# Artificial Intelligence-Based Diagnostic Aids in Optometry and Ophthalmology

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Disclosure: Dr. Armitage is a consultant to Carl Zeiss Meditec, Inc.

**Historical Perspective**

The mathematician Alan Turing is widely regarded as the originator of the modern concept of machine intelligence; and the term "artificial intelligence" was formally introduced by John McCarthy at the Dartmouth Summer Research Conference in 1956. Until recently, artificial intelligence (AI) was primarily the research domain of computer and data scientists, or it was employed for specific tasks within the medical professions. Even in cases where AI models were used in diagnostic devices such as optical coherence tomographers or widefield imaging platforms, the contribution of AI to a device's functionality was most likely unappreciated by the end user, despite AI having great potential in augmenting clinical reasoning in clinical settings.[1]

In November 2022, the large-language AI model Chat GPT was introduced to the world as an open-use resource. While several large-language models preceded ChatGPT and many have succeeded it, the release of the GPT model likely represents a watershed in terms of public interest in AI models. The factors driving the widespread interest and uptake of generative AI are multifactorial, not only in terms of language generation but also image generation and manipulation, and the computer vision-based analysis that underpins the system. It may be argued, however, that the inherent ease of use of generative AI and its ability to generate human-like language responses to plain language questions foster a level of affinity with the system. In fact, we may be close to the stage where the Turing test, the point where we are unable to differentiate between a human and a machine, has been achieved in terms of generative AI for both language and image generation.[2]

Notwithstanding our affinity for the nature of generative AI language models and their ability to scrape internet-based databases for information and present it in a human-like fashion, less conversational AI models play a vital role in undertaking tasks that humans cannot perform easily or that are particularly tedious or voluminous in nature. As with many new technologies, as the utilization of AI models transitions from the computer and data scientists who write the code to the health practitioners who use the system, new adopters tend to view the technology as a "black-box model" and are most interested in the output of the system. AI is not, however, flawless. It can provide incorrect conclusions, or in the case of generative AI, hallucinations, and nearly all models are limited to work under specific circumstances or situations.[3] Human performance is still superior to AI when clinical reasoning is required,[4] but by combining the strengths of human clinical reasoning with AI to develop an appreciation of the strengths and limitations of these AI models, it can apply this to individual patients. Clinicians and individuals training to be clinicians, need to become familiar with the fundamentals of AI models to develop some understanding of what occurs "inside the black box" to enable the AI model to inform their clinical reasoning strategies.[5] For reference, "black box model" describes models that are difficult to understand or interpret, such as those that use deep-learning algorithms like transformers or recommendation systems. In contrast, "white box" or "transparent" models are easier to understand because their internal workings and reasoning are more accessible.

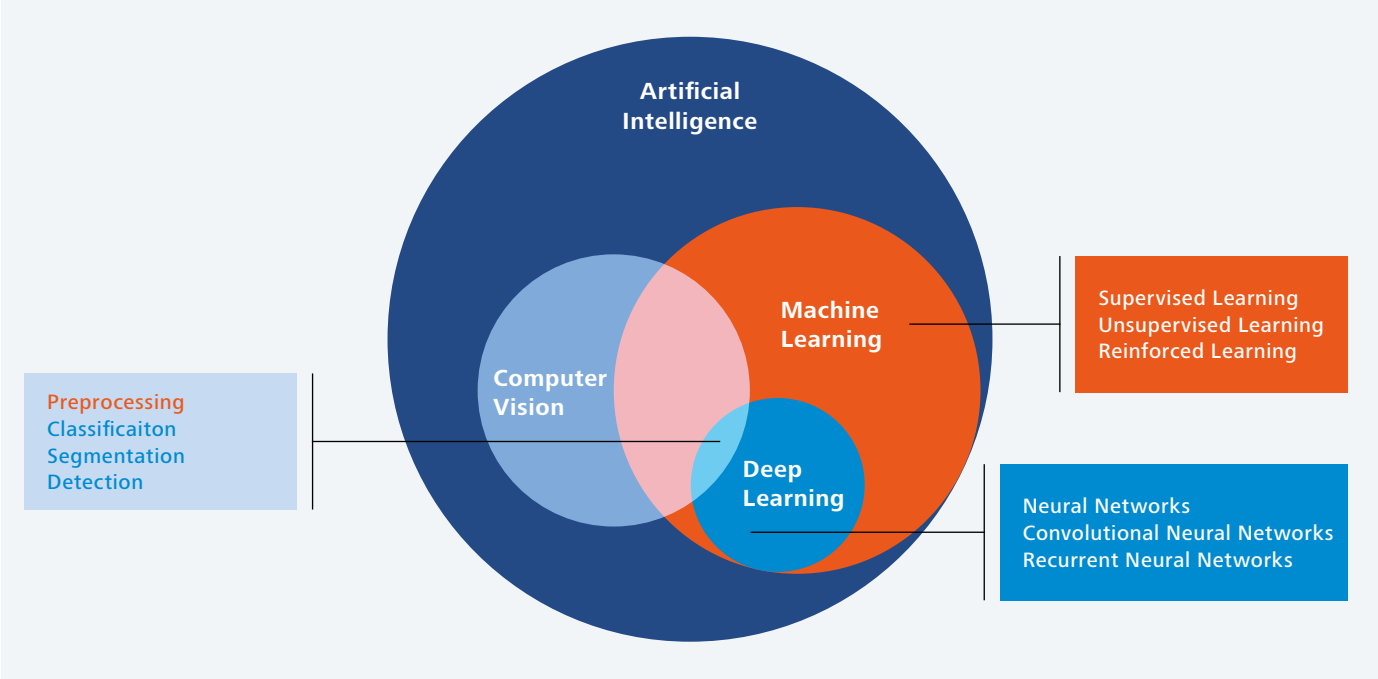


*Figure 1. Venn diagram illustrating the relationship between various aspects of AI and various modalities that reside within each group. Deep learning is a subset of machine learning, and computer vision intersects with both domains.*

**AI Use by Informed Clinicians Offers Improved Patient-Centered Care**

The adoption of AI-driven systems across the globe is widespread, and strategic use of AI promises increased productivity, accuracy, and precision in terms of task execution, as well as improved quality of life. From an ophthalmic health perspective, AI-driven image analysis is an incredibly promising area; however, several considerations need to be addressed before widespread implementation. When AI is used to monitor or improve human health, there is a clear need to validate its safety, efficacy, and suitability for the individual patient to maximize patient-centered care.

Regulatory oversight by health authorities across the globe (e.g., the Therapeutic Goods Administration in Australia; the Food and Drug Administration in the United States; the Medicines and Healthcare Products Regulatory Agency in the United Kingdom, and the European Medicines Agency across the European Union) register AI models as medical devices when the manufacturer can show that a specific model meets safety, precision, and accuracy benchmarks at a population level.[3] As practitioners, we need to be satisfied that an AI model is suitable for use in the diagnosis or management of ocular disease for the individual patient, given the circumstances of the patient sitting in our consulting room chair. To capitalize fully on the diagnostic assistance that AI can offer, optometrists, ophthalmologists, and orthoptists will benefit from a better understanding of how AI models work in order to make informed decisions about the suitability of particular diagnostic models for their patients.

**The Basics of AI**

AI is perhaps the term most widely used to describe the process by which computer programs analyze input and arrive at an output using machine-developed logic rather than logic baked in by the computer programmer. Figure 1 shows a high-level view of the AI landscape. Although all facets of AI have relevance to clinical ophthalmic practice, this article focuses on the computer vision and image analysis aspects of machine and deep learning, as they have the greatest potential to effect paradigm shifts in the near future.

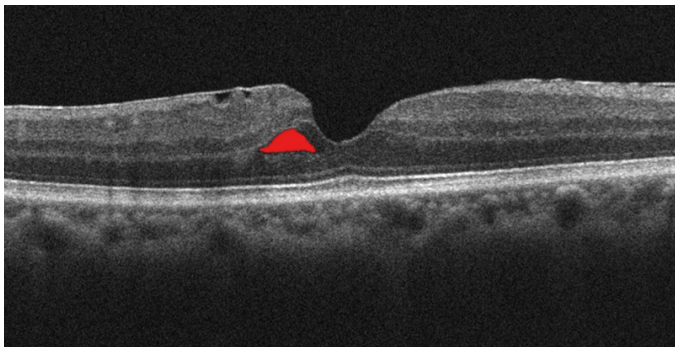

*Figure 2 Exemplar B-scan showing Central Serous Retinopathy (CSR) annotated manually with red fill which forms the ground truth that is used to train the AI model.*

Machine learning is the process by which software develops and refines algorithms to undertake a specific task based on training datasets. Machine learning algorithms are often based on decision trees and cluster analyses. Deep learning is a subset of machine learning and utilizes complex decision-making processes arranged much like the neural networks of the central nervous system. Review articles by Charng et al 2023[6] and Murphy et al 2024[3] discuss these in greater detail with reference to ophthalmic applications, and a review by Chakraboty[7] considers broader applications. In the context of image analysis, both machine learning and deep learning models are paired with computer vision which enables the algorithm to extract information from images. Computer vision also develops or uses image preprocessing routines to align image stacks, reduce noise, and improve contrast. An example of this may be the machine learning tools that have been implemented in optical coherence tomography (OCT) analysis software to automatically segment retinal layers.

**Training of AI Models**

Both machine learning and deep learning models require data to train the algorithm. In the context of image analysis, this usually represents a large (in the order of thousands) database of images, e.g., OCT B-scans, and information about the classification of the image that forms the ground truth. Figure 2 shows a single B-scan from a patient with an intraretinal cyst secondary to vitreoretinal traction and accompanying epiretinal membrane. Ground truth may be provided in terms of a classification, e.g., "disease," or as an annotation such as the red shading, depending on the type of AI model being developed.

Supervised models require annotation of features to provide an initial example of how the output should appear. In practice, this requires each image to be annotated, a laborious task, then the model typically uses regression-style algorithms to determine outputs. Unsupervised models do not require annotation, but they do require ground truth in the form of a classification. These models search for natural patterns in the data and use clustering style algorithms to generate the output. Reinforcement models can work with either annotated or unannotated data and are more complex than the former models, in that they interact with the environment and explore the data autonomously. The algorithm learns through a process of reward reinforcement for a correct response. Reinforced learning is a deep learning algorithm.

It is important to note that models usually work well only when the images or data are in the same form as those used for training. An AI model trained to detect diabetic retinopathy will ignore a choroidal melanoma, glaucoma, or any other pathology in the field, no matter how obvious it may be to humans. Models trained on emmetropes may fail in patients who are high myopes, and AI models trained on one manufacturer's device will not work reliably on images generated by another manufacturer's device.

As AI technology improves, there are moves to generate models that are trained on multiple forms of a disease or, indeed, multiple diseases. As the complexity of a model increases, however, so too does the computing processing overhead. As a result, these complex models often utilize a cloud-based server rather than local machine capacity, thus depending on reliable and high bandwidth internet connections to comply with cybersecurity requirements while maintaining patient privacy and confidentiality. Moreover, even though AI models can theoretically be continuously refined and features augmented, registered medical devices cannot change in their function (indication of use). Therefore, each version of the model requires regulatory approval to ensure that changes to the model maintain patient safety.

**Forms of AI Output**

Importantly, the type of model used affects the output provided to the end user. In practice, there are three major output forms: classification, object detection, and segmentation. Figure 3 shows these outputs graphically using the same OCT B-scan used in Figure 2.

Clinicians will immediately appreciate the usefulness of the segmented output because it enables them to cross-reference their own clinical judgment against the AI output and then make a final evaluative judgment. For example, many AI models may classify the small area of hyporeflectance just beneath the epiretinal membrane as an artifact caused by distortion of the retinal surface under the epiretinal membrane rather than a true cyst. Classification data are useful in flagging which B-scans are abnormal to enable clinicians to preferentially assess scans with potential pathological features. Non-clinically trained staff also may benefit from a classification-style output indicating that disease may be present and flagging a need for the patient to be examined by a clinician.

**Clinical Decision-Making Can Be Enhanced by AI Tools**

There is little doubt that AI image analysis is beneficial in eyecare delivery. In fact, AI data underpins many of the basic OCT analysis tools that optometrists and ophthalmologists take for granted. Retinal layer segmentation is achieved by an AI model in OCT, and, therefore, all layer thickness, TSNIT plots, macular GCC values, and RPE deviation models depend on the accuracy of this segmentation, as all analytics are derived from these assumptions/measurements. As such, clinicians are trained to sweep through the B-scans with layer visibility toggled on to ensure segmentation is accurate. As AI models become more advanced, it will be important for clinicians to continue to verify the clinical data themselves and to cross-reference their own clinical judgment with the information provided by the AI model. Outputs that flag particular B-scans or otherwise highlight areas that appear abnormal will increase efficiency while ensuring that human clinical judgment drives structured diagnostic reasoning. There is some concern that the rapid expansion of AI-assisted diagnostic tools may reduce the need for clinicians. After all, AI diagnosis of diabetic retinopathy is equal to or better than that achieved by humans[8] and AI can determine the sex of a patient from fundus appearance alone.[9] With the correct level of integration and training, however, these technologies may, in fact, enable us to increase our efficiencies.

At present, a large population of patients across the globe do not have access to trained eye health practitioners. In these regions, AI could be employed in screening programs for specific eye diseases, such as diabetic retinopathy, age-related macular degeneration, or glaucoma. Each revolution in eye care has enabled better patient care. With OCT, clinicians can view the retina in exquisite detail. OCT angiography reveals the structure of microvasculature, and widefield imaging and fundus autofluorescence enable clinicians to photo-document peripheral lesions and assess RPE function. All of these tools offer data that improve clinical reasoning capacity. Where access to well-trained eye health practitioners is possible, AI-assisted image analysis will not replace the need for human healthcare practitioners. Instead, it is a tool that will enable us to make more efficient clinical decisions based on high-quality data and to apply this data to drive better patient-centered care.

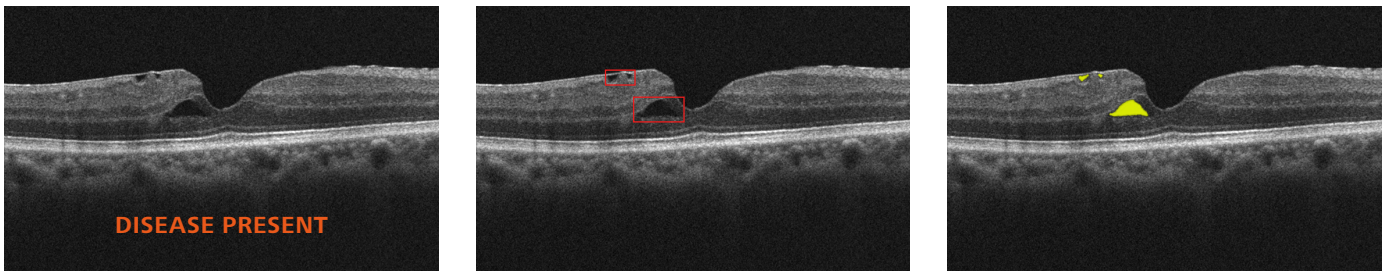


*Figure 3. Exemplar outputs for the OCT B-scan shown in Figure 2. The left panel shows a classification output indicating that disease is present, with no further information as to which aspects of the image have contributed to the decision. The middle panel shows an object detection output with a bounding box around the pathology. The right-hand panel depicts a segmentation output where the pathology is shaded yellow. Note that the area just below the epiretinal membrane may or may not be a true cyst, thus clinical judgment is required.*

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