

Welcome to Meeting 2018

- Visit the stalls
- Post on social media about today
Use **#ANUKconf**
Twitter: @AniridiaNetUK
- Sessions begin here at 10:00am

Please silence mobile devices



Housekeeping

- Fire exits and marshalls
- Toilets and baby changing
- Lunch at 12:00
- Break at 3:00pm
- Folder contents and slides are online
- Wifi Network: RFLPublic
 Password: Welcome123

Recording & Publishing

- There will be photographers and videographer.
- We will be posting words and pictures on social media
- We hope to be streaming live on the internet
- Consent forms have been filled in and will be assumed unless you tell us otherwise

Share

Post on social media about today

Use **#ANUKconf**

Twitter: @AniridiaNetUK

Facebook: @Aniridia Network UK



Aniridia Network

Keynote: Research into
possible drug treatments

Dr Mariya Moosajee

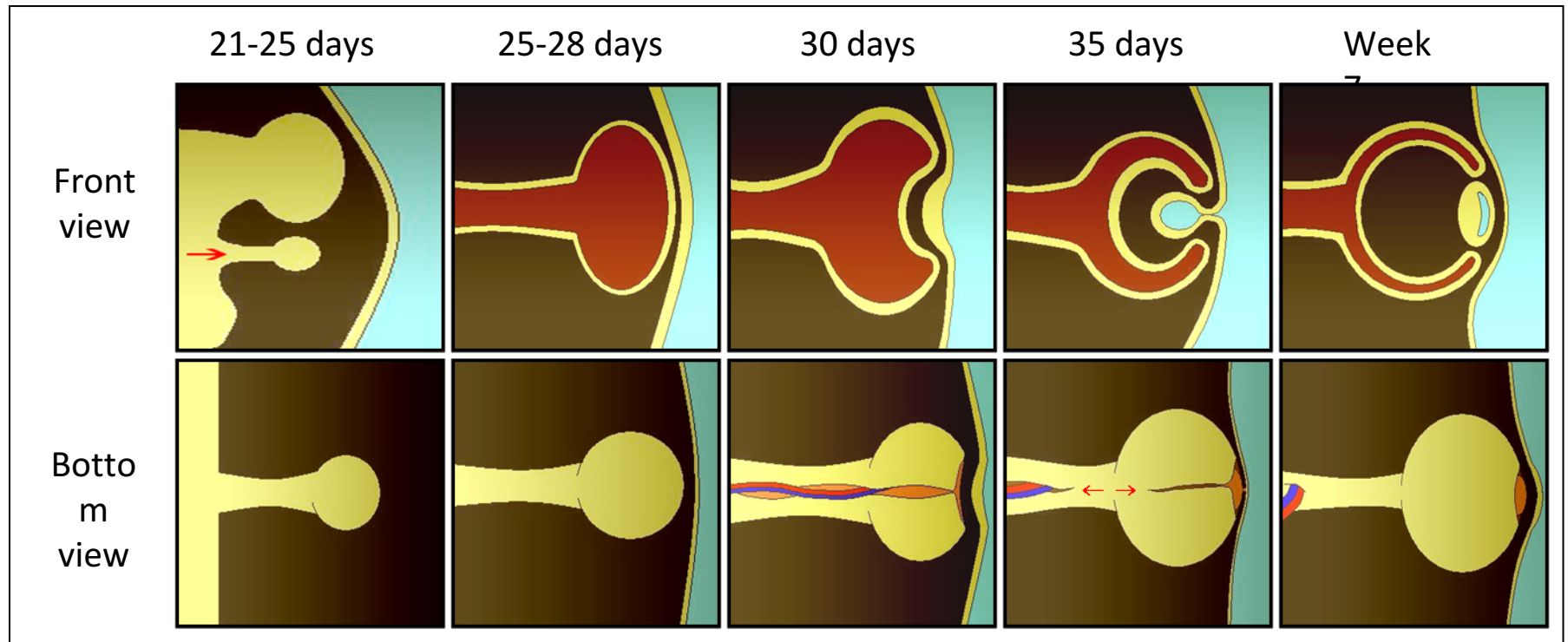
Web: aniridia.org.uk
Email: info@aniridia.org.uk

Twitter: [@AniridiaNetUK](https://twitter.com/AniridiaNetUK)
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Drug treatments for aniridia

Dr Mariya Moosajee MBBS BSc PhD FRCOphth
Consultant Ophthalmologist and Senior Clinical Lecturer
Moorfields Eye Hospital, Great Ormond Street Hospital for Children
and UCL Institute of Ophthalmology

Human eye development



Ocular maldevelopment

Causes >30% childhood visual impairment and blindness worldwide

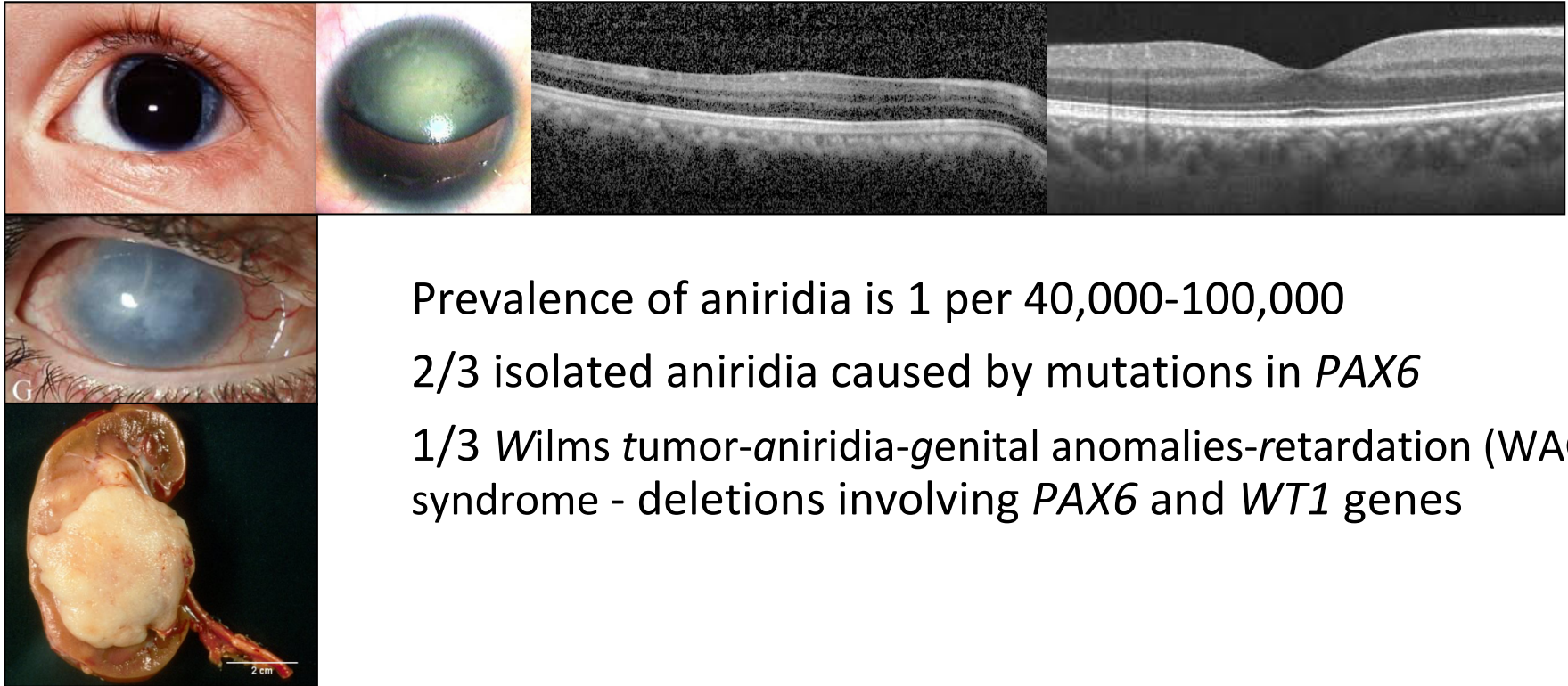
Most common cause of childhood sight impairment certification in UK

Includes:

- Anterior segment dysgenesis
- Congenital cataracts
- Primary congenital glaucoma
- Structural globe anomalies



Aniridia

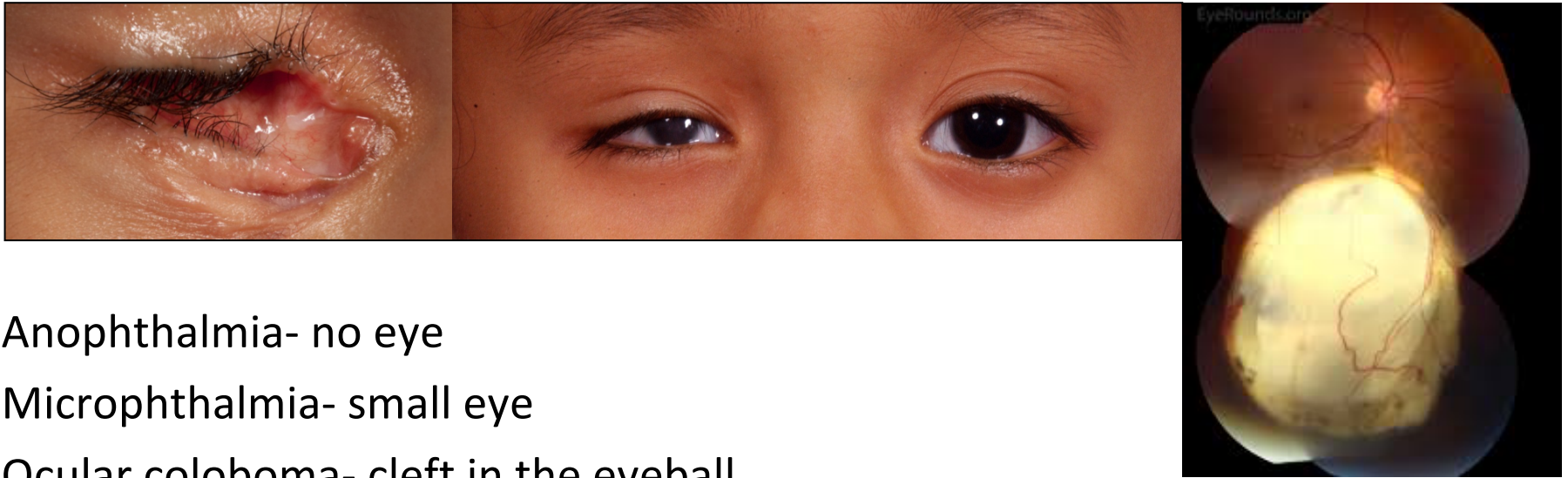


Prevalence of aniridia is 1 per 40,000-100,000

2/3 isolated aniridia caused by mutations in *PAX6*

1/3 Wilms tumor-aniridia-genital anomalies-retardation (WAGR) syndrome - deletions involving *PAX6* and *WT1* genes

PAX6 additional clinical features



Anophthalmia- no eye

Microphthalmia- small eye

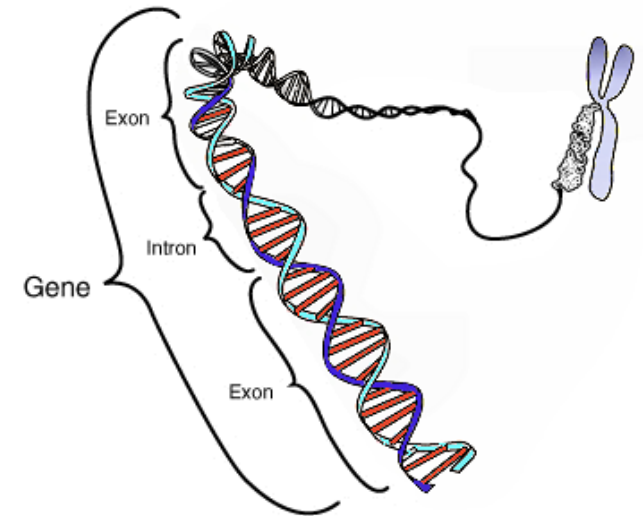
Ocular coloboma- cleft in the eyeball

Nystagmus with no iris defects

Genomic medicine

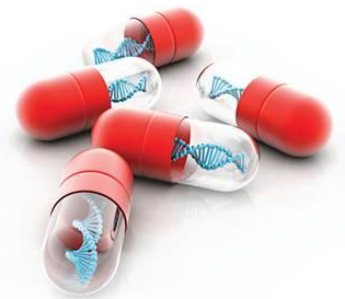
“An emerging medical discipline that involves using genomic information about an individual as part of their clinical care e.g. for diagnostic or therapeutic decision-making.”

Whole genome sequencing:
Screens all 3 billion letters of your genetic code
including 20,000 genes



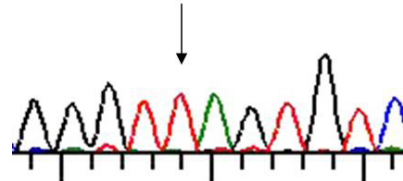
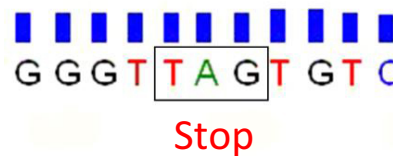
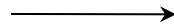
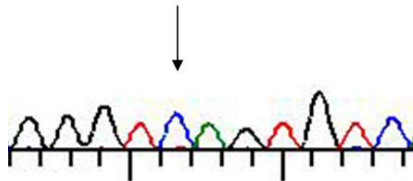
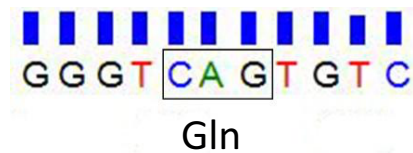
Benefits and application for aniridia

- Increased diagnostic yield
- Genetic modifiers- why some patients have more severe disease than others
- Prediction and prevention of associated disease e.g. *WAGR*, learning/behavioural/obesity
- Personalised medicine- based on genetic changes
- Pharmacogenomic profiling- response to drug treatment

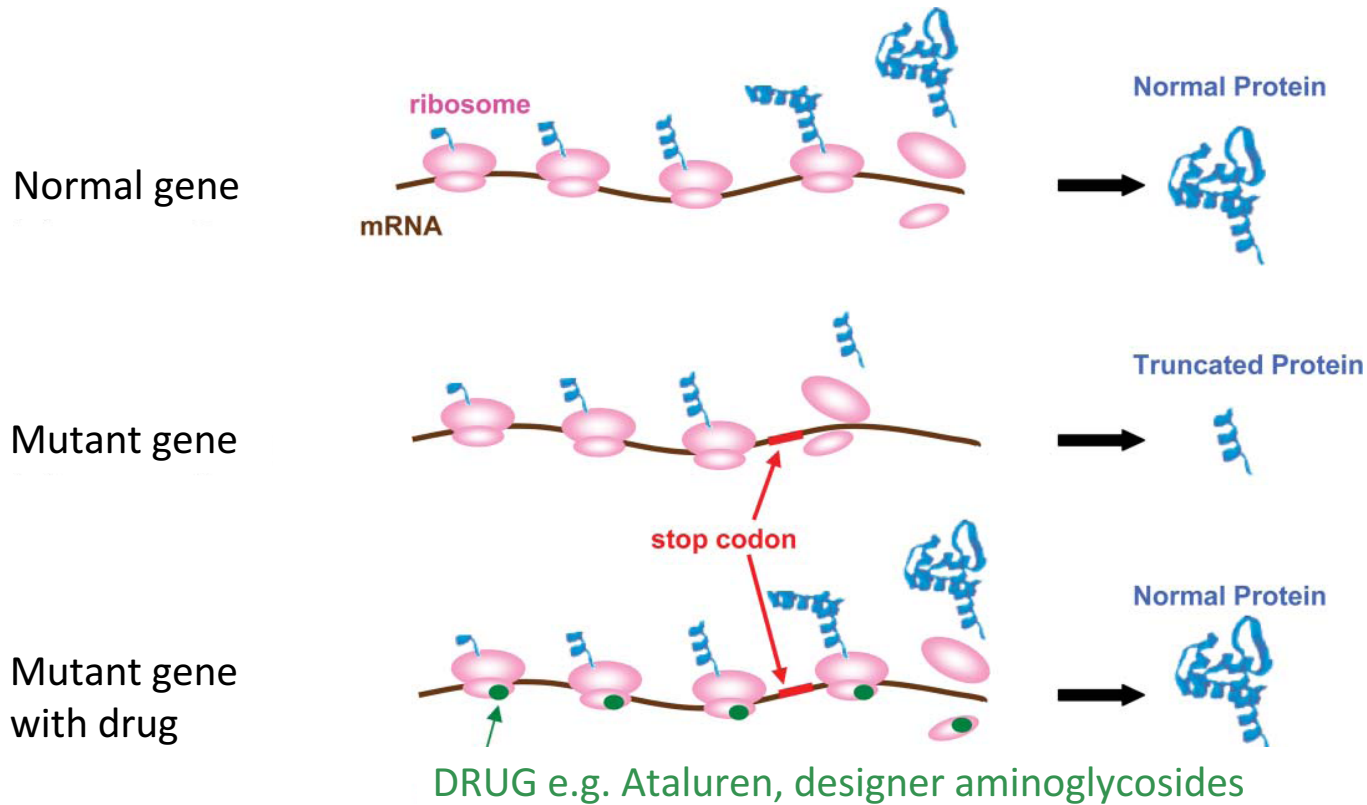


Nonsense suppression therapy

- Works on nonsense mutations- causes up to 70% genetic disease
- Disease- and gene- independent
- 40% of aniridia mutations



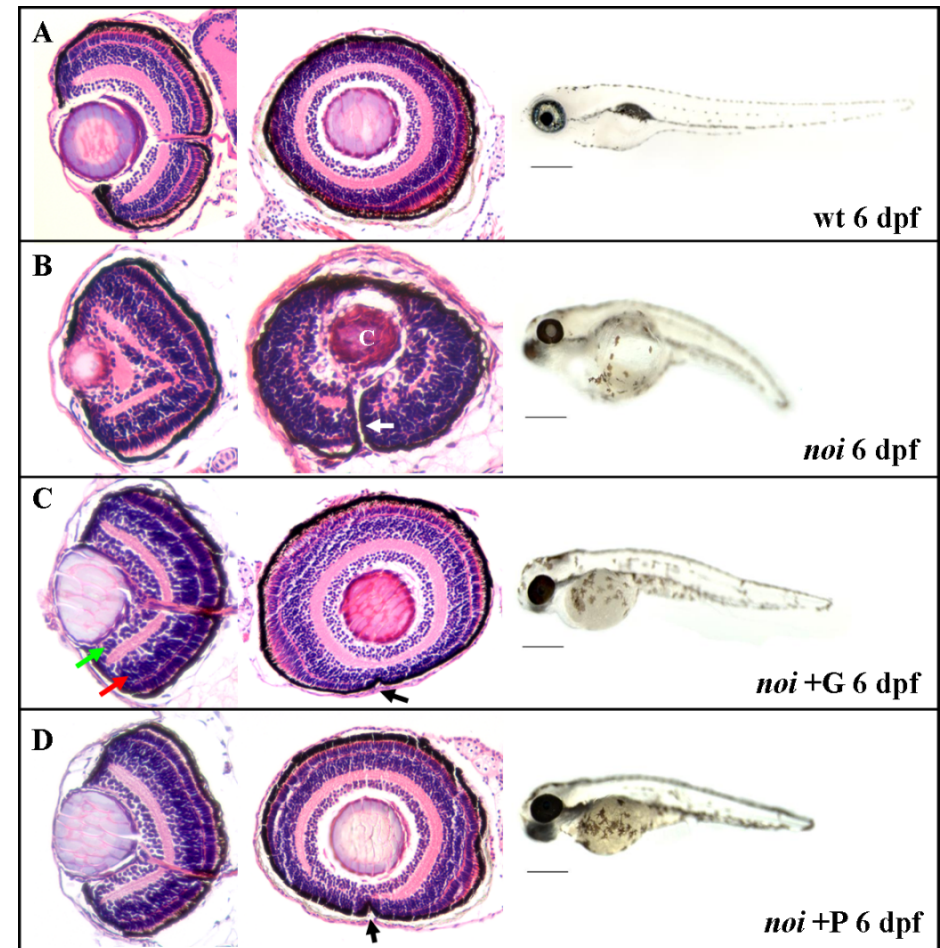
Nonsense suppression therapy



Treatment of ocular maldevelopment

Aminoglycosides for *PAX2* renal coloboma syndrome

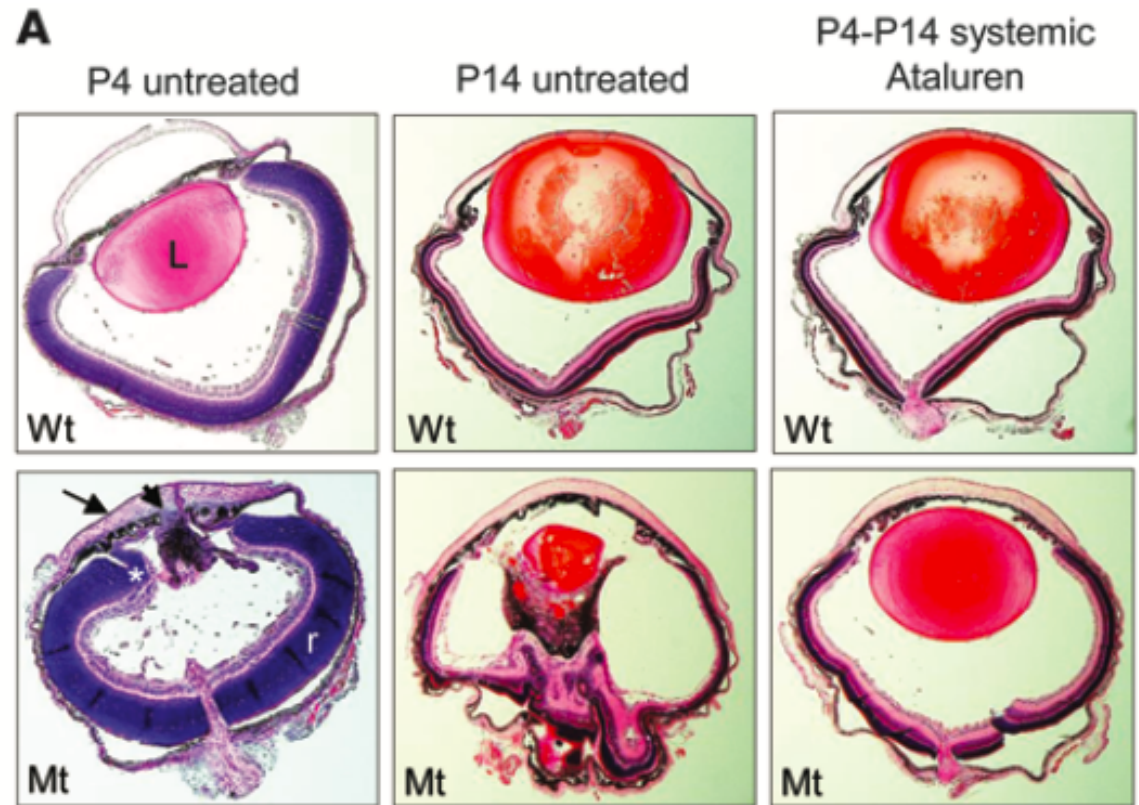
- *pax2*^{-/-} zebrafish
 - Ocular coloboma
 - Microphthalmia
 - Tx Aminoglycosides



Moosajee M *et al* Hum Mol Genet. 2008

Ataluren for *PAX6* Aniridia

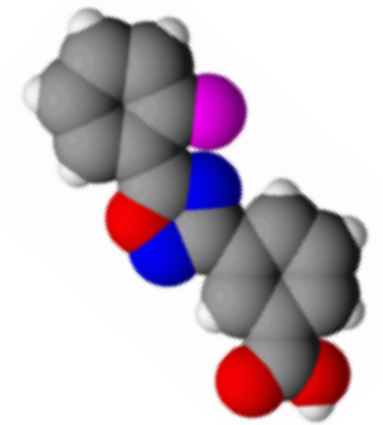
- *Pax6*^{-/+} Sey mouse
 - Aniridia
 - Post-natal treatment with ataluren
 - Developmental plasticity



Gregory-Evans CY *et al* J Clin Invest. 2014

Ataluren (PTC124)

- Large evidence base for readthrough of nonsense mutations in many animal and cellular disease models
- Phase 3 clinical trials for cystic fibrosis and Duchenne muscular dystrophy
- Safe and tolerable in children (>2 years) and adults
- Taken orally three times a day
- Minimal side effects- nausea, transient diarrhoea

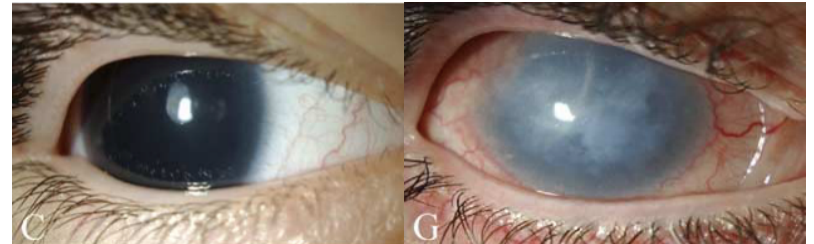


NICE approval in UK for Duchenne muscular dystrophy

- None of the children taking the drug lost the ability to walk over the 48 weeks of the trial compared with 8% on the placebo (0 out of 47 compared with 4 of 52).
- Predicted ataluren may delay loss of walking for up to 7 years.
- Patient experts said they had seen meaningful stabilisation or improvements in their child's mobility such as being able to get into and out of bed independently and go to school.
- If a child loses the ability to walk, greater deterioration follows requiring self-feeding, showering and going to the toilet. Delaying loss of walking could allow retention of normal adolescence.
- Treatment costs £220,000 per year.

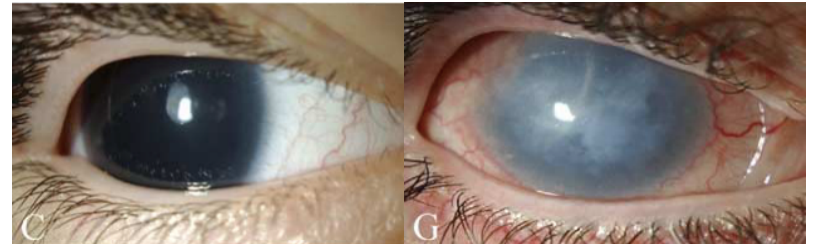
Phase 2 clinical trial for PAX6 nonsense-Aniridia

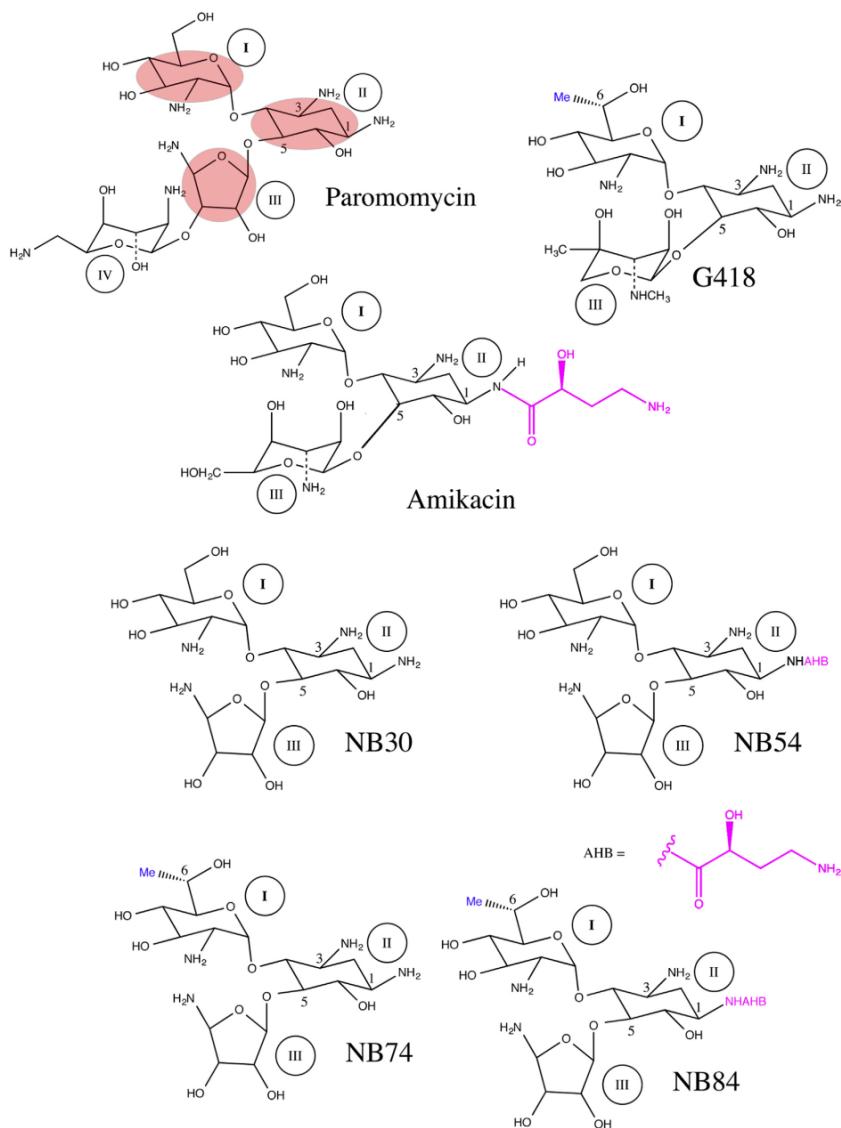
- Randomized, double-masked, placebo-controlled study
- Placebo/Drug for 48 weeks followed by open-label ataluren for another 48 weeks
- Oral dose of 10 mg/kg in the morning, 10 mg/kg at midday, and 20 mg/kg in the evening



Phase 2 clinical trial for PAX6 nonsense-Aniridia

- Primary outcome
 - Safety- adverse effects- body and eye
- Secondary outcome
 - Visual acuity
 - Corneal surface
 - Iris area
- End date Dec 2019- results in 2020

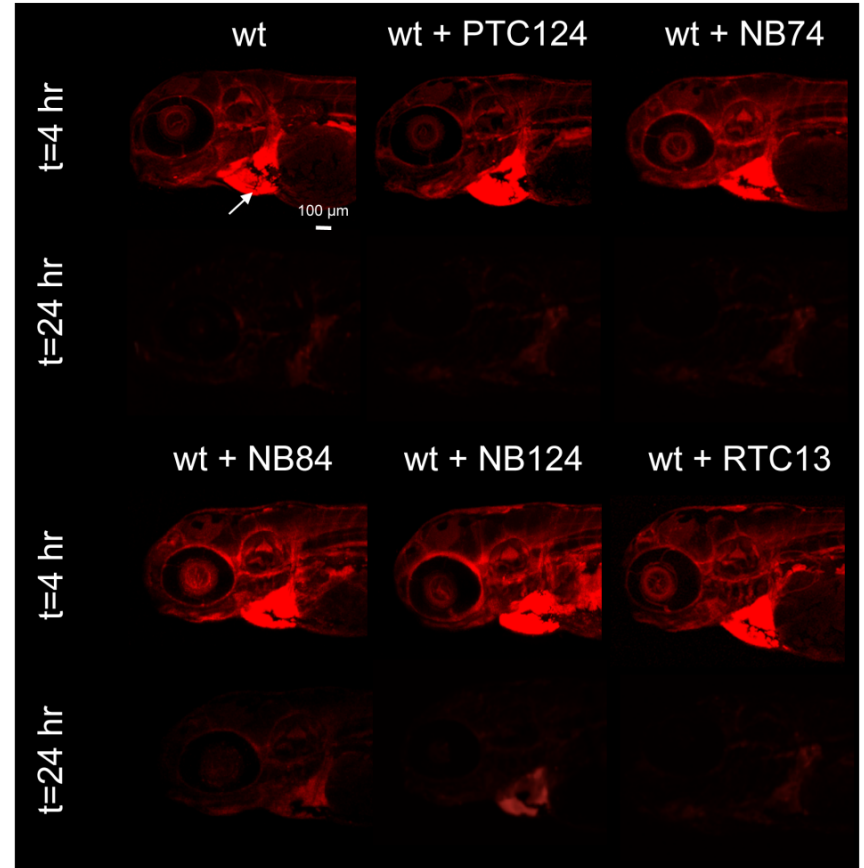
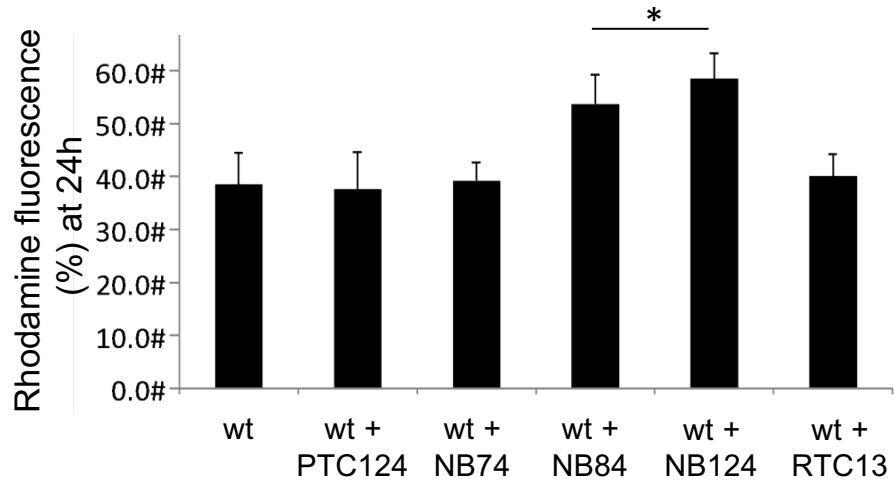




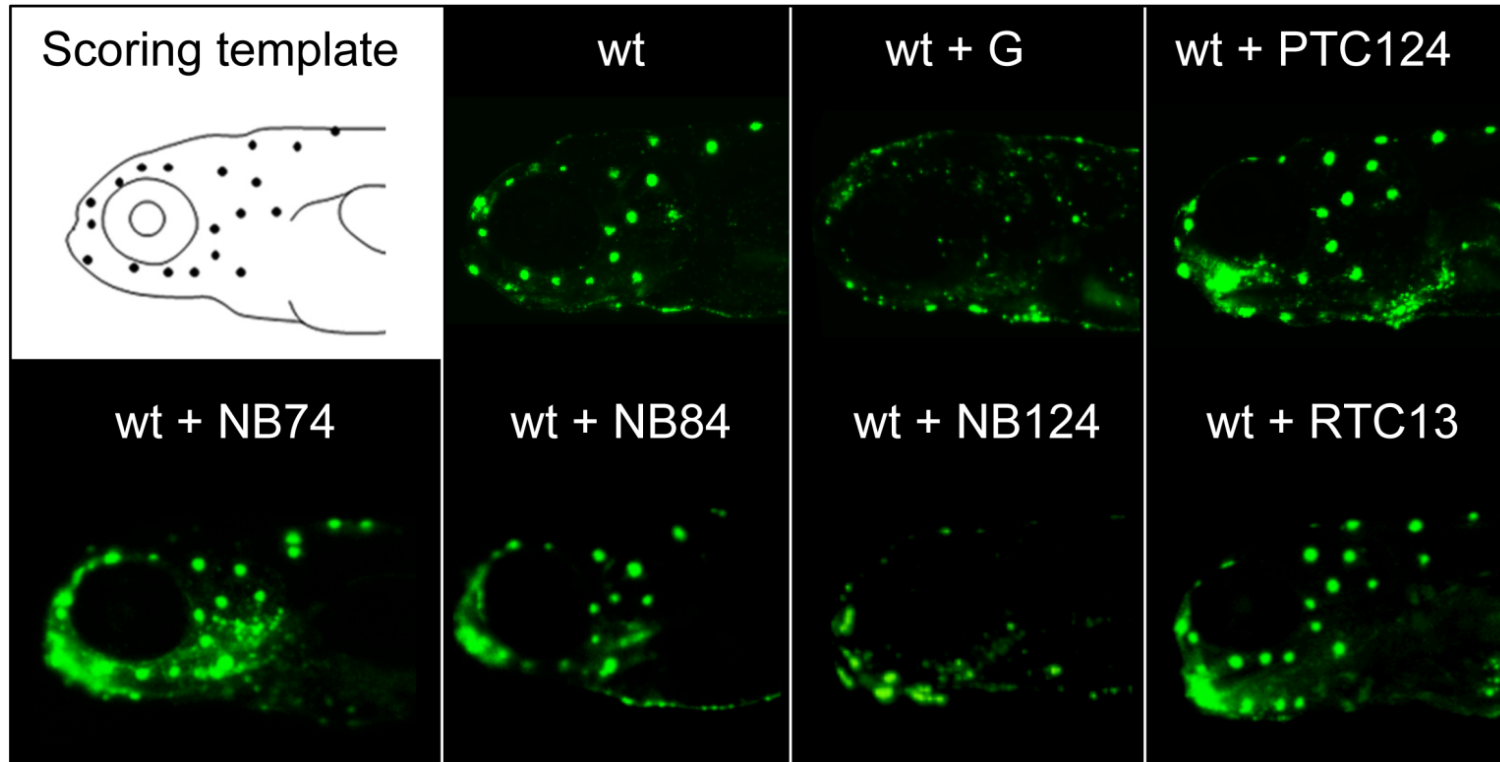
Other nonsense drugs

- My research group has tested a number of small molecule drugs
 - Designer aminoglycosides – NB74, NB84, NB124
 - Readthrough compounds – RTC13, RTC14
 - PTC compounds- PTC414

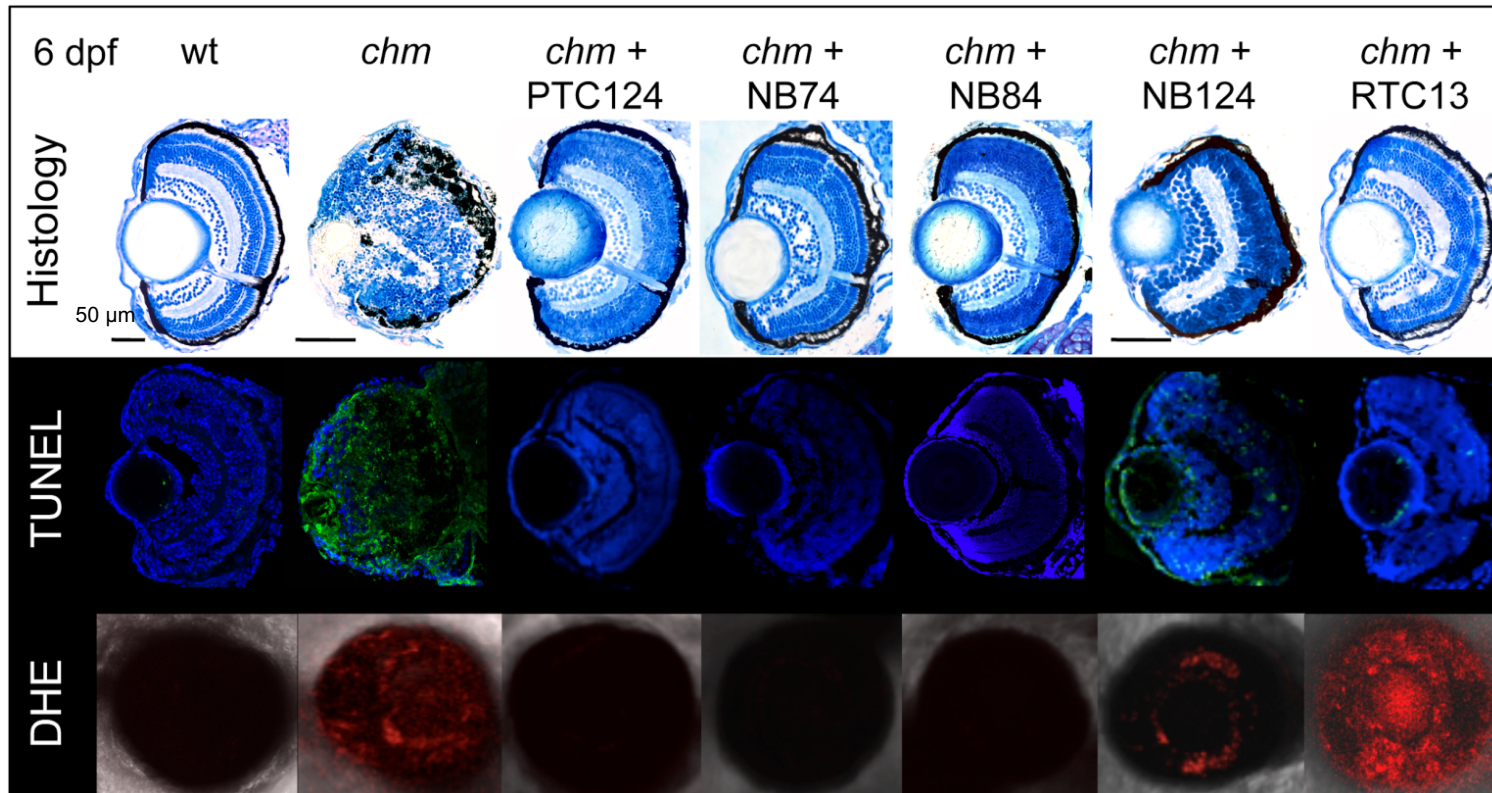
Safety screen using zebrafish- Kidney function



Safety screen using zebrafish- Hearing

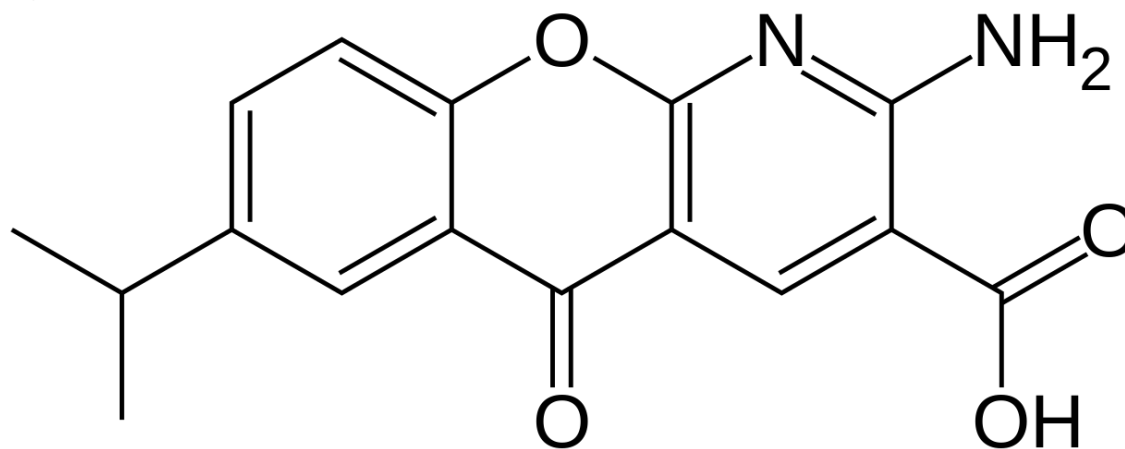


Drug screen using zebrafish- Eyes



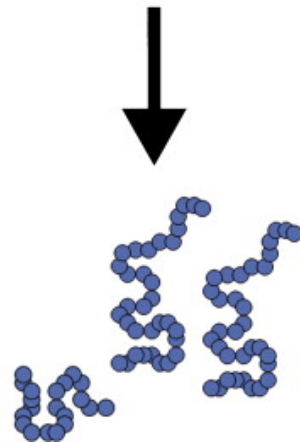
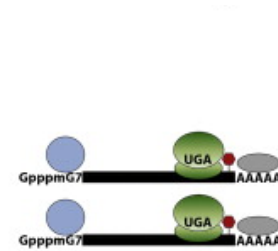
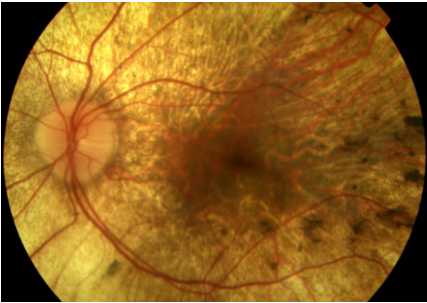
New candidate: Amlexanox

- FDA approved
- 20 years use for asthma and oral ulcers
- High-throughput drug screen
- Works on CF patient cells
- Repurpose if successful

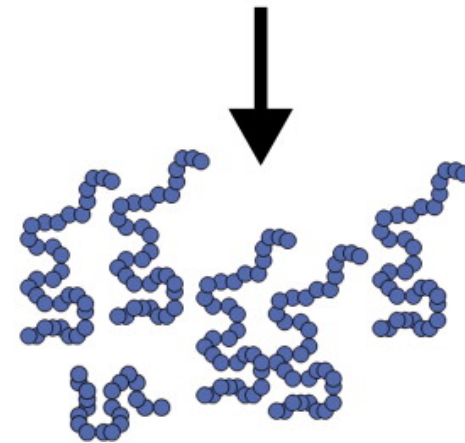
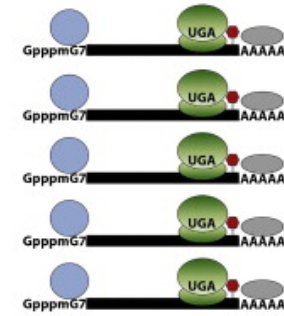


Amlexanox

- Dual
action

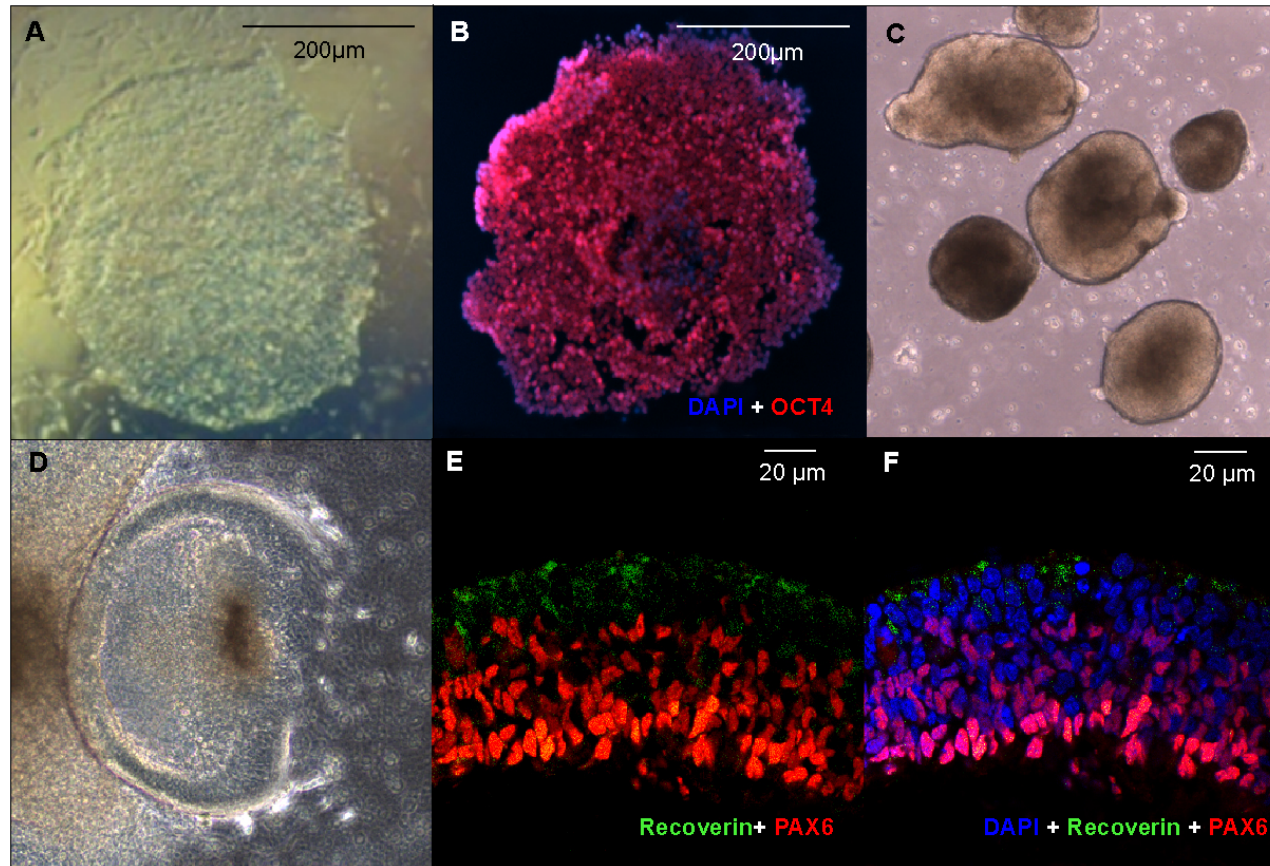


Ataluren:
nonsense
suppression

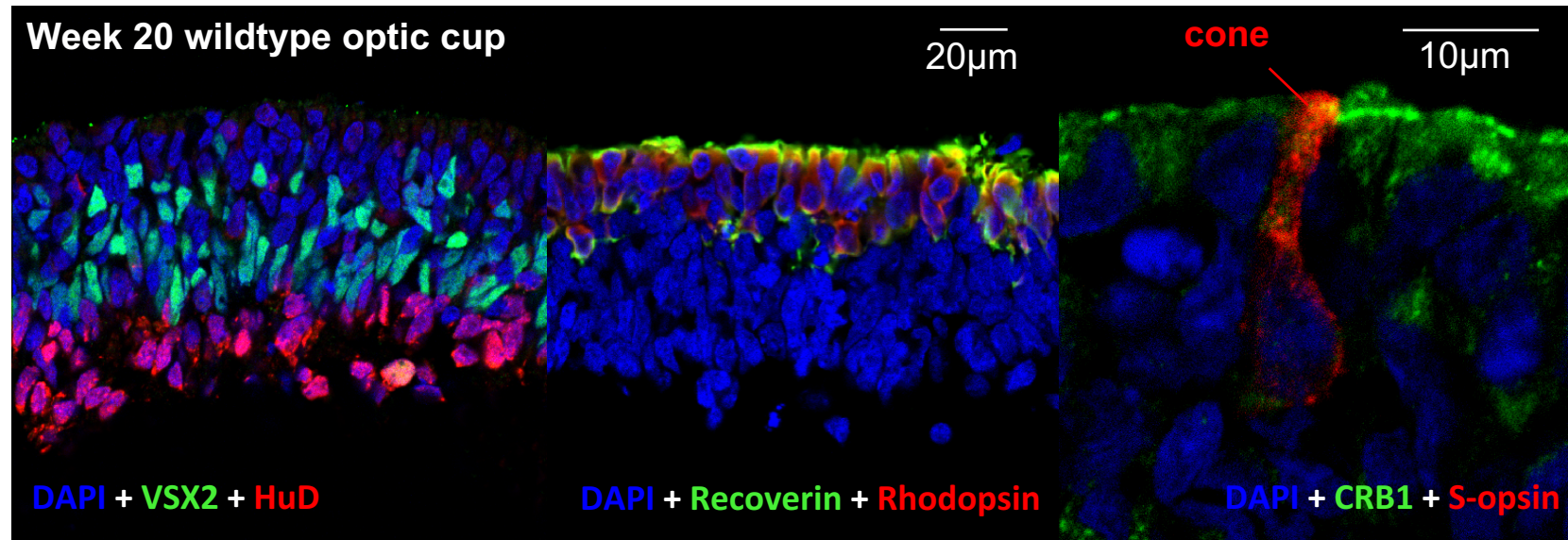


Amlexanox: nonsense-mediated
decay inhibitor and nonsense
suppression

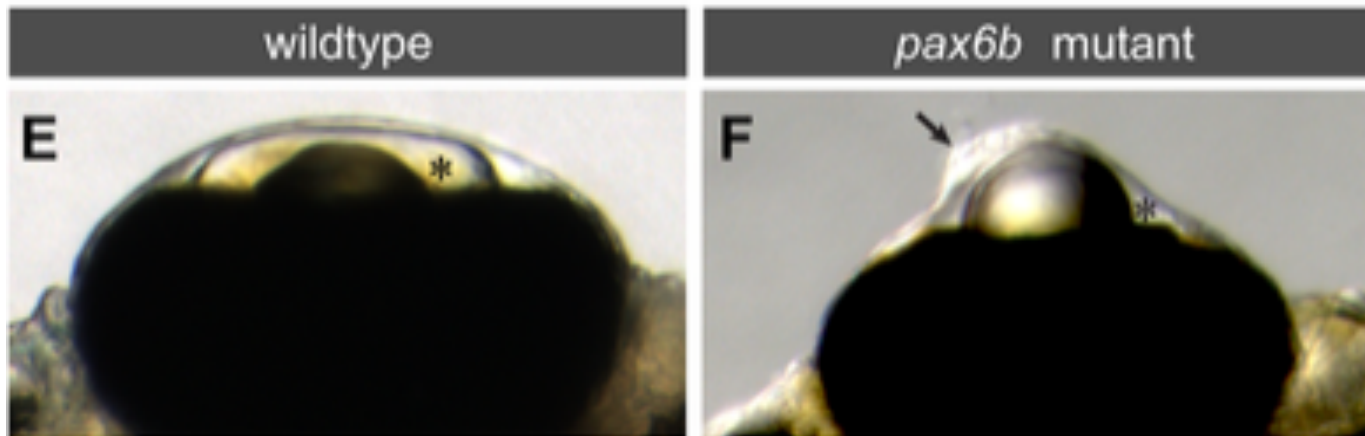
Disease models for aniridia- patient stem cell derived eye cups



Patient stem cell derived eye cups with light-sensing cells

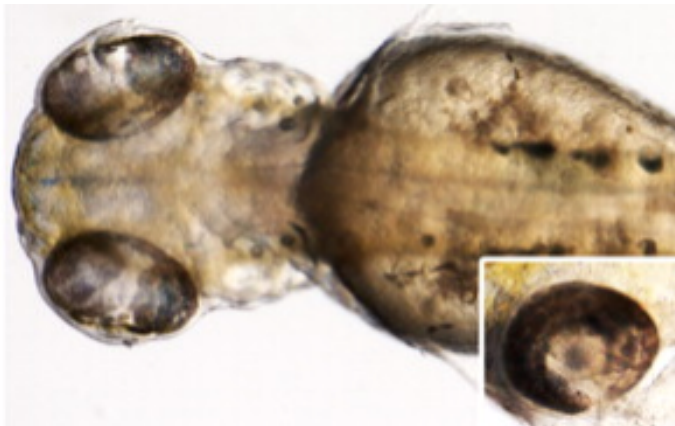


Disease models for aniridia- zebrafish

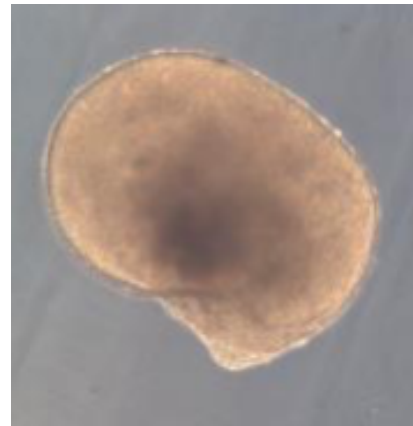


Takamiya et al PLOS One 2015

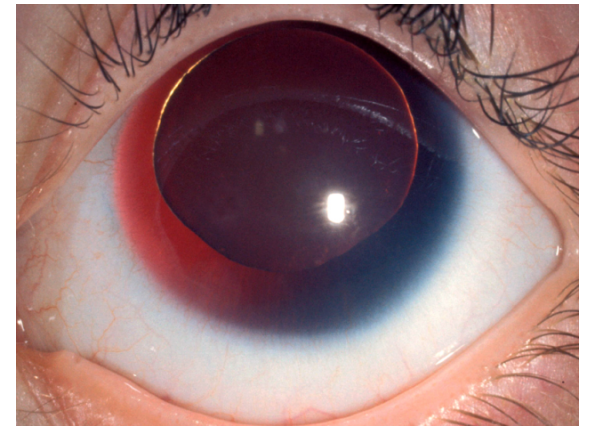
We can use these models to identify new targets and test therapies



High-throughput drug screen



Candidate compounds



Natural history and clinical trials

How can you get involved...

- Establish your genetic diagnosis
 - Helps develop therapies
 - Genetic counselling and family planning
 - Multidisciplinary care
 - Happy to see you at Moorfields- GP referral



- Participate in natural history studies- outcome measures for trials
- www.clinicaltrials.gov
- Fundraising

Conclusions

- Phase 2 clinical trial for ataluren underway- results in 2020
- Developing patient derived PAX6 disease models to further our understanding of aniridia
- Screening approved drugs may allow repurposing and faster translation - focus on amlexanox
- Please get involved and support research, treatments are becoming a reality



**Aniridia
Network UK**

Acknowledgements

Dr Rose Richardson Collaborators:
Dr Andreas Mitsios Prof Pete Coffey
Dr Nick Owen Prof Andrew
Webster
Maria Toms Prof D Fitzpatrick
Dhani Tracey-White Ms M Hingorani
Kishan Khambhaita Prof V Van Heyningen
Hajrah Sarkar Prof Steve Wilson



Thank you



**Aniridia
Network UK**

If you have any questions- please email me on:

mariya.moosajee@moorfields.nhs.uk



Aniridia Network

Keynote: Research into
possible drug treatments

Dr Mariya Moosajee

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Aniridia Network

Aniridia Network UK Annual General Meeting 2015

By ANUK Trustees

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Agenda

- [ANUK AGM minutes 2016](#) to be agreed
- Matters arising.
- Reports & Accounts.
 - Questions to officials
- Appointment of charity trustees
- Consultation on the foundation model for governance of the charity
- Any other business



Aniridia Network

Aniridia Network UK Annual Reports 2016/17 & 2017/2018

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Aniridia Network is registered charity 1176792 in England & Wales, operating as
Aniridia Network UK, 22 Cornish House Adelaide Lane Sheffield S3 8BJ

Charity registration

- At the 2015 AGM members voted to dissolve Aniridia Network UK unincorporated association (ANUK UA), and create a new one to allow us to register as a charity
- Implementation was delayed until autumn 2017 and there was no AGM in 2017 partly due to the handover
- Aniridia Network, a Charitable Incorporated Organisation (CIO), was created and registered by the Charity Commission for England and Wales in January 2018 by the incumbent trustees
- ANUK UA will be formally dissolved and the assets transferred to AN CIO by the trustees following its inaugural AGM

Management Roles

- James
 - Chief Executive
 - Head of Communications and Membership
- Katie
 - Head of Services and Information
- Emily
 - Head of Finance and Funding
- Martin
 - Head of Volunteering and Development

Services & Information

Katie Atkinson

Befriending 2016-17

- 13 individuals/families made contact with the Befriending Scheme or were contacted when they became members.
- 3 did not respond to emails.
- 1 preferred to continue to make links through Facebook.
- 6 were successfully linked to other families
- 3 requested links to Education support or other services

Befriending 2017-18

- 19 individuals/families made contact with the Befriending Scheme or were contacted when they became members.
- 4 did not respond to emails.
- 1 preferred to continue to make links through Facebook.
- 5 were successfully linked to other families
- 8 requested links to Education or other support
- 1 referred by a teacher for VI
- Many families now make contact through the Aniridic Family group on Facebook, which may be why we have fewer requests for the Befriending Scheme.

Education 2016-17

- We have aniridia pupil passport templates for children aged 0-18 years
- We produced Aniridia 0-16 Months VI Guidance for Practitioners, based on the Department of Education Early Years Framework.
- We had 3 requests for information e.g. Pupil Passports, Links to websites and Early Years Support.

Education 2017-18

- We produced a New Parent Information Sheet, giving links to a range of services and resources that support children with VI.
- In response to a grandparent asking for advice about suitable toys, we posted a blog entitled: Toys for visually impaired children.
- We have had 7 requests for information e.g. Pupil Passports, links to websites, Education & Health Care Plans (EHCP) and Early Years Support.
- Posts on the Aniridic Family group on Facebook, are contacted via email, if appropriate, and offered further support.

Enquiries

- 12 enquiries received in 2016-17
- 11 enquiries received in 2017-18
- Advice provided on a range of topics e.g. cataract and cornea operations, risk of inheriting aniridia, APD and assistance with applying for disability benefits
- Thanks to Melanie Higorani (Medical Adviser) and Veda-Jane Petre (Disability Rights Adviser) for their assistance

Aniridia Europe

- Attended the 3rd European Aniridia Conference was held 27th-28th August 2016, Duisburg, Germany
- Preparations are being made for the 4th European Aniridia Conference which will take place 24th-26th August 2018, Paris, France
- Sent representatives to several ophthalmology conferences across Europe. Katie and James represented AE and ANUK at the European Paediatric Ophthalmological Society Conference 2017 in Oxford, UK.
- Promoting the European Reference Network for Rare Eye Diseases and participating as a patient representative.



Patron's Report

- Attended 3rd European Aniridia Conference in Duisberg
- Suggested registry where to record details of the progress of their eye condition, as well as their PAX6 mutation.

This could help establish closer links between genetics and outcomes.

What do members think?

Registry

- Maintaining the PAX6 mutations database
 - Knowing your mutation may become useful when genetic therapies become available.
 - At MRC HGU in Edinburgh, we (Dr Isabel Hanson) have been maintaining a database of all reported PAX6 mutations for many years. There are >1000 individuals entered.

http://lsdb.hgu.mrc.ac.uk/home.php?select_db=PAX6

- Dr Mariya Moosajee discussed the need for this type of information earlier today



- Still working on new PAX6 mutations at Edinburgh is ophthalmologist /scientist Nikki Hall, who is here today



Rare Disease Day

- Shared Rare Disease Day 2017 and 2018 campaigns on social media
- [@PourHouseShrews](#) sold a awesome themed milkshake whose price included £1 donation



Events

- Conference 2016
- London meet up 2016
- London meet up 2017
- Ireland Conference 2017



Conference 2016

- Wallington, South London in May
- Organised largely by member Mary Cox and family
- 70 adults and 16 children attended
- Included an exhibition, food and crèche
- Sessions:
 - Morning - VI cricket and tandem riding
 - Afternoon – Presentations including:
Mobility, Living with aniridia, Blind Children UK, Living Paintings and 'Life, campaigning & politics & aniridia' by Emily Brothers
 - Evening – social event
- 60/40% very satisfied and great feedback comments on the impact the event has.

Ireland 2017

- First meeting in Ireland
- Held in at NCBI in Dublin in April
- 30 attended.
- Presentations and Q&A including via Skype:
 - President of Aniridia Europe, Rosa Sanchez de Vega
 - Ophthalmologist Barbara Kasmann-Kellner
- Plenty of time for chatting and networking.
- Organised by Eleanor Burke

Finance & Funding

Emily Nash

Finances 2016/2017

Opening Balance - £12,010.04

Closing Balance - £22,350.03

- Advance ticket sales for the May 2016 conference came to £2,650.37. Costs for the May 2016 conference totalled £830.10.
- £6,056.13 was deposited on 15 March 2017 as a closing balance from ANUK COOP bank account.
- Income from donations for the year was £1,134.13. The board had agreed to focus on increasing this for the next financial year 2017/18

Finances 2017/2018

Opening Balance - £22,350.03

Closing Balance - £31,753.59

- £600 has been invested in increasing awareness of ANUK in the Republic of Ireland. (Dublin conference/Aniridia Day.)
- £9,872.86 has been received through donations and charitable fundraising. Two charity events (bike ride and boxing match) with Justgiving pages raised over 50% of this income.
- £420 was received as reimbursement for travel costs relating to the 2015 European Summer Camp.
- £919.52 was transferred from our PayPal account into the bank account (previous conference ticket sales)
- £700 was used to bring the Trustees together for strategic planning and for the NCVO trustee conference.
- £1,269 spent on conference to date, £410.50 received in ticket sales (remainder will be in 2018/2019 accounts)

Key Financial Activities

- Centralising Funds
 - Coop account closed
 - Lloyds account opened
 - Transfer of PayPal funds
- Preparation for becoming Aniridia Network
- QuickBooks
 - Allocation of income/ expenses
 - Accounts preparation
- Purchasing insurance
- Research funds (Fight for Sight)
- Budget for 2018/ 2019

Research grant

- First time we have made a grant
- Aniridia Network pledged £7,500 towards £15,000 grant administered by Fight For Sight's Small Grant Awards
- Closing date was 27 March 2018
- 5 applications received
- Bid topics included genetics, sleep and correcting refractive error
- Feedback will be collated in April on proposals and decision will be made by 15 May 2017



FIGHT FOR SIGHT

The Eye Research Charity

Budget for 2018-2019

- Conference Costs - £4,600
- Conference Equipment - £750
- Research - £7,500
- Estimated running costs - £2,500
- Reserves - £3000

Total Planned Expenditure - £18,350

- Conference Tickets - £2,710.50
- Donations - £1,880 (Direct Debit)

Total Expected Income - £4590.50

Communications & Membership

James Buller

Membership statistics

Category	2015/ 16	2016 -18	Year on year increase				% of estimated affected people in the UK
			2015-16		2016-18		
familial aniridia	143	160	9	7%	17	12%	
sporadic aniridia	135	157	19	16%	22	16%	
aniridia but origin unknown	88	89	4	5%	1	1%	
Total with aniridia	366	406	32	10%	40	11%	28%
Traumatic aniridia	2	2	0	0%	0	0%	
WAGR	36	40	2	6%	4	11%	60%
None/unknown	316	355	51	19%	39	12%	
Total	720	803	85	13%	83	12%	

There are an estimated that there are 1344 people with aniridia in the UK.

Membership statistics

- Membership over 2 years is roughly the same as in the previous single year.
- Only a fraction of people with aniridia in the UK have contacted ANUK. Of course many more may have accessed our information without us knowing.
- New members are usually families with new-born cases of aniridia.
- James continues to manage membership but we need a dedicated volunteer to recruit and retain members.
- James and Keith are working on General Data Protection Regulation compliance and other database improvements.

Liaison/Public Relations

We sent representatives to:

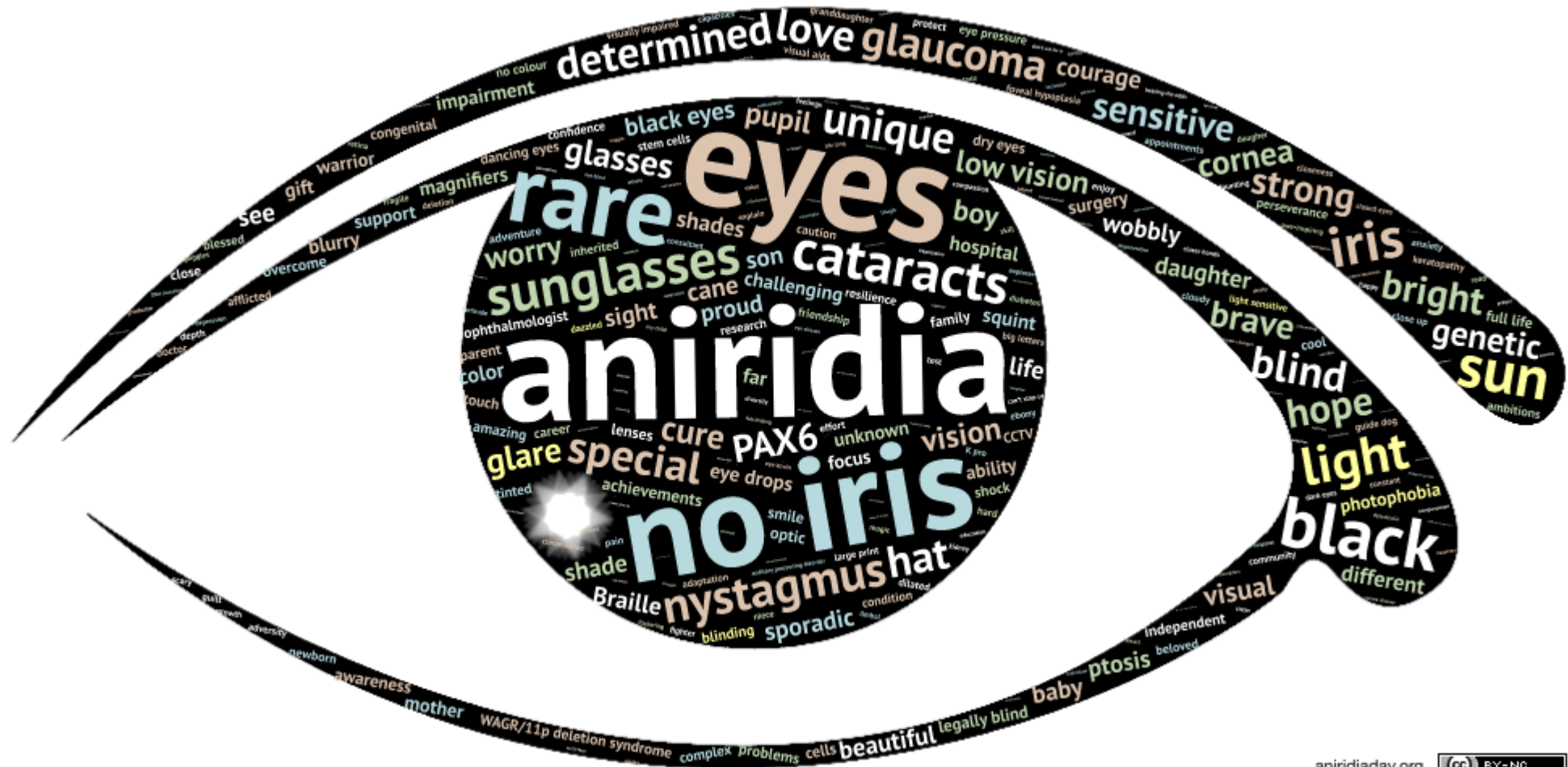
- Genetic Alliance UK Annual General Meeting.
- UK Genetic Disorders Leadership Symposium 2017 and 2018
- NCVO Trustee Conference 2017



Aniridia Day 2017

- Poetry competition
 - 10 submissions (vs 8 for rest of world)
 - Adult and child winners received Amazon vouchers
 - Katie co-judged international competition
- Eleanor, Marion and Una distributed leaflets to hospitals in Dublin and Belfast
- James held question and answer session on Facebook live
- Glen and James created a word cloud





Aniridia Day 2017

Promoted [#ShiningSuccess](#) campaign and many in the UK contributed

- Caryl & Mari (WAGR) won a pair of high quality sunglasses for the best story with an amazing 161 Likes/Loves



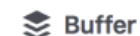
Like



Comment



Share



Buffer




Galina Gening, Glen Turner and 161 others

Website

Similar statistics in
2016/17 and 2017/18:

- 11,430 page views
- 6000 visitors
- 20 blog posts

Down compared to
previous years



The screenshot shows the homepage of the Aniridia Network UK website. At the top is the logo, which features a stylized eye with colorful figures holding hands around it, and the text "Aniridia Network UK". Below the logo is a navigation bar with links: Home, Blog, Events, About us, Services, Support us, About Aniridia, and Links. The main content area has a header with two links: "Thanks to the person in front" and "Kids complete Mini Great North Run in aid of aniridia". The main heading is "2016 Paralympians with aniridia", followed by a sub-header "Posted on September 18, 2016 by Aniridia Network UK". The text states that athletes with aniridia competed at the Rio 2016 Paralympic Games. A list of achievements follows: Mary Fisher (New Zealand) won gold in 100m backstroke S11; Amanda Dennis (USA) won bronze with her goalball teammates; Simon Richards (Canada) achieved the quarter finals with his goalball squad; and Cailin Currie (USA) came 10th and 15th in the 400m and 100m freestyle swimming S13 race. A "Join us" button is visible on the right. Below the main text, there is a section titled "Goalball Athlete - Amanda Dennis" with a video player showing a woman smiling. The video player has a "Student CNN News" logo in the bottom right corner. On the right side of the page, there are several promotional boxes: "Search", "Join us", "Subscribe", "Support us" (with links to donate or create a fundraising page), "eBay for Charity", "Shop for us!", "We recommend JULBO Kids sunglasses with loop SP4", and "BOOK: Aniridia and WAGR".

Aniridia Network UK

Home Blog Events About us Services Support us About Aniridia Links

← Thanks to the person in front Kids complete Mini Great North Run in aid of aniridia →

2016 Paralympians with aniridia

Posted on September 18, 2016 by Aniridia Network UK

Athletes with aniridia competed at the Rio 2016 [Paralympic Games](#).

- [Mary Fisher, New Zealand](#) won gold in 100m backstroke S11 with a world record time and 4th or 6th in freestyle swimming at other lengths
- [Amanda Dennis, USA](#) won bronze with her goalball teammates
- [Simon Richards, Canada](#) achieved the quarter finals with his goalball squad. [Read more](#) and [watch a video](#) about Simon.
- [Cailin Currie, USA](#) came 10th and 15th in the 400m and 100m freestyle swimming S13 race. Read a [local news article about Cailin](#).

Do you know any others with aniridia who took part?

▪

For more inspiring stories read our previous article about [people with aniridia at the London 2012 and previous Paralympic Games](#)

Goalball Athlete - Amanda Dennis

Student CNN News

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- [Sell on eBay and donate a percentage](#)

eBay for Charity

- [Get online shops to donate to us](#)

Shop for us!
Turn every pound you spend online into funds for us.

We recommend
JULBO Kids sunglasses with loop SP4

BOOK: Aniridia and WAGR

Email broadcasts sent

- London meet up 2016
- London meet up 2017
- March 2017 news
- November 2017 news
- January 2018 news
- March 2018 news & conference booking
- Conference booking reminder

Email broadcasts received

- Statistics from Conference 2018 booking email
 - 414 emails sent,
of these
 - 145 emails opened
of these
 - 30 clicked on at least one link

414 emails were sent



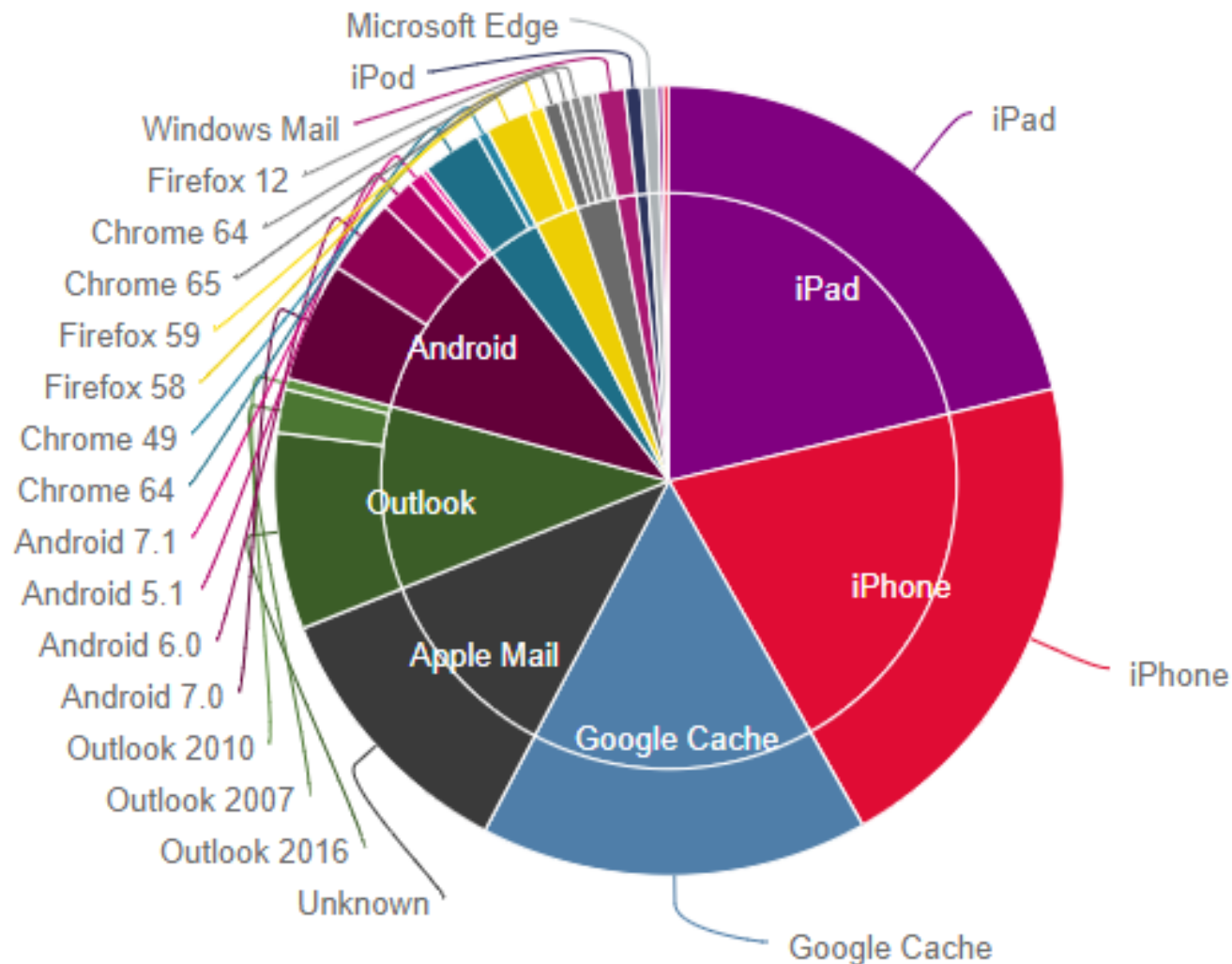
145 emails were opened



30 contacts clicked



Email broadcasts recipients

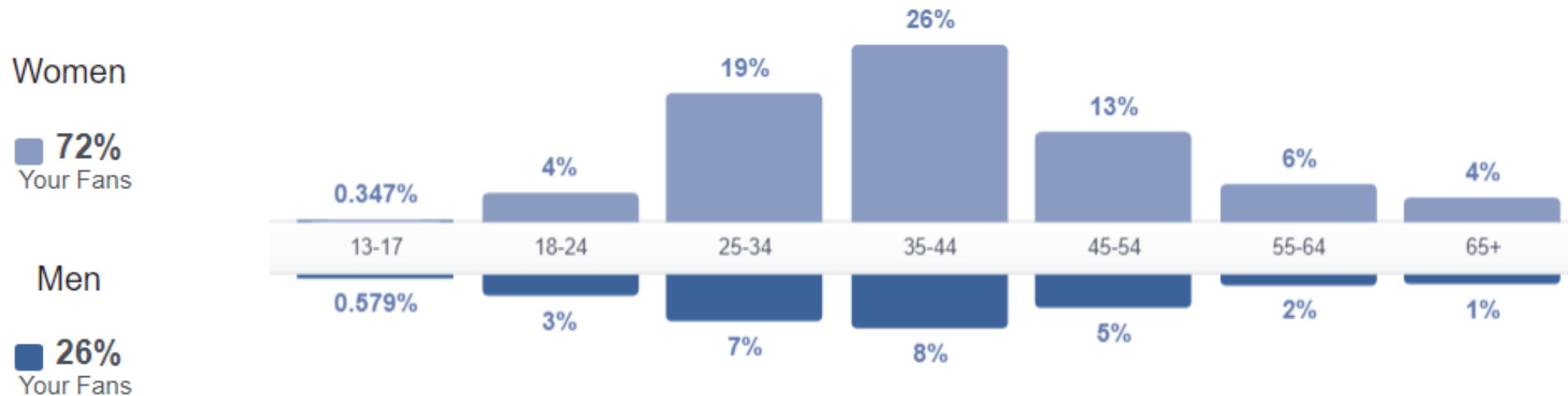


Social media

- Monitored and promoted mentions of 'aniridia'
- Promoted and engaged in Aniridic Family Facebook group
- 2016/17
 - Streamed and published recordings of Conference 2016 on Periscope/YouTube
- 2017/18
 - Glen volunteered to be Communications Officer
 - Regular reminder posts for setup about fundraising and volunteering




Facebook & Twitter

- Facebook

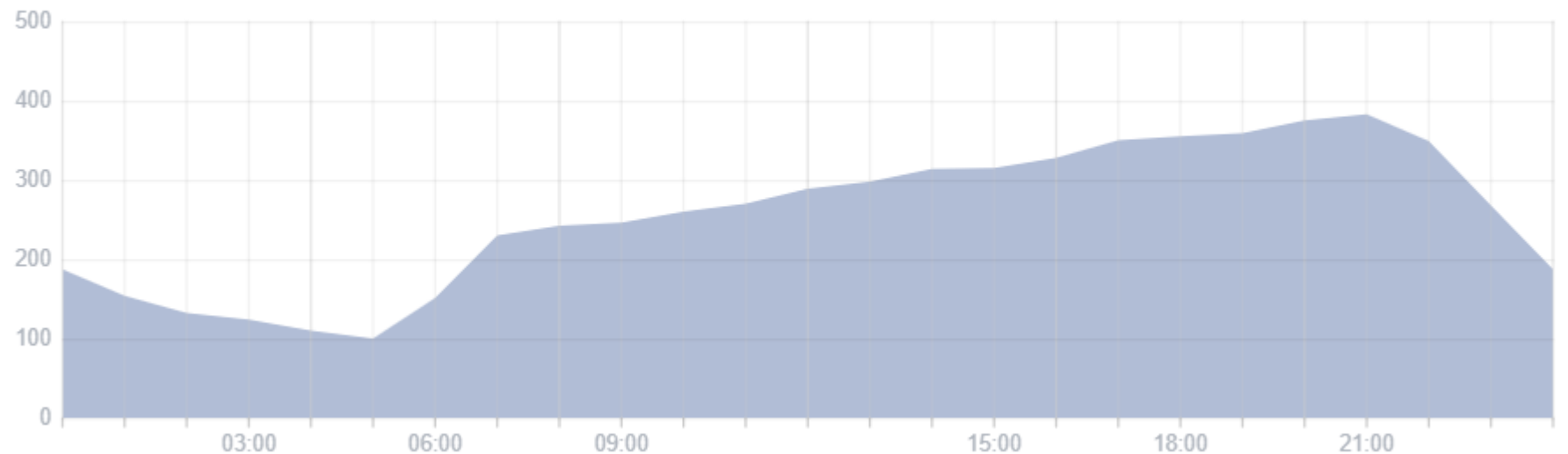


Site	2015/ 16	2017/ 18	Year on year increase					
			2015-16		2016-17		2017-18	
Facebook	705	850	98	16%	74	10%	71	9%
Twitter	597	718	80	15%	82	14%	39	6%

[Show All Posts ▾](#)Reach Post clicks Reactions, comments & shares









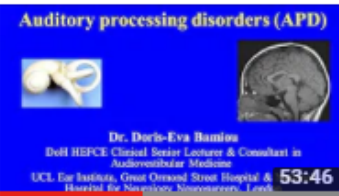

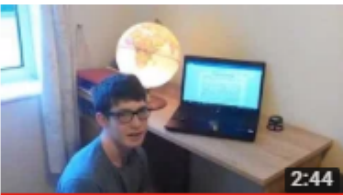

Type	Average Reach		Average Engagement	
 Link	376	<div></div>	42 17	<div></div> <div></div>
 Status	173	<div></div>	10 6	<div></div> <div></div>
 Photo	147	<div></div>	9 3	<div></div> <div></div>

TIMES



YouTube

- Published 14 new videos including sessions at Conferences 2011, 2015, 2016
- 69 subscribers

 Aniridia Network UK Aniridia: Check Your Knowledge <small>By James Buller and Jenny Lang</small> <div>53:12</div> <p>Aniridia: Check Your Knowledge 148 views • 1 year ago</p>	 Aniridia Network UK VI Tennis & Sports Discussion <small>By James Buller and Jenny Lang</small> <div>17:30</div> <p>Questions & Answers Session 2015 1 view • 1 year ago Private</p>	 Aniridia Network UK VI Tennis & Sports Discussion <small>By James Buller and Jenny Lang</small> <div>21:31</div> <p>Visually Impaired Tennis 1 view • 1 year ago Private</p>
 Aniridia Network UK One Vision - European Youth Exchange <small>By ANUK Trustees</small> <div>25:55</div> <p>One Vision European Youth Exchange 1 view • 1 year ago Private</p>	 Aniridia Network UK Aniridia Network UK Annual General Meeting 2015 <small>By ANUK Trustees</small> <div>42:22</div> <p>Annual General Meeting 2015 of Aniridia Network UK 29 views • 1 year ago</p>	 Aniridia Network UK Befriending: the story so far <small>Lyn Buller: ANUK Befriending Scheme Officer</small> <div>22:07</div> <p>Befriending No views • 1 year ago Private</p>
 <div>31:38</div> <p>Aniridia The Other Effects 72 views • 1 year ago</p>	 <div>8:39</div> <p>Starting university with aniridia 133 views • 1 year ago</p>	 <div>53:46</div> <p>Auditory processing disorders (APD) Auditory Processing Disorders & The PAX6 Gene 241 views • 1 year ago Subtitles</p>
 <div>6:43</div> <p>Katie on life with aniridia 85 views • 1 year ago Subtitles</p>	 <div>2:44</div> <p>Gleb: Living with aniridia 949 views • 1 year ago</p>	 <div>4:56</div> <p>Pupil Passports for aniridia 395 views • 3 years ago</p>

Volunteering & Development

Martin Fleming

Fundraising

We've broken our £5,000 target

- Matt Raised £5000
408 mile bike ride in 1 day
- David Raised £1866
First boxing match
- Other donations £500+
 - Kids Great North Run
 - Golden Wedding
 - Steelman Triathlon
 - School bake sale

Thank you to everyone who has given so generously to achieve a magnificent £7368.50!



Donations and commissions

Big events can provide a real boost to fundraising but the number can vary year to year so regular income is also important.

- Regular monthly giving from supporters continues and is a vital income stream for us.
- Give as You Live continues to give us a small but steady amount of income from online shopping.
- PayPal Giving Fund brings together donations made via PayPal Humble Bumble and eBay for Charity and is a steady growth area.

Volunteering

- Volunteering is our main means of filling roles to run the organisation and provide its services
- Important roles needing to be filled at this time include:
 - Membership Officer
 - Volunteer Coordinator
 - Trustee
- Many other volunteering opportunities are listed on the website for both long and short term tasks, and we welcome all levels of help.
- Lack of volunteers from among the members has led us to start advertising externally.

Volunteers thank you

We would like to thank all our volunteers for their work over the year:

- Liz Atkinson
- Eleanor Burke
- Lyn Buller
- Mary Cox
- Nicola Jamison
- Dave McKay
- Keith Spink
- Glen Turner
- David Woodward

Ireland

- Members delighted to have a local group as they felt quite isolated before and that their ophthalmologists had very little experience of aniridia.

Eleanor has

- fundraised £250.00
- interviewed on Dublin radio and the RNIB Northern Ireland 'Crack-On' podcast
- forged relationship with Fighting Blindness Ireland and RNIB NI to distribute information to their aniridia contacts
- been co-opted to the Board Need2Talk partnership a 5 year European project on emotional needs of visually impaired people in the rural areas.

Trustee Board 2016-18

- Ben Rendle stepped down in September 2016
- Emily Nash was co-opted in March 2017 after an appeal for volunteers with financial skills
- We would like to thank our trustees for their work over the year:
 - Katie Atkinson (Chair)
 - Emily Nash (Treasurer)
 - James Buller
 - Martin Fleming

Trustee recruitment

- We have one opening for a trustee to the board but we are ideally looking for certain skills/experience
- We are particularly keen for a trustee to be a secretary and/or a representative of younger members
- We are currently advertising the Secretary position on Trustee Bank
- Anyone wishing to nominate themselves or wishes to know more about what skills we are looking for get in touch

Questions to trustees?

Trustee elections

Nominations

- Katie Atkinson
- Emily Nash
- James Buller
- Martin Fleming

Governance consultation

- Membership
 - AGMs where trustees are elected to the Board
 - Who votes: Everyone, or Aniridics (representative), by household?
 - Extra administration to track and review membership
 - Benefits (consequences) of (non-)membership?
Don't want to exclude anyone
Why join?
- Foundation
 - No need for AGMs
 - Trustees co-opt on to the Board

Any other business



Aniridia Network

PAX6 and the Cornea: An Eye to the Future

Colin E. Willoughby

Prof of Ophthalmology, Ulster University & Honorary Consultant Ophthalmic Surgeon, Belfast

Honorary Professor, University of Liverpool

c.willoughby@ulster.ac.uk

Web: aniridia.org.uk

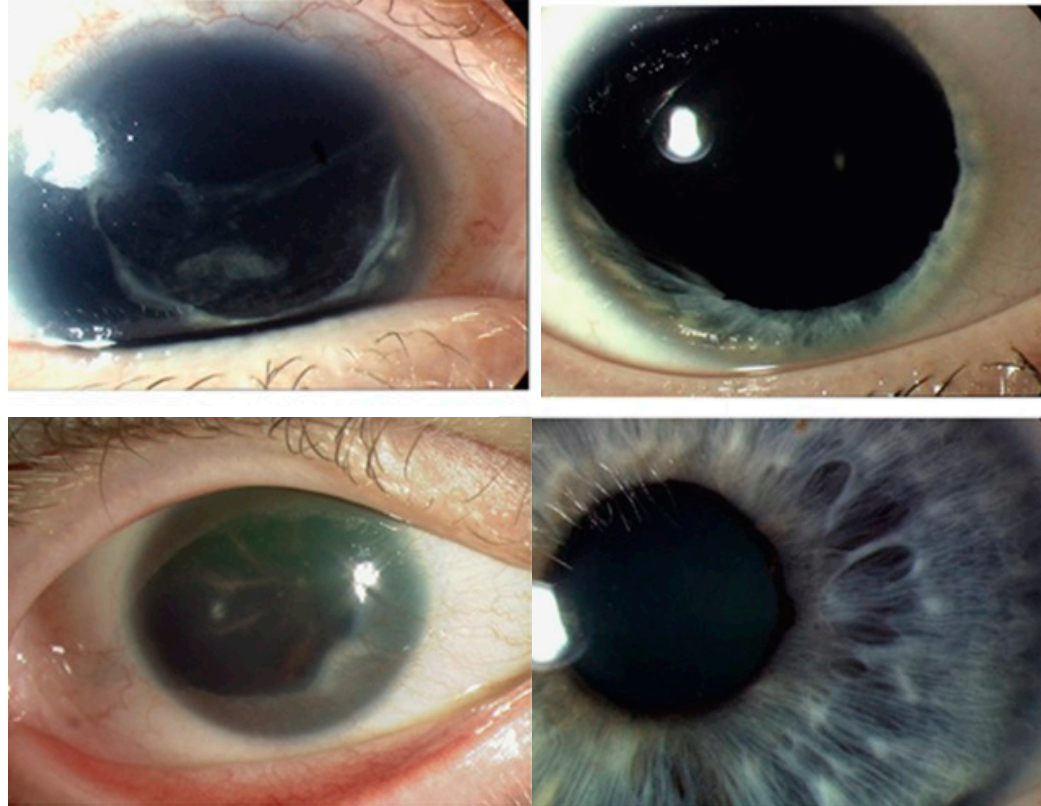
Email: info@aniridia.org.uk

Twitter: [@AniridiaNetUK](https://twitter.com/AniridiaNetUK)

Facebook: [AniridiaNetworkUK](https://www.facebook.com/AniridiaNetworkUK)

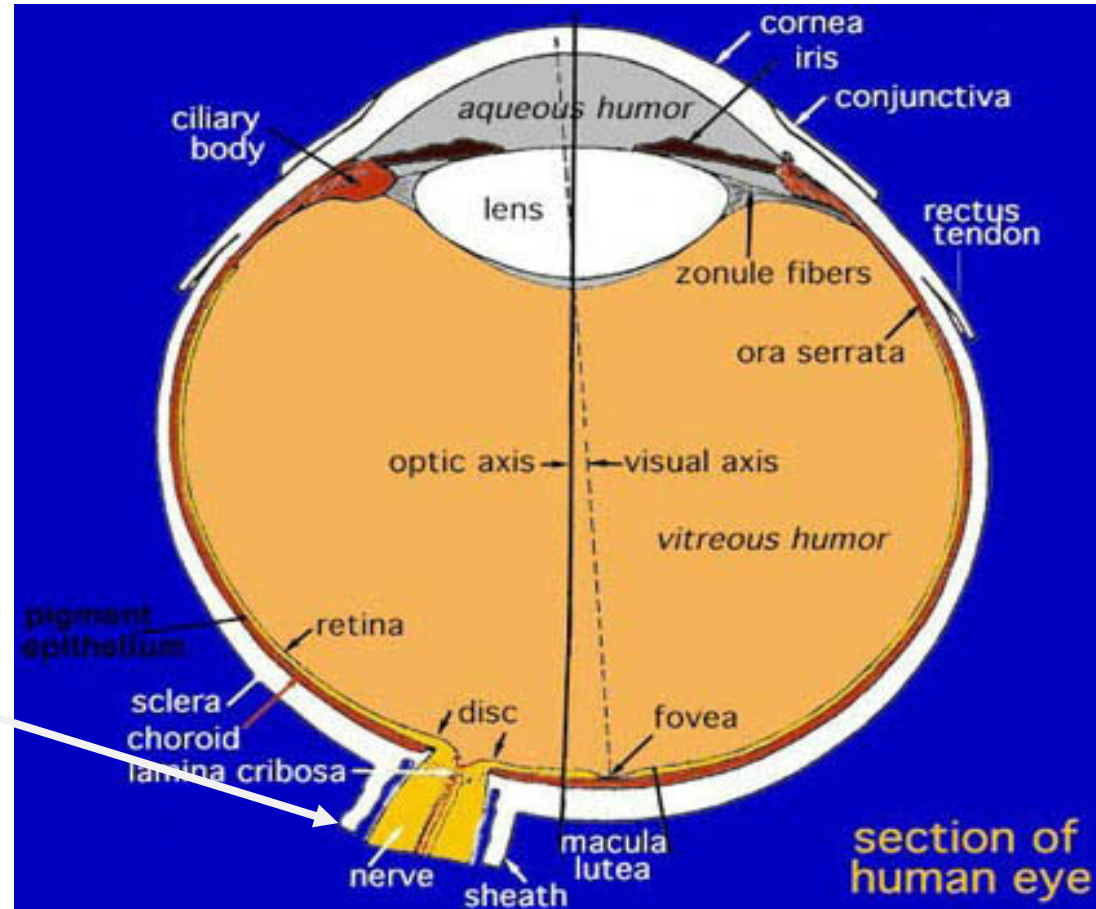
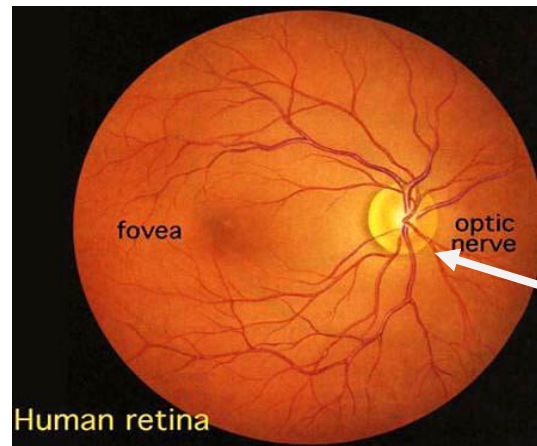
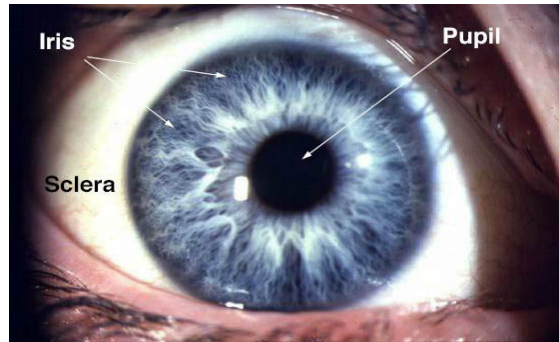
Aniridia Network is registered charity 1176792 in England & Wales, operating as
Aniridia Network UK, 22 Cornish House Adelaide Lane Sheffield S3 8BJ

Aniridia and the Eye

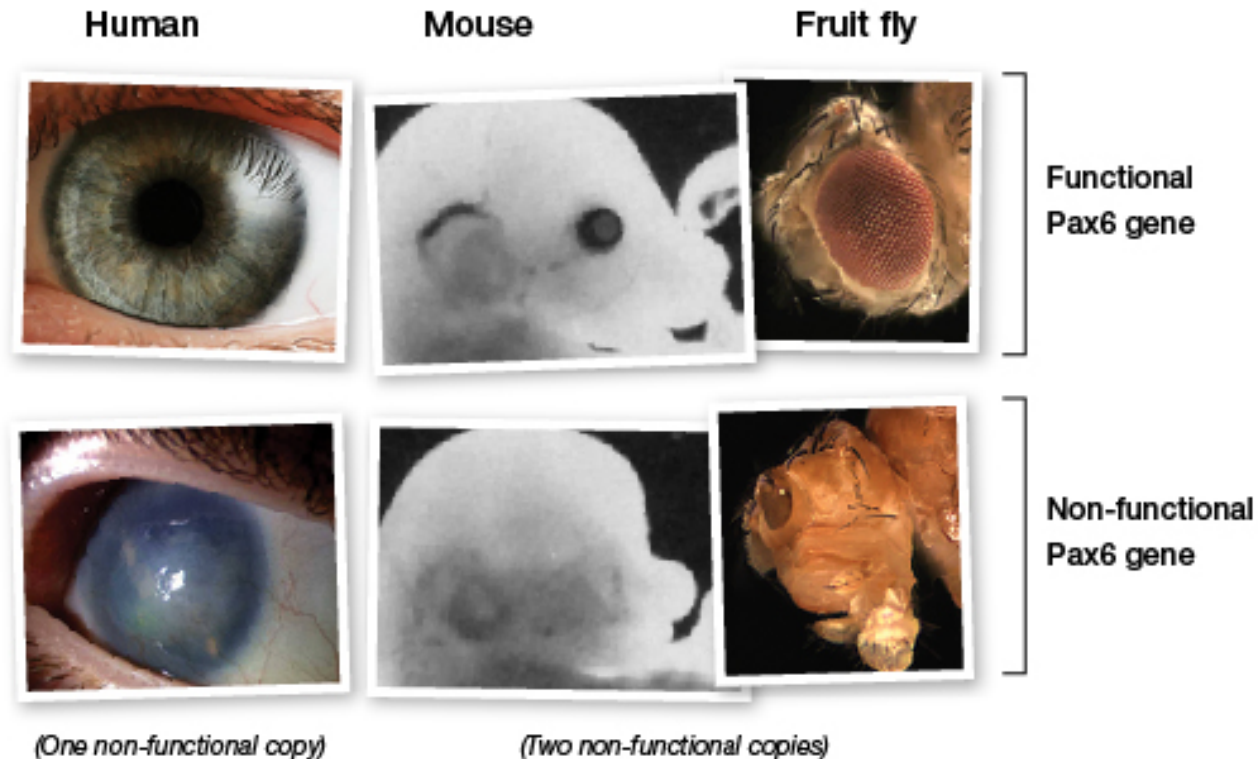


Images from Hingorani M, Hanson I, van Heyningen V.
Eur J Hum Genet. 2012

Human Eye Structure



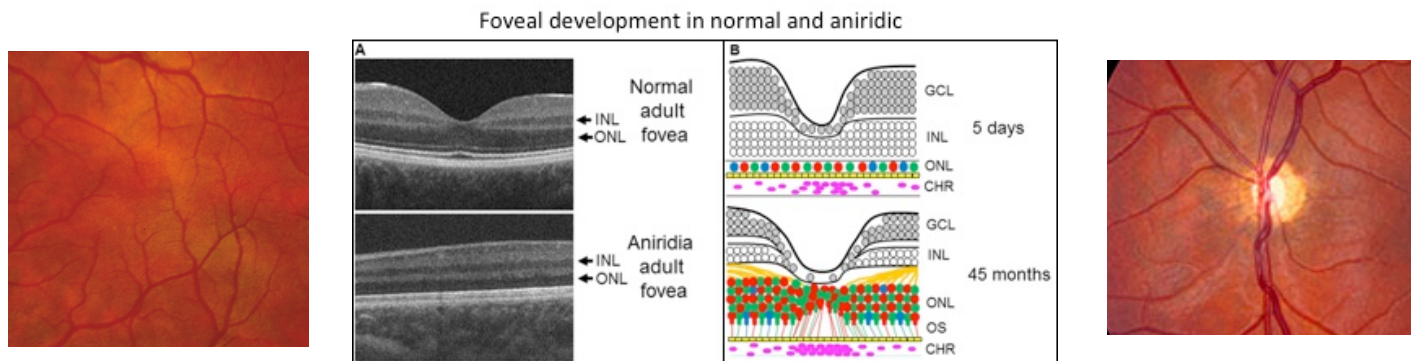
PAX6 and Eye Development



In humans aniridia results from genetic defects in PAX6

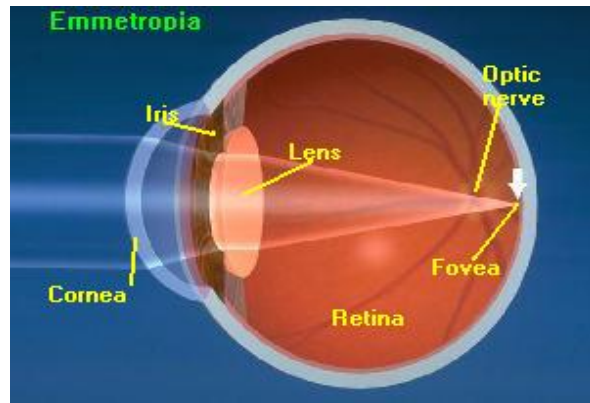
Ocular Features of Aniridia

- Aniridia is a pan-ocular or **whole eye disorder**
- Ocular features can occur at different stages of life: **birth, infancy, childhood, teens and adulthood**
- Major ocular features: **corneal disease, glaucoma, cataract, dislocated lenses, iris defects, foveal hypoplasia, optic nerve hypoplasia**
- Poor vision and nystagmus: **foveal hypoplasia and optic nerve hypoplasia**



Corneal Function

The cornea is the major refractive structure in the eye



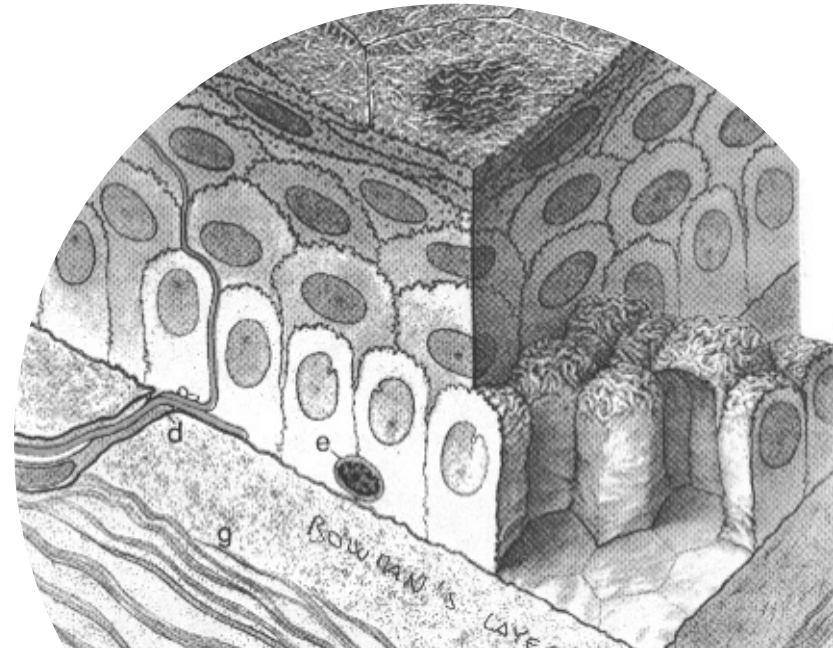
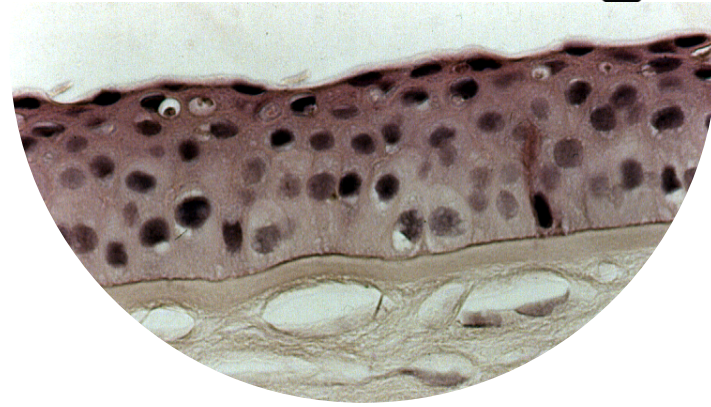
Vision is dependent on :

1. Corneal Shape
2. Corneal Transparency -

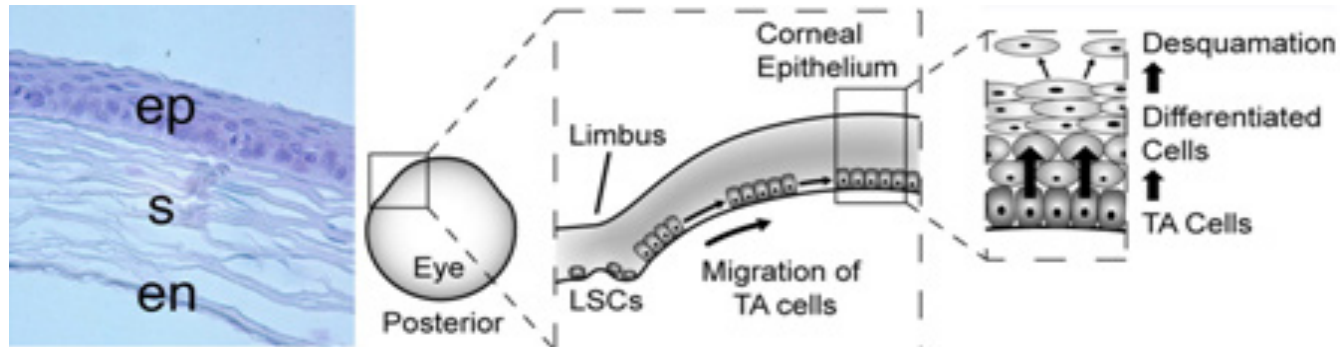
- a. **Intact corneal epithelium**
- b. Regular arrangement of corneal collagens
- c. **Avascular cornea**
- d. A functionally intact corneal endothelium to regulate corneal hydration

Corneal Epithelium

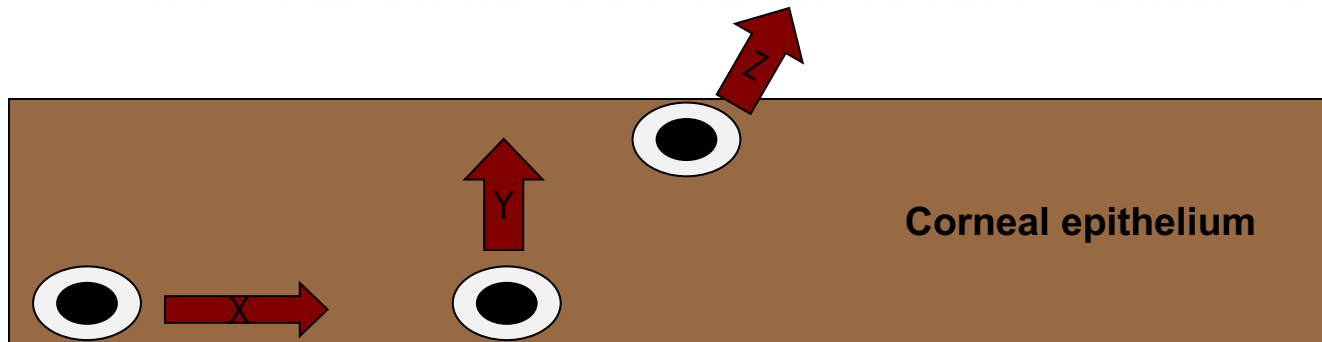
- 50 μm thick
- Three cell types:
 - superficial (1-2 layers)
 - wing cells (3 layers)
 - basal layers
- Basement membrane



X + Y = Z Hypothesis



Histology of Mouse Normal and abnormal maintenance of the adult corneal epithelium.



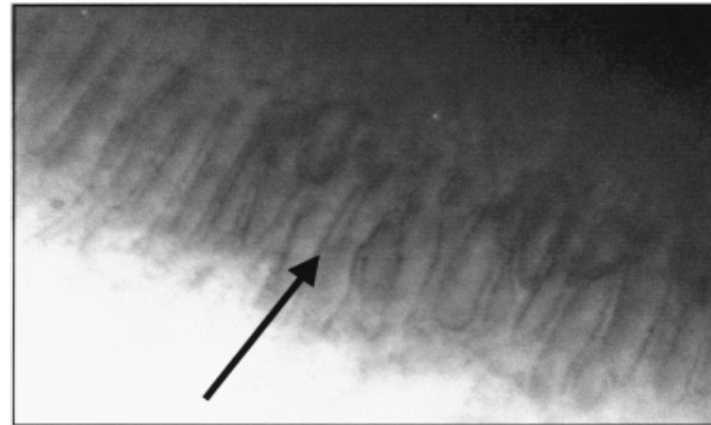
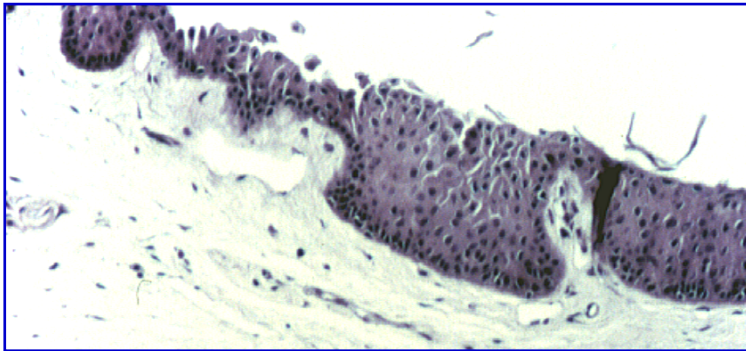
X = cell movement parallel to epithelial basement membrane

Y = cell movement perpendicular to epithelial basement membrane

Z = cell loss from epithelial surface (Thoft and Friend 1983)

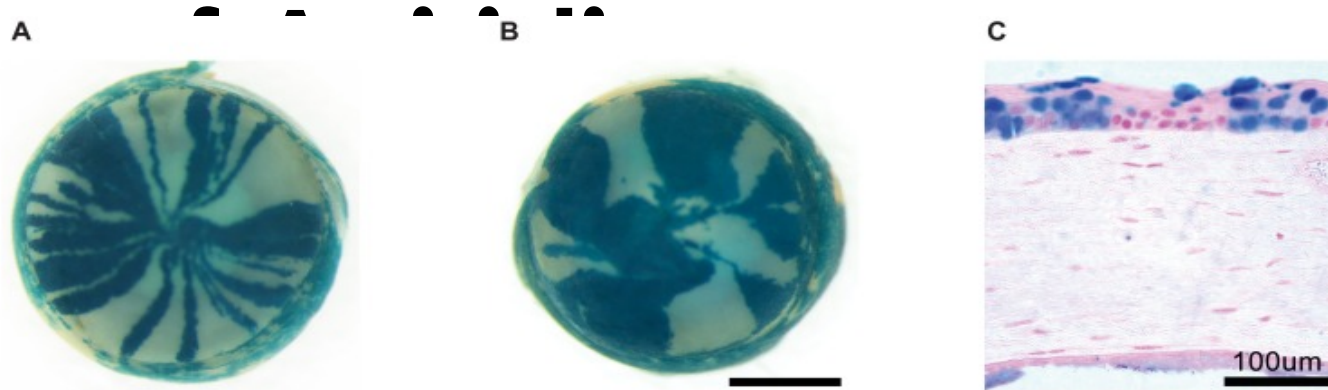
Corneal Limbus

Corneal epithelial cells originate from stem cells located in the limbal epithelium



palisades of Vogt

Corneal Disease in a Mouse Model

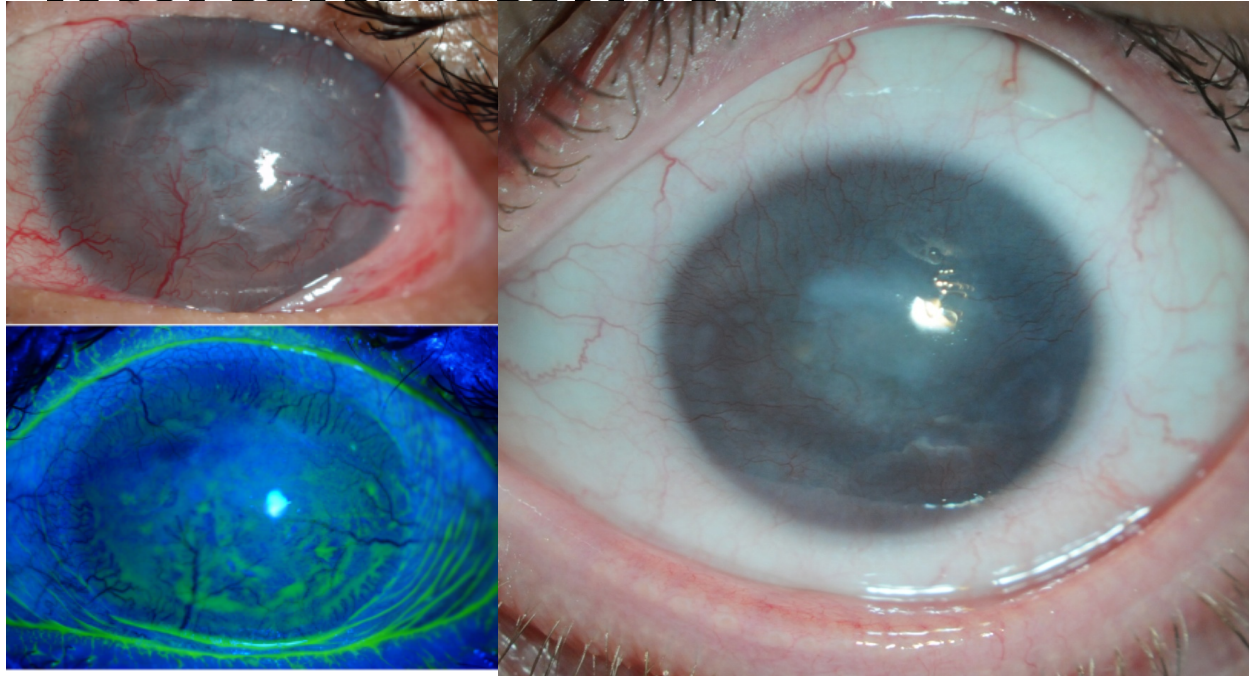


Dr John West and Prof Martin Collinson

mouse *Pax6*^{+/-} corneal abnormalities:

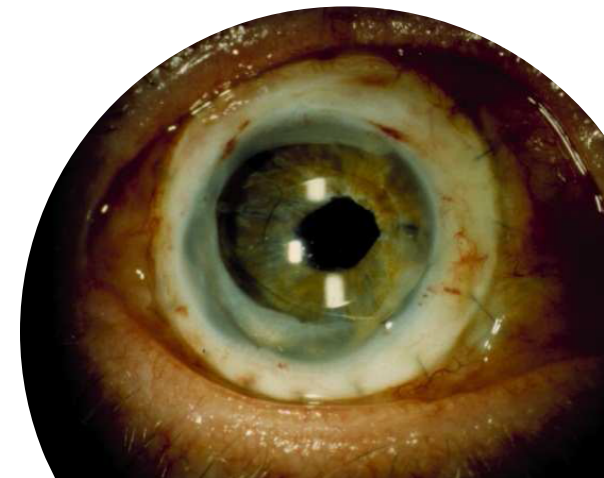
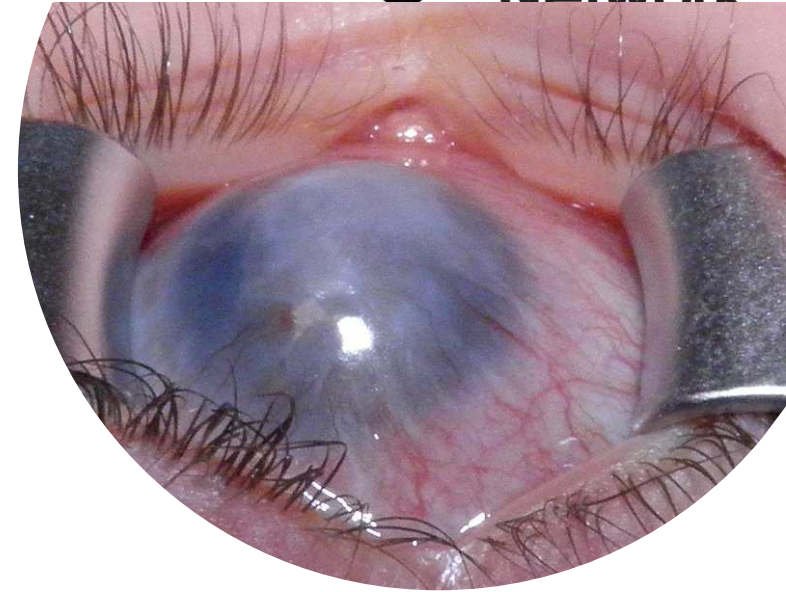
- arise during development (e.g. the corneal epithelium is already thinner than normal by embryonic day 18.5)
- other abnormalities arise during adulthood (e.g. blood vessels invade the corneal stroma, goblet cells accumulate in the corneal epithelium and centripetal epithelial cell movement is disrupted)
- Defects may result from limbal stem cell deficiency or increase corneal epithelial loss

Aniridia-Related Keratopathy



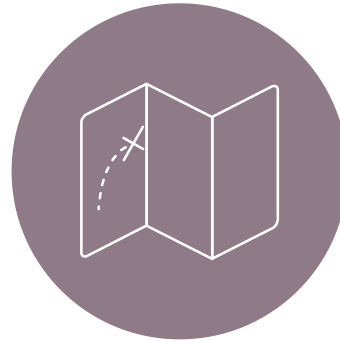
Aniridia-Associated Keratopathy

- Reduce incident light with tinted or photochromic glasses
- Preservative free lubricants
- Hyaluronic acid containing lubricants
- Autologous (or allogenic) serum eye drops
- Amniotic membrane
- Limbal stem cell reconstruction:
 - Living related donor
 - Non- matched organ culture donor
 - Keratolimbal allograft
 - allogeneic ex vivo cultivated limbal epithelial transplantation (allo-CLET)
- Corneal Surgery

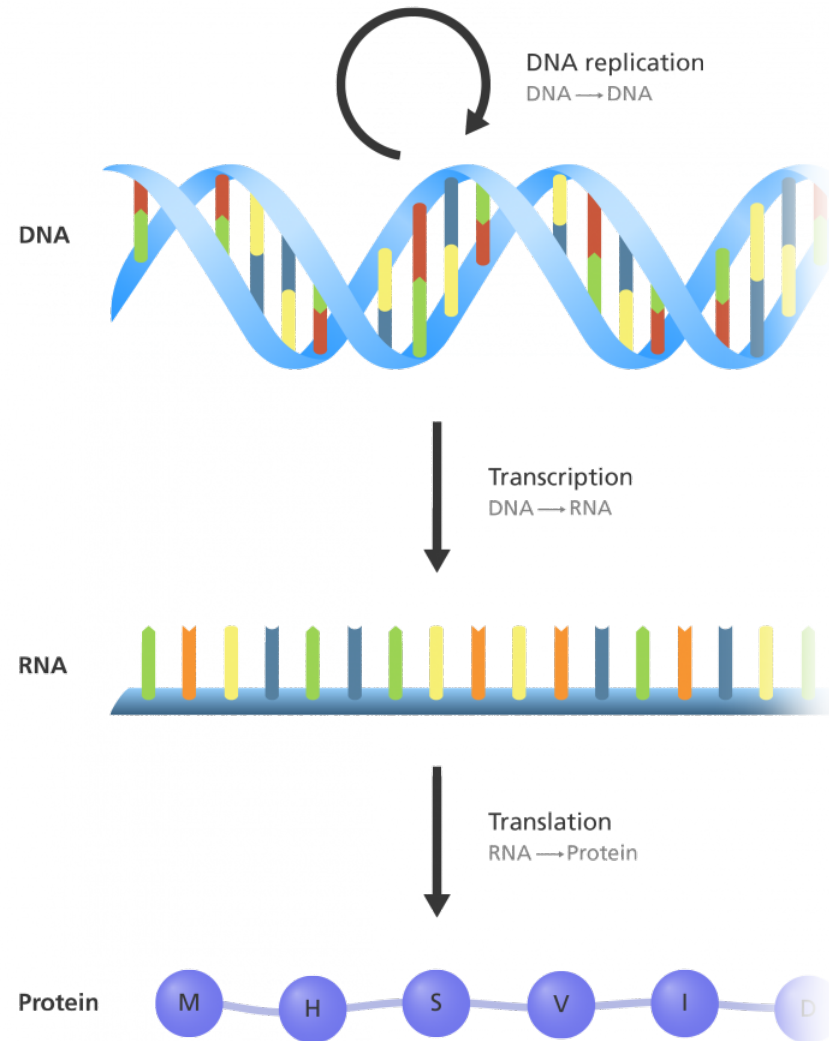
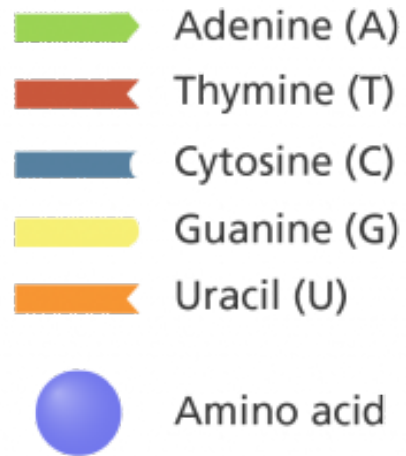


Morpholinos as Therapeutic Tools for Aniridia

Thanos Papadimitropoulos
PhD Researcher
University of Liverpool



- **Molecular Dogma**



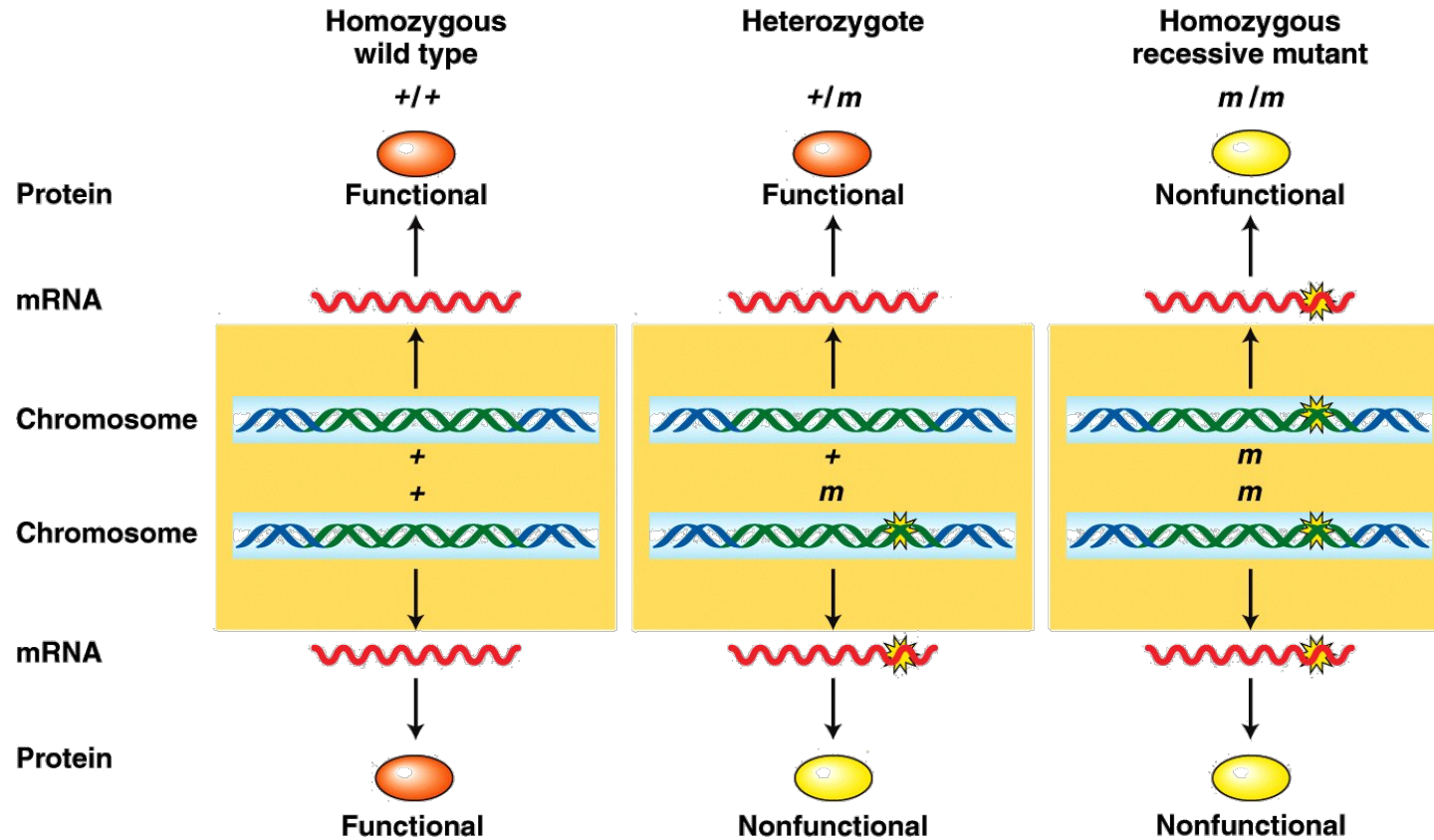


Figure 6-2
Introduction to Genetic Analysis, Ninth Edition
 © 2008 W. H. Freeman and Company

Haploinsufficiency

- Due to mutations on a single copy of a gene not enough protein is made; leads to abnormal or diseased state
- Developmental disorders, metabolic disorders, cancers, epilepsy
- Transcription Factors
- Control the rate of transcription from DNA to mRNA
- Many are involved in development
- Even small changes can lead to a cascade of events

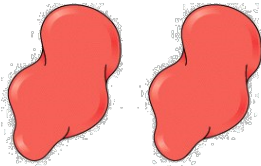


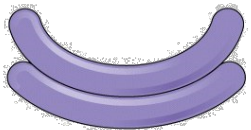
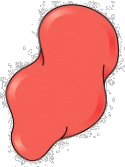
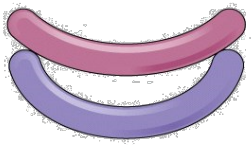
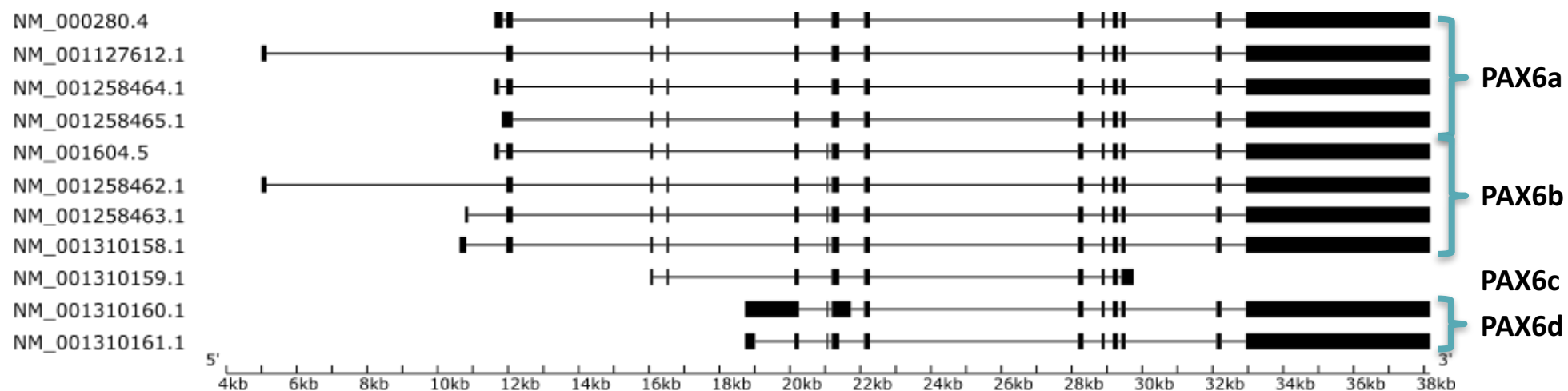
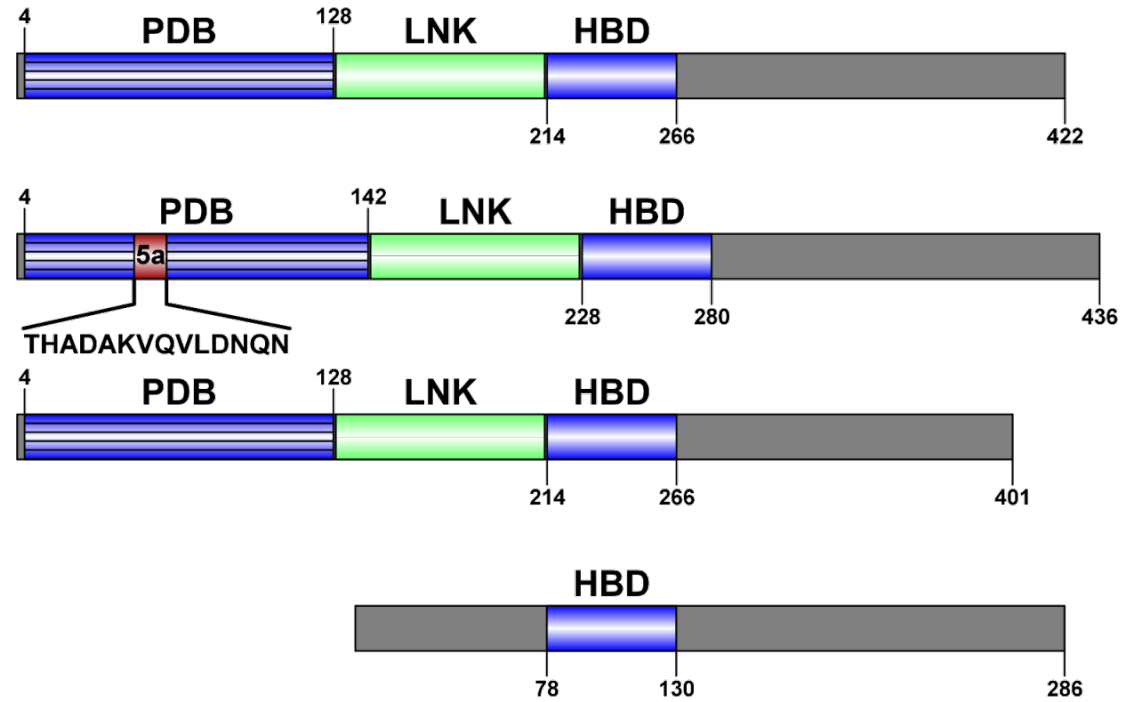
	Haploinsufficiency	Dominant negative
$+/+$	 2 "doses" of product	 Dimer
M/M	 0 "dose"	
$+/M$	 1 "dose" (inadequate)	

Figure 6-3
Introduction to Genetic Analysis, Ninth Edition
 © 2008 W.H. Freeman and Company



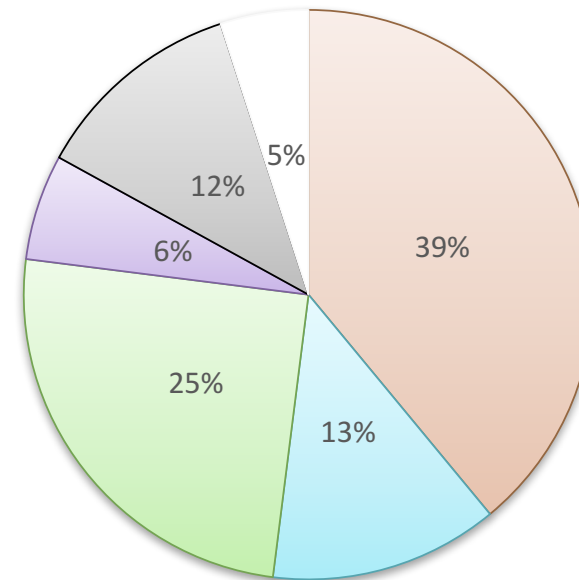
Legend:

■ Exon — Intron



PAX6

Percentage of mutation type

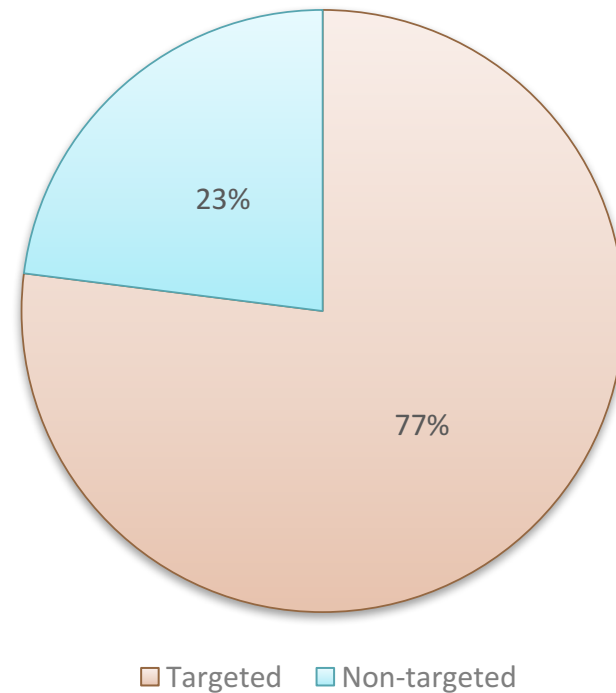


■ Nonsense
 ■ Splice
 ■ Frameshift Insertions & deletions
 ■ Inframe insertions & deletions
 ■ Missense
 ■ Run-on

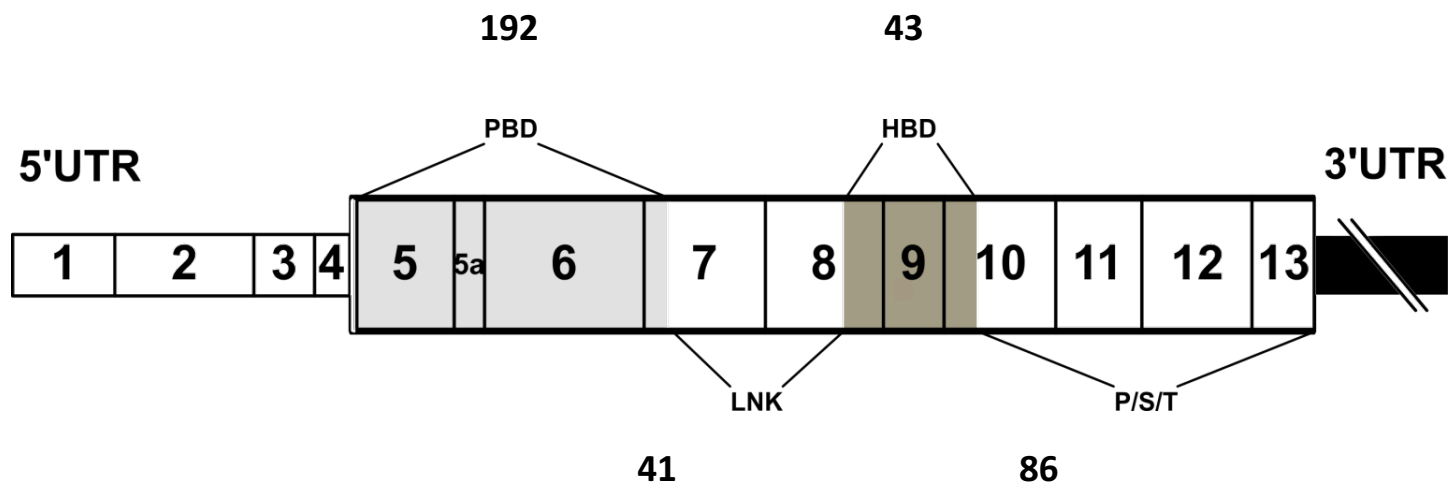
- **Master control gene in eyes, pancreas and central nervous system**
- **Regulates cellular proliferation, differentiation, migration and adhesion**
- **Its targets include *PAX6* itself and genes encoding other developmental regulators, cell adhesion molecules and structural proteins, such as lens crystallins and corneal keratins**
- **Most mutations lead to loss of protein-function**
- **Highly conserved at genetic level**

PAX6

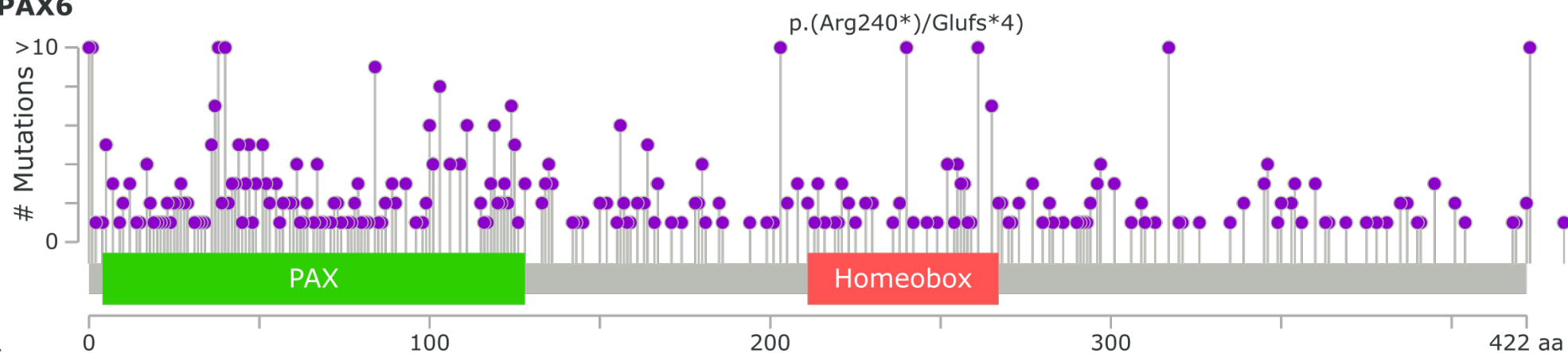
Percentage of saved types

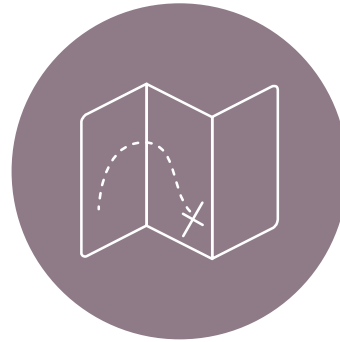


- Master control gene in eyes, pancreas and central nervous system
- Regulates cellular proliferation, differentiation, migration and adhesion
- Its targets include *PAX6* itself and genes encoding other developmental regulators, cell adhesion molecules and structural proteins, such as lens crystallins and corneal keratins
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PAX6

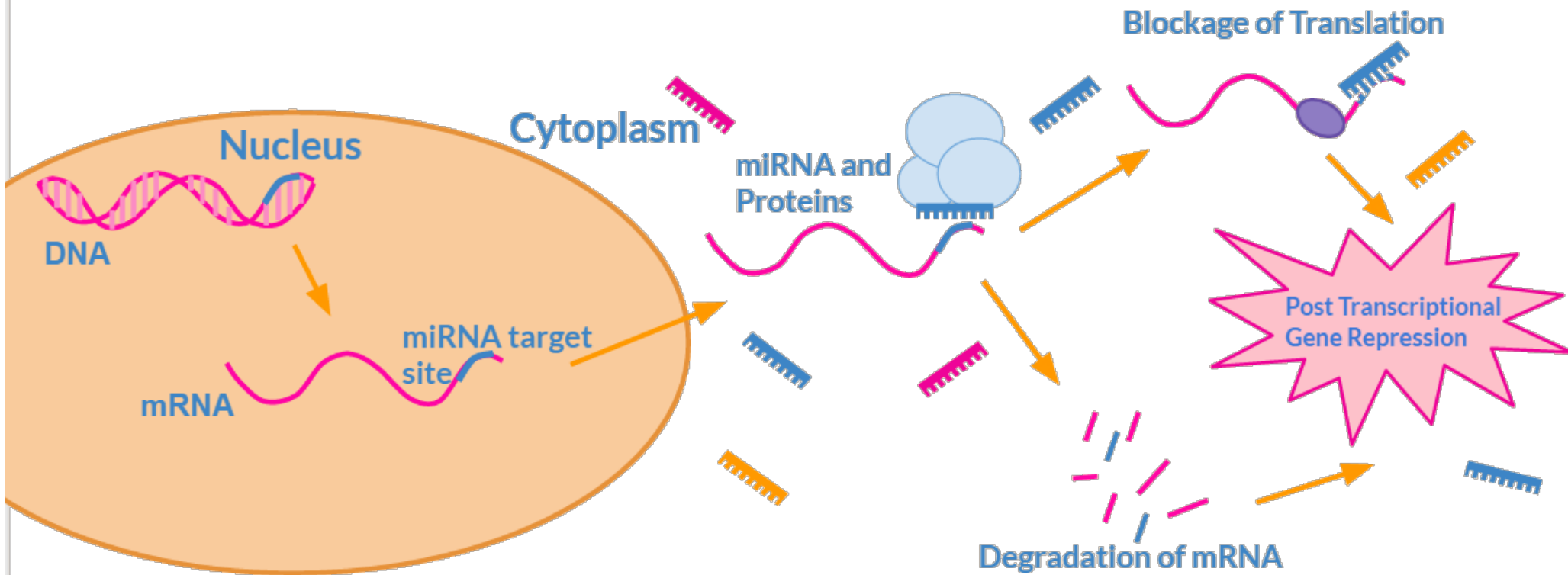




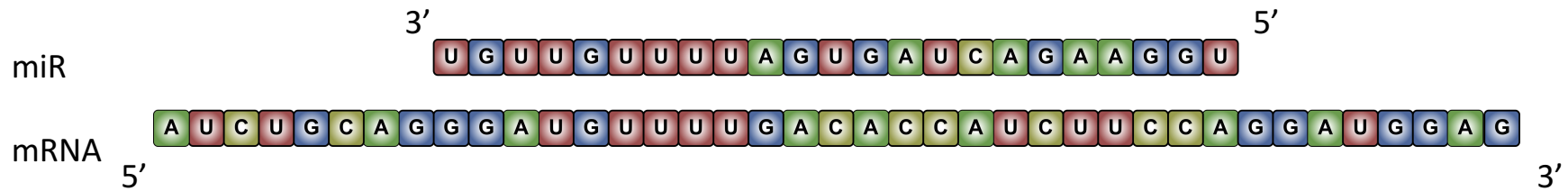
- **PAX6 levels**

*Can we increase PAX6
levels to treat Aniridia-
Related Keratopathy?*

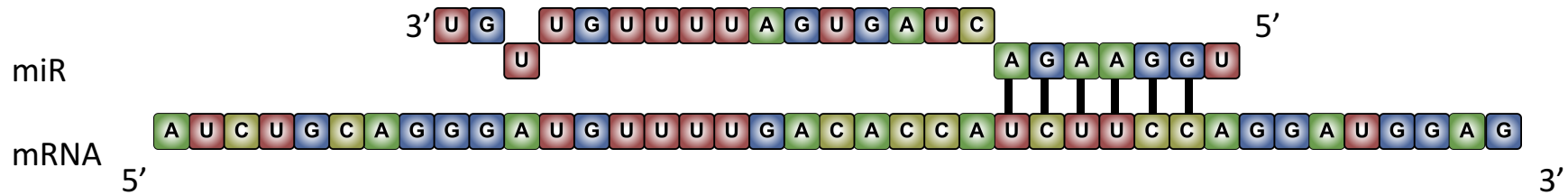
microRNAs



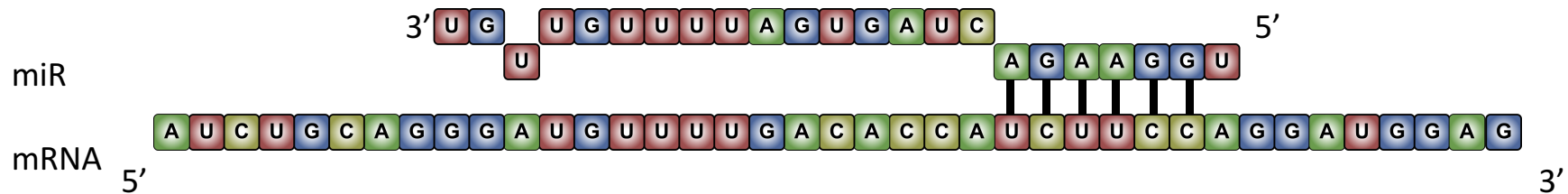
Blocking miRNAs



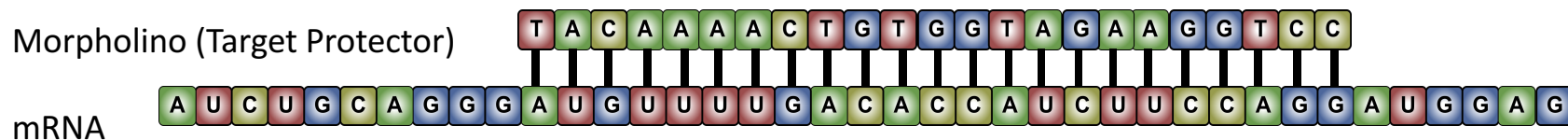
Blocking miRNAs



Blocking miRNAs



UUUAUCAAUGGGGUAUCUGCAGGGAUGUUUUGACACCAucuuuccAGGAUGGAGAUUAUUUGUGAAGACUUCAGU
AGAAU

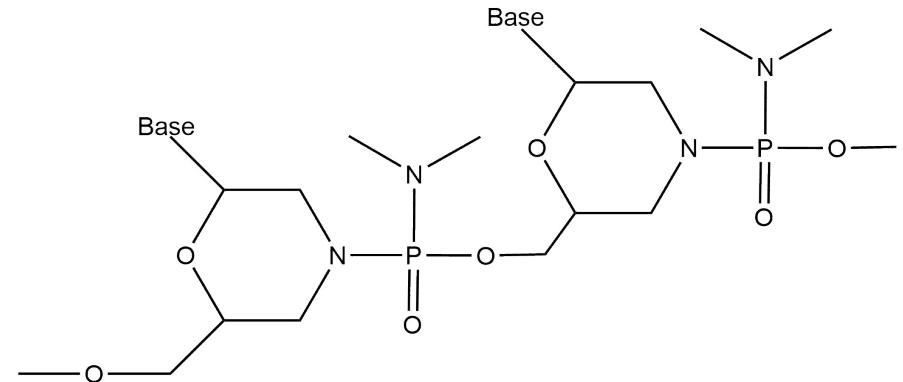
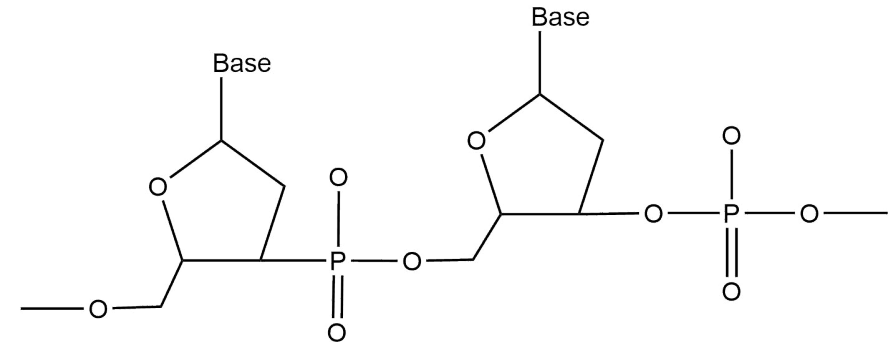


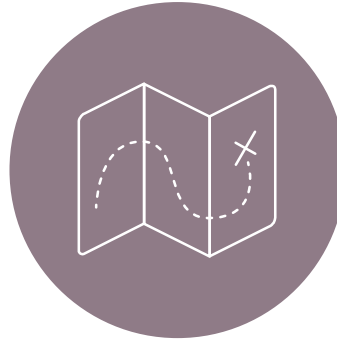
Morpholinos

- **Phosphorodiamidate morpholine anti-sense oligomer**
- **Deoxyribose rings – morpholine rings**
- **Anionic phosphodiester link non-ionic phosphorodiamidate link**
- **Watson-Crick base-pairing**

Benefits

- **Resistant to digestion by nucleases**
- **No degradation in serum or in cells**
- **No immune response**
- **No DNA methylation modification**
- **Endocytosis-mediated delivery method**

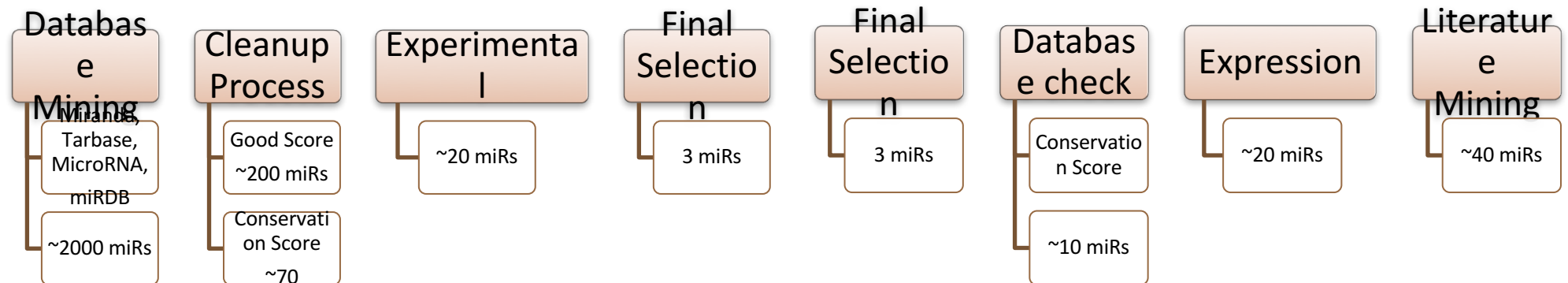




DESIGN PROCESS/METHODS

Picking the right target

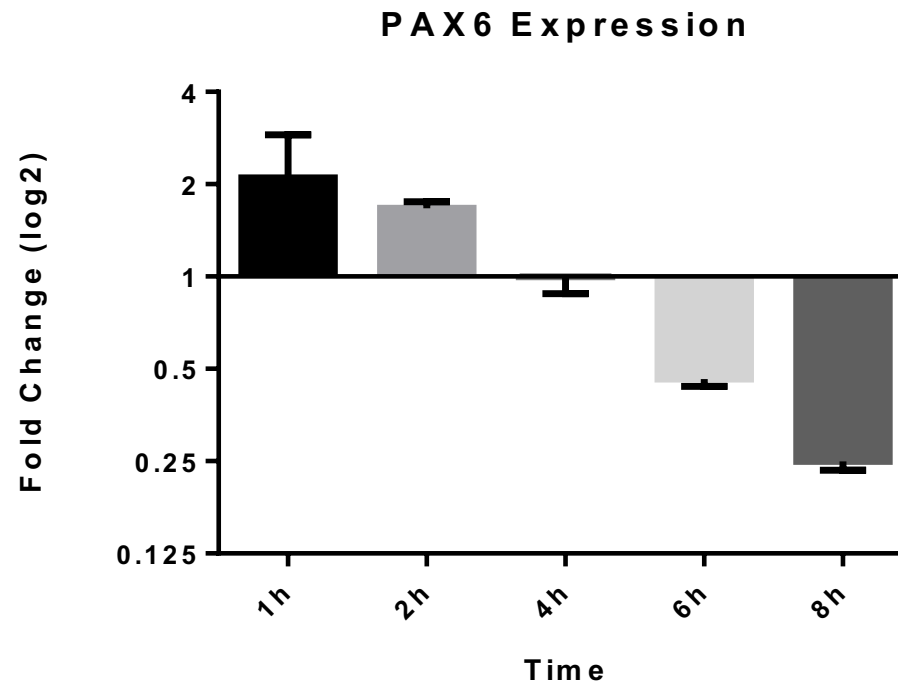
Elimination Process



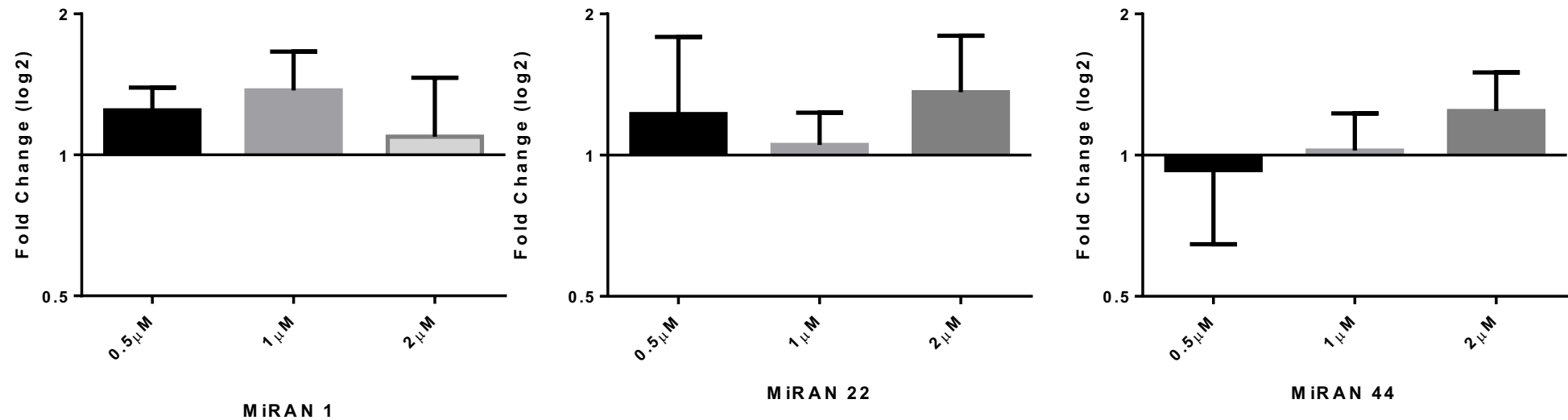


Results

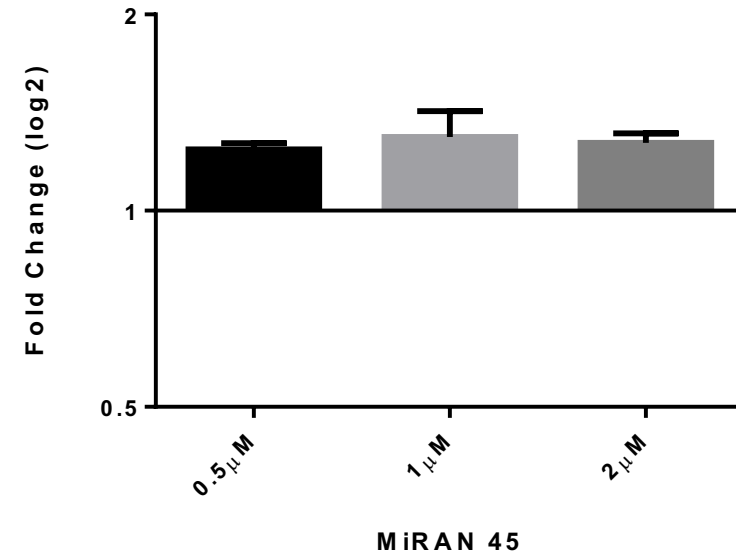
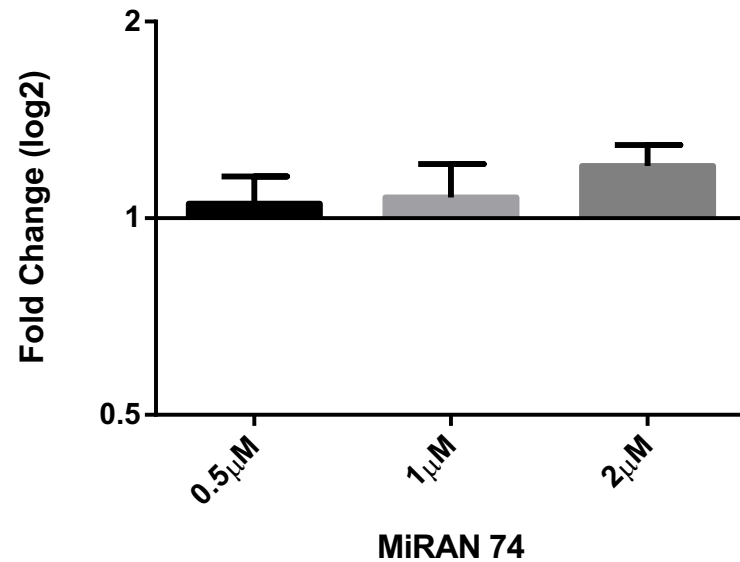
PAX6 Degradation Rate



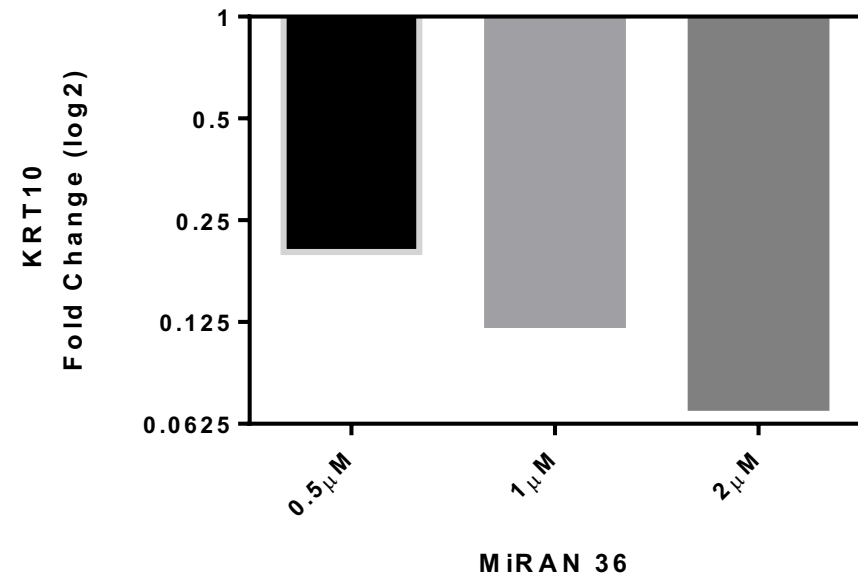
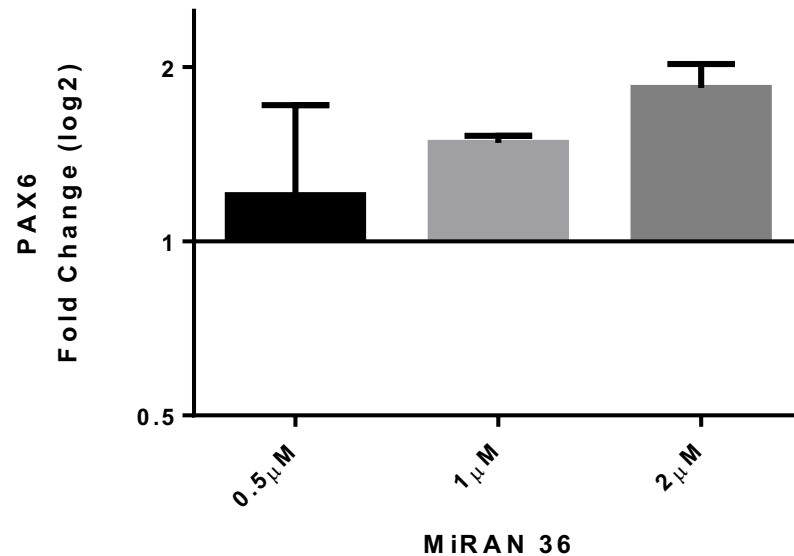
PAX6 Relative Expression Levels against control MO



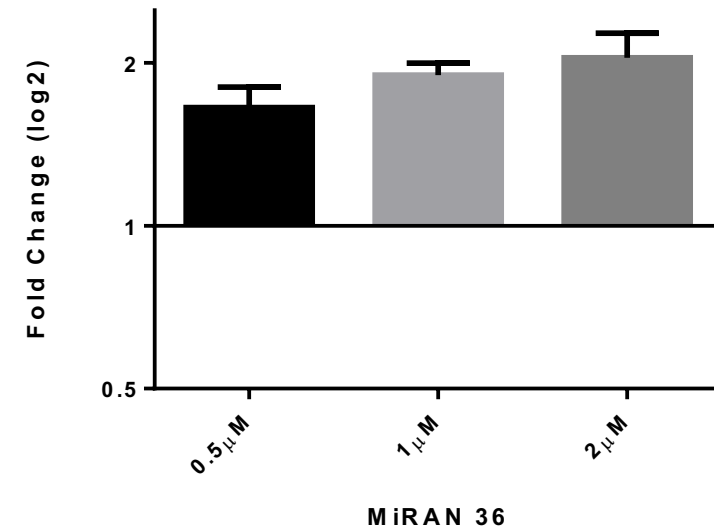
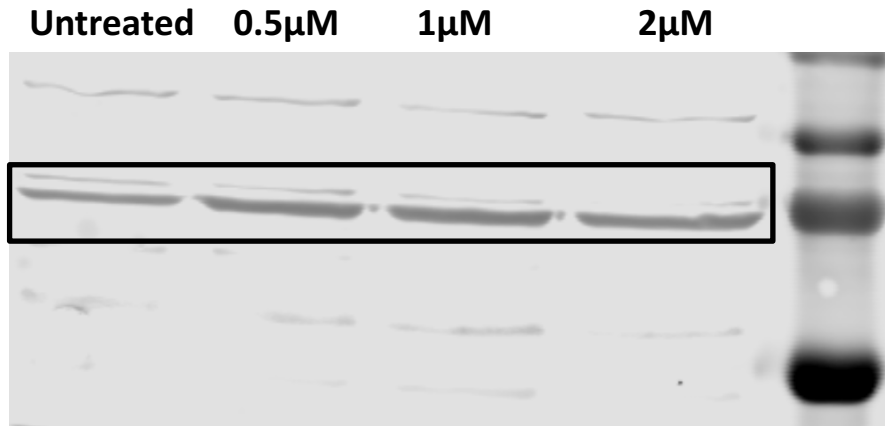
PAX6 Relative Expression Levels against control MO



PAX6 & KRT10 Relative Expression Levels against control MO



Pax6 Expression Levels against Untreated cells



Plans

- **PAX6 protein levels**
- **PAX6 targets**
- **Vivo-Morpholinos**
- **CRISPR PAX6-haploinsufficient limbal stem cells**
- **Delivery mechanism *in vivo***
- **Aniridia mouse**

Acknowledgments

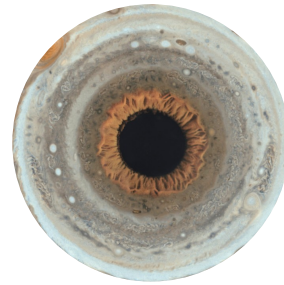
- **Dr. Kevin Hamill, University of Liverpool**
- **Prof. Colin Willoughby, Ulster University**
- **Dr Sajjad Ahmad, Moorfields Eye Hospital**

- **Dr Daniel Aberdam, INSERM, Paris**



**FIGHT
FOR SIGHT**

The Eye Research Charity®



Questions please?



Aniridia Network

PAX6 and the Cornea: An Eye to the Future

Colin E. Willoughby

Prof of Ophthalmology, Ulster University & Honorary Consultant Ophthalmic Surgeon, Belfast

Honorary Professor, University of Liverpool

c.willoughby@ulster.ac.uk

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Aniridia Network

Living with aniridia

Fern Lulham

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Facebook: **AniridiaNetworkUK**

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Raffle

- £1 to enter

Exhibition stalls

- Guide Dogs
- Nystagmus Network
- South East London Vision
- Mainline Instruments Ltd Stem cell researchers
- Aniridia Network:
 - Raffle, fundraising and volunteer
 - Make a donation
 - Buy merchandise and raffle tickets
 - Arrange to get a collection tin for a site
 - Information
 - Doctor consultation zone

Children and dogs

- Collect your children, the crèche will be closed during lunch
- Francis will help take guide dogs to spend on grass nearby

During the break

- Visit the stalls
- Hand in data check sheets and gift aid forms
- Post on social media about today
Use **#ANUKconf**
Twitter: @AniridiaNetUK
- Film a vox-pops
- Sessions begin here at 1:00pm

Lunch

- Sandwiches:
 - Roast Norfolk turkey with sage mayonnaise and salad
 - North Atlantic prawn, celery and rocket with classic cocktail sauce
 - Swiss Emmental cheese, tomato salsa and green salad (v)-
 - Smashed chick pea and kale pesto with grilled pepper (vg)
- Finger food:
 - Cajun spiced chicken skewer with burnt corn and coriander dip (gf)
 - Roasted Mediterranean vegetable skewer with fresh basil paste (vg) (gf)
 - Courgette, beetroot and feta tart (v)
- Special dietary sandwiches are individually packed and labelled.

During the break

- Visit the stalls
- Hand in data check sheets and gift aid forms
- Post on social media about today
Use **#ANUKconf**
Twitter: @AniridiaNetUK
- Film a vox-pops
- Sessions begin here at 1:00pm

Please silence mobile devices





Aniridia Network

Explanation of Access to Work

Geoff Taylor Meade

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Department
for Work &
Pensions

Access to Work



An Introduction to Access to Work

 www.gov.uk/access-to-work

Access to Work

- Access to Work was introduced in June 1994
- Is a National Programme delivered by Department for Work and Pensions
- To help overcome barriers that disabled people come across when moving onto or retaining employment.
- Provides a grant to cover additional costs over and above the requirements of making “reasonable adjustments” which an employer is legally obliged to provide under the “Equality Act 2010”
- A flexible programme that focuses on the needs of the individual.

What is a Reasonable Adjustment?

Summary

Where someone meets the definition of a disabled person in the Equality Act 2010 (the Act) employers are required to make reasonable adjustments to any elements of the job which place a disabled person at a substantial disadvantage compared to non-disabled people.

Employers are only required to make adjustments that are reasonable. Factors such as the cost and practicability of making an adjustment and the resources available to the employer may be relevant in deciding what is reasonable

3 Requirements of Employers Duty to make Reasonable Adjustments

There are 3 main considerations when determining what reasonable adjustments could be put in place for an employee

- Changes to the ways in which things are done in the organisation
- Changes to overcome the barriers created by the physical features of the workplace
- To provide extra / auxiliary equipment

Benefits of Access to Work

- Encourages greater independence
- Promotes work as being the best route to inclusion for disabled people
- Enables disabled people to work on a more equal basis with non disabled colleagues
- Encourages employers to recruit and retain disabled people by offering practical help.
- Provides advice to disabled people and their employers

Eligibility

Types Of Help



Eligibility Criteria

- Have a disability or health condition that has a long term, adverse affect on their ability to carry out their job
- Be over 16 years old
- Be in, or about to start, paid employment (including self employment)
- Live and work in England, Scotland or Wales
- Not be claiming Incapacity Benefit or ESA once they are in work (with the exception of higher permitted work)

Types Of Help

There are seven main elements within Access to Work:

- Special Aids and Equipment (SAE)
- Adaptations to Premises and equipment (APE)
- Travel to Work (TtW)
- Travel in Work (TiW)
- Support Worker (SW)
- Communication Support at Interview (CSI)
- Mental Health Support Service (MHSS)

Mental Health Support Service

Support is available for people with mental health conditions who are either:

- Going into work
- Absent from work as a result of a mental health condition
- Finding work difficult as a result of a mental health condition

The support available from Access to Work includes:

- An assessment of needs
- A personalised six month support plan, with detailed steps designed to keep a person in, or help them to return to work
- Signposting to relevant intervention and support services

This service is provided for Access to Work by Remploy

Examples of Mental Health Support

Coping strategies

This could cover a range of strategies and will depend on the individual's job role, their condition and how this impacts their work. For example:

- Keeping a mood diary
- Using memory aids such as Mind Maps, checklists
- Relaxation techniques when under pressure
- Developing a Wellbeing Recovery Action Plan
- Cognitive Behavioural techniques

Reasonable adjustments

- Development of Flexible working, phased return, etc.
- Putting in place a buddy or mentor
- Temporary reduction in targets or reallocation
- Additional time to complete certain aspects of job role
- Regular formalised 121 meetings to review concerns

Cost Sharing Financial Support



Access to Work Grants

The level of grant will depend on:

- Whether the applicant is employed or self employed
- How long they have been in their job
- The type of help required
- The size of company they work for

Access to Work provides the grant with which to procure the support that is needed, it does not provide the support itself.

Cost Sharing

- 0 – 49 employees attract no cost share
- 50 – 249 employees - £500 Threshold and 20% of the costs thereafter
- Over 250 employees - £1000 Threshold and 20% of the costs thereafter
- Costs above £10,000 will normally be met by Access to Work
- If there is a general business benefit a contribution will be sought in addition to any compulsory cost share

How to Apply Contact Details



How to Apply

- Disabled employee makes the application
- Telephone applications - Alternative arrangements can be made.
- The Customer Service Team will take basic details
- A stencil will be sent to an adviser who will call the customer and undertake an eligibility check and appraisal of need.
- Once the appraisal has been undertaken this will help the adviser make an informed decision to award support or identify if a further workplace holistic assessment is required before being able to identify what support is required (this will mainly be for specialist aids)

Further Information

Further information about Access to Work can be found on the internet at

www.gov.uk/access-to-work
[Employer's Guide to Access to Work](#)

You can contact the Customer Service Team with any questions on:

Telephone: 0345 268 8489

Textphone: 0345 608 8753

Email: atwosu.london@dwp.gsi.gov.uk

This presentation is intended to be a general guide to the principles underpinning the Access to Work programme and is not a full and authoritative statement of the law



Aniridia Network

Visual impairment, depression & mental health services

Dr Ian Petch

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Visual Impairment, Depression and Access to Psychological Therapies

Ian Petch

Aims

Visual impairment and depression

Are psychological therapy services used by people with a visual impairment?

Opportunities to improve access to psychological therapies?

Test the impact of training

Method

1. Review

- the literature
- national data

2. Focus groups

- people with visual impairment
- professionals
- lay individuals

3. Test a brief training workshop
to improve confidence

Depression

- Association between visual impairment and increased risk of depression
- How strong is that association?
 - Depends on the method used in the study
 - Diagnosable depression
 - Symptoms like depression

What explains the link?

- Visual functioning rather than visual impairment
- Depression is a strong predictor of visual functioning

- **Non-specific factors**

- economic status
- social isolation
- physical health co-morbidities
- reduced access to healthcare
- reduced access to valued activities

Impact on rehabilitation

- reduced use of rehabilitation services
- improved adaptation results in limited improvement in depression
- depression-specific interventions ?

Access to psychological treatments

- IAPT : Improving Access to Psychological Therapies
- 6% over age 65
- Problems recording all types of disability (80% of 1.27 million people have no record regarding disability)

People with visual impairment who do access IAPT

Equally as likely to

- remain in treatment
- engage in routine measurement of progress
- present with similar levels of depression and anxiety as those without a visual impairment

IAPT

(%)	'Sight'	Other disabilities	No disability	No code recorded
Enter Treatment	79	76	81	61
Complete Treatment	59	58	66	55
Reliable Improvement	59	54	59	61
Recovery	41	34	43	46

Stakeholder views

- 30 people with visual impairment
- 14 support staff
- 2 GPS with specialist interest in VI
- 2 ECLOs
- 2 Optometrists
- Clinical staff in IAPT – review of last 6 people with VI receiving treatment

Open interviews

- Disclosing mental health needs
- Staff recognising mental health needs
- Are services accessible?
- Do people with VI want what is offered?

What did we find?

- Depression is often not recognised among people with VI
- Trust and rapport
- Still adjusting to sight loss of depression?

- Readiness to discuss depression can change over time
- Staff often don't feel confident to talk to people about depression

- Support staff, family carers and peers are important to people with VI in supporting access to any type of service
- Peer networks and support outside of healthcare may be more accessible
- These settings might be a route to improving access to treatment

Training

- 50 people

Volunteer	8	(16%)
Befriender	6	(12%)
Rehabilitation Worker	18	(36%)
Social Care Staff	5	(10%)
Low vision clinic support	6	(12%)
Other	7	(14%)

Improve Knowledge and Confidence

- 1 Link between vision impairment and depression
- 2 Recognising depression
- 3 Responding to depression
- 4 Encouraging the person to seek professional help

Format

One workshop – 3 hours

(4 weeks later)

Action learning set – 1 hour

Outcomes

- Thank you Dr Gwynn Rees for permission to use her measures

Outcomes

	Increase	Decrease
Confidence recognising & responding to depression	86%	8%
Responding to depression	70%	24%
Barriers to recognition & responding	No change	

- *My limited knowledge of depression means that people may not always receive the best management for depression*
- *My poor knowledge of what to do if a person could be depressed means that they may not always receive the best care*

Workshop

One session was sufficient	84%
Made the signs and symptoms of depression clear	96%
I feel more confident bringing up emotional issues with people I support	98%
I know where and to whom I can refer people	98%
I intend to discuss the screening and referral system with my colleagues	96%
I intend to use the screening tool with my clients	90%
When I suspect clients are depressed I intend to suggest they consult their GP	90%

Conclusion

- First report in the UK
- A brief single training session for staff supporting people with visual impairment is feasible
- Improvements in confidence including directing an individual to further sources of support

- Increased risk of depression in VI
- Limited access to psychological treatments
- Increased recognition and support to access is needed
- Staff, peers, family and friends may be key
- Brief training is feasible and can enhance confidence



Aniridia Network

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Aniridia Network

Introduction to Nystagmus Network

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Nystagmus *Network*

Sue Ricketts

Executive Information and
Development Manager

Nystagmus **Network**

sue.ricketts@nystagmusnetwork.org



Nystagmus *Network*

Nystagmus **Network**
– we're here to support
adults with nystagmus,
parents and their
children

www.nystagmusnetwork.org

Founded in 1984

Vivien Jones

founded the charity to support families like hers. This is our charity's story:



https://www.youtube.com/watch?v=5F_Qfuatnso

Our objectives

- to provide **information** and **support**
- to raise **awareness** of the condition
- to fund **research**



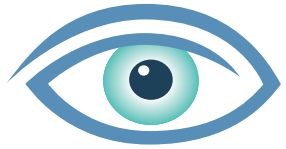
Nystagmus *Network*



Nystagmus *Network*

for information and support

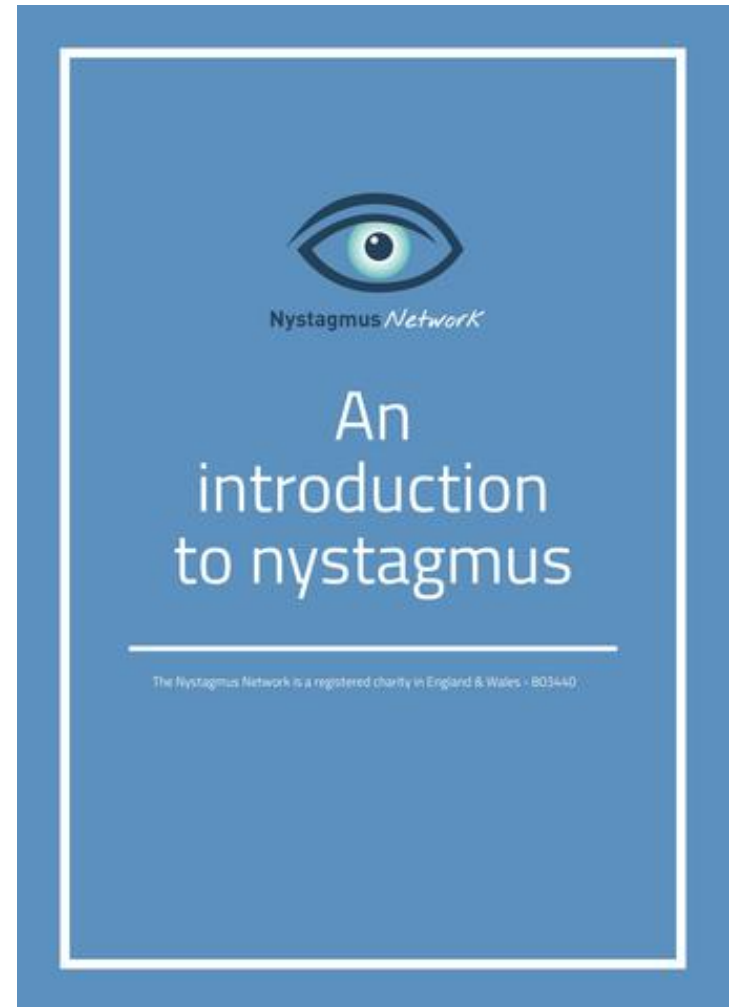
call **01427 718093**



Nystagmus *Network*

Our **free** to
download
booklets
are available
from

www.nystagmusnet.org



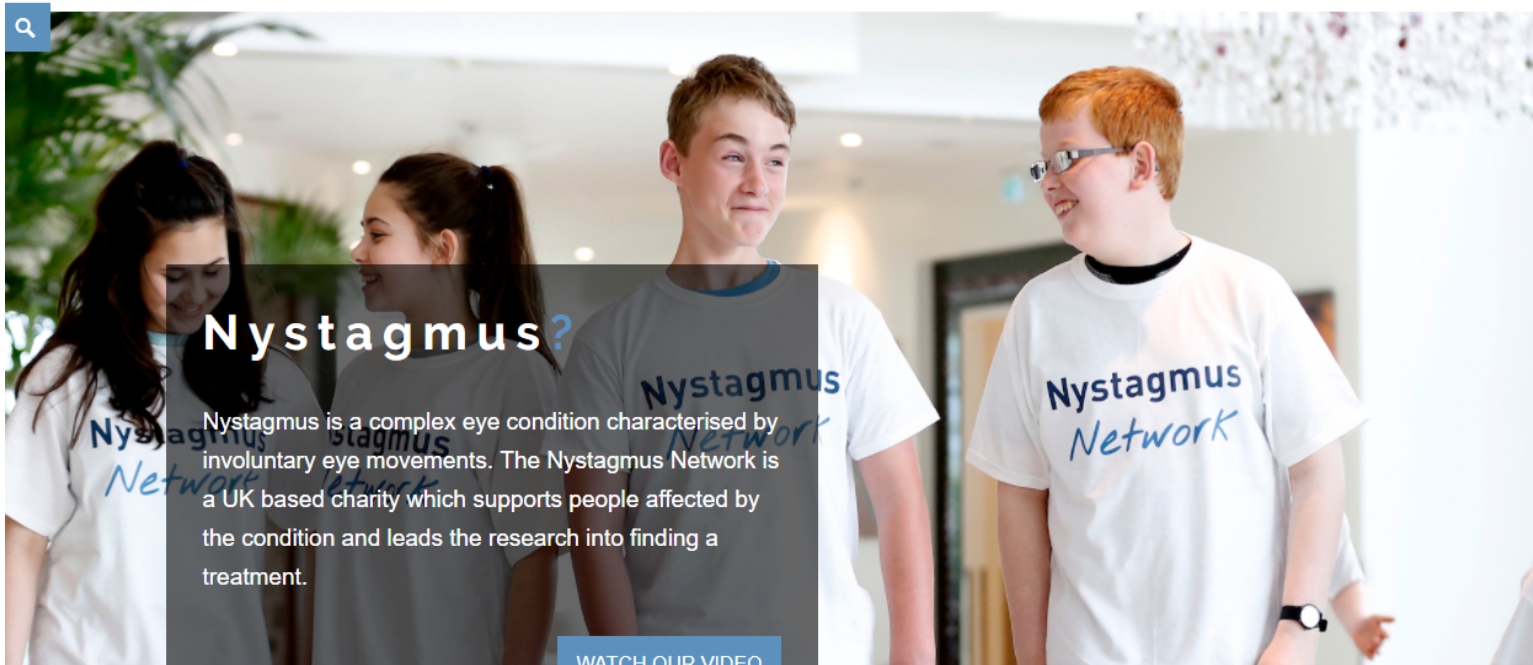


Nystagmus *Network*



**Education
advice,
guidance
and advocacy**

Coming soon - an **education support** information base





Nystagmus *Network*

We need to talk about nystagmus



Nystagmus *Network*

Nystagmus *Awareness Day* 20 June

Nystagmus *in the open* Garden Open Day

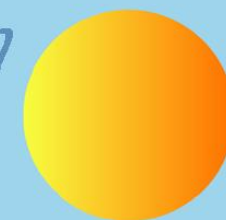
The Barn, Fenton

24 June

11am to 4pm

Nystagmus *Network*

Charity number 803440



cakes

crafts



plants

books



Entry £3 *per person*

Children *free*

Sorry, *no dogs*



Nystagmus *Network*



Research



Nystagmus *Network*

Julian Jackson and the **Big Blind Walk**



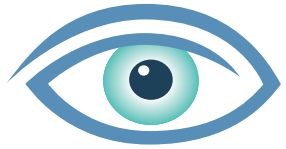
<https://bigblindwalk.com/>



Nystagmus *Network*

Join us for our annual **Open Day** Saturday, 29 September Birmingham





Nystagmus *Network*



Marsha
de Cordova

David Katz





Nystagmus *Network*

Nystagmus, **by George, 11**

- It means **my** eyes sometimes **move** without me wanting them to
- It means sometimes I find it **difficult** to see things
- It means I can only see about 3 metres **clearly**
- It **hurts** my eyes if I have to look at long distances

Did you know?

TV presenter,
**Richard
Osman**, has
nystagmus.
He also stars in
our video:



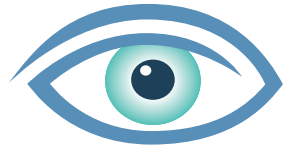
Nystagmus – the way we see it

https://m.youtube.com/watch?v=Ey-UD5Vzu_Q

In our video Harriet talks about the **support** she has at school



https://www.youtube.com/watch?v=Ey-UD5Vzu_Q



Nystagmus *Network*

What is **nystagmus**?

- involuntary **movement** of the eyes (wobbly eyes)
- a recognised **visual disability**
- a **lifelong** condition
- a **stand-alone** condition or associated with a variety of conditions or syndromes, such as Downs Syndrome and Albinism
- **incurable** - currently



Nystagmus *Network*

Social and emotional aspects of nystagmus

- **eye contact**
- **first impressions**
- **social cues**
- **recognise friends**
- **join in**
- **low self esteem**
- **isolation**
- **depression**



Nystagmus *Network*

Bullying and nystagmus

Bullying happens ...

... it happened to my daughter
and I talk to lots of other
parents who know it happens



Nystagmus *Network*

"I have
nystagmus.
At primary
school
I was called
'**alien eyes**'.
Do I look like
an alien to
you?"





Nystagmus *Network*



"At school I
struggled to
read at first.
Now I have a
degree in
philosophy"



Nystagmus *Network*

**Little charities can
make a difference**



Nystagmus *Network*



Thank you from Nystagmus Network



Nystagmus *Network*

Sue Ricketts

Executive Information and
Development Manager

Nystagmus **Network**

sue.ricketts@nystagmusnetwork.org

Refreshments

- Tea, coffee and cakes
- Cakes

During the break

- Visit the stalls
- Hand in data check sheets and gift aid forms
- Post on social media about today
Use **#ANUKconf**
Twitter: @AniridiaNetUK
- Film a vox-pops
- Sessions begin here at 15:45pm

Please silence mobile devices



Raffle

And the winners
are...



Aniridia Network

Growing up and moving to London with aniridia

Glen Turner

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Twitter: **@AniridiaNetUK**

Facebook: **AniridiaNetworkUK**

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Aniridia Network

Sleep and eye disease

Dr Iona Alexander

Web: **aniridia.org.uk**

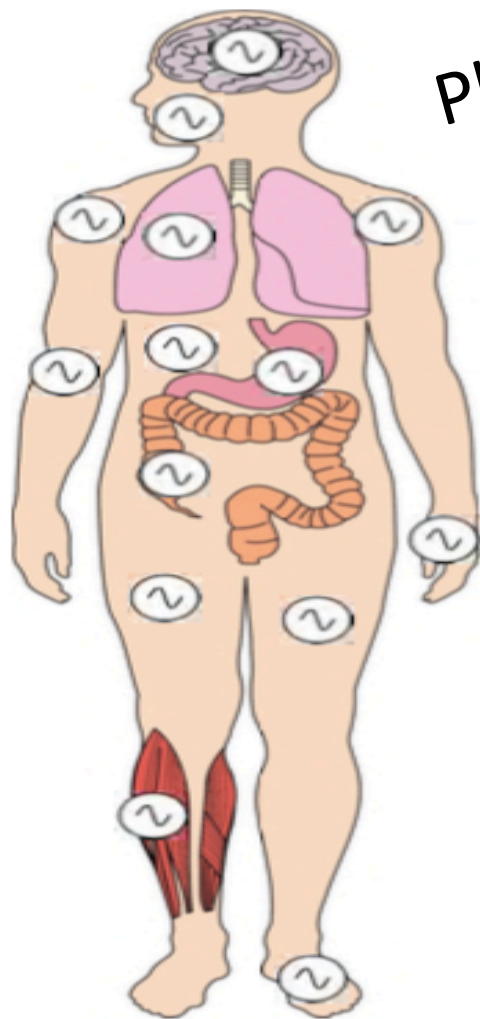
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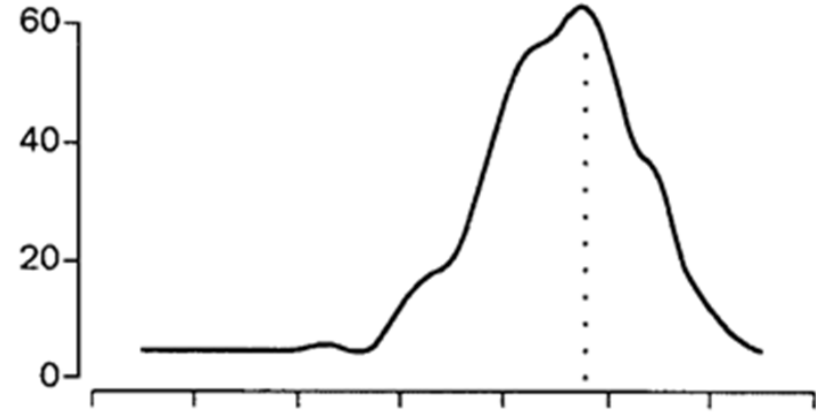
HORMONE -MELATONIN



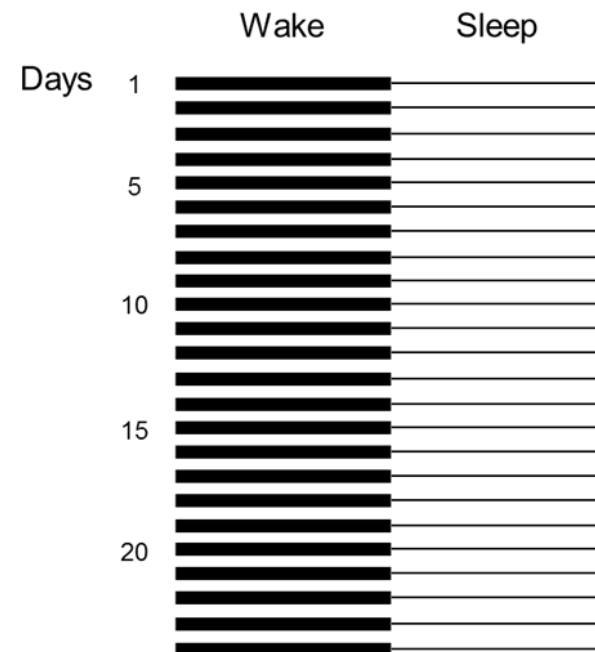
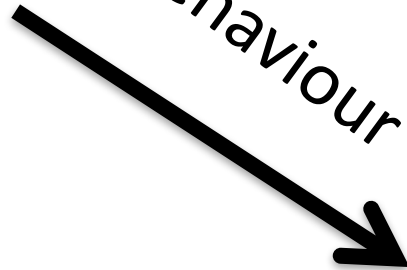
Physiology



A
Plasma melatonin
concentration (pg/mL)

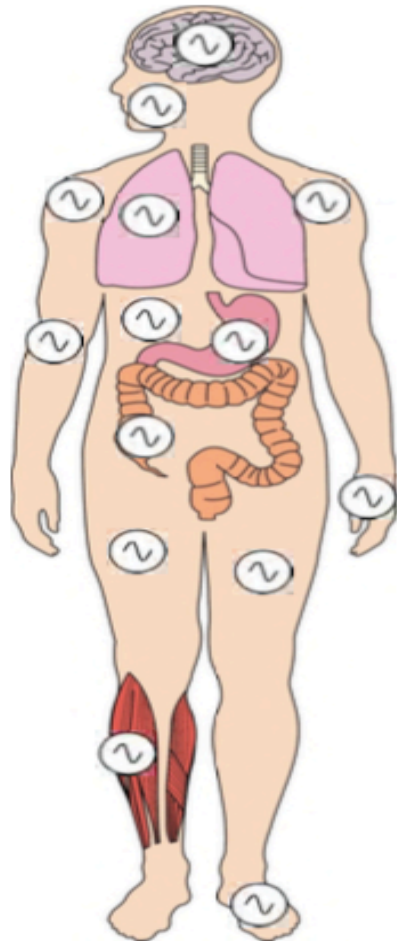


Behaviour



~24 hours

Suprachiasmatic Nucleus (SCN)

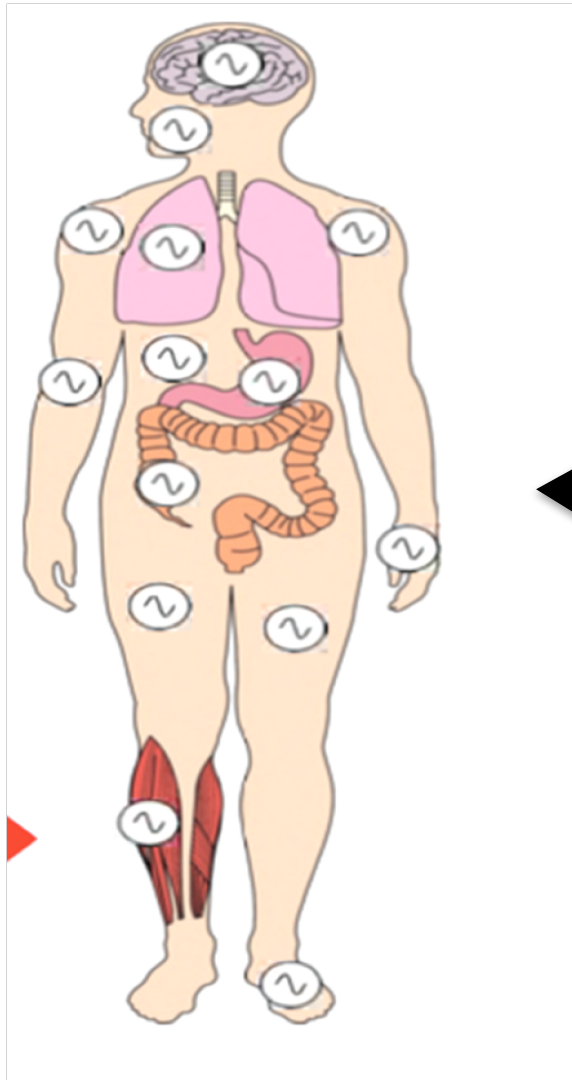


CARTOONSTOCK
.com

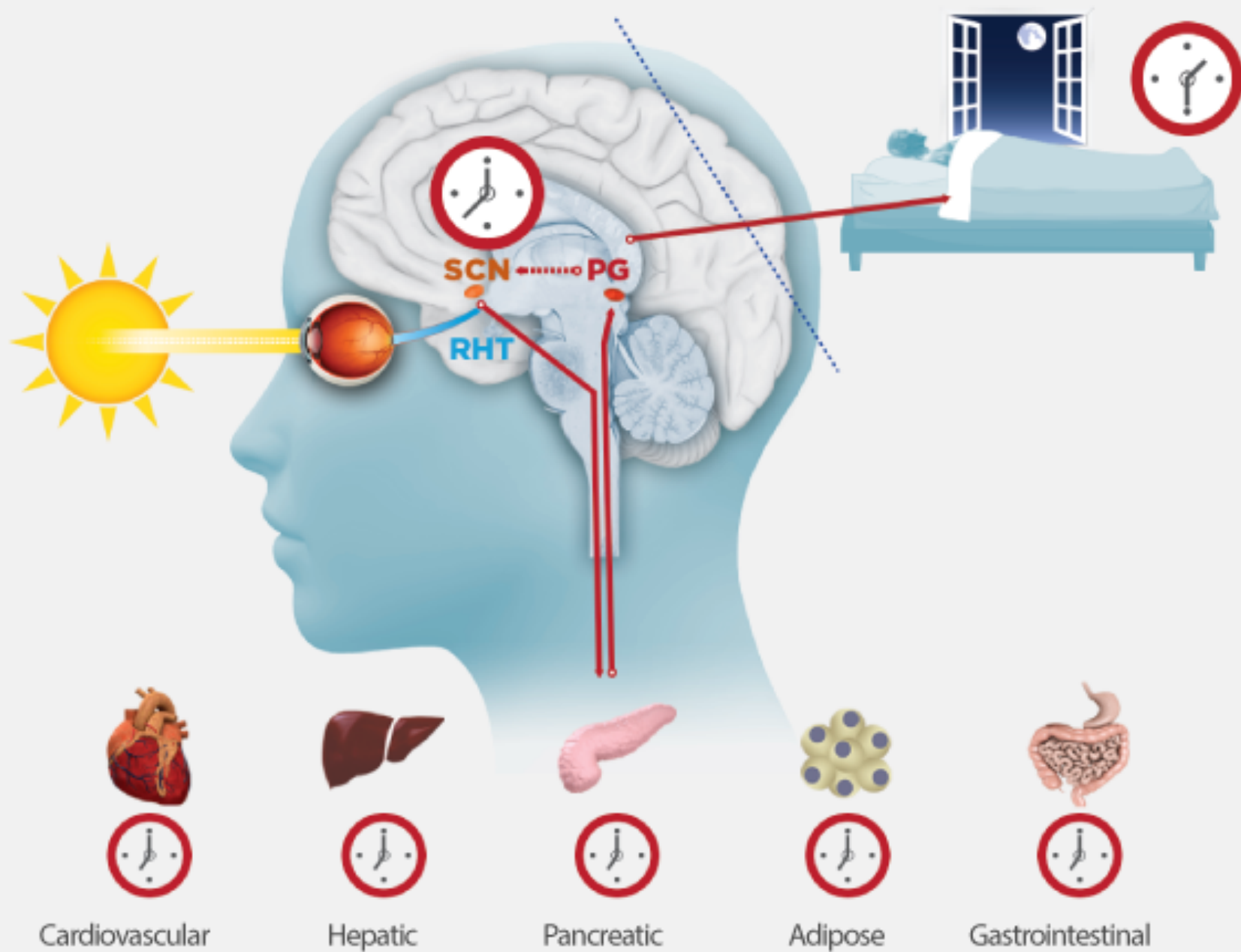
Search ID: jna1026

Body clock time

Environmental time



A clock needs to be set to local time



Why do we sleep?

- ✓ Sleep gives your body a chance to **REST and HEAL**
- ✓ Sleep allows chemicals in your brain to **BALANCE OUT**
- ✓ Sleep allows your brain to build **NEW CONNECTIONS**
- ✓ Sleep allows your brain to **PROCESS LEARNED INFORMATION** and **STORE** it in your **MEMORY**.

Consequences of sleep loss

**Unhappy
Grumpy
Inattentive
Anxious**

Disrupts body clock

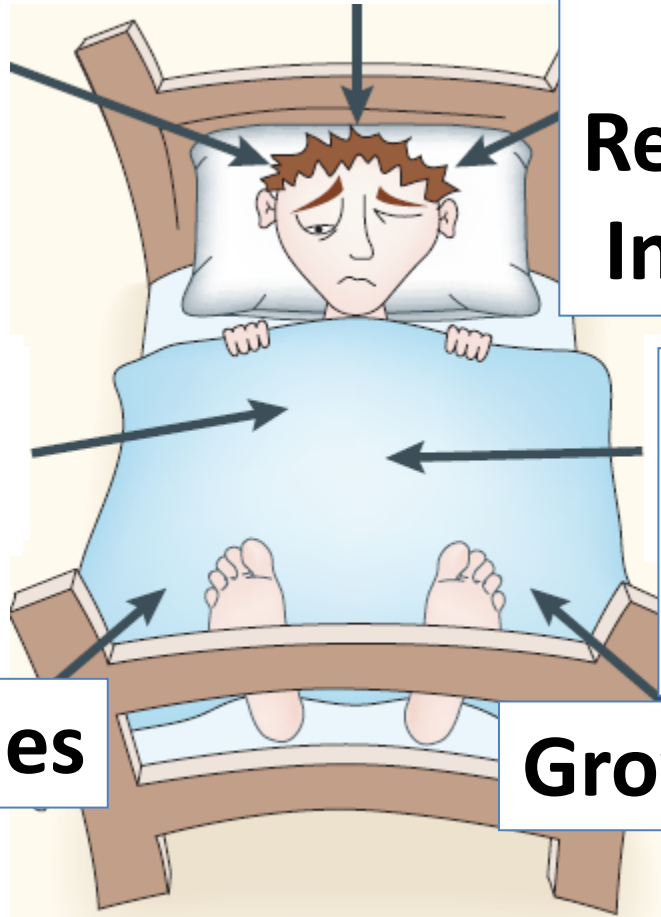
**Fatigue
Reduced learning
Increased errors**

**Weak heart
Getting fat**

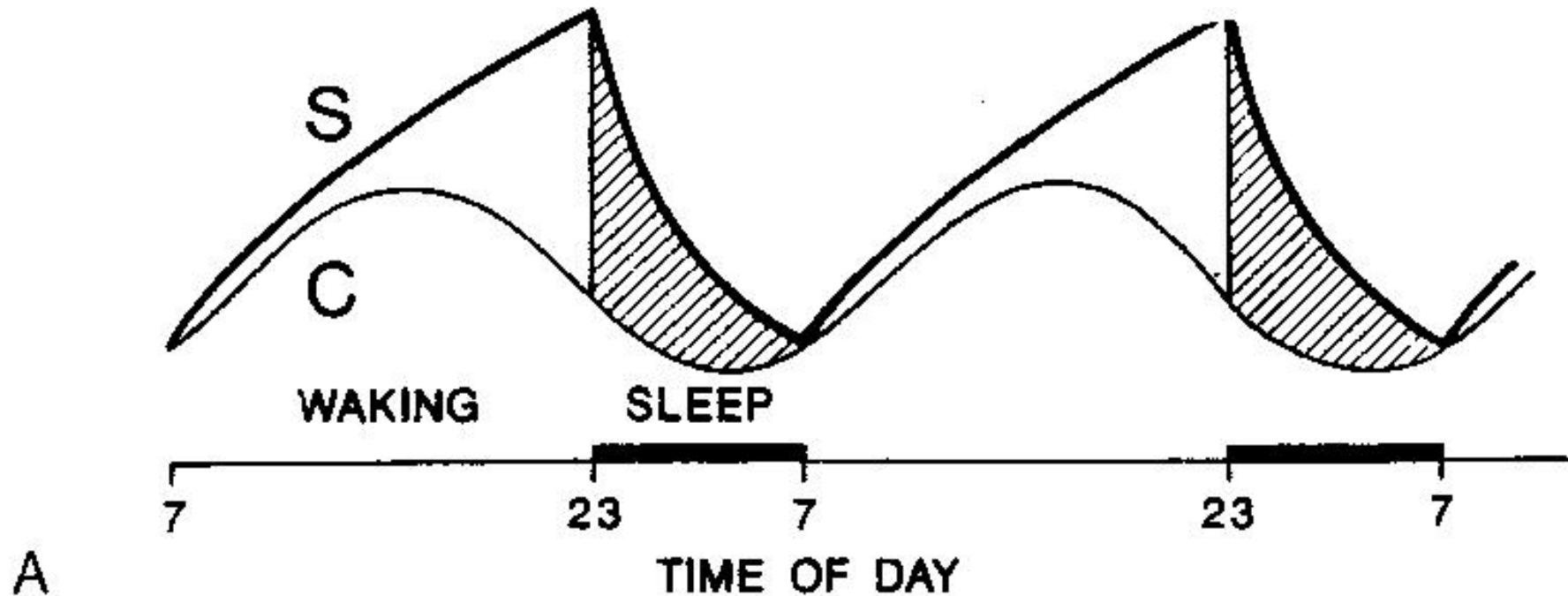
**Getting ill easier
Pain
Risk of diabetes**

Weak muscles

Growth suppression

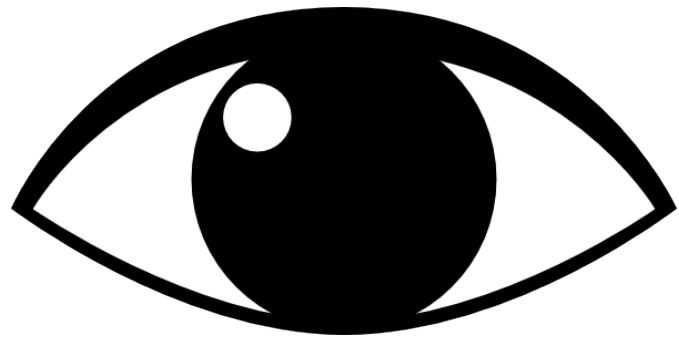


2-process model of sleep regulation (Borbély, 1982)



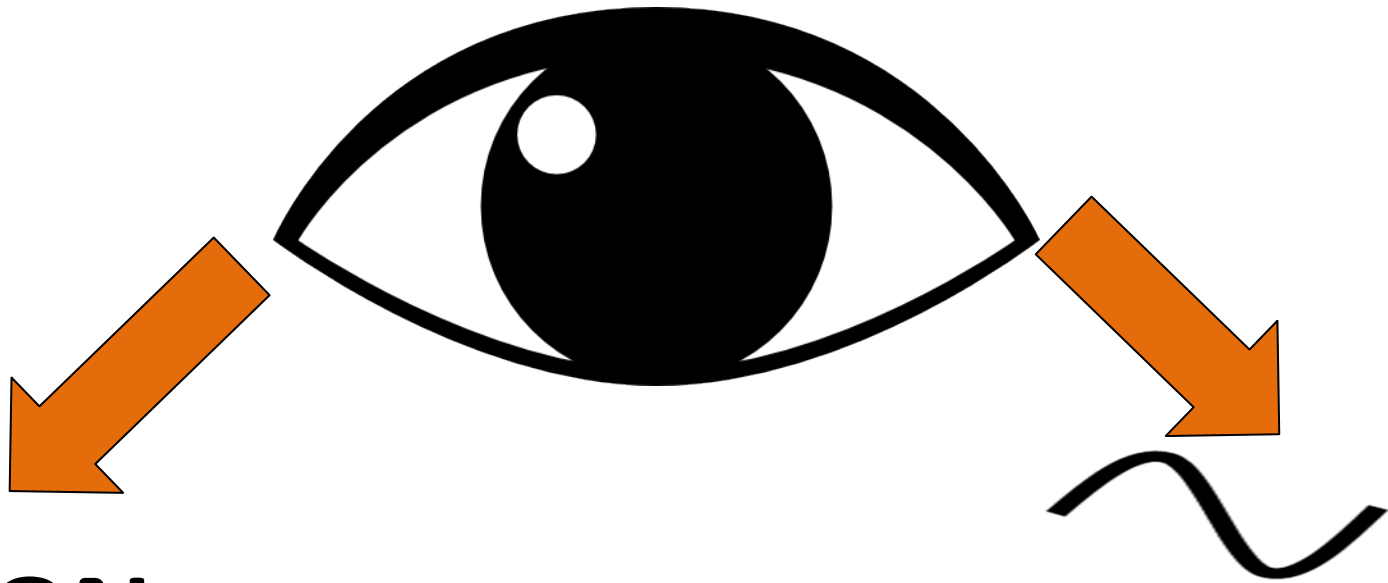
S= the need to sleep increases over the day

C = the system tells our bodies when to sleep (night time)

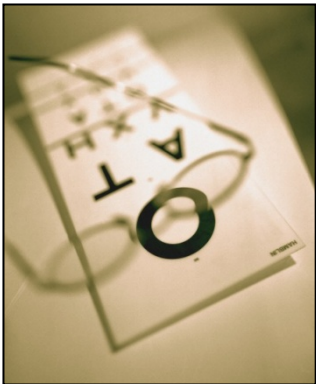


**The eye is important
to synchronise our
internal rhythms
with the external
world**

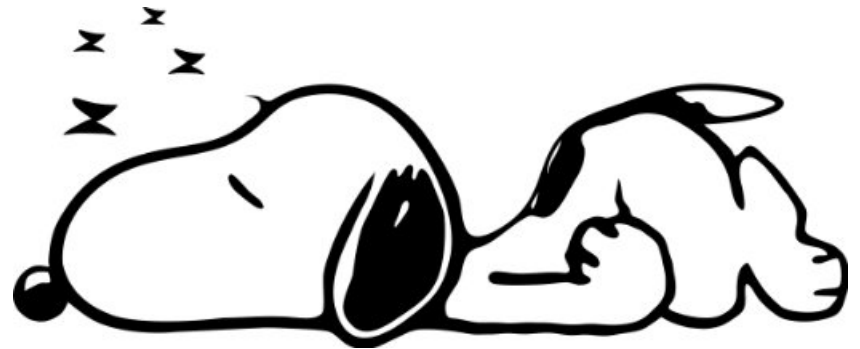
The Dual Function of the Eye



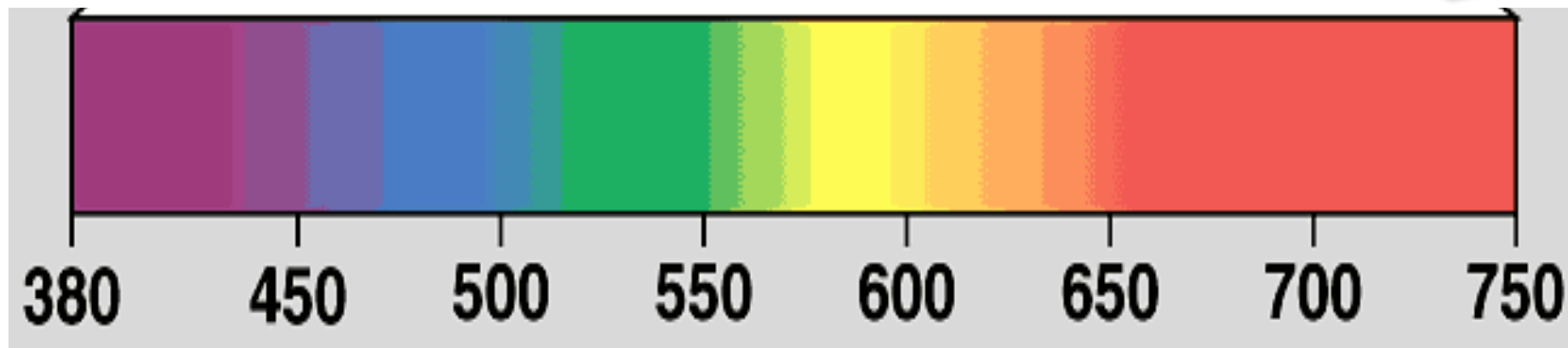
VISION



CIRCADIAN



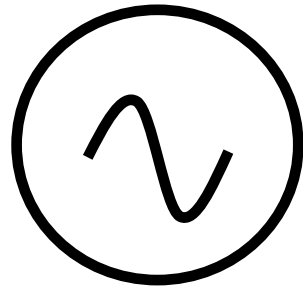
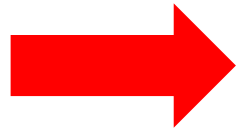
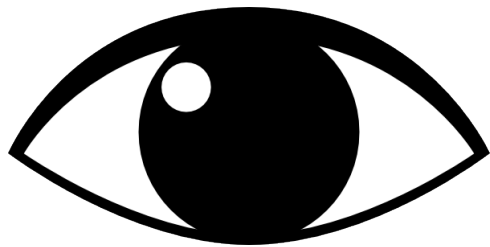
Visible Light



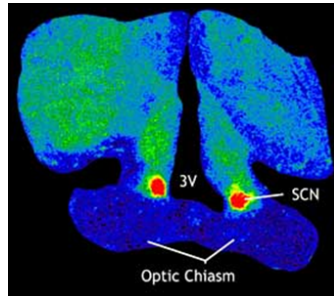
WAVELENGTH

~480nm (pRGC) ~

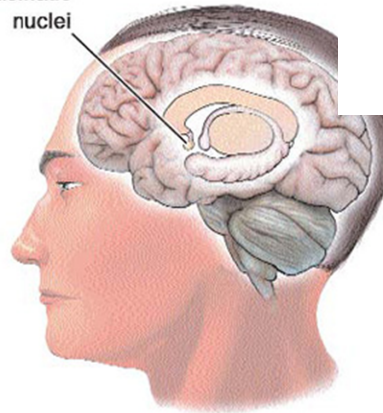
~480nm



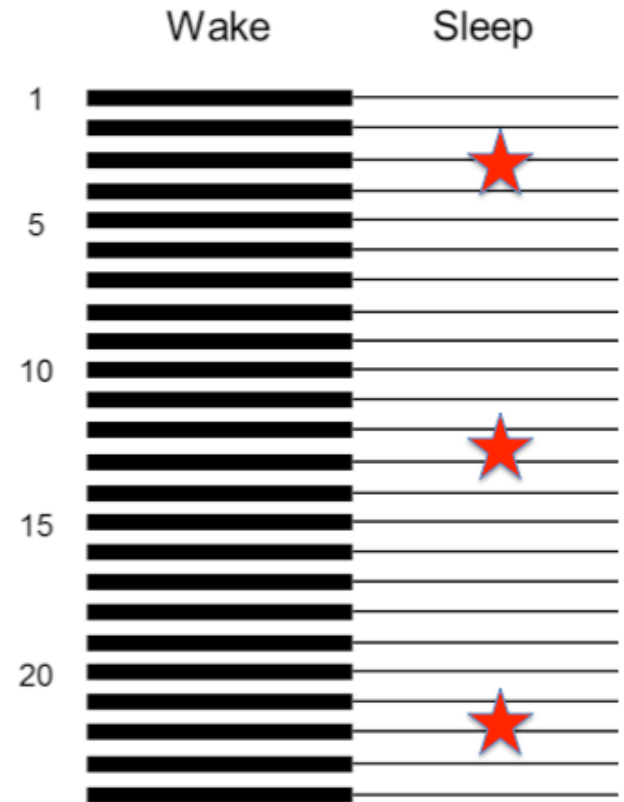
Body clock



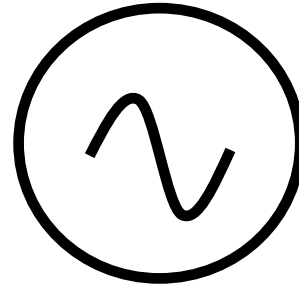
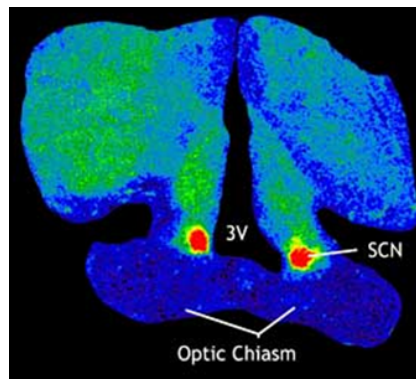
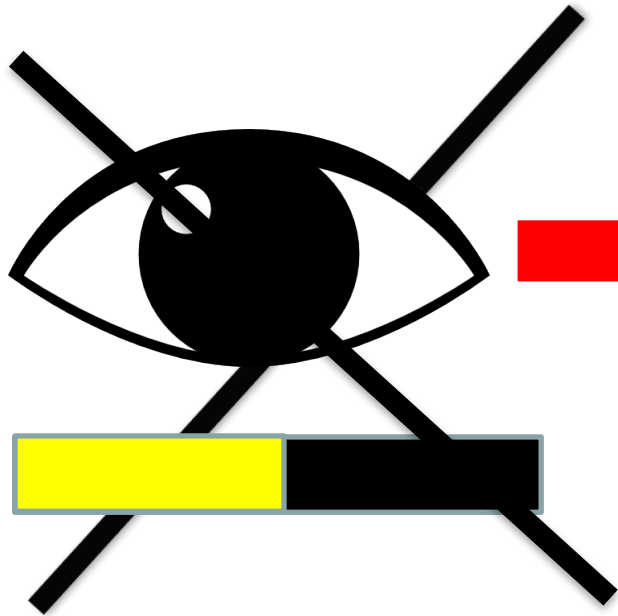
Suprachiasmatic nuclei



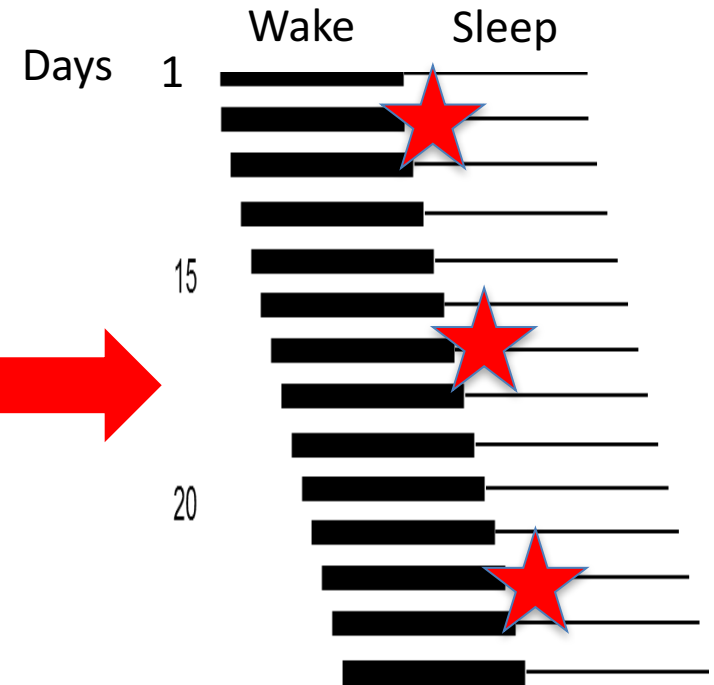
Days



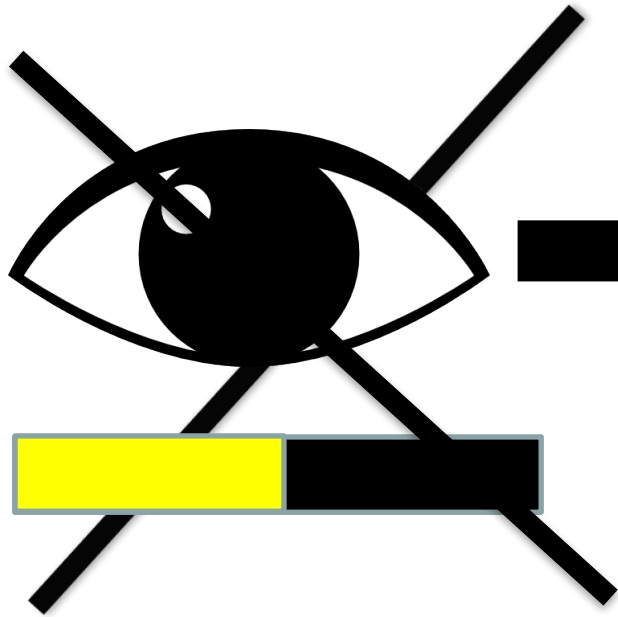
Activity Rest Cycle



Body clock



Freerunning



Photosensitivity



No access to light

Eye disease?

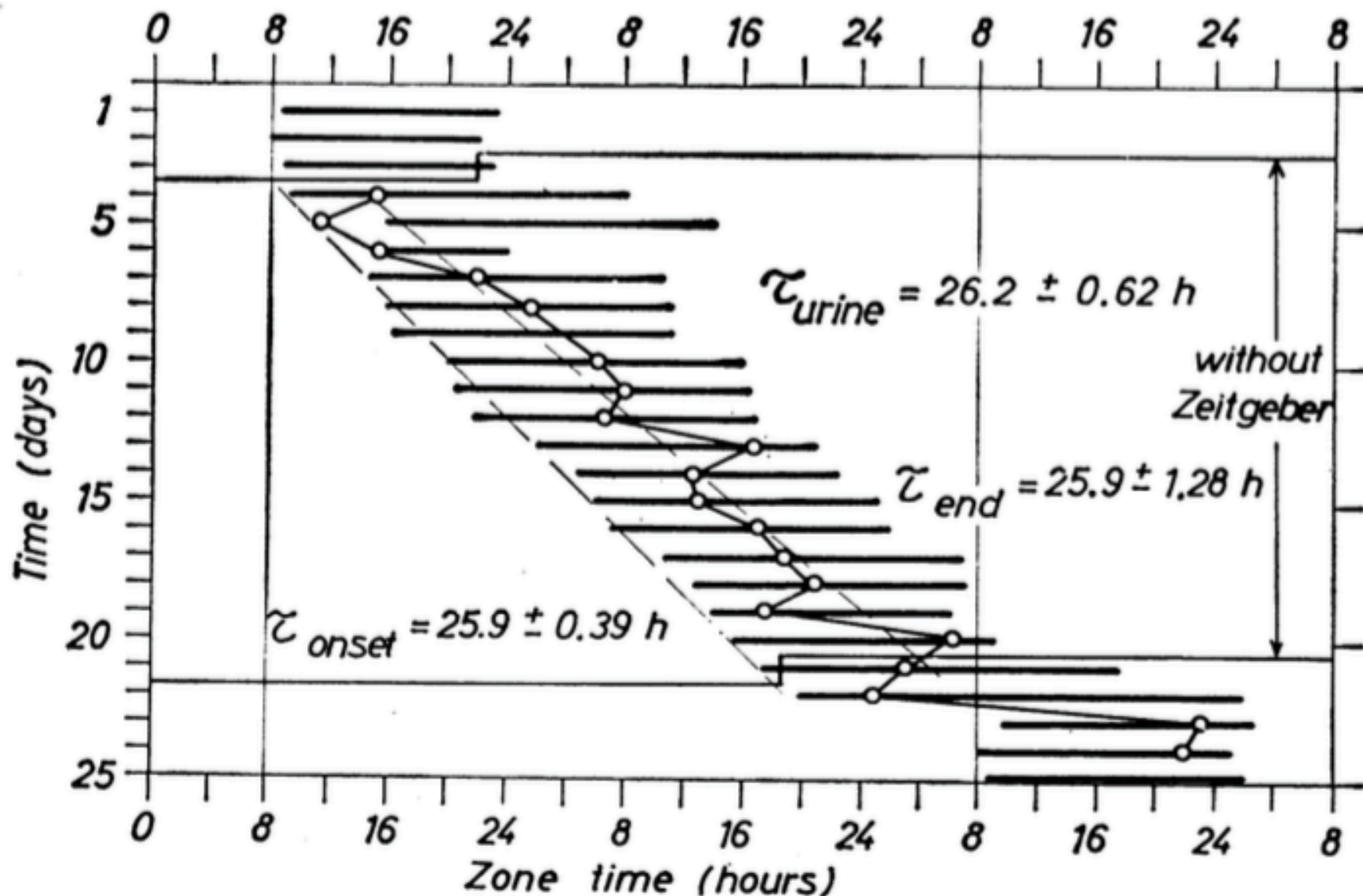
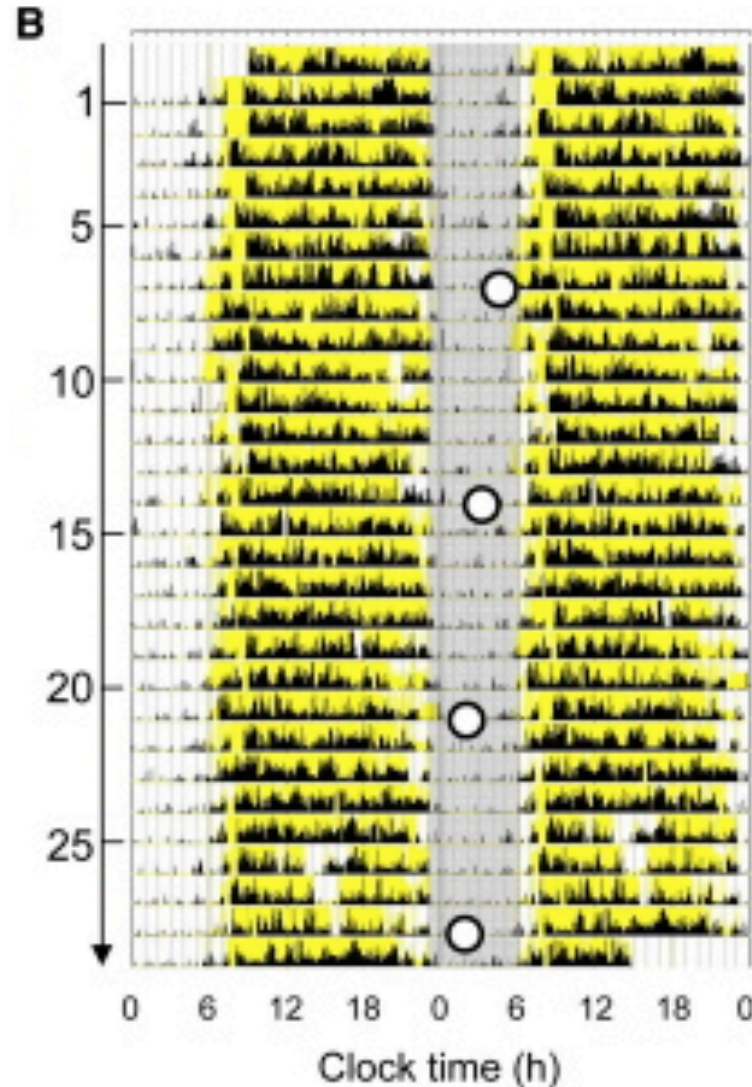


Fig. 4. Circadian rhythm of activity and urine excretion in a human subject kept for 3 days under normal conditions, then for 18 days in isolation, and finally again under normal conditions. Black bars, times of being awake; circles, maxima of urine excretion; τ , mean values of period for onset and end of activity and for urine maxima.

The laboratory

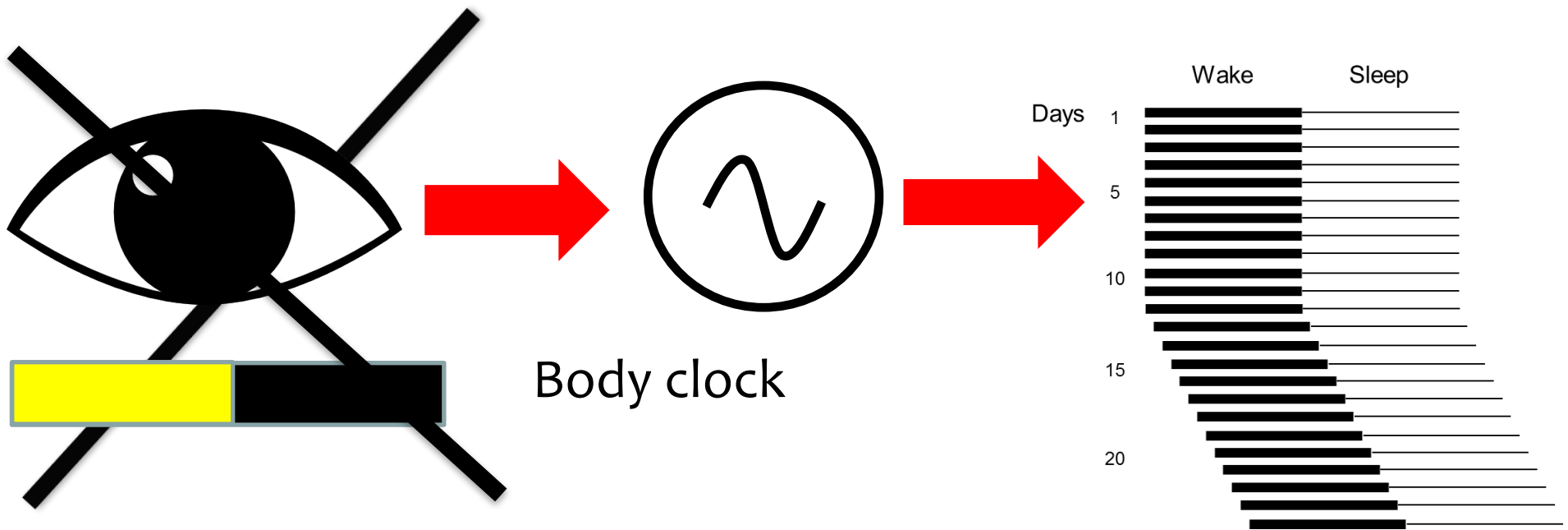


When is blind blind?



Zaidi et al., Curr Biol, 2007

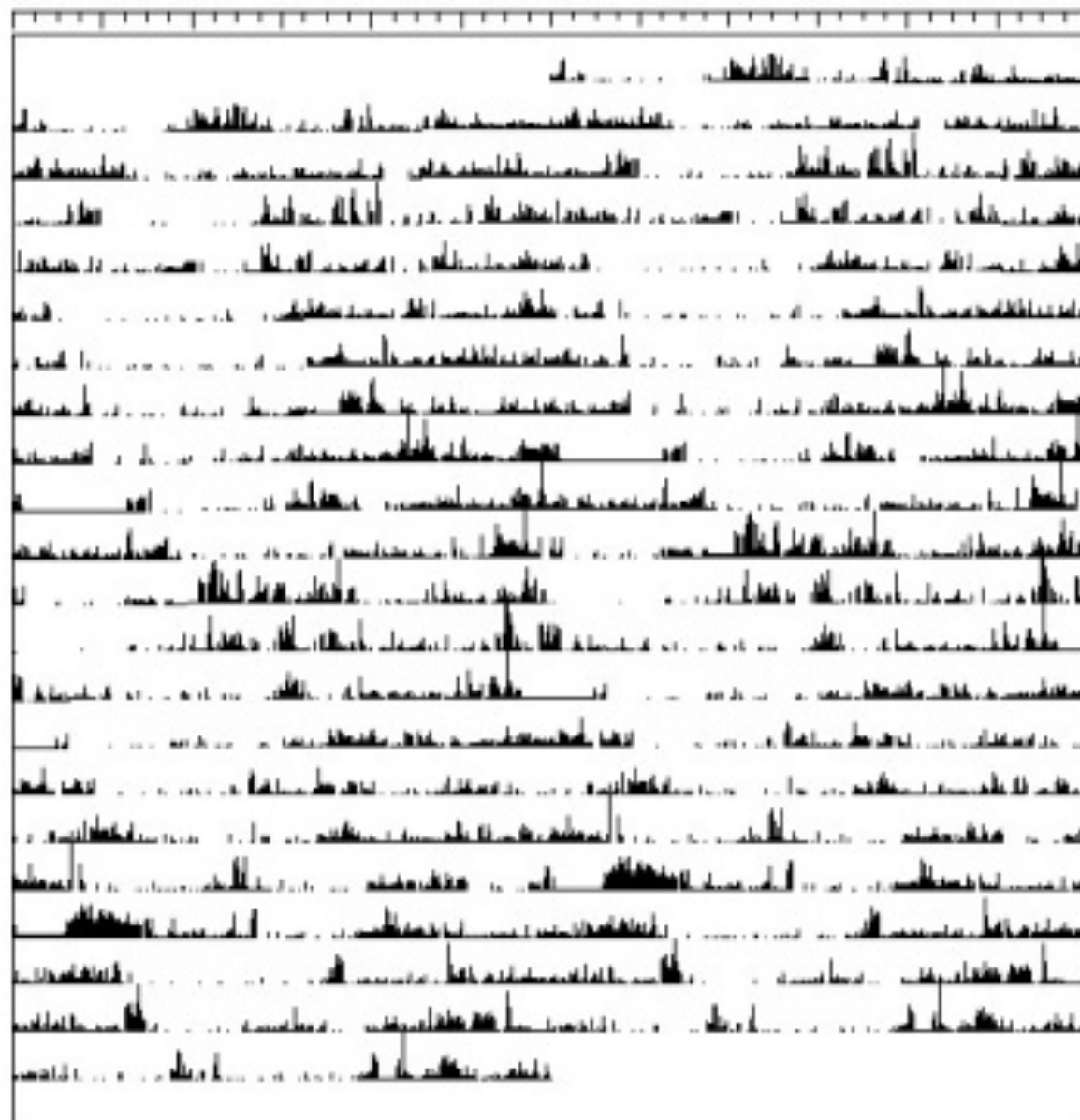
Anophthalmia



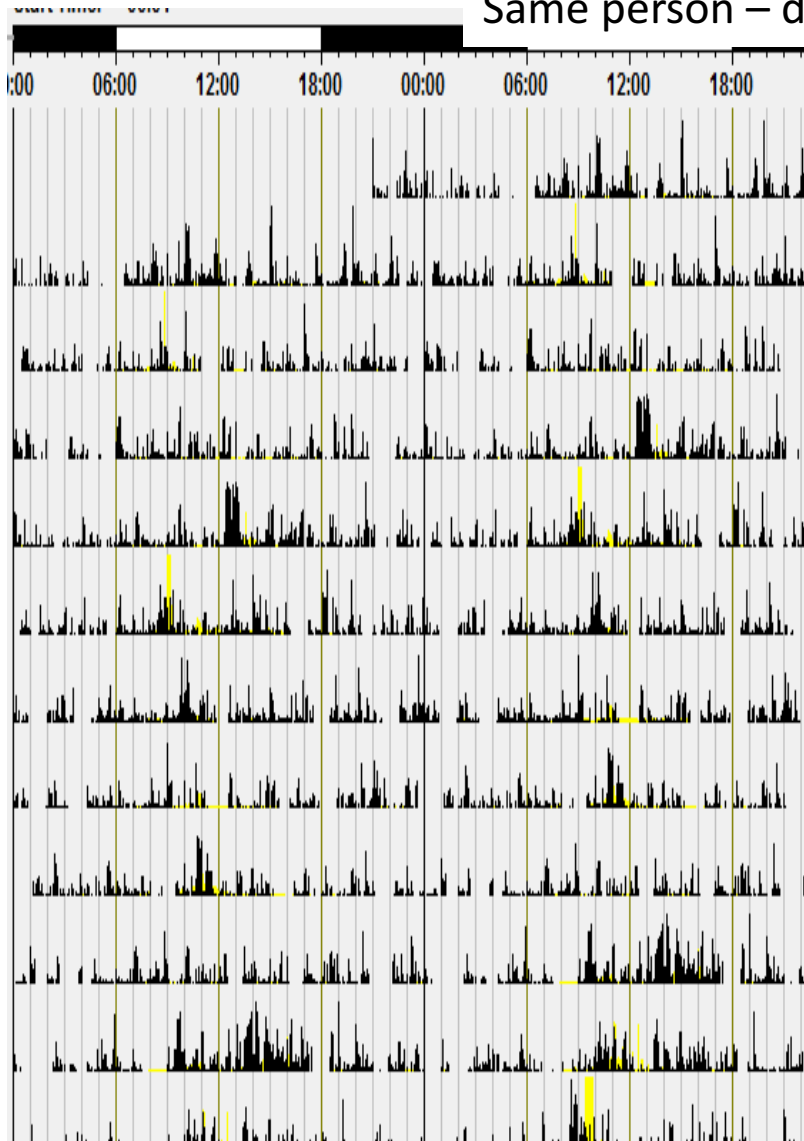
Freerunning

clock time

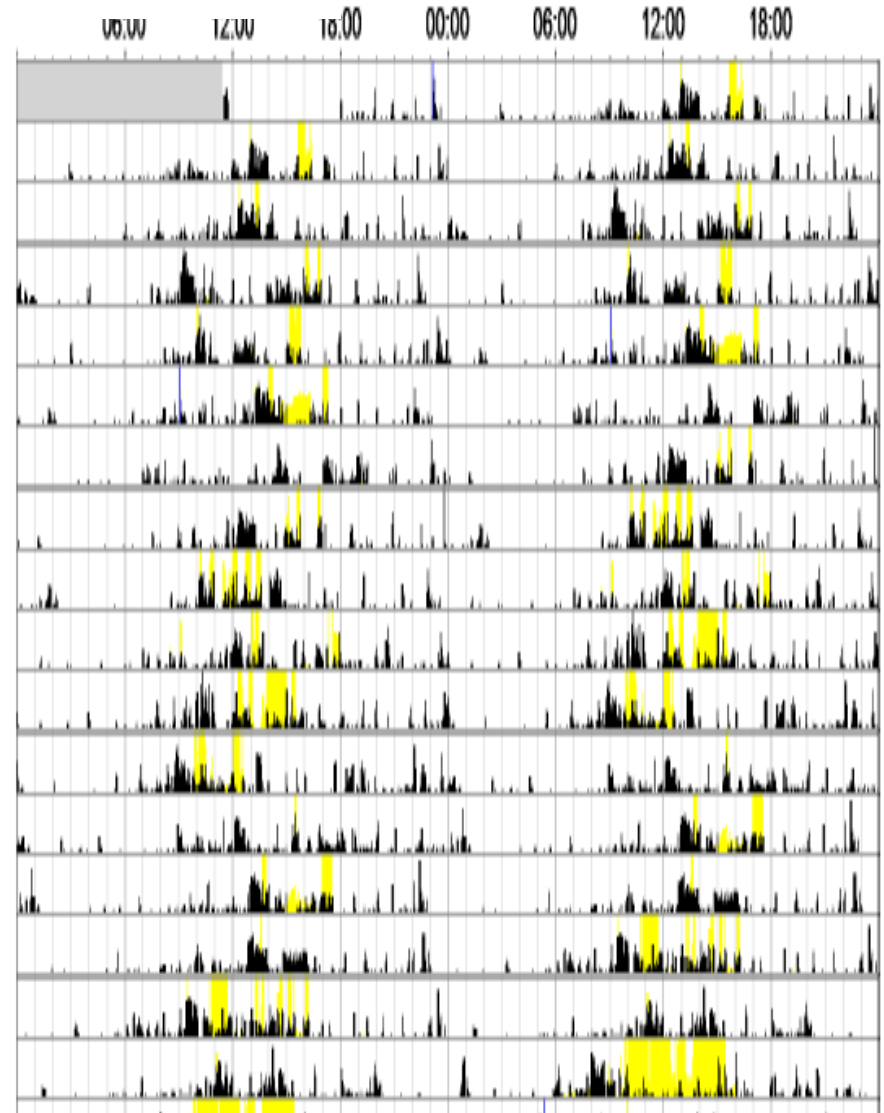
0 4 8 12 16 20 24 28 32 36 40 44 48



Same person – different circumstances



Home alone

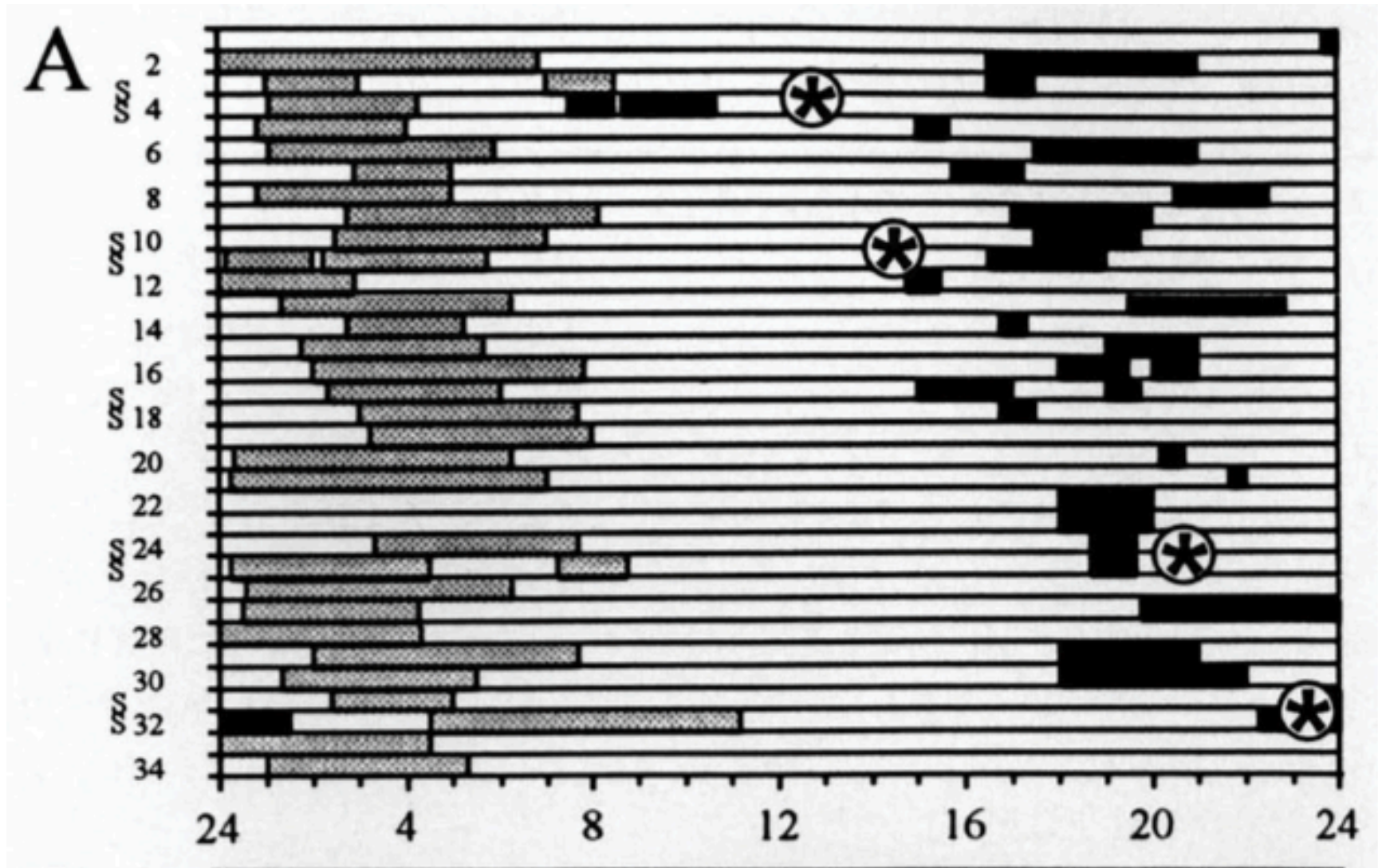


Husband at home

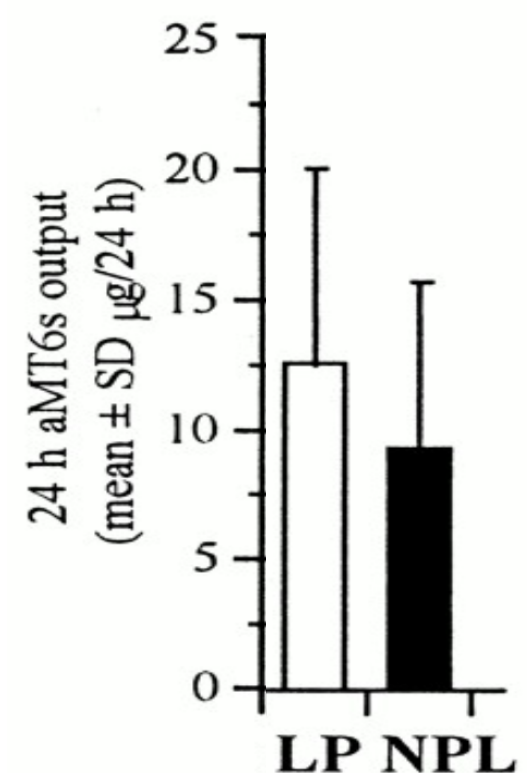
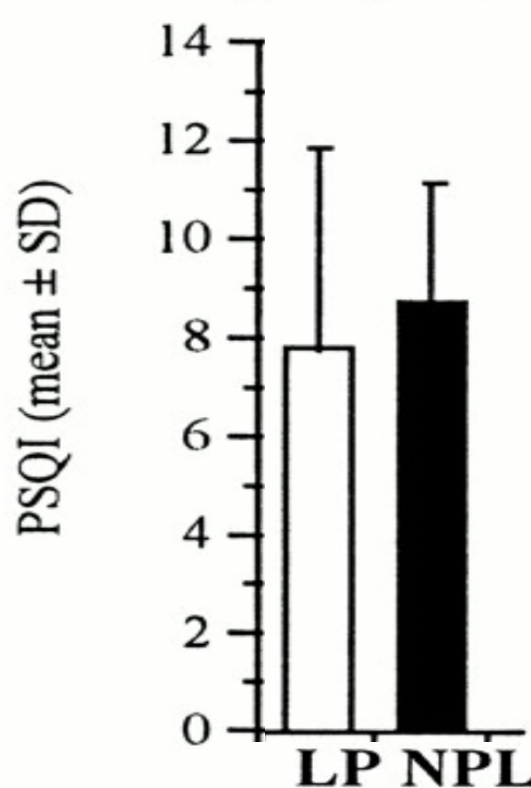
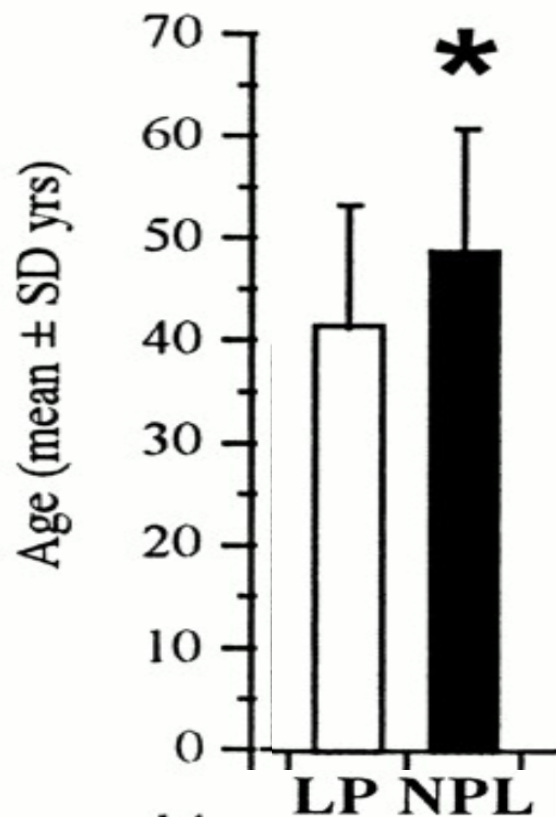
Does the habitual rest-activity period tell us anything about sleep?

How Blind?





Lockley Skene, Napping and melatonin in the blind 1997 journal of biological rhythms 12 pp 16-25



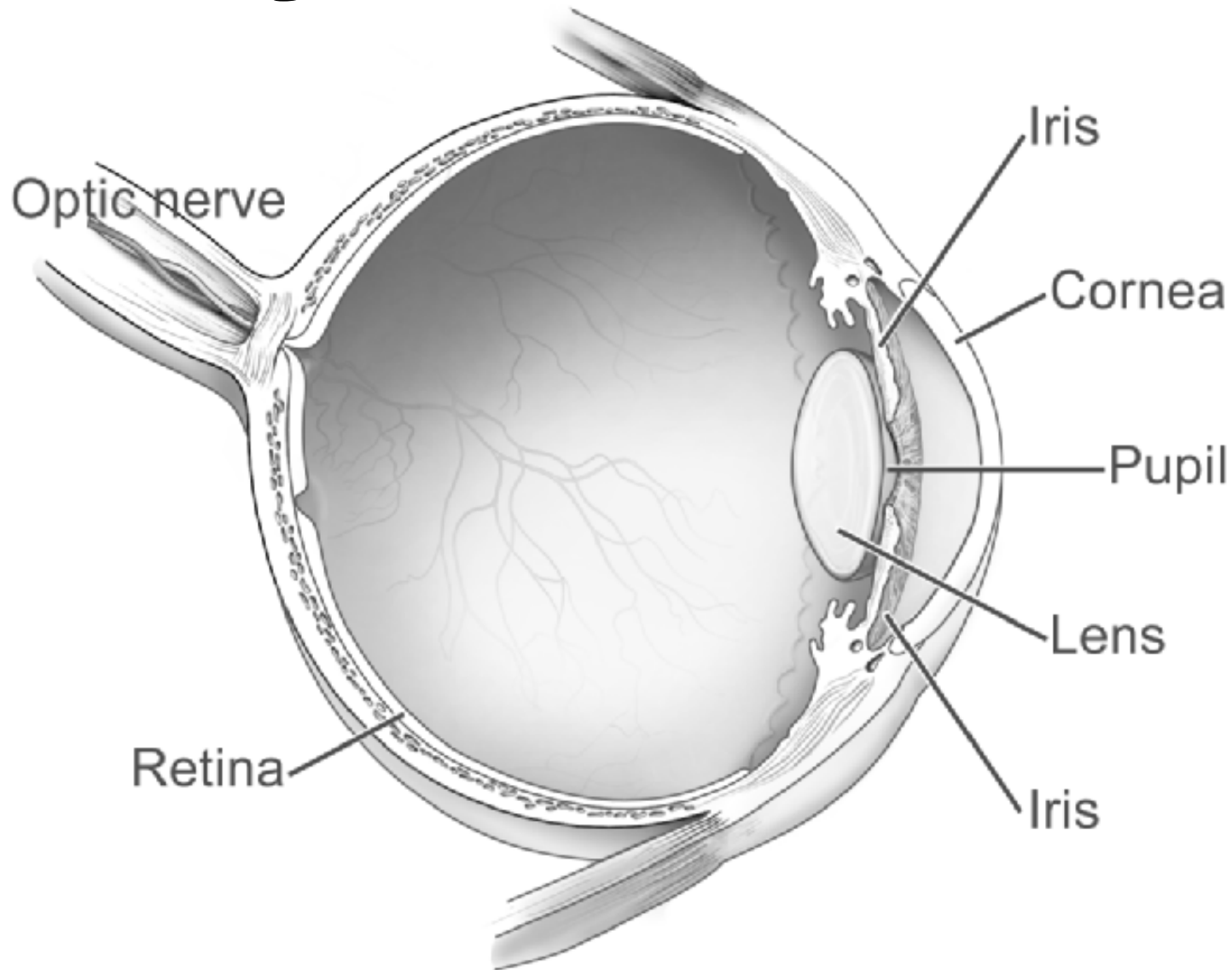
Lockley, S. W., Skene, D. J., Arendt, J., Tabandeh, H., Bird, A. C., & DeFrance, R. (1997). Relationship between melatonin rhythms and visual loss in the blind. *The Journal of Clinical Endocrinology & Metabolism*, 82(11), 3763-3770.

Severity of visual loss	Circadian type				Total
	Normally entrained	Abnormally entrained	Free running	Unclassified	
LP	NE		FR		
≥3/60 vision	4 (57)	2 (29)	0	1 (14)	7
Counting fingers	4 (80)	1 (20)	0	0	5
Hand movements	4 (100)	0	0	0	4
Perception of light	2 (67)	1 (33)	0	0	3
Subtotal	14 (74%)	4 (21)	0	1 (5)	19
NPL					
2 eyes present	4 (33)	5 (42)	2 (17)	1 (8)	12
1 eye present	2 (29)	0	5 (71)	0	7
0 eyes present	1 (9)	0	10 (91)	0	11
Subtotal	7 (23)	5 (17)	17 (57%)	1 (3)	30
Total	21	9	17	2	49

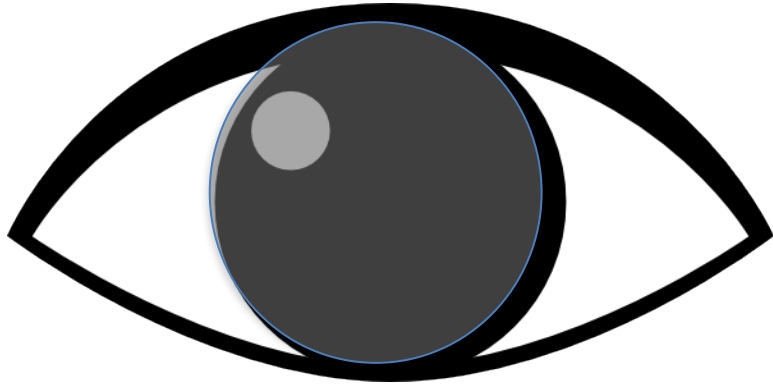
LP

NLP

Eye Health?



Cataract



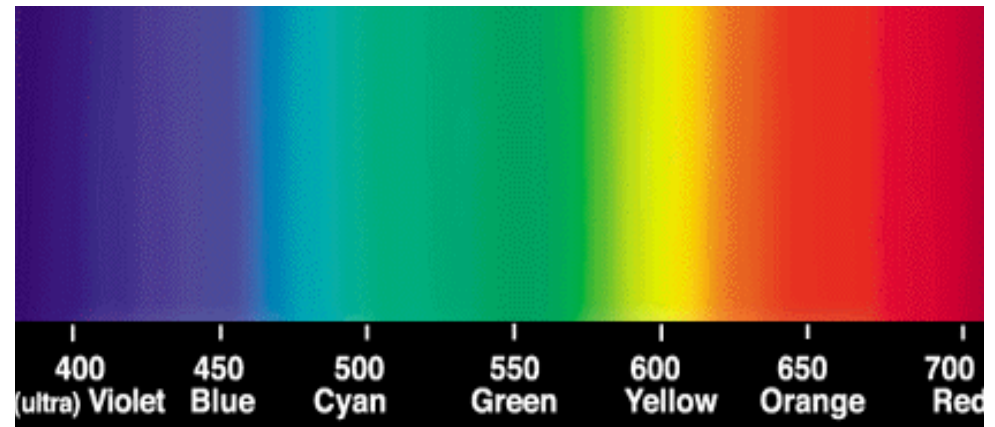
A cataract is an opacity of the normally clear lens, which is obstructing the passage of light.

It may develop as a result of aging, metabolic disorders, trauma or can be hereditary.

Clear or Blue filtering replacement lens?

- Short wavelength light is toxic to the retina and retinal pigment epithelium
 - selectively filtering out these wavelengths could be beneficial

- BUT.....



A study to compare a UV-blocking IOL and a blue-filtering IOL

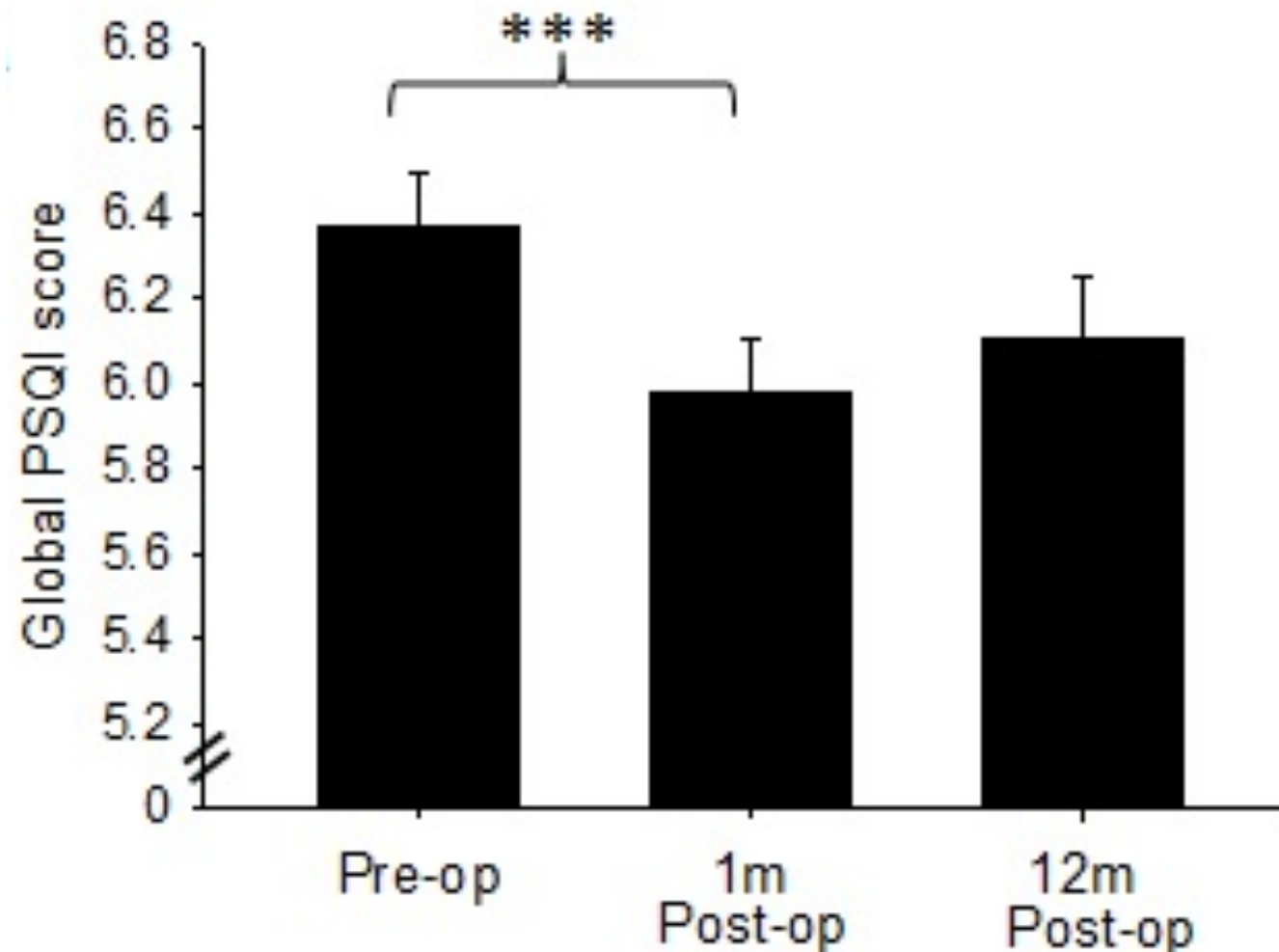
- Compare 498 received a clear intra-ocular lens (IOL) with
- 463 received a blue-filtering (BF) IOL over 3 time points.
- Pittsburgh Sleep Quality Index (PSQI) Questionnaire was completed :
 - 1 month pre-operatively
 - 1 month post-operatively
 - 12 months post-operatively

The PSQI is a validated, self-rating sleep questionnaire that asks about sleep quality over the **past four weeks** and scores **seven different sleep components** (sleep quality; sleep latency; sleep duration; habitual sleep efficiency; sleep disturbance; use of medication; daytime dysfunction) on a scale of 0-3. These component scores are added together to calculate a global PSQI score. **The higher the score, the poorer the sleep**

A PSQI score >5 indicates poor sleep.

Alexander et al Impact of cataract surgery on sleep in patients receiving either ultraviolet-blue blocking or blue-filtering intraocular lens implants Invest. Ophthalmol Vis Sci 2014 25:55(8) 4999 - 5004

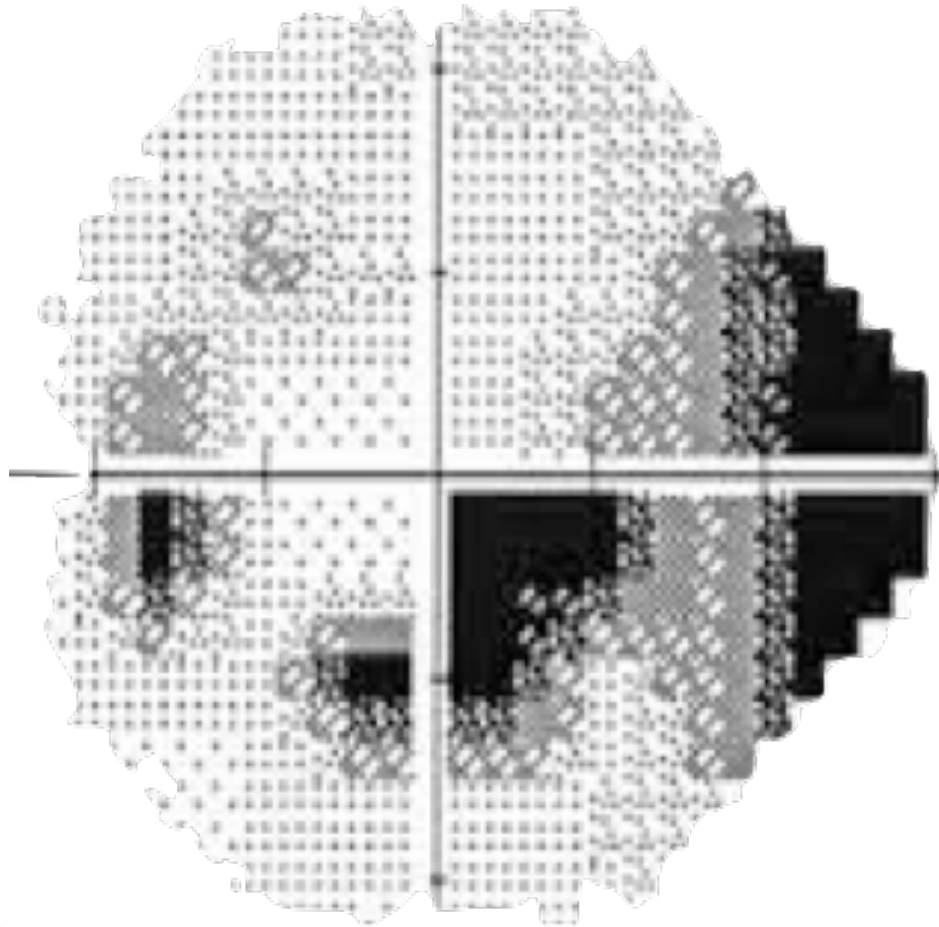




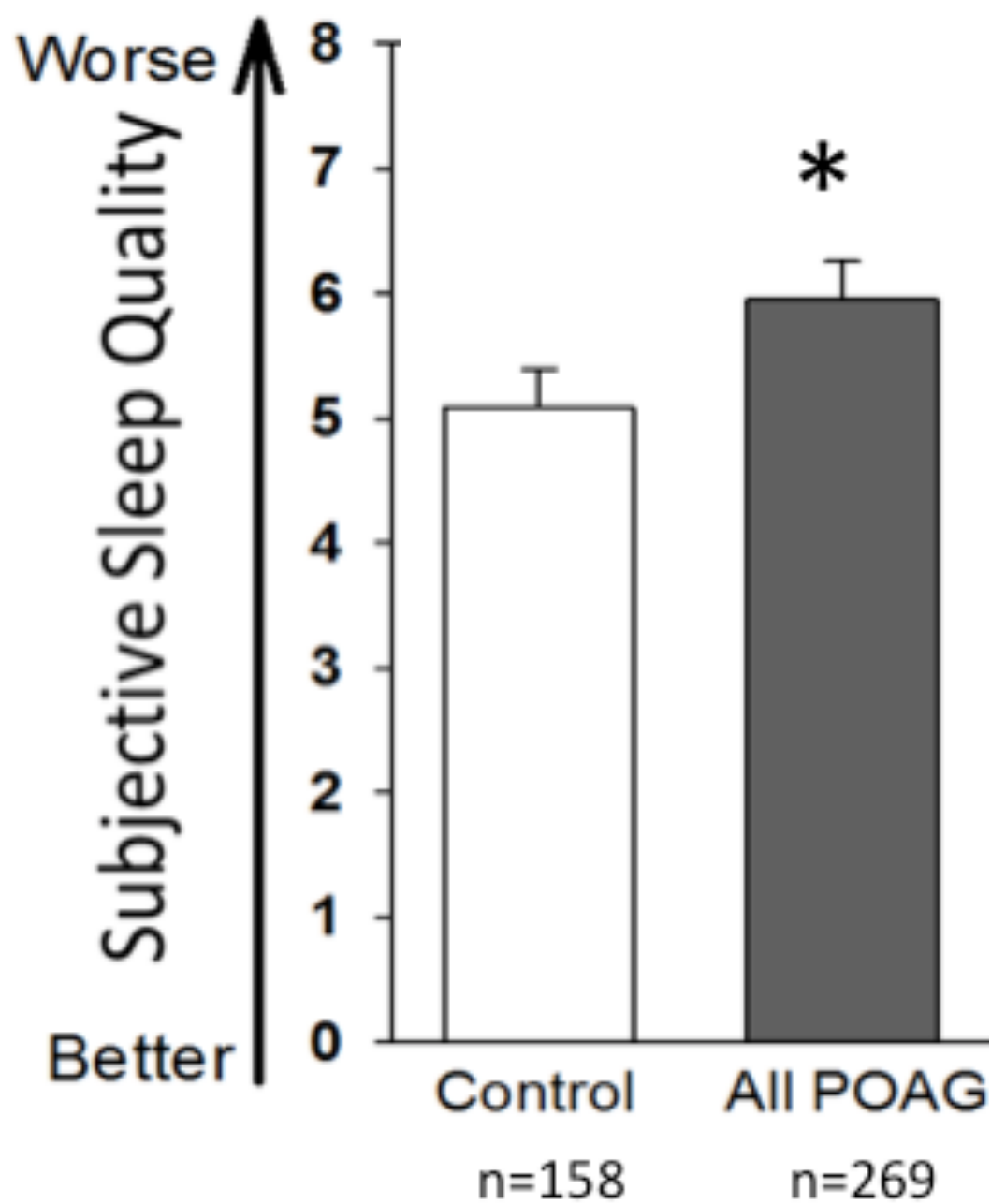
- PSQI score improved post-operatively irrespective of the type of lens
- Global improvement is not sustained over 12 months
- There was also an improvement in the component score sleep latency and this improvement is sustained at 12 months

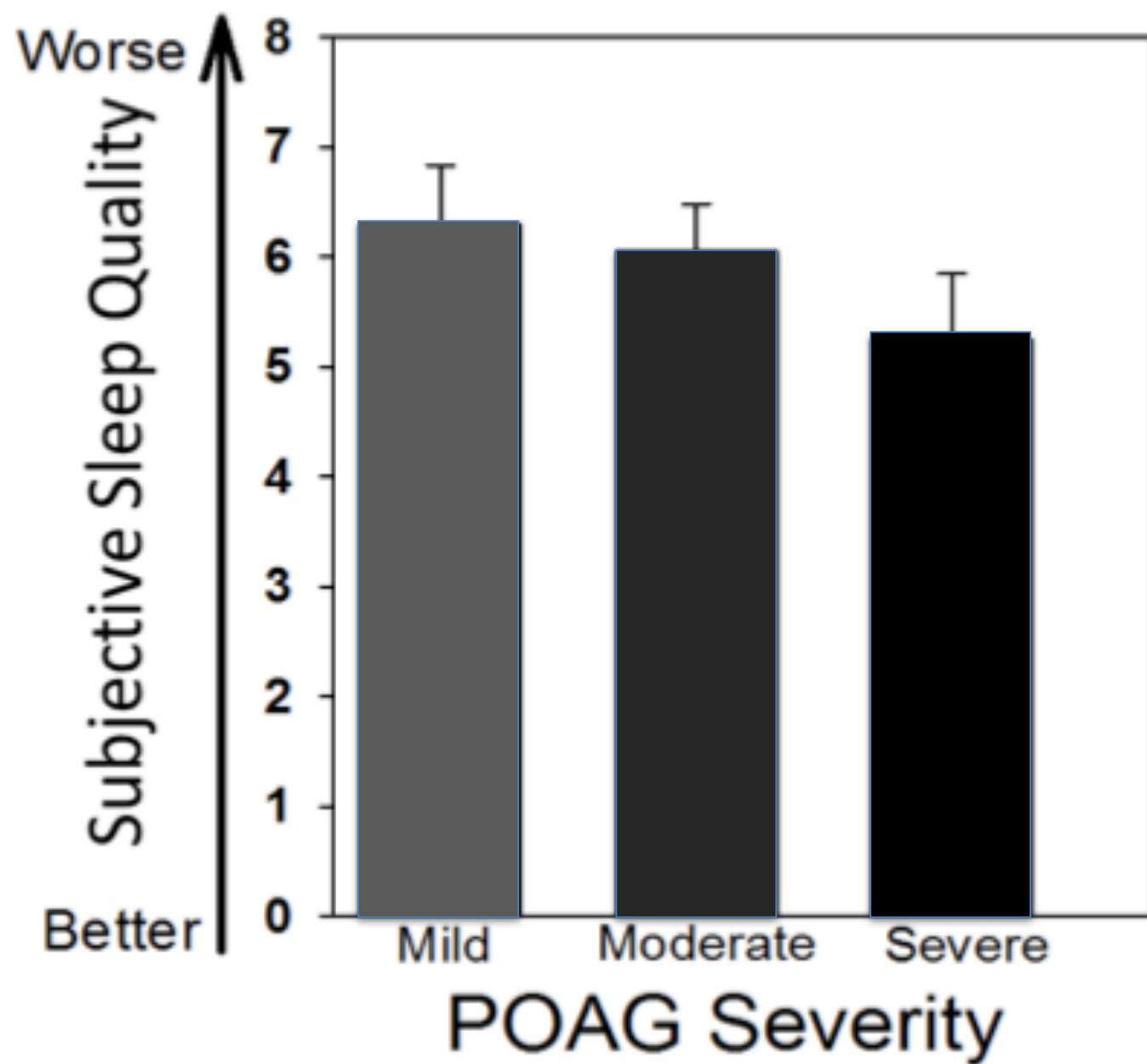
**Eye Disease - and
Severity?**

Primary Open Angle Glaucoma (POAG)



Optic nerve

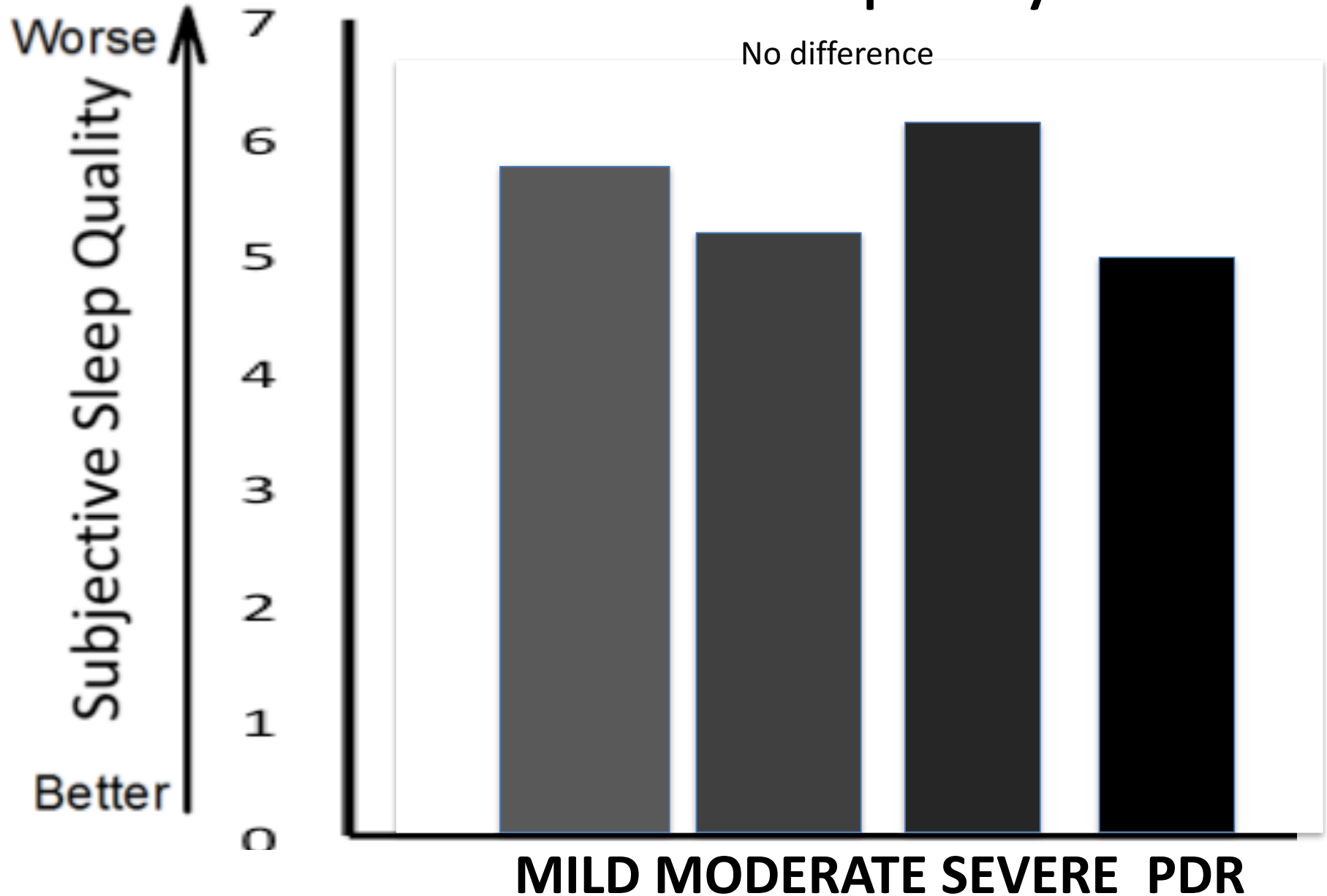




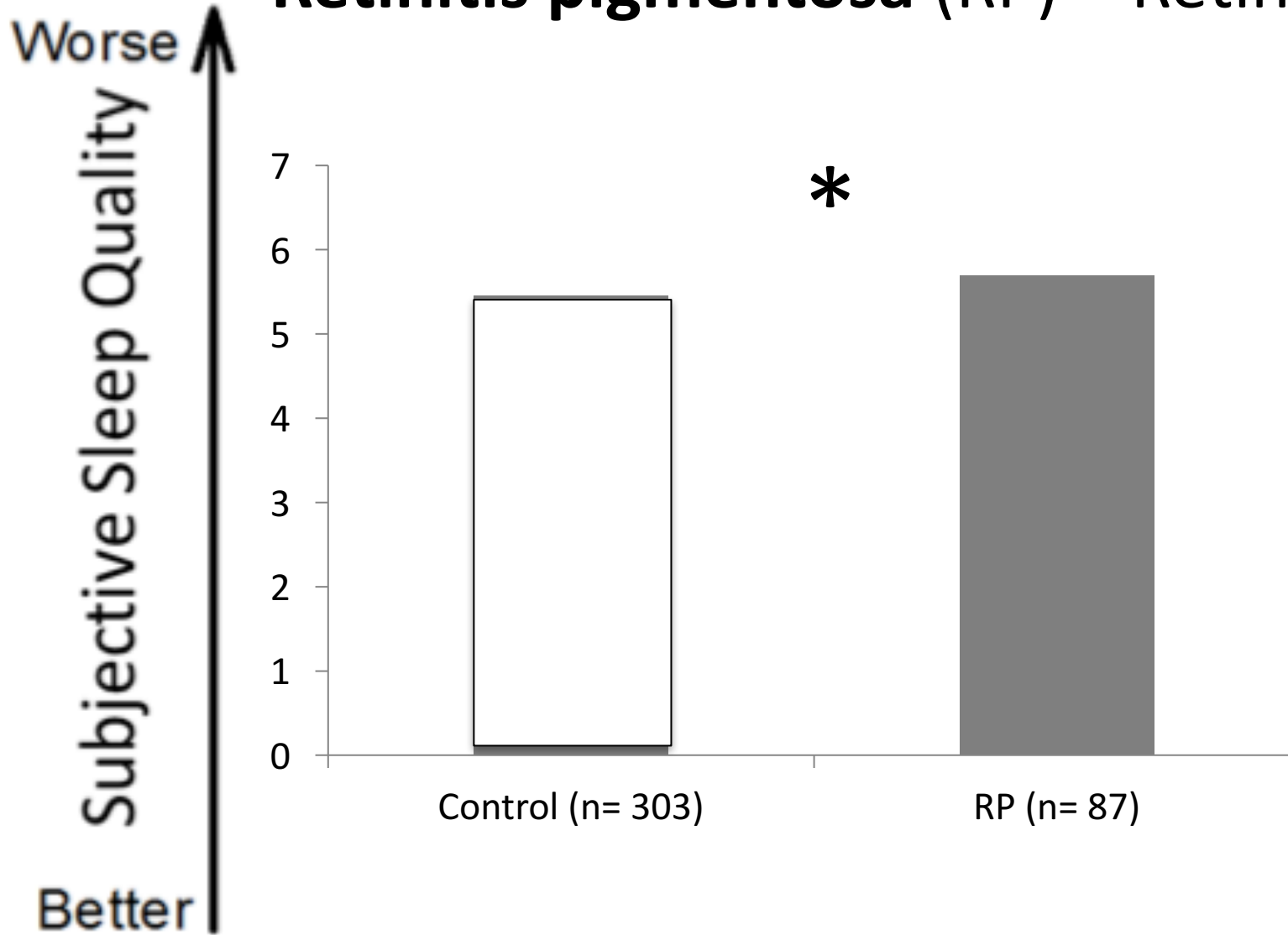
Diabetic Retinopathy - retina

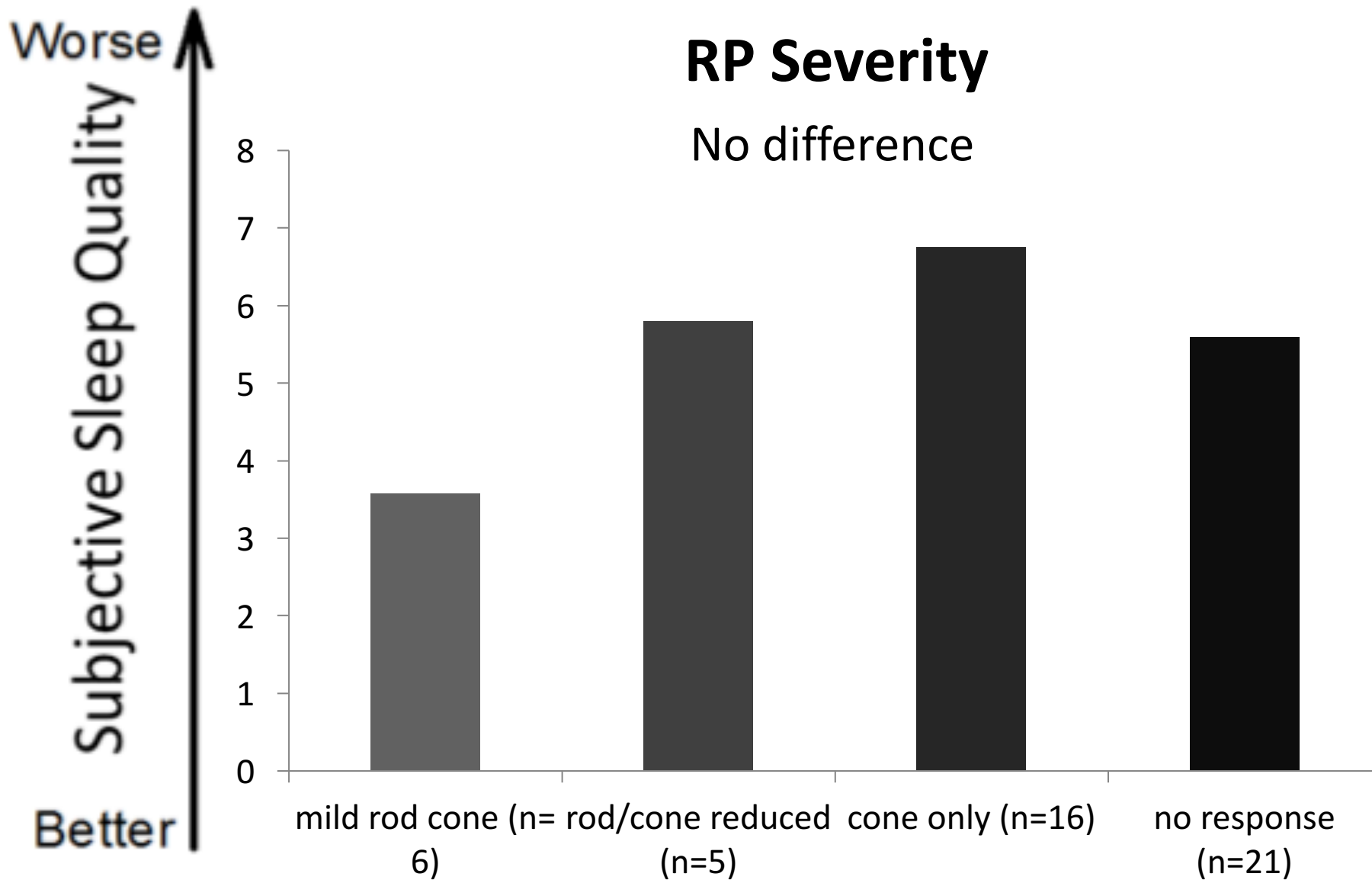


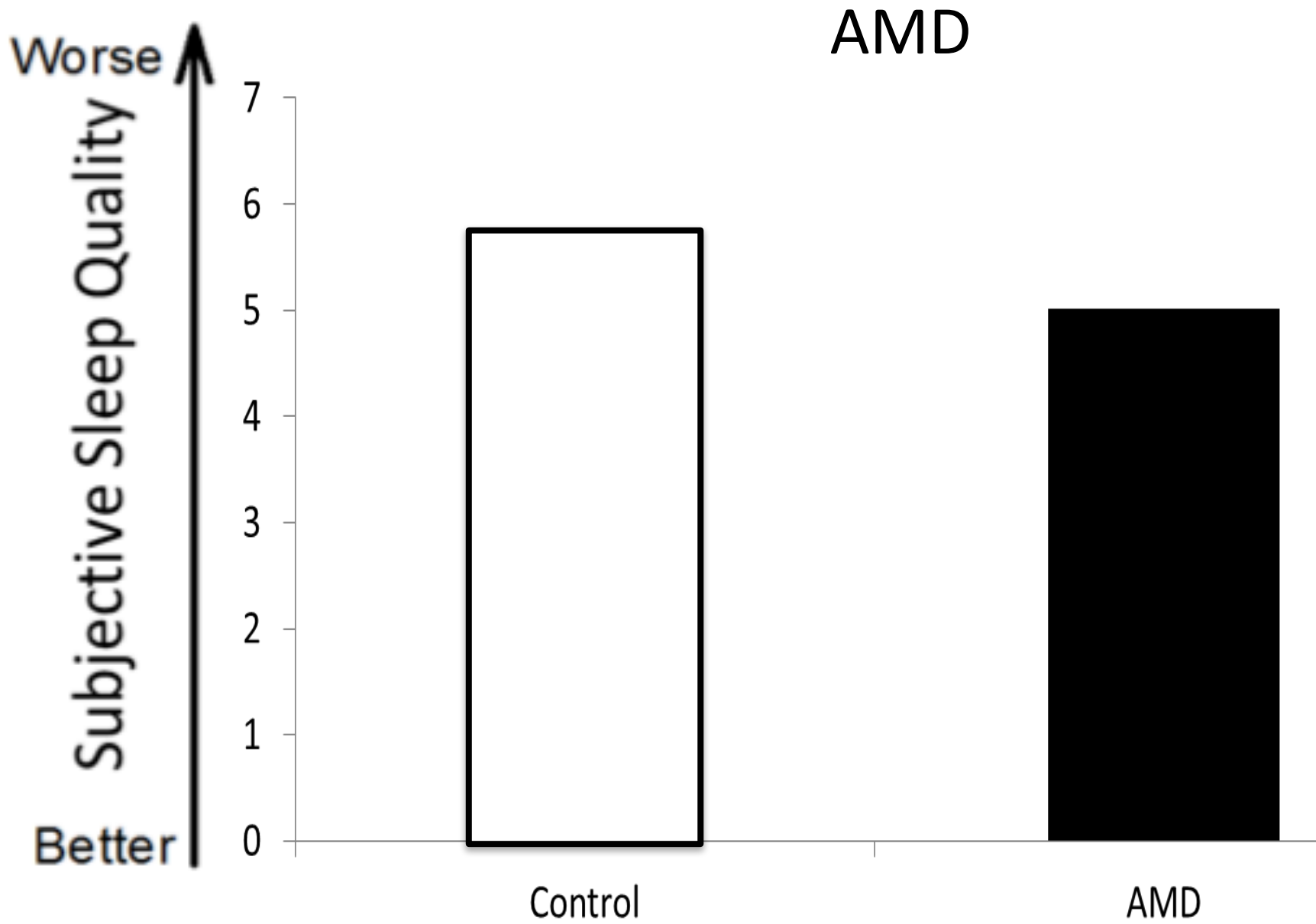
Diabetic Retinopathy



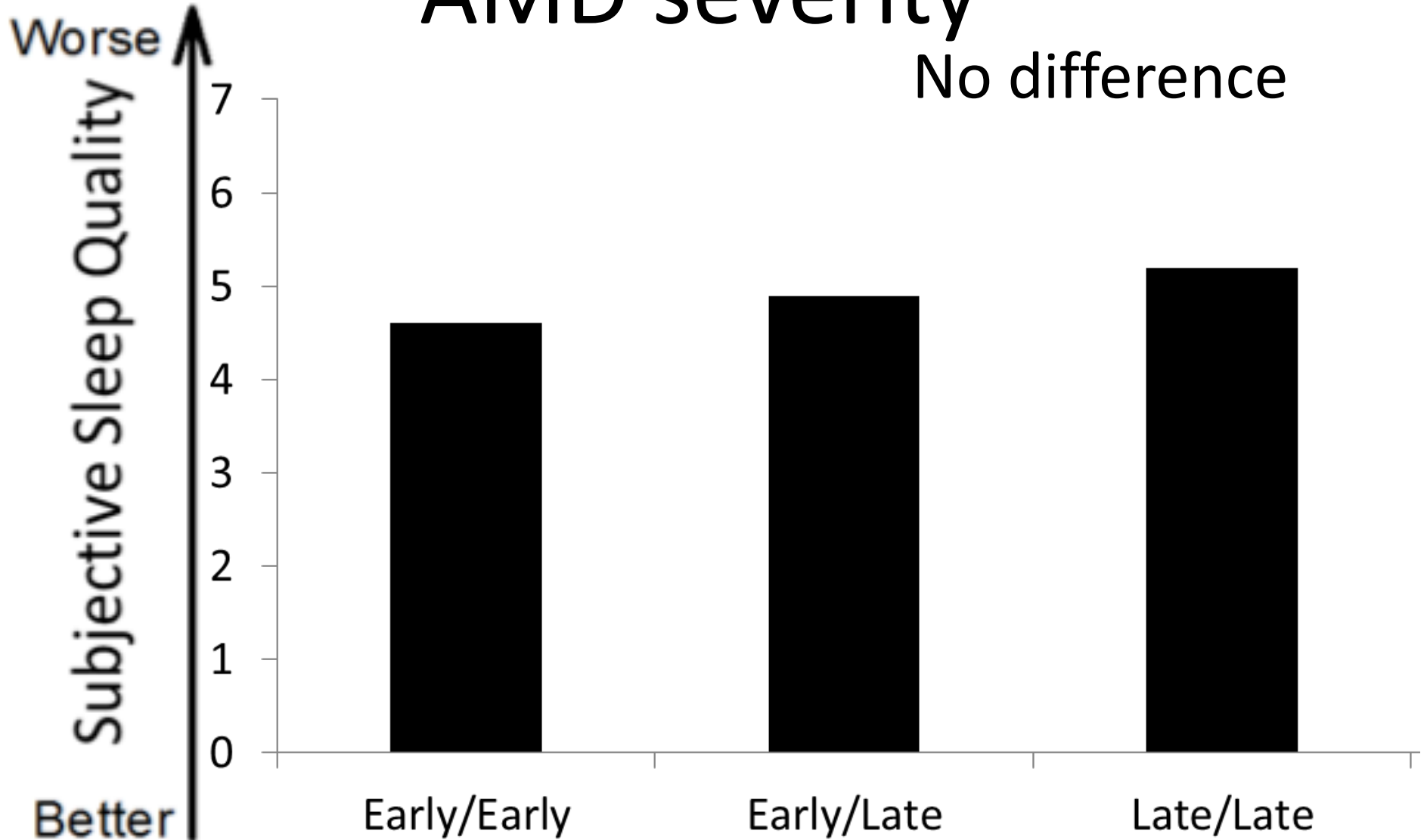
Retinitis pigmentosa (RP) – Retinal Cells







AMD severity



Eyes

- Cataract surgery leads to improved sleep which is not sustained over 12 months irrespective of lens type
- Glaucoma and RP are associated with worse sleep quality
- Diabetes and AMD doesn't seem to have an impact on sleep quality
- However – most our participants had a good visual acuity.

Further work

Melatonin Trial

Other eye conditions

Clinics – Eye Disease & Sleep
quality

ANIRIDIA?

- We don't know.

BUT.....

- Eye – Sleep?
- PAX6 – Pineal...Melatonin?

PAX 6 findings

- *Pineal Volume smaller in 37 individuals with PAX6+/- (including 9 individuals with aniridia) than 17 control participants
- * Morning Melatonin Lower in PAX6+/-

Greater parental report of sleep disturbances in children with *PAX6*^{+/-}
But probably only 2 or 3 with Aniridia

	PAX6 (n=16)	Controls (n=6)	P value
Sleep score	48 ± 6	41 ± 5	0.03

Table 1. Characteristics of Adolescents With *PAX6*+/- and Healthy Comparison Group.

Table 1. Characteristics of Adolescents With *PAX6*+/- and Healthy Comparison Group.

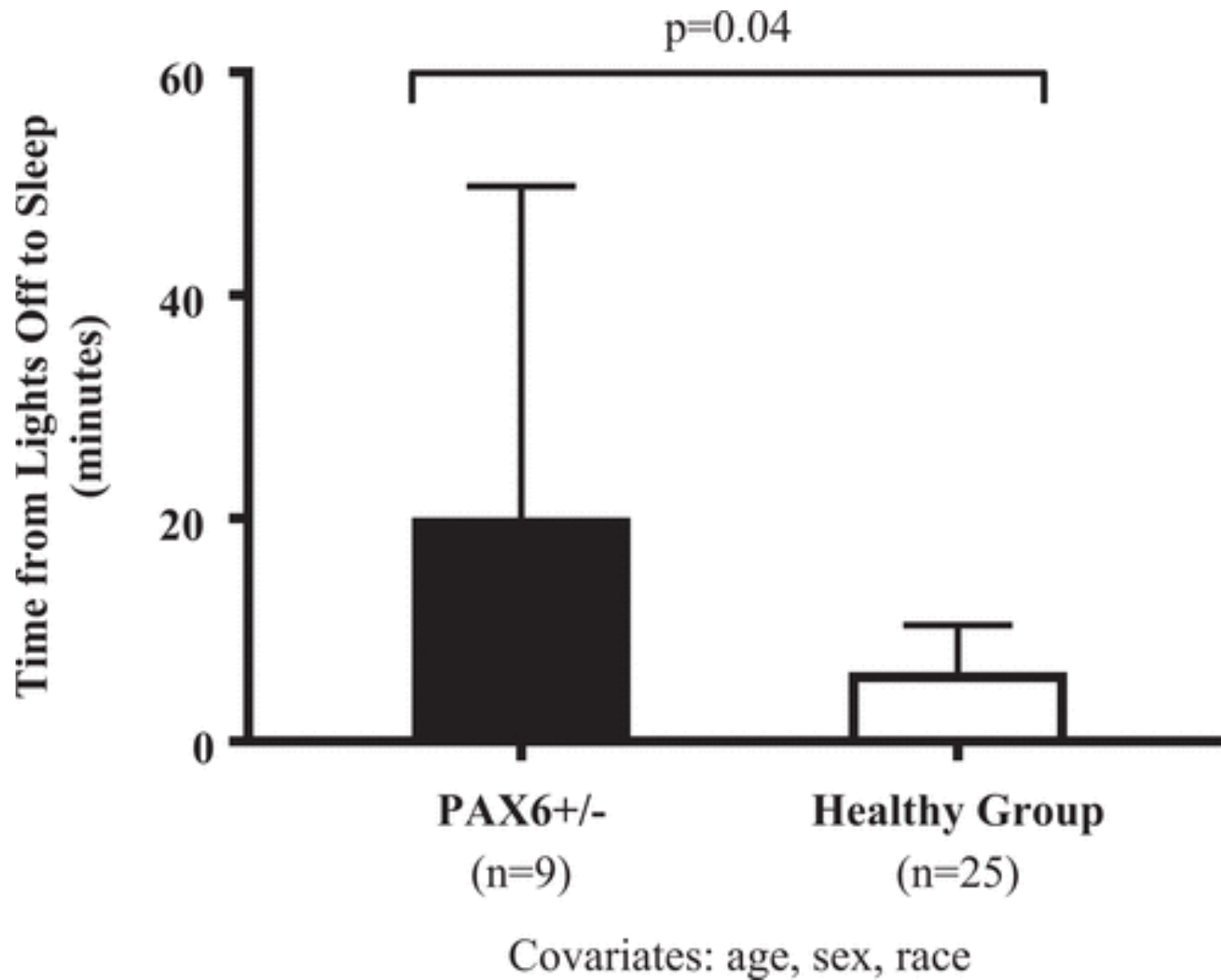
Characteristic	<i>PAX6</i> +/- (<i>n</i> = 9)	Healthy Comparison Group (<i>n</i> = 25) ^a	<i>p</i> Value
Age (years), mean \pm SD (range)	15.5 \pm 3.3 (10–19)	13.6 \pm 2.3 (10–18)	.07
Sex (% female)	66.7	44.0	.44
Race/ethnicity (%)			1.00
Non-Hispanic Caucasian	88.9	80.0	
Other	11.1	20.0	
CASQ total score, mean \pm SD	34.6 \pm 10.9	31.3 \pm 8.7	.39
PROMIS sleep disturbance, mean \pm SD	45.5 \pm 7.0	44.3 \pm 7.2	.67
PROMIS sleep-related impairment, mean \pm SD	45.7 \pm 10.2	50.2 \pm 7.1	.15
Bedtime, median (25th to 75th percentile)			
Overall	22:19 (21:34–23:41)	22:46 (22:25–23:48)	.11
Weekday	22:08 (21:23–23:33)	22:18 (21:56–23:32)	.25
Weekend	22:40 (21:38–01:33)	23:26 (23:04–00:14)	.14
Wake time, median (25th to 75th percentile)			
Overall	7:09 (6:43–7:30)	7:19 (7:00–8:00)	.11
* Weekday	6:36 (6:19–7:10)	7:05 (6:43–7:51)	.08
Weekend	7:24 (6:44–10:01)	8:16 (7:51–8:43)	.25
* Overall sleep duration, median (25th to 75th percentile)	7:42 (6:30–8:51)	7:55 (7:19–8:15)	.86
Sleep-onset latency (min), median (25th to 75th percentile)	1.8 (1.2–3.9)	2.4 (1.3–3.4)	.77
Lights off to sleep (min), median (25th to 75th percentile)	17.0 (6.1–74.4)	7.9 (2.5–13.5)	.07
Sleep efficiency (%), median (25th to 75th percentile)	91.2 (89.8–92.2)	91.1 (89.8–92.5)	.74

Note. Normally distributed variables are shown as mean \pm SD and were analyzed using independent samples *t* test. Nonparametric data are shown as median (25th to 75th percentile) and were analyzed using Mann–Whitney *U* test. Percentages were compared using Fisher’s exact test. CASQ = Cleveland Adolescent Sleepiness Questionnaire; PROMIS = Patient Reported Outcomes Measurement Information System. ^aPreviously reported (Hanish, Lin-Dyken, & Han, 2017).

Published in: Alyson E. Hanish; Joan C. Han; *Biological Research For Nursing* 20, 237-243.

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- Professor Susan Downes
- Russell Foster
- Sophie Marlowe
- Rob Purbrick
- Colm Andrews
- Sarah Stevenson
- Rupal Morjaria
- Rukhsana Safa
- Katharina Wulff
- John Salmon
- Alexina Fantato

All participants were recruited between 2012 and 2017 as part of the SOMNUS (“Effect of Ocular Disease on Sleep and Circadian Rhythms”) Portfolio Study.



Aniridia Network

Sleep and eye disease

Dr Iona Alexander

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Aniridia Network is registered charity 1176792 in England & Wales, operating as
Aniridia Network UK, 22 Cornish House Adelaide Lane Sheffield S3 8BJ



Aniridia Network

Psychological impact of aniridia

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Sofia's Diagnosis

Diagnosis was delayed

Concerns over photophobia were dismissed

Information on Aniridia was non-existent

Sofia's Diagnosis

A Rare Diagnosis with **no** support caused:

- Fear
- Stress
- Sadness
- Confusion

Rare Diseases

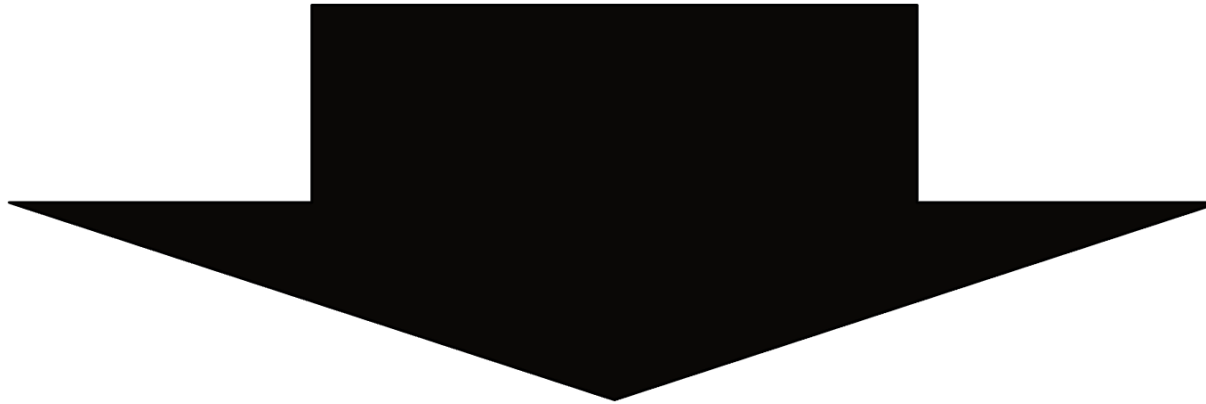
ARE:

Complex

Life-changing

Chronic

Challenging



FOR THE WHOLE FAMILY!

Rare Diseases

Care for a child with a rare disease involves a lifetime commitment and many times requires...

Change in
work pattern

Job loss

Income
reduction

Extra
responsibilities

Specialist
health literacy
& skills

Rare Diseases

The mental strain becomes too much for parents and results in:

CHANGES IN BEHAVIOUR

CHANGES IN MOOD

DEPRESSION

Aniridia

- Requires a multi-disciplinary team
- Such teams are not in place!
- Secondary conditions are even less documented than Aniridia
- Getting information and support require massive investment
- Devastating news and strain of discovery **DISTRESSES** family dynamics

Aniridia

**EMOTIONAL ROLLERCOASTER FOR
PARENTS IS OVERLOOKED**

Aniridia

The impact of an Aniridia diagnosis on the parents' mental health is neglected

Aniridia

With the focus, naturally, being on the children/new research and better treatments,

The parents' emotional well being is rarely discussed.

Aniridia

When parents' mental health is compromised:

- They will **not** be able to offer the best care possible to their Aniridic child
- They will **struggle** with coping mechanisms

Aniridia

- Their feelings **will affect** the family as a whole
- Their ability to seek treatment will be directly **influenced**
- They will feel **alone, stressed and isolated**

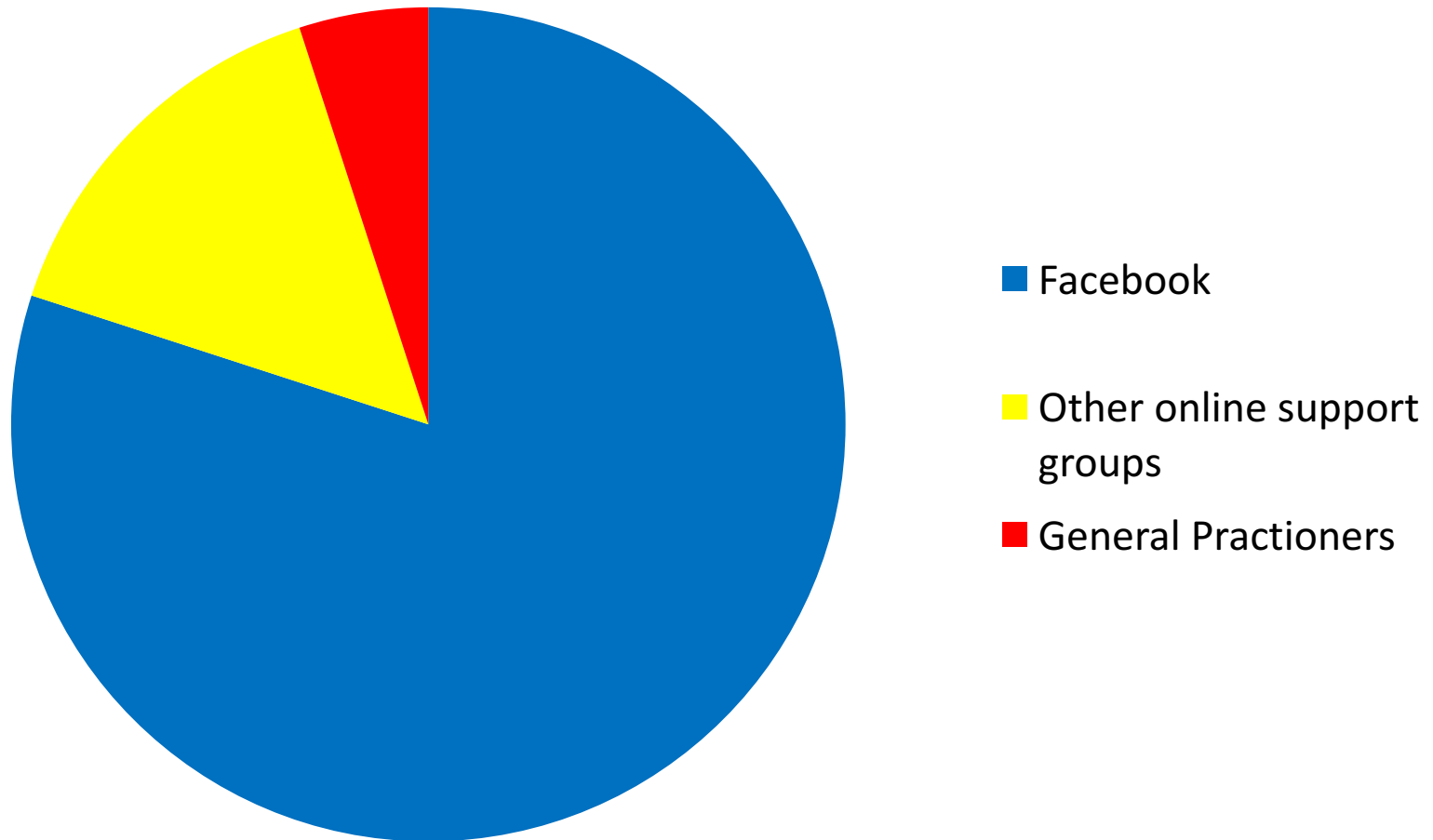
Aniridia

THE PSYCHOLOGICAL IMPACT OF ANIRIDIA ON PARENTS IS OFTEN **BURIED**

...with many of them reporting clinical
signs of depression!

Aniridia

Aniridia Resources



Moving Forward

Traditionally families received very little attention from clinicians/health authorities

In the last few years this is changing!

Moving Forward

- Aniridia Network UK
- Eurordis
- Aniridia Europe
- Rare Diseases UK
- The Royal National Institute for Blind People

ARE MAKING HUGE PROGRESS

Moving Forward

Families need:

- Guidelines in place between health professionals
- Early interventions to avoid diagnosis delays
- Psychological support and genetic counselling when receiving a diagnosis

Moving Forward

Families need:

- Updated information on Aniridia specialists in their country
- Better education for teachers and other professionals
- Direct communication and referral to Aniridia organisations

THANK YOU



Aniridia Network

Psychological impact of aniridia

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Thank you

- Resource For London
- Tinies – Iris Childcare!
- Stallholders
- Trustees

Special thank you

- Volunteers
- Speakers
- Conference Organiser Dave McKay

Say what you think

- Post comments and photos on social media: #ANUKconf
- Feedback by email
- Complete the survey that will be sent by email
- Write a blog post

Goalball & Social Evening

Coach leaves at
5:40pm

Guides

Ask for and offer
help getting to the
station

**Hope to see you at a future
event!!**

Organised by you?