

Downing Street Petition

The Marfan Forum has created another e-petition to Downing Street relating to Physical Education in schools, as many members of the forum have unpleasant memories of PE whilst at school.

If you would like to support this petition, follow the link below.

THE DEADLINE FOR SIGNING IS THE 2ND OF JUNE

We the undersigned petition the Prime Minister to raise awareness of Marfan syndrome and other connective tissue disorders in relation to a child's physical ability within physical education.

The recognition of the possible non-ability to take part in PE within school, due to suffering from Marfan syndrome, Loeys-Dietz syndrome & Ehlers-Danlos syndrome.

Many children with sometimes life threatening conditions are still being made to take part in PE in some UK schools. This still happens even with written information from the child's medical expert. Physical education teachers need to be made aware that when a medical consultant says No to PE, this means No.

Many sufferers are in the possibly fatal position of being at the point of Aortic dissection or pneumothorax if they were to take part in contact sports or even pushed beyond their physical capabilities.

PE staff also need to be aware that there might be a serious reason behind a child's reluctance to take part perhaps due to Muscular Skeletal problems or lack of energy.

The link for the petition is

<http://petitions.number10.gov.uk/PE-and-Marfans/>

Leave a gift in your Will to the Marfan Trust

A gift in your will to the Marfan Trust will help us save lives, by planning vital research far into the future. Together we can bring better treatment and more effective methods of prevention to all those suffering from Marfan syndrome.

Further details can be obtained by requesting a legacy leaflet from the Charity administrator.

Tel: 0208 725 1189 or
e-mail hdydyk@sgul.ac.uk

Pennine Cycleway

Darrell Whittle is taking part in the Pennine Cycleway in July and raising money for the Marfan Trust.

The route is 355 miles and goes from Derby to Berwick-upon-Tweed taking in some of the most spectacular yet remote countryside in the country. If you would like to sponsor Darrell, you can contact him via his blog,



<http://middleagecyclist.blogspot.com>

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, or prefer **not** to receive tickets in the future, please contact the Charity Administrator on
Tel: **020 8725 1189** or
e-mail hdydyk@sgul.ac.uk

Please return tickets and unsold books in the pre-paid envelope by 25 June 2010.

RAFFLE PRIZES

1st Prize - £100 cash

2nd Prize - £50 cash

3rd Prize - £25 cash

PLUS Other Prizes

In Memory

Donations received
in memory of

Liam Gash,

Mark Wheatley,

Jane Brogan,

Mrs Nora Thompson,

Victoria Pantling and

Elizabeth Ann Hardy.

MarfanTrust News

Spring/Summer 2010

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

www.marfantrust.org

Registered Charity No: 328070

Research Update

AIMS Medication Trial

The Aortic Irbesartan in Marfan syndrome study is now in the final planning stage with 21 UK centres collaborating. An information website has been created at www.rbht.nhs.uk/aims to answer initial questions from patients and doctors, and will be updated regularly.

Patients aged 6-40 years, unoperated, should ask at their next annual echocardiogram visit if they wish to be included, as we will be recruiting volunteers for a 2 year period, starting September 2010. It will take until September for the drug company to manufacture our order of Irbesartan, and for it to be distributed to the hospital pharmacies at participating centres.

The list of participating centres will be on the website by September. A paper printout may be obtained from our Trust Administrator, Helga Dydyk, in September.

Does Marfan Syndrome affect the Heart Muscle itself?

Our Research Fellow, Dr Anatoli Kiotsekoglou, has worked with many patients over the past 4 years and shown through her very careful echocardiogram studies that both the left and right ventricles (pumping chambers) can be affected over many years and cardiologists and surgeons need to be aware of this, and treat early signs of heart failure vigorously.

Studies in children show minor changes even in the first year of life, so once a better medication is proven effective, Anatoli's work will help us decide how early preventive medication should be started.

Since our last newsletter, Dr Kiotsekoglou has had another paper accepted for publication called; **Biventricular and atrial diastolic function assessment using conventional echocardiography and tissue-Doppler imaging in adults with Marfan syndrome.**

We are grateful to all patients who helped in these studies.

Marfan Syndrome and the eye

A new gene has been discovered which causes dislocated lenses. It is named ADAMTSL4. It is inherited recessively, and both unaffected parents must pass on a gene error to the affected child.

In a study of 35 patients with dislocated lenses attending Dr Child's clinic (most referred by Mr David Charteris, Moorfields Eye Hospital) half had fibrillin-1 mutations (Marfan syndrome) but a surprising number (one-quarter) with isolated EL had ADAMTSL4 mutations, and had normal hearts which do not need follow up. It is important to distinguish these two conditions so correct genetic counseling and management may be offered.

Aragon-Martin J, Ahnood D, Charteris DG, Saggari A, Nischal KK, Comeglio P, Child AH, Arno G. Role of ADAMTSL4 Mutations in FBN-1 Mutation-Negative Ectopia Lenis Patients. Human Mutation 2010 (accepted for publication).

E-Newsletter

Every day 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

WHAT WE HAVE FUNDED

1991 – Discovery of the Gene

A research fellow was funded and made a major contribution to the discovery of the gene on chromosome 15 responsible for this disease.

1997 – Pregnancy Associated Risks with Marfan Syndrome

A research nurse, who worked on a study of the associated risks of pregnancy in Marfan syndrome women.

2000 – 2010 – Sonalee Laboratory

The Trust made a major contribution to the funding of staff and equipment for the dedicated Sonalee Laboratory at St George's University of London. The laboratory is equipped with an ABI sequencer which enables the research team to screen patients and their families for mutations (gene errors) in the Marfan gene. In each family where a mutation has been identified direct benefit has resulted. Whole families can be screened by DNA sent through the post to the Sonalee Laboratory. The doctors are able to identify babies at birth who are carrying the mutation. The best available treatment can then be offered throughout life. Over 150 mutations have been contributed to the Paris – held database of Marfan mutations, and international

publications.

2009 Aortic Irbesartan Marfan Study (Medication Trial)

The Marfan Trust has guaranteed funds from its resources to ensure success of this trial.

International Gene Map

Funding of students who helped to discover gene mutations in UK patients, which were contributed to the international gene map.

Information Pamphlets

The Trust has produced a number of information pamphlets regarding psychological, rheumatological, pregnancy, ENT, dental and ageing problems in Marfan syndrome.

Summer Medical Student

Each summer, the six week Brian Adams Memorial Studentship is offered to a medical student to undertake a focussed project on an important aspect of Marfan syndrome. Results are publicised and published, and a patient/doctor information pamphlet produced and distributed by the Marfan Trust to the specialists concerned. Thereafter they are made available to patients, upon contact with Dr Child at St George's University of London.

Tel No: 020 8725 5248

Les & Jenni Vince

After 10 years and raising £60,000 for the Marfan Trust, Les and Jenni Vince have decided to step back from their fundraising activities. They will now concentrate on raising awareness about Marfan syndrome.

The Trust is extremely grateful to Les and Jenni for all their hard work and dedication in raising this extraordinary sum of money for our research projects, which has helped to improve the life of Marfan syndrome sufferers throughout the UK.



Back row, left, Nigel Dunwell, Nick Carolan Store Manager, Tesco Cleethorpes, Jenni Vince, Marie Peck Customer Services, Tesco Cleethorpes. Front row: Les Vince.

Messiah Concert

The Trust sponsored a performance of the Messiah at St Luke's Church, Chelsea last December, as a celebration of 20 years of research into Marfan syndrome and as part of the Music for Marfan campaign.

The event was a success and we hope to do something similar in the future, to raise funds towards the medication trial.



Steven Isserlis Concert

Sunday, 6 June 2010 at 7.30 pm, Wigmore Hall, London

Steven Isserlis, cellist, will be performing 4 Sonatas by Barber, Chopin, Schumann and Dohnányi with piano accompaniment at Wigmore Hall, London on Sunday, 6 June 2010. We have 18 stall tickets available for this concert at a cost of £15 each plus a donation to the Marfan Trust. If you would like to purchase a ticket(s), please contact the Administrator on 020 8725 1189 or e-mail hdydyk@sgul.ac.uk



Make a Regular Donation

Make a difference today by donating £5 a month by standing order and help us to plan a better future for Marfan syndrome sufferers.


How money raised by the Trust is spent

- ✓ Salary funding in order that vital research studies can be carried out.
- ✓ Purchase of consumables required for the various research studies.
- ✓ Maintenance of essential equipment for the research projects.
- ✓ Printing of the Trust's newsletter and distribution to patients and donors.
- ✓ Printing of information pamphlets for patients and the medical profession.

Setting up a standing order is easy, forms are available on our website or by calling the Charity Administrator on 020 8725 1189. Alternatively, you can set up a Direct Debit payment with your bank. If you wish to set up a direct debit, call or e-mail the charity administrator for details of the Trust's bank account.

MARFAN Patient-Physician Forum

First Annual Meeting of the

Liverpool Heart and Chest Hospital 

Marfans Patient-Physician Forum

Saturday 5 June 2010

10.00 – 13.00 followed by lunch

- What are connective tissue diseases?
- What are my treatment options?
- Should I have my family screened?
- Patient experiences of surgery
- Question and answer session

The Liverpool Heart and Chest Hospital is holding a Patient-Physician Forum on **Saturday, 5 June 2010**. The Forum is open to all and is free of charge.

To register call **Carol Baker** on Tel: **0151 228 1616**
e-mail: carol.baker@lhch.nhs.uk

For families with Marfans Syndrome and other Connective Tissue Disorders

Register for Forum free by contacting Carol Baker on tel: **0151 2281616**
e-mail: carol.baker@lhch.nhs.uk
(Hosted by the Liverpool Heart and Chest Hospital Aneurysm Service)

Music for Marfan

Put Saturday 16 October in your diary. Famous choral composer John Rutter is conducting a "Come and Sing" Day at Magdalen College Oxford, 10.30 am – 3.30 pm. The audience is the choir. Music provided for old and new choral pieces by John Rutter and others, with a few surprises!

Tickets £15 plus donation. Bring your own lunch, or restaurants nearby.