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The Use of Artificial Intelligence in the Diagnosis of Eye Diseases - a Review

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ABSTRACT

Introduction:

This review article consolidates current knowledge on the application of artificial intelligence (AI) in the diagnostic and therapeutic processes of ocular diseases, focusing on glaucoma, diabetic retinopathy (DR), cataract, and age-related macular degeneration (AMD). It also discusses the limitations of AI algorithms and highlights potential areas for future clinical research.

Materials and methods:

A literature review was conducted using the PubMed and Google Scholar databases with the following keywords: "glaucoma," "cataract," "diabetic retinopathy," "age-related macular degeneration," "artificial intelligence," "machine learning," and "deep learning."

Summary:

Diseases such as glaucoma, cataract, DR, and AMD significantly impact patients' quality of life. Factors like the growing number of patients, limited access to specialists, and time-consuming diagnostics have increased interest in AI-based tools. In recent years, machine learning (ML) and deep learning (DL) have contributed to faster, more objective diagnostics. In ophthalmology, AI enables automatic analysis of fundus images, prediction of disease progression, and remote monitoring. These solutions support early detection, individualized treatment plans, and improved access to care. AI is particularly promising in screening programs for DR, analyzing optic nerve structures in glaucoma, and enhancing precision in cataract surgery and AMD progression monitoring.

Conclusions:

AI applications in ophthalmology have the potential to improve early diagnosis, optimize treatment, and ease the burden on clinicians. Despite this progress, challenges remain—such as the opaque decision-making of AI systems, ethical issues, and integration into routine clinical workflows. Addressing these barriers will be key to realizing the full benefits of AI and guiding future research in this rapidly evolving field.

Keywords: Glaucoma, cataract, diabetic retinopathy, age-related macular degeneration, artificial intelligence, machine learning and deep learning.

Introduction

In recent years, the rapid development of artificial intelligence (AI) has been observed, along with a growing number of sectors deciding to implement it. The continuous advancement of medicine has led to the increasingly frequent use of AI algorithms in diagnostic and therapeutic processes. Numerous studies have demonstrated the high effectiveness of AI in medicine. One example is an AI algorithm developed for the detection of breast cancer based on mammographic images, created by specialists from Google Health and DeepMind. This algorithm, based on more than 28,000 mammographic images, identified the disease with an average of 12.1% greater accuracy than experienced radiologists and significantly reduced the number of false-positive diagnoses. [1] Another example is an algorithm developed using deep learning (DL), which, through the analysis of chest computed tomography (CT) images, was able to identify pneumonia specifically caused by COVID-19, achieving 94.93% sensitivity and 91.13% specificity. [2] Ophthalmology is a medical field that heavily relies on imaging during diagnostic and therapeutic processes. Researchers have noted that the enormous volume of generated images can support the development of complex algorithms that may subsequently be used in daily clinical practice. Existing studies have shown that AI can aid in the diagnosis and treatment of conditions such as diabetic retinopathy (DR), glaucoma, age-related macular degeneration (AMD), and anterior segment diseases such as cataract and keratitis. The ongoing evolution of individual components of AI, alongside emerging concerns among technology experts regarding the inner workings of specific algorithms, raises doubts about its applicability in the medical sector. Key considerations also include the safety and protection of patient medical data, ethical issues, and emerging technical and organizational limitations that may affect the potential integration of algorithms into clinical practice. The aim of this study is to provide a comprehensive overview of the current applications of AI in ophthalmology, with a particular focus on the diagnosis of glaucoma, AMD, DR, and cataract, with special emphasis on its diagnostic and therapeutic capabilities. In addition to reviewing the latest technological advancements, this paper also aims to identify the main barriers hindering the broader implementation of AI in clinical practice and to highlight future research directions that could contribute to the effective and responsible development of this field.

Fundamentals of Artificial Intelligence Functioning

Artificial intelligence algorithms can reduce subjectivity in the diagnosis of ocular diseases by analyzing and accurately comparing diagnostic imaging, such as fundus photographs, optical coherence tomography (OCT), or slit-lamp images. AI-assisted systems are capable of detecting subtle changes that may be overlooked by the human eye, which translates into faster and more accurate diagnosis of numerous conditions, including diabetic retinopathy, age-related macular degeneration, and corneal diseases. In addition to diagnostic applications, AI can also enhance the efficiency of ophthalmic institutions by supporting physicians in clinical decision-making, automating selected tasks, and allowing clinicians to devote more time to individualized patient care. Example applications of AI in medicine include image analysis—recognition of anatomical structures and pathologies in diagnostic scans—and expert systems, which are programs based on specialist knowledge operating according to predefined algorithms. It is important to note, however, that not all AI systems must be manually programmed by experts. Modern approaches increasingly utilize machine learning (ML), which enables algorithms to learn independently from collected databases without the need for explicitly defined operations. ML makes it possible to predict outcomes in new clinical cases by analyzing large volumes of medical data. Two main approaches are distinguished in ML: supervised and unsupervised learning. In supervised learning, the algorithm is trained on data that already contain assigned labels (e.g., an image labeled as showing a healthy retina or a pathological condition). Based on these labels, the model learns to recognize specific patterns and categories. In contrast, unsupervised learning involves data without prior labeling; the algorithm independently analyzes them to identify hidden relationships, clusters, or structures. In medicine—especially in diagnostic imaging—a crucial role is played by deep neural networks (DNN), particularly convolutional neural networks (CNN). CNNs are highly effective in image analysis due to their architecture, which mimics the functioning of the human brain. These networks consist of multiple layers of data processing, where initial layers detect simple features such as lines and edges, while deeper layers are capable of recognizing more complex elements, such as specific pathological changes in the eye. Key components of CNN architecture include convolutional layers, which act as filters scanning the image for specific patterns; the ReLU (Rectified Linear Unit) activation function, which accelerates and optimizes the learning process; and pooling layers, which reduce data dimensionality and enhance model efficiency. Owing to this architecture, CNNs achieve high accuracy in medical image recognition, making them extremely useful in the diagnosis of ocular diseases. [3][4]

Glaucoma

Glaucoma represents a group of disorders characterized by progressive optic neuropathy. It leads to characteristic damage of the optic nerve fibers and changes in the optic disc, resulting in visual field defects. The primary risk factor for glaucoma is elevated intraocular pressure (IOP). If left untreated, glaucoma can lead to irreversible blindness. [5] Artificial intelligence has been applied in glaucoma diagnostics, including the analysis of data from the Sensimed Triggerfish device—an innovative tool based on contact lenses that allows for continuous monitoring of IOP. Triggerfish measures corneal deformation due to diurnal fluctuations in pressure, providing more detailed insight into IOP dynamics than isolated office-based measurements. In one study, Martin et al. analyzed data from 24 prospective studies involving this device, applying machine learning techniques to evaluate the utility of Triggerfish in differentiating between open-angle glaucoma (OAG) and healthy eyes. Their results demonstrated that combining data from Triggerfish with ambulatory IOP monitoring offers a more reliable indicator of glaucoma diagnosis than either method used alone. AI algorithms proved effective in analyzing complex temporal data and identifying patterns specific to glaucoma, making Triggerfish a promising diagnostic biomarker. [6] However, despite encouraging outcomes, technological challenges remain—such as the rigidity and bulkiness of contact lenses embedded with electronic circuits, limited sensitivity to subtle pressure changes, and the complexity of their production. Addressing these issues could enhance the practicality of Triggerfish as a daily diagnostic and monitoring tool for glaucoma patients. [7] Fundus examination plays a critical role in glaucoma diagnostics, as it enables the assessment of changes in the retina and optic nerve head characteristic of the disease. Key parameters include optic disc cupping, cup-to-disc ratio (CDR), retinal nerve fiber layer (RNFL) thickness, as well as peripapillary atrophy and optic disc hemorrhages [8]. Traditionally, these features were evaluated manually using color fundus photographs. However, the development of AI—especially deep learning algorithms—has revolutionized this process. Since 1999, there have been efforts to use AI for the automated localization of anatomical structures in fundus images. Over the years, advanced models have been developed that are capable of detecting glaucomatous neuropathy features, such as elevated CDR, neuroretinal rim thinning, and RNFL defects [9]. For example, Li et al. developed a DL algorithm that achieved high sensitivity (95.6%) and specificity (92%) in detecting glaucomatous optic neuropathy based on color fundus photographs. [10] Other research groups, such as Bhuiyan et al., have created tools focused on CDR assessment to aid in screening for suspected glaucoma cases, although these models demonstrated lower performance compared to AI-based detection of other diseases, such as diabetic retinopathy. [11] Further studies, including one by Al-Aswad and colleagues, revealed that the AI-based Pegasus system achieved diagnostic performance comparable to that of experienced ophthalmologists—and even exceeded them in certain cases—underscoring its potential utility in screening programs.[12] Moreover, Masumoto et al. integrated fundus image analysis with visual field test results, allowing for more precise identification of glaucomatous eyes. This combined approach, incorporating both structural and functional data, represents the future of AI-supported diagnostics. Although the technology still requires further validation and optimization, it already significantly enhances the accessibility and efficiency of glaucoma detection, particularly in areas with limited access to specialists. [13]

Diabetic Retinopathy

Diabetic retinopathy develops in the majority of patients with long-standing diabetes, regardless of type, and may lead to significant visual impairment or blindness. The continuous rise in diabetes prevalence, alongside increased life expectancy of diabetic patients, has positioned ophthalmic complications as a leading cause of irreversible blindness in developed countries. One example of artificial intelligence application in DR diagnostics is the RetmarkerDR system, developed by a group of experts in Portugal as early as 2011. This system uses machine learning algorithms to detect characteristic DR lesions, particularly microaneurysms. The algorithm analyzes fundus photographs taken over various time intervals and compares them, enabling the monitoring of the appearance, persistence, and resolution of microaneurysms, thus aiding in the prediction of DR progression. In central Portugal, RetmarkerDR is employed in screening programs for diabetic patients, helping to preliminarily categorize individuals as either with or without DR. The system achieves a sensitivity of 97.9% in detecting proliferative diabetic retinopathy (PDR), which is comparable to the performance of ophthalmologists, although it has a relatively lower specificity—resulting in 47% false positives. [14] Pitatti et al. evaluated the effectiveness of the Retmarker DAIRET algorithm in automatically detecting DR among 637 patients, comparing AI results with manual assessments by ophthalmologists. Retmarker DAIRET achieved excellent sensitivity (100%) for identifying treatment-requiring DR and specificity of 80%, indicating high efficacy and potential utility in large-scale screening programs. These results suggest that Retmarker DAIRET could serve as an efficient and safe alternative to traditional ophthalmic exams in systematic DR screening. [15] Lupidi and colleagues assessed the performance of the portable Optomed Aurora IQ camera, integrated with the Selena+ AI algorithm, without the use of pharmacologic pupil dilation—simulating routine clinical conditions. The device captured a single 50° central retinal image in automatic focus and brightness mode. The objective was to determine the presence or absence of DR, as in conventional screening. The study demonstrated high sensitivity and specificity (96.8% each) with near-perfect concordance with ophthalmological diagnoses. Additionally, it confirmed that undilated images obtained with this device are of sufficient quality for effective DR detection. The Aurora IQ camera with Selena+ may serve as a valuable screening tool not only for ophthalmologists but also for primary care physicians, nurses, and technicians, especially in regions with limited access to specialized eye care services. Only four false-positive cases occurred, all associated with moderate cataracts. Although conducted on a medium-sized sample, the study indicates the need for further validation on larger populations. [16] In 2018, IDx-DR became the first fully autonomous AI algorithm in medicine to receive approval from the U.S. Food and Drug Administration (FDA), authorizing it to make diagnostic decisions independently of a physician. As of 2023, the algorithm is known as LumineticsCore. [17] In clinical trials, it demonstrated a sensitivity of 87.2% and specificity of 90.7%, supporting its diagnostic performance as comparable to that of experienced specialists. [18] The system requires two fundus images per eye, obtained using a non-mydriatic retinal camera. However, one of its limitations is a low tolerance for image quality—LumineticsCore classifies more images as unreadable than a human examiner, potentially leading to repeated testing or increased referral rates. Moreover, the system shows limited adaptability to atypical cases or patients with multiple comorbidities. These challenges highlight directions for future research and development. [19]

Age-Related Macular Degeneration

Age-related macular degeneration is one of the leading causes of visual impairment and irreversible blindness among the elderly in developed countries. By 2040, the prevalence of this condition is expected to reach 288 million individuals. [20] There are two primary forms of AMD: atrophic and neovascular. Atrophic AMD is the more common subtype and is characterized by the gradual degeneration and loss of the retinal pigment epithelium (RPE), photoreceptors, and Bruch's membrane. During the course of the disease, drusen—lipid and protein deposits—accumulate between Bruch's membrane and the RPE. In advanced stages, geographic atrophy (GA) develops, defined by clearly demarcated areas of RPE and photoreceptor loss. [21] Neovascular AMD (nAMD), a more aggressive and rapidly progressing form of the disease, accounts for approximately 10–15% of AMD cases. In nAMD, abnormal blood vessels form beneath the retina. Due to their structural immaturity and fragile walls, these vessels often leak, resulting in retinal edema, hemorrhages, and fibrotic scarring. [22] Niu et al. developed an algorithm capable of segmenting atrophic zones on optical coherence tomography scans and identifying structural features such as RPE loss, Bruch's membrane changes, and the presence of hyperreflective foci (HRF). The researchers also created predictive models that, based on baseline imaging characteristics, could forecast the progression and rate of GA expansion over time. The algorithm demonstrated segmentation accuracy comparable to that of expert clinicians and exhibited strong correlation between predicted and actual GA growth. This tool has the potential to personalize patient care by tailoring follow-up intervals and treatment planning according to the individual risk of atrophy progression. However, the model is currently limited by the lack of external cohort validation, short follow-up periods, and omission of clinical variables, which highlights areas for future research. [23] An interesting application of machine learning algorithms in AMD involves predicting anti-vascular endothelial growth factor (anti-VEGF) treatment needs in patients with neovascular AMD. Gallardo and colleagues utilized various ML models to analyze demographic, clinical, and imaging data to estimate the number of anti-VEGF injections required within the first 12 months of therapy. These algorithms showed promising accuracy in predicting treatment demand, with the best models achieving high precision in estimating the number of necessary injections. This suggests that ML approaches could be valuable in clinical practice for developing individualized treatment plans for patients with nAMD. Nevertheless, the study was limited by a small sample size, emphasizing the need for further research and standardization before such algorithms can be implemented in routine clinical care. [24]

Cataract

Cataract is defined as an opacification of the eye's lens and can be categorized as either congenital or acquired. Acquired cataractous changes typically result from the aging process or from metabolic disturbances within the lens caused by endogenous factors related to systemic or local diseases, as well as exogenous factors of physical or chemical nature. Cataract is estimated to be one of the leading causes of reversible blindness, contributing to moderate or severe visual impairment in approximately 52.6 million people worldwide. [25] The only treatment for cataract is surgical intervention, involving the removal of the opacified lens followed by correction of the eye's refractive power in the aphakic state. A crucial aspect of surgical qualification is the calculation of intraocular lens (IOL) power. Patients with atypical ocular biometry or a history of previous refractive surgery continue to pose a significant challenge for clinicians. Important artificial intelligence models applied in the diagnostic and therapeutic process of cataract include support vector machines (SVM) and multi-layer neural networks (MLNN). SVM is a method based on data prediction and classification, while MLNN combines elements from various AI algorithms to enhance performance in predictive tasks. [26][27] Sramka et al. evaluated the application of SVM and MLNN in IOL power calculations. Their findings indicate that these machine learning (ML) algorithms provide higher accuracy compared to traditional clinical methods. The use of these techniques holds promise for improving postoperative refractive outcomes, particularly in patients requiring precise IOL selection or in cases with atypical ocular dimensions. Implementing SVM and MLNN in clinical practice may also reduce the need for postoperative refractive corrections and minimize the risk of additional surgical interventions, thereby decreasing patient discomfort. [28] AI algorithms have also been applied in the diagnosis and treatment of congenital cataract. Lin and colleagues developed a screening model to identify infants at high risk for congenital cataract. The model is based on 11 easily accessible predictive factors. The most significant risk factors included a family history of congenital cataract, low parental education level, comorbid conditions, and the use of oxygen therapy or an incubator post-delivery. Data from 2,005 children were analyzed, and AI-based models demonstrated high performance and stability despite the limited dataset. The model underwent both internal and external validation. However, the authors emphasize that real-world efficacy requires further testing in populations with low congenital cataract prevalence (approximately 4.24 per 10,000 live births). This model could be particularly beneficial in under-resourced regions where access to traditional screening programs is limited. [29]

Limitations of Artificial Intelligence

Artificial intelligence holds significant potential in the detection and evaluation of ocular disease progression. The identification of individuals at risk of such conditions, supported by AI, may reduce the burden on healthcare personnel conducting screening examinations, and can save both time and costs. Despite its substantial promise, AI may not serve as a standalone solution due to various limitations and challenges, including algorithm output interpretability, medico-legal and ethical concerns, standardization and quality assurance, and the commercialization of AI systems as medical devices. [30] One of the main challenges in implementing AI into clinical practice is the variability in imaging quality. AI models are highly sensitive to image quality and may perform poorly when applied to lower-quality images. Variability in imaging may arise from differences in equipment, lighting conditions, or patient positioning, potentially leading to inconsistencies in the features used by AI models for image classification. [31] Another notable concern is the poor generalizability of AI models. While a well-trained model may demonstrate strong performance on data that align with the distribution of its training set, its effectiveness may quickly deteriorate when faced with out-of-distribution data. Even minor alterations in the data—often imperceptible to human observers—can significantly impair model accuracy. The development and validation of AI systems require access to large and diverse datasets, which can be challenging to obtain due to concerns regarding data privacy, security, and ownership. [32] Logistical considerations also present potential barriers. Education on the use and evaluation of AI systems should be incorporated into medical school curricula, and practicing clinicians will require targeted training to facilitate effective integration. Additionally, the recruitment of technical staff—distinct from existing clinical personnel—will be necessary to support collaboration with clinicians and ensure seamless implementation of new technologies. [33] Moreover, AI strategies must be transferable across platforms to accommodate input data from various devices, necessitating the adoption of standardized methodological approaches. [34] It is also critical to recognize that AI behavior may be unpredictable, regardless of its apparent safety, even when developers have adhered strictly to best practices. [35]

Summary

Artificial intelligence is increasingly applied in ophthalmology, particularly in the domain of image-based diagnostics of diseases such as glaucoma, diabetic retinopathy (DR), and age-related macular degeneration. AI models analyzing fundus photographs, optical coherence tomography scans, or slit-lamp images demonstrate high sensitivity and specificity in detecting pathological changes. In the case of glaucoma, tools such as contact lens sensors—e.g., the Triggerfish system—are utilized to enable continuous monitoring of intraocular pressure, potentially facilitating earlier diagnosis of the disease. AI also effectively supports the analysis of visual fields and fundus images, providing precise insights into glaucomatous neuropathy. In the diagnosis of DR, systems such as RetmarkerDR and Selena+ have shown high efficacy in identifying disease-related changes and supporting screening programs. In AMD, deep learning algorithms are being developed to segment pathological features on OCT scans and predict disease progression, thereby enabling earlier initiation of appropriate treatment strategies. It is important to emphasize, however, that AI also has its limitations. The quality of analyzed images significantly affects diagnostic accuracy, and models may struggle to generalize to new data that deviate from the training dataset. Implementation of AI in clinical practice additionally requires adequate training of personnel, the assurance of high-quality data, and strict protection of patient privacy. In conclusion, AI holds substantial potential to revolutionize the diagnosis of ophthalmic diseases. Nonetheless, its full integration into daily clinical practice necessitates continued technological and organizational advancements [30–35].

Disclosure

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