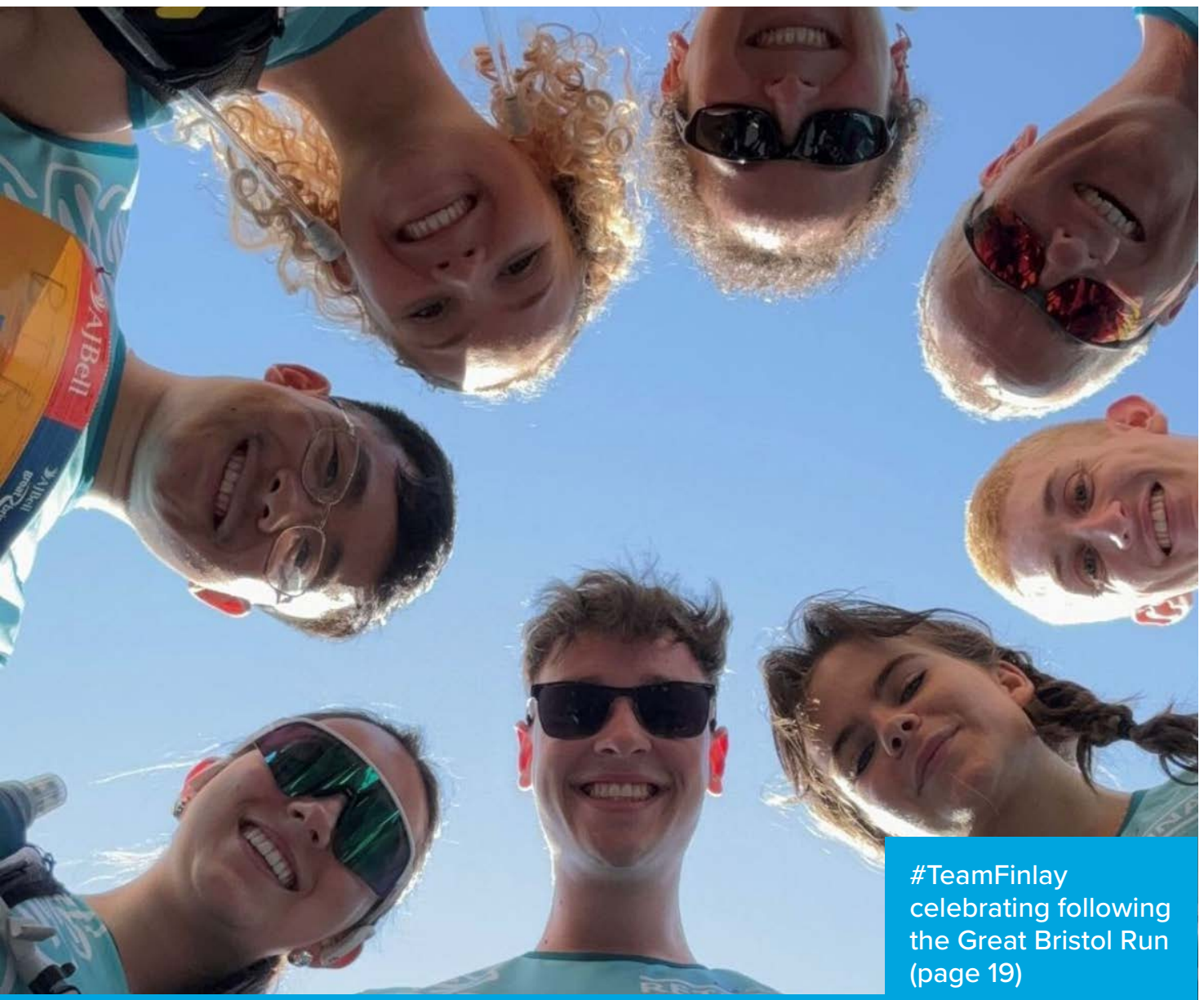




LOOK FORWARD

ISSUE 183 SUMMER 2024



#TeamFinlay
celebrating following
the Great Bristol Run
(page 19)

This edition includes exciting news about our Annual and Professionals' Conferences (pages 3 & 5). We hope as many of you as possible can join us in Manchester, or online, in September. You'll also find updates about the latest research (pages 12-13) and a feature on one of our funded researchers, Dr Jörn Lakowski (pages 20-21).



Our Annual Conference is heading north

We're halfway through the year and as I write this, summer appears to have arrived! It may not last but I'm enjoying it while I can.

This edition is packed with news about our upcoming Conferences in September (pages 3 & 5). For those of you receiving Look Forward in hard copy, you'll find a postcard inside to pop on your fridge to remind you to register to attend. I hope to see as many of you as possible in Manchester.

If you're not already playing the Retina UK Lottery, I'd encourage you to sign up. One line will cost you just £1 per week and you could win up to £25,000! Find out more on page 7.

This edition will arrive after the General Election and whatever the outcome, I hope that you were given the

opportunity to vote independently. We continue to work collaboratively as part of the Vision Partnership to address the challenges being faced by blind and partially sighted people. The Vision Partnership is made up of Retina UK, Blind Veterans UK, Glaucoma UK, Guide Dogs, Macular Society, RNIB, Thomas Pocklington Trust and Visionary.

If you have any questions or suggestions about our work, please email me on **chiefexec@RetinaUK.org.uk** or call our office on 01280 821334.



*Tina Garvey,
Chief Executive*

Look Forward is available as a hard copy, by email, audio (CD or memory stick) and in Braille. Get in touch if you'd like to receive a copy in one (or more) of these formats. If you no longer wish to receive Look Forward, please let us know by emailing **info@RetinaUK.org.uk**.



This newsletter has been funded through sponsorship by Janssen. Janssen has not been involved in the production, review or distribution of this material.

Retina UK is a charity searching for treatments and giving support every day, so everyone with inherited sight loss can live their best lives.

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Our ever-popular Annual Conference heads north

Register now

When: Saturday 28 September

Where: The Pendulum Hotel,
Manchester (M1 3BB)

Register: [RetinaUK.org.uk/
annual-conference](https://RetinaUK.org.uk/annual-conference)

You asked us to move our Annual Conference north and we've done just that! Join us in central Manchester on Saturday, 28 September for what promises to be an uplifting and informative day, and the great news is it's free to attend.

While we'd love you and your friends and family to join us in person, you can also register to watch the sessions live online.

The programme for the day includes:

- ***The Eye Care Support Pathway (ECSP) and what it means for you***
How this new guide and Retina UK can help you to access information, support and advice to help you live your best life.
- ***Harnessing the power of peer-to-peer support***
Listen to our community explain how Retina UK support services have improved their health and wellbeing.

- ***Supporting the next generation of research leaders: a discussion with Retina UK-funded PhD students***
Our three new PhD student grantees, funded by your donations, share their findings.
- ***Progress at the forefront of inherited sight loss research***
Join us as Professor Graeme Black takes a deep dive into the latest research into inherited sight loss.
- ***Research Q&A***
Your chance to put your burning questions to our panel of experts. Panel participants include Professor Graeme Black and Dr Panos Sergouniotis.

Come along and spend the day with others living with inherited sight loss, chat to our friendly staff and volunteers and meet exhibitors from across the sight loss sector.

Our conference is always popular and in-person places are limited so register today.

The venue is the Pendulum Hotel (M1 3BB) located just a short walk from Manchester's railway stations.

To register, please visit our website: **RetinaUK.org.uk/annual-conference** or call our friendly team on 01280 821334.

What's On

As we head into the second half of the year, we are excited to share our upcoming events and campaigns:



SEPT
2024

Christmas card launch

Support the work of Retina UK this Christmas by purchasing our brand new range of Christmas cards! Visit our online shop in September to support us this winter **RetinaUK.org.uk/shop**.

8
SEPT 24

Great North Run 2024

Six runners from #TeamRetinaUK will be taking to the streets of Newcastle! Could you be one of our runners in 2025? Call James on 07736 925174 to register your interest or email him at **james.clarke@RetinaUK.org.uk**.

18
SEPT 24

Annual General Meeting (AGM) – page 6

24
SEPT 24

Sight Village North-East (Leeds)

27
SEPT 24

Professionals' Conference (Manchester) – page 5

28
SEPT 24

Annual Conference (Manchester) – page 3

OCT
2024

Make a Will Month – pages 14-15

Leaving a gift in your Will means Retina UK can continue to support those with inherited sight loss, both now and in the future. Find out more at **RetinaUK.org.uk/legacy**.

10
OCT 24

Retina UK Day 2024

– page 11

Join us for the inaugural Retina UK Day and come together to help us raise awareness and funds on World Sight Day.

5
NOV 24

Sight Village South-East (Kensington, London)

29
NOV 24

Look Forward Winter 2024 edition

3
DEC 24

Big Give Christmas Challenge 2024

Double your impact with our annual Christmas campaign and have your donations to Retina UK doubled for one week only!

Sign up for our monthly e-Newsletter for news about upcoming events and more. Call us on 01280 821334 or email **info@RetinaUK.org.uk**.

Supporting you to support the inherited sight loss community

Register now for our Professionals' Conference

When: Friday 27 September

Where: The Pendulum Hotel,
Manchester (M1 3BB)

Register: RetinaUK.org.uk/prof-conf

If you are a professional supporting people with inherited sight loss then our CPD-accredited Professionals' Conference on Friday 27 September is for you. It's free to attend in central Manchester and online. Come along in person and network with your peers or attend online from your home or office.

The programme for the day includes:

- ***Working together to deliver the Eye Care Support Pathway (ECSP)***
How we can enable families to get the information, support and advice they need to live their best lives at all stages of their sight loss journey.
- ***The role of peer-to-peer support in achieving good outcomes for clients***
Our panel of lived experience volunteers and service users explain the positive impact peer to peer support has had on their lives.



- ***The importance of quality of life research: understanding the impact of inherited sight loss***
Exploring the findings of a new in-depth research study.
- ***Supporting you deliver equitable, accessible and inclusive services***
Practical advice from our expert speakers from SeeAbility, RNIB and BAME vision.

In-person numbers are limited for this popular annual event so early registration is advised.

To register, please visit our website: **RetinaUK.org.uk/prof-conf** or call our friendly team on 01280 821334.

Annual General Meeting notice 2024

For the first time, our AGM will be held fully online, to allow as many of our members as possible to take part. It will take place on Wednesday 18 September at 7.15pm.

Online, postal and telephone voting will be held in advance of the AGM, meaning all members who choose to do so can exercise their right to vote at a time, and in a format, that is accessible to them. **All members with voting rights will receive information on how to vote with this edition of Look Forward.**

We will present the results of the vote at our online interactive AGM meeting. All are welcome to attend the meeting where there will also be the opportunity to hear from our Chairman and Treasurer and to ask them, the Board and our Chief Executive questions.

Agenda

1. Welcome
2. Apologies
3. Approval of the minutes of the previous AGM (online vote)
4. Presentation of the annual report and accounts for the year ended 31 December 2023
5. Adoption of the Annual Report and Accounts for 2023 (online vote)
6. The appointment of the auditors (online vote)
7. Presentation of Retina UK activity in 2023 and 2024 overview
8. Amendment to the Charity's constitution to allow for the appointment of a President (online vote)
9. The appointment of trustees (online vote)
10. Any other business

New 'Tech Selector' tool

Tech Selector is a web-based tool, hosted on the Guide Dogs website on behalf of the Vision Partnership group of charities. It allows people to list technology products, apps and software that could be useful to them. They can select a number of different category filters to tailor the search results to best fit their needs and circumstances.

Once potential technology solutions have been identified, the site signposts to further information from a range of national and local sight loss organisations.

For more information about Tech Selector and its impact on the lives of blind and partially sighted people, please visit guidedogs.org.uk/tech-selector.

Have you joined our weekly Lottery?

Don't delay, play today!

Win up to £25,000 and feel great about supporting people with inherited sight loss.

One ticket a week can support parents and young people navigating the challenges faced after diagnosis.

Three tickets a week can help support our crucial Helpline, supporting people with practical and emotional help.

Five tickets a week can help fund groundbreaking research, giving hope to people living with inherited sight loss.

"Thank you so much. You don't realise how much it helped me to talk to someone who knows what I'm going through. I don't feel alone anymore. You have given me a lot to think about in terms of what I can do in the future".

Talk & Support service user

Responsible gambling

We're all about fun, but also about ensuring a safe and responsible gaming environment. Know the signs, play smart, and let's make every moment thrilling and enjoyable.

To help you enjoy the fun, we've put together some information about gambling responsibly on our website: RetinaUK.org.uk/lottery.

How to play



Embrace your lucky streak: Choose five numbers or entrust destiny to make the call with a Lucky Dip. Each line costs £1.



Increasing your lines boosts your chances to win, making a bigger difference.



Seamless payments through Direct Debit - a quick and secure setup ensures swift transactions, and if luck favours you, we'll share the news and celebrate your success!

Over 50% of each £1 comes directly to Retina UK.

Sign up now, set the ball rolling and be a catalyst for change!

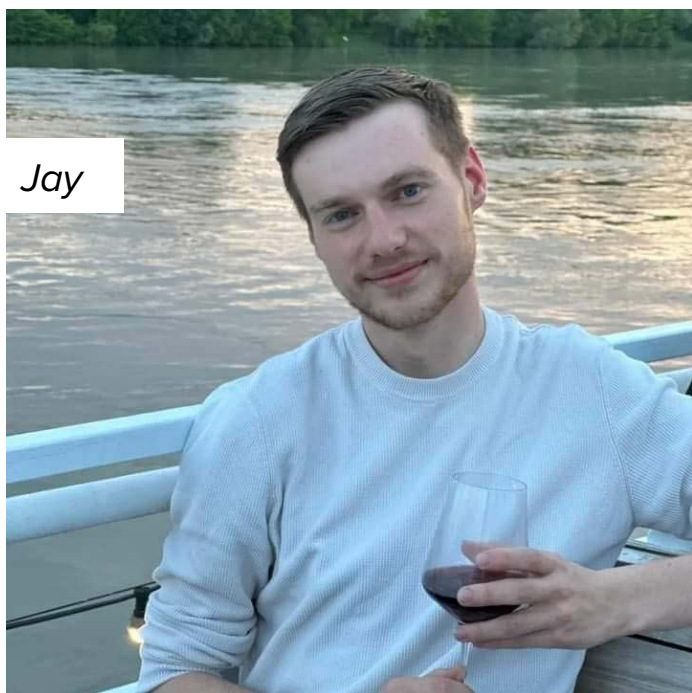
Just visit RetinaUK.org.uk/lottery or scan the QR Code and join the Retina UK Weekly Lottery.



GambleAware

Connecting young people

James (24), Sam (34) and Jay (27) connected with Retina UK this year. They are all living with retinitis pigmentosa (RP) and had never spoken to anyone with RP outside of their own families until now. They describe the positive impact of those conversations



Jay's eye problems first became apparent when he started primary school, but it wasn't until he started secondary school that he realised he was different to his classmates. "I was always a bit confused if we were in a drama studio and they turned the lights down and asked us to move around the room. Wait, how can people see? Where are they going, it's really dark!" Jay was diagnosed at 12 and following tests, his mum was found to be living with the same condition but with much milder symptoms.

Sam didn't notice any issues with his vision until the age of 15. He has other conditions which affect his health and at the time, it was thought that these were connected. He received a diagnosis of RP from his ophthalmologist but his genetic test wasn't able to determine the causative gene.

James was diagnosed in Paris and was confirmed to have the RPGR gene. He has two younger brothers living with the condition but the three of them have a considerable variation in their symptoms.

All three were introduced to Retina UK Information and Support Coordinator, Mark Baxter as a result of a phone call to our Helpline. Mark also lives with RP.

Jay said: "I got in touch with Mark, we got talking on the phone, and he invited me to a local peer support group day out to a museum in London. I got there and there were two or three people who were around the same age as me. It just blew my mind. Not just people my age, but men as well, who are living within a few miles of me! It was an amazing day out. I had the first



James

“Talking to Mark was such an amazing thing because it was my first time talking to somebody that really gets it! I know everyone can say they understand, but nobody understands it like someone who has it, and for me it was major. Having conversations with somebody who has the same eye disease and can really relate, that was a brand new experience for me!”

James

conversation in my life with someone who actually understands what’s going on inside my head. It was so surreal.”

Sam recently attended our Merseyside local peer support group. He said “speaking to other people who understand what it’s like to live with sight loss, sharing tips and information and being able to relate to them and vice versa, that was so comforting.”

If you are living with an inherited sight loss condition or a friend or family member of someone who is, connecting with others can be really empowering, whatever your age.

Information about all of our local peer support groups can be found on our website: **RetinaUK.org.uk/groups**.

We are committed to improving the support we offer for young adults and we are looking for people under the age of 30 to help us shape this work in the coming months.

If you would be interested in getting involved, please join our Lived Experience Panel by completing the form on our website: **RetinaUK.org.uk/get-involved/lived-experience/**.



Sam

Take on a challenge

Taking on a challenge for Retina UK is more than just a fundraising activity. It's a feeling of giving back, leading to a sense of achievement and crucially, it'll become something that you can treasure forever as a life-long memory.

Whether you want to take to the streets of a city near you for a running race, or you're more of a daredevil and throwing yourself out of a plane is more your thing, whatever the challenge – we'll be here to support you every step of the way. With lots of group hikes nationwide, perfect for friends, families or work colleagues, as well as a whole host of other brand-new events available on our website, there's something for everyone. You can also organise your own challenge for us! Could you walk 100,000 steps in a month or organise a fitness exercise class? The opportunities are endless and every penny that you raise will make a real difference.

Supporting Retina UK in this way allows us to continue supporting our community. Hazel Denby, who recently completed a skydive for us and raised



an incredible £1,000, loved taking on a challenge for Retina UK: *"The free-fall is a massive adrenaline rush and it's more like flying than falling, but it's exciting and intense! After the parachute goes up, there's a sense of calm and weightlessness, along with breathtaking views. I feel a real sense of achievement that I did it and raised some money for a charity close to my heart."*

We'd love to hear from you about your fundraising ideas, so please contact James on james.clarke@RetinaUK.org.uk or 07736 925174.

Visit our website to sign up for one of our upcoming challenges: RetinaUK.org.uk/challenge



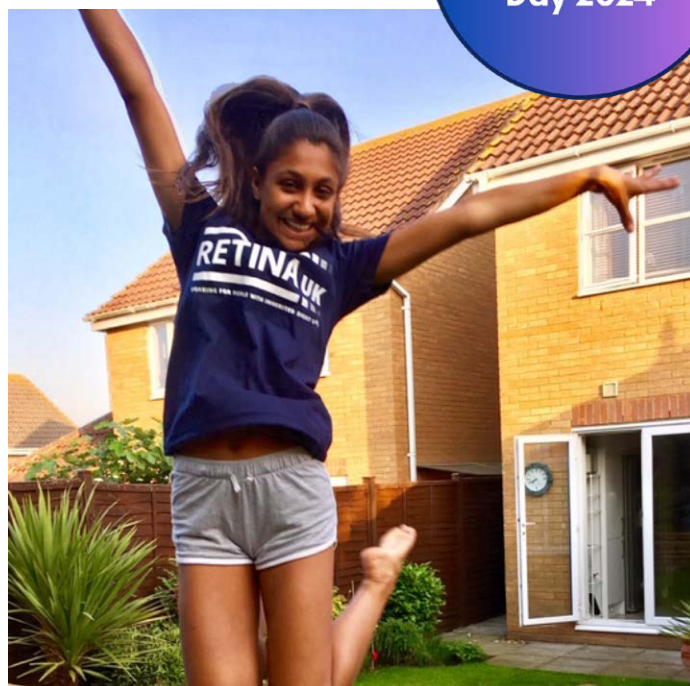
Sign up now for Retina UK Day 2024



On Thursday, 10 October, join us for the inaugural Retina UK Day where we're asking our community to come together on World Sight Day 2024 to support our cause and raise vital awareness and funds for Retina UK.

Do you (or someone you know) work or study in a school, college, university or business? Maybe you attend a local community group, church or club? Could you organise a bucket collection, quiz, event or challenge to take place on 10 October? Whatever the idea, we'll be here to support you as we come together, across the country, to raise awareness of inherited sight loss conditions on #RetinaUKDay 2024!

Save the date and we look forward to hearing your ideas on how you plan to mark this special day!



Find out more and register your interest in supporting us:

**RetinaUK.org.uk/
RetinaUK-Day**



Facebook fundraising

Are you celebrating a special birthday or occasion in the coming months?

Fundraising for Retina UK through Facebook is a great, easy way to support us, by asking your loved ones to make a donation as part of your celebrations. Your support allows us to fund our medical research projects and our support services, including our Helpline and

Local Peer Support Groups. From all of the Retina UK team, Happy Birthday in advance!

Set up your fundraiser today and find out more:



Email **james.clarke@RetinaUK.org.uk** or call him on 07736 925174.

Research round-up

Clinical trial for NAC launched

US-based Johns Hopkins university will begin a phase 3 clinical trial of NAC (N-acetylcysteine) for the treatment of retinitis pigmentosa (RP). The study will take place over 45 months and will enrol participants at sites throughout the US, Canada, Mexico and Europe.

NAC is an oral antioxidant. Antioxidants are molecules that help the body detoxify harmful by-products from metabolic processes. NAC is commonly used to treat conditions like cystic fibrosis and pulmonary disease.

In previous studies, NAC slowed retinal degeneration in RP and led to improvements in vision.

The study will establish long-term effects of NAC for people with RP including any side effects. The trial organisers do not recommend people with RP take NAC outside of the trial.

Moorfields Eye Hospital is hoping to become a UK centre for this trial.

A novel gene insertion therapy for Stargardt disease

US-based, biotechnology company, SalioGen Therapeutics are developing a treatment called SGT-1001 for Stargardt disease caused by mutations in the ABCA4 gene. Stargardt disease is an inherited retinal condition which primarily affects central vision and usually begins to impact the individual in childhood.

SGT-1001 is based on gene coding technology which uses a special enzyme and nano particles to insert the new genetic code into the recipient's DNA. These have a much larger carrying capacity than the virus traditionally used in gene therapy and so can accommodate very large genes like ABCA4. The company have said they plan to launch a clinical trial in the first half of 2025. This will involve a small number of participants and will probably only be at US centres.

Vitamin A supplementation has no benefit in RP

Research has determined that taking high dose vitamin A supplements does not slow vision loss in people with retinitis pigmentosa (RP). This new finding comes from re-analysis of data from a much older study. The trial originally found that taking vitamin A supplements slowed loss of vision. However, new analysis of the original data revealed that taking vitamin A had no overall benefit for people with RP. In addition, vitamin E supplements should be avoided due to its negative effect on retinal health.

It is advised that RP patients who have been taking vitamin A supplements for many years and feel that they are doing well, can continue under supervision of their doctor, but yearly liver function tests should be conducted. A healthy balanced diet is the recommended

approach, with no need to avoid any particular food.

CRISPR gene editing leads to improvements

A phase 1/2 clinical trial of CRISPR gene editing has generated positive results in participants with Leber congenital amaurosis (LCA), caused by mutations in the CEP290 gene. LCA is an inherited eye condition that appears at birth or in the first few months of life, affecting both peripheral and central vision.

The clinical trial involved 12 adults and two children. Participants received a single injection of the CRISPR gene editing therapy into the back of the eye.

CRISPR is a gene editing toolkit which acts as a guided scissor to cut a portion of the mutated genome to leave a functional gene. The trial marks the first-time gene editing has been shown to lead to improvements in the human eye.

Results showed that 11 out of the 14 participants had meaningful improvements in vision with no adverse effects. However, such findings establish proof of concept only. More testing over several years is required. It is unknown where and when the next phase of the trial will take place.

Nevertheless, these findings support the continuation of research on gene editing therapy for retinal disease.

Nanoscope announces topline results

Nanoscope Therapeutics has announced positive results from their two-year randomised controlled phase 2b gene therapy trial for late stage-RP. The treatment called MCO-010 is gene-agnostic, meaning that it could be appropriate for a large proportion of our community, no matter what their genetic diagnosis. Retina UK understands that trial participants' baseline was the ability to count fingers or less, meaning that the therapy is being aimed at advanced stages of sight loss.

MCO-010 led to improved visual acuity and visual function in participants after a one year follow up. After a 76 week follow up, visual acuity improved for patients in the high-dose group but there was no difference in visual acuity between the low-dose and control group at this point. There were no serious adverse effects.

Nanoscope have said they are possibly going to apply to the FDA later this year. However, it is extremely unusual for regulatory authorities to license a product after phase 2 trials only.

Meanwhile, other similar approaches are being investigated by companies such as Gensight, Bionic Sight and Kiora Pharmaceuticals, each with slightly different approaches and consequent pros and cons.

Support Retina UK and secure your legacy

Gifts left in Wills to Retina UK make a huge contribution to our research and support.



Many of our supporters choose to leave a legacy gift in their Will, ensuring we can continue to fund crucial research and support services for those affected by inherited sight loss. This October, we invite you to participate in “Make a Will Month.”

After you have made provision for your family and friends in your Will, we would be so grateful if you could consider leaving a gift, of any size, to Retina UK.

We recommend that you instruct a qualified solicitor to prepare your Will. You can find a comprehensive list of firms on the Law Society website (lawsociety.org.uk). You may also want to take a look at The Goodwill

Partnership who can visit you at home (if you live in England or Wales) to draw up a solicitor-provided Will (thegoodwillpartnership.co.uk).

Please note that Retina UK does not endorse any particular firm or firms. We recommend that you conduct your own research to identify the solicitor or provider who best meets your requirements.

If you already have a Will, you can sign a codicil document, which means a gift for Retina UK can be added as an amendment, without rewriting the whole Will. Your solicitor will be able to provide this document.

Your bequest

If you leave a legacy to a charity, these are entirely free of inheritance tax, and in some cases can even reduce the amount of tax burden on your other beneficiaries.

There are three types of bequests you could leave to a Retina UK:

- **Pecuniary** – this is a specific sum of money. Bear in mind that inflation could cause the value of a pecuniary bequest to lessen over time, so you might want to consider asking your solicitor to index link the gift, preserving its value.
- **Residuary** – this is a share of your remaining estate after all debts and expenses have been discharged and all pecuniary and specific bequests have been paid.
- **Specific** – you may have a particular item you wish to leave to the charity, perhaps some jewellery, artwork, furniture or even something larger such as a car or house.



“Whatever I leave the charity, it’s tax free. That really pleases me after paying taxes all my life. I know that all of the research projects supported by Retina UK have to go through a really thorough process, which includes the charity’s Medical Advisory Board. I know that my bequest will be used to make a difference for people living with inherited sight loss.”

Lynda Cantor, founder of Retina UK and Honorary President

If you would like further information on leaving a gift to Retina UK, or if you would like to tell us you have already named us as a beneficiary in your will, please call Jo Faulkner-Harvey, Head of Fundraising, on 01280 815900 or email jo.faulkner-harvey@RetinaUK.org.uk. Alternatively, you can visit our website RetinaUK.org.uk/legacy.

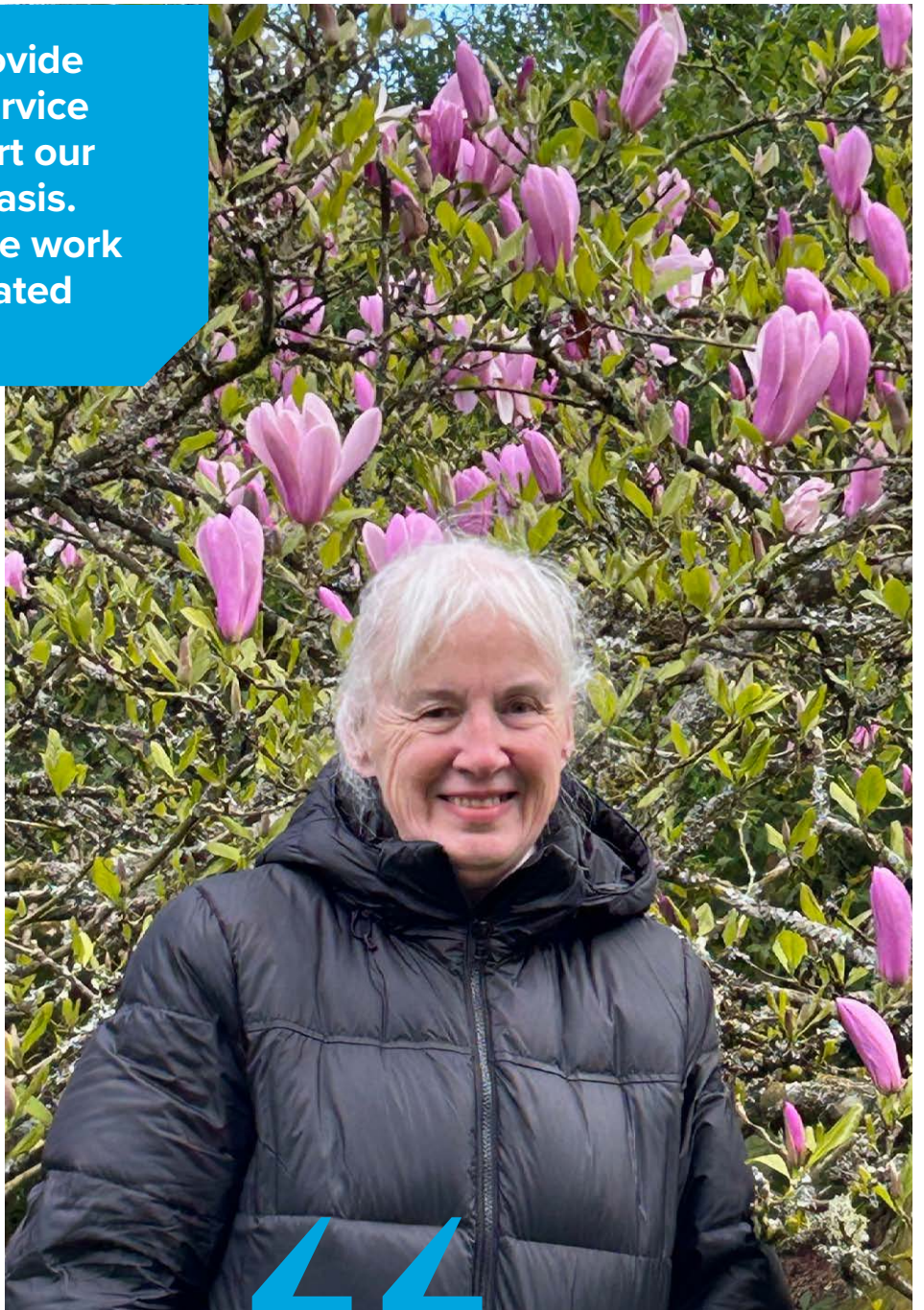
A job well done

Our office volunteers provide an incredibly valuable service to our charity and support our work on a more casual basis. We simply couldn't do the work we do without our dedicated team of volunteers.

Office volunteers help us in a variety of different ways. The type of tasks they undertake include data entry, envelope stuffing, recording our newsletter, trimming stamps, packing delegate bags for our Conferences and much more.

Catherine and Hilary have been part of our office volunteer team for around five years. They are both local to our office in Buckingham and first got in touch after seeing a poster asking for volunteers.

Volunteering has been at the heart of both of their lives from their mid-teens and they have a strong sense of service. Hilary describes it as follows:



"My late father always told me that you serve your country and your fellow countrymen. He was quite volunteering minded and is my driving force and my inspiration."

One of the attractions of office volunteering is the flexibility. It isn't a regular commitment at the same time every week. As Catherine said: "I don't have to do it, I can do it when I like and I can fit it in with what suits me."

She goes on: "Little jobs that I can help with make a big difference to the charity and give me a bit of entertainment and a bit of something different for a morning. Reading for the magazine is a task I quite enjoy. I knew nothing about retinitis pigmentosa before I started. I didn't even know what the name meant."

Hilary volunteers for multiple different organisations and also invigilates for local schools during exam periods.



"Just to make a tiny difference to somebody. That's a job well done. I enjoy meeting different people from all walks of life. From all abilities and different cultures."

She adds: "It is a tremendous privilege and honour to be involved in helping others, however small or large the contribution is. It is a value that can never be taken away from you."

Catherine would recommend volunteering at Retina UK. "One of the really good things about coming to the Retina UK office is that everybody is so friendly. Somebody's always making you a cup of tea. I just feel part of the team when I'm there. It's really nice."



Conference volunteers

If you are in the Manchester area and would be willing to volunteer your time on Friday 27 and/or Saturday 28 September at our Conferences, we would love to hear from you. Call Sadia on 07856 907463 or email **volunteering@RetinaUK.org.uk**.

If you are interested in registering as an office volunteer with Retina UK, please get in touch. Call our Volunteer Coordinator, Sadia, on 07856 907463 or email her at **volunteering@RetinaUK.org.uk**.

THANK YOU

#TeamRetinaUK



Our **TCS London Marathon** 2024 team made history in April, raising over £65,000 to become our biggest event in eight years! We're so proud and grateful to our 22 runners for joining #TeamRetinaUK and taking on such an iconic race!



5K A Day in May, our new virtual campaign, raised over £7,500 in May. A huge thank you to all of our participants, including Lucy McFarlane (pictured), for raising funds whilst staying active during National Walking Month.



Back in March, five #TeamRetinaUK supporters (Richard, Chloe, Alex, Jessica and Tom, pictured) raised over £2,500 for Retina UK by running the **Retford Half Marathon** in Nottinghamshire!



At April's **Shakespeare Marathon** in Stratford-upon-Avon, Dillon Thompson raised £757 for our vital work.



As well as completing the TCS London Marathon for Retina UK, **Alastair Davies** also went continental and raised awareness for us by completing the Paris Marathon in April!



Max Mountstevens, a 7-year-old supporter from Plymouth, organised a presentation about the work of Retina UK to his school classmates and raised £310 for us by taking on a 50-mile cycling challenge.

Thank you to everyone who has fundraised for Retina UK over the last few months.



Team Finlay, made up of 100+ friends and family members of Finlay Macleod, took on the Bristol Half Marathon and 10K in May. They have raised a staggering £100,000+ for Retina UK in memory of Finlay. We are so thankful for their fantastic support.

Our supporters at **NOCN Group** in Sheffield and Norfolk organised bake sales for us in their offices over the Easter holidays and their tasty treats raised funds for Retina UK.



Hope To Seaview, the annual walk on the Isle of Wight, has raised over £40,000 for Retina UK over the years. This year's event, organised by Colin and Linda McArthur, formed part of their 50th wedding anniversary celebrations. Congratulations and thank you for your fabulous continued support!

Alex Crockford, a fitness influencer and long-term supporter of Retina UK, raised an amazing £730 with his recent skydive! Thank you, Alex!



Hazel Denby, Mark Baxter and Mark Bailey collectively raised over £2,000 by taking to the skies recently, as they took on thrilling skydives for Retina UK!



A huge thank you to Ty-Lomas King (pictured), Josh Brazier and Zachariah Noble for raising over £3,000 for Retina UK by running the **Brighton Marathon** in April.

If you would like to get involved, we'd love to hear from you. Call James on 07736 925174 or email james.clarke@RetinaUK.org.uk. Details of all of our upcoming challenge events can be found on our website: RetinaUK.org.uk/challenge.

Please share your photos with us at fundraising@RetinaUK.org.uk or tag us on social media.

Spotlight on a funded research project: retaining

Night blindness and restricted visual field experienced by people in the early stage of retinitis pigmentosa (RP) present serious challenges, but what if we could ensure that sight loss didn't progress too far beyond this, leaving central vision intact and preventing complete blindness?

That's the question being asked by the scientists leading one of the newest Retina UK-funded research projects.

RP principally affects rod-shaped photoreceptors, the light-sensitive cells that dominate most of the retina apart from the very centre. Rod photoreceptors work well even in dim conditions and provide nearly all our peripheral vision. However, the high definition image at our central point of focus is generated by a cluster of cone-shaped photoreceptors. These cells are sufficiently different to rods to remain largely unaffected by many of the underlying genetic faults that cause RP.

Project leader Dr Jörn Lakowski from Southampton University explains: "It's been known for many years that the loss of rods precedes that of cones, and that cone death only happens once rod density diminishes beyond a certain point. This suggests that during the course of the disease, there is a loss of some sort of support signal from rods to cones."



However, many questions remain about the mechanisms underlying this "cone starvation". Dr Lakowski has extensive experience of working with retinal organoids, living mini-models of the human retina that can be generated in the lab from adult skin cells. He saw an opportunity to use these to address the significant gaps in our understanding of how cones are lost in RP.

"The concept of a rod-derived survival factor remains untested in the human context" Dr Lakowski told us.

central vision in RP



“If we want to develop therapies that will stabilise cones in the diseased retina, we first need to understand how they are maintained under healthy conditions; one wouldn’t try to fix a car without first understanding how the engine works.”

Dr Lakowski and his colleagues will therefore use the organoids to gain a much deeper understanding of the contribution of the rod-derived cone survival signal, not only looking at healthy systems but also those where a pivotal element of the cone survival pathway has been “knocked out”.

Cone survival is already receiving some attention in the research world, with US-based company Sparing Vision undertaking early-phase clinical trials of a gene therapy to boost cone health, but a complete picture of its efficacy won’t be available for several years. In the meantime, Dr Lakowski will be attempting to identify new survival mechanisms in cones that may present novel treatment targets, as well as testing chemical compounds from an established library of existing drugs.

“I very much hope that the Sparing Vision trials will bring positive news, but we won’t know the outcome for quite a while” said Dr Lakowski. “Our work will fill in the blanks in terms of data on the underlying mechanisms, as well as establishing for the first time a human model system for cone starvation in

RP. We can leverage this new tool to identify compounds that can prevent cone cell death, with the ultimate aim of developing a drug therapy. Because most of the compounds we will test are already approved for use in other conditions, any that show promise in our project could be more easily translated into a treatment for RP.”

It will be intricate work, requiring immense commitment. “Growing retinal organoids is very challenging” Dr Lakowski explained. “The cells are very sensitive and need constant attention, even at the weekends. Maintaining them is labour intensive and requires an extremely dedicated team. The actual experiments are the “easy” part! Ultimately though, we hope that stabilising cone cells will deliver significant benefit to those living with RP.”

With your support, Retina UK can continue to fund research projects like Dr Lakowski’s. Play our Lottery (page 7), take on a challenge event (page 10) or make a donation online **RetinaUK.org.uk/donate**. Thank you for your support.

A focus on Usher syndrome

Our friends over at Cure Usher Syndrome have been working to raise awareness and fund research. They describe their work in the article below:

What is Usher syndrome?

Usher syndrome affects 4 - 17 in 100,000 people worldwide and is the most common condition that affects both hearing and vision. It is a condition where the individual will have retinitis pigmentosa and hearing loss. It sometimes also affects balance.

It is still under diagnosed; children born with a permanent hearing loss are not always routinely offered genetic testing to identify whether they have an associated gene fault. This means that children who already have a hearing loss go on to develop a sight impairment, which can come as a real shock to both the children and their families. There are three types of Usher Syndrome which have varying traits and different genes responsible. Typically, the individual will first notice night vision problems and will go on to lose peripheral vision as the cells degenerate in the retina.

Cure Usher Syndrome have been raising awareness of this condition and funding research to find a cure. We have celebrated some massive

milestones and seen some huge leaps in how well known the condition is. From seeing a character on Eastenders with the condition, having Emmerdale's actors Laura Norton and Mark Jordon share their experience with their children's diagnosis on ITV Drama Queens and our founder Jo Milne being invited by His Majesty the King to Buckingham Palace, we are reaching new audiences. It is with more awareness of the condition that we can reach people who are in a position to make real change to the lives of people with Usher syndrome and we are working hard to enact change on many levels.

Cure Usher Syndrome has partnered with Mariya Moosajee from the University of London to help fund her research. Mariya and her team are developing pioneering work on viral gene therapy which is an approach that has been successful in treating similar inherited conditions. If you are keen to find out about this fantastic work you can read more at **cureushersyndrome.com**.

Living with Usher syndrome

Emma Handford describes living with Usher syndrome.

"It was a hard truth to learn when my sister and I were told we had Usher syndrome. Back in 2003 times were different and the condition wasn't as

widely known about as it is now. Having a hearing loss since birth and then finding out that my sister and I were losing our sight was devastating.

Thankfully with time, that initial crushing news has changed to acceptance and now hope.

Back then I was working as a plumber, with grand plans to run my own business. The diagnosis finally gave me some answers as to why I struggled to work in dim places. I had to quit when I realised that career wasn't one I could continue with and for a while I struggled with what the future could hold.

Things turned around when I started working for the paediatric audiology department. I had grown up visiting for my hearing aids and realised I could train to be an audiologist.

It took a few years and some hard work but the result was extremely rewarding. I now help other people with hearing loss and I campaign with the charity Cure Usher Syndrome.

Being able to use my own experiences has helped me become better at my job and it's so much nicer than having to work on pipes - although the perks of my plumbing knowledge has helped with costs when it came to doing our own bathroom!



What was initially a crippling diagnosis, has now served to be an advantage. I no longer hate the condition, I live with it, make allowances for it and I make the most of it. It helps that I do believe there will one day be a cure and with the work we do with Cure Usher Syndrome, I'm given hope.

For more information about Usher syndrome, including two webinars about the condition, please visit **RetinaUK.org.uk/usher-syndrome**.

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