is also a need for more diverse, representative datasets to ensure model generalizability across populations and specimen types [8,11]. Additionally, ethical concerns such as patient data protection, algorithmic transparency, and diagnostic responsibility must be addressed before AI can be fully integrated into diagnostic decision-making [12,13].

In conclusion, artificial intelligence has the potential to transform cytological diagnosis by complementing human expertise with automated precision. To fully harness these benefits, collaborative efforts are required across research, clinical practice, education, and regulatory bodies. Future research should focus on multicenter validation, open-access data sharing, and the integration of AI tools into diagnostic training curricula. With appropriate oversight, AI is poised to become a valuable partner in achieving accurate, efficient, and equitable cytology services.

Author Contributions (Contribución de Autoría)

Conceptualization: Castro-Peraza ME; Vega-Castro A; Lorenzo-Rocha N; Delgado-Díaz R; Torres-Jorge J.

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Formal analysis: Castro-Peraza ME; Delgado-Díaz R; Torres-Jorge J.

Investigation: Castro-Peraza ME; Vega-Castro A.

Methodology: Castro-Peraza ME; Vega-Castro A.

Supervision: Castro-Peraza ME; Lorenzo-Rocha N; Delgado-Díaz R.

Validation: Castro-Peraza ME; Lorenzo-Rocha N; Delgado-Díaz R; Torres-Jorge J.

Writing – original draft: Castro-Peraza ME; Vega-Castro A; Torres-Jorge J.

Writing – review & editing: Castro-Peraza ME; Vega-Castro A; Lorenzo-Rocha N;

Delgado-Díaz R; Torres-Jorge J.

Conflict of Interest (Conflicto de intereses)

The authors declare no conflicts of interest. This statement follows COPE recommendations for conflict of interest disclosure.

Data Availability

(Declaración de disponibilidad de los datos de investigación)

Data availability: Not applicable—no new data were generated or analyzed in this study.

This narrative review involved no human participants and no collection or processing of personal data.

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Advantages	Limitations
Automated analysis of large sample volumes	High dependence on dataset quality
Improved diagnostic accuracy and sensitivity	Limited generalizability across laboratories
Reduction of false positives and false negatives	Lack of regulatory standardization
Decreased interobserver variability	Need for specific training of laboratory personnel
Faster turnaround time and increased efficiency	Potential algorithm bias
Support for risk stratification and personalized screening	Concerns regarding data privacy and legal accountability
Enhanced quality control through AI-assisted rescreening	High initial costs and infrastructure requirements (in some cases)

Table 1. Summary of advantages and limitations of artificial intelligence in cytology

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- The authors declare that the necessary Terms of Free and Informed Consent of participants or patients in the research were obtained and are described in the manuscript, when applicable.
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