

Redefining healthcare through the eyes

BY ASHKAN ELIASY

he future of healthcare is being shaped by innovation in eyecare and in particular a field known as oculomics. This discipline leverages ocular biomarkers to provide insights into various health conditions, including cardiovascular diseases and psychological or neurological disorders [1]. Today, oculomics data are used to train artificial intelligence (AI) models to increase accessibility, accuracy and affordability of healthcare. For instance, the retina offers a unique non-invasive window into the body's vascular and neurological health. According to a recent review study, retinal scans can predict cardiovascular events with a minimum accuracy of 70%, showcasing the potential of ocular biomarkers in preventive healthcare [2]. As an ophthalmologist, optometrist, or primary care provider, understanding and integrating oculomics and related tools into your practice can significantly enhance patient care and open new avenues for early detection and personalised treatment.

Early detection and diagnostics

Early detection is a cornerstone of oculomics, providing a pathway for timely interventions that can save lives and reduce healthcare costs. Retinal imaging enables the early detection of systemic diseases such as diabetes, hypertension or cardiovascular conditions, and even kidney and neurodegenerative diseases [3–6]. This proactive approach allows for timely intervention, significantly increasing quality of life and reducing treatment costs by reducing the need for more complex or invasive procedures. The economic benefit of early detection is underscored by the projected growth of the digital health market, expected to increase from \$452 billion in 2023 to \$1.9 trillion by 2030 [7].

Today, significant challenges remain. For instance, studies have shown that optometrists can miss or misdiagnose retinal diseases in up to 39% of instances [8]. Primary care providers often lack adequate knowledge or confidence to effectively deal with eye problems, and survey results showed only 40% could confidently distinguish an abnormal optic disc [9]. Furthermore, ophthalmologists face challenges in detecting neurodegenerative diseases and age-related macular degeneration (AMD) in up to 25% of patients [10]. In all cases, diagnosis is often done when symptoms are notable, which is often too late. However, with the use of Al to interpret retinal images, a condition like AMD could not only be screened by all clinicians, but these systems could also predict which patients are likely to progress [11]. This capability not only facilitates earlier detection but also enables intervention at a stage when the disease is more manageable, potentially preventing severe complications such as vision loss.

Personalised and preventive healthcare

Oculomics plays a significant role in personalised and preventive healthcare. By analysing ocular biomarkers using advanced technologies such as AI, treatments can be tailored to individual patients based on their unique genetic and phenotypic profiles. Studies have shown that specific biomarkers in the retina can detect genetic mutations associated with AMD and other retinal diseases. For instance, the presence of certain angiogenic and stress-related factors in the vitreous fluid can indicate early stages of retinal degeneration, enabling early intervention and personalised treatment plans tailored to the patient's specific condition [12].

The personalised medicine market is projected to grow at a compound annual growth rate of 8.2%, valued at \$529 billion in 2023, driven by the potential to analyse vast data through AI, streamline drug development processes and reduce clinical trial costs [13]. This growth underscores the increasing importance of personalised approaches in managing complex diseases.

Regular ocular assessments contribute to preventive healthcare by identifying risk factors early, leading to lifestyle modifications and preventive measures. The economic benefit of preventive healthcare is significant, with potential long-term cost savings through the prevention of disease progression. For instance, detecting biomarkers of neurodegeneration or chronic inflammation in retinal diseases allows for timely therapeutic interventions, which can delay or prevent the onset of severe symptoms and complications [12]. Incorporating these advanced diagnostic tools into routine practice can help clinicians optimise patient outcomes by offering tailored, proactive healthcare solutions that address the unique needs of each individual, ultimately leading to better management of diseases and improved overall health.

Neurological and systemic health monitoring

The eye serves as a vital indicator for both neurodegenerative and systemic diseases, offering a unique window into a patient's

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overall health. Advanced ocular imaging techniques such as optical coherence tomography (OCT) and fundus imaging have been instrumental in diagnosing and monitoring various conditions:

Neurodegenerative diseases

Optical coherence tomography has shown promise in assessing disease severity and monitoring progression in multiple sclerosis by measuring retinal nerve fibre layer thickness, which correlates with disease activity [14]. Despite challenges in differentiating multiple sclerosis from other neurological conditions, OCT holds a significant prognostic value. For Alzheimer's disease, both OCT and fundus imaging have identified retinal biomarkers like changes in retinal thickness and amyloid plagues [15-17]. Fundus imaging, enhanced by AI, shows potential in assessing neurologic dysfunction in Parkinson's disease through the analysis of subtle changes in the retinal vasculature [18].

Systemic diseases

Ocular imaging techniques are equally valuable in diagnosing and monitoring systemic diseases. Diabetic retinopathy, a common complication of diabetes, can be detected early through retinal imaging, with Al-enhanced scans demonstrating over 90% accuracy [19]. Additionally, retinal imaging has been used to identify changes indicative of various conditions such as heart disease, kidney disease, autism, cardiovascular and cerebrovascular diseases, infectious disease, leukaemia and metabolic syndrome [20–27].

Data collection techniques

Advanced imaging and molecular techniques provide deeper insights that will further fuel oculomics, and consequently diagnosis and monitoring of neurodegenerative and systemic diseases through the eye. These methods provide detailed insights into retinal and choroidal structures, as well as the biochemical and genetic underpinnings of ocular health, enhancing the ability of clinicians to deliver personalised and effective treatments. Here are some examples:

- Fluorescein angiography: Highlights retinal blood vessels to diagnose diabetic retinopathy and AMD by detecting microaneurysms and non-perfusion areas.
- Indocyanine green angiography: Visualises the choroidal vasculature, useful for conditions like polypoidal choroidal vasculopathy.
- Electroretinography: Measures electrical responses in the retina to diagnose and monitor inherited retinal disorders such as retinitis pigmentosa.

- Adaptive optics: Enhances retinal image resolution, allowing for the visualisation of individual photoreceptor cells, primarily used in research.
- Hyperspectral imaging: Captures detailed spectral information from the retina to identify biochemical changes associated with diseases like diabetic retinopathy and AMD.
- Ocular coherence elastography: Measures mechanical properties of ocular tissues, aiding in the study of diseases like glaucoma and keratoconus.
- Proteomic and metabolomic analysis: Analyses ocular fluids to identify biomarkers for conditions such as dry eye disease, diabetic retinopathy, and neurodegenerative diseases.
- Genetic testing: Diagnoses inherited retinal disorders, enabling personalised treatment approaches based on the patient's genetic profile.

Potential integration of oculomics

Integrating oculomics into various healthcare platforms can significantly enhance diagnostic accuracy, personalised treatment, and accessibility to care. These technologies offer innovative solutions for both clinicians and patients, paving the way for a more comprehensive approach to health monitoring and disease management.

Telemedicine

Oculomics can be seamlessly integrated into telemedicine platforms to enhance remote healthcare delivery. This integration allows clinicians to conduct comprehensive eye exams and monitor ocular biomarkers from afar, making it easier to provide timely interventions for patients in remote or underserved areas. Teleophthalmology can include Al-assisted retinal image analysis or video games to screen for diseases like diabetic retinopathy and AMD, improving access to specialised care and monitoring treatment effectiveness.

Electronic health records (EHR)

Incorporating oculomics data into EHR can provide a more holistic view of a patient's health. By integrating imaging data and biomarkers into EHRs, with the help of clinical platforms, clinicians can track changes over time and correlate ocular health with systemic conditions. This can improve the accuracy of diagnoses and the effectiveness of treatment plans, leading to better patient outcomes.

Personalised medicine

Oculomics supports the advancement of personalised medicine by using ocular biomarkers to tailor treatments based on individual genetic and phenotypic profiles. Integration with genomic data and AI can help identify patients at risk for specific diseases and customise prevention and treatment strategies. This approach enhances the efficacy of interventions and minimises adverse effects.

AI and machine learning

Artificial intelligence or machine learning algorithms can analyse vast amounts of oculomics data to identify patterns and predict disease progression. These technologies can be integrated into diagnostic tools and imaging devices to provide real-time analysis and decision support for clinicians. Al-driven insights can lead to earlier detection of diseases like Alzheimer's, Parkinson's, and diabetes, improving patient prognosis, as discussed before.

Wearable health devices

Wearable devices, such as smart VR glasses, smart contact lenses and glasses, equipped with sensors to monitor ocular health, can or will continuously collect data on eye movements, intraocular pressure, glucose levels, and other biomarkers. Integration with mobile health apps allows patients and clinicians to track ocular health metrics in real time, facilitating early detection of anomalies and prompt medical attention.

Clinical decision support systems (CDSS)

Integrating oculomics into CDSSs can aid clinicians in making more informed decisions. Clinical decision support systems can provide alerts and recommendations based on oculomics data, helping to identify potential issues early and suggesting appropriate diagnostic tests or treatments without requiring complex or lengthy training or expertise. This can enhance clinical workflows and improve patient management.

⁶⁶ By understanding and utilising ocular biomarkers or tools that utilise them, you can play a crucial role in advancing the field of oculomics and shaping the future of healthcare ⁹⁹

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Research and development

Oculomics can be integrated into research platforms to facilitate the study of ocular and systemic diseases. By combining data from EHR, ocular imaging, genetic testing, and biomarker analysis, researchers can gain deeper insights into disease mechanisms and develop innovative therapies. Collaborative databases that pool oculomics data from various sources can accelerate discovery and translational research.

Public health and screening programmes

Oculomics technologies can be integrated into public health initiatives and screening programmes to identify at-risk populations and implement preventive measures. Large-scale retinal screening programs using portable imaging devices and AI analysis can detect early signs of systemic diseases, enabling community-wide health interventions and reducing the burden on healthcare systems.

Regenerative medicine and research

Regenerative medicine, combined with oculomics, offers potential in ophthalmology by leveraging advanced techniques such as stem cell therapy, gene therapy, and tissue engineering to more effectively repair and restore damaged ocular tissues. By analysing ocular biomarkers, clinicians can personalise these regenerative treatments to target specific genetic and phenotypic profiles, enhancing their effectiveness and minimising side-effects. This integration provides innovative therapeutic options for conditions like AMD and retinitis pigmentosa, significantly advancing our understanding and management of eye health.

Conclusion

Oculomics represents a transformative shift in healthcare, offering unprecedented opportunities for early disease detection, personalised treatment, and improved patient outcomes. As clinicians, embracing these technologies will not only enhance your practice but also contribute to the broader goal of a healthier, more equitable future. By understanding and utilising ocular biomarkers or tools that utilise them, you can play a crucial role in advancing the field of oculomics and shaping the future of healthcare.

The integration of oculomics into routine clinical practice, supported by robust technological innovations and a commitment to equitable access, promises a future where quality healthcare is more accessible, proactive, personalised, and effective. As the digital health market continues to grow and evolve, the role of ocular biomarkers in early detection, personalised medicine, and holistic healthcare will become increasingly prominent, offering a brighter future for patients and clinicians alike.

References

- Ghadiri N. Al & Oculomics: a new section for Eye News. Eye News 2024;30(5):58–9.
- Hu W, Yii FSL, Chen R, et al., A systematic review and meta-analysis of applying deep learning in the prediction of the risk of cardiovascular diseases from retinal images. *Transl Vis Sci Technol* 2023;**12(7)**:14.
- Ragab M, Al-Ghamdi ASA, Fakieh B, et al., Prediction of diabetes through retinal images using deep neural network. *Comput Intell Neurosci* 2022;**2022**:7887908.
- Arnould L, Meriaudeau F, Guenancia C, et al., Using artificial intelligence to analyse the retinal vascular network: the future of cardiovascular risk assessment based on oculomics? A narrative review. Ophthalmol Ther 2023;12(2):657–74.
- Wen J, Liu D, Wu Q, et al., Retinal image-based artificial intelligence in detecting and predicting kidney diseases: Current advances and future perspectives. *View* 2023;4(3):20220070.
- Bahr T, Vu TA, Tuttle JJ, lezzi R. Deep Learning and Machine Learning Algorithms for Retinal Image Analysis in Neurodegenerative Disease: Systematic Review of Datasets and Models. *Transl Vis Sci Technol* 2024;**13(2)**:16.
- Digital Health Market Size [2024]. Fortune Business Insights. https://www. fortunebusinessinsights.com/industry-reports/ digital-health-market-100227
- Ho S, Kalloniatis M, Ly A. Clinical decision support in primary care for better diagnosis and management of retinal disease. *Clin Exp Optom* 2022;**105(6)**:562–72.
- Gibson C, Roche E. A survey of general practitioners' knowledge and perceived confidence with clinical ophthalmology. *Ir Med J* 2014;**107(6)**:173–5.
- Neely DC, Bray KJ, Huisingh CE, et al., Prevalence of undiagnosed age-related macular degeneration in primary eye care. JAMA Ophthalmol 2017;135(6):570–5.
- 11. Chew E. Al-based systems can help identify rapidly advancing age-related macular degeneration [2023]. *National Eye Institute*. https://www.nei.nih.gov/about/news-and-events/ news/ai-based-systems-can-help-identify-rapidlyadvancing-age-related-macular-degeneration
- Micera A, Balzamino BO, Di Zazzo A, et al., Biomarkers of neurodegeneration and precision therapy in retinal disease. *Front Pharmacol* 2021;11:601647.
- 13. Personalized Medicine Market Size, Share & Trends Analysis Report By Product (Personalized Medicine Diagnostics, Personalized Medicine Therapeutics), By End-use, By Region, And Segment Forecasts, 2024 – 2030 [2023]. Grand View Research. https://www.grandviewresearch. com/industry-analysis/personalized-medicinemarket
- Swinnen S, De Wit D, Cleemput LV, et al., Optical coherence tomography as a prognostic tool for disability progression in MS: a systematic review. *J Neurol* 2023;**270(2**):1178–86.
- Sánchez-Puebla L, López-Cuenca I, Salobrar-García E, et al., OCT Imaging in Murine Models of Alzheimer's Disease in a Systematic Review: Findings, Methodology and Future Perspectives. *Biomedicines* 2024;**12(3)**:528.
- Yuan A, Lee CS. Retinal biomarkers for Alzheimer disease: the facts and the future. Asia Pac J Ophthalmol (Phila) 2022;11(2):140–8.

- Ma X, Wang X, Xiao Y, Zhao Q. Retinal examination modalities in the early detection of Alzheimer's disease: Seeing brain through the eye. *J Transl Int Med* 2022;**10(3)**:185–7.
- Zhang Y, Zhang X, Yue Y, Tian T. Retinal degeneration: a window to understand the origin and progression of Parkinson's disease? *Front Neurosci* 2022;**15**:799526.
- Grauslund J. Diabetic retinopathy screening in the emerging era of artificial intelligence. *Diabetologia* 2022;65(9):1415–23.
- Gupta K, Reddy S. Heart, eye, and artificial intelligence: A review. Cardiol Res 2021;12(3):132.
- Mitani A, Hammel N, Liu Y. Detecting Chronic Kidney Disease and Diabetes from Retinal Fundus Photographs. *Nat Biomed Eng* 2021;5:487–9.
- Bağcı KA, Çöp E, Memiş PN, Işık FD. Investigation of retinal layers thicknesses in autism spectrum disorder and comparison with healthy siblings and control group. *Res Autis Spec Disor* 2023;108:102242.
- 23. Abdollahi M, Jafarizadeh A, Ghafouri-Asbagh A, et al., Artificial Intelligence in Assessing Cardiovascular Diseases and Risk Factors via Retinal Fundus Images: A Review of the Last Decade. *WIREs* 2024;**14(6)**:e1560.
- Rim TH, Teo AWJ, Yang HHS, et al., Retinal vascular signs and cerebrovascular diseases. *J Neuroophthalmol* 2020;40(1):44–59.
- 25. Agrawal R, Weng RC, Fonollosa A, et al., Outcome measures for disease monitoring in intraocular inflammatory and infectious diseases (OCTOMERIA): understanding the choroid in uveitis with optical coherence tomography (OCT). Ocul Immunol Inflamm 2023;**31(2)**:374–92.
- Beketova T, Mordechaev E, Murillo B, Schlesinger MD, et al., Leukemic Retinopathy: A Diagnostic Clue for Initial Detection and Prognosis of Leukemia. *Cureus* 2023;**15(12)**:e50587.
- Baek J, Jung Y, Ohn K, et al., Association between localized retinal nerve fiber layer defects in nonglaucomatous eyes and metabolic syndrome: a propensity score-matched analysis. *Ann Transl* Med 2023;11(9):316.
- [All links last accessed December 2024]

AUTHOR



Ashkan Eliasy,

Investment Principal, Topcon Healthcare Inc., San Diego, USA.

SECTION EDITORS

Nima Ghadiri,



Medical Ophthalmology Consultant and Honorary Senior Clinical Lecturer, Liverpool, UK. nima.ghadiri@liverpoolft.nhs.uk

Arun James Thirunavukarasu,



Academic Foundation Doctor, Oxford University Hospitals NHS Foundation Trust; Clinical Research Fellow, Nuffield Department of Clinical

Neurosciences & Big Data Institute, University of Oxford; Rising Leader Fellow, Aspen Institute, UK.

ajt205@cantab.ac.uk

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