Gauchers NEWS

DECEMBER 2007

Gauchers ASSOCIATION

James Cox with his rabbit Spot

(see James' Gaucher Story on page 3)

Lysosomal Storage Disorder Centres

Addenbrooke's Hospital
Hills Road, Cambridge CB2 2QQ
Head of Clinic: Prof Timothy Cox
Tel: 01223 336 864
Fax: 01223 336 846
National Helpline: 01223 216 295

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
Fax: 0121 333 9998

Great Ormond Street Hospital for Sick Children
Great Ormond Street
London WC1N 3JH
Head of Clinic: Dr Ashok Vellodi
Tel: 020 7405 9200 ext 0075
Fax: 020 7813 8258

Hope Hospital
Department of Lysosomal Storage Disorders
Hope Hospital
Stott Lane, Salford
Manchester M6 8HD
Head of Clinic: Dr Steve Waldeck
Tel. 0161 206 4365 / 1419 / 1080.
Fax 0161 206 4036

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
Tel: 0161 922 2137 (9am - 5pm)
Fax: 0161 922 2137
Helpline: out of hours ring
0161 794 4696
and ask for the metabolic consultant.

Chairman's Forward

Dear friends,

Welcome to the December 2007 edition of Gaucher News.

As you will see this contains the usual mix of personal stories, fundraising ideas and updates on the clinical trials which are currently being undertaken.

This edition also launches the Susan Lewis Memorial Fund through which we are seeking to raise significant sums of money to enable the Association to bring doctors and other healthcare professionals from overseas to experience working alongside the teams at our National Centres of Excellence and thus develop their own skills that they can then use in their home countries.

We are looking forward in the year ahead to receiving a full report on the progress of the bone project and we hope that in the next newsletter we will be able to give a full update.

This edition also contains some exciting information on the ongoing clinical trials for new therapies for Gaucher Disease and we look forward to hearing further results in the future that will be of benefit to Gaucher patients.

The year ahead will also see the Eighth European Working Group on Gaucher Disease which takes place in Budapest in June. This is always an exciting forum and offers the opportunity for the world's experts to exchange ideas, to report on their research and to update on the clinical trials.

2008 promises therefore to be an important and exciting year for Gaucher patients and the Association is ready to meet the challenges that lie ahead. Thank you to all the members and friends who have come forward in response to my last request to become more actively involved. We are always looking for more people to help. Do give Tanya a call.

With every good wish to members and friends for the holiday season and for good health and happiness in 2008.

Yours,

Jeremy Manuel OBE
Chairman

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Edited by: Tanya Collin-Histed • Chairman: Jeremy Manuel OBE
Executive Director: Tanya Collin-Histed • Treasurer: Don Tendell
Fund Raisers Alan & Sharon Rosen and Melanie Lipson
Parent Co-ordinator: Susan Noe

My Gaucher Story, by James

James is 11 years old and has Type 1 Gaucher disease. James was diagnosed with Gauchers disease earlier this year; here is his story in his own words

'I live in Harrow with my mum, dad, sister and brother. My story begins earlier this year when I was in year 6 at primary school. My left knee started to swell and was painful; at first I thought it was a sports injury as I had just started playing table tennis at an after school club. In February I played table tennis for the whole day at the National Schools Championship where I was a zone finalist and during the day my knee got much worse.

From GP to Great Ormond Street Hospital

'My mum and dad took me to the doctor and I saw a trainee GP who was not sure what was wrong with me so he phoned the hospital for advice and I was told I had to go to the hospital straight away. At the hospital a doctor named Helen felt my tummy and said she thought I had a very enlarged spleen. The same day I saw another doctor called Mr Hyer who sent me to Great Ormond Street to the leukaemia ward to have a bone marrow biopsy.

'At Great Ormond Street hospital the doctors took a lot of blood and I nearly fainted as no blood wanted to come out of my veins that day. Later that day I saw a blood doctor and he told us that he had found some funny cells called 'Gaucher' cells in my bone marrow.

'We then had an appointment in the next few weeks to see Dr Vellodi and Niamh Finnegan the clinical nurse specialist. Dr Vellodi sent me for lots and lots of x-rays and scans. A few weeks later I had an appointment at GOSH in Kingfisher ward to start my Cerezyme infusions.

Cerezyme Infusions

'I now have my Cerezyme infusions every two weeks. After eight infusions of Cerezyme at the hospital I have started to have them at home. I am about to have the third one at home and Tracy my nurse is bringing a new nurse with her.

Telling my school friends

'After the summer holidays I started at my new High School and there were lots of children I did not know in my class. When I was at GOSH having one of my treatments Lee Mead who is the star of the musical 'Joseph' visited the ward and my dad took a picture of us together. I took this photo to school and used it to explain to my class about my cells and bone marrow. It made it easier to tell them as I was worried about them making fun of me as some children had done this in my primary school. But everyone liked the photo and my form teacher put it on the notice board for everyone to see.

Playing sports

'My spleen has gone down a lot but Dr Vellodi says I cannot do all sports yet. This has made me a bit sad as we have a big sports centre in high school and I cannot do all these things with my friends.'

Meeting other Gaucher children

'I would like to meet other people especially young people with Gaucher disease as it would be good to have other people of my own age to talk to.

Mum's Story by Angela Cox

You have read James' story and ours is almost the same writes James's mother Angela:

'Our perspective as his parents is slightly different as we were aware that things were not quite right for some time before his diagnosis. I work as a radiotherapy radiographer and this gave me an awareness of the implications of some of his symptoms…but you never want to fear the worst. James had heavy nosebleeds, easily bruised and other small problems and we had been alerted by his school welfare officer that they had problems stopping a nose bleed which had lasted a long while. However he always seemed to recover quickly.

The Journey Began

'When he first got sent to the hospital we were aware that they were looking for leukaemia before they indicated to us that this was what was wrong with him. That night at the hospital when he was first examined we felt devastated. Initially he was going to be admitted but there was a flood on the children's ward and we were sent home and had to return first thing next day.

James receiving his first Cerezyme infusion pictured with Lee Mead star of the West End Musical 'Joseph'

'He had ultrasound scans of his liver and spleen but the blood results did not add up to the leukaemia diagnosis. The doctors went through as many possibilities as they could think of but still nothing fitted. It was then decided he should have a bone marrow biopsy to clarify if it was leukaemia and if this was negative he should then be referred to the rheumatologists for further investigations. We then saw the Haematologist at GOS where the bone marrow biopsy was carried out and that afternoon was like a roller coaster ride…..Good news, no evidence of leukaemia or any other malignancy therefore we could just go back to our local hospital for a rheumatology referral.

'But as James recovered from the general anaesthetic we were called back in to see the consultant again and we both found ourselves having our spleens examined to see if they were enlarged too. More news from the lab…some cells had been found in his marrow that was identified as Gaucher Cells. We felt very relieved that it wasn't cancer and we had never heard of Gaucher Disease but understood the seriousness of storage diseases. More blood had to be taken and the results would take several days to come back to confirm the final diagnosis. In the mean time we were sent home and told we would be sent an appointment to see the Metabolic team at GOS in a few weeks.

What was Gaucher Disease?

'The roller coaster ride of emotions had only just started. Relief and heartbreak.

(continued on page 16)

Susan Lewis Memorial Fund

Jeremy Manuel writes:

'The constant focus of all of Susan's Gaucher activities was to help Gaucher patients and their families. She approached every meeting, conference or presentation with the question as to how to improve the position of patients both individually and collectively.

Susan (second row on the right) with Romania Gaucher patients and their families

'When we started the Association the patients' prime need was information, and one of the first things that Susan did was to gather information from all sources and set up a newsletter to disseminate everything that the Association knew or learned. She knew that the collective experience would help patients.

'It also quickly became clear that patients needed access to healthcare professionals who had experience in treating Gaucher Disease. In the UK relationships quickly developed with the doctors and their teams who had both an interest in Gaucher Disease and had exposure to Gaucher patients. Inevitably as more patients came to the Association for advice they were encouraged towards those doctors and other healthcare professionals who had such experience thereby enlarging the pool of patients these doctors were seeing. In our own way the Association therefore played a significant role in the development of these departments that were to become the National Centres and Susan was passionate in her beliefs that the establishment of the National Centres for the treatment of Gaucher Disease in the UK provided patients with the very best advice and treatment of their disease.

'As the internet became part of all our lives the Association (or more accurately Susan's husband David Lewis) developed its own website and approaches for advice and for help started to come in from all around the world.

'There are countless stories of patients (often parents of children with Gaucher Disease) who were desperate for help and contacted us and Susan did everything that she could in facilitating contacts with doctors and with Genzyme to see if treatment could be obtained for these often desperate cases. What again became evident was that these patients needed exactly the same as patients in the UK had. They needed information about the disease and its possible treatments and access to expert, physicians and their teams to manage and treat their disease.

'Much has been written about the work of the European Gaucher Alliance (EGA) to secure humanitarian aid for patients in Eastern Europe where enzyme therapy was not available to them. Susan was particularly proud of the fact that by the time of her death well over a hundred patients (now 114) were receiving free treatment through the ECAP programme.

'Through the establishment of national patient associations, through their contacts with other European patient groups and the European Gaucher Alliance Newsletter and of course through the internet, patients throughout the world have access to a considerable amount of information about Gaucher Disease.

'However what they do not always have is access to expert doctors and this is the area where the Association want to take action in honour of Susan together with her husband David. We are now launching the Susan Lewis Memorial Fund to provide grants and bursaries to doctors and other healthcare professionals from developing countries (particularly in Eastern Europe) to allow them to travel to UK Centres of Excellence to undertake mentoring and educational programmes in the treatment and management of Gaucher Disease.

Serbian Gaucher patients who receive ERT through ECAP, picture taken by Susan on a trip to Serbia in 2004

Susan on a trip to the Ukraine in 2003 pictured here (centre) with patients and their families

'The intention is the grants would cover travel, accommodation with a small daily subsistence allowance and the programmes will be individually tailored to take into account the applicants experience and special areas of Gaucher interest. Successful applicants will be expected on return to their countries to work with and assist others in gaining experience of Gaucher Disease.

'In this way we hope to be able to help to address in Susan's name a lack of expertise which may exist in countries where there are few Gaucher patients and where treatment until recently has not been available. In the name of Susan Lewis we want to help doctors and their teams achieve the excellent levels of expertise available in the long established centres so that patients in these countries can experience the excellence in treatment that patients in the UK receive.

'This proposal has been met with enthusiasm by the Centres in the UK by patient associations making up the EGA, and by industry who have expressed themselves very keen to support the project and to record their appreciation for all Susan has done in a tangible way.'

'I do hope members and their friends will be able to support this project. We are aiming to raise a significant sum to enable us to run this project for a number of years. By enhancing the skills of committed physicians interested in treating patients with Gaucher Disease we believe that we are continuing the work that Susan believed so important and keeping our focus on the needs of the Gaucher patient which Susan always did.

'We have established a special fund in which to receive donations. Please make cheques out to the Susan Lewis Memorial Fund and send them to the Association's Treasurer Mr Don Tendell at: Red Court, Beaconsfield Road, Farnham Royal, Slough, SL2 3BY.'

Travelling with ERT

Patients receiving treatment who are planning a holiday in the UK, taking an extended break abroad, or going to university, will need to think about how their treatment regime fits into their plans.

The Homecare Companies providing support to Gaucher patients offer the following services to help overcome the difficulties that patients may experience when travelling, studying or working away from home.

Storage: If patients are able (and willing) to transport the therapy in the UK or overseas, the Homecare Companies can provided a validated cold chain storage box that can keep the drug at between +2 and +8ºC for up to 48 hours. On arrival the drug MUST be transferred to a refrigerator at between 2 and 8ºC until required. If patients are staying in a hotel, cottage, apartment or with relatives a check must be made to ensure that there is a suitable refrigerator available for storage of the drug. Discuss travel plans with the Homecare Company so they can give suitable advice and provide cold chain boxes.

Supporting Papers: If travelling abroad, the Homecare Company can provide documentation for the customs authorities to help clearance of the drug and ancillaries through customs.

Nursing Support: If travelling on holiday in the UK, a nurse will, if required, visit the patient's holiday address or if more suitable, a local health centre to administer treatment.

Extended visits overseas: If travelling aboard for an extended period i.e. more than one month, the Homecare Companies can arrange to have the therapy delivered to a private address or healthcare facility. If nursing services are required to infuse the drug, the patient's own NHS Gaucher Centre and Homecare Company will help to arrange this.

Student travel: Many students like to take a gap year travelling. For Gaucher patients this may be possible but it will be necessary to adhere to a strict travel plan with specific treatment dates to prearrange therapy deliveries. The arrangements for this would need to be discussed with the patients Consultant. The Homecare Company would, if required, be able to make contact with overseas medical establishments and liaise with local medical professionals to discuss storage and administration.

Patients should also discuss destinations with their own consultants and ascertain country or area specific medical advice.

Going to University: If patients are going to college or university and living in student halls and there is nowhere suitable and safe to store the drug and ancillaries, or if a patient wants to keep this part of his/her life separate, the Homecare Company can arrange for the drug and ancillaries to be delivered to a local health centre and organise a healthcare professional to administer the drug. Alternatively, if a patient is happy to store the drug at his/her home whilst at university, and then simply change the delivery address to the new one.

The key to success when travelling with ERT is to plan ahead. Patients should inform their Gaucher Centre and Homecare Company of plans as soon as possible so that together necessary arrangements can be made for the delivery, storage and administration of the therapy.

Fundraising

Members Subscription Donations total £1582
We would like to thank all of our members who generously donated additional funds to the Association with payment of their annual subscription.

In Memory
Donations totalling £583 have been received in memory of **Dr Esmond Ball**, a member of the Association and father to three children with Gaucher disease.

The Family of **Clive Harries,** who had Gaucher disease has donated £40 to the Association on his birthday, Clive passed away in 1989.

The Ex-CID Officers Association donated £100 in memory of **Mr Cliff Smith**.

Donations raise £635
Generous donations have been received from; Barry Horowitz, Candace Fleischmann, M Grossman, The Ardwick Trust, Sharon Rosen British WIZO, Barbara Okada, H Henson, A Y Goldberg, Gloria Gordon,

Wedding Anniversary and Birthdays
In celebration of **Jean** and **David Bray**'s Ruby Wedding Anniversary, friends and family donated £540 for the Association. David and Jean are the maternal Grandparents of Emily who has Gaucher disease.

Eric Beecham generously donated to the Association on his significant birthday

Thanks go to The Manuel Charitable Foundation, Michael and Lisette Keats, Mandy and Russell Caller and Suzanne and Keith Barnett who celebrated the 50th birthday of the Association's Chairman **Jeremy Manuel OBE** with donations to the Association.

Special Thanks go to: 13 years old **Indie Culley** who rode her bike and raised £34 for the Association's neuronopathic fund. Indie is a friend Mia who has Type III Gaucher disease.

Effect of Miglustat (Zavesca) on bone disease in adults with Type 1 Gauchers disease

Published in the Journal of Clinical Therapeutics, 2007 a pooled analysis by Prof Gregory M. Pastores, Dr Deborah Elstein, Prof Martin Hrebicek and Prof Ari Zimran of three multinational open-label studies explores the effect of Miglustat on bone disease in adults with Type 1 Gaucher disease. In this report Dr Derralyn Hughes Lecturer in Haematology, Dept. Academic Haematology at the Royal Free & University College Medical School summarises the paper;

Dr Derralyn Hughes

'Gaucher disease, the most common lysosomal storage disorder, has multi-organ manifestations including those which are a consequence of bulk accumulation of undegraded substrate such as enlargement of liver and spleen and those that are more difficult to understand, such as bone disease. Bone manifestations can be amongst the most debilitating and disabling of symptoms of Gaucher disease. In the International Gaucher registry, clinical or X-ray manifestations have been reported among Type 1 Gaucher patients in 63% and 94%, respectively. The exact pathological mechanisms leading to Gaucher bone disease are not completely understood. However, three different problems have been recognised: focal disease such as osteonecrosis, local disease such as long bone deformity, and reduction in bone mineral density.

'Treatment of Gaucher-related bone disease includes both supportive care such as orthopaedic surgery, physiotherapy and analgesia, and Gaucher-specific therapies including enzyme replacement therapy (ERT) and more recently substrate reduction therapy (SRT). Compared to the effects of ERT on enlarged organs and low blood counts its effect on established bone disease may take longer to become evident. The effect of ERT with Imiglucerase (Cerezyme) on the bone mineral density in Type 1 Gaucher disease has been studied using data from the International Gaucher registry. Bone mineral density of Gaucher patients was found to be lower prior to therapy but increased gradually over time with treatment[1].

'The paper by Pastores and colleagues[2] explores the effect of Miglustat on bone disease, in particular the effect on bone mineral density and bone pain. Data were analysed from 72 patients all of whom had participated in one of three multi-national open labelled clinical trials of Miglustat therapy. Data were collected over a period of two years during which the patients were enrolled in the trials. All patients had received at least one dose of Miglustat 100mg tds (three times a day). Any patients who had had Bisphosphonates, which may also improve bone density, were excluded from the study as were any patients who had had a combination therapy with Miglustat and Cerezyme simultaneously. Of the 72 patients who were included in the analysis, 41 of them had previously received enzyme replacement and 31 were naïve to any Gaucher-specific treatment. Twenty patients had previously undergone splenectomy.

'The average age of the patients at the beginning of the study was 41 years. Thirty seven patients were male and 35 female. At the beginning of the study two thirds of patients had a low bone mineral density, two thirds had bone pain and almost 1 in 7 had history of bone crises, 1 in 5 had had surgery to the hip and 1 in 8 had avascular necrosis. The proportion of these patients with each of these bone-related issues was higher in the group who had previously received ERT which is probably a reflection of the overall severity of their Gaucher disease. Over the two year period of Miglustat therapy the proportion of patients without any bone pain improved from 54 to 86% in previously untreated patients and from 24 to 81% in patients previously treated with ERT. Results were similar in patients who had had removal of their spleen. No new bone crises, avascular necrosis or fractures were reported during the two years of follow up.

'The effects of Miglustat on bone mineral density were assessed by DEXA scan of the spine and hip The assessments were reported as a Z score which is the difference between the patient's bone density and that of a healthy person of the same age and sex. All patients had a scan at the beginning of the study and at least one follow up scan. During the time of this study the bone mineral density Z scores improved. This was seen as early as six months after starting Miglustat. The changes were maintained over the 2 years of the study. Patients without a spleen and those with very low bone density at the beginning, considered to be at high risk of bone loss, also showed improvements in their bone mineral density scores.

Possible Impact of Miglustat

'The data in this paper therefore suggests that Miglustat may have a beneficial effect both on bone pain and bone mineral density in patients with Gaucher disease. This paper adds useful knowledge to what is known about the effects of Miglustat on blood counts and enlarged organs and also suggests a further potential treatment option for patients with Gaucher-related bone disease.

'Pastores and colleagues emphasized the length of time required to observe changes in bone density in response to enzyme replacement therapy. They quote two studies one of which suggest that improvement of the bone density may require up to eight years of treatment and another where significant improvements are only seen at 4½ years[1;3] . Other studies show reduction in bone density in patients without a spleen after two years of ERT[4]. These studies, which were performed at

different times and in different patient groups, cannot be compared directly with the Miglustat study; however, it is intriguing that the response is seen at a comparatively early stage of six months in patients treated here. These results require confirmation. The early effects of Miglustat may suggest that there the effect of the drug on sphingolipid metabolism also affects bone turnover. This hypothesis requires further investigation.

In Summary

'The authors are open regarding the limitations of this study in that it is a pooled analysis of data and the original intention of the studies was not to examine the effect on bone metabolism. Nonetheless, it provokes challenging questions regarding the causes of Gaucher-related bone disease and potential methods of therapy. Data was not presented in the paper regarding whether there was any difference in those patients previously treated with enzyme therapy or not. It would also have been of interest to see the responses in those patients with combination therapy of miglustat and ERT since this is an important and frequently asked question of physicians and researchers in the field. The improvements in bone density observed in this study should encourage the design of further studies aimed at understanding the pathology and improving the outcome of bone problems in Gaucher Disease.'

References

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3. Ciana G, Addobbati R, Tamaro G et al. Gaucher disease and bone: laboratory and skeletal mineral density variations during a long period of enzyme replacement therapy. J.Inherit.Metab Dis. 2005;28:723-732.
4. Schiffmann R, Mankin H, Dambrosia JM et al. Decreased bone density in splenectomized Gaucher patients receiving enzyme replacement therapy. Blood Cells Mol.Dis. 2002;28:288-296.

Editors Note:

The information in this paper re-emphasises the importance of the National Bone Study which we plan to report on next year.

A Tool to Help Gaucher Patients' access Insurance

The Gauchers Association and the Genetic Interest Group (GIG) have been working together with the insurance industry to create a template to help patients' with Gaucher disease access life insurance. Melissa Hillier, Communications Manager at GIG describes the project to date:

'Insurance is an issue that is often raised by members of the Gauchers Association. Members highlight the difficulties and obstacles that they have in obtaining insurance and are keen to try and find a more consistent and reliable approach.

'GIG has been working with the insurance industry for a number of years to try and establish common ground and to work out how they can help patients with genetic conditions to obtain the cover they need without having to pay disproportionate premiums. Clearly some patients affected by some genetic disorders will not be eligible for insurance for various reasons, however, there are many others who due to their treatment programmes, have their condition well managed and under control.

'One of the difficulties that GIG often hears about is that insurance companies do not have any prior knowledge of the genetic condition the patient is affected by, consequently it can take a long time for them to make a decision as to whether or not insurance can be granted as further evidence maybe required by the insurance company.

What happened next?

'GIG began by looking at Life insurance and several workshops and meetings were held with Life insurance underwriters and actuaries as well as various patient organisations to establish current practices and experiences. Clearly there were difficulties on both sides: people felt that some insurance decisions were unfair and at the same time the insurance companies were finding it hard to obtain the information that they required. A tool to help both parties was needed and work began to create a ***'Supplementary Template of Information'*** that could be completed by the potential client and their doctor for sending with the usual application form.

Pilot project for Life Insurance

'Following these initial developmental stages GIG has been working with four of its members in producing templates for their specific conditions being; AMEND – the charity for patients with multiple endocrine neoplasia disorders, The Cystic Fibrosis Trust, The Haemochromatosis Society, and The Gauchers Association. The conditions were chosen for this pilot as they all have very different effects and patients have varying degrees of health, although for the majority, treatments have been developed that stabilise symptoms of the conditions. The ***Supplementary Template Information*** was checked and revised following comments by all parties until a format it was agreed that could be piloted with insurance staff.

'It was agreed that certain areas within the template should remain unchanged for all applicants, such as the description of the condition. The individual applicant is then able to explain how the condition affects them personally and a qualified health professional has to sign the form before it is submitted to the insurer.

'Template forms have now been drafted that hold generic information about the condition in question, with space for the applicant's hospital consultant (or GP if they are knowledgeable and/or carry out a majority of the patient's care) to record relevant details about the applicant's specific diagnosis, treatment and prognosis.

'Following an initial pilot with Swiss Re underwriters, the informal working party made amendments to the template and it was then sent out to a wider range of insurance companies in order for us to gauge the feedback from each one.

'Following discussions with DOUG (the Direct Offices Underwriting Group) feedback has now been given and although there are some discrepancies in the decisions that

(continued overleaf)

A Template to Help Gaucher patients' Access Insurance

(continued from previous page)

were made on the "mystery shoppers" there was nothing alarming or extremely different between the companies and the decisions that they made. There was a general recognition that getting information early can only be a good thing. There were however, concerns at how much, in practice, the use of templates would diminish the requests for further information. This very much also depends on the amount of life insurance that the applicant is requesting.

'We will be having further discussions following this feedback to see if we can accommodate any of the suggestions and we then hope that the forms will be available to be distributed by patient organisations so that any of their members wanting to apply for insurance will arrange to have a form completed for submission along with their initial insurance application.

'The insurance industry is keen for us to look at other areas of insurance such as critical illness policies and income protection. Marketing of the scheme across the whole insurance industry will take place following the pilot, through the help of those insurers working on this project and also the Association of British Insurers (ABI), the insurance industry body.

Travel insurance

'This pilot project is focusing on life insurance, but GIG would also like to assess whether the scheme could help underwriters in the travel insurance sector as this is an area where people have encountered particular problems.

'GIG has been in contact with the travel committee of the Association of British Insurers (ABI), to see how this work could be expanded for travel policies.

'Travel insurance is provided by many different companies and is rather separate from the rest of the industry. Their methods for securing medical information with which to make underwriting decisions are phone rather than paper-based and do not include input from medical professionals. It will be a challenge to find a way to integrate such systems so that getting travel insurance can become more straightforward for people with rare genetic conditions.'

The Genetic Interest Group (GIG) is a national umbrella organisation for all people affected by genetic disorders. *GIG's main aims are to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them and to stimulate research and development of products and services that will to address unmet medical needs.*

2008 LYSOSOMAL DISEASES AND THE BRAIN CONFERENCE

Hosted by The National Institutes of Health and The Children's Gaucher Research Fund

Thursday May 29th - Saturday May 31st 2008
(conference begins 3PM Thursday - concludes 3PM Saturday)
Hyatt Regency Hotel, Sacramento, California

General Information:
A two and 1/2 day purely scientific conference that will concentrate specifically on neuronopathic lysosomal storage diseases. The conference will focus on the pathogenesis, the blood brain barrier, and the treatment of neuronopathic lysosomal storage diseases.

Program Committee and Speakers
The program committee for the conference includes:
Dr. Tony Futerman - Dr. Roscoe Brady - Dr. Gregory Grabowski –
Dr. Raphael Schiffmann and Dr. Kondi Wong.
Speakers will include researchers presenting cutting-edge research on lysosomal diseases and the brain.

Poster Boards Welcome
The 2008 conference welcomes Poster Boards that will be displayed during the entire conference. In addition, there will be a special 2-hour Poster Board Breakout session. Further instructions for the Poster Board Session and all other conference details can be found at: **www.lysosomal-brain-conf.org**

Golden Bond for the London Marathon!

Fundraising Director Alan Rosen writes; The Gauchers Association announce that we have been selected for a Golden Bond for the London Marathon. This means that we have been allocated five running places in the Marathon for five years starting in April 2008. We are very pleased that we have been given this opportunity to raise money for our activities and research.

The 2008 London marathon will take place on Sunday 13 April, and we are delighted to be able to introduce our four runners already in place (see below). We still have one place to fill and are looking for our fifth runner. Would you like to run the marathon or do you know someone who would like to do this for the Association? If so please contact us by telephone: 01453 549231 or e-mail us: ga@gaucher.org.uk

Good luck to all of our runners over the next few months with their training. As members sit in their armchair in the warm comfort of their home they should think of Nick, Mike, Liz and Lee-Ann who will be pounding the streets in training. **Please support them as much as you can**.

The Association has now established a marathon fundraising page at www.justgiving.com to make it easy to support our runners. To make a donation all you have to do is go to the Just Giving website and type in the 'Gauchers Association'. Once you get to our site you will be guided through the process.

We will report on their achievements in the next newsletter.

I am Mike and I work for Metronet, I have run for at least 20 years and have entered many races including 2 marathons. My son Nick is a student at Westminster University in Harrow studying animation. He has been running for just over a year and wanted to run a marathon. Early this year close friends of ours had told us their son James had been diagnosed with Gauchers disease (see page 3) and it gave us an idea to run and raise money for this charity. With the Gauchers Associations help we have been lucky enough to be entered for this year's London marathon and are looking forward to a successful day.

I am Liz, a 33yr old nurse from Wolverhampton. I was never interested in running or sport until about eight years ago and since then to run London Marathon has just been a dream for me! I am a member of 'Get off Your Butts' motorcycle club and was asked by one of the members, Mandy Matthews, who is a member of the Gauchers Association, if I would run London to help raise money for the association. I am delighted to have been given one of your charity places so now I'm upping my training in earnest!

I am Lee-Ann, an Australian, living in London for the past 4 and a half years. I have witnessed the huge event that the London Marathon is and I am very excited to be participating in the 2008 London Marathon for the Gauchers Association. I work within the NHS and one of my work colleague's son James was recently diagnosed with Gauchers. Although I have completed 3 half marathons, this will be my first full marathon. I look forward to fundraising for the Gauchers Association.

Fifth Runner,
could this be you?

In the following pages we provide articles updating progress on the clinical trials for three potential new treatments for Gaucher disease and also report the announcement of the collaboration between Shire and Amicus on the development of Amicus' Chaperone Therapy:

Biotech Company Amicus reports Phase 1 Pharmacological Chaperone results

Amicus Therapeutics, of New Jersey, USA presented data from their Phase 1 clinical studies of AT2101 during the recent American Society of Human Genetics (ASHG) meeting in San Diego, California. Dr Richie Khanna led the experimental program and provides 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 â-glucosidase (GCase). All individuals with Gaucher disease make some active GCase enzyme; however, most *GBA* mutations 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 called 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 contains a quality control system that controls the flow of traffic of GCase and other enzymes within the cell. This quality control system allows only stably folded GCase to exit the ER and be sent to the lysosome where it is needed to break down substrate. Unstable GCase is retained in the ER and is unable to get to the lysosome. This results in a deficiency of GCase in the lysosome and allows accumulation of substrate.

Amicus researchers are investigating the ability of AT2101 to selectively bind to and stabilize GCase in the ER. Once stabilized, GCase may be able to meet the ER quality control requirements and be transported to the lysosome where it can work to break down substrate.

Key Findings

Key clinical findings presented by Amicus researchers at the ASHG conference include:

- AT2101 was well tolerated with no serious adverse events in healthy volunteers who received single doses and repeated doses
- A dose-dependent increase (up to 3.5-fold) in GCase was observed when healthy volunteers received AT2101 for 7 days
- GCase levels in healthy volunteers remained elevated for more than 7 days after the last dose of AT2101

Macrophage and Lymphoblast Cells Cultures

Amicus researchers derived macrophage and lymphoblast cell cultures from Gaucher patients (not treated with AT2101), and tested the effects of AT2101 on cellular GCase. The key findings from these experiments were:

- Incubation of the cells with AT2101 increased GCase levels in cells derived from 62 of 63 patients
- AT2101 increased GCase levels in cells derived from Type I and Type III Gaucher patients with 19 different genotypes, including the more common mutations of N370S, L444P, and 84GG

Phase II Studies

Phase II clinical studies are being conducted to further determine the effects of AT2101 in individuals with Gaucher disease. Amicus is conducting two Phase 2 clinical trials of AT2101 for Gaucher disease. One is a 4-week study designed to evaluate the safety and pharmacodynamic effects of AT2101 in Type I Gaucher patients who will discontinue enzyme replacement therapy for the duration of the study. The second is a 6-month study designed to evaluate the safety of AT2101 and its effect on parameters that are commonly abnormal in Gaucher disease. This study will be conducted in Type I Gaucher patients who have never received enzyme replacement therapy.

Editors note: Phase I studies are limited in determining whether a new drug is tolerated in a healthy human being and identifies any side effects. Phase II studies see if it is effective and further evaluate its safety. We await progress of trials for Phase III to determine whether this new potential treatment is efficacious.

GaucherKids.org!

The National Gaucher Foundation announced last month its newest online resource: GaucherKids.org! This website has been specifically designed for kids and young adults with any Lysosomal Storage Disorder (LSD). It will allow children and young adults, ages 6 and upwards, to enjoy the freedom of having a place to call their own so that they can communicate with one another. There is a bulletin board, chat room, places to upload pictures & videos, and two "Ask A Question" forums for them to ask their questions, specifically to two young people who have Gaucher Disease.

If you know a young person with a Lysosomal Storage Disorder, please pass the web address on to them so they can share and communicate with other kids. Click on http://www.GaucherKids.org to check it out!

Amicus and Shire Collaborate on Chaperone Compounds for LSDs

Amicus Therapeutics, of New Jersey, USA has announced that it has entered into a strategic collaboration with Shire Human Genetic Therapies, Inc., to jointly develop Amicus' three lead pharmacological chaperone compounds for lysosomal storage disorders. Shire will receive the commercial rights to these products outside of the United States. Amicus will retain all such rights in the United States.

The collaboration includes Plicera(TM) currently in Phase 2 clinical trials for the treatment of Gaucher disease and two other treatments currently in trials for Fabry disease and Pompe disease.

Under the terms of the agreement, Amicus will receive an initial, non-refundable licensing fee from Shire. Joint development costs toward global approval of the three compounds will be shared equally and Amicus will be eligible to receive an additional fee from Shire if certain clinical and regulatory milestones are met.

UK Gauchers Chairman ***Jeremy Manuel OBE*** comments; "It is interesting to see collaboration between two companies both previously working on separate and potentially competing therapies for the treatment of Gaucher disease coming together on the development of one of these treatments. We hope that the results of such collaboration will benefit the patients."

Update on Clinical Trials of potential 'Shire' Enzyme

Prof Ari Zimran and Dr Debbie Elstein provide an update and presents the 36 month data on the clinical trials on GA-GCB developed by Shire Human Genetics being carried out at Shaare Zedek Medical Centre, Israel and discuss the multi national phase trials III:

'Enzyme replacement therapy has become the standard of care for symptomatic patients with type I Gaucher disease. Gene-Activated' Human Glucocerebrosidase (GA-GCB) is the native sequence of human â-glucocerebrosidase, produced from a well-characterized, human cell line using proprietary technology belonging to Shire Human Genetic Therapies (Shire HGT) (formerly Transkaryotic Therapies, Inc.). We at the Gaucher Clinic in Shaare Zedek Medical Center has served as the single center for a nine-month Phase I/II study testing safety and efficacy in 12 adult patients with type I Gaucher disease (TKT 025) and are the primary treatment site for the extension study in the remaining nine patients (TKT 025EXT).

Dosing

'Of interest is the fact that the patients in the extension trial have been serially dose-reduced to half the original dose (all currently receive 30units/kg body weight/infusion), and are continuing to show improvement in disease parameters without evidence yet of plateauing.

36 month data

'At 36 months, the mean increase in hemoglobin from baseline was 19.7%; and the mean increase in platelet count from baseline was 133.3% change. The decrease in organ volumes will be assessed at 48 months, but the reductions at 24 months were impressive. Mean reduction in liver volume from baseline (by MRI) was 26.9% and in spleen volume from baseline (by MRI) was 70.9%. Similarly, reductions in the surrogate markers chitotriosidase and CCL18/PARC (most recently assessed at 27 months) were 79.7% and 68.3% respectively.

Safety

'GA-GCB continues to be well-tolerated: no patient has developed antibodies to GA-GCB, no pre-infusion medication was needed, and no drug related significant adverse events have occurred.

Phase III

'These results encouraged us to participate in two Phase III clinical trials with GA-GCB where the important endpoints are improved hemoglobin, platelet levels and organ volumes. One (TKT 032) is a multi-national double-blinded trial) for children and adults with symptomatic type I disease naïve to enzyme therapy randomized by dosage (45units/kg body weight/ infusion and 60units/kg body weight/ infusion). The second is also a multi-national trial (TKT 034) for paediatric and adult patients who have a minimum of two years experience receiving Cerezyme who switch-over to GA-GCB (as per the patient's current dosage). Based on our data, appropriate patients for this latter trial would include those who have not achieved the therapeutic goals expected within two years of enzyme therapy with Cerezyme® (Genzyme Therapeutics, Inc.), particularly normalization of platelet counts.

In Summary

'We have been pleased with the company's willingness to provide home therapy for those patients who request it, including two foreign nationals who participated in the original trial. This option underscores the safety of the product.'

Editors Note: Shire Human Genetics recently announced that they will be conducting a **Multicenter, Randomized, Double-Blind, Parallel-Group Study** to evaluate the efficacy and safety of GA-GCB administered every other week in comparison to imiglucerase in treatment naive patients with type 1 Gaucher disease.

Protalix's new Gaucher Drug in Phase III Clinical Trial

In the December 2006 edition of the Gaucher News, Dr Einat Almon, Vice President Product Development, Protalix Biotherapeutics reported on the Phase III trial on new enzyme replacement therapy produced in plant cells for patients with Type 1 Gaucher disease. Dr Almon provides a further update:

'In August of this year, Protalix Biotherapeutics launched a Phase III clinical trial for its human recombinant Glucocerebrosidase (prGCD), produced in plant cells. Leading clinical experts in Gaucher disease from Israel, the United States, Europe and other countries are participating in this study, aimed at assessing the safety and efficacy of prGCD as enzyme replacement treatment of Gaucher disease. The study is conducted under approved FDA protocol and was initiated at the Gaucher Clinic at Shaare Zedek Medical Center in Jerusalem lead by Professor Ari Zimran.

'Thirty Gaucher patients who have not been previously treated will be divided into two treatment groups, each receiving a different intravenous dose of prGCD (30U/kg; 60U/kg). The trial is designed to be double-blind, where neither the patients nor the physicians will know the dosage received until the trial period is ended and the data is collected for all participants.

'Phase III clinical trial for prGCD was initiated by Protalix after it received FDA approval to proceed directly from Phase I to Phase III. The trial was initiated after demonstrating the promising Phase I clinical trial results, which showed that prGCD was well tolerated by healthy human subjects, with all tests being within normal ranges and with no treatment-related adverse events. The potential therapeutic use of prGCD was further supported by Prof. Anthony Futerman of the Weizmann Institute of Science in Israel, who found that prGCD, which is produced by plant cells, is practically identical in its three dimensional protein structure to Cerezyme, which is produced from mammalian cells. Biochemical studies confirmed that prGCD has equal or superior biochemical and biological characteristics when compared to Cerezyme.

'Protalix, an Israel-based biopharmaceutical company, has developed a unique plant cell culture system for the production of human biotherapeutic proteins. Its ProCellEx™ system offers several advantages over mammalian cell culture systems in terms of manufacturing costs and safety. Protalix's new state-of-the-art manufacturing and research facility meets rigorous industrial and regulatory standards. Manufacturing takes place in closed plant cell bioreactors, completely free of animal derived factors. The company believes that prGCD will be the first of many enzymes manufactured with the ProCellEx™ system for various therapeutic purposes.

'Upon initiation of the trial, Dr. David Aviezer, President and CEO of Protalix, commented: *"Based on our extensive research and committed team, we believe that we have developed an alternative treatment that will not only be effective, but also safe and cost efficient when compared to the currently available treatment."*

EWGGD Announcement

The 8th Meeting of the European Working Group for Gauchers Disease will be held at June 4-7, 2008

Budapest, Hungary, Danubius Health Spa Resort Margitsziget

For further details and registration information go to: www.ewggd2008.com

Abstract submission

All abstracts should be submitted in English, only abstracts submitted online to the congress website will be considered. After completing all the required fields, please copy your abstract to the designated space on the site. Maximum word count for abstracts: 250 words. Please only use international English characters as characters specific to your country may not be readable at our end.

Deadline for abstract submission: March 1, 2008.

Acceptance of the abstracts will be confirmed in writing by April 1, 2008.

Research Update on Neurological Aspects of Gaucher Disease

Dr Ashok Vellodi, Consultant Metabolic Paediatrician at Great Ormond Street Hospital, London gives an update on current research being undertaken and publications;

Delivery of treatment to the central nervous system.

'There have been important advances in the treatment of Lysosomal Storage Disorders (LSD's) involving the brain and several papers have been published, most of them involving some form of gene therapy. However, the drawback of these methods is they direct administration, usually in the form of an infusion delivered via a pump. Needless to say, there are considerable reservations about applying this to patients, not least of all because of the invasive nature of the methods used.

'Recently, at least two groups have reported ways of achieving this by intravenous infusion.

'The phenomenon of "gene silencing" was briefly alluded to in my talk at the NGD conference in Wembley earlier this year (see June 2007 edition of Gaucher News). The molecule used to achieve this is called siRNA (small interfering RNA). One of the disadvantages of this method is that siRNA does not cross the blood brain barrier. Writing in the July 7 issue of *Nature*, Kumar et al describe, for the first time, a way of making this crossing[1]. They achieved it by taking a small piece out of the rabies virus protein and attaching it to the siRNA. Intravenous injection of this complex into mice resulted in effective gene silencing in the brain. Importantly, no side-effects were seen even after repeated administration.

'At the Society for the Study of Inborn Errors of Metabolism (SSIEM), held in Hamburg on 4-7 September, Dr Beverley Davidson from Utah reported during the Plenary session the results of intravenous gene therapy in the mouse model of MPS VII (which is another LSD in which there is a deficiency of the enzyme beta-glucoronidase). Mice with this disease were injected with the gene attached to a special virus, AAV-4 (AAV stands for adeno-associated virus). This virus enters the endothelial cells of the blood vessels of the brain. They found increased enzyme activity in the brain.

In summary: While not directly related to GD, these are very interesting developments, as they demonstrate, for the first time, a way of getting enzyme into the brain without having to directly inject it. An important point to make, however, is that larger animals need to be treated using this method, as the mouse brain does not seem to react to foreign substances entering it, while that of larger animals does.

However, the drawback of these methods is that they involve direct administration, usually in the form of an infusion delivered via a pump.

'There were also some interesting posters displayed at the SSIEM related to Neuronopathic Gaucher disease.

Peripheral Neuropathy

'One of most interesting was a study of peripheral neuropathy in untreated type I patients. This was a large multinational prospective 2-year study involving 8 countries. A total of 103 patients with GD1 between the ages of 18-75 were recruited. Standard neurological examination and nerve conduction studies were performed. A total of 11 patients (10.7%) had both clinical as well as electrophysiological evidence of polyneuropathy at baseline. Thirty-six patients had electrophysiological abnormalities, though not all had clinical signs/symptoms. So there were patients with abnormal signs/symptoms with normal electrophysiology, and vice versa. The incidence of neuropathy was higher than in the normal population. Only four patients had a co-morbidity (such as B12 deficiency). The authors conclusion was that careful clinical examination and testing were warranted in all patients with GD1. This is an ongoing study and further data will be available at 1 and 2 years[2].

In summary: These findings are just as pertinent to GD3 as to GD1 and careful follow up for neuropathy in patients is warranted.

Lung Involvement in Type III Patient

'A 9 year old girl with type III, homozygous for the L444 mutation, whose sister had died of Gaucher lung disease, was investigated for lung disease. She had been well controlled on ERT for 4 years. Despite having no symptoms, broncoalveolar lavage (BAL- a process in which the lungs are washed out with saline) and lung biopsy showed the presence of Gaucher cells. This shows that microscopic evidence of lung disease may be present even in the absence of symptoms[3].

In summary: This is an interesting case report. It illustrates the fact that the lungs may not be as easily accessible to ERT as other areas. Nevertheless, it is not clear whether such invasive procedures should be undertaken in children, particularly (as in this case) when the authors do not explain how the findings altered the management.

Effect of Enzyme Replacement Therapy on Rare Cardiac Involvement

'An 11 year old boy from Spain who was homozygous for the D409H mutation. This combination usually results in cardiac disease which is often fatal. This child had been treated since the age of 2 months and had developed no signs of heart disease. The authors suggest that this might have resulted from early commencement of ERT[4].

In summary: This was an interesting case report, as it suggests that the cardiac involvement in these patients may be prevented or at least considerably reduced by early ERT. However, one needs to be cautious about this interpretation. A comparison with other cases in the literature would have helped to strengthen the case. Also, it is unclear how the oculomotor apraxia was measured. This is important since it was stated that it had improved over the past 2 years.

Parkinsonian Symptoms

'A 67 year old man reported from

(continued overleaf)

Research Update on Neurological Aspects of Gaucher Disease

(continued from previous page)

Portugal who developed unsteadiness and weakness at the age of 60. Closer examination revealed more neurological signs including absence of horizontal and vertical eye movements, spasticity, and Parkinsonian features. He was found to be homozygous for the N370S mutation. He had no haematological problems, and his bones were normal[5].

In summary: Patients who have the N370S mutation are thought to have no neurological involvement, so this finding is very surprising. Importantly, full sequencing appears to have been carried out. Further assessment and reporting of this patient is very important.

Brains 4 Brain Initiative

'In the last edition of the Gauchers News (July 2007), Brains for Brains (B4B) a new European Initiative was reported. The B4B group have submitted a grant application to the European Union. The title was "Restorative Approaches for Therapy of Paediatric Neurodegenerative Diseases". This bid illustrates very well the close collaboration between the members of the group which consist of doctors, scientists and patient organisations. The outcome is expected in the early part of 2008.'

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3. Djordjevic M, Minic P, Djuricic S et al. Pulmonary involvement in patient with Gaucher disease type III after 4 years of enzyme replacement therapy. SSIEM 2007, Poster 476.
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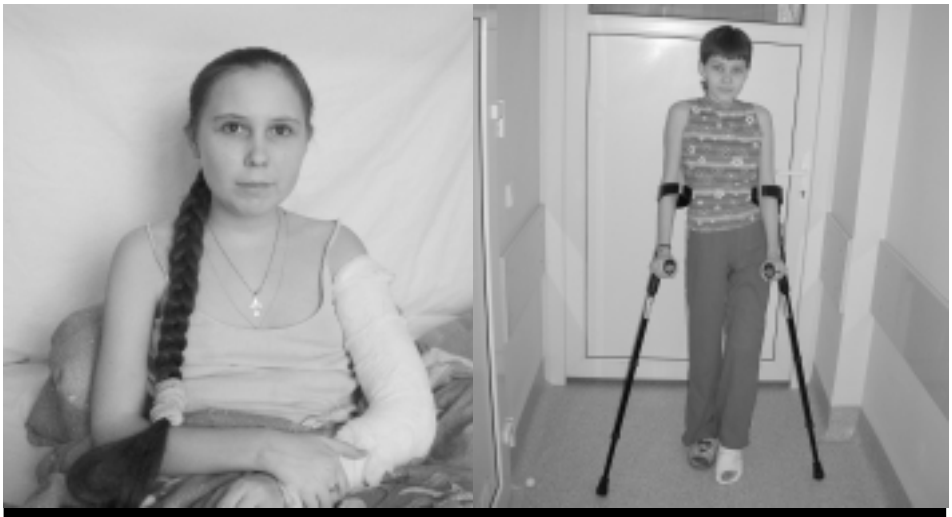
Russia Gets ERT into Ministry of Health Programme



Young Russian Gaucher patients who have suffered severe bone crisis prior to begining ERT

Marina Terekhova, President of the Russian Gauchers Association writes:

'This year has been very successful for the Association. We have managed to work with the Ministry of Health to get Gauchers disease and Cerezyme into a State programme which recognises expensive treatments for Russian people.

'On the 31st of October 2007 seven organisations (including the Russian Gauchers Association) dealing with the diseases recognised in this programme were invited to the Ministry of Health to participate in a meeting. At the meeting the State announced the amount of money available for the provision of treatments for next year.

'In the past patients with Gaucher disease had to register as disabled with the State to access treatment and for personal reasons many patients did not register as disabled and therefore could not access treatment. The new Ministry of Health programme will mean that every patient with Gaucher disease will receive Cerezyme without a disability status.

'By the end of 2007 there will be 86 gaucher patients receiving Cerezyme and in 2008 we plan to increase this to 140 patients.

'We are very proud of what has been achieved for Gaucher patients in Russia.'

NGF tribute to Susan Lewis

'Tanya Collin-Histed receives a tribute to Susan Lewis from Rhonda Buyers of the National Gaucher Foundation (NGF) at their recent patient conference in Atlanta in October 2007'

The plaque reads 'Susan Lewis: In Celebration of your life and the lives you've touched. Your passion and caring for others is your legacy to this world. Your light will shine forever.' National Gaucher Foundation

European Cerezyme Access Program (ECAP)

Written in memory of Susan Lewis by Selena Freisens

In the article below Selena Freisens of Genzyme Therapeutics describes the establishment and management of the ECAP programme which was the culmination of many years of activity by members of the European Gaucher Alliance on behalf of patients in Eastern and Central Europe who were unable to achieve treatment:

EGA Members with representatives of the Genzyme Corporation at the meeting in the Netherlands, during the Dutch Gaucher Association 20th Anniversary

From the moment of its creation, ECAP was driven by passion. It was born with passion, and was led by the passion of the people relating to it. And it became a mission for all of us involved: the mission to make a difference. This passion became mine from the first time I experienced it.

In 2003 the European Gaucher Alliance and patient representatives from Eastern Europe and the Balkan countries developed a common mission - to help Gaucher patients in those regions. In these countries reimbursement systems were not in place and treatment was therefore not available. The European Gaucher Alliance, who led the patients, had the vision and the leadership to make this mission live. And its passion was its heartbeat.

In the autumn of 2003, during the Dutch Gaucher Alliance 20th Anniversary meeting, patients and parents met Henry Termeer, CEO of Genzyme Corporation. His commitment to helping every patient, no matter their geographic location and financial situation, made way for a new programme with aim to provide treatment on a humanitarian basis for Gaucher patients in East European and Balkan countries, and, in the longer term, help build sustainable care systems for these patients. This is the European Cerezyme Access Programme (ECAP).

An independent Medical Advisory Board was created, consisting of five specialists in the field of Gaucher disease. Led by their common aspiration to help Gaucher patients, these specialists volunteer their expertise to evaluate the status of each patient in the programme and provide therapy recommendations to the treating physicians. Their dedication helped ensure the best possible care and outcomes for ECAP patients. To date, 114 patients have been approved for ECAP by the Medical Advisory Board, and around 80 patients currently receive regular yearly evaluation and treatment recommendation by the Board.

Seventy-five of these patients currently receive treatment, while five more will receive their first Cerezyme infusions in the near future. The treatment will also become available for patients in countries as far east as Uzbekistan and Kazakhstan. Twenty-five physicians from East European and Balkan countries are taking care of patients enrolled in the program. Thus, their dedication to patients has gone beyond care: many of them are working single-handedly to provide import documentation for Cerezyme, obtain necessary signatures and approvals, and sometimes even helping get the product through customs. Do that in an often bureaucratic and changing systems, that is true passion!

The commitment of Genzyme's employees involved in program application process and logistics has been crucial to make ECAP a sustainable program.While patient applications and approvals went rather smoothly, logistics often posed a challenge due to the difficulties encountered in importing humanitarian drugs into some of the countries. This required searching for other ways of getting the treatment to patients, but eventually the treatment was successfully shipped to 14 different countries in the region, either via the normal channels or with the help of organizations such as the Red Cross or the United Nations. However, shipping the treatment to some of the countries in the region still remains the biggest challenge for ECAP.

Finally, the passion of patients themselves and their families has been instrumental in helping speed up import processes and convincing authorities to support the programme.

And as our mission to support Gaucher patients in this region continues, ECAP can continue to thrive and provide guidance in the creation of sustainable care systems for patients, evolving referral and treatment centres, diagnostic centres, governmental programmes, and patient organizations. Over time, several countries involved in the programme have taken over the responsibility of reimbursement for the treatment, which has allowed some patients to transition from ECAP to receiving therapy through their own country's healthcare programme.

My Gaucher Story, by James
(continued from page 3)

We researched on the internet and met with good and very bad news…we wondered if one of the cancers might not have been easier to handle. However after a more logical approach to all the information available we formed a better understanding of the disease and the implications for James and the treatment available. James had a massive spleen and liver and moderate bone pain, involving his spine and other joints. No one wants their child to suffer and you wish you could have it yourself and not them. You feel helpless and cannot control events. However James has adapted really well and we wish we were as brave as he is. Now we are coming up to his 11th infusion and his Dad (Albie) and I will both be learning to do his infusions at home for him over the next few months? We have seen some improvement in James already and we hope this will continue. We are coming to terms with his condition and adapting our lives around it. In particular James is looking forward to a special trip to New York before Christmas with all his family.

Thank You
'We are grateful to so many people who have been a part of our journey over the past 10 months and would like to say thank you to; the GP at Simpson House, Dr Webb, Dr Hyer and Helen at Northwick Park Hospital, Dr Vellodi and Niamh Finnegan and the staff of Kingfisher Ward at GOS and to Tanya at the Gaucher Association for all their expertise, care, advice and general helpfulness.'

Genzyme and the National Commissioning Group form a Partnership

The NHS National Commissioning Group and Genzyme announced in November that they have joined together in a £7m partnership to support a specialised system of care for patients with Lysosomal Storage Disorders (LSDs).

The Partnership is to support the ongoing treatment at the seven designated hospitals in England for the care of LSD patients by The Royal Free Hospital, Great Ormond Street Hospital, The National Hospital for Neurology and Neurosurgery in London, Addenbrooke's Hospital in Cambridge, Birmingham Children's Hospital, Royal Manchester Children's Hospital and Hope Hospital in Salford.

Announcing the Partnership, **Dawn Primarolo**, Minister of State for Public Health said:

"We are delighted that we have been able to join in this Partnership with Genzyme for patients with lysosomal storage disorders and that we can jointly support patients living with these debilitating diseases. This Partnership will aid in sustaining our 'world class' commissioning of services for rare diseases through the NCG."

Paul Drohan, General Manager of Genzyme UK and Ireland said:

"Genzyme believes that the NHS has developed a highly effective model of care for patients with lysosomal storage disorders (LSDs) and that this is one of the best examples in the world of a universal healthcare system managing rare diseases. As enzyme replacement therapies are a key element of successful treatment of patients with LSDs, Genzyme is excited to have an opportunity to work closely with the NHS to support this service now and into the future."

Gauchers Association Chairman **Jeremy Manuel** said following the announcement:

"The Gauchers Association has always believed that the National Designation of the management and treatment of Lysosomal Storage disorders is the best for patients. When there were plans to dedesignate the original Gaucher service we fought very hard to ensure that it was retained and were particularly pleased that our efforts were successful and that it was extended to cover all LSD's. It is vital that patients suffering with these rare multi systemic diseases are seen and treated by experts and all efforts to support these Centres of Excellence in their internationally acknowledged work is supported and appreciated by Patients. The future will bring many new challenges including possible new therapies and the Centres need to be fully resourced to ensure that they are best equipped to face them. We share the Ministers enthusiasm for all actions (such as this Partnership) that will sustain the world class commissioning of this service which brings with it truly world class treatment of Gaucher Patients."

Nautical Nurses making Waves

As you read this two nurses from Great Ormond Street Hospital are on a long boating adventure. Metabolic medicine specialists Herdip Sidhu and Elin Haf Davies are rowing across the Atlantic together. Their journey which started on Sunday 2 December 2007 will take them 55 days to complete the Transatlantic Row. Through their gruelling efforts, they hope to raise £250,000 to help improve the lives of patients with life-threatening metabolic disorders such as cystic fibrosis and Gaucher disease. We wish them luck and look forward to their safe return in the New Year, we will report on their journey in the next edition of the Gauchers News.
You can also follow their daily progress via: http://www.woodvale-challenge.com/Atlantic+Rowing+Race+2007