

USING AI TO IDENTIFY PATHOLOGIC MYOPIA AND MYOPIC MACULOPATHY



How research on the use of AI can translate to future clinical applications.

BY YINING WANG, MD; TIEN YIN WONG, MBBS, MMED (OPHTH), MPH, PHD; AND KYOKO OHNO-MATSUI, MD, PHD

Pathologic myopia (PM) is a leading cause of irreversible blindness in East Asia and a growing global public health concern.¹⁻⁶ According to the International Myopia Institute, PM is characterized by excessive axial elongation associated with myopia, resulting in structural changes in the posterior segment of the eye, such as posterior staphyloma, myopic maculopathy (MM), and high myopia-associated optic neuropathy.⁷

To standardize the diagnosis of MM, a classification system known as the META-analysis for pathologic myopia (META-PM) was introduced in 2015 based on fundus photographs.⁸ This system categorizes MM into five grades (0 to 4) according to the severity of atrophic changes, with three additional “plus” features: lacquer cracks, myopic macular neovascularization (MNV), and Fuchs spot. Based on this classification, PM is defined as myopic eyes with MM equal to or more severe than diffuse atrophy and/or eyes with posterior staphyloma.⁹

Deep learning (DL) architectures—particularly convolutional neural networks (CNNs)—have demonstrated remarkable efficiency in detecting ocular diseases from fundus photographs, including the diagnosis and classification of PM.^{10,11} AI-powered DL systems not only offer automated classification, but also have the potential to enhance diagnostic efficiency, making them valuable tools for large-scale screening and clinical decision making (Figure 1).¹²

FUNDUS-BASED PM DETECTION USING AI

As a cost-effective and noninvasive imaging modality widely used in routine eye care, fundus photography

remains the primary choice for AI-driven PM detection. Its accessibility has facilitated the construction of large-scale datasets, enabling the development of robust CNN architectures. Additionally, the META-PM classification based on fundus imaging aligns well with AI model labeling requirements, further supporting automated PM detection.

A recent meta-analysis of 11 studies involving 165,787 eyes reported high diagnostic performance of AI-based tools in detecting MM and PM from fundus images, with an area under the summary receiver operator curve of 0.9905 and a pooled sensitivity of 0.959.¹³ Several representative studies illustrate the evolution of this field—from simple binary classification (ie, MM vs non-MM) to more detailed grading across all five META-PM categories. In 2021, a retrospective multicohort study using 226,686 fundus images from nine multiethnic cohorts across six regions developed DL algorithms for classifying high myopia and MM.¹² In the same year, two studies demonstrated the ability of DL models to classify MM across categories 0 to 4 and detect “plus” lesions.^{14,15}

The application of advanced computer vision techniques has enhanced the performance and efficiency of AI-based MM and PM detection. These innovations aim to reduce reliance on extensive manual annotations and explore the potential for fully automated diagnosis.

For example, Sun et al introduced a module that used the information of tessellated fundus and brightest image regions to assist in lesion localization using coarse-labeled images.¹⁶ Yao et al developed DeepGraFT, a classification- and-segmentation co-decision model that first applies

image masking to isolate the region of interest.¹⁷ This was followed by a binary classification for each MM category by Zhang et al that uses a technique known as self-supervised learning,¹⁸ which refers to the development of generalist models capable of adapting to various downstream tasks with significantly less annotated data, demonstrating promising performance in automated MM diagnosis and grading.

CHALLENGES TO AI IN PM SCREENING

Establishing Clear, Unified Definitions of PM and MM in AI-Based Studies

One of the major challenges with AI-driven detection of PM and MM is inconsistency in their definitions. In the previously mentioned systematic review and meta-analysis, 17 studies were included in the systematic review, and only eight explicitly stated the use of the META-PM classification for MM identification, while the remaining studies did not clarify which classification system was applied.¹³ This inconsistency complicates direct comparisons between models, as variations in diagnostic criteria can lead to significant differences in reported performance. Additionally, the lack of standardized definitions limits model generalization across diverse datasets and clinical settings. Future research should focus on establishing unified diagnostic criteria and standardized image labeling frameworks to enhance the reliability and applicability of AI models.

Evaluating DL Algorithms in Diverse, Real-World Settings

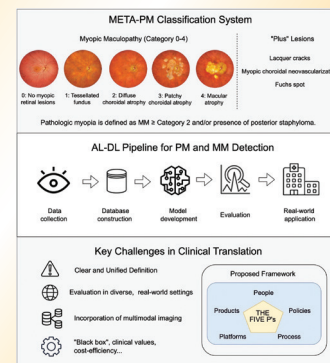
Despite the increasing availability of publicly annotated datasets for PM (eg, the Pathologic Myopia Challenge dataset and the Singapore Epidemiology of Eye Diseases study dataset),¹⁹⁻²¹ developing robust DL models for PM and MM diagnosis remains challenging. Variations in medical systems across different regions result in differences in the prevalence of PM and MM subtypes within study cohorts, affecting model performance and generalizability.

Recently, Qian et al introduced a publicly available dataset for MM diagnosis as part of the Myopic Maculopathy Analysis Challenge.²² This dataset comprised 2,306 fundus images for MM classification, with seven teams participating in the competition. However, all fundus images were exclusively sourced from Chinese patients, which may limit the model's generalizability to other populations. Future research should expand datasets to include multiethnic populations and evaluate model performance in diverse clinical environments to ensure real-world applicability.

Incorporating Multimodal Imaging

While the META-PM classification provides a standardized framework for identifying various stages of MM, it is solely based on fundus photographs, which presents

Scan the QR code to view this article online, which includes a schematic that illustrates the META-PM classification system used for MM and PM diagnosis, the development pipeline of AI-DL algorithms, and the key challenges to clinical translation.



potential diagnostic limitations. Fundus pigmentation variations among racial and ethnic groups can affect image interpretation, and other critical myopic macular pathologies, such as myopic traction maculopathy and dome-shaped macula, are not included. To address these gaps, an OCT-based classification has been proposed.²³

Recent studies have demonstrated that DL models based on OCT images can reliably detect PM and its complications, including MNV, dome-shaped macula, and tractional changes such as retinoschisis, macular hole, and retinal detachment.²⁴⁻²⁷ However, compared with fundus photography-based models, OCT-based AI research remains relatively limited. One major challenge is the lack of uniform diagnostic criteria and large annotated datasets, likely due to the complexity of PM and its diverse manifestations. Moreover, variations in OCT imaging systems used in real-world clinical practice pose additional barriers to the widespread implementation of these AI algorithms.

Despite the success of multimodal DL approaches in conditions such as glaucoma and AMD,²⁸⁻³³ their application in PM remains underexplored. Future research should focus on integrating fundus photography, OCT, and other imaging modalities to enhance diagnostic accuracy and provide a more comprehensive assessment of PM-related complications.

CLINICAL TRANSLATION

In addition to challenges related to AI model development and evaluation, systemic barriers remain in the clinical translation of AI-based DL algorithms for PM and MM detection. First, the “black box” nature of many AI algorithms continues to hinder trust and acceptance among clinicians. This challenge has given rise to the field of explainable AI, which focuses on developing models that not only achieve high accuracy, but also provide transparent, interpretable reasoning behind their outputs,

thereby enhancing clinical trust and usability.³⁴

The lack of rigorous clinical trials also limits our understanding of the true clinical value of these models—are they safe, effective, affordable, and relevant in the dynamic health care environment?³⁵ While many algorithms demonstrate high diagnostic performance in research settings, few have demonstrated meaningful clinical effect in real-world practice.³⁶ Furthermore, the seamless integration of AI tools into existing clinical workflows remains an obstacle. For example, it is unclear whether current PM and MM detection models are better suited for use in primary eye care settings or specialized settings, such as retinal clinics or high myopia centers. Determining the most appropriate clinical environment is essential for maximizing their utility and minimizing workflow disruption.

ADDRESSING THESE CHALLENGES

Gunasekeran et al proposed a comprehensive framework known as the “5Ps: People, Policies, Processes, Platforms, and Products.”³⁷ This framework outlines essential elements for the successful, large-scale deployment of medical AI solutions, such as those implemented in national diabetic retinopathy screening programs. Applying a similar framework to AI-based MM and PM detection systems may facilitate more effective, scalable, and sustainable integration into health care systems. ■

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YINING WANG, MD

- PhD Student, Department of Ophthalmology and Visual Science, Institute of Science Tokyo, Japan
- yiningwang241@gmail.com
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TIEN YIN WONG, MBBS, MMED (OPHTH), MPH, PHD

- Professor & Senior Vice Chancellor, Tsinghua Medicine, Beijing, China
- Director, Beijing Visual Science and Translational Eye Research Institute (BERI), School of Clinical Medicine, Tsinghua Medicine, Tsinghua University, Beijing, China
- Senior Advisor, Singapore National Eye Center, Singapore
- wongtiennyin@tsinghua.edu.cn
- Financial disclosure: Consultant (Astellas, Bayer, Boehringer-Ingelheim, Carl Zeiss Meditec, Genentech/Roche, Novartis, Opthea, Plano, Quareite Biopharm Research, Sanofi, Shanghai Henlius); Inventor/Patent Holder/ Co-founder (EyRIS, Visre)

KYOKO OHNO-MATSUI, MD, PHD

- Professor, Department of Ophthalmology and Visual Science, Institute of Science Tokyo, Japan
- k.ohno.oph@tmd.ac.jp
- Financial disclosure: Consultant (CooperVision, Eye Rising, Santen, Topcon)