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AI in Myopia

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Introduction

Myopia, nearsightedness, the most common type of refractive error, is an ocular problem of major public health proportions. Some 40% of adults and over 80% of children completing high school are myopic in urban areas of East Asian countries. Guo H et al, Self-reported myopia in Taiwan: 2005 Taiwan National Health Interview Survey, Eye (Lond.) 26(2012). While uncorrected myopia and refractive errors are major causes of visual disability in children, adults with high myopia are at significantly higher risk of losing their vision. (Saw S et al., Myopia and associated pathological complications, Ophthalmic Physiol. Opt, 25 (2005). Elongated axial length has been one of the most attractive endophenotypes investigated in myopia and heritability is the second metric that has consistently been examined. (Klein A et al, Heritability analysis of spherical equivalent, axial length, corneal curvature, and anterior chamber depth Beaver Dam Eye Study, Arch Ophthal, 127, (2009). More recently, using AI and deep learning to: (1) develop myopia subcategory patient datasets, (local and global), (2) achieve corresponding subcategory longitudinal follow-up, and (3) evolve mature deep learning segmentation tactics in fundus photo, OCT and fundus autofluorescence imagery as well as completion of Genome Wide Association Studies, all of these advances have brought myopic phenotypic and genetic causal elements into sharper view.

Methods

Dox gpt, Google Gemini, and Microsoft Copilot were queried for Pub Med and Google Scholar references in the last ten years on myopia epidemiology, pathology, economic considerations, and in the last five years for machine learning and deep learning myopia conferences, large group or foundation collaborations resulting in recommendations for myopia diagnostics, management and future studies. Comprehensive articles in myopia genomics, phenotypic/genotypic correlates and inherited retinal diseases were reviewed from the above mentioned search locations. AI Studies that achieved clinical train and test and validate with diverse, well culled patient datasets, with clear, comprehensive CNN architecture(s) definitions e.g. for fundus photos or OCT image classification, and or presented criteria that were weighed and considered for consensus by an august specialty body, after achieving higher accuracy metrics in studies, were favored for AI/Myopia presentations in this paper.

Results

Myopia Epidemiology, Pathology, Economic Considerations

Myopia with an expected global prevalence to almost 5 billion people by 2050, 49.8% of the world's population, is a common ocular condition [1,2]. High myopia with a refractive error of -6.00 diopters or greater may lead to glaucoma, cataract, retinal tears, and retinal detachments with vision threatening consequences. Progressive ocular axial elongation with pathological myopia may cause visual impairment in about 3% of the population and between 50-70% of myopes [2-6].

Traditionally, pathologic myopia is first visualized on fundus imaging and may include peripapillary optic atrophy, temporal flattening of the optic disc, posterior staphyloma, retinal pigment epithelium variegation and or atrophy and Fuchs spot, (gray to black central macula circular spot from choroidal bleeding, resulting in blurred or decreased central vision). Essentially, the most common cause of blindness in pathologic myopia is choroidal

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neovascularization, which if untreated can result in significant visual loss [2,3,9]. Reduced vision secondary to pathological myopia exerts a large impact on quality of life, social and emotional wellbeing and functional ability. Naidoo et al 2019, estimated the productivity loss among individuals with visual impairment from myopic macular degeneration in the US at $6 billion. Southeast Asia, South and East Asia, particularly, bear the greatest potential burden of economic activity loss from uncorrected and pathologic myopia [7].

Myopia Treatment Regimens Have Had Reduced Acceptance

E Prousali, et al. in 2019 reported that although in 18 treatment reviews involving 6400 children given 1% Atropine drops, followed by orthokeratology, and or novel soft contact lenses resulted in considerably greater 1 year myopic reduction compared to controls, doctors failed to uniformly embrace these treatments. This was possibly related to Atropine side effects or the doctors' belief that there was significant population heterogeneity [8]. 1% Atropine induced blurred vision with an odds ratio of 9.47 and hypersensitivity of 8.91, (which are considered high odds ratios) [8]. Thus, besides better methods to pick up pathologic myopia early, acceptable treatment regimens have not been largely accepted by the physicians and endemic populations [2,9].

Review of AI Model Subtypes Using Fundus Photos for Pathologic Myopia Detection

To help understand the evidence for AI diagnostic utility of examining fundus photos in pathologic myopia, especially in constrained areas, where expert clinicians are not available, Prashar and Tay in 2023 reviewed 17 studies in the systemic review and 11 studies evaluating 165,787 eyes from the meta-analysis. The pooled sensitivity was 95.9% and pooled specificity was 96.5% [2]. This included retrospective fundus image datasets from China, Turkey, Japan, USA, Singapore, Belgium, Indonesia, and Pakistan. Diverse AI model subtypes included CNN, convoluted neural networks and SVM, (SVM can replace Softmax as a closing convolving layer in CNN because it can optimally separate data classes of images) and keras, which can help code EfficientNet, which helps scaling a balance between depth, width and image resolution accuracy. Other different CNN architectural subtypes were also included in the review. CNN studies had a 0.960 sensitivity and specificity 0.999-1.0. Univariate meta-regression showed no statistically significant effect of training set size to account for heterogeneity [2]. Fourteen studies used direct labelling by expert ophthalmologists or retina specialists as the reference standard. 7 of 11 studies reported a comparison with human graders. These model sensitivities ranged from 0.719-0.986 and specificities from 0.972-0.998 [2]. Prashar and Tay [2] illustrated in the above review that AI screening methods for pathologic myopia were highly sensitive and specific. However, none of the studies reported by them included the implementation in clinical practice [2].

Clinical Realm validation of AI detection of Pathologic myopia with OCT and Retinal Fundus Photos

For clinical validation, Li et al in a prospective study from Zhongshan Ophthalmic Center, in 2019, obtained 412 OCT images from 91 high myopia patients. The CNN InceptionResnetV2 architecture was adapted to train four independent CNN models to identify four vision threatening conditions: high myopia: retinoschisis, macular hole, retinal detachment and pathological myopic choroidal neovascularization [10]. For all vision threatening conditions of high myopia the areas under the receiver operating curves were 0.961 or greater. The AI system achieved sensitivities equal to or better than the involved retina specialists and may be applied to large scale high myopia screening and patient care [10]. Tan, et al. in 2021 with retinal fundus photos validated deep learning algorithms for myopic macular degeneration detection in Singapore, China, Taiwan, India, Russia and UK with an AUC > or = 0.969 developing a blockchain based AI platform for secure data and model transfer across borders. The blockchain development may serve as a trusted platform for performance testing of future AI models across countries [11].

Zhu, et al. in 2026 [12] in a prospective longitudinal study including 375 children with bilateral high myopia, greater than -6.00 diopters, from Zhongshan High Myopia Cohort, following a median of 15 years, applied deep learning automated retinal vascular phenotyping to fundus photos. These photos were then integrated with longitudinal ocular biometry and swept- source optical coherence tomography metrics. Multivariate models were then used to identify independent predictors of pathologic myopia onset and progression. Pathologic myopia developed in 64 children or 17.1%. Independent predictors included lower retinal vessel density, (OR 9.69 CI 4.33-21.70); reduced fractal dimension, (OR 5.94 CI 3.07-11.50); narrower arteriolar and venular calibers, central retinal arteriolar equivalent (OR 3.44, 95% CI 1.86-6.34), central retinal venular equivalent, (OR 4.42 95% CI 2.24-8.71); thinner subfoveal choroid (OR 2.64 CI 95%, 1.56-4.45); faster early axial elongation (OR 1.72, 95% CI 1.17-2.50); accelerated early choroidal thinning, (OR 2.23 CI 95% 1.41-3.84); The Zhu et al. AI oculometric model, which characterized pathological myopia risk through retinal vascular and retinal layer growth dynamics over 15 years in myopic children had excellent discriminatory capability, (AUROC 0.96 CI 95% 0.92-1.00, improving to 0.98 CI 95%, with the biometric variables from the previous sentence [12].

The Zhu et al study illustrates that AI enabled retinal profiling of aging myopic changes in children not only offers a scalable risk stratification platform but also supports the potential for targeted therapy to slow or stop pathological myopic progression [12].

AI Prediction of Causative Retinal Genes in Inherited Retinal Disease (Eye2 Gene) from an input of patient retinal scans, including infrared, autofluorescence and SD-OCT

Looking at progressive retinal changes from the larger phenotypic-genotypic platform of inherited retinal diseases, IRD, Nguyen, et al. 2025 [13] retrospectively with a 10, 000 patient cohort from the UK and Japan sought to predict causative retinal genes from retinal images of patients diagnosed with IRD.

A brief retinal/ genetic digression to appreciate the myopia gene landscape.

In 2023, the International Myopia Institute, IMI, categorized myopia into two genetic mechanisms: (1) the vast majority, polygenic, genetic risk interacting with environmental factors and (2) monogenic early-onset high myopia due to single gene mutations, which can be isolated or syndromic [14]. This last group is estimated at 23-36% of high myopia patients globally and includes: (1) ametropic retinal dystrophies: Retinitis Pigmentosa GR/RPE/65, associated with increased axial length [15], congenital stationary night blindness (associated with 17 different genes of phototransduction, signal transmission, and RPE retinoid recycling) [16], blue cone monochromacy (unlike rod monochromism, which is autosomal recessive and affects all three types of cones, blue cone monochromism is X-linked recessive and only affects blue L-cones and M-cones; rods and S-cones are normal; prevalence 1 in 100,00 people). [17]; (2) connective tissue disorders **: Marfan syndrome [14], Stickler syndrome [18], Ehrler's-Danlos syndrome [19], (** screen for joint hypermobility and skin elasticity) and Knobloch syndrome (high myopia, glaucoma, retinal detachment occipital encephalocele) [20]; and (3) monogenic isolated high myopia: ARR3, the most common cause of Mendelian early onset high myopia [21]

Myopia Genomics and AI

Returning to Nguyen, et al, [13]. The Eye2Gene project, still in its early development, was divided into 8 work packages. The first was the development of a classification algorithm. Using Convolutional Neural Network, CNN, model, the N,10, which covers 70% of IRD, the most common IRD at Moorfields were selected, with the top five accuracy of at least 88%. They focused on high gene accuracy for the rarer genes [13]. The second step was to manually curate and segment the dataset provided by the Moorfields Reading Center IRD Segmentation expert Team. These segmented IRD datasets were useful for training multiple AI algorithms for Eye2Gene. They included patient photos of: SD OCT, infrared reflectance or short wavelength fundus autofluorescence. Other shape and distribution characteristics were also programed in so that a segmentation algorithm based on U-Net CNN could be developed from the dataset [13]. The classification algorithm was then combined with the segmentation algorithm to create an explainable accurate algorithm, which was then combined with a multinomial logistic regression and additional demographic items to enhance predictive value. Iterative adjustments were made to enhance confidence scores [13].

After looking at gene classification probability, segmented IRD features were translated to Human Phenotype Ontology, HPO, terms to support HPO-base phenotype prioritization using e.g. the Exomiser [22] approach, (Exomiser is a software that is affected by parameters such as allele frequency cutoffs and gene variant filter settings). Then they were assessed for phenotypic-driven variant prioritization to help solve cases with multiple candidate variants. This aided in achieving the ACMG annotation guidelines, namely that the patient's phenotype or family history is highly specific for a disease with a single genetic etiology [13,23]. Resource use and cost, considering standard care vs use of Eye2 Gene was modeled with cloud based software, following a QMS, quality management system, approach [13]. A Patient advisory group to contribute to decisions and result dissemination meets three times per year so that risks raised by participants will be added to the register for the QMS [13]. To learn about user experience, the Eye2Gene group will explore human elements related to expectations and outcomes [13]. SD-OCT image segmentation included: drusen, subretinal fluid, intraretinal fluid, cysts, subretinal hyper-reflective material, retinal pigment epithelial loss, choroidal hyper transmission, and foveal hypoplasia [13].

Although Nguyen, et al. [13] are still early in the protocol to execution phase, in examining their underpinnings, one can get a glimpse of the attention to detail involved in phenotypic genotypic research from the AI and genomic platforms. Voogelaar, et al. in 2025, [24] (after a comprehensive literature review (for the IMI – Myopia Genetics Report) including genome wide association studies, GWAS, rare variant analysis, functional genomics, and multiomics), have associated 1000 common genetic variants with refractive error and myopia. These genes have been implicated in pathways of retinal signaling, extracellular matrix remodeling, and neurodevelopment. This suggests that axial length may be a productive phenotype for further myopic gene inquiry and understanding [24]. New rare gene variants, for high myopia and syndromic myopia have been uncovered by whole exome and whole-genome sequencing studies [24].

Combined with environmental and demographic factors, polygenic risk scores show improved predictive power. In the myopia management realm, this suggests a role for educational adaptations, screen time, physical activity and metabolic and or inflammatory biomarkers in refractive error development [24] Patasova, et al. [25] to elucidate genetic factors that influence refractive errors obtained data on refractive error and age of first spectacle wear (AFSW) from the UK Biobank European in 340,318 subjects. Time to event analysis found significant genome wide associations at 44 independent genomic loci, many of which were previously associated with refractive error such as, GJD2 and LAMA2. (identified through GWAS, GJD2, Gap Junction Delta-2 encodes connexin 36, a protein that forms electrical gap junction channels in photoreceptors, bipolars, and amacrine cells [37] and LAMA2, an extracellular matrix protein crucial for basement membrane integrity, cell signaling in eye and neural pathways [38]. Both proteins play a keen role in axial length elongation and refractive error [25]. Patasova, et al. also identified six novel regions associated with AFSW, the most significant of which was on chromosome 17q for rs55882072. They learned that genes associated with AFSW were significant for central nervous system neurogenesis [25].

Jin, et al. [26] to integrate neural network causal attribution in their myopia AI model (and avoid the "Blackbox" nature of CNN outputs and attributions), used a pediatric ophthalmology cohort tracking over 3000 children longitudinally. To identify direct and indirect causal effects of input features in myopia progression, they categorized input neurons into three categories: isolated (input neurons that do not have any causal relationship with other input features, e.g. carbonated beverages or egg consumption frequency),

pure (input neurons that influence the output only through downstream mediators e.g. number of parents wearing glasses or gender), and confounded units (input neurons whose effect on the output is entangled with another causally related variable, such as axial length or cycloplegic refraction). Thus, variable input in different neuron entry categories achieved positions in the learned causal structure. This structure is based on targeted attribution strategies for each unit with a domain-adaptive meta learning approach for iterative estimation of causative effects under confounding bias. This causal attribution model allowed reconstruction of different interpretable causal pathways within the network, enabling mechanistic understanding of how each categorized input signal propagates through the CNN architecture and identified their individual causative relationship to the outcome [26].

This type of model transparency enhances digital deep learning and reliability. In addition, to uncovering underlying causal relationships among input features, a constraint based causal discovery algorithm, combining the PC algorithm with Degenerate Gaussian Scoring and dimensionality reduction was used [26].

A digression to explain analytic causal metrics in CNNs

(PC refers to the Peter-Clark algorithm used to develop a directed acyclic graph, dag. When paired with the Degenerate Gaussian Scoring, it provides a mathematically rigorous way to handle deterministic colinear variables or high-dimensional linear variables by allowing the joint variable distribution to collapse into a lower dimensional subspace. After conditional independence testing for skeleton extraction of graph variables, it identifies variables not in the conditioning sets and uses deterministic logical steps called Meek Rules [28] to orient the X and Y graph values without creating new co-variables. Degenerate Gaussian Scoring empowers causal discovery because it allows pure constraint based variable tests to swap for score-based metrics modified for rank deficient data, such as Degenerate Gaussians. In essence, PC in conjunction with the Degenerate Gaussian scoring helps isolate true deterministic independent causal drivers from redundant or downstream effects, allowing the causal graph generated to represent basal ancestral relationships rather than mathematical artifacts [26,27]. The final causal graph was constructed using the TETRAD software suite [29].

The directed acyclic graph is acyclic, meaning you cannot follow the causal arrows from say A to B to C, etc. in a circle. This respects the unidirectional flow of time in the convolving sequences. Thus, in causal discovery a dag is not drawn by hand connecting expert opinions. Special computer algorithms analyze the observational data and automatically learn the structure of the dag. Unlike conventional statistical metric associations, like Pearson correlation coefficients, R squared values or regression coefficients, (which e.g. may assume that if X correlates with Y, then Y correlates with X or e.g. in regression models which allow all or most variables into the model to control for them), dags work on interventional logic. It accounts for asymmetric X, Y changes, so if changing X changes Y, altering Y may not change X. Unlike regression models, dag uses math rules to pinpoint exactly which variables need to be controlled for and which ones can be omitted. [26,27],

YouTube Introduction to DAGS for Causal Inference, UCLA Office of Advanced Research Computing] Also, Pearson correlation, Chi-squared, Fisher's Z test are limited to continuous or discrete data and have difficulties with mixed domain data [26]. Thus. Jin, et al. [26] used the degenerate Gaussian with the PC algorithm, first proposed by Andrews [26,30], to explore and conclude on principal myopia causal drivers. Jin, et al's. [26] first binary classification model for myopia predictions using feedforward neural network was trained on the pediatric ophthalmic cohort dataset from Henan, China from 2011 to 2017. It consisted of 16 variables, spanning behavioral, physiological, dietary, environmental and genetic domains. The model achieved a 0.933 accuracy vs other traditional machine learning models, such as SVM, (0.863 accuracy), Logistic Regression, (0.935 accuracy), naive Bayes, (0.845 accuracy), K-nearest neighbors, (0.922 accuracy), and random forest, (0.936) [26].

Jin, et al. [26] demonstrated that black box neural networks can be elevated into causal substructures [31] such as the interpretable variable progression features of myopia. The model's ability to follow known physiological mechanisms over time particularly axial length and cycloplegic refraction which directly change the eye's focal characteristics help support its biological plausibility. The latter has keen implications for early intervention and individualized treatments [26]. By explicitly modeling conditional dependencies as well as basal ones in the causal paths, confounding factors are included in the early attribution framework and may be critically picked up as systemic bias or misattribution causal patterns. Conventional attribution methods such as SHAP or LIME [32-36] often fail with confounders or produce unstable saliency maps [26]. Jin, et al. with their causal attribution pathway including the PC algorithm, the directed acyclic graph paired with the Degenerative Gaussian Scoring provided a more rigorous analysis for CNN layer myopic variable generation even in the presence of confounding factors.

Conclusion

Polygenic and monogenic myopia are significant public health problems. The diversity of their presentations and global prevalences have challenged conventional early diagnostic and management methods. To address these gaps the International Myopia Institute and other outstanding research groups have carefully endeavored to establish credible representative datasets for machine and deep learning, especially CNNs, so that epidemiologic, pathologic, and genomic contributions to myopia can be analytically assayed and weighed for clinical relevance and causality. The contribution of inherited retinal diseases has also gained recent recognition. With the aid of accurate studies with well trained and validated datasets many of the difficult questions in prediction and management of myopia from the refractive side in children to the macular degenerative side in adults will be able to get answers.

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Conflict of Interest

None.

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