



Moorfields
Eye Hospital
NHS Foundation Trust



Genomics to Transform Glaucoma Care

Anthony Khawaja

Honorary Consultant Ophthalmologist & Professor of Ophthalmology

Moorfields Eye Hospital AGM

2nd October 2024



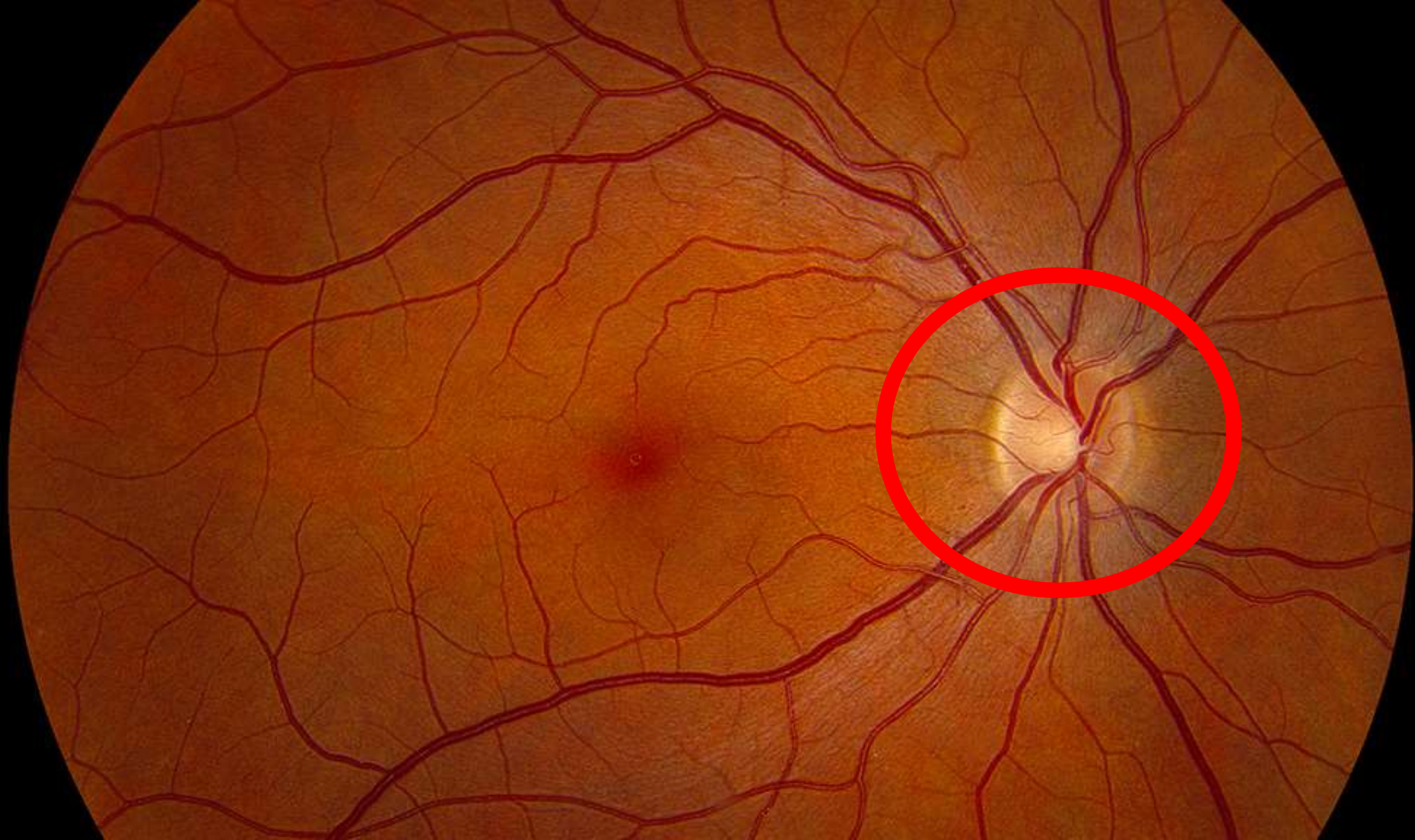
Early glaucoma

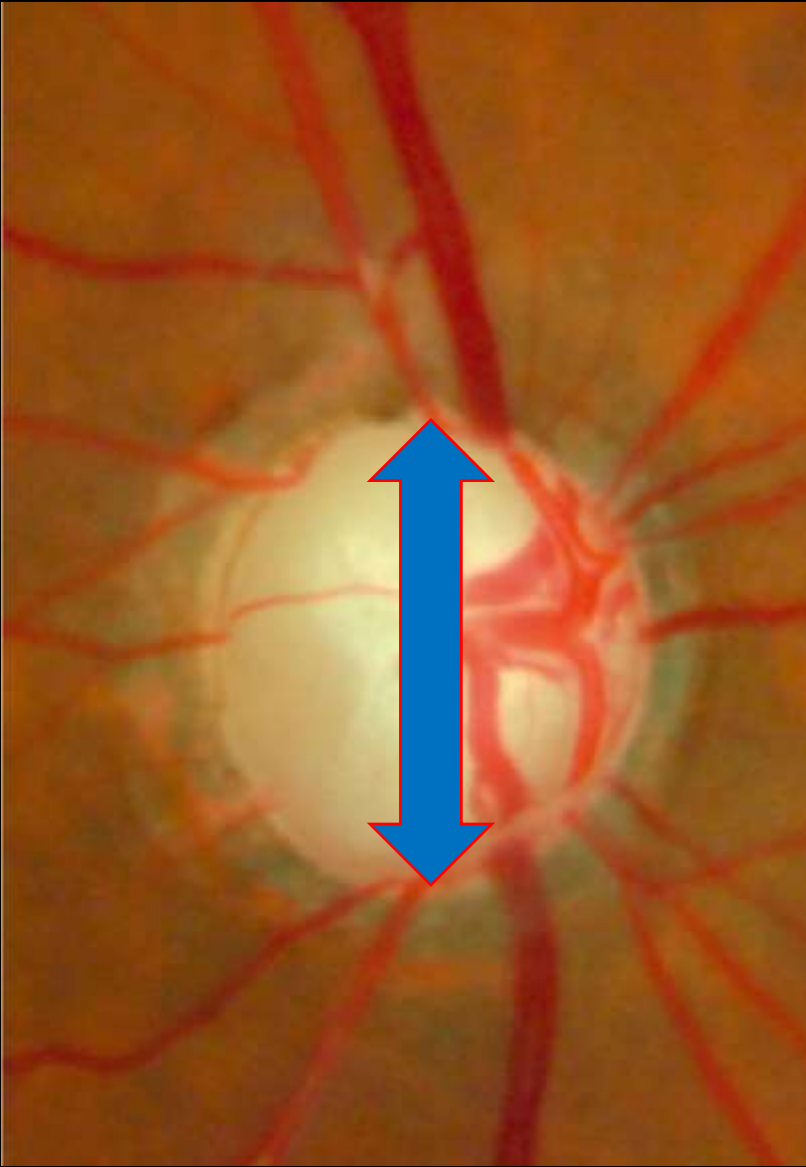
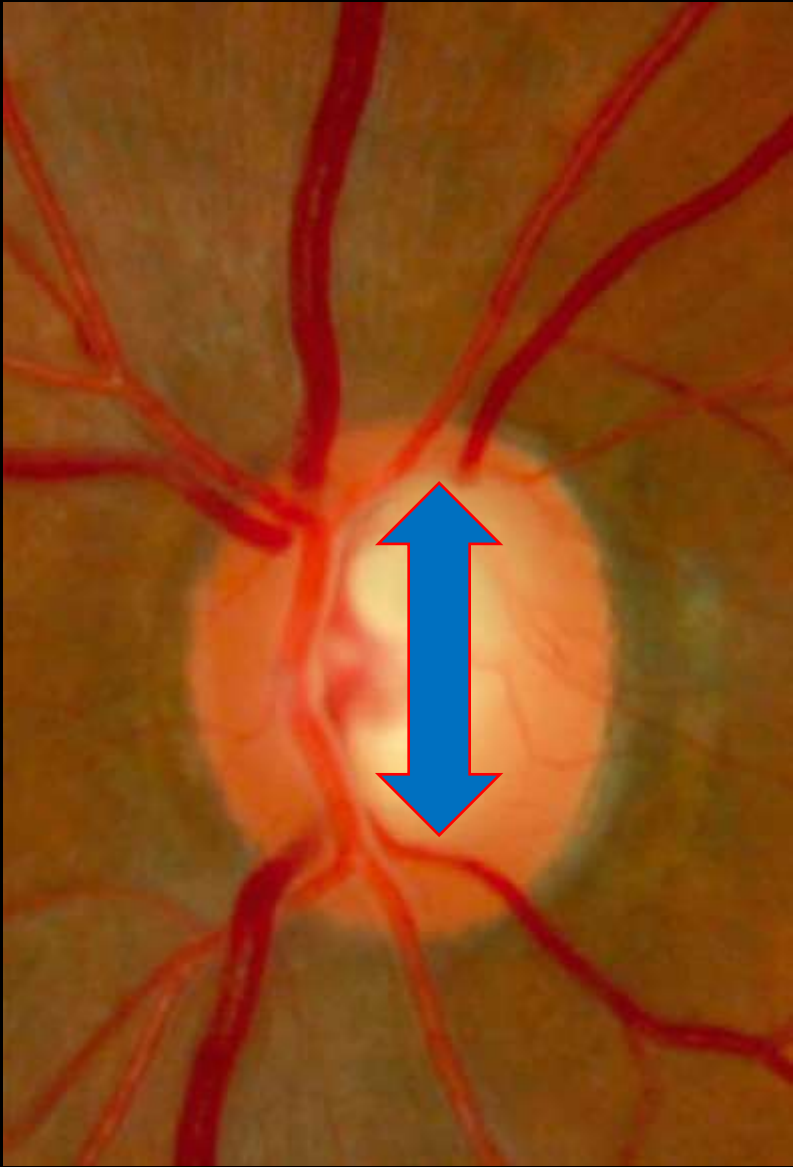
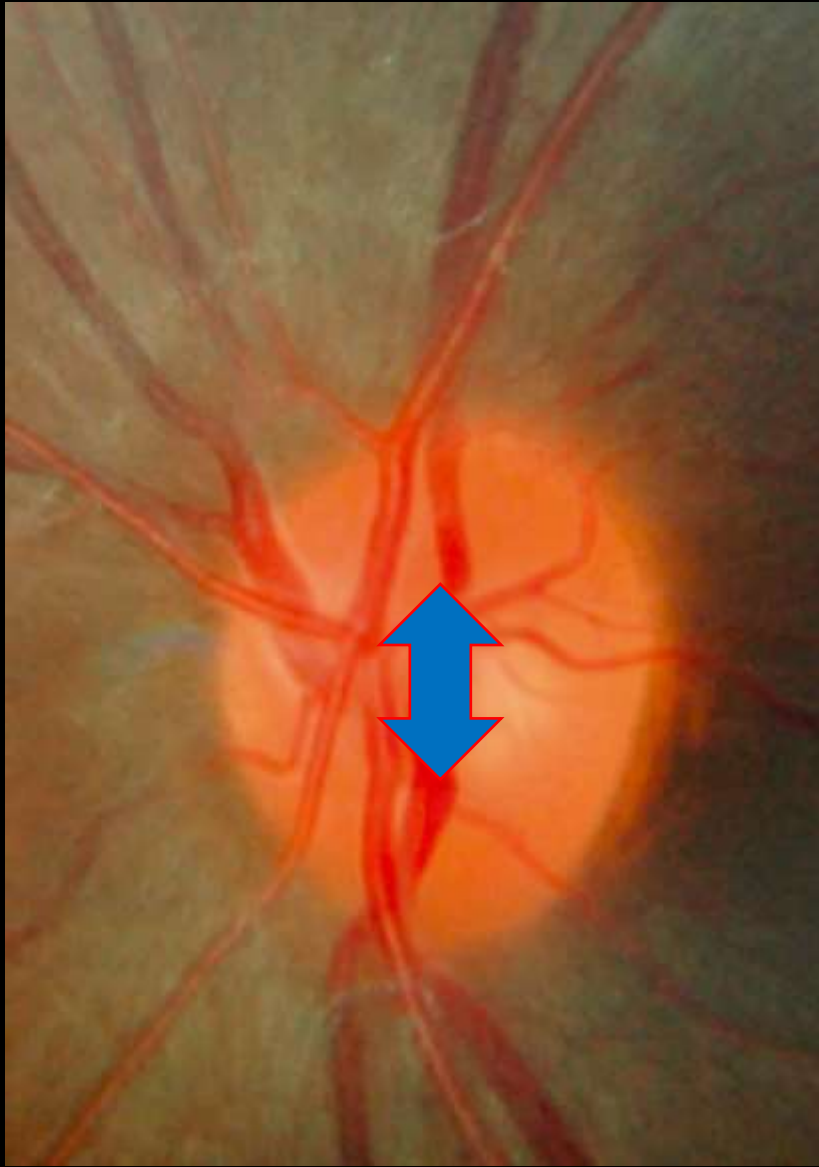


UK: £28 billion /yr

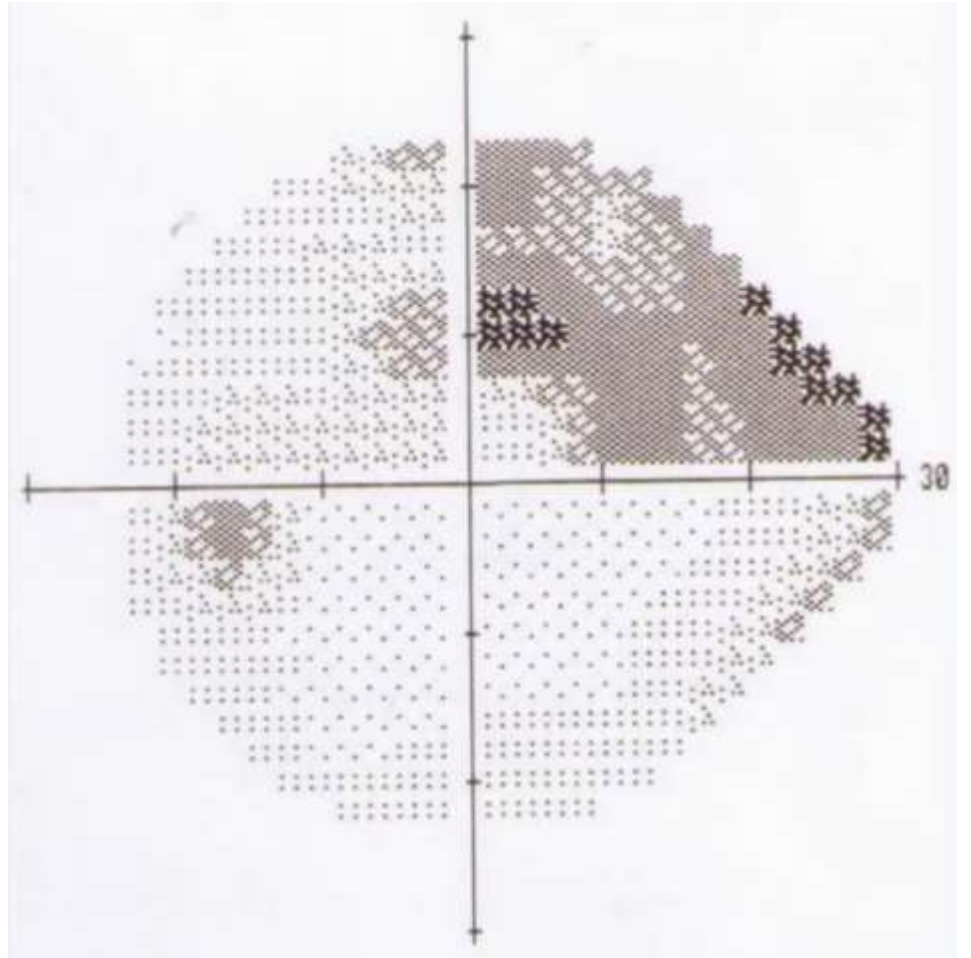
Late glaucoma





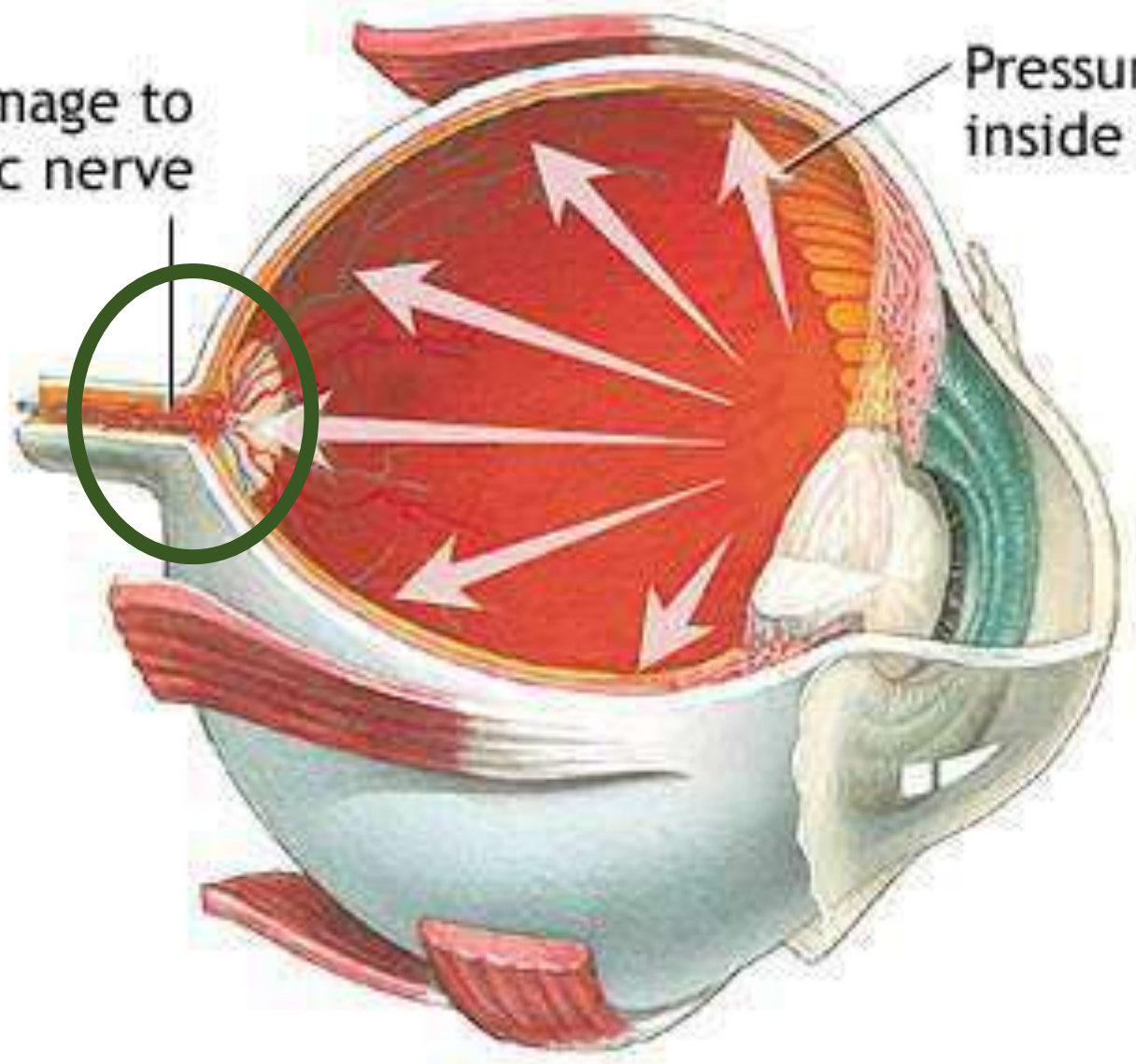






Damage to optic nerve

Pressure inside eye



Challenges



Challenges

#1: Screening is not recommended



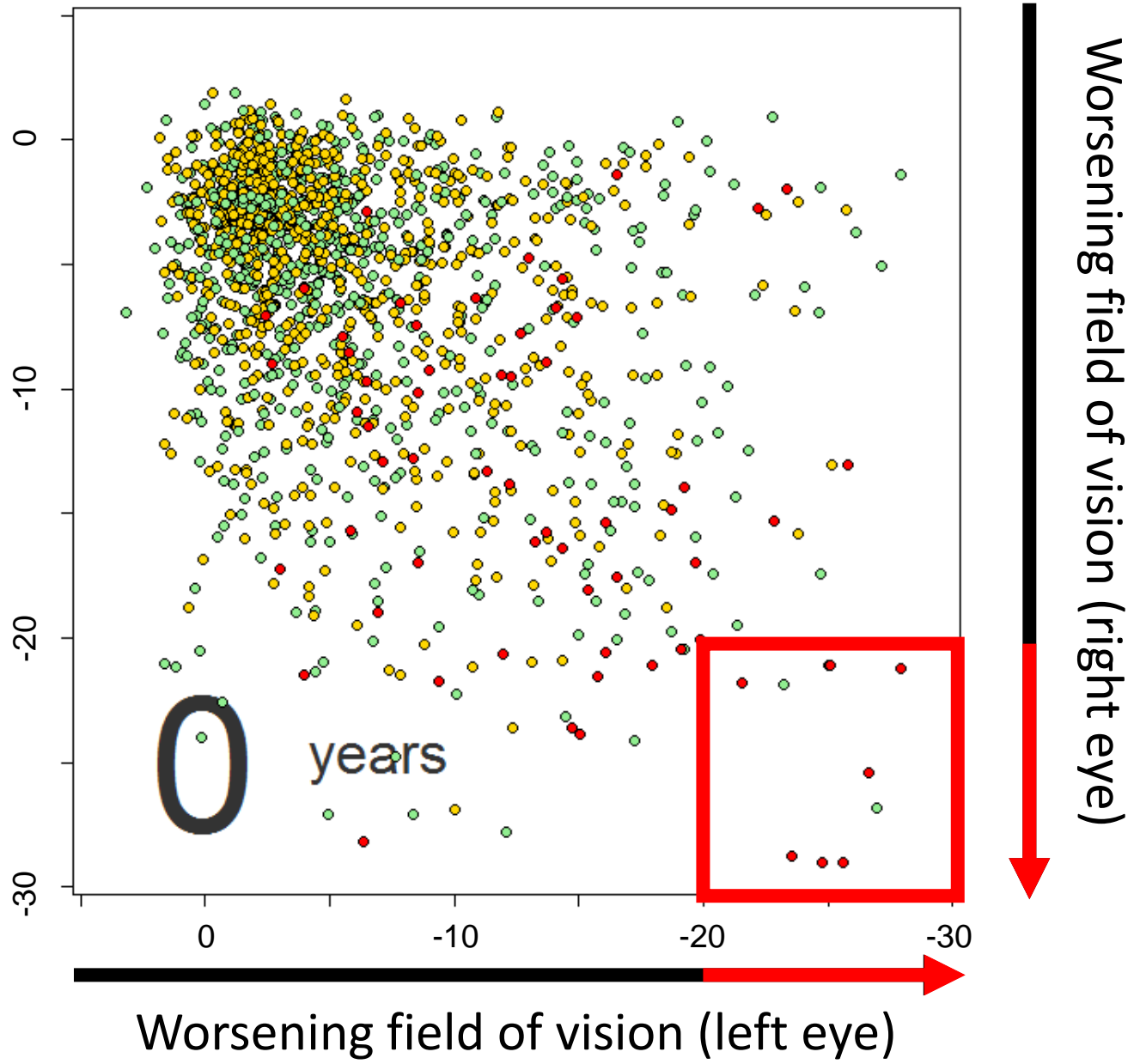


Challenges

#1: Screening is not recommended

#2: Risk of blindness varies





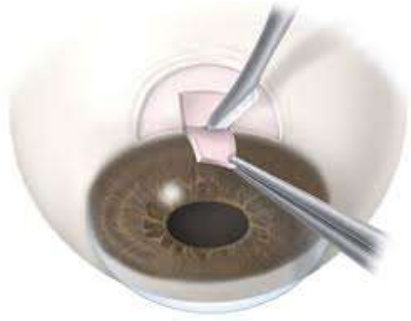
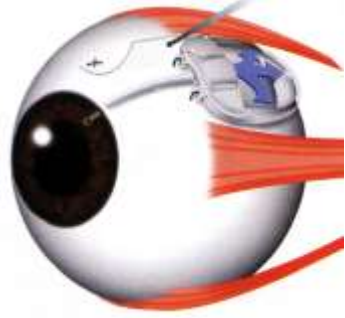
Challenges

#1: Screening is not recommended

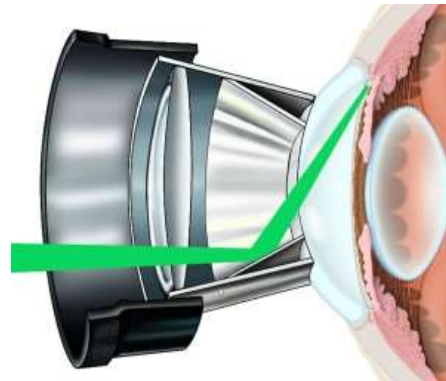
#2: Risk of blindness varies

#3: Treatment outcomes vary





Many treatments!



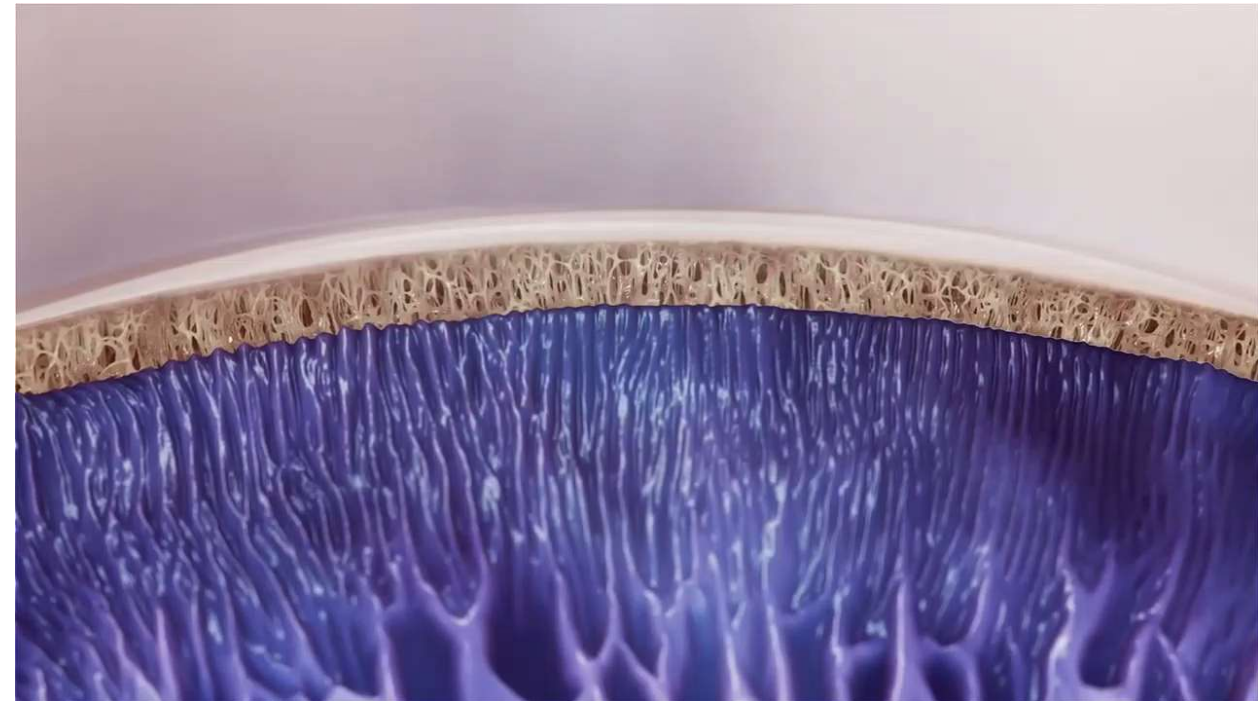
Selective laser trabeculoplasty versus eye drops for first-line treatment of ocular hypertension and glaucoma (LiGHT): a multicentre randomised controlled trial

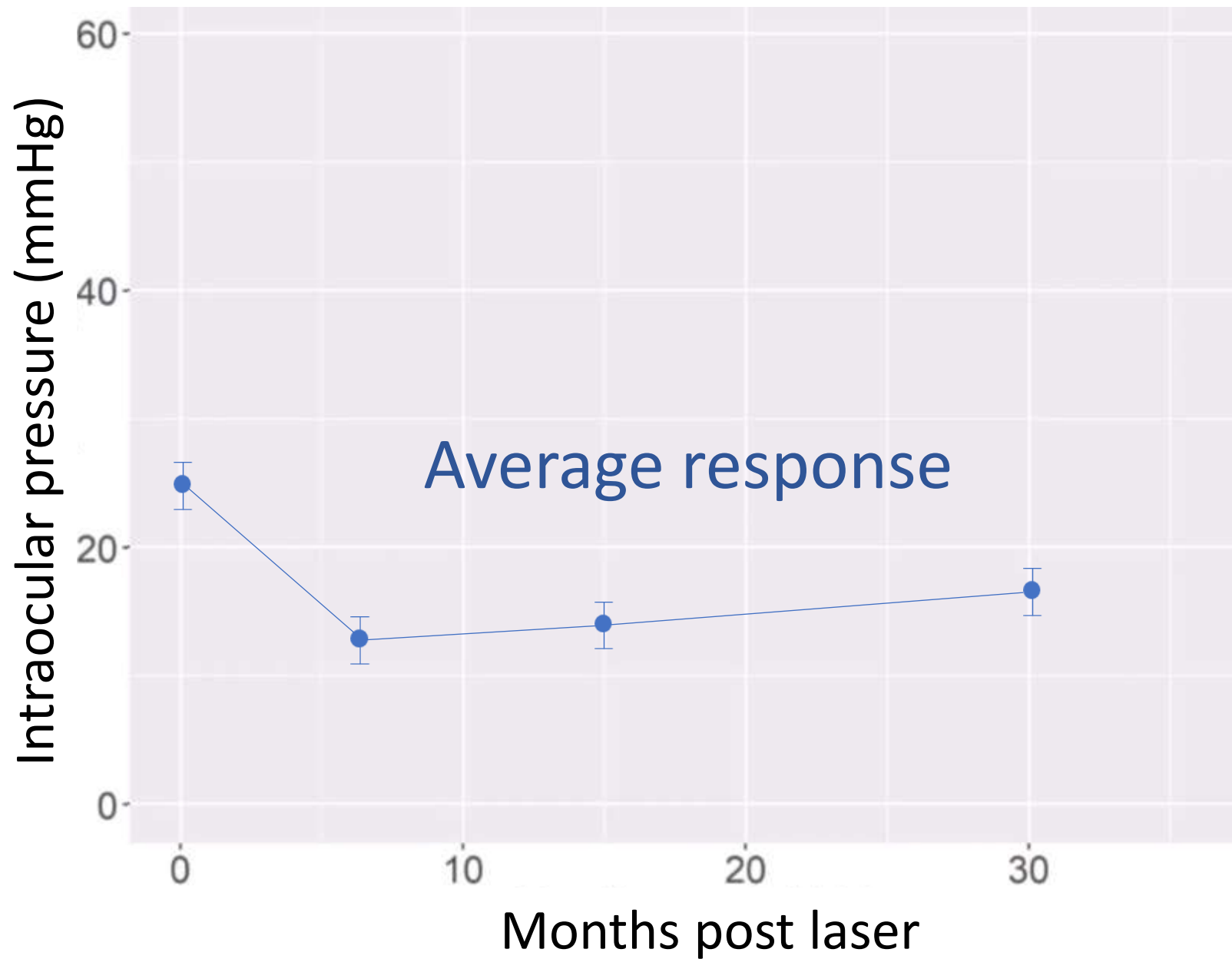


Gus Gazzard, Evgenia Konstantakopoulou, David Garway-Heath, Anurag Garg, Victoria Vickerstaff, Rachael Hunter, Gareth Ambler, Catey Bunce, Richard Wormald, Neil Nathwani, Keith Barton, Gary Rubin, Marta Buszewicz; on behalf of the LiGHT Trial Study Group*



Vs







wellcome collection



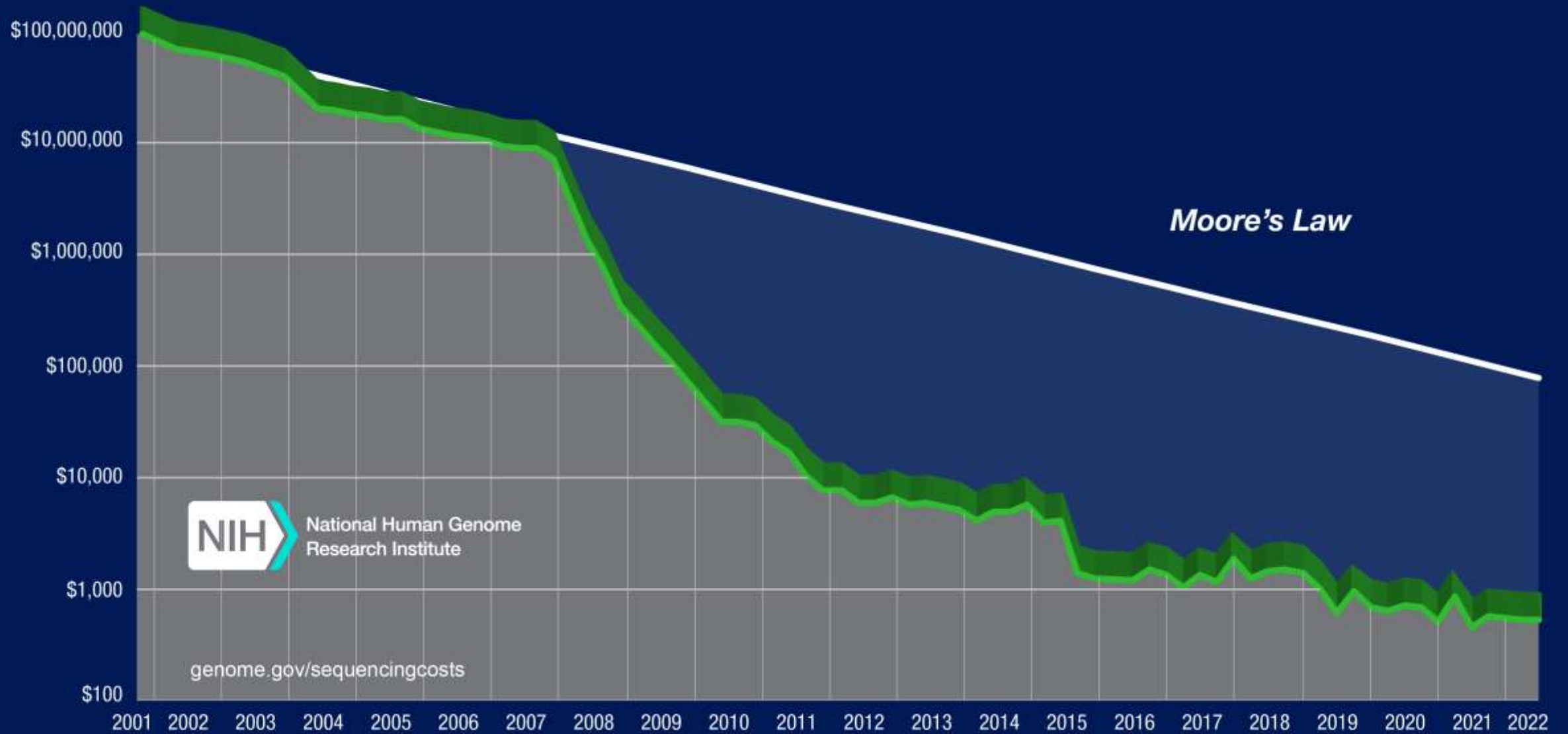


Genomic data represented as a grid of vertical columns of characters. Each column contains a single character, likely a nucleotide (A, C, G, T) or a gap symbol (N). The characters are arranged in a regular grid pattern, with some gaps or irregularities in the spacing between columns. The data appears to be a sequence alignment or a set of genomic coordinates.



**3 billion letters
a/c/g/t**

Cost per Human Genome



Superd

Shop Categori

Home > Health > Home Tes



The best DNA gift
with the most to give.



£63

+ FREE gift wrap
Ancestry + Traits

£99

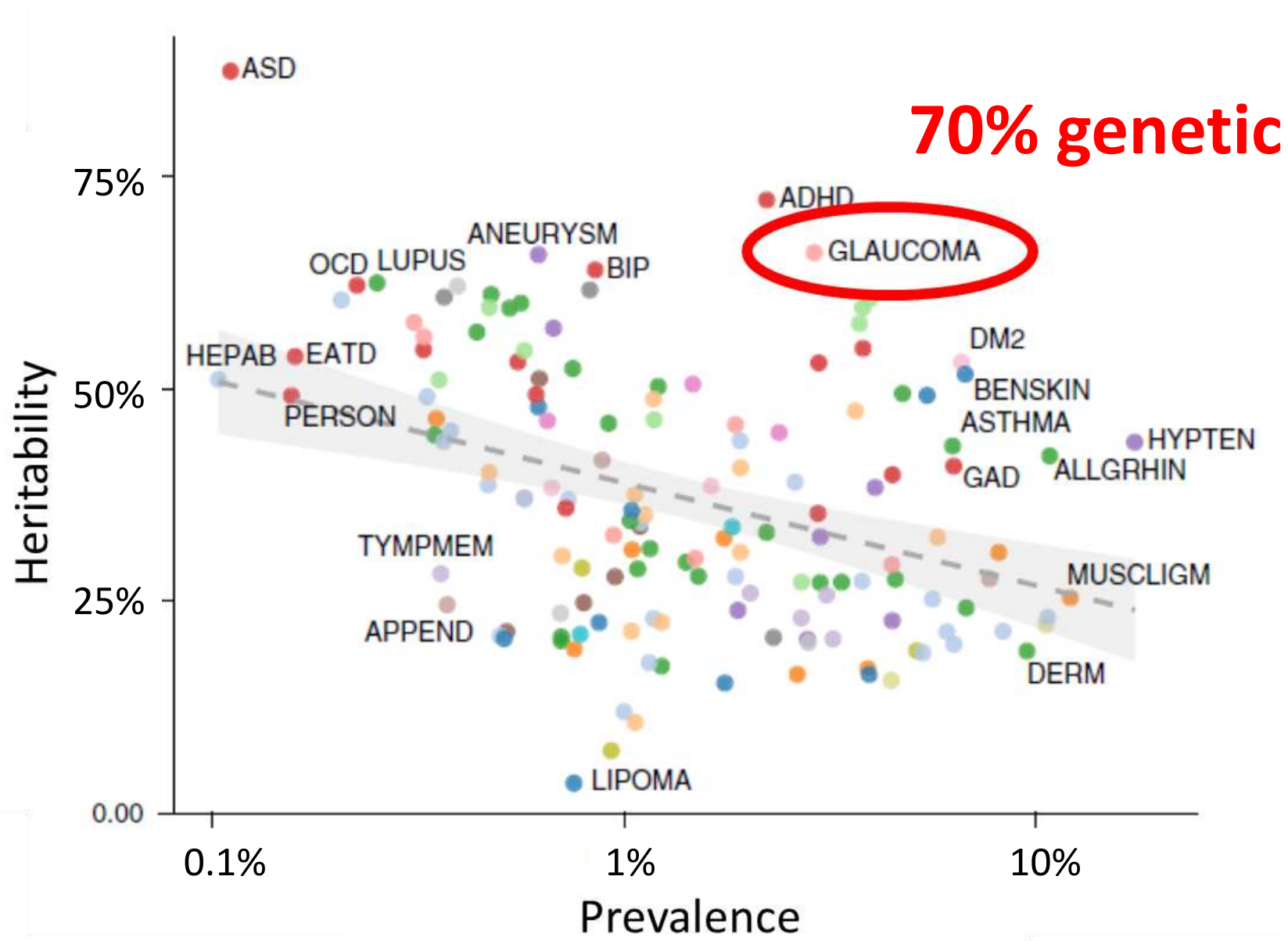
+ FREE gift wrap
Health + Ancestry

Search produc

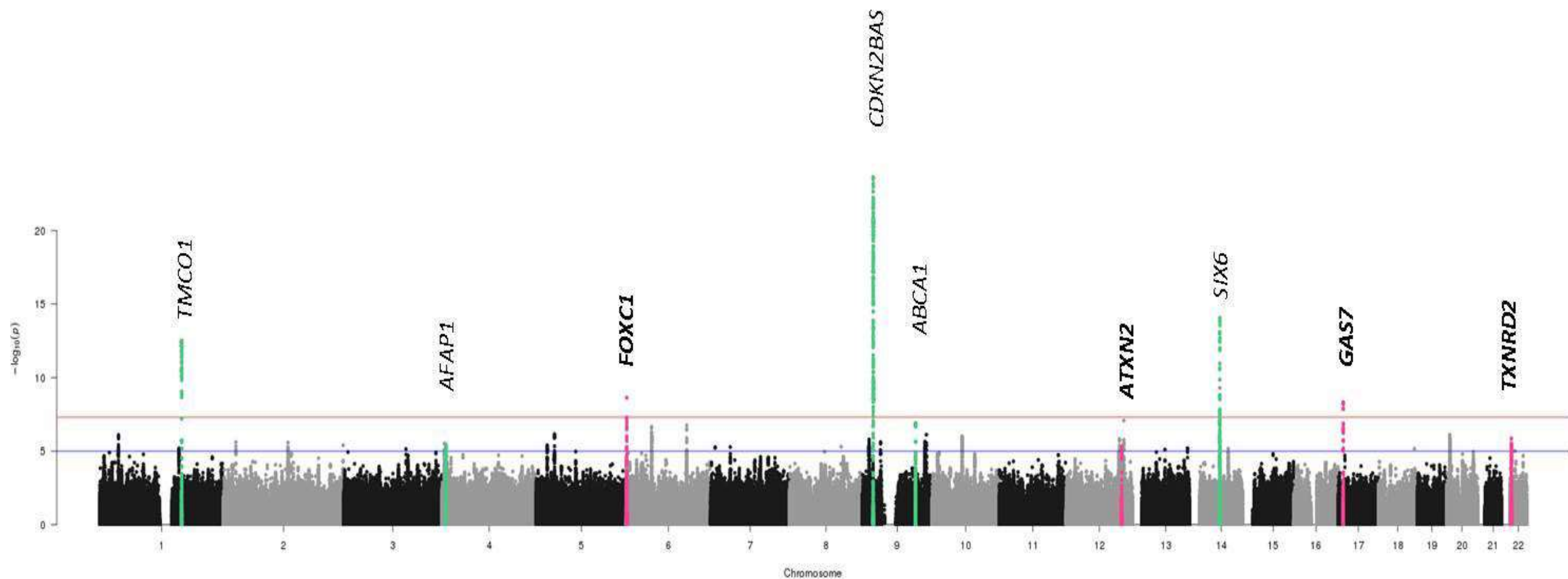
Store & Services

Home Service:

shlist



Genome-wide association analysis identifies *TXNRD2*,
ATXN2 and *FOXC1* as susceptibility loci for primary
open-angle glaucoma



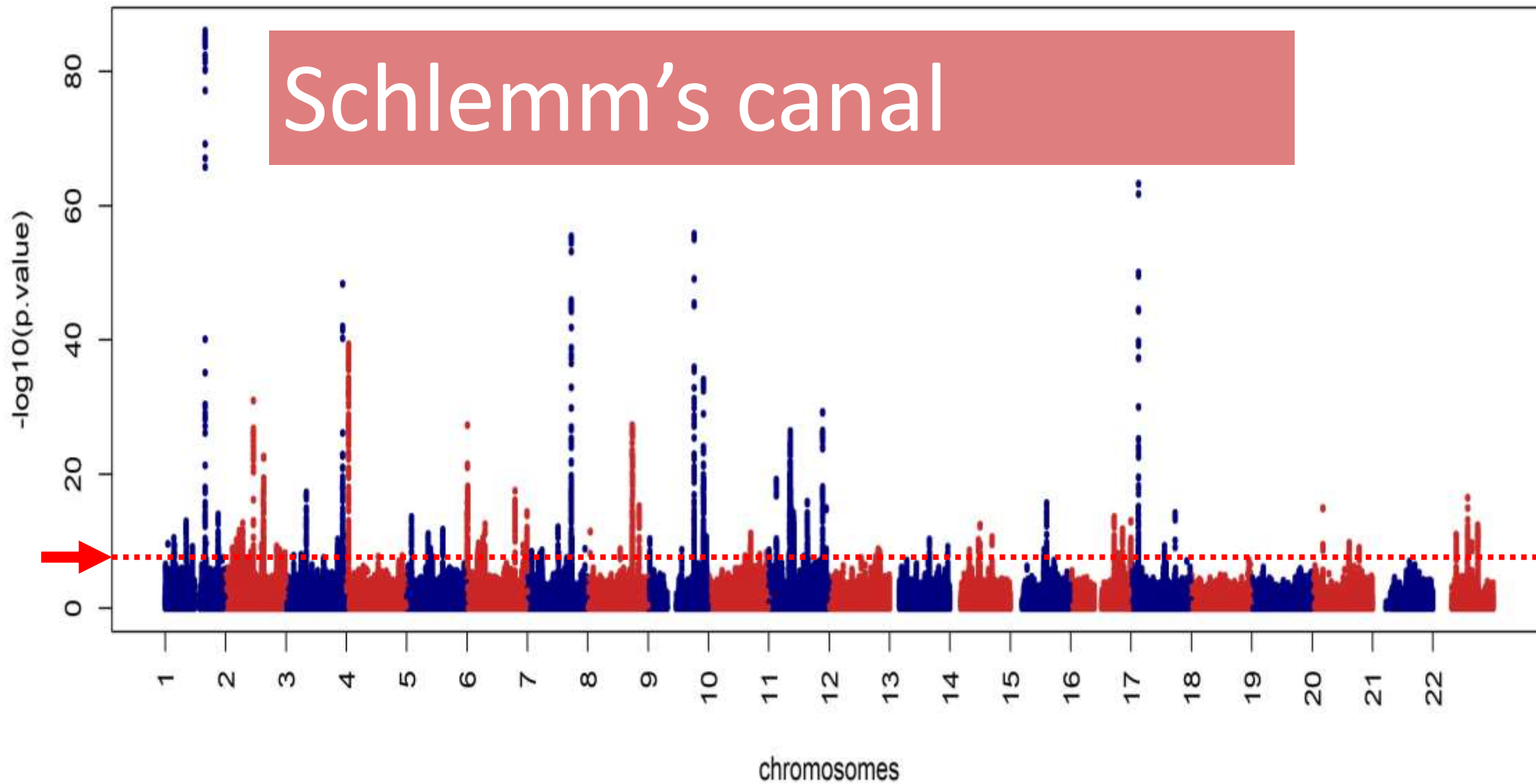


500,000 participants

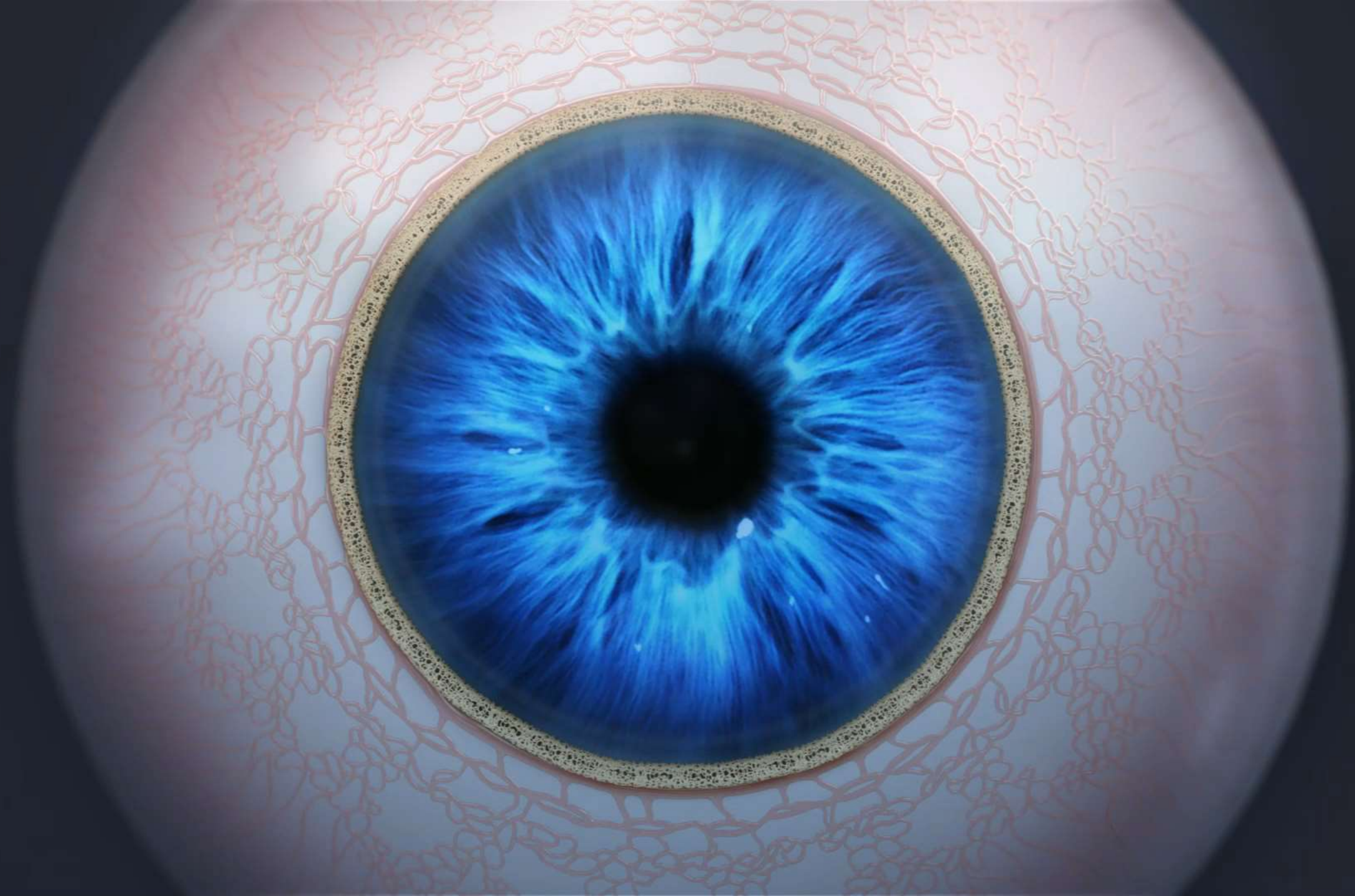
22 centres

Eye measures supplement





Khawaja *et al*, Nature Genetics, 2018

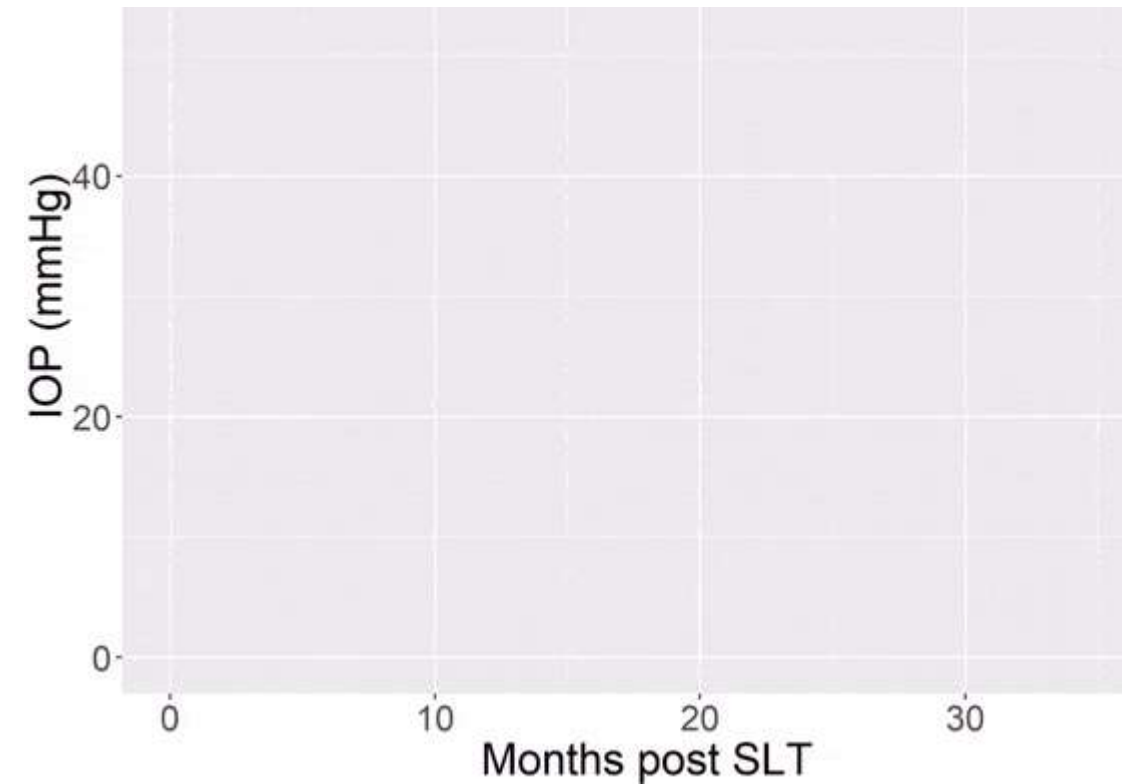
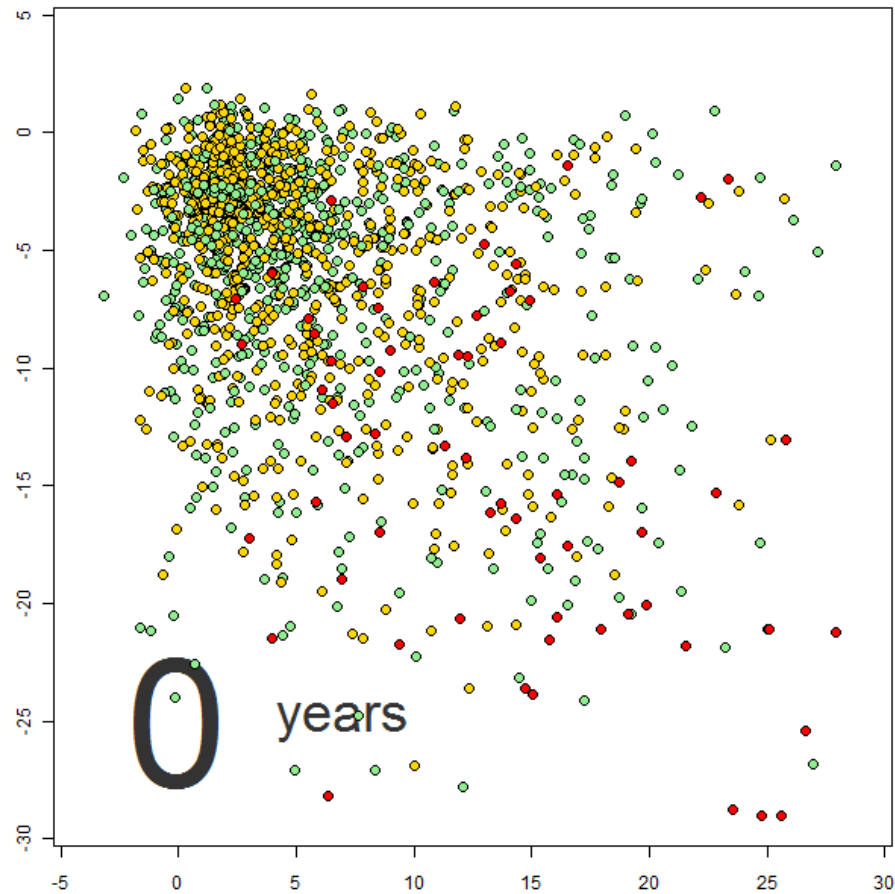


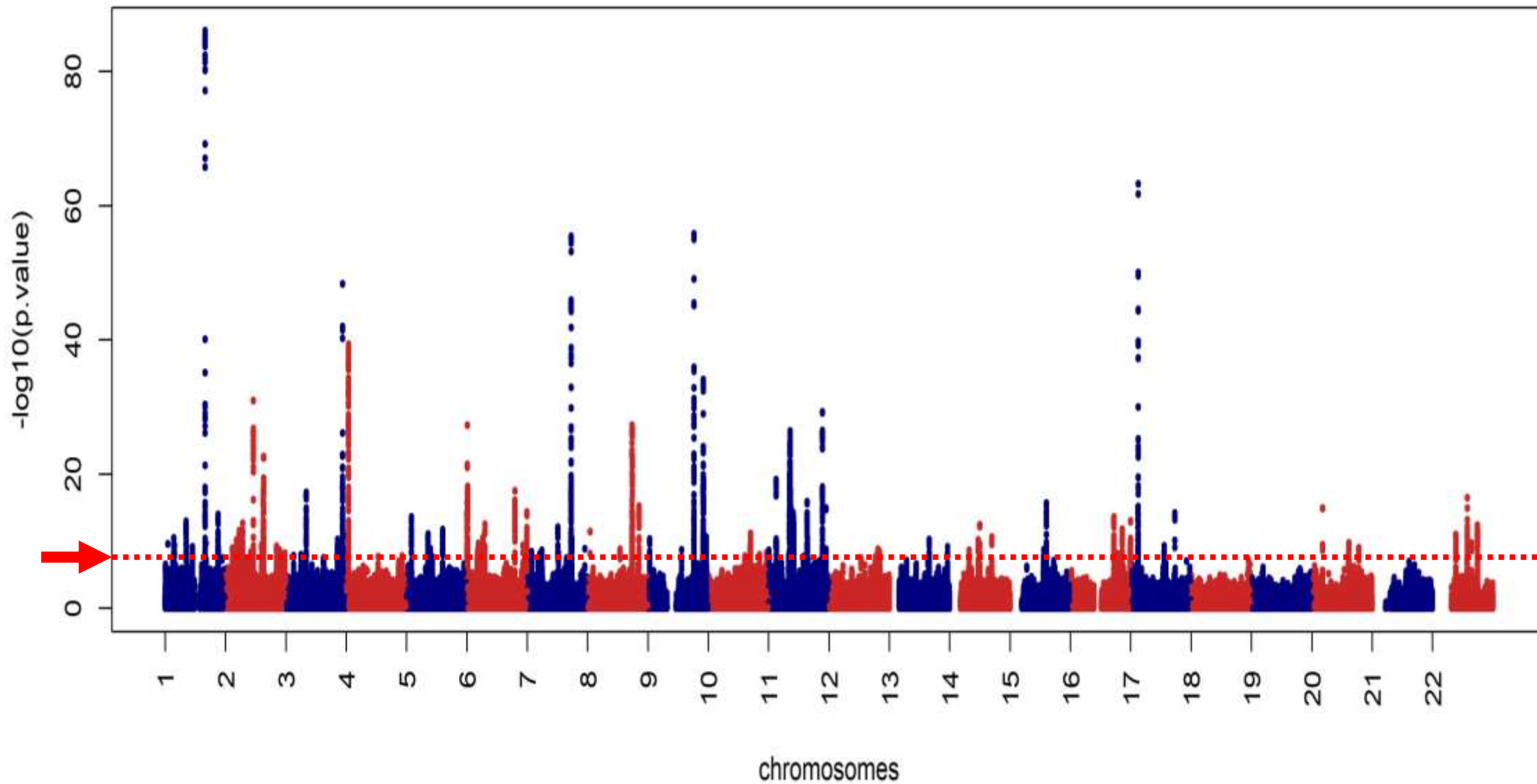


Moorfields Glaucoma BioResource



Moorfields Glaucoma BioResource

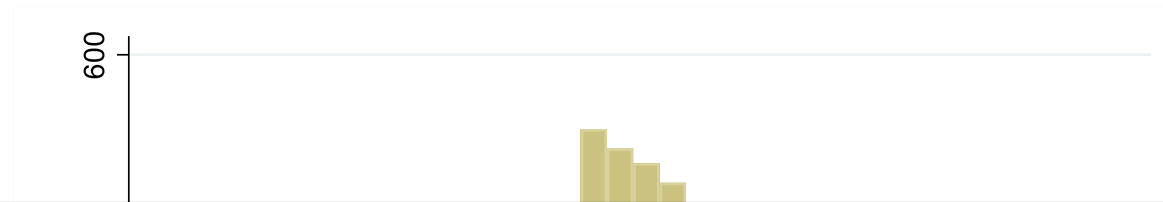




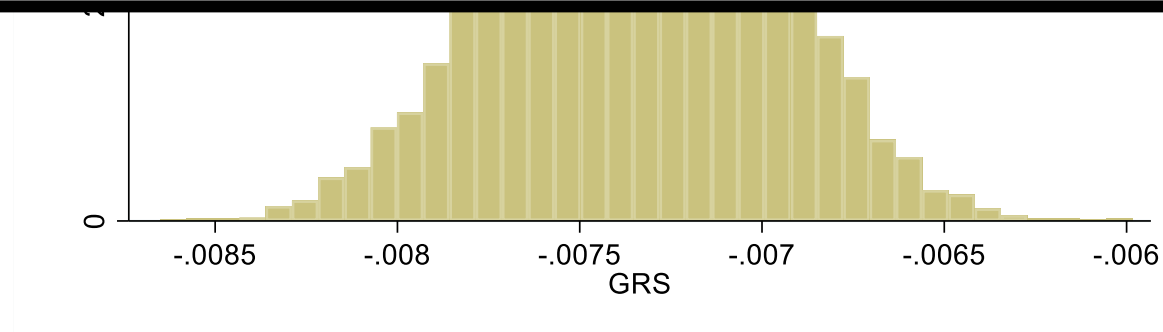
Khawaja *et al*, Nature Genetics, 2018

One number!

One number!

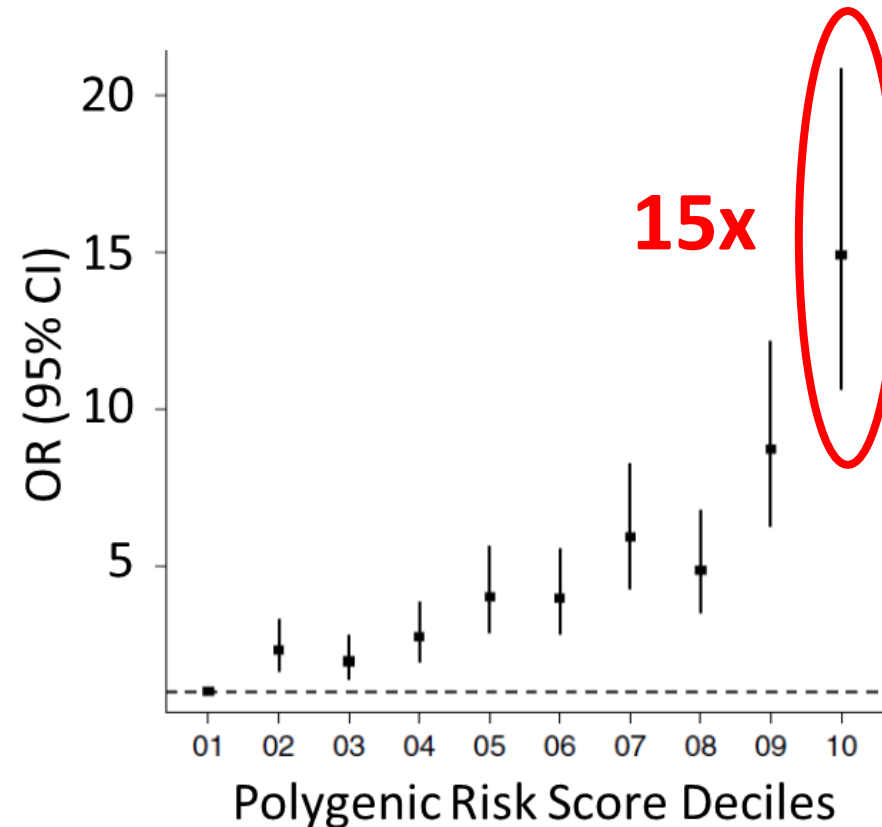


PRS - Polygenic risk score



Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression

Jamie E. Craig^{1,40}, Xikun Han^{2,3,40*}, Ayub Qassim^{1,40}, Mark Hassall¹, Jessica N. Cooke Bailey⁴, Tyler G. Kinzy⁴, Anthony P. Khawaja⁵, Jiyuan An², Henry Marshall¹, Puya Gharahkhani²,



What can I do to help
with my glaucoma?



A word cloud featuring various health and lifestyle terms. The words are arranged in a non-linear fashion, with some appearing larger than others. The colors used are purple, green, and grey. The terms include: activity, food, outdoors, vegetables, vegetarian, tea, coffee, sunlight, fibre, fruit, smoking, drugs, Mediterranean, drink, fat, diet, nutrition, sedentary, exercise, alcohol, medications, modify, and meat.

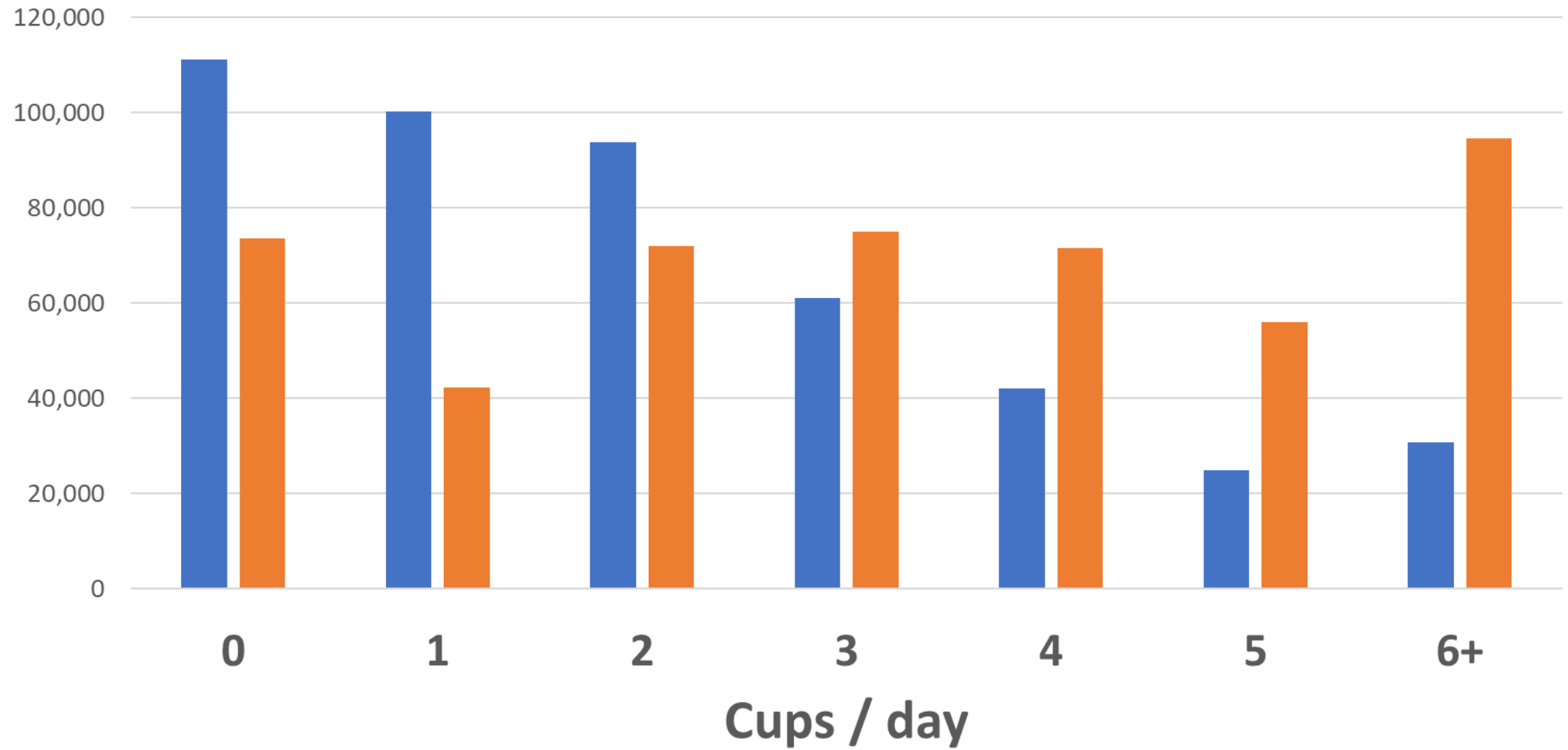
activity food outdoors
vegetables
vegetarian tea
coffee sunlight fibre
fruit smoking drugs
Mediterranean drink fat diet
nutrition sedentary
exercise alcohol
medications
modify meat

Modifiable Risk Factors for Glaucoma Collaboration





■ Coffee ■ Tea



Caffeine derivation



Oxford WebQ instrument covering **24-hour recall**



Instrument repeated up to **5 times** between 2009-2012

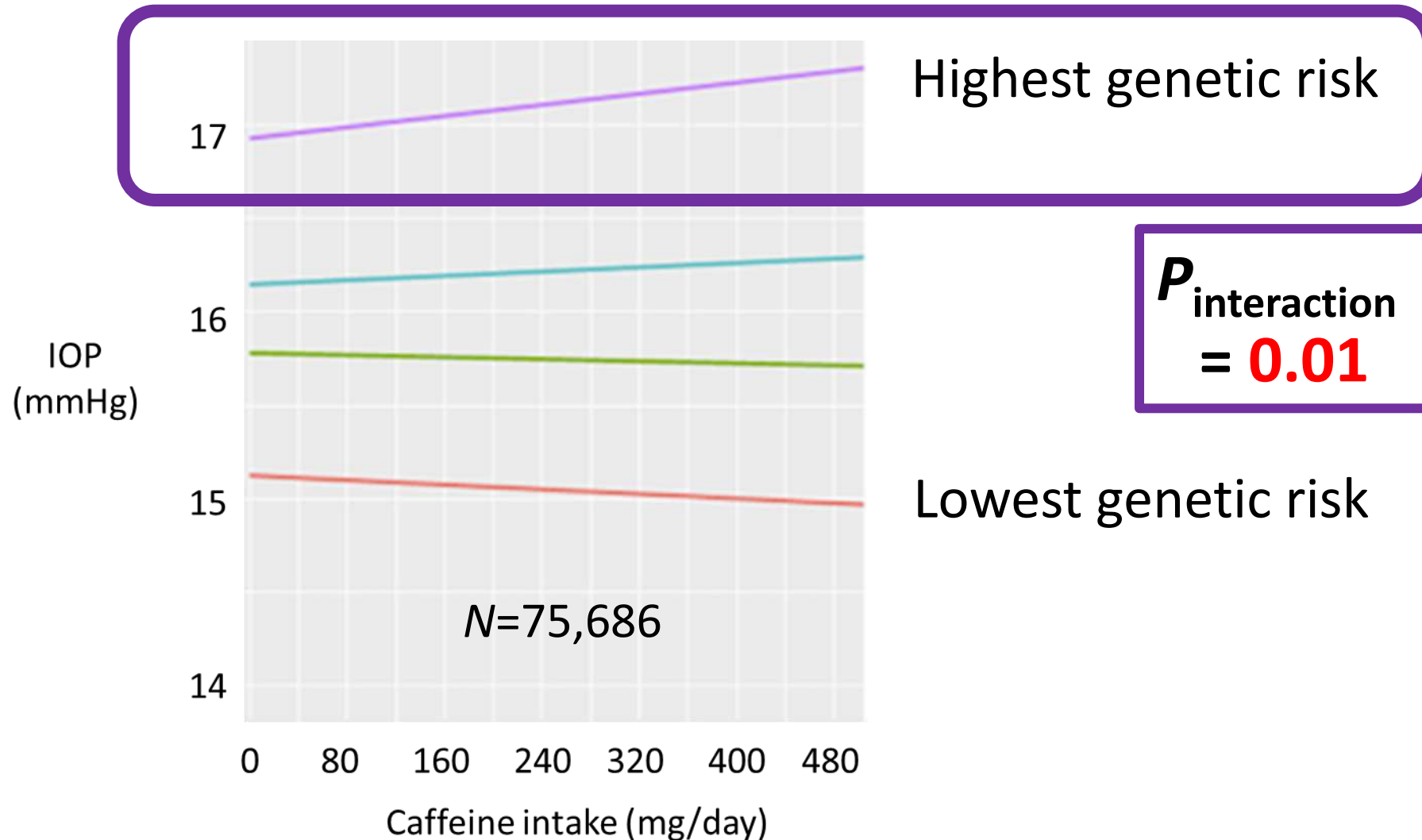


19 questions on caffeine containing foods and beverages

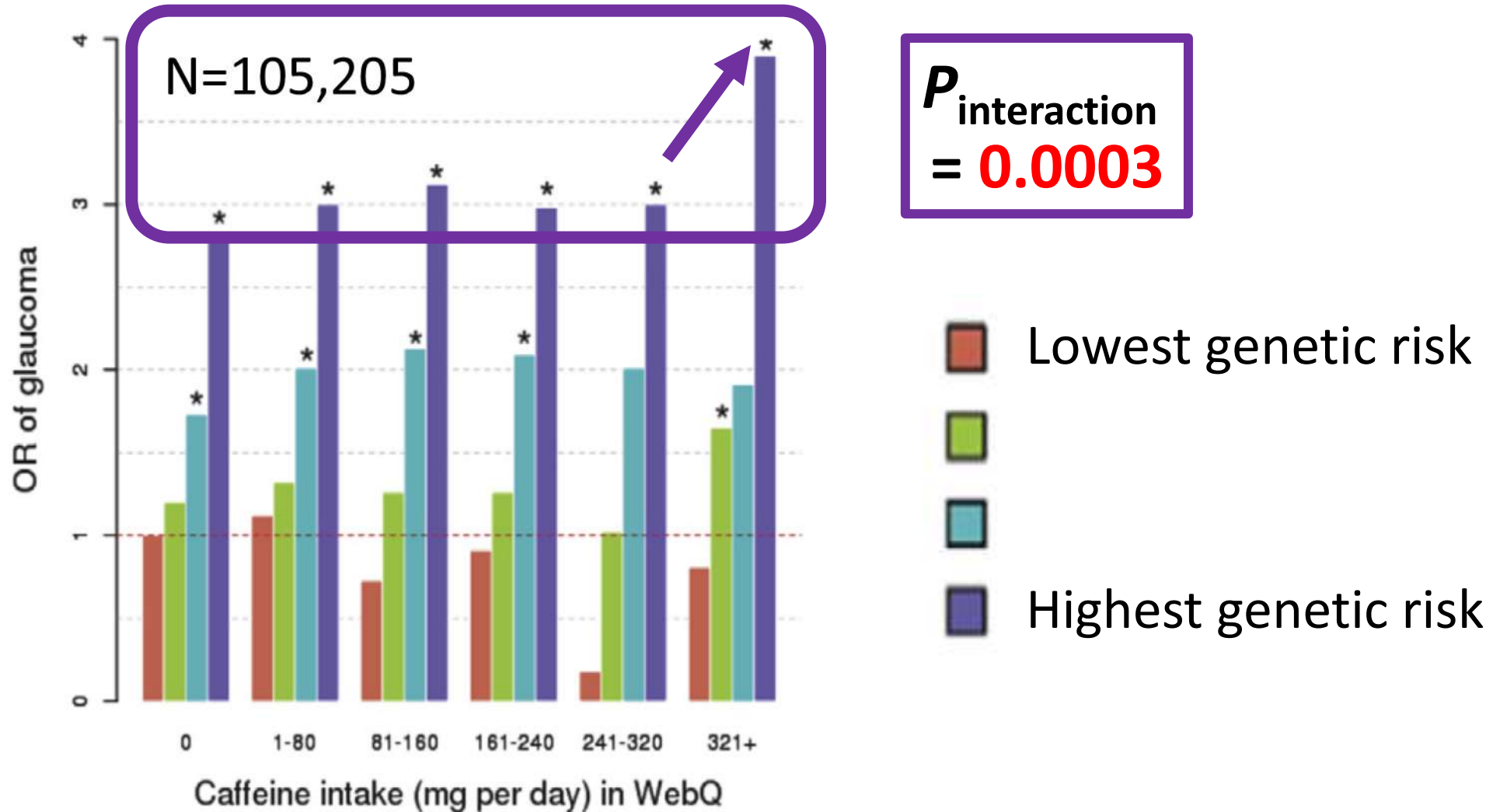


Total caffeine consumption calculated using published reports on caffeine content.

Caffeine and eye pressure



Caffeine and glaucoma





Glaucoma

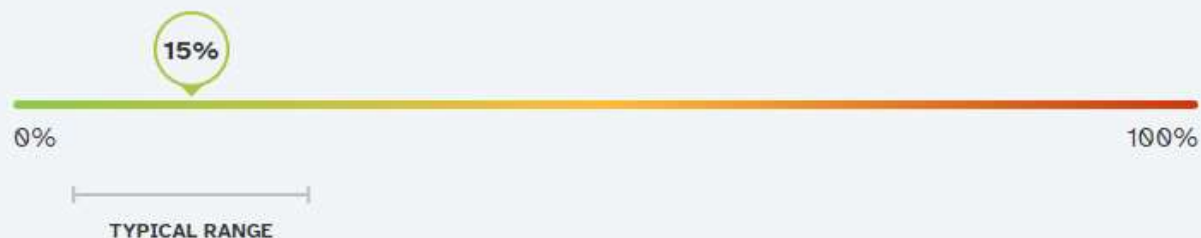
 POWERED BY 23ANDME RESEARCH

Glaucoma is a group of eye conditions that occurs when the nerve in the back of the eye (called the optic nerve) is damaged, most commonly due to high eye pressure. Optic nerve damage can cause gradual vision loss that may start as blind spots or loss of side (peripheral) vision and worsen over time.



Anthony, your genetic result is associated with a **typical likelihood** of developing glaucoma.

An estimated **15%** of people with genetics and other factors like yours develop glaucoma **by their 80s**. This is based on data from male 23andMe research participants of Northern African/Central & Western Asian descent.



This estimate is based on currently available data and may be updated over time.

Ways to take action

Experts agree that for people who have glaucoma caused by high eye pressure, it is important to reduce eye pressure to slow vision loss. Evidence suggests that healthy lifestyle habits may help manage eye pressure.

- Get regular, moderate exercise. Some exercises may increase eye pressure, like inverted yoga positions or headstands. Talk to your doctor about an appropriate exercise program.
- Wear eye protection to avoid injuries, such as when using power tools or playing racket sports.
- Limit caffeine consumption.

Talk to your doctor if you notice any changes to your vision, such as reduced side (peripheral) vision or blind spots. If you have concerns about glaucoma, talk to a healthcare professional about whether getting regular dilated eye exams is right for you. You may be referred to an eye specialist, such as an ophthalmologist.



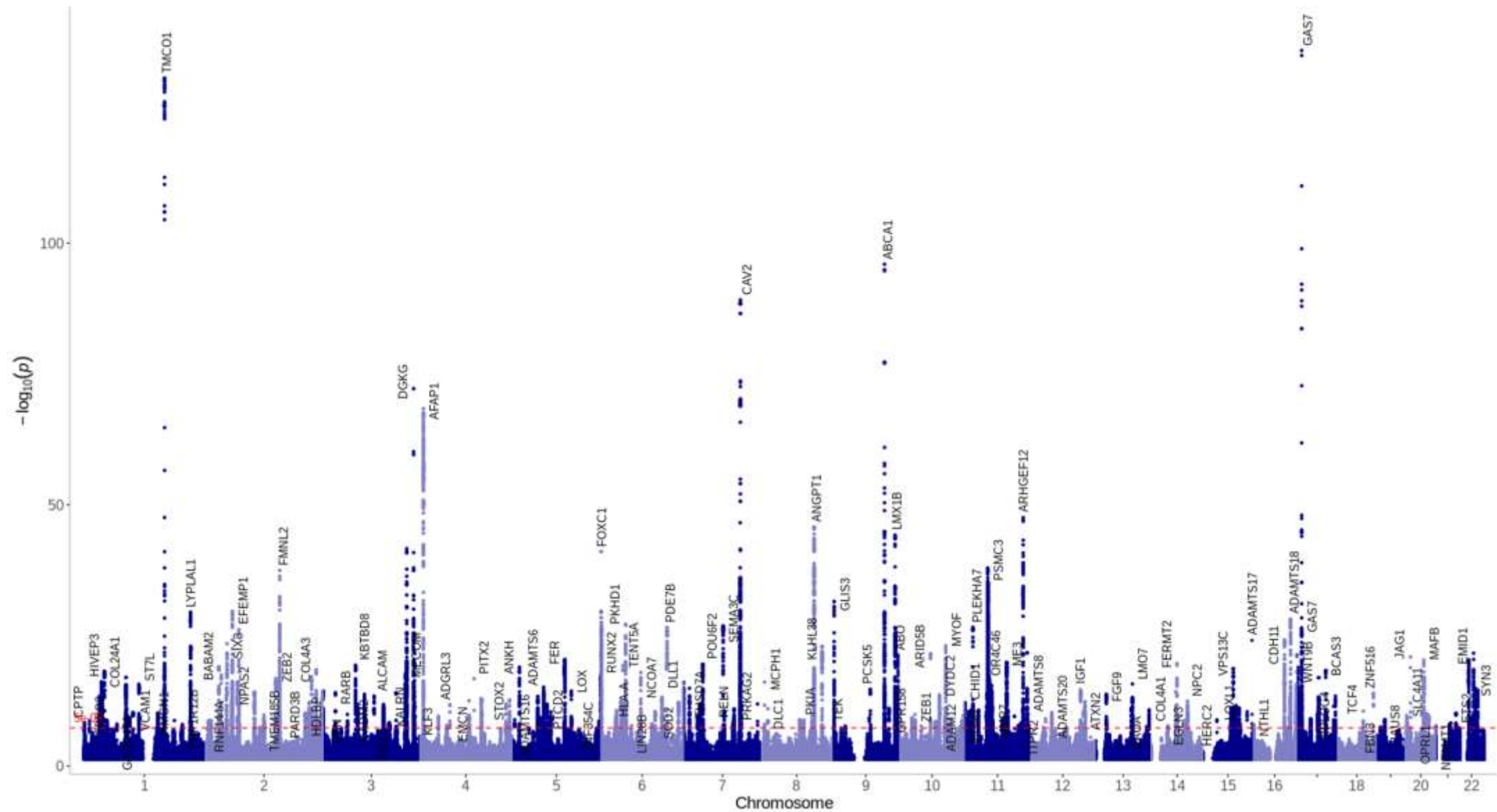
+
Our
Future
Health



£20/p

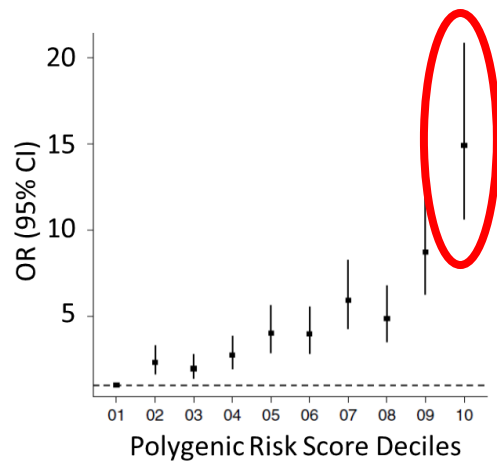
**5 million
adults**



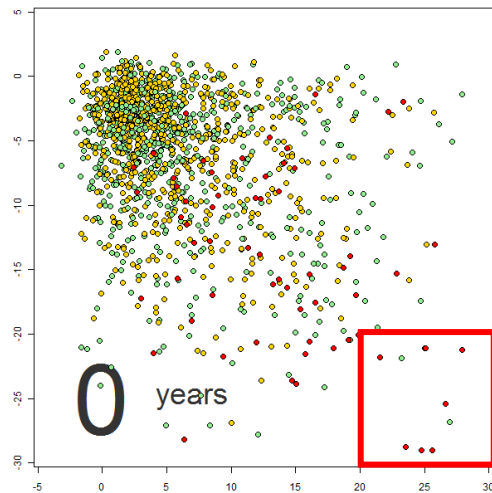


Genomics for personalised glaucoma care

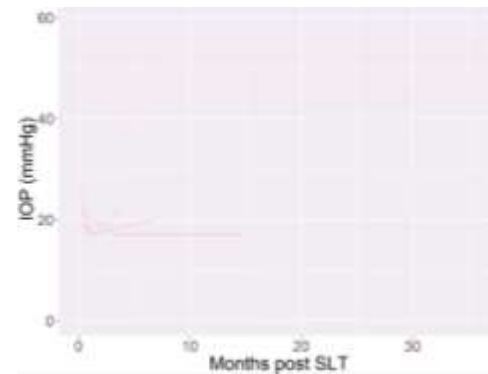
Population screening



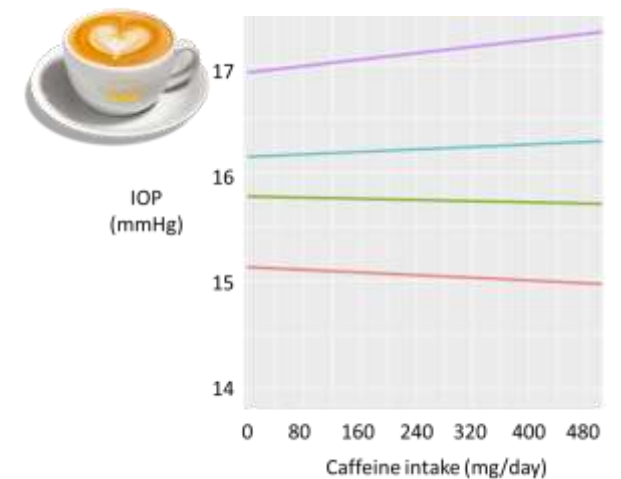
Risk stratification



Personalised treatment



Personalised lifestyle advice





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UCL Institute
of Ophthalmology

Thank you



UK Research
and Innovation

NIHR | Moorfields Biomedical
Research Centre



Moorfields
Eye Charity



 **The Lister Institute**
of Preventive Medicine



Any questions?



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Thank you for attending Moorfields Annual General Meeting

**The presentation and a recording of the meeting will
be posted on www.moorfields.nhs.uk**



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