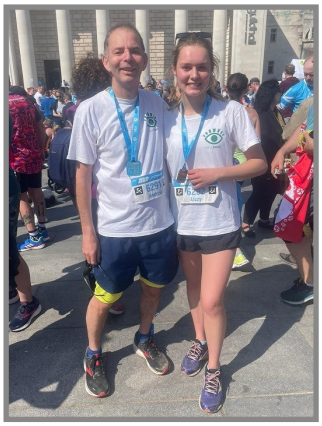
Gift of SIGHT

JUNE 2022 NEWSLETTER

Dear friends and supporters



My heartfelt thanks to those of you who sponsored my daughter Lizzy and myself for our ABP Southampton half marathon. We managed to raise £1000 which was a great result—for us and for our research studies.

We are now recruiting patients at Southampton Eye Unit who meet the entry criteria to participate in a new clinical trial for 'wet' age-related macular degeneration (AMD). We hope this will lead to better outcomes for patients affected with this type of the disease. In addition, over the next two to three years we will be testing whether gene therapy will maintain vision for patients with 'dry' AMD. This will be the first time we have actually performed this procedure in our eye department and is an exciting step forward. This work builds on our genetic studies identifying which genes are faulty in AMD. June 2022 also marks the end of our multinational recruitment to the Pinnacle study of patients with early AMD. We have recruited around 400 patients who will be followed for up to 3 years and hope that by doing this we will find novel insights into the progression pathways for this disease. Further clinical trials are being developed in Southampton which we anticipate will be rolled out over the next year.

My PhD student, Dr Rebecca Kaye, has been awarded the Faculty of Medicine Michael Arthur clinical academic trainee prize. This was for a publication showing that healthy people with genetic risk factors for AMD have thinner retinal layers, suggesting that the disease starts much earlier in life than previously suspected. This work and details of our Pinnacle study can be read about in two recently published papers available here and through this link. None of this work would have been possible without generous donations to the Gift of Sight Appeal.

Your support has again proved immensely beneficial to our laboratory team as Gift of Sight funds have enabled us to send an expensive camera to the USA for repair. The repairs, costing £35,000, will allow us to continue a vital laboratory study to investigate a potential treatment for AMD. We would be unable to continue the project without this piece of kit and we're immensely grateful for your help.

Finally, I am delighted to share snapshots from early career researchers and PhD students on the following pages. I know you will share my pleasure that we are helping the next generation of visual scientists become confident and independent, ensuring the continuation of vision research.

Thank you again for all your support and interest for our work.

Andrew Lotery

Andrew Lotery MD, FRCOphth
Professor of Ophthalmology
University of Southampton

Dr Rebecca Kaye

Hello, I'm Rebecca Kaye a PhD Student and Clinical Research Fellow working with Professor Lotery and Dr Jorn Lakowski. I'm an academic clinician, which means I have taken time away from my clinical duties as an Ophthalmologist to conduct a PhD.

My research surrounds Age-Related Macular Degeneration (AMD), specifically using stem-cells derived retinal pigment epithelium (RPE) to model the disease. We are interested in the role of a novel AMD genetic target called TSPAN10 and how it impacts the RPE. To understand this we have generated stem cells in which TSPAN10 is 'knocked-out', using a technique called CRISPR/cas9. These new modified stem cells have been differentiated into RPE. Interestingly, so far we have seen changes in the level of pigment these cells have and we are very excited to continue this research. Eventually we hope to understand how TSPAN10 functions in the RPE and its contribution to AMD development.

Alongside laboratory work I recently had a paper published in Scientific Reports investigating the role of AMD genetic changes on retinal thickness. This work was presented at The Academy of Medical Sciences Clinical Academics Training Conference and, as mentioned by Professor Lotery, also won the University of Southampton Michael Arthur Clinical Academic Trainee prize.

As a clinical academic it's hard to balance research and clinical time. The COVID-19 pandemic particularly affected the amount of surgical training available for trainee ophthalmologists, meaning lots of trips to the eye simulator for surgical simulation training. I do still have some clinical duties to keep my skills in check, including patient recruitment for the PINNACLE trial. Ultimately it would be fantastic to produce research in the lab that I can directly transfer to my clinical practice. Gift of Sight has been fundamental to this work in many ways. Thank you.

Dr Ellie Keeling

I worked for five years with Arjuna Ratnayaka to research the underlying mechanisms associated with age-related macular degeneration. There is a waste disposal system within the retinal pigment epithelium (RPE) which removes used up photoreceptors. This process is damaged with age and other AMD risk factors including a high fat diet, which causes the accumulation of incompletely degraded photoreceptors within the RPE, leading to cell death. This work has generated numerous publications and Ms Rebecca Miller is now continuing the project and is completing her PhD looking at how we can reboost this system as a potential therapeutic target area.

I am now working in paediatric vision research with Jay Self to screen for compounds which can overcome genetic mutations associated with albinism. Albinism patients have reduced pigment in their eyes as well as hair and skin. This reduction in pigment is caused by alternations to the Tyrosinase gene. We have created a cell culture model which contains these genetic mutations and found that the cells themselves have pigment levels that are significantly lower than the unmutated sister cultures. Interestingly, these levels match the amount of pigment seen in patients with each mutation. We are now screening a panel of FDA approved drugs to see if any can increase pigment levels.

Thank you for your donations to Gift of Sight which have supported my journey through the labs and ensuring we have up to date and reliable equipment.

Sarah MacDonald

I completed my integrated masters in Genetics at the University of York, where I also had the opportunity to delve into neuroscience. My specific area of interest is rare human genetic disorders with a neurological component.

I am coming to the end of the first year of my three year PhD investigating new treatments for children who have albinism. This lifelong condition affects the production of melanin, the pigment that colours skin, hair and eyes. The reduced amount of melanin can cause eye problems because it is involved in the development of the retina, the thin layer of cells at the back of the eye.

However, there is hope. Children's eyes continue to develop during the first 18 months of their life and previous research at the University of Southampton showed that L-DOPA, a drug that is usually used to treat Parkinson's Disease, could alter eye development if it was given at this critical time. My PhD is expanding this research and analysing how the structure of the eye – in particular the synapses around the retina – change once the drug has been given at either a low, medium or high dose. With just preliminary data coming through I can already see an impact. I now need to analyse this data to see how we can use it for the benefit of patients but it's a big step in the right direction.

I'm amazed by the generosity of someone who has given a donation to fund my project for three years and I want to make sure that my work makes an impact and helps as many people as possible. On a personal level it was also really important to me to undertake a project that would have a positive impact on people's lives. To help with the foundation of that knowledge and understanding of a rare disease is a prospect I find very exciting.

Catherine Robertson

Responsible for your sense of sight, cones cells are the receptors in the retina and convert the light that enters your eye into electrical signals that can be decoded by the vision-processing centre of the brain. Cone-rod dystrophy (CRD) is a group of inherited eye disorders that affect the cone cells and lead to vision loss over time. There are over 30 types of CRD caused by genetic changes in several different genes that can be inherited in many ways.

My research is investigating the role of the gene GNGT2 in the development of cone cells and how they absorb light.

Around one in 4,000 people are affected by CRD and experience debilitating sight loss, having a marked effect on their quality of life. My project is using human stem cell derived organoids to model the retinal developmental process, to identify the genes that influence cone cells. This could help us find a potential target from treatment.

I'm incredibly grateful for the support people have shown this project - knowing that the funding for my PhD has been donated brings an extra sense of commitment and has encouraged me to give back. I know other members of our team have raised money for Gift of Sight through running events and that's something I would like to get involved in too.

I'm very proud to be part of the team here in Southampton.

FORTHCOMING EVENTS

The EYE-land Fete 31 July 2022 2.00pm—4.30pm

At The Island, Greatbridge, Romsey SO51 0HP (thanks to Christopher & Sarah Saunders-Davies)

The event will support children's eye research. **Entry £2.50 (Under 3s free).**

Registration details are given on our website www.giftofsight.org.uk/events

Refreshments, stalls, games for children of all ages, Tombola, 'Meet the Scientists' and stories for children read by 'Granny Bean'

Opening by Lady Mary Fagan at approx. 2.15pm

Kindly sponsored by Butterflies Healthcare VITEYES LIFE IN SIGHT

Please phone Ailsa for details if you'd like to come and help at the fete. 023 8059 9073.

Card readers will be available for paying for tea etc. but if you can please bring cash for games.

Gift of Sight 'Beating Blindness online lecture : 21 September 2022 6.00pm—7.00pm

Following the success of our on-line Lecture in 2021, our colleagues in our University Events team are organising another Lecture to give further updates on how our research is moving forward, leading to the potential development of new treatments for some blinding eye conditions.
Full details of the event will be added to our website and shared on social media.

Professor Andrew Lotery will be speaking at the Open Sight Eye Contact Exhibition on Wednesday 28 September 2022 at Southampton Football Club's St. Mary's Stadium, Southampton, SO14 5FP. We'll be there with our Gift of Sight stand so do please come and chat to us.

WE REGRET THAT WE ARE NO LONGER ABLE TO COLLECT MILK BOTTLE TOPS.

Contact: Ailsa Walter | T:**023 8059 9073**
Email: **info@giftofsight.org.uk** | Web: **www.giftofsight.org.uk**

To donate:

By card: Tel. 023 8059 7239

By cheque: Please post to Gift of Sight, Office of Development and Alumni Relations, Building 37, Highfield, Southampton, SO17 1BJ

Online: https://donate.giftofsight.org.uk/

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