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**FIGHT  
FOR  
SIGHT**  
EYE RESEARCH

# news summer 08

News for Fight for Sight supporters – Edition 05

## World's first gene therapy trial results

**Thanks to Fight for Sight supporters, a landmark in eye research was made recently when researchers from the UCL Institute of Ophthalmology and Moorfields Eye Hospital announced results from the world's first clinical trial to test gene therapy treatment.**

Doctors have not only proven that the experimental treatment is safe, but that it has also improved the sight of one of the three patients involved. Fight for Sight is delighted to have helped fund this important research.

### **Gene therapy can improve sight**

The initial year-long trial involved injecting healthy



copies of the missing RPE65 gene into the retina of patients with Leber's congenital amaurosis (LCA), a form of inherited retinal disease that results in poor night vision from an early

Following the treatment, the three patients involved underwent a series of tests. They all achieved levels of vision at least equivalent to before the operation, but one patient, Steven Howarth, 18, pictured, benefited from significantly improved night vision.

age and gradual sight loss as the retina deteriorates over time. There are currently no effective treatments available for LCA.

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(continued from page 1)

Professor Robin Ali led the research team, which included eye surgeon, Mr James Bainbridge and retinal specialist, Professor Tony Moore.

### “A significant milestone”

Professor Ali commented on the findings: *“Showing for the first time that gene therapy can work in patients with eye disease is a very significant milestone. This trial establishes proof of principle of gene therapy for inherited retinal disease and paves the way for the development of gene therapy approaches for a broad range of eye disorders.”*

He added: *“These results give us great confidence that this technique is safe and can bring real benefit to patients with impaired vision. While we’re very excited about the results it’s important to emphasise that gene therapy is still an experimental treatment not yet generally available to patients. The technique will be tested in other patients with LCA and we also hope to begin trials for other forms of retinal disease in the future.”*

Thanks to your ongoing support, Fight for Sight continues to help fund the team to further the clinical trials.

## New drug to cure blindness

**Age-related macular degeneration (AMD) and diabetic retinopathy are the leading causes of visual impairment in developed countries. AMD affects 25 – 30 million people worldwide<sup>1</sup> whilst diabetic retinopathy is one of the fastest growing diseases in developed countries, now accounting for 5% of world blindness<sup>2</sup>.**

Fight for Sight awarded a 3-year project grant of £140,000 in 2006 to Professor David Bates from the University of Bristol to pursue his research into abnormal blood vessel growth. Professor Bates and his team have identified a naturally occurring form of vascular endothelial growth factor (VEGF), which prevents the formation of new blood vessels in the retina. The exciting news is that a drug is now being developed for testing in clinical trials to treat wet AMD, due to start at the end of the year.

### Understanding abnormal blood vessel growth

The charity awarded a clinical fellowship grant of £136,000 over 2 years in 2006 to Dr Andrew Scott, below, from the UCL Institute of Ophthalmology. Dr Scott is also researching the causes of abnormal blood vessel growth in the retina.

Dr Scott explained: *“Abnormal blood vessel growth in the retina is a response to a lack of oxygen reaching the retina. The new vessels that grow are faulty and tend to leak causing complications. We’ve discovered, however, that the retina can grow healthy blood vessels in the absence of a certain immune cell in the body. My work involves understanding this process and the role of immune cells in new blood vessel growth in the retina.”*

The findings from Dr Scott’s research will help doctors to better understand retinal diseases and will contribute to the development of new treatments.

<sup>1</sup> AMD Alliance <sup>2</sup> WHO



## Transplanting retinal cells to restore sight

Inherited retinal disease and age-related macular degeneration (AMD) are major causes of sight loss in the UK caused by the deterioration of photoreceptor cells. Fight for Sight is funding several projects into finding new treatments for AMD. One of these projects is described below.

### Regenerating photoreceptor cells

The exciting news for patients is that scientists now know how to transplant neural stem cells that develop into new photoreceptor cells. Although this work is in its early stages, the therapy looks promising and Fight for Sight is delighted to be supporting research in this field.

The charity awarded a 3-year project grant of £140,000 starting in July 2007 to Dr Jane Sowden from the UCL Institute of Child Health and Professor Robin Ali from the UCL Institute of Ophthalmology, pictured above. The team is researching methods for developing new photoreceptor cells for transplantation and recruited Dr Jorn Lakowski to join the project last year.

### Replacing diseased cells

Although the diseased retina is normally unable to replace the lost photoreceptors, the team has discovered that they can be regenerated



from immature cells transplanted into the retina.

Dr Sowden explained: *“The new work we are doing will provide an important step towards the development of stem cell transplantation therapy for treating retinal disease.”*

She added: *“So far we have isolated stem cells from the eye and are now working on identifying conditions that encourage these cells to produce new photoreceptors. Once we are confident that we can develop enough stem cells to the right stage for optimal generation of new photoreceptors then our aim is to transplant them to repair the degenerating retina.”*

## New techniques for diagnosing retinal diseases

Fight for Sight awarded a 3-year project grant of £140,000 in 2006 to Professor Andrew Stockman from the UCL Institute of Ophthalmology. Professor Stockman is developing new techniques for understanding and diagnosing retinal dysfunction, as well as for the evaluation of specific treatments.

### Understanding photoreceptors

Professor Stockman explained: *“We are measuring the reaction of the photoreceptor cells to flickering lights amongst patients with specific genetic deficits in their photoreceptor cells. We’ll be comparing these results with patients with no known genetic deficits to gain*

*insights not only into the nature of the various dysfunctions, but also into the normal workings of the rod and cone photoreceptors.”*

### Improving treatment

*“We are also active in assessing the effectiveness of potential and novel disease treatments, including the exciting new gene therapy treatments of patients with Leber’s congenital amaurosis,”* added Stockman.

This research will improve doctors’ abilities to make definitive diagnoses in patients with retinal dysfunction, improving the counseling given to patients in the clinic.

## Gene research into retinitis pigmentosa



**Retinitis pigmentosa (RP) is a group of progressive genetic eye conditions in which abnormalities of the photoreceptors or the retinal pigment epithelium (RPE) lead to gradual loss of sight. Around 18,000 people in the UK are affected by the disease.**

Understanding the genetic causes of RP has been the focus for much of the research into the disease. Scientists have now identified more than 100 mutations in the gene for rhodopsin, a substance in the retina that is responsible for seeing in dim light conditions. These mutations account for 15% of all types of retinal degeneration. So, whilst progress has been made there is still much to do to fully understand the disease.

### Mutations in rhodopsin

Professor Michael Cheetham, pictured, was awarded a 3-year PhD Studentship

of £66,000 in 2005 and appointed Maria Kosmaoglou to study why mutations in rhodopsin cause blindness and how this may be overcome.

Professor Cheetham explained: *“Cells have special proteins called molecular chaperones that monitor the correct development of substances such as rhodopsin. We have identified a chaperone that increases the removal of faulty rhodopsin. The more this chaperone is added the more faulty rhodopsin is disposed of. This suggests that in the future we will be able to use chaperones to prevent photoreceptors dying because of faulty rhodopsin.”*

*“We’re now trying to find other chaperones, which might be used to correct faulty rhodopsin. Once we’ve identified the key ones then they will become targets for future therapies,”*

### Second grant awarded

Fight for Sight awarded a second grant to Professor Cheetham to pursue his research into RP. In 2006 he received a 3-year project grant of £140,000 to study mutations in the gene RP2, which lead to a severe form of X-linked RP found primarily in males.

Professor Cheetham said: *“We are researching the function of gene RP2 and its role in photoreceptor function. This understanding will give a greater insight into how the retina functions and how we may repair any defects caused by the faulty gene.”*

### Improving treatment for RP patients

Professor Cheetham’s research will be used to offer improved diagnosis and genetic counseling for patients and may also lead to the development of new treatments for RP.

He said: *“We hope to see the impact of our research on patients within the next 5-10 years.”*

## New tests for optic nerve disease

Optic nerve disease is second only to cataract as a cause of worldwide blindness and early diagnosis is vital to minimise sight loss. Good vision depends as much on the image-forming mechanisms of the eye as on the health of the optic nerves, which carry the signals from the eye to the brain.

### Improving the way patients are diagnosed

Current testing methods for optic nerve disease measure the patient's visual field. These tests are often unreliable and sensitivity is poor. Dr Mithu Storoni, pictured, from the National Hospital for Neurology and Neurosurgery, London, received a 3-year clinical fellowship of £157,000 from Fight for Sight in 2007 to develop more effective testing methods based on pupillometry.

Dr Storoni explained: *"We are designing a new test that relies on measuring the pupil's reaction to a light. We will compare the results of this new test with current test results.*



*This is an important project that could change the way patients with optic nerve disease are diagnosed and managed."*

She added: *"Our new test will be painless, more*

*comfortable, and much faster than current tests. If our research is successful, we could start seeing our results implemented in clinics within just a few years."*

## Friends rally round to give Jessica the gift of sight



**Three-year old Jessica Levinson, pictured, is a mischievous, little bundle of life like most other toddlers. There is one big difference though;**

**Jessica has been diagnosed with the incurable genetic condition, Leber's congenital amaurosis (LCA), which results in severe visual impairment from birth.**

### Hope for Jessica

The good news is that major progress has been made recently in the field of gene therapy to treat LCA (see front page for more information). This groundbreaking new treatment offers real

hope to children like Jessica who are born with inherited retinal disease.

### More research is still needed

It is early days yet and there is still much more work to be done before gene therapy becomes widely available. Janine Baird and Stacey Triplow, family friends of Jessica, are determined to help this little girl to see by raising money for more vital research.

Janine and Stacey from Ashington, Northumberland, organised a charity disco and raffle on 7 June 2008. Janine said: *"Jessica is a wonderful, bright, loving, little girl. We want to do as much as possible to enable her to see. Organising this event is our way of helping."*

**To contribute towards Janine and Stacey's campaign or for more information on organising your own fundraising event contact Annu Mayor on 020 7929 7755 or email [annu@fightforsight.org.uk](mailto:annu@fightforsight.org.uk)**

## 'Tommy's Tucker' Recipe Book



Eight-year old Tommy Salisbury's mother, Emma Salisbury, and his grandmother, Dot Grindley, have embarked on a new fundraising venture with the launch of 'Tommy's Tucker', a book of over 100 favourite recipes from friends and family.

### Funding for choroideremia research

All proceeds from the book will go to Fight for Sight to fund research into choroideremia, a progressive genetic eye disorder that affects Tommy. Emma and Dot have raised more than £93,000 so far for choroideremia research through coffee mornings, family discos and golf days.

Emma said: *"We're really excited about 'Tommy's Tucker'. Friends and family have been helping us to pull it together and we hope it will raise more money for Fight for Sight."*

### Order your copy today

Tommy's Tucker costs just £3 including postage and packaging. For more information email [tommystucker@googlemail.com](mailto:tommystucker@googlemail.com) or call Emma on 07984 790 285.

## Golfers hit a 'hole in one' for eye research

Members of the men's section at Blacknest Golf Club in Blacknest, Hampshire, were pitching and putting throughout 2007 in aid of Fight for Sight, their nominated charity of the year.

Dr Bob Andrews, Men's Captain, is pictured below presenting a cheque for £1,700 to the charity's Chief Executive, Michèle Acton, on 16 March 2008.

He commented: *"Sight is so important and especially for golfers – it would be devastating to lose it. There is a real need for more eye research and the more money we can raise for Fight for Sight, the sooner scientists can find new treatments for blindness."*

### Charity of the year

If you would like to nominate Fight for Sight as your charity of the year on behalf of your golf club, school, employer or other local organisation, contact Annu Mayor on 020 7929 7755 or email [annu@fightforsight.org.uk](mailto:annu@fightforsight.org.uk)

This is a great way to raise the charity's profile and more money for eye research.



## Mother races to save son's sight



Helen Gittings, above left, ran the Reading Half Marathon on 2 March 2008 to raise funds for Fight for Sight. Helen's 15-year old

son, Matthew, is awaiting test results that are likely to confirm he has retinoschisis, an X-linked genetic eye disorder, that can result in deterioration and, or, loss of sight.

Helen and her fellow runner, Jodie Sanders, have raised over £2,500 so far for the charity, which includes a £1,000 donation from Goodman UK, their employer.

Mr Jeff Pulsford, CEO of Goodman UK, said: *"We were very pleased to support Helen and Jodie in the Reading half marathon. Knowing that Helen's son, Matt, may have a genetic eye disorder and being able to support such a worthwhile charity as Fight for Sight was a pleasure."*

## A tribute

**Three long-term supporters of Fight for Sight sadly died earlier this year. Our condolences are with the family and friends of Mrs Pat Caro, Lady Prideaux and Mr John Winstanley.**

### Mrs Patricia Mildred Mary Caro

Mrs Pat Caro was born in London but grew up with her father in Antwerp and Paris and later travelled widely throughout Europe with her second husband, Mr Tony Caro.

Pat held a number of different jobs during the Second World War including working at Cornbury Manor and for the BBC French Service. She later came to play an important role in Fight for Sight's Wiltshire Committee after her two younger children developed Stargardt's macular dystrophy. She worked tirelessly as a

volunteer for the charity for over 20 years, helping to raise thousands of pounds for eye research.

Pat died on 29 March 2008 after a courageous struggle with cancer. She is survived by her four children, ten grandchildren and six great-grandchildren.

### Lady Prideaux

Lady Prideaux, a Vice-President of The Iris Fund for the Prevention of Blindness, passed away in January 2008 aged 96 years. After the charity merged with Fight for Sight she continued her support for eye research as a Patron of the new charity.

Lady Prideaux was married to the late Sir John Prideaux OBE who was President of The Iris Fund, Treasurer of St Thomas' Hospital and subsequently Chairman of

the Special Trustees of St Thomas' Hospital. Their son, Christopher, was Chairman of The Iris Fund.

### Mr John Winstanley

Mr John Winstanley MC TD FRCOphth, distinguished soldier who fought at Dunkirk, El Alamein, Kohima and Burma, and later, leading ophthalmic surgeon at St Thomas' Hospital, died in January 2008 aged 88.

Mr Winstanley helped to establish St Thomas' as one of the country's premier ophthalmic teaching and research establishments. He was a long-term supporter and Vice President of The Iris Fund (later the British Eye Research Foundation) and Patron of Fight for Sight.

# 2008 Events Diary

We're looking for people to take part in forthcoming events and races. To get involved contact Rowena Finn on 020 7929 7755 or email [events@fightforsight.org.uk](mailto:events@fightforsight.org.uk)

July	05	Mike Brooke begins his sail around GB for LCA research
July	02	Annual Golf Day, Warwickshire Committee, Stoneleigh Deer Park
	06	British 10K, London
	12	Emma Salisbury's coffee morning for choroideremia research, London
August	08	Big Band Evening, Bath Committee, Midford
	09-10	The Mazda London Triathlon, London
September	05	Fight for Sight Annual Clay Pigeon Shoot, Newbury, Berks
	09	Hydro Active Women's Challenge, locations around the country
October	02	Annual Autumn Fair, Wiltshire Committee, Frome, Somerset
	05	'Run to the Beat' Half Marathon, London
	12	Royal Parks Half Marathon, London

## Congratulations to our Marathon runners



17 supporters completed the 2008 Flora London Marathon for Fight for Sight on 13 April 2008. Congratulations and thank you all for your great efforts

An incredible £32,000 has been raised so far, all of which will be used to fund more research into blindness and eye disease.

Mr Fred Moore, pictured second from right, ran the Marathon in three hours and 57 minutes and said: "It was an exhilarating experience thanks to the incredible support from friends, family and crowds lining the route. And there was the knowledge that I had raised money for Fight for Sight, which does so much in the important field of eye research. It is an experience that I will never forget."

### Thank you to all our runners below:

Akin Adenubi	Sonya McCullough
Matt Boyles	Arthur Montgomery
Chris Cooper	Fred Moore
Tony Fryer	Benjamin Reid
Anthony Haines	Kate Taylor
Andrew Hill	Rachel Thompson
Samantha Jerrom	Manish Vaid
Pete Khoo	Sam Wilkinson
Jan Knight	

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Royal Patron: HRH The Duke of York KG President: The Right Hon Baroness Boothroyd OM PC

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