

# Gauchers NEWS

JULY 2008

Gauchers ASSOCIATION



Jo Bardoe, pictured centre with her daughters Mia, left, Skye, right and the Valentine's Ball Committee  
(see full story page 6)

# Lysosomal Storage Disorder Centres

## Addenbrooke's Hospital

Hills Road, Cambridge CB2 2QQ

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Fax: 01223 336 846

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## Birmingham Children's Hospital

Diana, Princess of Wales Children's Hospital

Steelhouse Lane, Birmingham B4 6NH

Head of Clinic: Dr Chris Hendricks and

Dr Anupam Chakrapani

Tel: 0121 333 9999

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## Great Ormond Street Hospital for Sick Children

Great Ormond Street

London WC1N 3JH

Head of Clinic: Dr Ashok Vellodi

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## Hope Hospital

Department of Lysosomal Storage Disorders

Hope Hospital

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Manchester M6 8HD

Head of Clinic: Dr Steve Waldeck

Tel. 0161 206 4365 / 1419 / 1080.

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## National Hospital, London

Charles Dent Metabolic Unit, Box 92

National Hospital for Neurology  
and Neurosurgery

Queen Square, London WC1N 3BG

Head of Clinic: Dr Phil Lee

Fax: 0207 2092146

## Royal Free Hospital

Pond Street, London NW3 2QG

Head of Clinic: Dr Atul Mehta

Tel: 020 7830 2814

Fax: 020 7830 2313

## Royal Manchester Children's Hospital

Willink Biochemical Genetics Unit

Hospital Road, Manchester M27 4HA

Head of Clinic: Dr Edmond Wraith

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Helpline: out of hours ring

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and ask for the metabolic consultant.



# Chairman's Forward

Dear Friends,

Welcome to the Summer 2008 edition of Gauchers News.

With this edition you will be receiving a special supplement that we are publishing on behalf of the European Gaucher Alliance of the proceedings of the European Working Group on Gaucher Disease which was held in Budapest on 4<sup>th</sup> to 7<sup>th</sup> June. This meeting was a huge success bringing together the world's leading clinicians, scientists, patient associations and pharmaceutical companies working in the field of Gaucher Disease. We heard presented advances in the clinical management of patients, details of scientific developments and information about the ongoing clinical trials for the four potential new therapies for Gaucher Disease. Of particular importance were the discussions on seeking solutions for unmet medical needs, promoting more scientific research and on encouraging young doctors and scientists into the field of Lysosomal Storage Disorders. Patient groups from 24 European countries (and South Africa) were represented at the Meeting of the European Gaucher Alliance which agreed to formally become constituted as a company limited by guarantee.

The launch of the Susan Lewis Memorial Fund was received with great interest and enthusiasm and, you will see the leaflet inviting applicants in the EWGGD supplement. We were also particularly pleased that Eleana Pavlova won second prize in the poster competition for her presentation on the bone study which members and friends have enabled through their generosity.

The next few months will also be busy for patients and the Association. Increasingly members will be invited to join clinical trials - the decision to participate will be very personal. The European Neuronopathic Gaucher Disease Conference in Northampton on 28<sup>th</sup> till 30<sup>th</sup> November next promises to be an extremely important event with a full programme, updating information and providing advice and support.

We are also working hard on our new website which I hope will be fully functional by the end of the year.

I extend special thanks to those who answered my call to support the Susan Lewis Memorial Fund and to become involved in our activities. The Executive Committee are always grateful for all the support and help provided by members. If you are able to help, do contact us.

With all good wishes.

Jeremy

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# Biomarkers of Avascular Necrosis in Adult Patients with Gaucher Disease - Development of Predictive Testing

*Prof Timothy Cox of the Department of Medicine, University of Cambridge at Addenbrooke's Hospital reports on the progress of the National bone study funded by the Gauchers Association:*

Dr. Elena Pavlova and Mrs. Jane Tindall were supported by the Gauchers Association with funds awarded nationally to the four national designated Gaucher centres working on Gaucher disease. Dr. Pavlova presented her work at the 8<sup>th</sup> meeting of the European Working Group on Gaucher Disease held in Budapest June 3-7 2008. Dr Pavlova's work was chosen for a presentation having been one of two papers presented by her and colleagues at the meeting; she was awarded the second prize at the meeting for the related poster presentation (a summary of Dr Pavlova's talk can be found in the EWGGD summary newsletter that accompanies this newsletter).

## **Scale of the problem**

Even in the era of effective enzyme, in Europe today, Gaucher disease is principally a disease of bones. These manifestations occur largely in those patients whose condition was established before enzyme therapy could be introduced – either because it was unavailable or because the diagnosis was not made in time.

As part of this project **Dr Patrick Deegan** in his clinical survey conducted with Jane Tindall and colleagues, surveyed 100 adult and 13 paediatric patients with Gaucher disease nationally at the four original participating centres.

This research was funded generously by the Association and was recently reported in outline at the Association 15<sup>th</sup> Anniversary Conference in January last year. Dr. Deegan set out the burden of disease in the UK and described the frequency of the modeling deformities, avascular necrosis (osteonecrosis or bone crises), the bone thinning (osteoporosis), and osteolysis, the regional disease in which bone is destroyed focally at sites of Gaucher tissue within the marrow cavity.

## **Bone Crises**

Of all these conditions, **avascular necrosis** appears to be the most disabling and, once established, the most difficult to treat. It frequently leads to destruction of big joints particularly the hips but also the knees and shoulders; it may also cause diffuse pain by affecting the difficult-to-visualise bones of the pelvis. Avascular necrosis is associated with severe episodes of pain at the time with no findings either on clinical examination by the doctors -or by conventional X-rays- in the first instance; only MRI and more sophisticated methods of diagnostic imaging can identify the affected bone during the crises. Careful doctors know that this is a diagnosis based on experience - and on simply believing the patient.

We know that avascular necrosis ('bone crises') occur particularly at critical points of growth and are common at puberty but they also occur at other times. **Dr Deegan** in an analysis of the survey data as part of this project has shown that they occur more frequently close to the time that a spleen was removed in the old days as a treatment for Gaucher disease.

The manifestations of disease in the bone reflect both the release of powerful enzymes from the lysosomes (as shown in part by **Dr. Mary Teresa Moran** – whose research was also funded by the Association) and cytokines studied by us and numerous other groups. These factors influence local events in bone but also have widespread effects on the skeleton; they are believed in some way to impede a critical supply of blood. Some believe that abnormal blood coagulation and abnormal capillary supply in the bone reflect a blocking of the blood supply with accompanying death of living bone tissue.



*Prof Timothy Cox*

## **The Need for Research into Biomarkers**

The frequency of bone crises appears to be markedly reduced by enzyme therapy. At the same time, existing biomarkers measured in the blood at visits to hospital (plasma chitotriosidase and CCL18/PARC – discovered jointly by Dr. Mary Teresa Moran with **Prof. Aerts's** group in Amsterdam) reflect the activity and bulk of Gaucher cells in other organs such as the spleen. We thus propose that these and other components that change in the blood might be able to predict the risk for occurrence of these complications in the bone. Certainly there is a longstanding association between severe bone disease in patients with Gaucher disease and the prior removal of the spleen before enzyme therapy was introduced.

## **Predictive biomarkers**

At present specialised biological "markers" are used in the clinic to follow the patients' responses to treatments. In fact currently licensed treatments for Gaucher disease (Cerezyme and Miglustat) reduce the amounts of the markers chitotriosidase and CCL18 in the plasma and are used by many centres to follow the course of the condition during treatment. However, at the

*(continued overleaf)*

present time clear evidence that abnormalities in these biomarkers, predict a particular complication for Gaucher disease is lacking.

Other candidate biomarkers which can be used to follow the course of the condition and possibly may have a role in predicting complications have been under consideration from the research carried out mostly by European investigators particularly those in the Netherlands and elsewhere. These cytokines or chemokines mediate inflammation by blood cells at sites where Gaucher disease is active. They can be measured and some of them have been shown to be elevated in untreated patients with Gaucher disease.

**We have then a problem in determining the clinical value of serum biomarkers:-**

- Does measurement of these biomarkers play any role in helping doctors distinguish between patients with and without the bone manifestations of Gaucher disease?
- We also ask whether or not of certain biomarkers be used to treat the treatment response?
- Most importantly, does the extent to which the biomarkers are elevated in the blood predict the *risk* of developing of bone complications?

These questions, though simple to ask are difficult to answer – particularly in the era in which most patients are receiving appropriate treatment. Thus any attempt to determine the value of biomarkers is hampered by the effects of pre-existing intervention. Given that this is not an ideal world and that we do not have a large bank of serum from patients before treatment, enabling us to measure the biomarkers in advance and then measure the outcome of the patients' illness over a long period of time subsequently, we are forced then to interpret and analyse changes in samples we already have.

**Details of the Project**

To get a handle on this problem, Dr Elena Pavlova catalogued the independent data collected by Dr

Deegan and Sister Tindall from the UK patients at the various centres. She also used a technique to measure numerous candidate and known biological markers of Gaucher disease in the patients' serum.

Our clinical database has been established on more than 100 patients – 100 adults and 13 children. This database has unbiased clinical information including the results of physical examination, radiological assessment of the bones and clinical laboratory data that were routinely recorded. At the same time, the research team collected samples of serum, plasma and urine from the 100 adult participants and Dr Pavlova analysed the potential biomarkers in these samples.

Here we report on the characteristics of 100 adult patients

- 60 female, 40 male
- Average age: 49 years
- Average age at presentation: 17 years
- Average age at diagnosis: 21 years.
- History of avascular necrosis: 43
- Spleen removed: 44
- Those with type 3 disease: 4
- Enzyme replacement therapy: 92
- Genotype: 13 out of 89 in whom genotype known were homozygous N370S.
- Average Zimran severity score: 10.5

Of this group of 100 adult patients, 84 were receiving enzyme replacement therapy without ongoing avascular necrosis; a minority receiving enzyme replacement therapy (8 patients) had ongoing episodes of avascular necrosis.

Of the 8 patients who were not receiving enzyme replacement therapy, none of these fortunately had developed avascular necrosis.

**Laboratory Studies**

The analytical methods used multiplex flow cytometric bead array assays to measure 15 cytokines simultaneously on a very small sample of serum; in addition Dr Pavlova developed a separate flow cytometric bead assay for CCL18/PARC which gave very good operating characteristics – and required some development.

**Results**

Several serum were found to be

elevated significantly in the patients with Gaucher disease compared with healthy control subjects serum biomarkers of several cytokines that were stable in stored serum and plasma. This included macrophage chemotactic protein 1 (MCP-1); macrophage inhibitory protein 1a and 1b (MIP-1b and MIP-1a); interleukin 8. Dr Pavlova found that those patients who gave a history of avascular necrosis that had been determined in an unbiased way before the analysis had a significantly elevated concentrations of both MIP-1b and IL8 as well as CCL18/PARC.

The study of the receiver operating characteristics for these cytokines showed indeed that they had some considerable specificity and sensitivity in detecting those patients with avascular necrosis. The best results were found with Interleukin 8 which at best had a highly significant performance with 78% specificity and 60% sensitivity for detecting avascular necrosis where a cut off value of greater than 25 picogram/ml was chosen. However, as they stand, these values would mean that about 30% of the patients would be mis-characterised.

We then asked the question: Are the biomarkers associated with the achievement of therapeutic goals – in this case avoidance of avascular necrosis?

Indeed Interleukin 8, CCL18/PARC, MIP1a and MIP1b were all significantly elevated in patients who had continued to develop avascular necrosis after receiving ERT compared with those who did not have avascular necrosis while receiving enzyme therapy We then further asked the question – could we distinguish between patients who had had avascular necrosis **after** enzyme replacement therapy, and those who **had no** avascular necrosis after enzyme therapy- compared with those who had never had avascular necrosis. In this case, the significance was seen strongly in favour of those who developed avascular necrosis, whose measured cytokine and chemokine concentrations were significantly higher.

We thus concluded that avascular necrosis of bone with a failure to meet a key therapeutic goal was associated with elevated serum cytokines including the one discovered by Dr

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Mary Teresa Moran in conjunction with Professor Aerts and colleagues in Amsterdam and now in current use as CCL18/PARC. MIP1a, MIP1b and Interleukin 8 were also correlated with failure to meet this therapeutic goal.

#### **Reservations and plans for the Development of this Work**

We are aware that there may be some flaws in the design of this study, it is a cross-sectional study and makes assumptions, however is the best available method to give a proper survey of bone disease in the UK as seen in our Gaucher patients.

What the study shows to our great surprise is a strong association between biomarkers, whose abundance correlates with Gaucher disease activity (and decrease with specific enzyme treatment), and occurrence of the disabling complication, avascular necrosis. In all the cases studied, the bone events occurred many months or years before the serum samples were collected.

#### **Future Extension of the Study**

We are now planning to set up

prospective studies to determine whether the serum proteins can truly serve as predictive biomarkers for this complication. This would allow us to establish target values of the biomarkers so that enzyme replacement therapy and other treatments can have standard limits which should be attained in order to reduce the risk of avascular necrosis.

The study then is very important in setting the ground rules for an important piece of work which would add further lustre to the development of therapeutic goals and, if we may say so here, to the work supported by the Association through the specialist centres where treatment is given.

Members of the Association will be pleased to know that a grant has been obtained in collaboration with investigators in Germany, Italy and Sweden further to investigate aspects of the burden of Gaucher disease and other lysosomal disorders in the European population under the Framework Package 7 funded by the European Union. The very recent award of this grant should enable us to do prospective studies as demanded by the findings here in several large

populations of Gaucher patients including the UK, in an attempt to answer these key questions definitively. We feel that such studies relate very much to the daily experience of patients in their own lives. We have been very fortunate in having the support of the Association to launch this study and when the further work has been done will undertake not only to report back but to implement any new standards of care that will be implicit in the findings.

#### **Envoi**

On behalf of all the centres, all staff participants remain enormously grateful to the Association for its incredible support; we are also touched by the confidence placed in us to provide a credible national service for its patients nationally. We feel that strong expectations are always in the air! Nonetheless, we are persuaded that to maintain the highest international standards of practice and advancement for all patients with Gaucher disease, an active on-going programme of research, at all times in partnership, is essential.

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## **Wednesdays' Child Grant to Support Arts Programme**

*Since 2006, in partnership with Great Ormond Street Hospital (GOSH), the Gauchers Association has been running 'Aunty Elin Days' involving a group of girls with neuronopathic Gaucher disease. Over the two years the Association has organised four days out which have started with an activity (which have included an open top bus trip) followed by an afternoon session with Elin Davis, Niamh Finnegan and Lynne Cross, Specialist Nurses from GOSH to discuss issues important to young patients including school, friendships and living with a chronic illness.*

'The Association is extremely grateful to Wednesdays' Child, a North London Charity who have generously supported this project since it started.

Wednesdays' Child have agreed to continue their support with a further grant for a one year art programme for children and young people with neuronopathic Gaucher disease. In the past the project has only been open to a small group but with this grant the Association is now able to open the arts programme to all children and young people with neuronopathic Gaucher disease. The programme will offer the children and young people a choice of different art forms i.e. drama, African drumming, mask making, music and street dancing.

'In addition at the forthcoming European Neuronopathic Family Conference to be held on the 28 – 30<sup>th</sup> November in Northampton the neuronopathic patients and their siblings will have a taste of what will be available to them when they take



*The girls enjoy a day out in London*

part in a drama workshop on the Saturday. During this week end the children and young people will also be asked to design their own arts programme for the following year.

'For further information please contact Tanya Collin-Histed on: 01453 544740 or e-mail: [Tanya@gaucher.org.uk](mailto:Tanya@gaucher.org.uk)'.

# Valentine's Ball raises £21,000 for Gauchers Association

*Since my daughter Mia was diagnosed with type 3 Gauchers disease 7 1/2 years ago at the age of 13 months I have had a strong desire to do something, anything, to help raise funds and awareness of Gauchers, writes Jo Bardoe. Here Jo describes how together with a group of friends she organised a Valentines Ball near her home in West Harting;*

## The Opportunity

'An opportunity came my way when Elin Haf Davis, who had taken such great care of Mia and our family during the Type 3 drug trial, decided to row the Atlantic with Herdip Sidhu to raise money for metabolic medicine at Great Ormond Street Hospital. I decided to set up a committee comprising of a group of eight talented and hard working friends from Harting and Petersfield.

## Our Aim

'We aimed to raise awareness and as much money as possible for the Gauchers Association's Type 3 Gauchers disease fund and help raise money for the "Nautical Nurses".

It was imperative that this local event provided excellent value for money and in doing so I was keen to try to make between £10,000 - £15,000 which would be equally divided between the two charities.

'The Tithe Barn, a stunning local venue was provide free of charge on the 15<sup>th</sup> February and before we knew it we were planning The Valentines Ball.

'We sold the tickets very quickly. Everyone seemed to be keen to help us in our mission and have the opportunity to enjoy a great party.

'We had a tremendous response from local companies and in total over 100 amazing raffle prizes were offered by local and national companies. The most exciting raffle prize was two return tickets to New York courtesy of Virgin Atlantic.

'We ran silent and live auctions and these prizes too were very exciting. They included a holiday in Dubai, tickets to Wimbledon and Stella Artois, a day on the set of Midsomer Murders, an Elite Dinner cruise with Bateaux London, Case of Champagne, VIP tickets to a top London night club and many more.

'Some special friends generously donated all the alcohol. A catering company provided an exquisite three course meal that everyone raved about.

'We had a top London DJ and asked guests to email their top "ball tracks"

'We spoke to local newspapers and radio stations and even had a live interview with the nurses at sea on Radio 5 Live.

## The Evening

'Although the Nautical nurses didn't make it back in time to celebrate with us, the Valentines Ball was a great success. We raised over £28,000 for the two charities on the night and what a night it was! Even some of the teenagers we recruited to work on the night asked not to be paid.

## Other Gauchers Events

'I hadn't expected such an amazing response. It was quite overwhelming.

'A family previously unknown to me held their own Gaucher fundraiser. They had an ex guitarist from Slade play in their house and charged an admission fee raising over £500 for Gauchers. Another friend Steve Brenkley raised £650 running the London Marathon. In addition £2500 was donated by the Bill Butlin Charity Trust: £1000 by the Erica Leonard Trust: £1000 by the William Brake Charitable Fund: £300 from friends: £500 by our local Church for their Lenten appeal and £250 from Anthony Lodge.

'I was delighted with the outcome. It was well worth all the hard work and would have been impossible without such a dedicated committee.

**Steve Brenkley, cricket correspondent of the Independent on Sunday and a friend of the Bardoe family attended the Valentine Ball and gives his personal perspective on the events:**

'It began with an idle chat over coffee in the back garden. It ended with a Valentines Charity Ball that raised in the region of £30k which comes from money raised directly and indirectly from specific donations as a result of the Ball.

'More than 250 guests, all in best bib and tucker, gathered at the Tithe Barn, Ditcham to support the Great



**Jo, Mia and Skye Bardoe pictured with their grandmother and Dr Ashok Vellodi at the Valentines Ball**

Ormond Street Hospital and the Gauchers Association.

'The highlights of a happy evening were two auctions – one live involving conventional bidding, the other silent. Guests were gently but expertly parted from their money by the auctioneer Simon Lush who was at his finest in securing £1200 for a day out at the Queen's Club tennis tournament.

'The ball was born over Jo Bardoe's kitchen table. Her elder daughter, Mia, is eight and has Gaucher Disease. It is a rare condition, an inherited metabolic disorder preventing the proper functioning of cells and organs, it is rare and needs research which needs money. To raise the money it needs fund-raising events and it needs publicity. Over their lingering morning coffee Jo and friends formed a committee and decided to act. Jo's husband Christian was dragooned later.

'Many people rallied round the cause, companies gave auction prizes, individuals offered their time and skills. The measure of willingness to help could be gauged by the fact that nearly 100 people or organisations were thanked in the ball programme.

'The ball was run in tandem with the Transatlantic Row by two Great Ormond Street nurses. Their efforts raised more than £190,000. And the only small blight on a wonderfully organised evening was that the pair did not quite make it back in time to celebrate. Apart from a determined effort to donate cash, they missed some truly bizarre dancing.

'Thank you to all those companies and individuals who contributed to the success of this event by providing services ,raffle and auction items.'

# Nower Hill High School Charity Fun Day raise over £1,000

**Charlotte Cox, sister of James Cox who has Type I Gaucher disease reports on a Charity fun day she organised at Nower Hill High School in Pinner where Charlotte is a Media Studies Teacher:**

'We found out my little brother James had Type One Gaucher Disease in April 2007. In all honesty I had never heard of Gaucher Disease, and had no clue what it was; so the first thing I did was 'Google' it.' For the first few days all we read was bad news. It was both frustrating and very confusing not knowing what Gaucher was, if it could be treated or if it was life threatening?

'We came across the Gauchers Association website and it gave us lots of information and advice which helped us all understand the disease, and re-assured us that although it could not be cured, it could be treated.

'What this made me realise was, not only, was is this disease rare, but reliable information about it is not always readily accessible and I wanted to ensure that people who may be in our position one day have access to useful advice and reassurance, avoiding the horror stories that scared us so much. By supporting the work of the Gauchers Association through fundraising we can ensure that up to date information is always available.

## **Raising Awareness**

'I am a teacher in a High School and 6<sup>th</sup> Form College, and we get our pupils involved in lots of charity events

each year, many of which seem quite faceless, so I decided to ask my pupils if they would like to help me raise some money and awareness about Gaucher Disease and The Gaucher s Association.

'We started by producing a PowerPoint presentation which could be shown to all pupils in every form, telling them about the 'Charity Day' that we were organising on Thursday November 22<sup>nd</sup> 2007 .

'From the moment it was circulated I had pupils coming to ask me about my brother and Gaucher Disease. They wanted to know what it was, if my brother was okay, if my family was okay, if I was okay – some of the boys even offered to take James out at weekends to play football with him to cheer him up.

'Their enthusiasm and compassion was amazing, and with the help of other members of staff and the pupils we prepared the 'Charity Day' to help three charities, which had helped people in our school. These were The Gauchers Association, Diabetes UK and The Odyssey Cancer Trust.

## **A Very Success Event**

'The day was a massive success, with 6<sup>th</sup> form pupils having their legs



waxed, teachers being sponged and a sponsored silence, all in aid of The Gauchers Association. Along with seven other members of staff I had the privilege of being sponged, which although was very cold in the middle of November, was great fun.

'As a school we are proud supporters of The Gauchers Association, and in organising this 'Charity Day' we raised money and awareness for three very worthy causes. A total of £1021 was donated to the Gauchers Association by Nower Hill High School.'

**Editors Note: Special thanks go to Charlotte and all at Nower Hill High School. If you would like to organise a similar event at your school, college or work place please contact Tanya Collin-Histed.**

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## Fund Raising events raise £33,500

**Donations, School charity fun days, a Valentines Ball, six London Marathon runners and donations received in memory of Tom Downes and Peter Crump, have achieved an amazing £33,500 in fundraising efforts in the last six month, writes Don Tendell, Treasurer of the Gauchers Association. I would like to thank everyone individually for their efforts.**

### **In Memory**

Donations totaling £900 have been received from family and friends in memory of Tom Dowes, a member of the Association who sadly passed away in April.

Family of friends donated £140 in memory of Peter Crump, a member of the Association and father of a Gaucher patient.

### **Donations raise £405**

Generous donations have been received from; Melanie Lipson, Diane Peters, Lynne Cross, Harrow on the Hill Lodge.

### **Neuronopathic Fund Donations raise £907**

Thanks go to Helen Cavanagh, Barnato Lodge, St James Church in Lancashire, D Bray, and Bohemian Beads.

# Five Runners raise money for the Gauchers



James Cox cheers on the runners

**92,000 people applied to run this year's London Marathon and Mike, Nick, Liz, Eleanor, Sean and Steve braved the streets of London on Sunday 13<sup>th</sup> April for the benefit of the Gauchers Association as six of the 34,000 who took part.**

Despite two heavy downpours the runners were cheered on by hundreds of thousands of supporters who lined the streets along the 26 mile course.

Below our six runners describe their experience of the day:

**Mike Ingram** who ran with his son **Nick** and writes for both of them "From last Christmas when Nick and I decided to run the London Marathon for James Cox who has Type 1 Gaucher disease we hoped it would be worth it and the occasion didn't let us down. The support and atmosphere around the whole course was an incredible and an unforgettable experience, one that we'll never forget, and to share this with my son and for James was brilliant. We managed to finish in four hours and 26 minutes and collected £1450 for the Gauchers Association, Thank you."

**Eleanor Edwards** said after completing the marathon "It has been an amazing experience to run the London marathon this year. It was my first marathon and I hope it won't be my last. On the day I was very nervous but the support at the start and all around the course was great. My family and friends waited at mile 26 so I had to reach the end and I was so pleased to see them! I finished in five hours 27 with sore legs and a few blisters but I have a lovely medal and pictures to show for it! I am very grateful for the support I have received with both my training and sponsorship...thank you!"

**Stephen Brenkley** is cricket correspondent of the Independent on Sunday and a friend of Mia Bardoe who has Type 3 Gaucher disease. He reports; "Somewhere

between the 14<sup>th</sup> and 15<sup>th</sup> mile was a rock and a hard place. Give up then and slope off home, sparing the pain that was undoubtedly to follow but knowing that if I did you would have to live with a job half done.

'I chose to go on, aware that the next few hours would seem like half a lifetime but that after it was done there might, just might, be some sense of enduring reward. The balance was tilting in favour of going on driven by all the good wishes and the pledges of cash towards the Gauchers Association.

'Then it happened. It wasn't quite a Damascene conversion with the ankles screaming blue murder and the knees saying much worse. A chap passed on the inside looking serene: he was on elbow crutches and he had iron rods where his legs had once been.

'There was no choice after that. If he could do it and look and sound so blissfully happy there could be no giving up. To say that the rest of the course was a breeze would be a stretch but it was a pleasure. The crowds had thinned so that it was a normal Metropolitan Sunday by the time number 32582 approached the finish after seven hours, finishing with what passed for a flourish. There were still some welcome cheers.

'But no matter it was a happy day, it was worth it. Thanks to all for the sponsorship and the kind words. The orange cap which the inhabitants of the Hartings in West Sussex have seen bobbing up and down with excruciating slowness these past few months is now officially retired.'

**Sean Turnbull** reports after his success: "On the morning of the race an auntie phoned to wish me



Sean Turnbull celebrates his



Eleanor Edwards finishes with

# raise £9,000 for s Association



achievement



with a smile

luck and remind me what an achievement it was to get this far, to survive the months of training, blisters, aches, pulls, tweaks and pains and if I didn't finish it wouldn't be the end of the world. Of course it wouldn't be I agreed...but of course it would, of mine anyway. All that mattered was finishing and not just for me, for the Gauchers Association!

'I have so many memories of the day. The sudden mad dash from the loos as the race was about to start. The bands and the PA systems blasting music along the route, children offering their hands for a hi-five, people taking the time and effort to cut oranges or have bowls full of jelly babies and chocolate to offer a much needed and welcome sugar boost. The deafening noise that was made for the Masai Warriors. Being passed by a gorilla and seeing too many Borats! The conversations along the way and the taps of encouragement for the walking wounded.

'I ran in personalised Gauchers Association t- shirt and as such would frequently hear a random "come on Sean!" I would turn in the direction of the voiced encouragement to see a complete stranger smiling at me, often with thumbs up, willing me on – just fantastic.

'Seeing my family at mile 17 gave me the biggest lift and it was as I pulled away from them (after a kiss or two of encouragement from my wife) I heard my eldest son (4 1/2) shout "yeah for daddy". Suddenly I had tears streaming down my face as the emotion of the occasion overwhelmed me, but it was then that I knew I would finish.

'Meeting Tanya Collin-Histed and James Cox and his family on

Horseguards Parade reminded me why I was there and as we talked about the day I thought what made it great was the determination of runners and crowds alike that despite it all, we would succeed.

'At the end of the day and on the tube home a boy aged nine or ten looked at my finishers medal and said "wow, you've run the marathon haven't you!" I looked at my wife, smiled and so began another marathon conversation."

**Liz Fullwood** writes: "I finished in 4hrs 48mins and could have cried as I crossed the finish line, at 17miles it seemed like an impossible feat and to finally get there was what I consider one of my greatest ever achievements. I expect my total raised to be around £1200, so far I have received £800 and am hunting the other sponsors down."

The Association thanks to all its members, friends and supporters who helped raise over £9,000 for the Gauchers Association. Special thanks also go to our six wonderful runners who endured 26 miles in wet conditions to help us in our work.

**Editor's Note:** Photographs courtesy of Chris Ingram: [www.cjiphotosandimages.com](http://www.cjiphotosandimages.com)



Sean Turnbull, Tanya Collin-Histed, Mick and Nick Ingram

# Brains for Brain (B4B)

*Dr David Begley from Kings College, London reports on the 2<sup>nd</sup> Annual B4B meeting that took place in Frankfurt on the 22<sup>nd</sup> March 2008:*

'This was almost exactly one year after the founding meeting in Madrid in 2007. During the year many things have happened. The organisation has consolidated considerably during the year and it has been decided to form the B4B group into a legal entity, a Foundation. Also during the course of the year a substantial donation to B4B has been received from an Italian family specifically for research into Mucopolysaccharidosis. We see this as a beginning to our coordinated research programme. The plan is for B4B to work in close collaboration with Family Associations and Patient groups and organisations such as the Global Organisation for Lysosomal Storage Disorders (GOLD) and the European Study Group for Lysosomal Storage Disorders (ESGLD). In this way we hope to progress research into a better understanding and improvements in treatment for Lysosomal Storage Disorders.

A number of scientific presentations were made at the Frankfurt meeting but in addition, an important component of the meeting was to discuss the groups approach to the forthcoming research call from the European Union FP7 programme for research projects starting in 2009. Calls for the 2009 programme will be made in July 2008 and applications need to be submitted in November. We already have a good idea of the areas in which calls will be made and it is important to start planning our applications as soon as possible. Two calls will be made which are very suitable for B4B to address and the plan is to submit applications to both. One is in the Rare Diseases call under the "Rare Neurological Diseases" topic with up to 6 million Euros available for each project. The second is in the Brain Related Diseases call under the "Understanding the Blood-Brain Barrier (BBB) to Improve Drug Delivery to the Brain" topic with up to 3 million Euros



*Discussions continue over dinner*

available for each project. It is largely due to the lobbying activity of B4B and family associations including the UK Gaucher Association that these specific calls have been made by the EU, which are extremely relevant to Lysosomal Storage Disorders and their understanding and treatment. It is very gratifying that groups of like-minded people can influence European research plans in such a productive and practical way.

'B4B have a number of plans for the future which we hope will lead to improvements in understanding and treatment.'

**Editors note: Brains for Brain (B4B) has a website at: <http://www.brains4brain.eu>**

## LSD and the Brain

*The second 'Lysosomal Diseases and the Brain' conference was held in Sacramento, California, from 29 – 31 May 2008, Prof Tony Futerman of the Weizmann Institute of Science, Israel writes:*

'More than 100 scientists, physicians and students, and a few families of patients with type II and III Gaucher disease attended the conference. Unlike most conferences in this field, the 'Lysosomal Disease and the Brain' conference focuses almost entirely on basic science. The two questions that the conference attempted to address were (1) why does accumulation of metabolites in lysosomes cause disease, and specially neurological disease, and what are the underlying molecular and cellular mechanisms, and (ii) what new therapeutic approaches might arise out of the basic science? These questions were addressed over two days of intense and stimulating discussion, in which some of the leading researchers in the area of Lysosomal disease participated.

'New to the 2008 conference was an attempt to bring researchers who were relatively new to the Lysosomal storage disease world. Thus, a talk was given on glucosylceramide biology

(glucosylceramide is the metabolite that accumulates in Gaucher disease), on lysosomes biology, and autophagy – none of these areas has been addressed in previous conferences. In addition, significant discussion was devoted to mouse models of neuronopathic Gaucher disease and to disease pathology in other related Lysosomal storage disease. As a result of the excellent talks, and the stimulating discussions, a number of putative novel research directions were defined. One of the most exciting discussions concerned the complexity of glucosylceramide biology in cells, which turns out to be much more intricate than once thought. Time will tell whether this complexity is somehow involved in Gaucher pathology. Another stimulating discussion concerned why similar metabolites gives widely differing pathologies in different diseases. Finally, the possibility was raised that multiple types of lysosomes might be present in different cells, and that they



*Prof Tony Futerman*

could differentially accumulate different metabolites, thus indirectly affecting disease pathology. This latter issue is sure to be intensely pursued by researchers over the coming years.

'The Sacramento conference was a great meeting, with talks given at the highest level. The main focus of this meeting is basic science; however, there were also fascinating talks on novel therapeutic approaches, including talks on generic chemical chaperones, and on combination therapies. I congratulate the Children's Gaucher Research Fund for having the vision to understand that basic science is the first step I finding a cure for these lethal diseases, however long it takes.'

# Joint Lysosomal storage diseases patient meeting held at the Royal Free Hospital

*On Sunday, 15<sup>th</sup> June 2008 Dr Atul Mehta and Dr Derralyn Hughes together with their team at The Royal Free Hospital in London organised the first ever joint LSD Patient Meeting.*

Dr Mehta and Dr Hughes organised the programme to ensure that the day focused on the patients and the welcome was given by UK Gaucher Association Chairman, Jeremy Manuel, who praised the organisers on this initiative and reminded those present of the benefit of the designation of the seven National centres which ensures that all patients with Lysosomal Storage Diseases are seen by experts.

Dr Derralyn Hughes presented to the audience of over 100 an introduction and overview to LSD's explaining their causes and effects and detailed both existing and emerging therapies and the way they operate. Although the manifestations of each of the diseases are quite different they are all diseases which result in the storage of material leading to the physical symptom.

Moving presentations were made by patients with each of the disorders and for many present this was a first occasion when they had heard about the experiences of a patient living with one of other conditions. The presentations were deeply personal and in many ways were most uplifting. One participant said "I did not know much about the other conditions but the challenge of coming to terms with

a rare inherited long term disease that may have a treatment but no cure is very similar for all of us even though our symptoms may be quite different."

Dr Rob Anderson of the Health Technology Agency spoke about the National Collaborative Study of the Lysosomal Storage Diseases that is shortly to be starting. He explained the rationale behind what is to be a longitudinal study to capture information about the various diseases (the study is to start with collecting data on Gaucher Patients, MPS1 Patients and Pompe Patients with Fabry Patients to follow) and a lively discussion ensued over the benefits of such study and the possible risks to patients.

After a well earned lunch generously supplied by Marks and Spencer, participants broke into separate disease specific workshops.

Dr Mehta opened the Gaucher workshop with a discussion on emerging new therapies. He explained the various different products starting clinical trials (and announced that The Royal Free had been "opened" as a trial centre for the Protalix Enzyme) and he led a discussion about how a patient would decide whether or not to go into clinical study.

Professor Timothy Cox addressed the increasingly relevant but difficult topic of how Gaucher Disease should be classified. He presented an overview of the differing manifestations of Gaucher Disease from individual personal case studies and with the recognition of differing neurological symptoms developing in Gaucher patients which raised the question whether it is still correct to rigorously classify Gaucher Disease into "types."

Finally, Dr Patrick Degan presented the initial results of the Bone Study commenting on the fact that in many ways Gaucher Disease is a disease of the bone and that whilst enzyme replacement therapy is able to reverse symptoms in organs (such as the spleen) and whilst there is some evidence that further bone disease may be prevented by enzyme replacement therapy it is not possible to reverse bone damage. He said that he was hoping to publish more data from the Bone Study in due course.

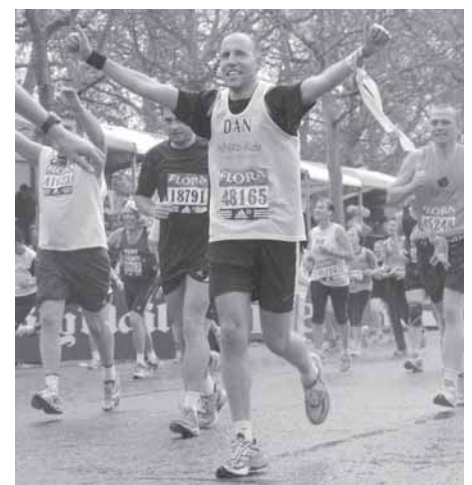
After tea a patient from each of the three workshops gave a short presentation of what had been discussed and what they had gained from the day. The Meeting concluded with a vote of thanks given by Mrs Christine Lavery, Chief Executive of the MPS Society.

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## Dan Brown to run the New York Marathon for the Gauchers Association

On Sunday 2<sup>nd</sup> November Dan Brown (pictured right finishing the 2006 London Marathon) who has Type 1 Gaucher disease will line up with around 40,000 and run the 26 miles across five bridges and through five boroughs of New York City with more than two million spectators cheering them on. Dan who was diagnosed with Gaucher disease in September 2003, is an active member of the Association, speaking at family conferences and attending Executive Committee meetings.

If you would like to support Dan to run the New York marathon please visit the Association's Just Giving page at: <http://www.justgiving.com/gauchers>



**Dan completes the 2006 London Marathon**

# Novel enzyme replacement therapy for Gaucher disease: On-going phase III clinical trial with recombinant human glucocerebrosidase expressed in plant cells

*In the December 2007 edition of the Gauchers News Dr Einat Almon, Vice President of Product Development, Protalix Biotherapeutics reported on the Phase III trial on new enzyme replacement therapy produced in plant cells for patients with Type I Gaucher disease. Dr Almon provides a further update:*

'Protalix Biotherapeutics, is currently performing a Phase III clinical trial to assess the safety and efficacy of prGCD in Gaucher disease patients, in major medical centers worldwide including Shaarei Zedek Jerusalem, Morningside Medi Clinic Johannesburg, NYU New York and Royal Free hospital in London. The phase III clinical trial is aiming of recruiting 30 adult (older than 18 years) untreated Gaucher patients.

'Recently, Protalix Biotherapeutics announced that it intends to initiate a double-blind (this is where both the patient and the treating physician are not aware of the dose of treatment given), follow-on extension study as part of the Company's on-going phase III clinical trial of its prGCD. Eligible patients who have successfully completed treatment as part of the pivotal phase III clinical trial will be offered the opportunity to continue to be treated with prGCD at the same dose that they received in the trial. The objective of the proposed extension study is to compile additional

information relating to the long term safety and efficacy of prGCD.

"We are encouraged by the clinical results to date, and look forward to reporting the results of the phase III clinical trial when they become available" said **Professor Ari Zimran**, M.D., Director of the Gaucher Clinic at Shaare Zedek Medical Center in Jerusalem and Principal Investigator for the trial. "The Gaucher disease community has a keen interest in developing new treatment options, particularly treatments that will be less expensive." Professor Zimran is a member of the Company's Scientific Advisory Board.

'In addition, a switch-over study to prGCD is to begin soon, for stable patients who have been treated with the currently available ERT treatment for at least two years and have been on the same dose for at least six months.

'Prior to the pivotal trial, Protalix completed a Phase I clinical trial under FDA Investigational New Drug (IND) approval to assess the safety of prGCD



in healthy volunteers. All doses administered to subjects in the Phase I clinical trial demonstrated a good tolerability profile. The prGCD product has at least equal in-vitro efficacy to current product in the market and both have similar three dimensional structures (protein crystallization)

'Protalix is a publicly traded clinical stage biopharmaceutical company (AMEX: PLX) that is focused on developing and manufacturing of recombinant therapeutic proteins. Protalix uses its proprietary plant cell culture and bioreactor technology for the expression of recombinant therapeutic proteins, and is currently developing several biopharmaceutical products. The lead product under development is prGCD, an enzyme for the treatment of Gaucher disease'.

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## Biotech Company Amicus announces preliminary Phase II Pharmacological Chaperone results

*Amicus Therapeutics of New Jersey, USA presented preliminary data from a phase II study of AT2101 during the recent American College of Medical Genetics (ACMG) meeting in Phoenix Arizona. Dr. Eugene Schneider, Medical Director for the Gaucher Program at Amicus Therapeutics, provided the following update;*

'AT2101 is an orally administered pharmacological chaperone currently under investigation for the treatment of Gaucher disease.

'Gaucher disease is a lysosomal storage disorder caused by genetic mutations in the *GBA* gene that lead to a deficiency of the lysosomal enzyme acid  $\beta$ -glucosidase (GCase). Most individuals with Type 1 Gaucher

disease are thought to make GCase; however, the majority of *GBA* mutations in this population result in the production of misfolded or unstable GCase.

'The role of GCase is to break down a substrate known as glucocerebroside in a compartment within the cell known as the lysosome. GCase is made in a different compartment in the cell

called the endoplasmic reticulum (ER) so it must be transported, or "trafficked" from the ER to the lysosome. The ER has a quality control system that regulates GCase trafficking within the cell. This quality control system allows appropriately folded GCase to exit the ER and be sent to the lysosome, where it is needed to break down substrate. Misfolded GCase may be retained in the ER and thus unable to get to the lysosome. This results in a deficiency of GCase in the lysosome leading to accumulation of substrate.

*(continued on opposite page)*

### Key Findings

The primary objective of this Phase 2 study was to evaluate the safety and tolerability of different doses and dosing regimens of AT2101. The secondary objective was to evaluate the biological effects of the investigational treatment by measuring various laboratory markers relevant to Gaucher disease.

'A total of 30 men and women were enrolled in this Phase II study in sites across the United States. Several mutations were represented including the common N370S and L444P mutations. Subjects were on enzyme replacement therapy (ERT) for an average of nine years before entering the trial, and were temporarily discontinued from ERT to receive AT2101 for the four-week treatment phase of the study.

The key preliminary findings from the clinical trial as reported at ACMG include:

- AT2101 was generally well-tolerated at all doses evaluated and no serious adverse events were reported;
- GCase activity measured in the white blood cells was increased in 20 of the 26 subjects with evaluable GCase data;
- Five out of six subjects who did not show a clear increase in GCase activity were either in the lowest dose group or the group that was dosed least frequently;
- In this short-term study, the levels of relevant markers of Gaucher disease were maintained; these included platelet counts, chitotriosidase activity, hemoglobin levels, substrate levels, and pulmonary activation-related chemokine (PARC) levels.

'The preliminary results of this study support further evaluation of AT2101 in patients with Gaucher disease. A six-month Phase II study in individuals who are naïve to ERT or substrate reduction therapy (SRT), or who have not received either ERT or SRT for at least 12 months, is currently ongoing at sites in the United States, Germany, Israel and the United Kingdom'.

## Just Giving raises in excess of £4000 for the Association

Last year the Gauchers Association teamed up with justgiving.com to provide a quick and easy facility to donate money online. We are delighted to tell you that to date we have received donations in excess of £4,000 through this website.

The site is simple to use, just type in [www.justgiving.com](http://www.justgiving.com) and search for the Gauchers Association. Then follow the instructions on the screen and once you have made your donation you will automatically receive a confirmation and receipt. The Association also keeps a full record of who has donated funds in this way.



Members and their families who wish to raise funds for the Gauchers Association can use the site to make their own personalised web pages. All that is needed is to log onto the site; go to the area marked 'Get Your Free online Fundraising Page' and click the button 'Get Fundraising'. For more information on using the site call Tanya Collin-Histed on: 00 44 1453 549231 or e-mail: [ga@gaucher.org.uk](mailto:ga@gaucher.org.uk)

## Remember you can raise funds for the association whilst you shop online

[www.easyfundraising.org.uk](http://www.easyfundraising.org.uk) is a FREE service where you can shop at your



favourite online stores to raise funds for

the Gauchers Association at no extra cost to you. You still deal directly with each retailer as you would normally, but simply by using the links from the Easyfundraising site each retailer will make a donation to the Gauchers Association. This is how it works:

**Firstly** Register at

[www.easyfundraising.co.uk](http://www.easyfundraising.co.uk) so the site knows that you wish to support the Gauchers Association. Registration is completely FREE.

**Next**, login using your username / password. This is how the system recognises who you are and which cause benefits when you make purchases.

**Finally**, click any of the retailer links provided and then shop just as you would normally.

## European Neuronopathic Gaucher Disease (Type III) Family Conference

Will held on November 28 – 30, 2008 at the Hilton Hotel, Northampton, UK

**Presentations will include;** research, audiology; personal stories; behavioral issues, and clinical management.

Members, families, friends, healthcare professionals and Industry representatives are welcome to attend the Saturday day time conference and a diner on the Saturday evening. The Sunday morning programme is open to families only.

A supervised arts and drama programme will be organised for children and young people to attend over the week end.

For further details and a programme of the weekend please contact Tanya Collin-Histed on: 00 44 1453 549231 or Email: [Tanya@gaucher.org.uk](mailto:Tanya@gaucher.org.uk)

# Serbian Patient's Meeting

*Tanya Collin-Histed, Executive Director of the Gauchers Association reports on the Serbian Gaucher Patients' meeting which took place on 18 and 19 April in Belgrade;*

'At the invitation of **Darinka Sulic, Dr Ashok Vellodi** and I were guests at the Serbia Gauchers Association's National patient meeting held in Belgrade on the 18<sup>th</sup> and 19<sup>th</sup> April. To coincide with the meeting leading local Gaucher physicians **Dr Maja Djordjevic** (who is a paediatrician) and **Prof Nada Suvajdzic** (who is a haematologist treating adult), from two Children's Hospital's in Belgrade organised a patient clinic at which Dr Vellodi was their guest and examined three Type III Gaucher patients.

## Presentations

'Dr Ashok Vellodi gave a presentation to medical students and doctors at the Mother and Child Hospital in Belgrade on Neuronopathic Gaucher disease and its clinical management. This was warmly received and provoked much discussion.

'At the patient meeting on the Saturday morning I explained the structure and workings of the UK Gauchers Association and the purpose and aspirations of the European Gaucher Alliance. **Vladimir Tomov** Chairman of the Bulgarian Gaucher Patient Organisation described the

development of the Bulgarian Association and how they have been working with other Rare Disease Patient Associations in Bulgaria to develop a National Alliance of People with Rare Disease. A total of 30 patients, family members and doctors attended the meeting. Dr Maja Djordjevic and **Dr Pedrag Rodic** (a doctor working with Prof Nada Suvajdzic treating adult Gaucher patients) described the manifestations of Gaucher disease to patients and spoke about the progress of the clinical treatment of paediatric Gaucher patients in Serbia on Enzyme Replacement Therapy.

'There are 37 Gaucher patients in Serbia, 22 of which currently receive enzyme replacement therapy through Genzyme's European Cerezyme Access Programme (ECAP). The remaining 15 patients have mild disease and are currently untreated but they are being closely clinically managed by their doctors in local hospitals.

The discussion following the presentations highlighted that the Serbian Patient Association's face a number of challenges. They discussed how to develop channels of communication with the Ministry of



*Serbian Patient meeting*

Health, to lobby for government funding to pay for the treatment for Gaucher patients and those with other rare diseases, and seek to improve adult management of Gaucher patients both nationally and locally. Currently patients have to travel to Belgrade to receive their treatment every fortnight, but many of these patients and their families live a long way from Belgrade. The Patient Association in partnership with the expert doctors in Belgrade hospitals determined to work closely with doctors in local hospitals to encourage them to administer patient's treatment.

'Dr Maja Djordjevic hailed the meeting as a great success. She said "the Serbian Gaucher Association have a number of challenges for the future but the support of our friends from overseas is a great help. The sharing of experiences with Dr Vellodi will certainly help to improve the quality of life for the young Type III Gaucher patients, I am grateful for him taking the time to visit us."

# Jeremy Manuel elected Chairman of the EGA

*At a meeting of the Members of the European Gaucher Alliance held in Budapest on 3<sup>rd</sup> June 2008 representatives from patient groups from 24 European countries unanimously agreed to adopt a constitution and to incorporate the European Gaucher Alliance as a formal body.*

Until then the EGA had been an informal structure without officers or structure but those present felt that the time had come to formalise and to become an official body. The first Board of the EGA was elected and consists of:-

- Yossi Cohen (Israel);
- Tanya Collin-Histed (UK);
- Gil Faran (Israel)
- Anne-Grethe Laurdisen (Denmark);
- Lars Magnusson (Sweden);
- Jeremy Manuel (UK);
- Pascal Niemeyer (Germany);
- Wojtek Oswiecinski (Poland);

- Fernanda Torquati (Italy).

UK Gaucher Association Chairman, Jeremy Manuel was elected the first Chairman of the EGA. He commented:-

"It is a huge honour and privilege to be elected as the first Chairman of the EGA and I am delighted that I have a great team who have committed themselves to driving forward the aims and objectives of the EGA. Significant challenges lie ahead. It has become apparent that the issues that affect the Gaucher patients are not limited by national boundaries. Health officials are constantly looking to see



*The EGA Board*

how health care is provided in neighbouring countries. Pharmaceutical companies are global organisations. Doctors and scientists are increasingly working together on collaborative projects so it is right that there should be a formal structure in place for patients so that they may work together on a Pan European basis to their mutual advantage. Together we will strive to enhance the position of the Gaucher patient in Europe and in all other parts of the world."

# Romanian Patients Participating in the Clinical Trial for Gaucher Disease in Jerusalem

*Dr Debbie Elstein, from the Shaare Zedek Medical Centre, Israel provides an update on the happenings at our Gaucher Clinic in Jerusalem:*



*Monica and her husband Daniel*

'The topic, I believe, is appropriate to the UK Patient Association Newsletter because it really is an outgrowth of the spirit and vision of the late **Susan Lewis** who inspired us all to think about patients with Gaucher disease who are less able to receive optimum care. Despite suffering from severe manifestations of Gaucher disease herself, Susan was a staunch advocate of outreach to all patients who need specific therapy for Gaucher disease, and as such, has left us a legacy of an ethical imperative to continue her vision.

'We would like to tell you about a brother and sister with Gaucher disease from Romania, **Bogdan** and **Monica**, whom we met when in Bucharest to evaluate possible candidate for the then TKT (now Shire HGT) clinical trials in type I Gaucher disease. Both were in their early 20's and had known about their disease for many years, and met the commonly accepted criteria for enzyme therapy. Both suffered from massive enlargement of the spleen and liver and Monica in particular had impressive bone involvement. Nonetheless, both of these young

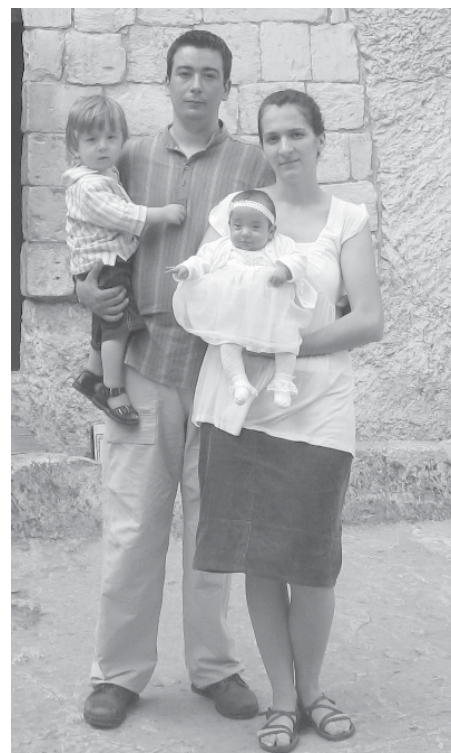
people were attending university and working to support themselves.

'Bogdan and Monica elected to come to Jerusalem in April 2004 and enroll in the seminal clinical trial with velaglucerase alfa (previously GA-GCB), the new infusible enzyme. This was not an easy decision, both in terms of the commitment to the trial, flying to Israel every other week, and politically, because this was the time of heightened terrorist activity in our country. Bogdan in particular immediately fell in love with Israel and insisted on taking courses in spoken Hebrew. Both also had to keep their grades up in school at home, and indeed by the end of the trial, Bogdan graduated with a degree in mechanical engineering and Monica in physiotherapy.

'The results of the clinical trial with velaglucerase alfa were excellent and these two patients, like the rest of the participants, benefited by significant reductions in spleen and liver volume and improvement in hematological features. Monica has also undergone some orthopedic management in our hospital that has improved her quality of life.

'Both Bogdan and Monica have continued into the extension phase of the velaglucerase alfa trial, now in its fourth year, and are continuing to do well.

In fact they are doing very well indeed: Monica married her high school sweet-heart and started to receive her infusions in Bucharest with periodic visits to Jerusalem for routine evaluations. Bogdan, on the other hand, met and married a young



*Bogdan with wife Thea, son Ariel and daughter Batel*

Romanian woman in the church he attended every Saturday when in Israel, and they now live and work in Israel and have a beautiful son, Ariel, whose name is one of the biblical names for Jerusalem, and a little princess named Batel, which means a daughter of God.

These life-affirming stories are the story of our Gaucher community: it was Susan Lewis who inspired us to reach out across the globe to help all patients, it was TKT/Shire, that facilitated a treatment option for non-nationals in our Jerusalem center, and it was the extraordinary fortitude of the patients who refuse to let their disease status get in the way of living a full and happy life.'

## Shire Human Genetic Therapies and Amicus Therapeutics Partner in Gaucher Disease

In November 2007, Shire Human Genetic Therapies (HGT) and Amicus Therapeutics announced a collaboration to jointly develop three compounds from Amicus Therapeutics. This partnership leverages Shire HGT's experience in supporting global access to treatments for patients. In addition to Shire HGT co-developing the three compounds with Amicus, Shire HGT will be responsible for registration and commercialization in all geographic areas outside of the US, including Europe, and Amicus will be responsible for registration and commercialization in the US.

# Nautical Nurses cross the Atlantic and Elin takes up new challenge

*In the December edition of the Gauchers News we reported on the adventures of two nurses from Great Ormond Street Hospital, Elin Haf Davies and Herdip Sidu who planned to row across the Atlantic together to raise funds to help improve the lives of patients with life threatening metabolic disorders such as cystic fibrosis and Gaucher disease. Elin Haf Davies reports on their extraordinary adventure;*

'After 77 days 7 hours and 37 minutes of rowing, Herdip and I finally landed in Antigua (pictured below). It was the end of two years hard training, planning and life encompassing commitment which involved more emotional highs and lows than either of us could have possibly imagined. Herdip suffered severe dehydration as a result of sea sickness and sun stroke - which required an intravenous fluid infusion. She later also scalded herself with boiling hot water, so our nursing skills came to be very useful. I also suffered from excruciating tooth ache to match the pain in her rear end as a result of pressure and salt sores!

'Rowing an hour and a half on, and



an hour and a half off, day and night continuously resulted in major sleep deprivation, which was one of the hardest aspects of the challenge, that and being pushed back by the wind what had taken hours to row! But it was all worth it for the fundraising total achieved. The money is still coming in but at least £190,000 has already been raised, not forgetting what we will get from the sale of the boat. The money will fund two metabolic research projects led by Dr Ashok Vellodi and Professor Peter Clayton at Great Ormond St Hospital.

## New Challenge

'After such experience however, I am now already signed up for my next

challenge at the end of this year. Landing in Antigua and seeing my family was a very emotional experience for me. When I later landed in Gatwick airport (pictured left) to be welcomed my friends, close colleagues and children and families from GOSH, I really was overwhelmed, and inspired to go and do something else like it, as soon as possible. So I am doing the Sahara marathon, or six to be precise! A total 250km over six days in the heat of the desert. This time I have decided to raise funds for the Gaucher Association. I had such a great response when I ran the London marathon in 2007 for the Gauchers Association, that I am now keen to show my support by raising more money for the charity.'

## Congratulations

The Gauchers Association would like to congratulate Elin and Herdip on this amazing and altruistic achievement, we are delighted that Elin has chosen the Gauchers Association to benefit from her next challenge. The Sahara challenge will take place on 26 October later this year. We would like to wish Elin all the success in this forthcoming adventure.

If you would like to make a donation in support of Elin please visit the Association's Just Giving page at: <http://www.justgiving.com/gauchers>

## New Charity Christmas Cards for Sale

The Gauchers Association is pleased to announce that in September we will be launching our first ever Charity Christmas card. The design has been drawn by one of our younger members and was selected from a number of entries received.

### Packs of Cards

Individual packs are available for purchase. Each pack contains 10 cards (1 design), these are available at £5.99 per pack (postage & packaging is £2.50 per order for UK and Channel Islands and £3.50 for Eire and Europe).

To place an order simply call Tanya Collin-Histed on: 00 44 1453 549231 or email [ga@gaucher.org.uk](mailto:ga@gaucher.org.uk)

Orders will be sent out in late September.

