



Personalised healthcare in ophthalmology

Toolkit for patient leaders Core slide deck





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About this document

Why?	The presentation aims to demystify the topic of personalised healthcare in ophthalmology, by proposing a shared definition and exploring the value that personalised healthcare strategies can bring to people living with eye disease, their caregivers and broader healthcare systems
Who?	<p>The content is aimed at patient advocacy groups and patient leaders to help:</p> <ul style="list-style-type: none">• Introduce people living with eye disease to the concept of personalised healthcare• Inform and guide patient groups around advocacy and policy initiatives/activities related to personalised healthcare in ophthalmology
What?	The content provides a background on personalised healthcare in ophthalmology and the impact that personalised healthcare strategies can have
How?	This document can be downloaded, read and presented to, or shared with, audiences at meetings, individual briefings or used as part of discussions



Understanding personalised healthcare in ophthalmology





Key terms and definitions

Key term	Definition
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. ¹ <i>Also known as AMD.</i>
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 ² .
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. ³ It could help implement personalised healthcare in practice. ^{4,5} <i>Also known as AI and machine learning.</i>
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. ^{6,7} New biomarkers in the eye are being discovered. ⁸ <i>Known as genetic information and molecular marker.</i>
Colour fundus photography	A non-invasive imaging test that takes a series of coloured images of the inner surface of the eye. The results can help guide diagnosis and monitor the progress of eye disease. ⁹ <i>Also known as CFP.</i>
Deep learning	A category of AI in which machines are developed to learn from experience, like people do. The machine uses algorithms to detect patterns and relationships in large datasets, training itself each to improve outcomes. ¹⁰ <i>Also known as DL.</i>



Key terms and definitions

Key term	Definition
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. ¹¹ <i>Also known as DME or ‘diabetes-related macular oedema’, as preferred by the patient community.</i>
Disease stratification	The process of categorising people into sub-groups based on the mechanisms of their disease or how they respond to treatment. ¹²
Genomic testing	A test that looks at all the genes that make up a person (known as the genome) from a sample of a person’s saliva, cells or blood. It can guide diagnosis, screening and decisions about treatment of eye diseases. ^{13–15} <i>Also called biomarker testing, genomic profiling, molecular testing, somatic testing.</i>
Macula	The part of the eye that controls sharp, straight-ahead vision. ¹⁶ The thickness of this region (macular thickness) can help diagnose and monitor some eye diseases. ^{17,18}
Optical coherence tomography	A non-invasive imaging test that uses light to take detailed images of each layer in a retina. The results can help guide diagnosis of eye diseases and decisions about treatment. ¹⁹ <i>Also known as OCT.</i>
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. ²⁰

What is personalised healthcare in ophthalmology?



*An approach based on a person's **unique health needs**, including the genetic information of their eye disease, as well as their lifestyle and environment²¹*

*An **evolving field**, gradually moving away from a traditional approach to eye disease care toward a targeted, tailored approach²¹*

An approach which responds to an individual's rehabilitation, socialisation and mobility needs²¹

*Its aim is to find **prevention** and **treatment** strategies tailored to each individual patient, to successfully **treat their specific eye disease** and **prevent vision loss**²²*

Applications of personalised healthcare in ophthalmology



Identify people at risk of eye disease earlier, (such as those with the potential to develop inherited retinal diseases), and tracking retinal degeneration to prevent further vision loss^{21,23,24}

Eye disease screening & prevention

Adopt digital tools to monitor symptoms remotely, provide regular check-ups and access to care through virtual appointments and develop our understanding of how different eye diseases are progressing³⁰

Digital health tools

Detecting & diagnosing eye disease

Earlier and more accurate detection and diagnosis of eye disease to ensure prevention of further, treatable vision loss^{25,26}

Personalised treatments & care

Help select the most effective treatment options and regimens for better outcomes and experiences, developed from an understanding of genetic information²⁷

Research & development

Collect and analyse large quantities of patient data to help accelerate research and development of new solutions and treatments for eye disease, improve the quality of clinical trials and ensure necessary research is being conducted in rare diseases to help transform patients' lives^{28,29}





How can personalised healthcare transform ophthalmology?



Technology advances are paving the way for personalised healthcare



Advances in diagnostic tools and technologies have transformed ophthalmological care from a traditional approach to a precision approach²¹ – this marks an important step towards personalised healthcare, but there is still a way to go...

Past: Traditional approach

Treatment limited to surgical procedures (e.g. laser photocoagulation), which often has poor visual outcomes and high safety risks.^{21,31} In addition, limited access to digital health tools and slow adoption of solutions for those without access to an optometrist.^{32,33}

Present: Precision approach

Using cutting edge technology (e.g. AI and genetic 'biomarker' testing) to more accurately diagnose eye disease and tailor care with targeted treatments (e.g. anti-VEGF treatments used for wet-AMD, which have helped to preserve vision which, in the past, may have been lost completely).^{30,34,35}

Future: Personalised approach

Tailoring care for every person's unique eye disease, taking into account the genetic information of the condition, and the person's lifestyle and environment – providing '**the right treatment to the right person at the right time**'.^{35–37} In addition, providing access to ophthalmological care through new digital health tools.^{32,33}

Advances in eye imaging, AI and digital technologies

The main pillars for achieving personalised healthcare in ophthalmology



Artificial Intelligence (AI) tools could help diagnose eye diseases, predict how eye disease might progress, and predict treatment responses^{3,4,30}



Biomarker testing could utilise a patient's genetic information to help predict disease incidence, identify at risk individuals and understand disease mechanisms³⁸

Emerging **digital health tools** and efforts to enhance **research and development** could also help realise the potential of both AI and biomarker testing within ophthalmology²⁴

Understanding AI



Artificial Intelligence (AI):

- A broad area of science which aims to develop computers and machines that have the ability to **use information as people do** – to display intelligence³
- Expected to have a dramatic impact on medicine, by improving our ability to **diagnose disease** and select the **best treatments** for individual patients^{4,35,37,39}
- Allows large amounts of data to be analysed very quickly, empowering healthcare professionals to make more effective, and efficient, treatment decisions^{10,23,40}
- 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 earlier²⁴



Ophthalmology has been at the forefront of this revolution²³



AI: the driver of personalised healthcare in ophthalmology



One main category of AI that holds promise in personalising ophthalmology care is **deep learning**:

- Where machines **learn from experience**, like the human brain does^{4,10}
- Uses algorithms to detect patterns and relationships in large datasets, each time the machine trains itself to **improve outcomes**^{10,23}
- Can collect large amounts of retinal images, which can be used to **characterise changes** in the eye and distinguish between different diseases – to help doctors make better informed decisions about care^{4,10,23}

It is also expected that AI-assisted scientific discovery could improve the efficacy of clinical trials⁴⁰



Example: CFP and OCT imaging

- Deep learning has taught machines to estimate **macular thickness** from colour fundus photography (CFP) and optical coherence tomography (OCT) images⁴
- This can improve **accuracy of diagnosis**³⁷
- Images may also provide more clues to help predict how a person's eye disease might **progress** or **respond to treatment**⁴

*For AI to be effectively implemented in ophthalmology, it is vital that patient communities work with healthcare institutions to **built trust around the sharing of health data***³⁰

Biomarker testing for personalised healthcare in ophthalmology



Biomarkers are substances found in cells (e.g. genes) that provide **important information** about an eye disease^{6,7}



Researchers are continually identifying potential biomarkers for AMD and DME^{8,38}

Biomarker testing:



From eye images together with AI tools^{41,42}



Comprehensive genomic profiling tests on ocular fluid^{21,28,43,44}

Biomarkers can:^{21,38,42,43}

- more accurately diagnose disease stage
- predict disease progression
- predict response to treatment
- 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

Example: VEGF pathway biomarkers

- Vascular endothelial growth factor (VEGF) is a protein which influences the growth of blood vessels³⁸
- Sometimes cells produce too much VEGF, causing new blood vessels to grow in the eye – this can result in vision loss³⁸
- Treatments that block VEGF function (anti-VEGF treatments) are the gold standard for treating AMD and DME^{34,38}
- Testing for biomarkers that are involved in the VEGF pathway can help predict responses to VEGF treatment^{38,45}

Digital health tools to implement personalised healthcare



Digital health tools

- As digital technology progresses, it could become possible to **monitor eye conditions** from home, as well as **track lifestyle information** that affects vision like quality of sleep, mood and activity levels⁴⁶⁻⁴⁸
- This data, collected or analysed using AI, could help detect disease progression and help people receive treatment earlier²¹
- It could also reduce healthcare visits and travel costs, which in turn has a wider benefit by reducing the burden on hospitals²³



Example: AI-enabled telemedicine/virtual clinics

- Artificial intelligence (AI) could be used to detect a positive optical coherence tomography (OCT) remotely²³
- This could then trigger a direct appointment to a hospital-based injection clinic²³
- This potentially bypasses the need to visit a hospital, enabling early treatment²³

R&D: The wider application of personalised healthcare



Personalised healthcare doesn't just benefit individual patients

Personalised healthcare can help the wider ophthalmology community, by accelerating **research and development** (R&D), through:²⁵

Collecting **large amounts of datasets** from clinical trials and real-world settings^{23,49}



Development of improved **imaging and genomic technologies**^{25,49}



Biomarker testing for patient selection and stratification²⁵



Research into **rare diseases** and why disease develops²⁹



Which can result in:

A better understanding of eye disease

Providing a complete and detailed picture of every eye disease at different stages to allow research into more tailored treatments^{23,40,49,50}

More effective clinical trials

Improving patient selection methods which can lead to more effective results^{40,49}

More efficient clinical trials

Identifying specific patient populations for smaller, more streamlined trials, which can also reduce costs and high rates of failure^{40,49}

Collecting data from people living with eye disease is critical to achieve this²⁸



What is the value of personalised healthcare in ophthalmology?





Who is impacted by personalised healthcare?²⁵



*People at risk
of eye disease*



*People living
with eye disease*



*Loved
ones*



*Healthcare
professionals*



Researchers



Society

Everyone!

Potential benefits of personalised healthcare in ophthalmology



Improved **health outcomes** due to more accurate, earlier and more personalised detection, diagnosis and treatment of eye disease^{25,26}



Empowered patients and families through managing potential health risks and disease knowledge^{25,51}



Improved **quality of life** for people with eye disease and their families^{25,51}



Potential lower **financial impact** and reduced time off work²⁵



Promoting health and wellbeing of **society** and better use of healthcare resources²⁵



To **track retinal degeneration**, keeping closer track of inherited retinal diseases²³



To provide broader ophthalmological care which takes into account the **personality, lifestyle** and **environment** of each patient²¹





How does this affect us?






Current landscape for personalised healthcare in ophthalmology



In some areas of eye care, we are already witnessing positive change and adoption of more personalised approaches...

...But for many countries, putting personalised healthcare into practice will require a **fundamental change in how care is delivered and managed**

			
Policy	While genetic testing is not commonly practiced in ophthalmology, many EU nations have shown intent to move towards personalised treatments in this field ⁵²	The use of genetic testing is not currently recommended by the American Academy of Ophthalmology ⁵³	The current NICE guidelines do not refer to genetic testing for AMD – however, it is practiced for some other genetic eye diseases ⁵⁴
Science/R&D	Many EU countries are looking to establish dedicated centres in which genomic testing is carried out, while others have this practice in other therapeutic areas ^{52,55,56}	New research is being conducted within personalised medicine with the launch of the Precision Medicine Initiative in 2015 ⁵⁷	NHS launched the <u>Genomics Medicine Service</u> , which aims to offer genome sequencing as part of routine care ⁵⁵ Access to testing is also available through Genomic Laboratory Hubs ⁵⁵



Where do we go from here?

The future of personalised healthcare in ophthalmology looks bright but there is still work to be done to implement it – you can play a key role in helping achieving this goal

Challenges

Security and privacy concerns around data sharing among patients⁵⁸

Lack of awareness or knowledge around personalised healthcare among healthcare professionals (HCPs)⁵⁹

‘Untapped data’; there are large quantities of data available that could have a great impact on healthcare – but it exists outside of medical systems⁵⁶

More dedicated centres are needed to carry out genomic testing on a greater scale²¹

Call to action

Work together with healthcare institutions to gain greater levels of patient trust in the gathering and storing of genetic information

Inform HCPs on the benefits of personalised healthcare and new technologies and their applications; this will require the collective efforts of academic institutions, pharmaceutical companies and experts

Help patients and the public understand the importance of lifestyle, nutrition and environment data on health, and how tracking and collecting this data can benefit everyone

Encourage governments and policymakers to devote funding to the development of genomic testing facilities



Where do we go from here? (continued)

Challenges

There is always a potential barrier surrounding new technologies – it is important to verify that they are precise enough for general use, and HCPs are prepared to adopt them.⁶⁰

It is often difficult to provide ophthalmological care in rural areas, where infrastructure or access to technology is limited.³³

There are currently only treatments available for the wet-form of Age-Related Macular Degeneration, and so treatment does not occur early enough.⁶¹

There is a lack of research into rare retinal diseases.⁵⁷

Call to action

Work with HCPs to teach them about the benefits of new technologies and encourage adoption.

Ensure the adoption of digital health tools to provide more widespread access to ophthalmological care.

Continue research into new technologies that will help detect macular degeneration earlier in patients, with emphasis on genotypes within inherited retinal diseases.⁶²

Provide research opportunities for all retinal diseases.

Thank you



Appendix





Helpful resources

Here we provide a comprehensive list of supporting assets and additional resources that provide more information on personalised healthcare in cancer for patient leaders and the cancer community:

- [An introduction to Personalised Healthcare for the eye](#)
- [The value of new diagnostics and personalised medicine](#)
- [PHG Ophthalmology and genomics](#)
- [Harnessing research data for personalised healthcare](#)



References (1 of 6)

1. National Eye Institute. Age-Related Macular Degeneration. [Internet; cited December 2022]. Available from: <https://www.nei.nih.gov/learn-about-eye-health/eye-conditions-and-diseases/age-related-macular-degeneration>
2. RNIB. Anti-VEGF treatment. [Internet; cited December 2022]. Available from: <https://www.rnib.org.uk/eye-health/eye-conditions/anti-vegf-treatment>
3. SAS Analytics Software. Artificial Intelligence: What is it and why it matters. [Internet; cited December 2022]. Available from: https://www.sas.com/no_no/insights/analytics/what-is-artificial-intelligence.html
4. Daniel Shu Wei Ting et al. Artificial intelligence and deep learning in ophthalmology. British Journal of Ophthalmology, 2019, Volume 103, Issue 2, Pages 167–175. Available from: <https://bjo.bmj.com/content/103/2/167>
5. Kevin B Johnson et al. Precision Medicine, AI, and the Future of Personalized Health Care. Clinical and Translational Science, 2021, Volume 14, Issue 1, Pages 86–93. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7877825>
6. Susette Lauwen et al. Omics Biomarkers in Ophthalmology. Investigative Ophthalmology & Visual Science, 2017, Volume 58, Issue 6, Pages BIO88–BIO98. Available from: <https://iovs.arvojournals.org/article.aspx?articleid=2628930>
7. National Cancer Institute. Dictionary of Cancer Terms – Molecular marker. [Internet; cited December 2022]. Available from: <https://www.cancer.gov/publications/dictionaries/cancer-terms/def/molecular-marker>
8. Science Daily. New biomarkers may detect early eye changes that can lead to diabetes-related blindness. [Internet; cited December 2022]. Available from: <https://www.sciencedaily.com/releases/2021/08/210813105533.htm>
9. The University of British Columbia. Color Fundus Photography. [Internet; cited December 2022]. Available from: <https://ophthalmology.med.ubc.ca/patient-care/ophthalmic-photography/color-fundus-photography>
10. Forbes. What Is Deep Learning AI? A Simple Guide With 8 Practical Examples. [Internet; cited December 2022]. Available from: <https://www.forbes.com/sites/bernardmarr/2018/10/01/what-is-deep-learning-ai-a-simple-guide-with-8-practical-examples>
11. Macular Society. Macular Oedema. [Internet; cited December 2022] Available from: <https://www.macularsociety.org/macular-disease/macular-conditions/macular-oedema/>



References (2 of 6)

12. Ulster University. Stratified Medicine? [Internet; cited December 2022]. Available from: <https://www.ulster.ac.uk/doctorscollege/find-a-phd/research-areas/stratified-medicine>.
13. National Human Genome Research Institute. Genetics vs. Genomics Fact Sheet. [Internet; cited December 2022]. Available from: <https://www.genome.gov/about-genomics/fact-sheets/Genetics-vs-Genomics>
14. Genome Education Programme. Genomics in Ophthalmology. [Internet; cited December 2022]. Available from: <https://www.genomicseducation.hee.nhs.uk/genomics-in-healthcare/genomics-in-ophthalmology>
15. Kathryn P Burdon. The utility of genomic testing in the ophthalmology clinic: A review. Clinical & Experimental Ophthalmology, 2021, Volume 49, Issue 6, Pages 615–625. Available from: <https://onlinelibrary.wiley.com/doi/abs/10.1111/ceo.13970>
16. Medical Eye Center. The Macula of the Human Eye. [Internet; cited December 2022]. Available from: <https://www.medicaleyecenter.com/2018/03/18/macula-human-eye>
17. Divakar Gupta, Sanjay Asrani. Macular thickness analysis for glaucoma diagnosis and management. Taiwan Journal of Ophthalmology, 2016, Volume 6, Issue 1, Pages 3–7. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5602122>
18. Glaucoma Today. Macular Thickness Measurement for the Detection of Glaucomatous Progression. [Internet; cited December 2022]. Available from: <https://glaucomatoday.com/articles/2015-sept-oct/macular-thickness-measurement-for-the-detection-of-glaucomatous-progression>
19. American Academy of Ophthalmology. What Is Optical Coherence Tomography? [Internet; cited December 2022]. Available from: <https://www.aao.org/eye-health/treatments/what-is-optical-coherence-tomography>
20. All About Vision. The Retina: Where Vision Begins. [Internet; cited December 2022]. Available from: <https://www.allaboutvision.com/resources/retina.htm>
21. L F Porter, G C Black. Personalized ophthalmology. Clinical Genetics, 2014, Volume 86, Issue 1, Pages 1–11. Available from: <https://onlinelibrary.wiley.com/doi/10.1111/cge.12389>
22. Genetics Home Reference. What is the difference between precision medicine and personalized medicine? What about pharmacogenomics? [Internet; cited December 2022]. Available from: <https://medlineplus.gov/genetics/understanding/precisionmedicine/precisionvspersonalized>



References (3 of 6)

23. Jill Hopkins et al. Delivering personalized medicine in retinal care: from artificial intelligence algorithms to clinical application. Current Opinion in Ophthalmology, 2020, Volume 31, Issue 5, Pages 329 –336. Available from: https://journals.lww.com/co-ophthalmology/fulltext/2020/09000/delivering_personalized_medicine_in_retinal_care_6.aspx
24. J Yim et al. Predicting conversion to wet age-related macular degeneration using deep learning. Nat Med. 2020 Jun;26(6):892-899. doi: 10.1038/s41591-020-0867-7. Epub 2020 May 18.
25. Charles River Associates. The benefits of personalised medicine to patients, society and healthcare systems. [Internet; cited December 2022]. Available from: <https://efpia.eu/media/362040/cra-ebi-efpia-benefits-of-pm-final-report-6-july-2018.pdf>
26. Science Daily. New biomarkers may detect early eye changes that can lead to diabetes-related blindness. [Internet; cited December 2022]. Available from: <https://www.sciencedaily.com/releases/2021/08/210813105533.htm>
27. NHS England – personalised medicine. [Internet; cited December 2022]. Available from: <https://www.england.nhs.uk/healthcare-science/personalisedmedicine>
28. Roche. An introduction to personalised healthcare for the eye. [Internet; cited December 2022]. Available from: https://www.roche.com/research_and_development/what_we_are_working_on/ophthalmology/personalised-healthcare-eye.htm
29. 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. doi: 10.1159/000501374. Epub 2019 Aug 5.
30. Ji-Peng Olivia Li et al. Digital technology, tele-medicine and artificial intelligence in ophthalmology: A global perspective. Progress in Retinal and Eye Research, 2020, DOI: 10.1016/j.preteyeres.2020.100900. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7474840>
31. Vincent Daien. Evolution of treatment paradigms in neovascular age-related macular degeneration: a review of real-world evidence. British Journal of Ophthalmology, 2021, Volume 105, Pages 1475–1479. Available from: <https://bjo.bmj.com/content/105/11/1475>
32. Tham YC et al. New digital models of care in ophthalmology, during and beyond the COVID-19 pandemic. Br J Ophthalmol. 2022 Apr;106(4):452-457. doi: 10.1136/bjophthalmol-2020-317683. Epub 2021 Mar 22.
33. EyeNews UK. The challenges of rural optometry and how independent prescribing has helped. [Internet; cited December 2022]. Available from: <https://www.eyenews.uk.com/media/8045/eyefm17-optometry-v2.pdf>



References (4 of 6)

34. Yoshihiro Takamura et al. New Strategies for Treatment of Diabetic Macular Edema. Journal of Ophthalmology, 2018, Volume 2018. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6120282>
35. Genentech. Seeing the unseen with Artificial Intelligence. [Internet; cited December 2022]. Available from: <https://www.gene.com/stories/seeing-the-unseen-with-artificial-intelligence?topic=ophthalmology>
36. Antonio I. Fernández-Domínguez et al. Plasmon-Enhanced Generation of Nonclassical Light, ACS Photonics, 2018, Volume 5, Issue 9, Pages 3447–3451. Available from: <https://pubs.acs.org/toc/apchd5/5/9>
37. Trishan Panch et al. Artificial intelligence, machine learning and health systems. Journal of Global Health, 2018, Volume 8, Issue 2. Available from: <https://jogh.org/documents/issue201802/jogh-08-020303.pdf>
38. Nathan G Lambert et al. Risk Factors and Biomarkers of Age-Related Macular Degeneration. Progress in Retinal and Eye Research, 2016, Volume 54, Pages 64 –102. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4992630>
39. Xue-Li Du et al. Application of artificial intelligence in ophthalmology. International Journal of Ophthalmology, 2018, Volume 11, Issue 9, Pages 1555–1561. Available from: <https://pubmed.ncbi.nlm.nih.gov/30225234>
40. Front Line Genomics. Artificial intelligence systems in ophthalmology. [Internet; cited December 2022]. Available from: <https://d4-pharma.com/artificial-intelligence-systems-in-ophthalmology>
41. Retina Specialist. How Imaging Can Identify Predictive Biomarkers of AMD. [Internet; cited December 2022]. Available from: <https://www.retina-specialist.com/article/how-imagine-can-identify-predictive-biomarkers-of-amd>
42. Sajib Saha et al. Automated detection and classification of early AMD biomarkers using deep learning. Scientific Reports, 2019, Volume 9. Available from: <https://www.nature.com/articles/s41598-019-47390-3>
43. Frank S Ong. Personalized Medicine in Ophthalmology: From Pharmacogenetic Biomarkers to Therapeutic and Dosage Optimization. Journal of Personalized Medicine, 2013, Volume 3, Issue 1, Pages 40 –60. Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3947950>
44. Eveline Kersten et al. Systemic and ocular fluid compounds as potential biomarkers in age-related macular degeneration. Survey of Ophthalmology, 2018, Volume 63, Issue 1, Pages 9–39. Available from: <https://www.sciencedirect.com/science/article/pii/S0039625716302624>



References (5 of 6)

45. Irina Balikova et al. Genetic biomarkers in the VEGF pathway predicting response to anti-VEGF therapy in age-related macular degeneration. *BMJ Open Ophthalmology*, 2019, Volume 4. Available from: <https://bmjophth.bmj.com/content/4/1/e000273.info>
46. Retina Today. Putting vision monitoring in the hands of patients with AMD. [Internet; cited December 2022]. Available from: <http://retinatoday.com/2017/06/putting-vision-monitoring-in-the-hands-of-patients-with-amd>
47. Steven R Steinhubl et al. The emerging field of mobile health. *Science Translational Medicine*, 2015, Volume 7, Issue 283. Available from: <https://www.science.org/doi/10.1126/scitranslmed.aaa3487>
48. Medical Technology. Eye On The Prize: Ophthalmology's Digital Future. [Internet; cited December 2022]. Available from: https://medical-technology.nridigital.com/medical_technology_jun20/ophthalmology_digital_future
49. Davide Cirillo, Alfonso Valencia. Big data analytics for personalized medicine. *Current Opinion in Biotechnology*. 2019, Volume 58, Pages 161–167. Available from: <https://www.sciencedirect.com/science/article/pii/S0958166918301903>
50. Roach L. American Academy of Ophthalmology. Precision Medicine. [Internet; cited December 2022]. Available from: <https://www.aao.org/eyenet/article/precision-medicine>
51. Giulia Sedda et al. Challenges and innovations in personalized medicine care. *Future Oncology*, 2019, Volume 15, Pages 3305–3308. Available from: <https://www.futuremedicine.com/doi/pdf/10.2217/fon-2019-0284>
52. Cécile Méjécase et al. Practical guide to genetic screening for inherited eye diseases. *Therapeutic Advances in Ophthalmology*, 2020, Volume 12. Available from: <https://journals.sagepub.com/doi/10.1177/2515841420954592>
53. AAO, Genetics and Age-Related Macular Degeneration. [Internet; cited December 2022]. Available from: <https://www.aao.org/eye-health/diseases/age-related-macular-degeneration-amd-genetics>
54. NICE guidelines, Scenario: Confirmed age-related macular degeneration. [Internet; cited December 2022]. Available from: <https://cks.nice.org.uk/topics/macular-degeneration-age-related/management/confirmed-amd/>
55. National Health Service. NHS Genomic Medicine Service. [Internet; cited December 2022]. Available from: <https://www.england.nhs.uk/genomics/nhs-genomic-med-service>



References (6 of 6)

56. Graeme C Black et al. The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet Journal of Rare Diseases, 2021, Volume 16, Issue 11. Available from: <https://www.ndcn.ox.ac.uk/publications/1174546>
57. The Precision Medicine Initiative. [Internet; cited December 2022]. Available from: <https://obamawhitehouse.archives.gov/precision-medicine>
58. Bernd Blobel et al. (2016). Patient privacy and security concerns on big data for personalized medicine. Health and Technology. 6. 10.1007/s12553-016-0127-5. Available from: https://www.researchgate.net/publication/295399085_Patient_privacy_and_security_concerns_on_big_data_for_personalized_medicine
59. Chou AF, et al. Barriers and strategies to integrate medical genetics and primary care in underserved populations: a scoping review. J Community Genet. 2021 Jul;12(3):291-309. doi: 10.1007/s12687-021-00508-5. Epub 2021 Feb 1. PMID: 33523369; PMCID: PMC7849219. Available from: <https://pubmed.ncbi.nlm.nih.gov/33523369/>
60. EyeNews UK. Technology in ophthalmology – a promising future and what we need to know about the regulations. [Internet; cited December 2022]. Available from: <https://www.eyenews.uk.com/education/trainees/post/technology-in-ophthalmology-a-promising-future-and-what-we-need-to-know-about-the-regulations>
61. Treatments - Age-related macular degeneration (AMD) (NHS). [Internet; cited December 2022]. Available from: <https://www.nhs.uk/conditions/age-related-macular-degeneration-amd/treatment/>
62. Ellingford JM et al. 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. doi: 10.3390/genes11111274.