

Marfan Trust News

AUGUST 2004

91 Perryhall Road, Orpington, Kent BR6 0EF

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

www.marfantrust.org Registered Charity No: 328070



Welcome from our Chairman

It has been a long time since we reported on the progress of the charity, for which I apologise. All the Trustees are volunteers and with all our work commitments and our commitment in raising funds for further research into the Syndrome, I hope that you would agree the wait demonstrates progress on all aspects in maintaining the Trust in forming a solid foundation for the research to continue on a number of fronts as well as making the condition more widely known.

To start I would wish to emphasise the objective of the Trust, that is to support medical research into the condition so that more is known about it and its severity. This will enable medical staff to treat the patients in the short and long term. All money raised goes to the various research aspects of studying and informing patients and medical staff.

All Trustees give their time voluntarily for which I am proud and publicly thank them for all their time in support of the Trust.

During the last year George Mold retired as a Trustee. For many years he supported the Trust both in time and by donating many gifts that enabled us to use these as raffle prizes. George has been replaced by Dr Don Wijetunge. Until his retirement, Dr Wijetunge was a very experienced doctor in the Accident & Emergency Department at St George's Hospital. He still gives his time willingly, often by helping Third World Countries and undertaking medical operations to reduce suffering in those countries. In addition, Dr Caroline Carr from Moorfields Hospital joined at the same time where her medical research background has proved very beneficial.

As the Trust is growing in the number of projects it undertakes and the various activities in support of this, the Trust decided to employ a part time administrator based at St George's Hospital Medical School. I am pleased to report that after a vigorous selection campaign, Helga Dydyk, has been appointed as Administrator for the Trust and

started her new role in April. In addition to financing medical staff and students at the St George's Hospital Medical School in the research of this condition, the Trust partly funded (£40k; trust contribution) an analyser which will enable the diagnosis of the severity of the condition to be known in a much shorter timescale than previously undertaken.

Trustees

Mr Les Tippin (Chairman)

Mr Alan Savage

Dr Don Wijetunge

Mr Roger Hill

Dr Michael Carr

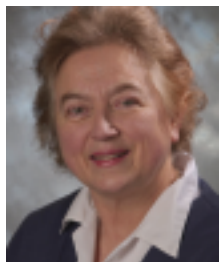
Dr Caroline Carr

Medical Director

Dr Anne Child MD

Marfan Trust
Supporting Research into Marfan Syndrome

Research Up-date



Dr Child reports progress in the various projects using the dHPLC Wave nucleic acid fragment analysis system, at the heart of the Marfan syndrome research programme.

With 2 laboratory staff working full time we have increased the number of mutations discovered from 90 to 123. A publication is being prepared. Each mutation confirms the diagnosis, and permits the NHS to offer a lifelong management programme to prevent sudden death. Screening for the mutation in close family members reassures those who are unaffected, and identifies those who need further care. Mutations have been reported to the up-

dated international database held on computer in Paris, thus increasing the International Gene Map.

In order to increase the understanding of this condition, some mutations of great interest have been referred to Professor Cay Kielty, University of Manchester, who will study why some produce eye disease, others heart disease and others neonatal Marfan syndrome, through functional studies of the fibrillin-1 produced.

In addition questionnaires regarding health problems in older patients (over 50) are still being analysed, and a publication is in preparation. Answers to these questions will guide our future research. How can we ensure the best health for patients in their later years?



One of our Team...

Dr Paolo Comeglio using the dHPLC WAVE analyser to identify mutations in the Marfan Syndrome gene for fibrillin-1.

SIR JOHN TAVENER

TAVENER AT 60

**Friday, 19 November 2004
Barbican Hall, London**

PROGRAMME

Tavener	Butterfly Dreams (London première)
Tavener	Schuon Hymnen (UK première)
Tavener	Supernatural Songs (London première)+
Tavener	The Lamb
Tavener	Birthday Sleep
Tavener	Pratrirúpa (world première)*

Susan Graham mezzo-soprano +
Ralf Gothóni piano *

Stephen Layton conductor
English Chamber Orchestra
Polyphony

To celebrate the 60th birthday of Sir John Tavener, one of the most successful living composers, the English Chamber Orchestra, Polyphony and guests perform a selection of Tavener's music.

The Trust is working closely with Sir John Tavener, who has Marfan Syndrome, on promoting his latest work, which coincides with his 60th birthday this year. Sir John has written a piano concerto, **Pratrirúpa**, which will be premiered at the Barbican Concert Hall in London on the 19 November 2004. We are hoping to be able to promote the work of the Trust and its funding requirements for the future at this exciting concert.

We have purchased a block of tickets for this prestigious event. The ticket prices are £20 and £25 each, which includes a donation of £5 towards the Trust.

If you would like to purchase tickets for the concert, please complete the enclosed form, together with the number of tickets you require and your payment, and return to the address on the form. **Ticket applications must be received before the 30 September 2004.** As we have a limited number of tickets available, they will be sold on a first come basis.



Les & Jenni Vince RAISE £20,000

Five years ago Les & Jenni Vince started raising money for the Marfan Trust. To-date they have raised an incredible £20,000 by holding Tombolas and Raffles in their home town of Grimsby. They normally hold 15 fundraising events a year and for 3 days at Christmas they have a stall at their local Tesco store and always dress up for the occasion. Last Christmas they dressed up as Snowmen!!

Les was diagnosed with Marfan Syndrome in 1978. When he was born in 1952, Marfan Syndrome had hardly been heard of. Up until the age of 13, Les was of fairly normal height and then suddenly he shot up from 5 ft to 6ft 6ins in 2 years. He carried on growing more slowly until he reached his present height of 6ft 10 ins.

He was the youngest of four boys. Sadly his mother died suddenly of a massive heart attack when she was 59 years old and his eldest brother, also died suddenly at the age of 43. It was then that the fatal connection with Marfan Syndrome was established.

Over the years, Les has had 41 operations, from a right hip replacement (his hip was finally replaced after 5 attempts), to the retina in his right eye becoming detached

(it took 6 operations before doctors managed to save his sight). Unfortunately he was not so lucky when the same thing happened to his left eye. Attempts to repair the left eye have failed, leaving him with very little vision in this eye. Les now has problems with early signs of glaucoma.

Les is still able to walk but he also relies on a wheelchair to get around. Even though he is less mobile, this has not deterred him from his fundraising work and he is determined to carry on raising money for the Trust and for the general public to be made more aware of Marfan Syndrome.

My fellow trustees and I are extremely grateful to Les and Jenni for the money they have raised so far and we wish them well with their future fundraising activities.

PUBLICATIONS

The Trust has produced a number of pamphlets for the medical profession, these are:



- Dental Aspects of Marfan Syndrome
- ENT Aspects of Marfan Syndrome
- Growing Older with Marfan Syndrome
- Musculoskeletal Problems with Marfan Syndrome
- Pregnancy in Marfan Syndrome
- Psychosocial Aspects of Marfan Syndrome

In addition, the British Heart Foundation has produced 2 books about the condition. Although these books were first published 3 years ago, they are still good background reading into the condition and of the work that has been undertaken to prolong the life of sufferers.



- **Marfan Syndrome**
Information for people with Marfan Syndrome, parents of children with Marfan Syndrome and interested health professionals



- **The Marfan Syndrome**
A Clinical Guide

If you would like a copy of any of the booklets, write to the Charity Administrator at the address below and consider sending a small donation to cover postage.

For further information about the work of the Trust please contact:

Helga Dydyk, Charity Administrator, Department of Cardiological Sciences
St George's Hospital Medical School, Cranmer Terrace, London SW17 ORE
Tel No: **020 8725 1189** Fax No: **020 8725 3416** e-mail: **hdidyk@sgms.ac.uk**

FUNDING REQUIREMENTS

As the Trust grows and takes on more research activities in fully understanding the condition and informing all the different medical organisations of its symptoms and preventative measures now available, we need to raise at least £90k per annum to maintain the team at St George's Hospital Medical School.

The Trustees are committed to raising this amount and any donation you may wish to make will be very much appreciated by the Trustees.

For the Future

The Trust wishes to form closer links with other medical research organisations undertaking research into Marfan Syndrome. This will allow a better understanding of the research being undertaken and the hope of improved knowledge of the condition and its prevention.

Acknowledgements

Over the past 8 years the Abbeydale Trust has sponsored the summer student post which has in the past undertaken vital analysis work from the various research projects and co-written a number of our pamphlets for the medical profession.

Fundraising

Lands End to John O'Groats Bike Ride

In August 2003 Becky Towler, Alex and Rory Riddel, cycled from Lands End to John O'Groats in 11 days raising £7,391 for the Trust. On behalf of the Trustees many thanks to Becky, Alex and Rory for the money raised.

2003 London Marathon

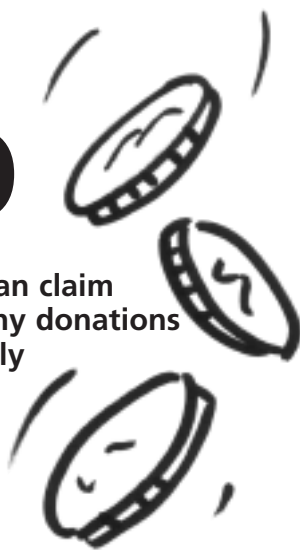
Roger Hill's daughter Harriett ran the London Marathon last year and gave £500 sponsorship that she raised for the Trust. Our sincere thanks to Harriett.

Best wishes to you all. Les Tippin, Chairman

GIFT AID

Remember, if you are a taxpayer we can claim money from the Inland Revenue for any donations made via the Gift Aid scheme. Currently for every £1 you donate to us the Inland Revenue gives us an additional 28 pence. To make a donation in this way you will need to complete a declaration form.

Gift Aid forms are available by telephoning **020 8725 1189** or visiting our website.



STOP PRESS

Marfan Trust Christmas Cards will be on sale later on in the year.

We do hope you will support the Trust by purchasing these cards.

Further details to follow.

www.marfantrust.org

Website Status



The website is now up and running and the address is;

www.marfantrust.org

The present information on the site is very basic and we intend to add to it as time permits but it is a vehicle for the future, which we intend to expand.