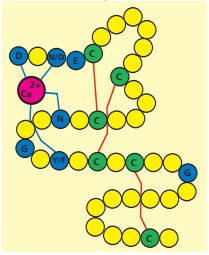


What We Have Funded

1991



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

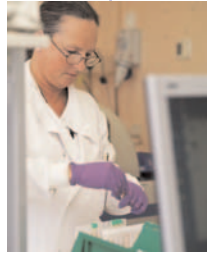


1997:

Pregnancy Associated Risks with Marfan Syndrome

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

1998



1998:

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 publicized and published, and a patient/doctor information pamphlet produced and distributed by the Marfan Trust to the specialists concerned.

2000



2000:

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 180 mutations have been contributed to the Paris – held database of Marfan mutations, and international publications.

2001



2001:

Information Pamphlets

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

2002



2002:

International Gene Map

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

Raffle

The Trust receives no government grants and relies entirely on donations and the generous help of our supporters.

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 on Tel No: 020 8725 1189 or e-mail hdydyk@sgul.ac.uk.

Please return tickets and unsold books in the pre-paid envelope by **2 June 2006**

1st Prize
£200 cash

Draw
The draw
takes place
7 June

Plus Other prizes

2nd Prize
Weekend voucher
for 2 at any
Hilton Hotel
in the UK
or Ireland

Plus Other
prizes

3rd Prize
£50.00

4th Prize
Wallace & Gromit
poster signed
by
Nick Park

Marfan Trust News

SPRING/SUMMER 2006

PATRONS: SIR MAGDI YACOUB FRCS, MR ANTHONY LATTER MA

www.marfantrust.org

Registered Charity No: 328070



Chairman's Report

The Newsletter this period looks at our core responsibility of funding Research projects to identify, categorise and support those patients who have been diagnosed with the Marfan condition.

The Trust was formed in 1991 when we helped fund the initial study into the gene. The year 2000 saw the creation and opening of the dedicated laboratory at St George's University of London. This facility is the heart of the Trust programme where, over the years, the detection of the gene has been refined to a level now that was not possible 5 years ago. The detection rates now achieved offer patients the possibility of early treatment in terms of medical advice/support that will ultimately assist in prolonging their lives.

Our additional work in keeping the medical profession informed through the issue of dedicated medical pamphlets is another important facet of our work.

In 2002 we contributed to the international Gene Map, which enables doctors/geneticists to diagnose the symptoms of Marfan syndrome in all its forms. Our involvement with this particular area of research, is in my view, of particular importance and interest. As the Trust has limited reserves I am extremely keen to collaborate with all Research Centres to ensure continuous exchanges of knowledge on the condition.

Unfortunately in 2005/6 we failed

to satisfy the National Lottery fund with our submission for funding. As this was the last time the lottery organisation would consider a medical research project it was a major disappointment for all those that worked so hard to put together a very creditable proposal. However, late in 2005 we were very pleased to receive financial support for two specific projects from 2 charitable organisations. One project is for the continuation of research into the ultrasound identification of the condition and the second is to undertake a study into the extra or missing heart beats which can cause sudden death in Marfan syndrome.

Our intention over the next period is also to support a 5 year drug trial due to start later this year. If the drug, which aims to strengthen the tissue, is proven to be successful, it will protect the aortic wall structure to help prevent aneurysm and dissection.

Over the past few years the Trust has worked hard to raise the profile of Marfan syndrome both within the medical profession and among those who are or may be affected. In doing so we have managed to secure larger

donations and thus grown our research base in a number of areas. We will continue with our endeavours to support the community of those affected and to enable a better informed medical community to ensure that all patients are well serviced in terms of best medical treatment for the various conditions that the Marfan syndrome exhibits. However, in order to maintain this level of research we must continue to call upon all of our resources to support the Trust. We are currently up-dating our website in a more user-friendly format, with information, advice and contact points. The new website will also allow donations to be made through a verified secure portal which will be simple to use and we hope will encourage all those who visit the site to make a donation.

We are continuing to promote the Trust through a range of activities and would welcome all those who would like to participate in support of our work. For those who took part in our activities during 2005 to raise funds, may I say a very big thank you and encourage you to support us again in 2006 and beyond.

Research Update

Tissue Doppler Echocardiography

Regarding tissue doppler echocardiography in Marfan syndrome our research fellow, Dr Anatoli Kiotsekoglou, funded by the Bluff Field Charitable Trust and the Henry Smith Foundation is using new ultrasound techniques to look at heart function in Marfan syndrome. Preliminary results from 20 patients are being carefully analysed. Altered left ventricular pumping function is noted. This may be a new measurement which can be used to assess whether medication is improving heart function or not in the future. **Further volunteers who have not had heart surgery are required, and anyone with Marfan syndrome interested in participating should contact Anatoli directly by telephone on (020 8725 3480),** or e-mail her at anatoli9@hotmail.com. The study involves a half-day visit to the Unit, with expenses paid.

The Role Of TGFBR 2 Gene In Familial Ascending Aortic Aneurysm

We know from our work in the Sonalee laboratory that we have achieved a high yield of mutations using our dHPLC Wave machine to analyse the Marfan syndrome gene. The yield at present is 92%, however 8% of the families refuse to yield their genetic secrets! These families have drawn attention to the fact that there may be other genes involved and the first such gene (TGFBR 2) has been identified by the Marfan research group in Paris, in a large French family. Other Marfan syndrome groups from around the world find that this gene explains approximately 1 in 50 families who have ascending aortic aneurysm, but not classical Marfan syndrome. TGFBR 2 interacts with fibrillin-1, the Marfan gene and so it all makes good sense that this causes aneurysm. We are at present seeking funding to analyse a group of forty families with ascending aortic aneurysm who have not demonstrated mutations in the Marfan gene. A pilot study of 10 such families reveals no TGFBR 2 mutation. We need to establish what contribution this new gene makes in the UK population.

Preimplantation Genetic Diagnosis

For families with an identified fibrillin-1 mutation, who want to plan to have unaffected children, the most acceptable method is through preimplantation genetic diagnoses (PGD). This is available through most primary care trusts (GPs) on genetic grounds. If the husband has Marfan syndrome, then pregnancy in a normal wife can proceed normally. However if it is the wife who has Marfan syndrome, her cardiac risk may increase in pregnancy, and has to be assessed carefully beforehand. We are at present trying to develop a programme of PGD for Marfan syndrome patients within the UK.

Interested couples should contact Dr Child directly at St George's, University of London, SW17 0RE (tel no: 020 8725 5248 or e-mail achild@sgul.ac.uk).



Last year our runners raised a marvellous £4,007. This was the first time that the Trust had applied for charity places at a major running event. A big thank you to all those who participated in this race for the Trust.

We have again secured places for this year's race which takes place in Portsmouth on Sunday, 22 October 2006.

If anyone is interested in running in this event and raising money for the Trust, **please contact the Charity Administrator, Helga Dydyk on tel no: 020 8725 1189 or e-mail hdydyk@sgul.ac.uk,** for further information. An entry fee of £27 is required for each runner.

Thank You...

Marfan Trust Christmas Cards

Thank you to all of you who bought Marfan Trust Christmas Cards last year. More cards will be on sale later in the year.

In Memory

Donation received in memory of Steven Mark Don.

A big thank you to Mark Kay and John Whennan from Loughton Fire Station (Red Watch) who ran independently in the Great Run South and raised £160 for the Trust.

Marfan Trust T Shirts



Support and raise the profile of the Trust by buying a Marfan Trust T shirt. They come in sizes, small, medium, large and extra large and are priced at £5 each.

To order your T shirt, call the Charity Administrator on 020 8725 1189 or e-mail hdydyk@sgul.ac.uk.