RETINA UK

ISSUE 166
WINTER 2018

LOOK FORWARD

FIND OUT MORE ON PAGE FOUR

We are Retina UK!

We're delighted to present your refreshed *Look Forward* as part of our name change from RP Fighting Blindness to Retina UK. This is an exciting time for us, and we're pleased so many of our community have supported us in this process. You can learn more about the charity's updated name and image later in this edition, and we encourage you to visit our new website **www.RetinaUK.org.uk**

Our name change isn't all that's been happening however; we've been busy preparing for 2019, assisting our amazing volunteers with their fundraising efforts and supporting pioneering research. Read on to learn more!

A new look for *Look Forward*

This issue of *Look Forward* is a very special one, as it is the first issued under our new name of Retina UK.

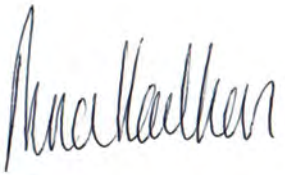
What an incredible year it has been for us, and I'm so delighted our new name and look has been so welcomed by the community. I truly believe it will help us reach even more people living with inherited sight loss, help us support more healthcare professionals and discover new potential partners for our work.

We are still committed and passionate about our core objectives; supporting the inherited sight loss community with the very best services and information, and funding leading researchers.

We are your charity and your support and input is vital to ensure we get it right. Do consider getting involved at our events or helping us raise funds for our pioneering research and unique support services.

Please do feel free to get in touch with me on **chiefexec@RetinaUK.org.uk** if you have any feedback or comments about the charity; we always welcome your thoughts.

Thank you for your ongoing support.



Tina Houlihan, Chief Executive

Retina UK funds medical research into inherited sight loss and offers a range of information and support services to those affected.

Retina UK, Wharf House, Stratford Road, Buckingham MK18 1TD
E: info@RetinaUK.org.uk · **T:** 01280 821334 (Office) · **T:** 0845 123 2354 (Helpline)

Do you follow us on Facebook and Twitter? Search for Retina UK on Facebook to find our page and various groups, and follow us on Twitter at **@RetinaUK** for the latest news and updates about the charity.

Registered charity number 1153851

One donation – twice the impact!

theBigGive.org.uk
CHRISTMAS CHALLENGE 2018

It's back! Once again, we are participating in the Big Give Christmas Challenge. This year we aim to raise even more money to fund the wide range of pioneering research we are currently funding, and have set an ambitious target of £30,000!

What happens during the Christmas Challenge?

Donations made to the project online on the Big Give website between noon on Tuesday 27 November and noon on 4 December will be doubled at no additional cost to our supporters.

Donations are doubled from a match funding pot provided by some amazing funders and supporters, as well as our Big Give Christmas Challenge Champion.

What do I need to do?

- Make a note in your diary for noon on 27 November and visit **www.RetinaUK.org.uk/BigGive**
- Follow the link to make your donation between that time and noon on 4 December.
- Spread the word amongst friends and family, and encourage them to donate during the campaign too.
- Follow us on social media for updates, videos and more.

If you are able to donate to our work this festive season this is the way to do it! Your contribution will go twice as far, and will help us drive progress towards effective treatments for inherited sight loss.

If you have any questions about the campaign or the project, or have any difficulty accessing the website to make your donation, please contact Deborah Laing, Senior Fundraising Manager, on 01280 815900 or email **deborah.laing@RetinaUK.org.uk**

Thank you so much for your support!

Retina UK – the new name

Wednesday 17 October was an exciting day for our charity – it marked the culmination of 18 months' hard work, investigation and consultation; it was when we changed our name from RP Fighting Blindness to Retina UK.

Many of you will be aware of the long in-depth process we went through to reach this important point; we wanted to ensure our community and supporters were behind the idea, and that we considered all options carefully.

Our Extraordinary General Meeting held in May revealed 86 per cent of members who took part in the voting process were in favour of adopting the name Retina UK, and our new look. We were delighted at this result, and it strengthened our resolve that this was the right path to take.

RETINA UK
WORKING FOR PEOPLE WITH INHERITED SIGHT LOSS

We felt moved to explore the concept of a name change in 2017 because it was becoming increasingly clear medical terminology was changing. Whereas in the past 'RP' covered all inherited sight loss conditions as an umbrella term, more and more people are being given more precise diagnoses – for example being told their condition is Stargardt disease, or Leber congenital amaurosis or Best disease. This meant many people didn't realise the charity RP Fighting Blindness was their charity, there to support them and funding research related to their condition.

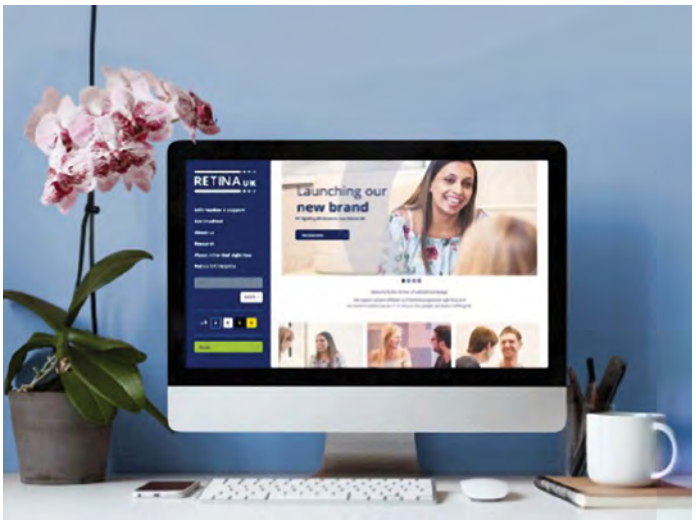
It was essential we addressed this. As understanding of inherited sight loss conditions accelerates thanks to the work of charities like ours, this concern was becoming more apparent. Under the direction of our Board of Trustees, we employed expert consultants to explore this issue for us, and make recommendations. This ultimately lead us to the conclusion that it was right to change our charity's name.

for RP Fighting Blindness

Undertaking such a project can prove controversial, so we were delighted when so many of our community supported us. Many of you called us during the consultation process to find our more, ask about the process and share your views.

> ***"Our new name and logo mark a pivotal moment for our charity and we are hugely excited by the opportunities that lay ahead."***
> *Tina Houlihan*

As part of the new image, we've had a new website built which you can find at **www.RetinaUK.org.uk** – we encourage you to visit our new site and explore the layout and wealth of information it holds. Our new website performs better on tablets and smartphones, as well as having been updated and re-authored where necessary.



Chief Executive, Tina Houlihan, said the new brand marked a step change for the charity. "Our new name and logo mark a pivotal moment for our charity and we are hugely excited by the opportunities that lay ahead. Our aims remain unchanged; we simply want to do more of what we do best, which is funding and stimulating ground-breaking medical research and providing high quality support for those affected by inherited sight loss conditions."

Should you have any queries about our name change or would simply like to give us feedback, please contact Thomas O'Neill, Communications & PR Manager, on 01280 821334, email **thomas.oneill@RetinaUK.org.uk**

Dinner raises £220,000 for research

The Eyes on the Future Gala Dinner, which took place last month at the Sheraton Grand London Park Lane, was hugely successful, raising more than £220,000 for our medical research project investigating RDH12-related Leber congenital amaurosis (LCA).

Organised by the family and friends of Vicky, a four year old affected by the condition, the event featured the visually impaired comedian Chris McCausland, soprano singer Lizzie Capener, a cocktail reception, luxury three-course dinner, a soul band and silent and live auctions. The 300 guests included industry leaders and senior executives at investment banks and funds, law firms and insurance firms in the City of London.

All proceeds from the event are funding our new £350,000 three-year project led by Dr Mariya Moosajee at UCL's Institute of Ophthalmology and Moorfield's Eye Hospital, which aims to study the disease mechanisms and identify potential treatments. The project was recently featured on the BBC's documentary *NHS at 70.*

The event was sponsored by Arrow Global, esharelife, Petroleum Equity, P&G Group: People with Disabilities, Lavazza and Allen & Overy, and supported by Barclays and Citi.

We'd like to say a huge thank you to everyone who helped make this incredible event such a success!

Challenge Events Calendar 2019

Why not make 2019 the year you get involved with some of our fun challenge events and raise money for Retina UK? We offer a wide range of activities you can get involved with no matter what your ability or where you live.

Call our Fundraising Events Manager, Emily Webb, on 01280 815900 or email **emily.webb@RetinaUK.org.uk** for details about any of upcoming challenges!

DATE	LOCATION	EVENT
3-8 February	Sahara	The Sahara Challenge
25 February	Thorpe Park	Half Marathon
10 March	London	The Vitality Big Half
12-13 April	North London	Tough Mudder
28 April	Virgin Money	London Marathon
4-5 May	London West	Tough Mudder
11 May	Midlands	Tough Mudder
18-19 May	Midlands	Tough Mudder
25-26 May	Edinburgh	Marathon Festival
June	Anywhere!	Retina UK Virtual Run
15-16 June	Scotland	Tough Mudder
7 July	Wales	5k/10k/half marathon/full marathon
27-28 July	London	London Triathlon
27-28 July	Yorkshire	Tough Mudder
8 September	Newcastle	Great North Run
13 October	London	Royal Parks Half Marathon
11-13 October	Hadrian's Cycleway	Retina UK Tandem Cycling Challenge
17-18 August	South West	Tough Mudder
7-8 September	North West	Tough Mudder
14 September	London South	Tough Mudder
21-22 September	London South	Tough Mudder

OXSIGHT

Helping People with Low Vision Gain Greater Independence

Dave and his wife, had always been keen travellers, taking in views from all over the world. Sadly, as each year past, his vision decreased.

Dave lost his sight due to a degenerative eye condition called Retinitis Pigmentosa.

In September 2018, Dave and his wife attended a wedding in Slovakia. Not only did they enjoy watching the couple tie to knot. Dave was able to use the modes within his OxSight Prism glasses to see faces and details of the wedding.

The next day, they travelled to a mountainous area on the Polish borders,

"We had a terrace where we stayed, I could SEE the mountains when I walked out first thing in the morning - It was amazing!"

"On top of the hill was a ruined castle, we had visited here before but I have never been able to see it, when I put my glasses on and looked up I was able to see it!"

OxSight Prism glasses have enabled Dave to see the world again after years of vision loss.

OxSight smart glasses are designed to enhance vision for people with peripheral vision loss caused by conditions such as Glaucoma, Diabetes, Retinitis Pigmentosa and other degenerative eye diseases.

Upcoming events:

Coventry - 20th November

Sheffield - 27th November

Visit: **www.oxsight.co.uk/events** for up to date event timings and locations

01865 580255 • care@oxsight.co.uk • oxsight.co.uk

Get in touch to request additional information or sign up for a demonstration

Investigating a revolutionary gene therapy technique for Usher syndrome

Thanks to support from our wonderful community, including those who participated in the Big Give Christmas Challenge last year, we have been able to commit funding to a three-year research project into a potential gene therapy treatment for Usher syndrome, led by Dr Mariya Moosajee at UCL Institute of Ophthalmology.

Usher syndrome is the most common cause of deaf-blindness worldwide, with a significant proportion of cases being caused by mutations in the USH2A gene. USH2A is a large gene, too big to be carried by the viruses traditionally used to deliver gene therapy. Dr Moosajee and her team will therefore work on adapting a non-viral gene delivery system known as S/MAR for the potential treatment of the condition.

The researchers have already generated a synthesised version of the instruction sections of the healthy USH2A gene, which can be inserted into the S/MAR vector delivery system. A person affected by Usher syndrome has also volunteered to donate some skin cells, which can then be converted into stem cells and used to create a "retina in a dish" model of the disease.

The team will be able to study the effects of USH2A mutations in this human cell model, as well as evaluate the impact of delivering healthy copies of the gene via S/MAR vectors. A zebrafish disease model will also be used to help the researchers investigate the effects of the treatment on visual function, toxicity and long term gene expression.

The successful use of S/MAR vectors could revolutionise the treatment of a wide range of inherited retinal diseases, not just Usher syndrome, by providing a safer and more applicable form of gene therapy, so we very much look forward to seeing the outcome of this project.

Widespread errors in “proofread

Mistakes in “proofreading” the genetic code of retinal cells is the cause of a form of inherited blindness, retinitis pigmentosa (RP), caused by mutations in splicing factors. This new understanding of the disease process, published in *Nature Communications*, is leading to the development of a gene therapy for RP caused by splicing factor defects. The work, led by Professor Majlinda Lako at Newcastle University, investigated how a common form of inherited blindness, RP, is caused by genetic defects in splicing factors.

Splicing factors are important protein components of the gene proofreading or “splicing” mechanism that is found in all cells. Some sections of our DNA, known as introns, are removed or spliced out by the cell during protein construction, so that only the final intelligible genetic code remains. This is because the introns do not actually provide any meaningful instructions for making proteins. Variations in splicing can cause very different consequences on the formation or function of cells, including retinal cells.

The scientists were able to create a “retina in a dish” using stem cells derived from the skin samples donated by RP patients at the University of Leeds. This cell model enabled the team to compare retinal cells to others in the body. These cells are normally very hard to obtain as they would previously have had to be donated from the retina, usually after death. Using this model, the researchers have shown that defects in splicing factors cause defective proofreading of components of the editing machinery itself. This counter-intuitive effect results in a “vicious cycle” of disruptive misinterpretation of the genetic code. The formation and functions of a special type of retinal cells, retinal pigment epithelial (RPE) cells, are the most severely affected. These cells are essential for supporting and nourishing photoreceptors (rod and cone cells),

ing" cause inherited blindness

so when they go wrong the light-processing function of the retina breaks down, resulting in sight loss.

The study shows, for the first time, how genetic defects in splicing factors cause variations in the proofreading of retinal genes, leading to defects in retinal cell function and their eventual degeneration in RP.

Professor Lako's team went on to show that CRISPR-Cas9 gene editing could be used to correct the genetic defects in a particular splicing factor. This also corrected the function of the RPE and rod and cone cells in their laboratory model, indicating a potential pathway to future treatments.

Professor Lako said: "This research gives us much deeper and broader insights into how splicing factors cause RP, enabling the next step in our research – the design of gene therapies for future treatments."

Co-author Professor Colin A. Johnson, from the University of Leeds, said: "We've been puzzled by the genetics behind these unique forms of inherited blindness for over 20 years. Our study is the first to really make sense of how these conditions develop, and I'm now very hopeful that this will lead to clinical trials for new treatments within five years."

Tina Houlihan, Chief Executive, Retina UK said: "We are delighted that our support has enabled the group to publish these important findings in *Nature Communications*. We look forward to seeing the development of this work through our newly awarded grant, which will allow Professor Lako and the team to further understand the mechanisms underlying this type of RP and progress towards the targeted treatment strategies our community needs."

Professor Mike Cheetham from UCL, scientific advisor for Retina UK commented: "It has been a conundrum why genetic changes in ubiquitous and highly conserved 'splicing factors' cause RP. This exciting work is a major step towards understanding how these changes in splicing factors lead to RP, and was only possible by using stem cells made from affected individuals and turning them into a 'retina in a dish'. There is still much to learn about why splicing factors are so necessary for the retina to function and how we might repair, or treat, this in individuals with this type of RP, but this work will focus research on the right models and pathways for future development."

The research was led by four teams in collaboration with 38 researchers worldwide: Prof. Majlinda Lako, Prof. Colin A. Johnson, Dr. Sushma Nagaraja-Grellscheid and Prof. Reinhard Luehrmann and Dr. Sina Mozaffari-Jovin.

You could be a winner!

Don't miss out on your chance to win £500, or one of our other fantastic prizes, just in time for Christmas!

The tickets for this year's Christmas raffle were posted out last month, so if you've received them and want to take part please send them back in with your payment as soon as you can – the deadline is Wednesday 12 December, with the draw taking place on Friday 14 December.

If you haven't received tickets, but would like some, contact our friendly fundraising team on 01280 815900 or email fundraising@RetinaUK.org.uk. You can also request tickets via our website at **www.RetinaUK.org.uk**

This year's amazing prizes include:

- **First prize – £500 cash!**
- **Mary Poppins Afternoon Tea for two at the Taj Hotel in London**
- **Four tickets to visit St Paul's Cathedral**
- **A copy of *Little Me* – comedian Matt Lucas' autobiography and signed by the man himself!**
- **A mini Christmas hamper**

Last year we raised almost £20,000 through our raffle, and this year you could help us raise even more to support ground-breaking research and vital support services.

Thank you and good luck!

Hope to Seaview

The Isle of Wight County Press

The inaugural *Hope to Seaview* fundraising event took place on Sunday 10 June on the Isle of White and was a huge success!

Organised by Retina UK Trustee Colin McArthur and his wife Linda, 70 amazing people took part to raise money and awareness of inherited sight loss conditions, by either walking a 12-mile route along the coastal path or cycling 50 miles around the island. Everyone donated to enter and many also collected sponsorship or made further donations, all of which will go towards our research fund.

At the end of the route in Seaview, the Town Crier welcomed everybody back and each walker received a medal and certificate. Everyone was feeling proud of themselves and pleased to have taken part in such an enjoyable and well-organised event.

A massive congratulations and thank you go to Colin and Linda, Mari and Mark from the Salix and everyone who worked hard to make this event happen. The total amount raised for this year's event was an incredible £15,068!

Next year's *Hope to Seaview* will take place on Sunday 9 June; if you're interested in finding out more you can email **fundraising@RetinaUK.org.uk** or call 01280 815900.

Introducing our volunteer

Here at Retina UK we have some truly amazing volunteer Ambassadors, who help to spread the word about our important work and share their inspirational stories.

Seema Flower – *visual awareness and disabilities consultant*

Seema aims to inspire people to achieve their full potential through her training, coaching and counselling. She lost her sight at an early age, but has not let this stop her building a successful chain of businesses and a thriving property portfolio. She often features in the media discussing issues affecting disabled people and entrepreneurial challenges. Seema, who is married with a ten year old daughter, said: ***"I have not let my sight stop me doing what I want, and I do not see obstacles as obstacles, but as opportunities. Having a disability, whether it is being blind, or any other disability should not hold you back in life or when seeking employment."***

Steve Bate MBE – *Paralympian*

Steve is a Paralympian who was diagnosed with retinitis pigmentosa (RP) in 2011, and won two gold medals and one bronze medal at the Paralympic Games in Rio de Janeiro in 2016, smashing the world record in the 4km pursuit. In addition to these sporting achievements, Steve has completed a number of remarkable challenges, including a solo ascent of El Capitan, a 3,000-foot-high climb in Yosemite in California. Despite his huge success, Steve considers himself a normal guy, and aims to inspire others living with sight loss to dream big. As Steve says: ***"Almost anything can be achieved if you are willing to work hard."***

Ambassadors

Bhavini Makwana – *activities coordinator at East London Vision*

Bhavini (and her lovely guide dog Colin!) has been involved with Retina UK for many years, and her work with us enables her to achieve her aim of empowering others living with similar conditions to her own. She organises a large number of events throughout the year, bringing people together to share their experiences and learn new skills. Bhavini has found the support Retina UK provided to her in the past invaluable: ***"The charity helped me find my confidence within myself and made me realise you can still achieve your ambitions even with RP."***

Victoria Claire – *professional sculptor and public speaker*

Victoria was diagnosed with RP at the age of 19, and registered blind at the age of 29, but has not let her visual impairment hold her back. A professional sculptor who has exhibited throughout the UK, as well as a keen surfer, skateboarder and musician, Victoria has made it her mission to share her story with others living with similar conditions. She was honoured to be offered the Ambassador role with the charity: ***"I first came into contact with Retina UK many years ago, when my parents encouraged me to seek out more information about my condition. Now that the charity is poised to increase the level of support it provides to people throughout the UK, it seems the perfect time for me to come on board and share my experiences."***

"My artificial vision system, the Argus® II, is the most amazing, life changing invention"

In 2009, Keith was one of the first patients in the UK to receive an Argus II Retinal implant at Manchester Royal Eye Hospital. The implant restores some useful vision to blind patients with RP.

Keith has been totally blind for many years due to RP. Diagnosed in his 20s while he was working as a butcher, he was registered blind in 1981 and was forced to give up work.

"The first thing I really remember seeing when my Argus II implant was switched on, was a bonfire night party with the grandchildren on November 5, and I was more excited than the kids.

"When they let the fireworks off I could see flashing lights and rockets and big fireworks going off in the night sky – it's the first thing I had seen for 25 years. It was a new world. It was wonderful. I had a few tears that day."

For more information about Argus II and the NHS, please call UK freephone 0800 520 0925

Second Sight

This advertisement is paid for by Second Sight and its inclusion in this newsletter does not imply any preferential endorsement by Retina UK

Send us your stamps

You can help support our work easily at home – just by saving the stamps from your post!

The used stamps on your morning post have a value to stamp collectors all over the world. We can also raise funds from unused stamps, stamp collections and first editions.

Perhaps you could collect stamps at work, or ask friends and family to help. If possible, please cut your stamps out of their envelopes, leaving plenty of room (5–10mm) around them, and post them to the address below:

Retina UK Stamp Appeal, Retina UK, Wharf House, Buckingham MK18 1TD

Scientists reverse congenital blindness in mice

A team of scientists led by Dr Bo Chen at the Icahn School of Medicine at Mount Sinai have successfully reversed congenital blindness in mice by changing supportive cells in the retina called Müller glia into rod photoreceptors. The findings, published in the journal *Nature*, advance efforts toward regenerative therapies for blinding diseases such as age-related macular degeneration and retinitis pigmentosa.

Researchers have long studied the regenerative potential of Müller glia cells (MGCs) because in some species, such as zebrafish, they divide in response to injury and can turn into photoreceptors and other retinal neurons. The zebrafish can thus regain vision after severe retinal injury.

"Between four and six weeks after the reprogramming, the blind mice regained their vision to some degree."

In mammals, MGCs are not so adaptable and cannot spontaneously differentiate into retinal neurons, meaning that retinal damage is often irreversible. However, Dr Chen's team successfully developed a two-step gene transfer process that reprogrammed mouse MCGs and forced them to develop into rod photoreceptor cells.

After this two-step reprogramming, the team found that the new rod photoreceptors were integrated into the existing retinal structure. They saw no difference between these new cells and the original rod photoreceptor cells. The cells sensed light, which then triggered information to be sent to the visual cortex (brain) and restored function of the visual pathway.

Between four and six weeks after the reprogramming, the blind mice regained their vision to some degree, although the researchers could not measure the level of improvement, and must do further testing to find this out.

"This is the first report of scientists reprogramming MGCs to become functional rod photoreceptors in the mammalian retina," said Dr. Thomas Greenwell, program director for retinal neuroscience at the National Eye Institute.

"Rods allow us to see in low light, but they may also help preserve cone photoreceptors, which are important for colour vision and high visual acuity. Cones tend to die in later-stage eye diseases. If rods can be regenerated from inside the eye, this might be a strategy for treating diseases of the eye that affect photoreceptors."

Retina UK Challenge Series

Join us for our 2019 Retina UK Challenge Series! Each month take on a different challenge to raise money and awareness to support our work. For every challenge completed you will receive a unique pin badge. Collect them all and we will send you something special to help you display them! There are many ways you can interpret each month's challenge. Below are some of our ideas:

Juicy January:
Swap alcohol and/or fizzy drinks for juices and smoothies – ask friends, family members and colleagues to sponsor you to keep it up for the whole month! Hold a smoothie making competition or make your own signature smoothie and sell it. Or why not have a night in with friends?

Flipping February:
Hold a pancake flipping competition. Charge a small fee to enter, and offer a prize for the best flip! Send in videos and you could feature on our social media channels. You could even hold a second competition for the best or most unusual pancake topping!

Marathon March:
Run/walk/cycle/horse ride 26.2 miles during March! Whichever way you decide to take part you can be a part of Marathon March! Track your distance and send in your results to us! You could ask people to sponsor you per mile or for the whole challenge.

Arty April:
There are so many possibilities with Arty April! You could hold a drawing/painting competition, put on an art workshop, photography competition or put on a play!

Musical May:
Put on a music quiz (name that tune), hold a fundraising concert or how about a karaoke competition?

– new for 2019

Jogging June:
Sign up and be part of our first virtual run! Set your own target and track how far you run. Like with Marathon March, you could ask people to sponsor you per mile!

Jiving July:
Dance for Retina UK! Hold a dancing competition and send in your videos for the chance to be featured on our social media channels. Or you could hold a danceathon and ask people to sponsor you per minute!

Afternoon Tea August:
Hold an afternoon tea or cake sale for Retina UK. Charge people to attend, and you could even hold a raffle alongside it.

Swimming September:
Swim once a week or set yourself a target to swim a certain number of lengths during the month.

On your bike October:
Brand new event for 2019! Our official tandem cycling challenge! Watch this space for details, or email **emily.webb@RetinaUK.org.uk** to be the first to find out more.

Non-uniform November:
Take part in a dress-down or non-uniform day and ask people to donate to take part.

Deck the halls December:
Take part in a Christmas activity for Retina UK! From carol singing to Santa runs there are a lot of options available!

If you'd like more information on any of the challenges, contact Emily or Leanne on 01280 815900 or email **fundraising@RetinaUK.org.uk**

Volunteer with Retina UK

Would you like to make new friends, gain confidence, or get experience for work or further education? If so, consider volunteering for Retina UK.

We currently have over 70 volunteers across the UK who support us in a variety of ways. There are many different ways you can get involved:

- Joining our telephone and email help-line team
- Being part of our telephone and email support service
- Hosting a peer support group
- Attending information events on be-half on Retina UK
- Supporting our social media activities
- Public speaking
- Helping at marathons or other events
- Allowing us to share your stories about sight loss and your experiences
- Assisting with administration or office work
- Lending us your professional skills, knowledge or services

Without our amazing volunteers we wouldn't be able to undertake the variety of work we do on behalf of the inherited sight loss community.

If you would like to find out more please email **volunteering@RetinaUK.org.uk** or call us on 01280 821334 and speak to Clair our volunteer coordinator.

"Becoming a volunteer for Retina UK is one of the best things I have ever done."
Khadeja Ali, Retina UK Helpline Volunteer

Articles for the Blind Scheme

Did you know we can post this newsletter to you with no postage costs to our charity under Royal Mail's Articles for the Blind (AFB) scheme? All you need do is notify us you are blind or visually impaired - we cannot make that assumption - and that you are happy for us to send *Look Forward* to you under this scheme.

Please email info@RetinaUK.org.uk or call us on 01280 821334 and ask us to record this information on our database. If you already receive this newsletter from us marked Articles for the Blind, there is no need to do anything.

The money we save on postage costs will be invested in our information and support services and research projects.

Giving in memory of a loved one

Making a donation or choosing to fundraise in memory of someone special can be a wonderful way to pay tribute to them, whilst helping Retina UK to fund pioneering medical research and vital support services. We are here to help you every step of the way, whether you choose to collect donations at a funeral or memorial, or set up an online donation page.

Retina UK supporters Christine Reed and Angela Forbes collected over £500 in memory of their mother Rosemary Treen at her funeral. Rosemary had a long association with our work, having been part of the original Devon & Cornwall Branch of our charity from its inception.

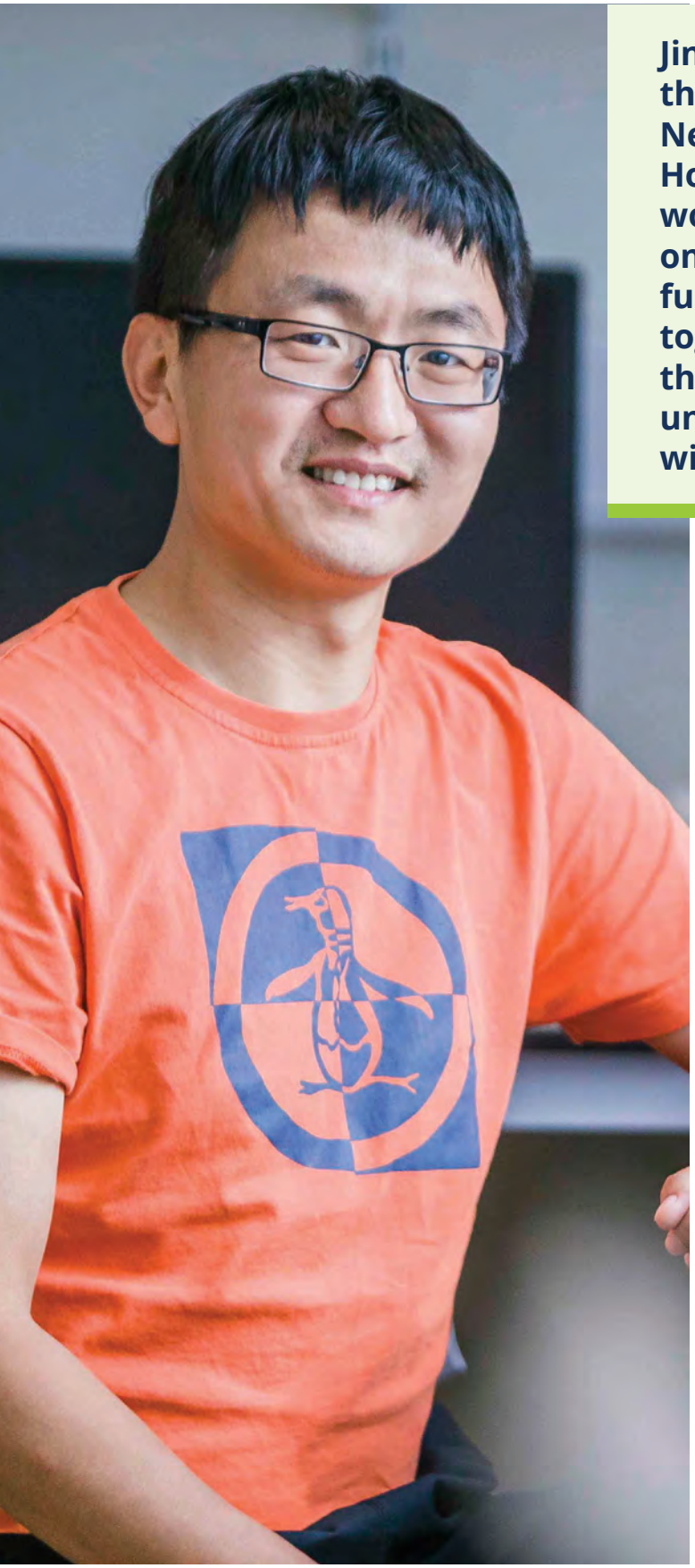
"It was a lovely way to honour our mother's memory, supporting a charity so close to her heart." Christine Reed and Angela Forbes.

Coming soon...

the brand new Retina UK online shop! Buy Christmas cards, t-shirts and merchandise at **www.RetinaUK.org.uk/shop**

Our 2018 Christmas cards will be available shortly!

Meet the Researcher: Jing Yu



Jing Yu is a Research Assistant at the Nuffield Department of Clinical Neurosciences at the John Radcliffe Hospital in Oxford. He is currently working with Prof Susie Downes' team on the RP Genome Project, an initiative funded by Retina UK, which brings together leading research centres across the UK to increase knowledge and understanding of the genes associated with inherited retinal disease.

Jing works in bioinformatics, an interdisciplinary field that develops methods and computer software tools for analysing large amounts of biological data and is an essential component of the search for new genetic causes of eye disease. Here he tells us about his work, his motivations, breakthroughs and challenges.

"My job is akin to the work of a detective. The clinical notes on a patient's disease are like the description of a crime scene, and the genetic changes found by closely examining their genetic code are like tens of thousands of suspects.

"In order to find the one or two genetic changes that are responsible for the disease, I need to ask a series of questions of each 'suspect'. For example: is this mutation rare in the general human population? (only rare mutations can cause rare diseases);

and: has this gene been linked to a similar disease?

"The workload can be massive considering the number of patients in the study – over 600 and growing – and the idea of manually inspecting each of the thousands of genetic variants in each patient is unbearable! That's where I come in as a bioinformatician. I write code to instruct computers to inspect millions of mutations and only report the ones that are interesting. I am also good at representing data visually, which has been key in delivering analysis results to scientists and doctors.

"Before working with Prof Downes, I had been involved in lab-based research on bacterial genetics. However, I had become more and more intrigued with computer coding and number crunching, so I taught myself bioinformatics. Then I saw this job advertised, looking for someone with my mix of experience, and I jumped at the chance.

"Because of my work, they will be more likely to know which gene is faulty, the chance of the disease passing down to their children and, in some cases, how the disease will progress."

"To be honest, the fact that the work was in inherited retinal disease was not a major factor to start with, but now that I have learned about how these diseases affect patients, I feel a great responsibility to help these families however I can. I had never realised how eager patients are to know what is going wrong with their genome, until I met some of them at a public event hosted by Retina UK last year.

"Because of my work, they will be more likely to know which gene is faulty, the chance of the disease passing down to their children and, in some cases, how the disease will progress. Our research also brings hope. Gene therapy has made huge progress in recent years, bringing with it the very real possibility that inherited sight loss could be stabilised or even cured, but it will depend on knowing the identity of the faulty gene for a particular patient.

"Our work has produced some exciting developments. Early on in my time here, I wrote some code that has not only led to the speedy genetic diagnosis of many of our patients, but has also become part of a larger project called Phenopolis (phenopolis.org). This is a web-based, user-friendly bioinformatics tool that could be used by researchers in other fields, not just inherited retinal disease.

"I love this job, so much so that I have declined a couple of more secure posts so that I can keep working in this area. People affected by inherited sight loss make an immense contribution to my research: without their kind collaboration I would have no data to work on, and would solve nothing. After all, how can a detective work with zero evidence?"

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