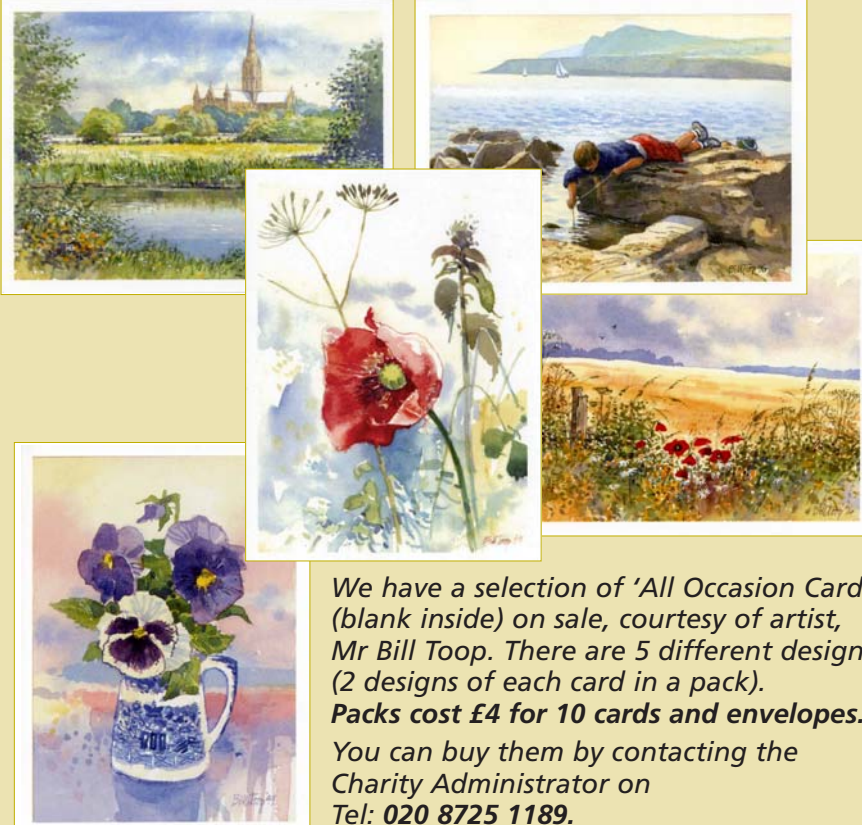


## All Occasion Cards



We have a selection of 'All Occasion Cards' (blank inside) on sale, courtesy of artist, Mr Bill Toop. There are 5 different designs, (2 designs of each card in a pack). Packs cost £4 for 10 cards and envelopes. You can buy them by contacting the Charity Administrator on Tel: 020 8725 1189.

## IBOX PHOTOGRAPHY



Ibox7 Photography is raising money for the Trust by selling digital images which can be used as desktop backgrounds, loaded onto digital picture frames or printed to hang on the wall.

For further information visit their website:

[www.photo.iboxsolutions.com](http://www.photo.iboxsolutions.com) or visit our shop on the Marfan Trust website [www.marfantrust.org](http://www.marfantrust.org)

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# MarfanTrust News

Autumn/Winter 2010

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

[www.marfantrust.org](http://www.marfantrust.org)

Registered Charity No: 328070

## Research News

### Sonlee Laboratory

Dr Gavin Arno, assisted by Dr José Aragon-Martin has been busy finishing the analysis of a group of patients attending the Marfan syndrome screening clinic who do not appear to have mutations in the Marfan gene fibrillin-1.

Briefly, we have now screened 14 patients who previously did not appear to have a mutation in the fibrillin-1 gene.

We evaluated a newly developed procedure to study the mRNA of the fibrillin-1 gene to look for possible mutations not visible by studying the whole gene at the DNA level. We have not identified any new mutations using this technique and we would not recommend its use in the future.

A more useful technique that can be added to the standard fibrillin-1 gene screen is Multiplex Ligation-dependent Probe Amplification (MLPA) which detects large gene deletions that

may go undetected by screening the 65 exons of the fibrillin-1 gene. This method has shown greater success and is now offered by a number of laboratories throughout the UK. These results are being prepared for publication.

### New Papers

Our recent study of the causes of ectopia lentis in patients with no FBN1 mutation has reported the importance of a recessively inherited gene called ADAMTSL4. First described in a Turkish family, it has been found in our families from Sweden, Asia, Afghanistan and Pakistan, as well as English families.

Aragon-Martin, J. A., Ahnood, D., Charteris, D. G., Saggar, A., Nischal, K. K., Comeglio, P., Chandra, A., Child, A. H. and Arno, G. (2010), Role of ADAMTSL4 mutations in FBN1 mutation-negative ectopia lentis patients. Human Mutation, 31: E1622-E1631. doi: 10.1002/humu.21305

### Medical Student Activities

Mr Nelson Kamali was awarded the Brian Adams memorial studentship for 2010 to extract DNA from frozen blood samples of Marfan syndrome patients and



Dr Gavin Arno on left Postdoctoral Supervisor of Sonlee Laboratory discussing DNA analysis of Marfan gene with Summer Project Medical Students Nelson Kamali and Kate Hughes.



PCR machine with commemorative plaque used for DNA preparation kindly donated by the Finkletaub Family in memory of Daniel, former Treasurer of the Marfan Trust.

### DANIEL FINKLETAUB MEMORIAL ESSAY PRIZE

The first annual Daniel Finkletaub Memorial essay competition for undergraduate medical students throughout the UK was launched in September. The essays can be on any aspects of Marfan syndrome. **The prize is £250.**

Daniel was Treasurer of the Trust for over 8 years and his family are sponsoring the competition

## 2010 Raffle Prize Winners

Thank you to everyone who bought tickets for our Summer Draw which raised £1,147 for the Trust.

The winners of the first 3 main prizes were:

1st Prize	£100 cash	Mrs Valerie Coughlin
2nd Prize	£50 cash	Mr Roy Williams
3rd Prize	£25 cash	Mrs J Brambleby

Winners of the other prizes were:

Marfan Trust Bear	– Mr M Hemsley
Gift Token (donated by Andy Sole)	– Ms A Bennett
MP3 Player	– Mrs T Greene
Anais Anais eau de Toilette	– Mr A Gash
Crystal Bottle Stopper	– A Warnes
Book – 'Take Chelsea Home'	– Mr R Towne

### New Website

We have redesigned our website which now has more information about Marfan syndrome, diagnosis and living with the condition.

The website also gives details of our current and future research projects and all the Trust's leaflets and booklets are now available electronically.

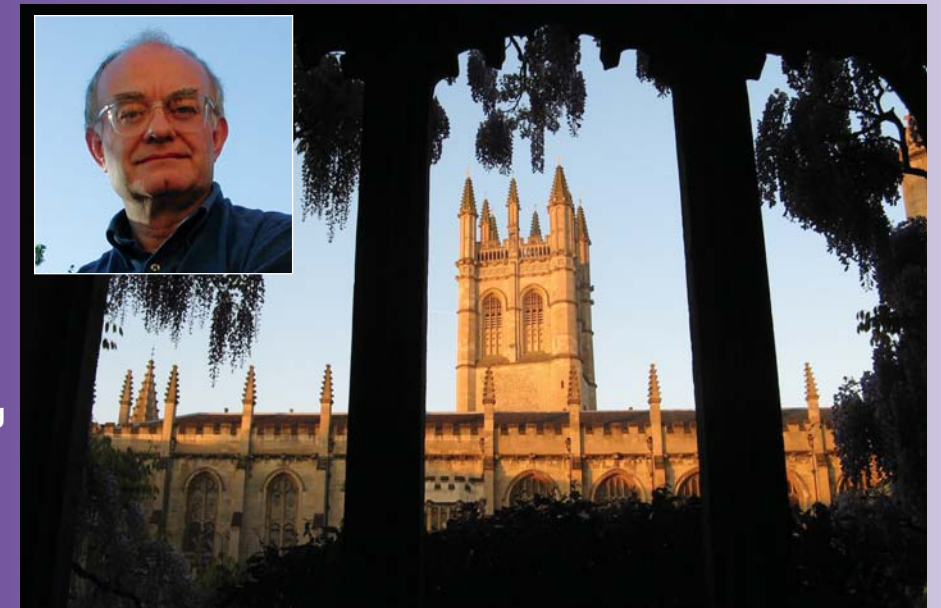
[www.marfantrust.org](http://www.marfantrust.org)

## John Rutter Singing Day at Oxford

John Rutter, famous choral composer, is holding a Singing Day at the historic Magdalen College in Oxford on Saturday, 16 October 2010, in aid of the Marfan Trust.

So if you like singing, come along to what will be a great event. Tickets £15. Contact the Charity Administrator on 020 8725 1189 or visit the Trust's website:

[www.marfantrust.org](http://www.marfantrust.org)



their families, now available for future research projects. DNA gradually deteriorates with time, but once extracted, is preserved for many more years. Also, practically speaking, the amount of freezer space saved is considerable. Nelson has also contributed to a future publication on genetic causes of non-Marfan adolescent scoliosis.

**Miss Kate Hughes**, recipient of the second summer scholarship, won a prize for the best student molecular genetic project for her essay 'Genetic causes of Ectopia Lentis'. This work also won her co-authorship of a published paper. She will voluntarily return in November to do a short project on "Genotype-Phenotype Correlation in Ectopia Lentis" providing material for a second publication on this topic.

### **Eighth International Marfan Syndrome and Related Disorders Symposium**

Dr Anne Child attended the 8th International Research Symposium on Marfan syndrome September 11th -14th in Warrenton, Virginia just outside Washington, USA. Held every few

years, this meeting of the original 8 groups who together found the gene for Marfan syndrome in 1991, attracted scientists from all over the world.

Dr Child presented work of the Sonalee Laboratory in 2 sessions, one on the causes of ectopia lentis and the second, a co-presentation of the AIMS trial (with Dr John Dean, Consultant Geneticist, Aberdeen). A panel discussion about aims, protocols, problems, solutions, and possible meta-analysis of all medication trials underway internationally followed, and helped plan the collaborative analysis of results from these trials in which patients of each nation are eagerly participating. Panel members represented trials in USA, UK, France, Belgium, Italy, Netherlands and Taiwan.

### **AIMS Medication Trial**

Final approvals to proceed with this national UK 21 centre medication trial will be available by November 2010, and the first patients invited to join the trial at their routine Marfan clinic visits, from early 2011 onwards for a 2 year period. For further information see

[www.rbht.nhs.uk/aims](http://www.rbht.nhs.uk/aims).

### **Publications**

Dr Anatoli Kiotsekoglou has written 2 new papers recently:-

#### **"Effect of aortic stiffness on left ventricular long-axis systolic function in adults with Marfan syndrome"**

Anatoli Kiotsekoglou, Samir K Saha, James C Moggridge, Venediktos Kapetanakis, Bart H Bijnens, Michael J Mülle, John Camm, George R Sutherland, Ian B Wilkinson and Anne H. Child\* Hellenic Journal of Cardiology.

#### **"Assessment of aortic stiffness in Marfan syndrome using two-dimensional and Doppler echocardiography"**

Anatoli Kiotsekoglou, James C Moggridge, Samir K Saha, Venediktos Kapetanakis, Malini Govindan, Francisco Alpendurada, Michael J Mullen, John Camm, George R Sutherland, Bart H Bijnens and Anne H Child. Journal of Echocardiography.

This material will also form part of her thesis.

### **Freemasons Grand Charity**

The Trust has recently been awarded a grant of **£2,500** from the The Freemasons Grand Charity, which we are very grateful for, especially in this tough economic climate.

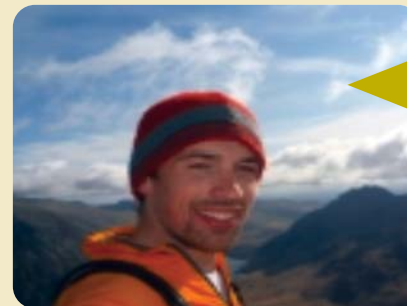
# Fundraising



### **Darrell Whittle**

#### **Pennine Cycleway Challenge**

Darrell completed the 344 mile Pennine Cycleway Challenge in July and has raised **£2,043** for the Trust. Darrell's cousin died from Marfan syndrome in his 30s. He was undiagnosed and left a wife and two children who both have the condition.



### **David Buglar**

#### **Paddy Buckley Round (60 miles, 47 Mountains in 24 hours)**

David took part in this gruelling event, which covered most of Snowdonia in Wales. He has raised **£1,695** for the Trust. David took part in this event in memory of his sister-in-law, Victoria Pantling, who had Marfan syndrome.



### **Jonathan Best**

#### **Sky Dive**

Jonathan did a Sky Dive in August and raised **£1,294** for the Trust. A close friend of his died suddenly last year at the age of 27 from Marfan syndrome.



### **Philip Coles**

#### **44 Mile Hike**

Philip is taking part in a 44 mile hike from Inkpen Beacon in Berkshire to Eling Tide Mill in Hampshire, the weekend of the 6-7 November and is hoping to raise **£1,200**.

Philip's brother Russell died suddenly at the age of 27 from Marfan syndrome. The family were not aware that he had this condition which had remained undiagnosed by doctors.

You can sponsor Philip by visiting his page on Virgin Money Giving, <http://uk.virginmoneygiving.com/philipcoles-marfantrust>.

Darrell, David, Jonathan and Philip used the Virgin Money Giving website to raise funds for the Marfan Trust. Between them, they have raised £4,758 for the Trust.

### **Other Fundraisers**

Rosemary Peters with the help of Alan Gadd and friends cooked a meal for guests and raised £846 for the Trust.

### **St Teresa Catholic Primary School, Stoke-on-Trent**

During Lent children from this school participated in various sponsored activities and very kindly donated £100 to the Marfan Trust from funds they raised.

We wish to express our sincere thanks and gratitude to all the above people for their fundraising efforts. The money they have raised will help to improve the life of Marfan syndrome sufferers throughout the UK.

Virgin Money Giving

<http://uk.virginmoneygiving.com/philipcoles-marfantrust>

Now on sale

## Marfan Trust Christmas Cards



We have 5 new designs. Cards cost £4 for a pack of 10 cards.

If you would like to order any cards, please complete the enclosed order form and return it to the Charity Administrator in the pre-paid envelope. Alternatively, you can order Christmas Cards via our website, [www.marfantrust.org](http://www.marfantrust.org)

Please support the work of the Trust by buying our Christmas cards

## Join us on FACEBOOK

The Marfan Trust is now on FACEBOOK. We would encourage all our members and supporters to join our group. Please share this page with your friends and family so we can all help create support and increase awareness of Marfan syndrome. Visit us:

[www.facebook.com/pages/Marfan-Trust/132589376774336](http://www.facebook.com/pages/Marfan-Trust/132589376774336)

## Virgin Money Giving

The Trust has opened an account with Virgin Money Giving for anyone taking part in sponsored events and wanting to raise money for the Trust.

Using the internet is becoming the most effective form of fundraising. It's easy to set up an internet sponsorship form which can then be e-mailed to friends and even to those that live on the other side of the world!



[virginmoneygiving.com](http://virginmoneygiving.com)