Visionaries

Fighting Blindness Newsletter | Summer 2020

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FIGHTING BLINDNESS

Cure. Support. Empower.

Welcome

Dear members, supporters and friends,

I hope this newsletter finds you healthy and happy. The past three months have been so difficult for so many, but as a nation I believe we can be proud of what we have achieved.

Personally, I'm also proud of what our team have achieved. Within days of lockdown commencing, our Insight Support Service moved face-to-face counselling sessions to telephone and our groups all went online. We also set up a virtual coffee morning which is proving particularly popular – within weeks it was fully subscribed.

The fundraising team quickly adapted to their new normal and this month we're running a virtual fitness challenge – Steps for Sight. We want you to walk, jog or run a half or full marathon in June. Find out how to sign up on the back page!

However, if a holiday sounds more appealing than a marathon, our brilliant ambassador Victoria Smurfit has you covered. She wants to raise €20,000 to help us through this difficult period. She's giving away a three night break for two people on the Channel Islands! Full details can be found on the next page.

Throughout the crisis we have not lost sight of our mission to find cures and treatments for sight loss. I'm delighted to report that in recent months three of our funded research teams have published new findings in scientific journals.

You can read about the developments on pages four through to six.

Our team was saddened to hear of the passing of Llura Gund in March. Llura was married to Gordon Gund and together they founded Foundation Fighting Blindness in America. The couple are firm friends of Fighting Blindness and provided inspiration and support to our own founders. May she rest in peace.

I would also like to extend the team's condolences to Fighting Blindness member Padhraig McLaughlin on the recent loss of his father Jack McLaughlin. Ar dheis Dé go raibh a anam.

Finally, I want to thank everyone who has donated, signed up for our Private Members Draw, renewed their membership, shared our content with friends, volunteered their time or completed a survey. Your support is vital at a time of crisis and we are so grateful.

Kindest regards,
Kevin Whelan
CEO

Win a holiday to the beautiful Channel Islands!

Our dedicated ambassador Victoria Smurfit is offering you the chance to win a three night break for two in the Blonde Hedgehog hotel in Alderney.

The lucky pair will also win return flights to the island on a private plane! The prize includes flights from Dublin to London.

To win, simply donate €5 to Victoria's JustGiving page. Help her reach her fundraising target of €20,000. All of the money raised will be used to fund our vital research and support services.

If you would like to enter over the phone using your debit or credit card, you can call a member of the team on 01 678 9004.

A winner will be randomly selected on Tuesday 30 June. Good luck!

Zebrafish studies reel in answers on how we see

A team of researchers at University College Dublin (UCD) has dedicated significant time to understanding how the visual cycle works inside retinal cells.

The outcomes of their studies have provided further insight into the different mechanisms within cone cells that are responsible for enabling vision in bright light.

This research team used a zebrafish – a small tropical fish – as their eyes are very similar to humans. Specifically, they used a strain of zebrafish that has been shown to rely exclusively on cones for their vision. They then blocked the function of some proteins known to play a role in the visual cycle.

For example, they blocked the function of RPE65 in zebrafish using Emixustat, a drug currently in phase 3 clinical trials for use in Stargardt disease. Within the retina, RPE65 has an important job in switching 'inactive' dietary vitamin A to 'active' light sensitive vitamin A.

The zebrafish were then placed in the dark for a period of time and their responses measured immediately following exposure to light, 30 minutes after light adaptation and 4-6 hours after light adaptation.

This enabled the scientists to understand what is regulating immediate, early or late phases of cone phototopic vision.

The study found that when RPE65 is blocked, zebrafish had poorer vision when exposed to light immediately after a period in the dark.

Professor Breandán Kennedy, who led this research consortium, said, "A puzzle for vision researchers is to understand how cone photoreceptors continuously recharge with light-sensitive vitamin A during daylight conditions. Surprisingly, we discovered that if the zebrafish with impaired vision due to emixustat treatment, were exposed to light for 30 minutes or longer, they regained full vision.

"This indicates that RPE65 is required at night to provide light-sensitive vitamin A for vision in the morning. But, during the day, RPE65 is not required, and the eye uses light to regenerate the light-sensitive vitamin A – a clever, efficient solution."

Through the incorporation of strong public and patient involvement in research, this is a motivating example of how the priorities and lived experience of those affected by visual impairment can complement the scientific expertise and demonstrates the essential integration of all perspectives to yield more relevant and impactful research. By knowing how vision works, it becomes much easier for us to fix, and in our case to develop treatments that will prevent or restore vision.

The full research study was published in the Journal of Biological Chemistry.

Identification of irregularities in Stargardt gene

Collaborators, led by the Fighting Blindness funded researcher Prof Frans Cremers, have identified the changes in the ABCA4 gene responsible for Stargardt disease in 448 people.

The research was made possible through the collaboration of 75 scientists, some of which we proudly fund, across 21 countries around the world. Irish researchers based at Trinity College Dublin were part of this collaborative effort and Irish patient samples were used in this study.

In their research paper, the team describe their cost-effective methods that allowed them to sequence the entire ABCA4 gene in more than 1000 samples. This new method replaced conventional genetic testing which analyses only certain parts of the gene, the region that is responsible for the production of the ABCA4 protein, for example.

Incredibly, the team of researchers identified 105 previously unknown changes (sometimes termed mutations) following sequencing of the entire gene.

These irregularities affect normal ABCA4 gene function resulting in central vision defects and identification of these changes has significantly improved the knowledge-base of the processes through which Stargardts occurs.

In addition to changes in the gene, 16 new deletions (loss of parts of the gene), were also identified.

These new findings were found in 27% of 448 genetically resolved cases, which researchers believe indicates a high number of these genetic changes are associated with Stargardt disease developing.

It is thanks to the generosity of our members and the general public that we are able to fund this vital research.

Among the Irish effort in this study was Fighting Blindness funded PhD student Laura Whelan: “As part of this momentous study, I worked alongside an international team to detect changes in DNA that may cause Stargardt disease. I gained invaluable skills that are now being used by our research team in TCD Genetics and could also be applied to other forms of visual impairment in Ireland in the future.

“Affordable DNA sequencing technology used here to analyze one gene implicated in Stargardt disease will now be used soon to sequence more than 200 genes involved in inherited blindness, having a positive impact for many.”

The full research study was published in Genetics of Medicine.

Potential new therapeutic target for eye disease

Research led by a team at Trinity College Dublin, with funding from Fighting Blindness, has pinpointed a potential new therapeutic target for treating retinal degeneration.

The researchers have discovered that a protein called SARM1 involved in neuronal cell injury may also have a role in the progression of retinal degeneration.

In their study the team, led by Dr Sarah Doyle, first generated genetic mouse models of retinitis pigmentosa (RP) by blocking the function of the rhodopsin gene, an important gene for vision.

Using this disease model they overexpressed the amounts of SARM1 that is produced in the cell. This was shown to drive and accelerate photoreceptor death in the mouse model.

In contrast, when they then genetically suppressed the expression of SARM1 in the cell, there is a delay in photoreceptor death in the disease model.

Furthermore, at advanced stages of disease and with the suppression of SARM1, there still seemed to be evidence of cone function.

Ultimately what the results suggests is that by deleting SARM1, you can promote rod and cone cell survival.

Explaining the significance, Dr Sarah Doyle said: "Our research indicates that SARM1 is likely to be a key executioner in the process of retinal degeneration, because if we remove it from our experimental model system this has the effect of delaying the photoreceptor cells from dying.

"This is an important finding because the first steps involved in processing 'light into sight' take place in the photoreceptors. As a result, losing photoreceptors ultimately equates to losing vision.

"For this reason, interventions that prevent or delay photoreceptor cell death are critical to preserve sight for as long as possible in people with degenerative retinal diseases."

This work will undoubtedly continue to grow and progress even further over the next while with the potential to advance into a study that may develop an intervention that would slow or delay vision loss in retinal degenerations.

The full research study was published in Life Science Alliance.

A message from Peter O'Toole, Support Services

Over the last three months we've been delighted to witness an overwhelming sense of solidarity and support fostered by a strong and engaged peer community.

We believe this has helped our service users to re-establish a defined structure, some degree of routine and a sense of belonging and connection during these uncertain times.

We also believe that this very spirit of support and connection is something that will endure after the health crisis passes. We would like to thank all of our dedicated volunteers, we couldn't do it without you and we really appreciate your hard work.

Insight Support Services counselling update

Within two days of the closure of the office we had contacted all our clients and transferred our face to face counselling to a service delivered exclusively by telephone.

Telephone counselling has always formed part of our service and it's a preferred mode of communication for many who are unable to travel to our counselling centre.

Our Need to Talk counselling service, which offers free face-to-face counselling in the border counties of Ireland, also made the same transition. Our counsellors and clients located in this region have adapted well to the transition and we continue to receive referrals.

In addition, we developed a "Reach out" service. It provides the comfort of having a regular phone-call or a once off chat with one of our team to help people stay connected.

Our team are available to check in on how individuals are coping and provide help to find appropriate resources. In the last eight weeks we have engaged with 80 people in this manner and the service has been greatly appreciated.

For anyone who is need of counselling support, we would be happy to have an initial chat to discuss the options available. Please contact us on 01 674 6496 or email us on insight@fightingblindness.ie.

Covid-19 changes to support and social groups

To address the difficulties relating to isolation, Insight Support Services has prioritised our focus on ensuring that our clients and service users stay connected during these difficult times.

We have always provided a number of peer-focused services, such as our Technology Exchange Club, Mindfulness sessions and peer support groups. All of these have moved to online communication, primarily using Zoom as a platform.

Using a new mode of communication can be difficult. To help with this, our team have provided training on how to use Zoom to over 150 individuals, both on a one to one basis and in a group setting. We received very positive feedback and participants told us that the skills learned have added to their ability to engage, not just with our support groups, but also with family and friends.

Practically every support group we run has increased participant numbers since the health crisis commenced.

Our Technology Exchange Club has more than doubled in numbers of those engaging with the service. Attendance at our Mindfulness sessions on Wednesdays has increased by 400%.

Sligo based VIP group looking for new members

As part of our engagement in the border counties through our Need to Talk project, we've recently started a peer group in Sligo.

This group came together as a result of our recent Living with Sight Loss course held in Sligo. Since the start of the Covid-19 crisis, they have moved their meetings online and use the free platform Zoom to connect.

The group is open to people living with sight loss in Sligo. To find out more please contact Tina Leonard at tina.leonard@fightingblindness.ie or call 01 674 6496.

Connections Coffee Mornings

During Covid-19 we became aware of a need for people to engage with each other, often just driven by a need to talk, share stories and experiences.

To meet that need, we launched our virtual Connections Coffee Morning eight weeks ago. An instant success, the group is already fully subscribed! The success has been largely based on the willingness of participants in providing guidance, tips, support and advice to each other.

Resources and ideas are exchanged; many make arrangements to meet online during the weekend and new friendships are being forged.

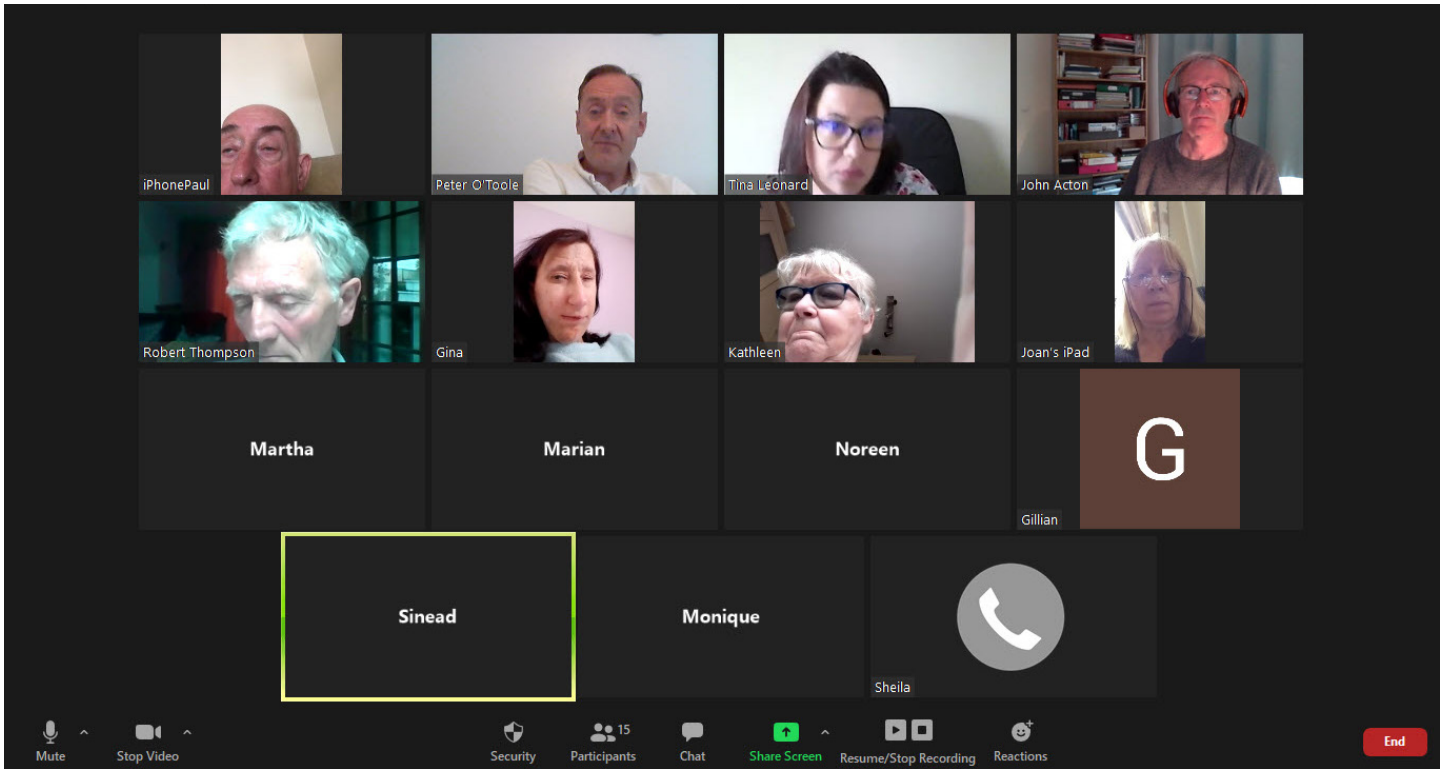
The group love to share stories of what they're eating and drinking and new recipes and ideas for cooking and baking are regularly circulated!

Members of the Fighting Blindness staff team often join the group to discuss research, health advice, fundraising and promotion.

In addition, guest speakers such as Stuart Lawlor from Sight & Sound and Paul Norton form Sightless Cinema have joined the mornings to discuss additional services and resources available through other organisations.

Our move to online engagement has presented an opportunity to extend our support and engagement to a much wider audience nationally.

In our Connections Coffee Morning alone, we have participants from Dublin, Waterford, Wicklow, Cork, Westmeath, Meath, Donegal, Wicklow, Sligo, Cavan, Galway and Limerick!



Luxturna™ - reimbursement of gene therapy

Luxturna™ (voretigene neparvovec) is the first gene therapy approved for the treatment of children and adults with vision loss as a result of mutations in both copies of the RPE65 gene.

Conditions caused by this type of mutation include very rare forms of Leber congenital amaurosis (LCA) and retinitis pigmentosa (RP).

Novartis, the pharmaceutical company responsible for Luxturna™, has now applied to the HSE to have this drug made available in Ireland.

Because this drug is new and considered to have a high cost, it must undergo what is called a full Health Technology Assessment (HTA), a process carried out by the National Centre for Pharmacoeconomics (NCPE).

They will evaluate and assess all evidence surrounding Luxturna™ including the effectiveness of the drug, cost effectiveness and budget impact.

The role of patient involvement, through the sharing of personal perspectives, in decision-making processes like this is being increasingly recognised.

As part of the HTA, Fighting Blindness will submit a supporting document that will highlight directly to the decision makers, the true lived experiences and impact of these conditions.

The NCPE will then include the Fighting Blindness Submission of Evidence in their final report to the HSE.

It is the HSE Drugs Committee that makes the final decision on whether or not to fund Luxturna™. The reimbursement of new and expensive drugs in Ireland is not a straight forward process. It can be complex and difficult at times. We anticipate a response from the HSE towards the end of the year.

Given the rarity, there are just a small number of people in Ireland who may be eligible for Luxturna™. However, this process paves the way for future therapies that have the potential to treat a wider group of people living with retinal disease in Ireland.

To inform our submission, Fighting Blindness are consulting with the community to gather information about the life long impact of living with an inherited retinal degeneration and the impact a new medicine might have on quality of life.

The survey is open to people living with retinitis pigmentosa (RP), Leber congenital amaurosis (LCA), including their families and carers.

The closing date for completion is **Monday, June 29, 2020.** Take the survey here.

For further information, you can contact research@fightingblindness.ie.

Impact of Covid-19 on people living with sight loss

In May we launched a survey to find out how Covid-19 is impacting people living with sight loss in Ireland.

We shared the survey with our email subscribers, on our website and across our social media channels. We also made a small investment in social media advertising to reach as many people as possible.

We wanted to capture a snapshot of the impact and give people an opportunity to have their voice heard.

The results will help us campaign on behalf of people living with sight loss and strengthen the case for research and support funding.

We had a total of 126 respondents. We would like to thank everyone who took the time to complete the survey. We appreciate both your time and willingness to share your experience with us. Your involvement and engagement in our work is crucial, there is strength in numbers.

The survey closed on Wednesday 10 June and we will release the findings in the coming weeks.

We're the its4women.ie charity of choice!

We're delighted to tell you that insurance company its4women.ie has chosen Fighting Blindness as one of its charities of choice.

The three year partnership will provide not only financial support to our organisation but also lots of new publicity as their share our campaigns across their social media channels.

The its4women.ie team are taking part in our Steps for Sight challenge this June and they're fundraising to support people living with sight loss in Ireland.

Speaking about the challenge, Gary McClarty, Chief Executive of MCL InsureTech, the parent company of its4women.ie, said: "We chose Fighting Blindness as one of our nominated charities earlier this year and we have been supporting the team in their efforts to save sight and change lives.

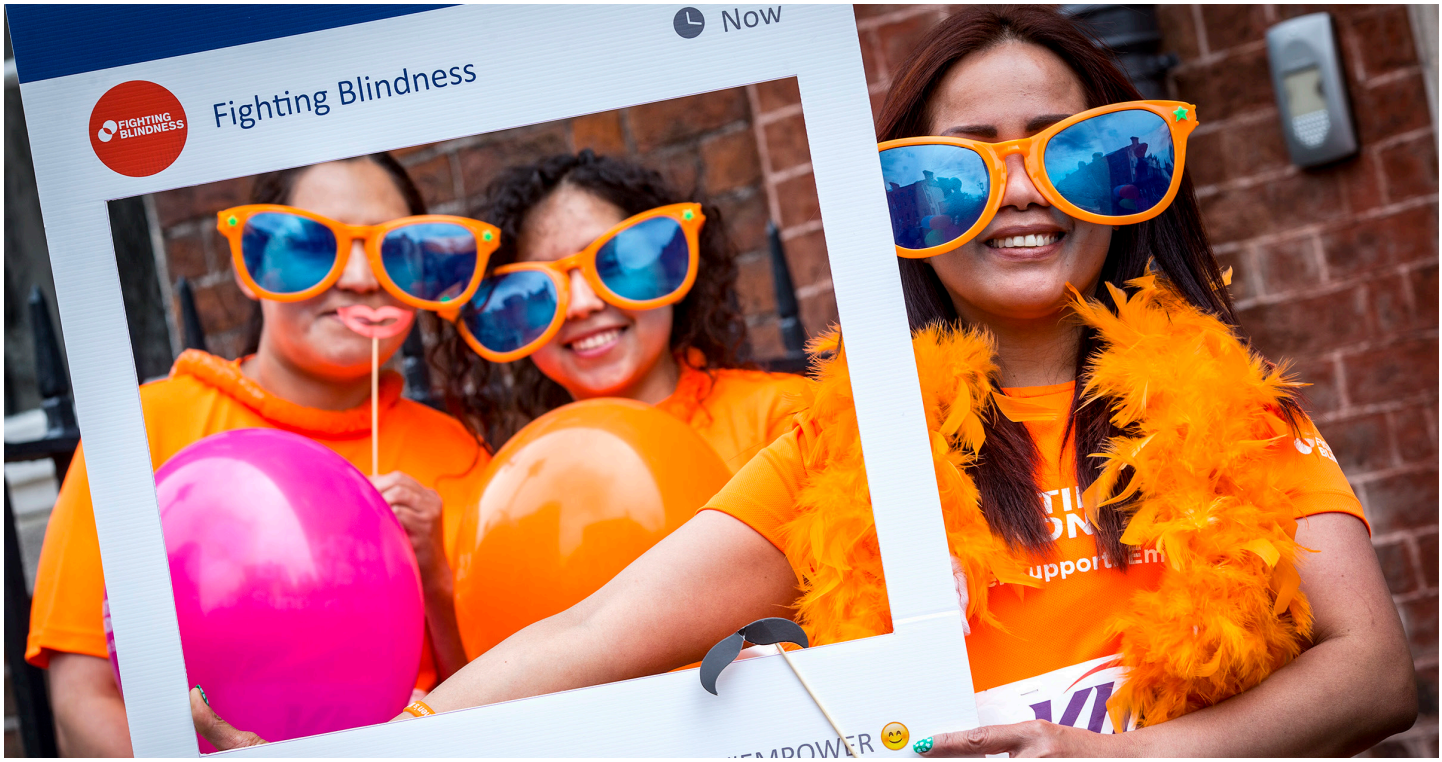
Since the outbreak of the Covid-19 pandemic, we have become acutely aware that the charity sector is facing huge challenges as campaigns have been called off due to distancing protocols.

"Fighting Blindness is doing amazing work to support those living with blindness but also invests in life changing research and we want to ensure they can continue that work during what is a very stressful time for people living with sight loss.

"Our team is looking forward to working with Fighting Blindness over the coming years to help them achieve their goals of supporting and empowering people with sight loss."

We look forward to working in partnership with its4women.ie to delivering outcomes that change lives for the better. Find out more about Steps for Sight on the back page.

We need your help to keep fundraising virtually!



We could not do what we do without you, our fundraising heroes! As many events have been cancelled due to Covid-19 restrictions, virtual events have become an incredibly popular way of fundraising.

Here's a list of fun ideas to inspire you! As with any event, you can make your fundraiser more powerful by setting up your own page on JustGiving, Everyday Hero or Facebook.

Zoom quizzes
Video conference calls have become the new normal when it comes to staying in touch with friends and family. Platforms such as Zoom are perfect for hosting pub quizzes from home. Ask participants to donate a small amount to your fundraising page in exchange for the link to take part.

Virtual fitness
Now more than ever we use devices and apps to track our fitness, why not challenge yourself to run, jog or walk and raise funds online by asking friends, family and co-workers to sponsor you.

Virtual birthday parties
Birthday fundraisers are incredibly popular across social media, why not take it to the next level and host a virtual birthday party on Zoom.

You can invite all your friends to make a donation to your Facebook birthday fundraiser in place of gifts and come up with some fun virtual games and activities.

If you have a question about running your own virtual fundraiser, please contact Rachel at fundraising@fightingblindness.ie.

Update on our 2020 events calendar

Like other charities and organisations across the country, we have revised our calendar of events in light of Covid-19. The decision to postpone or amend events was not taken lightly and the safety of our members, supporters, community and team is our number one priority.

Vision 2020

Vision 2020 is the 13th International Conference by the International Society for Low Vision Research and Rehabilitation. The conference was due to be held this July but will be postponed until Sunday 11 – Thursday 15 July 2021. Visit www.vision2020dublin.com

Vision Walk 2020

Vision Walk is a four day walking festival due to take place this October in Marbella, Spain. The event has been postponed to October 2021. To find our more and sign up for 2021, please email fundraising@fightingblindness.ie.

Paris2Nice 2020

The organiser of the charity cycle Paris2Nice recently took the difficult decision to postpone the event until 2021. If you've recently rediscovered a love for cycling during Covid-19 you now have lots of time to train for the six day cycle through France!

Retina 2020

This year we will run our Retina conference as an online event this October. We're disappointed that we can't meet in person but we know that the online version will be just as engaging and informative. We will announce dates and speakers over the coming weeks.

STEPS 4SIGHT

FIGHTING BLINDNESS Empowered by its4women.ie

Take part in Steps for Sight this June!

You still have time to join our Steps for Sight virtual challenge! We're calling people of all ages and abilities to get moving in June and support people living with sight loss in Ireland.

You can walk, jog or run a half marathon (21km) or a full marathon (42km) over the month of June.

Entries cost €20 and you'll receive an e-bib, certificate of completion and an e-medal. You could be in with a chance to win some brilliant prizes too. Everyone who takes part will be entered into a raffle to win a €100 One4all voucher and the entrants with the fastest times in both challenges will win a €50 One4all voucher.

Don't forget to set up a Facebook fundraiser or a JustGiving page and share with your friends, as the top fundraiser will win a hamper full of goodies!

To register visit Steps4sight.com. If you have any questions, please email Rachel at fundraising@fightingblindness.ie.

Proudly empowered by its4women.ie

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