

Research Report

It has been another highly productive quarter for the Marfan Trust research programme. PhD Student Anatoli Kiotsekoglou has had a further paper accepted for publication (*The unravelling of primary myocardial impairment in Marfan syndrome by modern echocardiography* by Anatoli Kiotsekoglou, George R Sutherland, James C Moggridge, Dariush K Nassiri, John Camm and Anne H Child: Heart, published online 17 Feb 2009).

Anatoli is visiting an echocardiography unit in Sweden to improve her skills, which will be useful in further research on the topic of 'How does Marfan syndrome affect the heart in childhood and adulthood'.

Two medical students were selected to receive the Brian Adams memorial studentships for 2008 and 2009. Miss Dana Ahnood has presented a poster on her topic "The role of fibrillin-1 mutations in ectopia lentis (dislocated lenses)" at St George's Research Day. This is being written as a scientific publication. Interestingly, she has discovered that although we find 92% of fibrillin-1 mutations in patients with classical Marfan syndrome, in patients with dislocated lenses as their presenting feature, fibrillin-1 mutations are only

found in 60% of cases, suggesting that a second and possibly other genes can cause ectopia lentis without producing the heart problems found in Marfan syndrome.

This work is being conducted in conjunction with Mr David Charteris, Consultant Ophthalmologist, Moorfields Eye Hospital.

Miss Sultana Hasso has performed TGFBR2 gene screening. This gene, which acts on the same pathway as fibrillin-1, has been shown to cause ascending aortic aneurysms. Her topic was "What is the incidence of TGFBR2 mutations in ascending aortic aneurysm patients who do not have a fibrillin-1 mutation?" She has established an incidence of 1 in 20. This correlates well with

the larger study performed by the Paris Marfan syndrome database group. These patients have different diagnostic signs (wide set eyes, cleft palate, associated congenital structural heart disease such as septal defect, tortuous widened arteries.) Since this test is expensive, we will recommend it first in patients who have these signs, and will recommend fibrillin-1 screening tests for patients who appear to have Marfan syndrome features. This very practical project will guide our clinical assessment and mutation analysis of patients who come to our aneurysm screening clinic. Sultana's work will be incorporated into a larger paper addressing this problem to be written later in the year.

Boxing Day Dip 2008

The Lions Club of Sunderland kindly donated £480 from their annual Boxing Day Dip. Lucy Gash, (Liam Gash's sister) took part in this event.



Liam Gash inspires a charity show



Lucy Gash (middle in blue) and friends with two members of the band, The Futureheads. The Futureheads headlined a gig in Sunderland to raise awareness of Marfan syndrome that claimed the life of 22-year-old Liam Gash whilst backpacking on a trip to Australia. Since Liam's death in 2006, family and friends have raised more than £51,000 for Marfan charities in the hope that the disease, which was undiagnosed in Liam, will be more easily detected. The Futureheads joined other Sunderland bands at a charity event, entitled Three Night Stand, to raise money for the cause.

In Memory

Donations received in memory of Jane Brogan, Julia Palmer, Liam Gash, Daniel Finkletaub, Dr Martin Briggs and Victoria Pantling.

Daniel Finkletaub

It is with sad news that the Trust was informed of the sad loss of Daniel Finkletaub. Daniel was Treasurer of the Trust for over 8 years and we were very grateful for his straightforward approach and advice in running the affairs of the Trust during this time.

BBC RADIO 4

Charity Appeal

The appeal that was broadcast in October last year raised £5,500 for the Trust, but importantly, more people have now heard of Marfan syndrome.

SGUL ref: 00978

Marfan Trust News

Spring/Summer 2009

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

www.marfantrust.org

Registered Charity No: 328070



Participants attending the Marfan Meeting

On Saturday 25th October 2008 my husband Mike and I hosted an informal Marfan meeting at the Dragonfly pub in Newport. We started at midday and had a whole end of the pub to ourselves, which was ideal.

The people attending were mostly members of the Marfan Friends World forum- which can be found at www.marfan-friends-world.org.uk We had people attend from all over the UK as well as friends from Dublin and even all the way from the USA! In total we had just under 30 people, which was a lovely turn out. Veterans of previous meetings were very impressed with this!

The day stayed wonderfully informal, with people chatting comfortably throughout the day. This was my first Marfan meeting and I was quite nervous, but everyone was genuinely lovely and

we've made some great new friends. Mike and I enjoyed selling raffle tickets, as well as the T-shirts that the Trust had kindly donated to us, and I drew the raffle tickets ably assisted by all the lovely children that were there.



Ellie Bull

www.marfan-friends-world.org.uk

Raffle!

Enclosed with the Newsletter you will find raffle tickets. Please support the work of the Trust by buying raffle tickets in order that our vital research can continue.

If you would like additional tickets, please contact the Charity Administrator: Tel 020 8725 1189 or email hdydyk@sgul.ac.uk Please return tickets and unsold books in the pre-paid envelope by 24th June 2009.

1st prize £100 cash, 2nd prize £50 cash, 3rd prize £25 cash plus 5 x Marks & Spencer £10 gift cards.

We carried on right until last orders, making it an 11 hour meeting for some! It was a lovely day though, of great interest and support all round and it wasn't over, as the following morning, just about everyone came from their hotel to our house for a little while to say goodbye, and to meet our pet Dalek!

In total we've raised £145 for the Marfan Trust, which we were both surprised and delighted by- thank you to everyone!

Ellie Bull

Student programme with

University of Connecticut

Dr José Aragon-Martin, who has successfully completed his PhD studies this year in a collaborative project regarding Glaucoma genes, is assisting Dr Gavin Arno in the Sonalee Laboratory, studying messenger RNA abnormalities in Marfan syndrome. He is also assessing the possible role of the second gene reported for ectopia lentis (ADAMSTL1-4) in a Jordanian family. José's work will combine with Dana Ahnood's work to produce a published paper.



As a token of our appreciation of your continued support, enclosed with the newsletter is a complimentary Marfan Trust pin badge.

Further badges are available at a cost of £1 per badge.

Silent Auction

We have many exciting and unique prizes to choose from. Why not take a look! Thank you to all those who generously donated items to the auction.

It's really easy to take part.

If you are interested in bidding for any of these items, complete the enclosed form and return in the envelope provided.

All bids must be received by 31 May 2009.

Marfan Trust Teddy Bear
Reserve Price £20



Reserve Price £150



Lily Cole's Designer Jeans

Pair of low rise jeans donated and signed by the model Lily Cole. (Size: 32 inch waist, 36 inch leg) £150

Reserve Price £150



Unframed professional photograph (Size A3) of Alhambra in Spain



Wallace & Gromit

A3 signed original drawing of Wallace & Gromit by Nick Park the creator

Reserve Price £1000

Week at a UK Holiday Home Bond

1 week for 2 people at a holiday flat at locations in the UK and the Republic of Ireland. Dates available subject to availability. Reserve Price £350

Tie-Dyed Silk Scarves
in various colours (27" square or 70 cms square)
Reserve Price £15 each



2 Tickets to Kew Gardens London + Souvenir Guide
Reserve Price £40

Odeon Cinema Tickets Reserve Price £50

Round of golf with Les Tippin, Chairman Marfan Trust in the Bromley area Reserve Price £50

Apple Nano Ipad (2) Reserve Price £150 each

2GB MP3 Player Reserve Price £25

Goodwood Races 2 passes to Gordon Enclosure at Goodwood Races valid from May-October (not available Wednesday or Saturday of the Festival Meeting in July) Reserve Price £50

Professional Tooth Whitening Treatment
Dental Practice based in Cobham, Surrey
Reserve Price £400



Holiday flat in Switzerland

Location: Vercorin, Val d'Anniviers, Canton Valais. Fully furnished and equipped. Sleeps 4. The offer comprises accommodation for 2 weeks (Saturday to Saturday). The successful bidder must organise his/her own transport. 2 hour drive from Geneva airport.

Reserve Price £350

Available dates 2009

September, October and November

Available dates 2010

9 January to 6 February 2010, and 6 - 20 March 2010 and the months of May and June 2010.

Tie-Dyed Silk Stole
(72 ins x 19 ins or 184 cms x 4 cms)
Reserve Price £30



Holiday Bungalow on the Isle of Arran

Week in a Bungalow at Whiting Bay on the Isle of Arran with wonderful sea views. Sleeps 7. Date to be agreed subject to availability.

Reserve Price £500



2 hours professional tennis coaching in either Buckinghamshire or south west London Reserve Price £20

Man's Milano 100% Silk tie in pink Reserve Price £30

Belle Fleur elegant boxed scented candle from Space NK London Reserve Price £12

Champagne picnic for 4 from Peni Black Caterers - south west London area Reserve Price £100

An Italian supper or lunch for 6 catered in your own home (south west London area only) Reserve Price £150

A guided walk followed by a picnic in Happy Valley, South Coulsdon, Surrey (10 places only) Reserve Price £15 per person

"Hands on or hands off: the nature and process of economic policy in Hong Kong" signed by Anthony Latter Reserve Price £15

"Dear Fatty" By Dawn French Reserve Price £10

Marie Claire Seasonal Kitchen Cookbook Reserve Price £20

Barnabas Children's Bible Reserve Price £15

Tractor stencil book Reserve Price £5

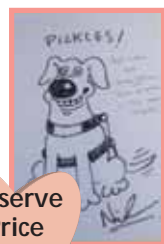
Reserve Price £150



Framed A3 professional photograph of The Black Musician

Wallace & Gromit

A4 photocopy of Pickles the Guide Dog from Creature Comforts Nick Park creator, signed by the voiceover artist Sarah Williams



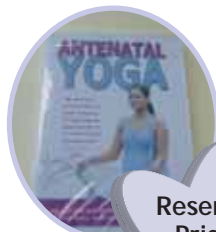
Reserve Price £25

Reserve Price £20



Crystal Golf Ball Wine Stopper containing laser image of golfer

Antenatal Yoga DVD
Signed copy of Relaxation and Exercise in Pregnancy Tricia Robinson



Reserve Price £10

Cosmetic Dentistry
Professional cosmetic dentistry by a Dental Practice based in Cobham, Surrey
Reserve Price £1,000

Gentleman's Made To Measure Shirt
By Couch & Hoskin Ltd, Bespoke Tailors, London
Reserve Price £150

£25 John Lewis Gift Voucher Reserve Price £25

£26 Debenhams Gift Card Reserve Price £26

2 x £10 Book tokens Reserve Price £10 each

£20 Marks & Spencer Gift Card Reserve Price £20

£15 Theatre Token Reserve Price £15

Anais Anais Eau de Toilette Reserve Price £20

Toy puppets, cow, reindeer, octopus Reserve Price £10 each

Fisher Price laugh and learn puppy dog Reserve Price £22

Set of Fisher Price tub time tugboat bath toys Reserve Price £12



Tour of Downe House, home of Charles Darwin

Throughout 2009, the life and work of Charles Darwin is being celebrated to coincide with what would have been his 200th birthday year. Visit Downe House and discover the man and the revolutionary theory that changed our understanding of the world.

Guided tour with Dr Child (Saturday or Sunday) with refreshments (Maximum 4 in group). Nearest Train station Bromley South or parking on site.

Reserve Price £12 per person