

Marfan Trust News

autumn / winter 2007

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

www.marfantrust.org

Registered Charity No: 328070



Les Vince with his wife Jenni, at Buckingham Palace after collecting his MBE

MBE for Les Vince

On 23 May 2007, Les Vince accompanied by his wife Jenni, received his MBE for services to the Marfan Trust from Her Majesty the Queen at Buckingham Palace.

Les is thrilled to have been awarded an MBE and to have met the Queen is probably his greatest achievement.

Message from the Chairman



Les Tippin, Chairman

The Trust is delighted that our most prolific fundraiser Les Vince received his MBE. His dedication in raising substantial amounts of money on a regular basis (£3,500 so far this year) is very much appreciated and enables the Trust to continually fund the various research projects. Although Les received this most prestigious award I would like to mention that he did not do this alone and is always supported by his wife Jenni, who supports Les 100% on his various fundraising events.

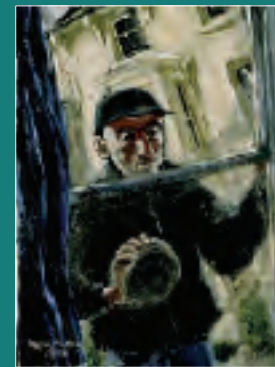
On a slightly sadder note, our chief research analyst Dr Paolo Comeglio has decided to return to Italy with his young family and continue his research work in his native country. Paolo has been in the Trust's laboratory for 7 years and his experience and dedication will be

missed. However, we are now assisted by Gavin Arno, who has been working alongside Paolo for the past 3 years, ensuring all knowledge and techniques have been transferred to Gavin. We wish Gavin a successful and challenging time in the Marfan laboratory.

The Trust over the past 6 years has been analysing and perfecting the technique of finding the gene mutations to establish if the patient has Marfan syndrome or not. In consultation with the NHS, they have now agreed to take over this service. This allows our team to concentrate on our research projects, now that we believe the technique identifies 92% of those patients who have the condition.

The Trust believes that the years of funding resources supporting this area, has taken the understanding of the condition to a satisfactory first conclusion and one the Trust can be very proud of. We won't rest on our laurels and are continually funding other research areas connected to Marfan syndrome as indicated in the newsletter.

Stephen Maddams



The Trust would like to thank the family of Stephen Maddams, an artist who died from Marfan syndrome

in 2002, for profits donated from an exhibition of his work in April of this year.

Most of Stephen's paintings were based on him watching people going about their daily jobs.



If you would like to have a look at some of Stephen's work, his website is: www.stephen-maddams.org.uk

In Memory

Donations received in memory of:

Eileen May Dyche,
Florence Virginia Fitz-Earle
and Margaret Bayne

research

Neonatal Marfan Syndrome: How Does It Differ from Paediatric Marfan syndrome?

This study by St George's medical student Mr Benedict Scoones, aimed to review infants with neonatal Marfan syndrome diagnosed in the Sonalee laboratory at St George's, University of London. The laboratory is supported by the Marfan Trust. These infants were compared to case reports in the literature, and paediatric cases (under the age of 5) also diagnosed in the Sonalee laboratory. The aim was to draw a clear picture of what constitutes neonatal Marfan syndrome.

Generally, neonatal Marfan syndrome is a diagnosis made at or shortly after birth, since the child is severely affected in the eyes, heart and skeleton. By definition, because of the severity of the condition in these infants, they are affected due to new mutations in the fibrillin-1 gene, often confined to the neonatal region of exons 24 to 32. It often results in death by 2 years of age, despite medical attention. Diagnostic signs include crumpled ears and loose skin as well as a wizened or aged appearance especially of the face. Infants can have joint contractures

and emphysema (obstructive lung disease). The cardiac problems can include aortic dilation but more commonly, valve insufficiencies (most commonly the mitral valve) are the main health problem.

This condition must be differentiated from Beals syndrome, Isolated Ectopia Lentis (dislocated lenses), Homocystinuria and Shprintzen-Goldberg syndrome.

A review of all 193 mutations found in the laboratory revealed 15 in children tested under the age of 5, due to clinical suspicion of Marfan syndrome.

Of our 6 most severely affected children diagnosed in the newborn period, all suffered from arachnodactyly, joint contractures and mitral insufficiency. Five of the six had the typical aged facial appearance described in neonatal Marfan syndrome. Four of the six had ectopia lentis. Five of the 6 died by age 4, 2 dying at age 4: both had had previous valvular surgery without which they would not have reached the age of 4. In the other 9 cases the most

common features were joint laxity and ectopia lentis, in addition to typical cardiac features.

Literature Review

25 cases of neonatal Marfan syndrome in the literature have been reviewed and compared to our cases. This reinforces the idea that if a newborn has a mutation in the neonatal region of the gene, together with joint contractures, typical facies and mitral valve insufficiency, this does confirm the diagnosis and also the prognosis. Surgery may provide hope for some cases, but all too often it is a high risk situation. Because of the sporadic nature of the neonatal form, children will continue to be born with this condition. Using these guidelines however, physicians can be guided to a more accurate diagnosis and therefore prognosis of the child in their care, which helps in counselling the family.



Reference

Elcioglu NH, Akalin F, Elcioglu M, Comeglio P & Child AH: Neonatal Marfan syndrome caused by an exon 25 mutation of the fibrillin-1 gene. Genet Couns. 2004; 15(2):219-25.

Irritable Bowel Syndrome in Marfan Syndrome

We are grateful to those patients who completed a postal questionnaire for St George's University of London medical student Benedict Scoones. Comparison to community controls has shown that there are significantly higher rates of abdominal pain and bowel disturbances in Marfan syndrome. Control patients were matched to Marfan syndrome patients by age and sex. This study was approved by our Research Ethics Committee. Of 217 patients, 118 returned questionnaires, a response rate of 54%.

Abdominal pain was significantly more common in Marfan syndrome patients than in controls (p less than 0.005). Men with Marfan syndrome were more likely to suffer from constipation than women with Marfan syndrome. Depressed patients were more

likely to suffer from Irritable Bowel Syndrome, and more likely to use laxatives. It is postulated that bowel symptoms are due to deficiency of fibrillin-1 (an essential component of elastin), which is an important fibre in the bowel wall. Further studies are required to determine the effect of bowel symptoms on patients' quality of life. It is also important to raise awareness of irritable bowel syndrome with doctors who care for Marfan syndrome patients, particularly cardiologists, gastroenterologists and primary care physicians.

Benedict Scoones has presented this original data to the British Society of Gastroenterologists where it was received with great interest. He is now preparing a leaflet for distribution.

Diagnostic testing

As a result of the Regional NHS laboratory taking on the diagnostic testing, this has left us with more time available to concentrate on our important research work.

Our thanks to the Greater London Genetic Testing Network and technician Louise Walton.

Hello to all.....

I am Trevor L. Bostock (Trev.) and am 49 years of age. I have Marfan syndrome which was not diagnosed until I was 23yrs old. I was told that I have many symptoms of Marfan syndrome. My older brother by 2 years and younger sister by 2 years do not have Marfan syndrome. They both have grown up families and their children are free of it. My parents and extended families do not have Marfan syndrome nor know of any affected ancestors. I have a spontaneous mutation; I prefer to be known as the unique one of the family.

I have double curvature of the spine and very small depressed upper body, and only 5 ft. tall. This means my lung function is restricted. I had my aortic heart valve replaced with a metal one. I have ticked away like a little bomb for the last 16 years. My eye sight is affected, having first dislocated lenses in both eyes which were removed, resulting in my wearing very thick spectacles. Later



Janet and Trevor

the retina in my right eye detached resulting in total blindness in that eye. I had just recovered from the shock and anger of being blind in one eye; when the left also detached. It took seven operations to restore my sight.

I have long gangly limbs, the joints in my hands, feet, shoulders and left hip are also troublesome. I have a high mouth palate with all the associated

problems with my teeth. Despite all my problems I have always tried to live a near normal life as possible.

Janet, my wife of 27 years is my greatest companion and friend. We took the decision not to have children. The risk of another Marfan syndrome victim was too much for us to consider. I realise having a family is a deeply personal choice between two people and they must follow their own feelings in such matters. I had to retire from work 10 years ago due to increasing medical problems. We live full and happy lives and I don't think now, that I have time to hold down a job. I did not get a fair deal in life; but then, life is not fair. I have learnt to live with the hand I was dealt. I have many accounts of this, but the greatest one is to keep a sense of humour and bright outlook on life. If any one would care to contact me about anything relating to Marfan syndrome, I would be most happy to hear from them.

Best wishes. Trevor Bostock

Trevor can be contacted by e-mail: t2jbostock@tiscali.co.uk

Prenatal diagnosis is now available. Contact achild@sgul.ac.uk

I was born with Marfan syndrome

which has affected my eyes, heart and joints. Though being visually impaired and having a love of sport, I play golf with the English Blind Golf Association and every year I compete in the British Blind Open and the British Blind Masters and I have just returned from Melbourne with my caddy, competing in the Australian Blind Open Golf Championships.



Sarah Williams and Ozzie

Every Thursday I ride at 'Riding for the Disabled' and I compete in the regional and national dressage championships of Great Britain on my horse, Winston. I am currently 6th in the country.

I love sailing and I sail at Chew Valley Lake disabled sailing club every Saturday. I also sail on the Tall Ships for the disabled and on yachts around the Solent with RYA Sailability Blind Week. Last year I obtained my competent crew yachting certificate.

I have Ozzie, a 5 year old guide dog and I am a School Liaison Officer for the Guide Dogs and I carry out three public speaking engagements each week at schools, rotary clubs and church groups etc. I do all the media for the Guide Dogs which includes many

radio and television interviews as well as writing numerous magazine articles, e.g. Woman's Own, Woman's Weekly and Health and Sports magazines. I also do the voice of 'Pickles' the guide dog in ITV's Creature Comforts for Aardman Animation. My friend, Nick Park the creator, interviewed me then he drew the character and made it out of plasticine. He then animated the character and it takes a day to do 3 seconds of filming Pickles. Pickles is the logo for the Guide Dog's organisation 'Shades for a Day' scheme, which is a big fundraising scheme organised by the charity.

I have spent most of my life working with underprivileged and blind children as well as orphans in Romania, refugees in Yugoslavia, street children in New York, homeless children in Peru and Nicaragua and a childrens hospital in Bolivia. I spent 7 years teaching Braille at blind schools in Australia, New Zealand and Kenya. In October I am going to work with Aids orphans in South Africa.

Through undertaking many challenges and opportunities which involve taking many risks and going out of my comfort zone, I have had the most wonderful, enjoyable and rewarding life imaginable.



Pickles

fundraising



Runners for MARFAN were: (L-R) Gloria Pantling, Ann Buglar, Mandy Pantling, Ian Pantling and David Bulgar

Pantling family road race

The family and friends of Victoria Pantling, who has Marfan syndrome, took part in the Great East Anglia Run 10k road race on Sunday, 6 May 2007 around the historic centre of King's Lynn.

Victoria was diagnosed with Marfan syndrome last year after being rushed into hospital with a near-ruptured aorta. Victoria and her family are now actively campaigning to raise awareness of the condition. £483 was raised by the family and their friends and Victoria's brother-in-law, Andrew Bartram took part in a 46 mile walk along the Pathfinder

Long Distance Footpath and raised £230 for the Trust.

A big thank you to all the family and friends for raising this money.

Any one undertaking a fundraising event for the Marfan Trust will be supplied with free T-shirts, sponsorship forms (if required) and literature on Marfan syndrome and the work of the Trust.

Contact the charity administrator on 020 8725 1189 or e-mail: hdydyk@sgul.ac.uk



THE GREAT BRITISH DUCK RACE

The winning Duck numbers are: 157791, 23300, 107146



The remaining 27 prize winners are:
23495, 8427,
32707, 5932,
39440, 88491,
81462, 51777,
46791, 19035,
85596, 108095,
91601, 73853,
57887, 26171,
119323, 121924,
107176, 78914,
82033, 27292,
66495, 48203,
53664, 37230,
90857, 55204.

In total 148 ducks were bought on behalf of the Marfan Trust, raising £285

A big thank you to all those who adopted a duck in the Great British Duck Race, which was held on the 2 September 2007.

Saving our planet

Everyday we are encouraged to reduce energy and waste, therefore if you have access to a computer then sign up to receive our newsletters electronically.

Not only will this help to reduce our posting and printing costs, it will also be a contribution to cutting down on waste.

If you decide to receive an electronic version of the newsletter, please let the Charity Administrator know by e-mailing, hdydyk@sgul.ac.uk.

Marfan Trust Christmas cards

are now on sale.

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

Not only will you be supporting research into the cause and possible prevention of Marfan Syndrome, you will also raise the profile of the condition for the majority of people who have never heard of it. 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.



★ Summer Draw

★ Thank you to everyone who bought tickets for our Summer Draw which raised £856 for the Trust.

The winners of the first 4 prizes were:

1st Prize	Mrs D Carter	£200
2nd Prize	Mr Williams	£50
3rd Prize	J Blackman	LCD Telephone
4th Prize	P Pinnock	£25

Other Prize Winners

6 piece kitchen knife set	Mr A Myers
19 piece kitchen knife set	Mrs V Coughlin
Pair of binoculars	Mr A J Sanderson
10 pc kitchen knife set	Mrs Chidlaw
Mini Digital Camera	J Sharratt
Rechargeable Spotlight	J Storr
Remote Control Car	Mr R McInerney
Calculator Pen	C Dando
Sim Card	Flair Listing

Sponsored walk in memory of Jane Brogan

Five work colleagues of Jane Brogan, who died from Marfan syndrome last December, took part in the Glasgow 10k race on the 24 May and raised £755 for the Trust.



Can you help?

If any of our readers work for a company or organisation that has a charitable committee, we would be very grateful if you would nominate the Trust to receive a donation.

The majority of corporate companies prefer employees to nominate a charity, rather than an unsolicited request direct from the Marfan Trust.