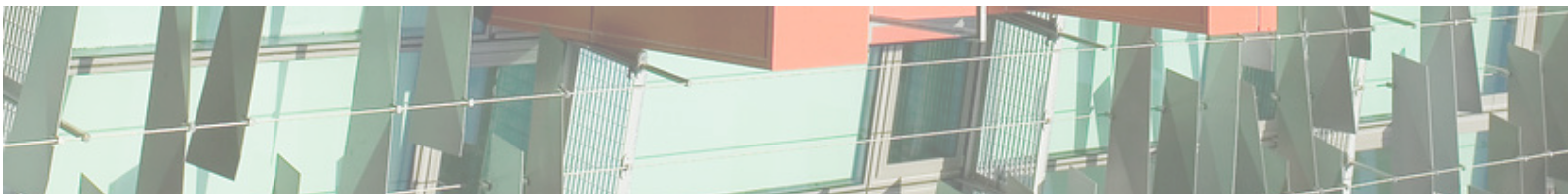
LONDON'S GLOBAL UNIVERSITY

Eye2Gene: Information for patients

UCL INSTITUTE OF OPHTHALMOLOGY

Moorfields Eye Hospital NHS Foundation Trust



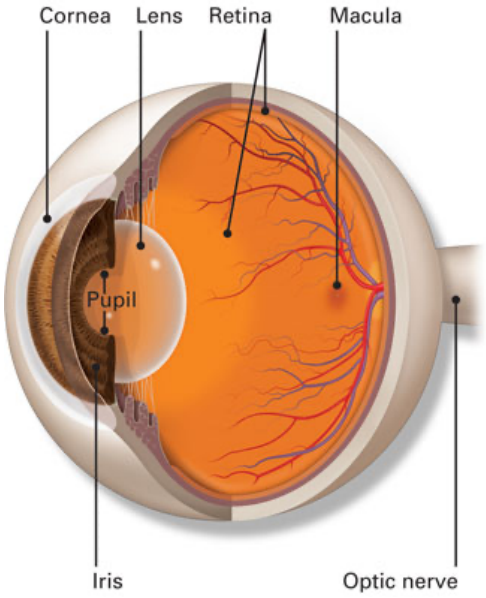
Background: the eye

You can think of the human body as a complex machine. The instructions for running it are stored in your genes. Genes are like blueprints that tell the body how to function correctly. Everyone inherits a mixture of their genes from each of their parents, which is why we look similar to them.

Genes store information about how a specific part of the body should work. Sometimes the information contained in these genes is faulty, so that part of the body may not function normally. These “faulty” genes are inherited from our parents, hence the disease is called “inherited disease”.

We see the world around us using our eyes. The back of the eye contains a layer called the retina, which converts light into information for the brain to understand.

The retina is incredibly important, because if it does not work correctly, the quality of your vision will be impaired. There are many diseases that can affect the retina, including those caused by faulty genes. If you have a faulty gene that stores some information about the retina, this could affect your sight and give you an inherited retinal disease, or IRD. There are about 30,000 individuals affected by over 300 inherited retinal diseases in the United Kingdom. IRDs are the one of the leading causes of blindness in children and young adults in the United Kingdom.



The process of being diagnosed with an inherited retinal disease is called the patient journey, which for many patients can take over 5 years. It may start with an optician noticing the patient’s vision worsening, or signs of damage when they examine the retina. This leads to further tests trying to work out which specific gene could be faulty. Only then can a doctor explain which disease affects the patient’s sight (the diagnosis), and how it may change over time (the prognosis).

Unfortunately, this process does not always work well. It requires attending many appointments with expert professionals, which can be difficult for patients who live far from the UK hospitals specialising in diagnosing IRDs. Being diagnosed usually takes years, which is stressful for patients who do not understand why their vision is reduced or worsening. It also makes getting help, such as claiming benefits, harder. There are currently over 10,000 individuals in the UK living with IRDs who have not received a diagnosis, some of whom are stuck in this journey.

Being diagnosed with inherited retinal disease will always be challenging for patients. However, it could be made much easier with some improvements to how they are looked after by the healthcare system.

Background: AI

You may have already heard of artificial intelligence, or AI, as it is frequently mentioned in the media. It is helpful to think of AI as a computer program which is built to perform a specific task. Importantly, it gets better at that task the more it does it. This means that AI becomes more accurate as it gains experience. The amazing ability of artificial intelligence to learn over time is used to solve problems in a wide range of industries. This includes self-driving cars; online banking; and of course, medical research.

In recent years, there has been great interest in using artificial intelligence in hospitals.

At the moment, when doctors think a patient may have an inherited retinal disease, they will look at photographs and scans of the affected retinas. Some IRDs cause unique, specific patterns of damage to the retina, which can be used to work out which disease affects the patient. Being able to understand scans like this requires a lot of experience: the most experienced doctors are called consultants, and there are only approximately 50 specialising in IRDs in the UK. This means they can only see a limited number of patients, causing a significant delay.

We currently rely on doctors diagnosing patients by identifying certain disease-associated patterns in scans and photographs of damaged retinas. Given that artificial intelligence can learn to identify these types of patterns, and get better at it over time, could it learn to recognise signs of disease? Previous research says yes. Many scientific studies

have already shown AI can identify specific diseases affecting the eye using this information, including inherited retinal diseases.

The solution

Eye2Gene is a research project based at Moorfields Eye Hospital and University College London. The aim is to build artificial intelligence that can look through photographs and scans of retinas, and accurately predict which inherited retinal disease may affect the patient. Once thoroughly tested, it can then be used to help make decisions about their care.

For example, using Eye2Gene in a high-street opticians may help identify individuals with IRDs earlier, so they can be referred directly to specialist consultants in hospitals. Or, it could be used in laboratories to assist scientists trying to understand the results of genetic tests.

The plan

The Eye2Gene project will start by collecting anonymous information from patient records in four eye hospitals: Moorfields; Oxford University; Royal Liverpool; and Tokyo Medical Centre. Using patient data in research is always taken extremely seriously, and only allowed after approval by specialised organisations who work directly under the government. Eye2Gene research is funded by the National Institute for Health Research (NIHR) and has been authorised by the NHS Health Research Authority, who ensure research is fair, responsible, and ethical.

💡 Data is information, which computers can understand

The patient data will be carefully sorted by experts, including doctors specialising in inherited retinal diseases. They will highlight important features, eg in photographs that show signs of damage to the retina, and identify which inherited retinal disease caused it. This is similar to how such diseases are regularly diagnosed in hospitals.

To build the artificial intelligence system, it is first given photographs and scans of damaged retinas, and told which inherited disease caused it.

During this stage, the AI is learning which bits of information are important for recognising signs of disease, similar to how we learn. Next, it will try to copy the human experts. After being shown brand new scans and photographs, the AI will predict which disease they show. Researchers will tell the system how accurate those predictions were, and the whole process will be repeated over and over again. This continues until Eye2Gene can look at this patient information, and reliably identify which gene may be faulty.

Finally, Eye2Gene can apply to be certified by another specialised organisation, the Medicines and Healthcare products Regulatory Agency (MHRA), who regulate medical devices in the UK. If approved, it can then be used in places like medical laboratories and opticians, to look over patient data and help make decisions about care.

About the project

Eye2Gene was originally created by two scientists with backgrounds in artificial intelligence and ophthalmology, Dr Nikolas Pontikos and Dr William Woof.

Dr Pontikos studied computer science, bioinformatics, and statistical genetics at university. These degrees are about understanding computers, and using them to explore biological information, like human health. This interest carries over into his professional experience, as Dr Pontikos currently manages a research team based at UCL and Moorfields Eye Hospital of which Dr Woof is part of. Their aim is to make patient care more personalised and effective using healthcare data.

Dr Woof was diagnosed with an inherited retinal disease (USH2A-retinopathy) when he was 18 years old. Since then, he has maintained a strong interest in his condition, including involvement in patient support groups around Manchester, where he is based. Dr Woof studied mathematics and artificial intelligence at university, which sparked a strong interest in using the technology to help patients with inherited retinal disease.

Dr Pontikos and Dr Woof initially discussed Eye2Gene a few years ago, waiting until they were both available to formally begin the project, which is funded by a £1.3 million grant from NIHR.

Timeline

2019: Dr Nikolas Pontikos starts working on the Eye2Gene prototype.

2020: Dr William Woof joins the team and helps improve the prototype.

2021: Dr Pontikos applies for funding to the NIHR and the Wellcome Trust to support further development of Eye2Gene. The NIHR agree to fund the project for 3 years.

2022: The NIHR funded project officially kicks off. First meeting with patient group take place. Data transfer from participating hospitals to the Eye2Gene system can start.

2023: Working prototype of the Eye2Gene system is further developed, with the ability to explain to users how it makes decisions.

2024: Eye2Gene prototype is sufficiently developed to begin the regulatory approval process so that it can be used as software as a medical device.

Contact details

More information can be found on our website: www.eye2gene.com

If you are interested in the project, please contact: n.pontikos@ucl.ac.uk

Authorship

Written by Mr Nathaniel Kabiri and edited by Professor Michel Michaelides and Dr Nikolas Pontikos, February 2022