What is personalised healthcare in ophthalmology?

Personalised healthcare is an approach that is centred on an individual's health needs - taking into account genetic information as well as personality, lifestyle and environment, and their rehabilitation, socialisation and mobility needs.[1] The aim of personalised healthcare in ophthalmology is to find prevention and treatment strategies tailored to each individual, successfully treating specific eye diseases, and preventing vision loss.[2]

Advances in diagnostic tools and technologies have transformed ophthalmological care from a traditional approach to a precision approach, allowing for more personalised treatments in the future:[1]

Traditional healthcare approach

Treatment limited to surgical procedures (e.g. laser photocoagulation, a type of laser surgery for the eyes) which often has poor visual outcomes and high safety risks.[3]

Precision healthcare approach

Using cutting edge technology (e.g. artificial intelligence and genetic 'biomarker' testing) to more accurately diagnose eye disease and tailor care with targeted treatments (e.g. Anti–vascular endothelial growth factor (anti-VEGF) therapy used for wet age-related macular degeneration (AMD), which have helped to preserve vision which, in the past, may have been lost completely).[4–6]

Personalised healthcare approach

Tailoring care for every person's unique eye disease, taking into account the genetic information of each patient and their condition, as well as the person's lifestyle and environment.[6–8]

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Through what methods could personalised healthcare be achieved in ophthalmology?

Artificial Intelligence (AI)

AI, based on deep learning, is a broad area of science which aims to develop computers and machines that have the ability to use information in the same way that people do.[9]

It is expected to have a dramatic impact on medicine, by improving our ability to diagnose disease and select the most suitable treatment for individual patients.

It allows large amounts of data to be analysed very quickly, empowering healthcare professionals to make more effective, and efficient, treatment decisions.[10]

While the widespread use of AI in ophthalmology is still something for the future, one potential application is to more accurately predict the development of wet-AMD, allowing doctors to diagnose and start treatment earlier.[11]

Genetic Information (Biomarkers)

Biomarkers are substances found in cells (e.g. genes) that provide important information about a specific eye disease.[12,13]

By using detailed eye images and gathering genetic information, it is possible to carry out ‘biomarker testing’.

This could allow us to analyse the genotypes of patients with inherited retinal diseases (for example, for people whose disease runs in their family), making it possible to track their retinal condition with the aim of leading to an earlier, more accurate diagnosis of disease.[14]

In addition, analysis of biomarkers could allow us to predict disease progression and the effectiveness of a specific treatment.[15,16]

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Digital Health Tools

As digital technology continues to develop, it could become increasingly possible for patients to monitor their retinal conditions from home, as well as track lifestyle information that affects vision like quality of sleep, mood and activity levels.[17]

Technological solutions, such as virtual appointments and consultations, could improve access to eye care services – potentially reducing the burden on people living with retinal conditions who may not easily be able to attend face-to-face consultations.[18,19]

Research and Development

Research and development is also fundamental to the development of personalised healthcare, this includes:

- Collecting large amounts of datasets from clinical trials and real-world settings (this, in turn, could ensure more effective clinical trials going forward).[20]
- Ensuring valuable research is put into all forms of retinal disease, including why a certain disease develops and what causes it.[21]
- Development of improved imaging and genomic technologies.[22]
- Biomarker testing for patient selection and stratification.[23]

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What are the potential benefits of personalised healthcare in ophthalmology?

Improved health outcomes due to more accurate and more personalised detection, diagnosis and treatment of eye disease[24]

Empowered patients and families through managing potential health risks and disease knowledge[24]

Improved quality of life for people with eye disease and their families[24]

Potential lower financial impact[25]

Promoting health and wellbeing of society and better use of healthcare resources[24]

The ability to track retinal degeneration[20]

The potential to capture important information earlier, which could prevent disease progression[24]

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Conclusion

The future of personalised healthcare care in ophthalmology certainly looks bright – but work still needs to be done to implement it. **Partnerships**, **collaborations** and **integrated solutions** always play an important role in drug discovery and development – and this certainly rings true within personalised healthcare and ophthalmology.

Many patients are now able to access some form of genomic testing.[26,27] For more information on genomic testing and personalised healthcare tools that might be available to you, please speak with your healthcare team.[28]

Glossary

1. Age-related macular degeneration - An eye disease that can blur central vision. It happens when aging causes damage to the macula, and comes in both dry and wet forms.[28]
2. Anti-VEGF treatment - Anti-VEGF treatments are a group of medicines which reduce new blood vessel growth. They can be used to treat a number of eye conditions that cause new blood vessel growth or swelling under the macular area of the retina.[29]
3. Artificial Intelligence - The ability of computers and machines to use information as people do. Computers and machines can process information to identify patterns, solve problems and answer questions. Artificial intelligence can analyse and interpret large amounts of data. It could help implement personalised healthcare in practice.[6]
4. Biomarker - A substance found in the genetic information of cells (e.g. genes) that provides important information about an eye disease. Biomarkers might be able to detect early damage to the eye.[12,13] New biomarkers in the eye are being discovered.[15]
5. Diabetic macular oedema - An eye disease that can blur and distort central vision. It happens when diabetes damages blood vessels in the eye, causing fluid to leak and build up in the macula.[29]
6. Retina - A thin layer of tissue at the back of the eye that senses light and sends signals to the brain to allow a person to see. It contains many layers.[30]

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References

1. Porter LF, Black GC. Personalized Ophthalmology. Clinical Genetics. 2014; 86(1):1-11.
2. Genetics Home Reference. What is the difference between precision medicine and personalized medicine? What about pharmacogenomics?
3. Daien V. Evolution of treatment paradigms in neovascular age-related macular degeneration: a review of real-world evidence. British Journal of Ophthalmology, 2021;105:1435-1479.
4. Li JO et al. Digital technology, tele-medicine and artificial intelligence in ophthalmology: A global perspective. Prog Retin Eye Res. 2021; 82:100900.
5. Takamura Y et al. New Strategies for Treatment of Diabetic Macular Edema. Journal of Ophthalmology, Volume 2018.
6. Genentech. Seeing the unseen with Artificial Intelligence.
7. SAS Analytics Software. Artificial Intelligence: What is it and why it matters.
8. Fernández-Domínguez AI et al. Plasmon-Enhanced Generation of Nonclassical Light, ACS Photonics, 2018; 5(9):3447-3451.
9. Panch T et al. Artificial intelligence, machine learning and health systems. Journal of Global Health. 2018; 8(2):1-8.
10. Ting DSW et al. Artificial intelligence and deep learning in ophthalmology. British Journal of Ophthalmology. 2018; 103(2):167-175.
11. Johnson KB, et al. Precision Medicine, AI, and the Future of Personalized Health Care. Clinical and Translational Science. 2021; 14(1):86-93.
12. Yim J, et al. Predicting conversion to wet age-related macular degeneration using deep learning. Nat Med. 2020; 26(6):892-899.
13. Lauwen S et al. Omics Biomarkers in Ophthalmology. Investigative Ophthalmology & Visual Science. 2017; 58(6): BIO88–BIO98.
14. National Cancer Institute. Dictionary of Cancer Terms – Molecular marker.
15. Ellingford JM, Hufnagel RB, Arno G. Phenotype and Genotype Correlations in Inherited Retinal Diseases: Population-Guided Variant Interpretation, Variable Expressivity and Incomplete Penetrance. Genes (Basel). 2020 Oct 29;11(11):1274.
16. Science Daily. New biomarkers may detect early eye changes that can lead to diabetes-related blindness.
17. Li JO, et al. Digital technology, tele-medicine and artificial intelligence in ophthalmology: A global perspective. Prog Retin Eye Res. 2021 May;82:100900. doi: 10.1016/j.preteyeres.2020.100900. Epub 2020 Sep 6. PMID: 32898686; PMCID: PMC7474840.
18. Retina Today. Putting vision monitoring in the hands of patients with AMD.
19. Steinhubl SR, et al. The Emerging Field of mobile health. Sci Transl Med. 2015; 7:283: 1-12.
20. Hopkins JJ, Keane PA, Balaskas K. Delivering personalized medicine in retinal care: from artificial intelligence algorithms to clinical application. Curr Opin Ophthalmol. 2020 Sep;31(5):329-336.
21. Cursiefen C, et al (EVI Steering Board). Unmet Needs in Ophthalmology: A European Vision Institute-Consensus Roadmap 2019-2025. Ophthalmic Res. 2019; 62(3):123-133.
22. Cirillo D, Valencia A. Big data analytics for personalized medicine. Current Opinion in Biotechnology. 2019; 58:161–167.
23. Villani E, et al. Decade-Long Profile of Imaging Biomarker Use in Ophthalmic Clinical Trials. Invest Ophthalmol Vis Sci. 2017; 58(6):BIO76-BIO81.
24. Charles River Associates. The benefits of personalised medicine.
25. Vellekoop H. et al. Guidance for the Harmonisation and Improvement of Economic Evaluations of Personalised Medicine. PharmacoEconomics. 2021; 39: 771–778.
26. Roche. An introduction to personalised healthcare for the eye. [Internet; cited November 2021].
27. Black GC, et al (ERN-EYE study group). The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet J Rare Dis. 2021; 16(1):142.
28. National Eye Institute. Age-Related Macular Degeneration. [Internet; cited June 2021].
29. National Eye Institute. Macular Edema. [Internet; cited August 2022].
30. All About Vision. The Retina: Where Vision Begins. [Internet; cited December 2021].

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