

Celebrating 15 Years of the Gauchers Association

On the evening Saturday January 20 2007 the Gauchers Association's 15th Anniversary, 138 guests celebrated at a gala Dinner held at the Wembley Plaza Hotel during the week end conference.

Association Members with family and friends, doctors, nurses and representatives from the Industry attended the Association 15th anniversary gala dinner. Jeremy Manuel OBE, Chairman of the Association welcomed guests.

Keynote speaker Sir Michael Rawlins, Chairman of the National Institute for



A String Quartet from the Royal College of Musicians entertain the guests during the gala dinner

Health and Clinical Excellence (NICE) gave the after dinner address and finished his speech by congratulating the Association on what it had accomplished in its 15 years (a full report on Sir Michael's

speech can be found on page 4 of this newsletter). Unfortunately Anne Begg MP who was also due to speak broke her foot on the way to the conference so was unable to attend. Prof Cox however praised the association's work and proposed a toast to the future of the association.

During the day on Saturday 20 January, 65 delegates attended a neuronopathic patient day and listened to presentations on auditory processing; causes and pathophysiology of nGD; results from the recent Type III trial with Zavesca; the blood brain barrier; personal stories; neurological features and complications of nGD and the future of nGD patients. Families included patients from Denmark; Germany and Bulgaria.

On Sunday 21 January the main conference took place 'Studies, Treatment and Costs – Preparing for the Future'. Prof Ari Zimran from the Shaare



Guests at the Gala Dinner enjoy the evening

Zedek Medical Centre in Jerusalem gave the opening presentation. Prof Timothy Cox, Dr Patrick Deegan, and Dr Atul Mehta from the UK national Gaucher Centres all gave presentations. Other speakers included Hanna Hyry of Cambridge University, Dr Edmund Jessop from the National Specialised Commissioning Advisory Group (NSCAG) and personal stories from Dan Brown a patient with Type 1 Gaucher disease and Jo Bardoe the mother of Mia Bardoe who has Type III Gaucher disease.

A full report of all the conference presentations can be found in the Gauchers Association 'Conference Supplement' included with this newsletter and can be viewed on the CD attached. In addition you can view the conference in full on the GOLD website at: www.Goldinfo.org. Go to the 'Education & Information' section and follow the instructions.



Families listening to the presentation on neuronopathic Gaucher Disease



Young Gaucher patients enjoy the childrens activity programme

Gala Dinner Speaker Sir Michael Rawlins

A Gala Dinner was held on Saturday 20 January 2007 at the Wembley Plaza Hotel to celebrate the 15th Anniversary of the Gauchers Association. The guest of honour and keynote speaker at the Dinner was Prof Sir Michael Rawlins, who has been Chairman of the National Institute for Health & Clinical Excellence (NICE) since its formation in 1999, Sir Michael is also the Chairman of the Advisory Council on the Misuse of Drugs and an Honorary Professor at the London School of Hygiene and Tropical Medicine, University of London, and Emeritus Professor at the University of Newcastle upon Tyne.

Gauchers Association Chairman Jeremy Manuel OBE, in welcoming Sir Michael thanked him for agreeing to be with the Association on their special anniversary and for his support and encouragement during the NICE appraisal into ultra orphan diseases ensuring that the patients view was represented at all times.

Below is the text of Sir Michael's speech to the diners;

'It is both a pleasure and a great privilege to be invited to join you on this special occasion.

'Although I have been practicing medicine for 40 years, I have never looked after someone with Gaucher disease. The reason is because it is very rare. It is described in small print in most medical text books. But one of my old teachers once said "if you have a disease that is small print in the text books it is large print to you" and this is my point. Just because you have a rare disease you are no less worthy of high quality care by doctors, nurses and the NHS than anyone else. And it is a point that the Gauchers Association has made over the years forcefully and effectively.

'Despite my own lack of clinical experience of Gaucher disease I have played some part in its treatment. In the early 1990's, I was Chairman of the Committee on Safety of Medicines (the body responsible for advising on which new drugs should be put on the market and whether existing drugs should stay on the market). During my Chairmanship an application was made to licence Ceredase as a treatment for Gaucher

disease. I remember it vividly for three reasons:

- It was a novel and ingenious approach to treating the condition.
- It appeared to be remarkably effective.
- Only a very few people have been treated with it and far fewer than we would have normally expected. But we recognized that the rarity of the condition made it impossible to expect large numbers.

'One particular concern, though, was that Ceredase was manufactured from human placentas. It needed 20,000 placentas to treat one patient for a year. Our worry was that viral infections might be transmitted to patients. We recognised that the benefits to the patients seemed to outweigh these theoretical risks, so we licensed it. In the event our worries were groundless as no viral infections have ever been passed on. And now the new recombinant product, Cerezyme, has replaced Ceredase which rules out the possibility of viral contamination. Ceredase and now Cerezyme have provided very real benefits to patients.

That might have been the end of my involvement with Gaucher disease and enzyme replacement therapy but, two years ago, NICE were asked how it might approach the evaluation of drugs generally for the treatment of very rare diseases. As part of the investigation we decided to use Gaucher disease and its treatment with Cerezyme as an exemplar. We chose Cerezyme because it



Sir Michael Rawlins

had already been used in Britain for 10 years and studying it now would be non-threatening to its continued availability under the NHS. In other words, whatever conclusion we might draw, no one was going to withdraw its use on the basis of our findings.

'Although still unpublished by the Government, we believed (and still believe) that expensive treatments for very rare diseases should be made available under the NHS. This conclusion would I believe safe guard the position of people with very rare disease and, as importantly, offer encouragement to scientists and pharmaceutical companies to continue to develop new treatments. In conducting our enquiry we had enormous help from physicians like Prof Timothy Cox, Genzyme Pharmaceuticals who manufacture Cerezyme, and of course the Gauchers Association.

'Your Association has done an enormous amount for its members. It provides massive support for patients and their families; it has been extremely affective in ensuring the views of patients are heard – and acted on – at the highest levels of the NHS and Government; and it played a critical role in helping us at NICE in developing our approach to the provision of treatment for very rare diseases generally.

'So in paying tribute to the Association, and congratulating you all on what you have accomplished, I ask you to raise your glasses to the 'Gauchers Association'.

Fifth Alan Gordon Memorial Award

On Sunday 21 January 2007 at the Association's 15th Anniversary Conference, Robert Gordon, son of the late Alan Gordon the Association's founder Treasurer, presented Professor Hans Aerts with the 5th Alan Gordon Memorial Award in recognition of his sustained and distinguished contribution to the understanding and treatment of Gaucher disease and his commitment to patient welfare.

In presenting the award Robert Gordon said: 'Professor Hans Aerts is well known to the Gaucher Association, as he is to patients, patient associations and investigators throughout the world.

'Professor Aerts is the cofounder and chairman of the European Working Group for the Study of Gaucher's Disease, the only body that holds truly international meetings independent of particular companies, but his real job is that of Full Professor of Biochemistry in the University of Amsterdam Medical Centre in The Netherlands. This is a very senior academic post to which he was appointed at a very young age.

'Professor Aerts is distinguished for many discoveries in the science of Gaucher disease - a field to which he has dedicated his entire independent professional life.

'He is credited with the first full purification of the enzyme that is deficient in Gaucher disease, using an antibody that was developed by him and his then mentor in the Netherlands, **Professor Joseph Tager**. This reagent was given free to the Genzyme company and was instrumental in the commercial production of Ceredase- the first and dramatically successful treatment available for Gaucher disease.

'Professor Aerts has made numerous other discoveries of direct value to Gaucher patients and his group is well known for discovering biomarkers which are in wide international use for monitoring responses to treatment. It can be fairly stated that over the last 10 years, Professor Aerts' group has made a series of the most original and research findings in Gaucher disease of any laboratory in the world; many of these have been of direct application to clinical care.

'But Professor Aerts is much more than an inspiring biochemist in the backroom: he has always been engaged with patients and

doctors in the fields of Gaucher disease. Indeed he is a true father figure to the international Gaucher community- and above all to patients.

'As a result of his leadership, we have had the publication of guidelines for the treatment of those cases where Gaucher disease affects the nervous system; the position statement on miglustat at a time of great controversy -and now a further review with guidelines for the treatment of patients safely in the home environment- to ensure access to patients throughout Europe. He is always clear that patient representatives should be full participants -and co-authors- of these important policy documents.'

Thank you

In accepting the award Professor Aerts said: 'Thank-you for this award, it really means a lot to me, as mentioned Gaucher Disease has become part of life as well. In 1983 I was still a Chemist, a Biologist and I had never heard of Gaucher disease when Professor Joseph Tager called me up and offered me a one year position to purify Glucocerebrosidase, the enzyme deficient on Gaucher Disease and I got stuck. In one weeks time on the 28 January (in 1882) Philip Gaucher first described Gaucher disease and I am today in 2007 with you celebrating your 15th Anniversary.

Contact with Patients

'In the laboratory where I worked I came into close contact with patients and patient association and in my very first week two Dutch families with Gaucher Disease visited the laboratory and took the initiative to set up the Dutch Gaucher Association. Over the next year every three months there was a patient meeting in the lab and then later at another venue and I had a lot of contact with a growing number of Gaucher patients in the Netherlands. In 1983 there



Robert Gordon son of the late Alan Gordon presents Prof Hans Aerts with the Alan Gordon Memorial Award

was no therapy for Gaucher disease and therefore we had very intimate contact with patients. It was this contact and the development of treatment that inspired me to go on and start the European Working Group for Gaucher Disease (EWGGD).

'This prize is very important to me. One of the patient associations that have driven most of the development in Gaucher disease is your Association in the UK. I would like to express my deepest admiration to Susan Lewis and Jeremy Manuel in bringing forward Gaucher disease on an International level as well.

Gaucher Disease at the Highest Political Levels

'Two days ago on Friday morning one of our highest officials, the National Minister for Insurance systems and the former Defense Minister who is responsible for distributing money amongst academic medical institutes visited my University. The Dean of the University selected the topic of Gaucher disease to have presented; and this man knew about Gaucher disease and was aware of the treatment and expressed the need to find similar approaches in the treatment of other rare inherited metabolic diseases.

'Awareness in the Netherlands of Gaucher disease is very good and I hope that it is the same here in the UK. The Gaucher Association has played a very important role in raising the awareness of Gaucher disease and therefore this prize is extremely valuable to me. Thank you very much'

Gaucher Art Day

On Friday 1st June ten families with Type III Gaucher disease visited Careology's head quarters in Oxford to see some of the paintings which now take pride of place around their offices. Tanya Collin-Histed reports;

At the Gauchers Association's 15th Anniversary conference, children were asked to paint pictures for Homecare provider Careology; seven of these paintings have now been framed.

To celebrate the displaying of the paintings Careology invited families to their Oxford offices. Guests arrived at 11.30am and after refreshments the children were entertained by an entertainer, had their faces painted and did some magic. They were then taken on a tour of the offices viewing the paintings along the route and staff explained the homecare process from prescription through to delivery and nursing services. After lunch there was more



entertainment for the children during which the parents were able to catch up with each other.

At 1.30 the ice cream van arrived and after everyone had enjoyed an ice cream they boarded an open top bus for a tour around Oxford. The sun was shining and everyone enjoyed the ride.

Returning to Careology at 3pm the families left and made their way back home.

This event is part of the Association's commitment to support families by arranging for them to get together in a non medical environment to share experiences and strengthen friendships amongst parents and patients. The Association would like to thank the staff at Careology for their warm hospitality.



Gaucher patients and their families at the art day in Oxford

Tea at the House of Commons

At the Gauchers Association conference in January, Emily Wallrock won a raffle prize of Tea at the House of Commons with Anne Begg MP, she reports on her visit;

On 13 March 2007, I was lucky enough to visit the House of Commons to have tea with Anne Begg MP. This was as the result of a raffle prize that I had won at the Gauchers Conference in January. Anne met me in the grand reception area, known as Central Lobby, at one end of which is the House of Commons, and at the other, the House of Lords. These are distinguished by their colour schemes: red for the Lords and green for the Commons.

We had tea amongst many other MPs and their guests in the Pugin Room, which is named after the architect of the Palace of Westminster. This room has beautiful views of the River Thames.

Anne then took time out of her busy schedule to take me on a tour of Parliament, explaining many of the traditions and stories

behind the building. One of the places we visited was Westminster Hall, which is the oldest part of Parliament. The walls were built in 1097 and the hall is one of Europe's largest medieval halls with an unsupported roof. Once used as a law court, the hall has held several notable trials, including that of Sir William Wallace (1305), the Gunpowder Plot conspirators (1606) and King Charles I (1649). Today the hall is often used for important State occasions such as the Queen's Golden Jubilee and the lying-in-State of the late Queen Elizabeth the Queen Mother, both in 2002.

We were also fortunate to be able to sit for a while in the House of Lords, which that day was debating its own future and possible abolition. It was interesting to observe the speeches and arguments being



Emily Wallrock and Anne Begg MP on the terrace at the House of Commons

made. I imagine that this is a fairly unique experience - although with Parliament being such a dynamic place, similarly crucial issues are being debated and decided every day.

All in all it was a great day. Anne is an inspirational person, full of energy and leads an incredibly busy and interesting life without letting the fact that she has Gaucher Disease get in the way.

Royal Free Announces Study to investigate possible links between Gaucher and other blood disorders

Researchers from the Lysosomal Storage Disorders Unit at the Royal Free Hospital are pleased to announce a new project which explores the possible links between Gaucher Disease and another blood conditions including multiple Myeloma. Dr Atul Mehta, Consultant Haematologist at the Royal Free explains the aims of the study;

'There is evidence of a possible link between Gaucher Disease and haematological conditions where the immune cells are abnormal resulting in high levels of a serum protein called immunoglobulin. The conditions where this has been best described in Gaucher Disease are polyclonal gammopathy (PG), Monoclonal gammopathy of unknown significance (MGUS) and Multiple Myeloma. PG and MGUS are benign conditions which are not associated with symptoms and which do not require treatment. Multiple Myeloma, however, is a form of bone marrow cancer which can result in anaemia, kidney disease and bone disease and requires treatment. Although comparatively rare an increased incidence of multiple Myeloma has been documented in patients with type 1 Gaucher disease.

'Our study aims to understand the relationship between Gaucher Disease and the increased levels of

immunoglobulin as seen in PG, MGUS and Myeloma. We also will be investigating whether there is increased levels of immunoglobulin in people who are carriers of Gaucher Disease and also whether there is any evidence of Gaucher mutations in patients with MGUS and Myeloma. Other aspects of the project will examine the relevance of abnormal immune function, and alterations within the bone marrow microenvironment to the development of Myeloma in Gaucher Disease.

Dr Mehta and Dr Hughes are excited to welcome Dr Rob Ayto to work with them in this project. Dr Ayto is a haematology registrar training in North London and will work in the laboratory and clinic for 2-3 years. We are grateful to Myeloma UK for funding this post and hope that our findings will be relevant to not only patients with Gaucher disease but to any patient with MGUS or Myeloma.'



(pictured left to right) Dr Derralyn Hughes, Dr Rob Ayto and Dr Atul Mehta,

NSCAG Designation for Lysosomal Storage Diseases

On 28 March 2007 the National Specialised Commissioning Advisory Group (NSCAG) announced that the Department of Health had received Ministerial approval for the extension of the designation of the Lysosomal Storage Diseases services in England for a further four years from April 2008 to March 2012.

This announcement means that every Gaucher patient in England will continue to receive their clinical care at an NSCAG designated centre. The designation also continues the cover for the cost of providing treatment and access to homecare services nationally.

The NSCAG centres in England designated for the treatment:

Paediatric Gaucher patients are the: Great Ormond Street Hospital, London, The Royal Manchester Children's Hospital, Birmingham Children's Hospital, Addenbrooke's Hospital, Cambridge and **for Adult Gaucher patients are the:** National Hospital, The Queen's Square; The Royal Free Hospital, London; Addenbrooke's Hospital, Cambridge, Hope Hospital, Manchester

Scotland, Wales and Northern Ireland

NSCAG already has an arrangement whereby Scottish patients can be seen in the designated English centres for diagnosis and management. The decisions on funding treatment however remain with the patient's Local Health Board which is arranged through the National Services Division in Edinburgh.

Welsh and Northern Irish patients can also be seen in designated English centres for diagnosis and management, although the cost of this access and treatment are funded through the Health Commission Wales and the Northern Ireland Health and Social Security Boards respectively.

The Gauchers Association has teamed up with justgiving.com to provide a quick and easy facility to donate money online. The site is simple to use, just type in 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 will keep a 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'. If you would like more information on this please call Tanya Collin-Histed on: 00 44 1453 549231 or e-mail: ga@gaucher.org.uk

Raise funds for the Gauchers

Association whilst you shop

by using **easyfundraising**
.org.uk

www.easyfundraising.org.uk is a FREE service where you can shop at your favourite online stores and at no extra cost raise funds for the Gauchers Association. 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 cashback donation to the Gauchers Association.

For example, spend £25 with WH Smith and 3.5% will be given to the Association. In that way you will have raised £0.88, at no extra cost to your purchase. Make any purchase from Woolworths and 2.5% will be donated. Insure your car with The AA and raise £30.00, or purchase a mobile phone from Vodafone and earn £29.00.



To start please Register 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.

In Memory

Friends and family collected £620 instead of flowers at the funeral of Alfred Elliott aged 87. Alfred was the granddad of Tanya Collin-Histed (The Gauchers Association's Executive Director) and great grandfather to Maddie Collin who has Type 3 Gaucher disease. Sadly, Tanya's grandmother died four weeks later after a long battle with cancer. All money raised will go to the Neuronopathic fund.



Thanks go to:

Alan and Sharon Gardner who together with friends and family raised £750 for the Neuronopathic Fund with a pre-Christmas drinks party at their home in Essex.

Denise Stevenson, F Harman and Sue Curtis from **Hair & Son** who donated £135 for the Association instead of sending Christmas cards.

Sharon Stokes and friends who held a coffee and cake morning in the village hall in Carlby, Stamford and raised £350.

Donations raise £2090

Generous donations have been received from: Lucy Jameson, Richard Solomon, Sante Communication, Sue Cowan, Richard Serlin and Hope Samuel, Susan Sackier, The Bentley Charitable Trust, Sue Cohen, RD Cohen Charitable Trust, Redington Investments, W Sloan, G Troman, Fordeve, The Alfred & Nina Meyer Trust, Jonglore Properties, Don Tendell, Keith and Joy Moore, SRB Painters and decorators.

Motor Cycle Club raises £750 for the Association

On Saturday 21 April the 'Get off your Butts' Motor Cycle Club held their annual Charity night at Edwards St Club in Cannock. This year Mandy Matthews and members of the motor cycle club kindly agreed to dedicate the evening to the Gauchers Association and Microcephaly, both causes are associated with members of the club.

The evening was a great success with over 200 bikers enjoying lots of music and dancing. The venue was donated free of charge and a local DJ gave his time. Many local shops and businesses donated prizes for the event.

£750 was donated to the Gauchers Association.



Mandy Matthews (Pictured far left) hands over the cheque to Don Tendell

Elin Davis Completes the London Marathon in 5 Hours and 18 minutes and raises over £3,900

Elin, a clinical research nurse at Great Ormond Street Hospital braved the streets of London on Sunday 22nd April with 46,500 other runners.

Below Elin describes her experience of the day:

"We arrived at 9:38am; just in time to join the masses!"

*Mile 1
I felt GOOD!*

*Mile 3
I developed a headache!*

*Mile 5
my trainers started sticking to the road where all the spilt Lucozade was becoming warm and sticky!*

*Mile 10
I ate my warm and soggy banana which I'd carried around with me carefully so far...*

*Mile 13
I had to take my t shirt off and wrap it around my head to protect me from the sun...*

*Mile 17
I started to run with my fingers in my ears to protect me from the excruciating high noise level coming from the crowd...*

*Mile 25
I was stopped by a steward and told I had to have my race number showing... (I think it was a polite way of telling me to cover myself up!)*

... and finally I stumbled over the finishing line!"

The Association would like to express its thanks to all its members, friends and supporters... who helped Elin to raise over £3,900 for the Gauchers Association.



Elin Davies displays her London Marathon medal.

In Memory of

A Personal Tribute by Jeremy Manuel OBE

When I told David, Susan's husband, that I wanted to add a personal tribute to Susan in Gauchers News he told me that I had to keep it short as Susan did not want to be "canonised" after her death. He said that Susan had said she would only want the minimum of fuss made and that we should not be saddened by her passing.

I should have expected this because it is characteristic of Susan's humility and modesty but it gave me a real dilemma because I had so much to say about Susan from the 15 years of us working together. There are countless stories to tell, incidents to recount but the restriction forced me to focus not on Susan's achievements through the Association but on the essence of our professional and personal relationship.

The fact that Susan was super efficient, highly organised, a stickler for detail, demanding the best of herself at all times (and delivering it) is well known. Her journalistic background meant that she was rarely seen without her notepad jotting down information and action points. It was these skills that got the Association off the ground, collecting information, raising the profile, making contacts and disseminating it all for newly identified patients. But for Susan this was not just a process it became a mission. She worried about individuals, she learned their names, details of family members, medical circumstances and she would often call me to discuss a particular case. She was anxious not to offend or tread on toes and was determined for each and every individual to find the best course of action. I rarely saw her angry but when she was it was only because she had perceived injustice on behalf of a deserving patient. In the early days when health authorities would deny treatment arguing it was experimental and latterly when she feared the potential impact of the NICE review on access to expensive treatments for Gauchers patients and those with other LSD's she was stirred into passionate action.

This always led her to giving of herself. In many ways a very private person she was willing to bear her own story because she felt she had to – to protect treatment and to secure the future for others. I accompanied her to the presentation she made to the Citizen's Council of NICE when they were carrying out their review of treatments for ultra orphan diseases using Gaucher Disease as the model. I know how hard it was for her to make this presentation. It went against her natural instinct to respond to every enquiry of her health with "I'm okay" for her to publicly recount the painful and life threatening incidents in her life but she knew she had to do this for others.

And what was it like to work with her? The answer is that together we went through the full range of motions. At times it was challenging – we would brainstorm and devise strategies for action, the best way to present our case to authorities, public bodies, and industry; it could be unbearably sad and urgent when trying to help cases of very

ill people both in the UK and overseas: it could be elating when hearing that a patient had finally received treatment; it could be heart warming and inspirational when hearing of selfless acts of kindness of members and their friends and it could (and perhaps this sounds strange bearing in mind the subject matter of many of our discussions) be fun. Susan had a great sense of humour and if something made her giggle all around her would share the laugh.

We became a team working together almost intuitively, but Susan took the great burden of the work on herself. We would constantly talk. We joked that if we did not speak at least four times a day one of us had to be away and our conversations often strayed from Gauchers matters. I certainly enjoyed the intellectual debate that often flowed. We did not always agree but we respected each other's viewpoint.

From the time of the formation of the Gauchers Association until July 2005 we did everything together and when she told me that she intended to retire on her 60th birthday I knew that I could not dissuade her. With other executive members we set about the task of restructuring the Association so that we would be able to continue as we had done before. Susan told me on more than one occasion how delighted she was with the way Tanya had filled the gap. Although we no longer spoke so often as four times a day she was always available to give help and advice.

Susan was interested in people. She was concerned and cared for them and there are countless individuals in the UK and overseas who have benefited from that concern. I miss our chats, the pride with which she spoke of her sons, her niece and nephew and great niece and great nephews and her genuine delight when hearing of my children's achievements.

Susan was physically small and slight in stature but huge in personality and achievement. She was modest of the impact of her work, sometimes incredulous that "little me" as she would describe herself was standing up to Ministers of State, captains of industry or challenging learned Professors.

Professor Cox has said that she was an exemplar of the maxim of turning "private pain into public gain" and indeed the Gauchers world is a better place for Susan having lived (albeit for too short a time) in it.

We all will miss her, I will miss her clear thinking and focus on what needed to be done, her judgment, her sparkle, her concern, our debates and her advice. Most of all I will miss her friendship and her personal kindness. It has been my good fortune to have known her and to have shared so many experiences with her.

On behalf of all members and friends I send condolences to David, to her sons Duncan and Matthew and to her sister Ellen and my hope that the outpouring of affection and great esteem from around the world that has followed her death is a source of comfort to them.

f Susan Lewis



A Letter to Susan Lewis (from Joan Grantham)

Susan, this is strange. I am sitting down to write about you on one of your days - bright sunshine, blue skies and just a tiny breeze, and my first thought is 'I'll ring Susan to see what she thinks'. That's how it is without you now. The phone calls have stopped. The smile - always the smile - is in my mind's eye only. The voice 'Hello Joan, it's Susan here - how are you?' is in my head, everyday without fail, and it is all so tangible that I feel somehow you are not gone. That I will still go to King's Cross and get in your car so we can have lunch. Gin and tonic and olives for me first, kindly provided by David, under a tree in your Hampstead garden. And then, Susan, your lists of all that needed to be discussed and dealt with that day. And much was achieved - under such circumstances how could it not?

So much has been written since your death. I thought of adding to these words with a catalogue of things that we accomplished through our offices. But brevity is best. We spoke about patient issues, funding, political issues and fundraising matters with the charity dinners in College, auction of paintings and a musical afternoon and you were always there ready to help and listen. Time was not kind to you Susan and I always felt you knew it would not be, but you still gave your time freely and with such good grace.

When you rang the office you would often say that you needed to speak to Prof but you wanted my opinion first. When I gave it you always listened carefully. That's the kind of friend you always were - loyal, kind and thoughtful.

In the last winter months, when you were so ill, you wished that the sun would shine and that we could sit under a tree. You asked me to promise that in July on a visit to Seattle for my son's wedding, we raise a glass of champagne and drink to you on your birthday in the sunshine. On my visit to you in April, with Ellen and David's kind help, you spoke to me of Seattle again; you were so poorly that day but still concerned. On 14th July, Susan, the sun will shine and we will drink to you and remember you on that day of double celebration.

Thank you for everything: the thoughtfulness, the humour, the fun and most of all the friendship.

Sleep well, Susan, sleep well.

Below are extracts from numerous letters and cards received by the association and Susan's family following her death;

I feel honoured to have had the privilege of meeting Susan. When I think of Susan all I can remember is her smile which will remain with me forever.

Naheed Qureshi

I first spoke to your wife 2 years ago when our son was un-expectantly diagnosed with Gauchers disease. Susan was very kind and helpful to us, being very positive about Joseph's care and treatment that he would receive. I will always be grateful to her for answering all my questions and being reassuring.

Susan Yarrow

I was so sorry to hear the news about Susan. She was a wonderful woman who, almost single handedly put Gauchers disease on the map in the UK. She was the first person I ever spoke to who had the same disease as I did and her calm, good sense was so reassuring for me. I know I am not the only person who has a great deal to thank Susan for.

Anne Begg MP

I have such fond memories of a courageous lady, strong yet small, and with an ever present smile! She was always ready to listen, and never pre-judging, but had a sharp mind. Her care for other Gaucher patients, their families and relatives was legendary. Funny, but now that I reflect, she seldom made any reference to her own condition, instead focusing on this or that Gauchers Association issue, what a selfless lady.

Dr. Paul Schofield.

Susan was an inspiration to us all. She changed my life. She changed the lives of my husband and our five children. Without her hard work with the Gauchers association I would never have known of Cerezyme treatment and therefore would never known "normal" health as I do today. I will always be eternally grateful for all her help and encouragement.

May she rest in peace.

Susan Cowan

Tributes continued from page 11

Susan made so much difference to my life and the lives of so many others. She will be remembered in my heart always.

Evelyn Ashdown

She was a great woman, with a great soul, a sense of humour and intelligent; a sensitive woman, who was always ready to help. Her laugh and smile cannot be forgotten.

**Prof Mia Horowitz,
Tel Aviv University, Israel**

You will all be feeling this loss-such a lovely, gentle and courageous lady, whose smile was never far away.

Lesley Greene, CLIMB

She did so much to make society aware of the disease and fought for treatment. She will be greatly missed. It was her visit to my office in the late 80's or very early 90's that instigated my own involvement with research on Gaucher disease.

Professor Bryan Winchester

Susan was such a special person and I had the utmost regard and respect for her. She accomplished so much for the entire Gaucher community around the world.

**Rhonda Byers,
National Gaucher Foundation, USA**

Susan has been not only for me a partner in this journey we undertook in Gaucher disease many years. She was more. With her delicate yet assertive way of describing her illness and the hopes after the coming of the enzyme replacement therapy, she has definitively inspired my job at Genzyme. With others like Ria, Fern, Robin in the US and many more, I consider Susan as an integral part of the success of transforming Gaucher disease into a treatable illness.

They say that "the show must go on", however our "Gaucher world" will not be the same without Susan.

Carlo Incerti, Genzyme Therapeutics

I share in our collective sadness in the Gaucher community throughout the world. Susan was an inspiration and what she exemplified was precious. She had vision

that was very evident when I first met her in 1991 in Amsterdam and courage that few of us can match. Her wisdom in all matters, whether, public policy, advocacy or politics was transcendental and always with the singular goal of improving the lives of people affected by Gaucher disease not just in UK but throughout the world. It was an enormous privilege to know Susan, to be inspired by her and guided by her extraordinary wisdom. Her memory will for me and many others, be a lasting source of inspiration.

**Professor Pramod Mistry,
Yale University School of Medicine**

Susan has been for me a friend, a guide and in a way also a role model. I will miss her a lot. We all owe her many of our achievements in the struggle for the patients.

It was an honor for me to know her, to work with her and to share with her very difficult moments connected to our common "interest": Gaucher disease.

**Dr Raul Chertkoff,
Medical Director, Protalix Biotherapeutics
(Former Chairman of the Israeli Gaucher
Association)**

I was saddened to hear that Susan Lewis passed away. She was a warm, compassionate, and remarkable woman who accomplished much on behalf of the world-wide Gaucher Community.

Few have done as much for us as she did. I feel privileged to have known Susan and will miss her.

Sam Shponka, U.S.A.

We are cognisant of the enormous contribution she made for the patients with Gaucher disease, not only in the UK but world-wide. She was of great help to our society on many an occasion and indeed the British society has been a role model for ours and thus we are greatly indebted to her.

May I wish you all a long life and may you be spared any further sorrow.

**Hylton Sevitz,
South African Gauchers Association.**

Susan was a role model of fight, tenacity and devotion for all the patients with Gaucher.

She was a sensitive, generous person who

cared about all the patients and for us, the ones from Romania; she was a real support through many of the problems we encountered over the years.

She was not only enthusiastic and energetic, but also deeply caring which is shown by her tireless work on behalf of the Gaucher patients.

**George Sinca,
Romanian Gauchers Association**

Her generosity and dedication to others as well as her struggle for helping sufferers of gaucher disease will always be remembered. All members of the Israeli Gauchers Association send their thoughts and prayers to Susan's family and all the members of the UK Gauchers Association at this difficult time.

**Yossi Cohen,
Chairman of the Israeli Gauchers
Association**

I am sure that her spirit will remain with us, with her distinguished personality, human character and ability to give what ever she can for any human being, I am personally learned many things from her, she was with Fernanda who motivate me to do our JGA, so not me only but all JGA members, patients and families feel so sorry for her absence, we will continue what she started, she will be always with us.

**Dr Mohammed Antaki,
Chairman of the Jordanian Gaucher
Association**

Susan was a beacon of optimism and humanitarianism and loving kindness.

She will be sorely missed, by the people who knew her intimately, and also by those who knew of her compassion and good deeds.

She will be missed by those who only knew her by reputation as the one person in an impersonal system, who would always find time to listen and a way to be helpful.

Susan has a great lady who never let her personal issues de-rail her from her main purpose, and that was always, caring for her fellow patient.

We will always view any compassionate therapy program in the world of lysosomal diseases as a personal tribute to Susan, because Susan was the pioneer to champion the cause of treatment for all who need it.

**Prof Ari Zimran and Dr Debby Elstein,
Shaare Zedek Medical Centre, Israel**

Award Granted to Prof. Mia Horowitz of Tel Aviv University in Memory of Ellie Carter

In the December 2006 edition of the Gauchers News the Association invited research applications in the area of Type 2 Gaucher disease for consideration.

The Association are pleased to announce that after three applications were considered a three year grant has been awarded to Prof. Mia Horowitz of the Department for Cell Research and Immunology, Tel Aviv University, Israel. The project will contribute towards the understanding of the molecular mechanism underlying brain cell death in neuronopathic Gaucher disease.

The funds to support this research have been raised by the family of Ellie Carter who died on 9 February 2004 aged seven months. Ellie had Type 2 Gaucher disease. In memory of Ellie her parents Jill and Ian, set up ELF, the Eleanor Lily Foundation, to raise funds

to go towards research into understanding more about Type 2 Gaucher disease.

In accepting the research grant Prof. Mia Horowitz said "I was delighted to realise that I was the recipient of this award from the UK Gaucher association, which will support our efforts toward understanding the molecular mechanism underlying brain cell death in neuronopathic Gaucher disease. As part of this project, we intend to study what are the consequences of the endoplasmic reticulum associated degradation of the mutant glucocerebrosidase forms. We believe that the accumulation of mutant glucocerebrosidase forms in the endoplasmic reticulum of Gaucher patients with neuronopathic forms of the disease lead to unfolding protein response and cell death. We would like to test this hypothesis. During these



Ellie Carter

processes mutant glucocerebrosidase is supposed to interact with several proteins, which we would like to unravel."

Research at the Weizmann Institute in Memory of Emily Downes

Emily Downes died in 2003 aged one year. Emily had Type 2 Gaucher disease. Funds in Emily's memory to be used towards research into understanding more about Type 2 Gaucher disease.

The Gauchers Association has now awarded Prof Tony Futerman of the Weizmann Institute, Israel a grant in memory of Emily to further his research program into understanding the pathological mechanisms at work in the brains of type 2 patients, and in particular, in nerve cells.

Prof Tony Futerman said; 'There has been some dispute in

the field about whether nerve cells are themselves affected in type 2 Gaucher, but we have provided data over the past few years showing defective calcium homeostasis in neurons from animal models of type 2 GD and also in human brain (obtained post-mortem). We are continuing to define the biochemical pathways that are



Jeremy Manuel pictured on a recent trip to Israel with Prof. Tony Futerman and Einat Vitner a PhD student working in Prof Futerman's laboratory funded by the Children's Gaucher Research Fund

affected as a result of these changes in calcium homeostasis, and this award will help us to pursue these studies, with a view to eventually identifying novel therapeutic approaches'.

Bone Disease in Adults with Type 1 Gaucher Disease

Published in the Journal of Bone and Mineral Research, 2007 data from the International Collaborative Gaucher Group (ICGG) reveals the impact of treatment on bone disease. In this report Dr Suyash Prasad Senior Associate Medical Director, Genzyme Therapeutics, UK summarises the full clinical paper;

'This X ray picture is of the legs and hips of an adult with Gaucher Disease Type 1.

'It demonstrates the weakened structure and deformities of the thigh bones characteristic of Gauchers disease, along with surgical repair of the heads of both femur bones, where the hip joint is formed.

'Complications of the bones and joints are common in Type 1 Gaucher disease. There is often a reduction in bone mineral density which increases the risk for bone fractures and osteoporosis in such patients. A study by Wenstrup and colleagues¹ published recently in the Journal of Bone and Mineral Research, found that therapy with imiglucerase (Cerezyme[®]) significantly increases the bone density of adults with type 1 Gaucher disease. There is some thought therefore, that achieving a normal bone density should be a therapeutic goal for people with type 1 Gaucher disease as this may prevent serious and irreversible bone complications.

'Bone disease occurs frequently amongst individuals with type 1 Gaucher disease. This occurs as a result of Gaucher cells infiltrating the bone marrow. Bone disease in Gaucher, is a considerable cause of poor health and long-term disability. Indeed skeletal manifestations may be the chief complaint in some patients, and can have greater impact on quality of life than other features of Gaucher. Common complications of bone in Gauchers include bone pain, pathological fractures (ie fractures secondary to underlying disease) and joint collapse. In children with Gaucher, bone disease may result in poor growth and delayed development.

'The infiltration of bone marrow by Gaucher cells, results in a reduction of bone



mineral density (BMD - an indicator of the strength of bone); this reduced BMD is a typical finding in Gaucher Disease. This leads to a weakening of the bone structure, and an inability of the bony skeleton to perform its anatomical and physiological functions optimally. A technique known as dual-energy x-ray absorptiometry (DXA) is the

most widely used method for assessing bone density. It is a simple technique that involves an individual receiving an extremely small dose of radioactive isotope (less than one tenth the dose of radiation of a chest x ray); the individual then lies on a firm couch, whilst a mechanical arm passes over the body and takes an image of the hips and spine.

'To determine the effect of enzyme replacement therapy, imiglucerase, on bone density in people with type 1 Gaucher disease, Wenstrup and colleagues¹ analysed data from all type 1 Gaucher adults enrolled in the International Collaborative Gaucher Group Registry, who had bone density measurements determined by DXA. Included in the study were 342 people who received imiglucerase, and 160 people who did not receive imiglucerase therapy for Gaucher disease. The analysis found that adults with type 1 Gaucher disease have an increased risk of osteoporosis compared to a healthy population. If left untreated, this risk of osteoporosis among people with Gaucher disease ranged from approximately 10 to 30% in women and 10 to 25% in men.

'In both groups (ie those that did, and those that did not receive imiglucerase therapy), at baseline, bone density was significantly lower than the normal population. In patients not treated with imiglucerase, the BMD showed no

improvement or a slight decline over time. In those treated with imiglucerase, bone density improved over time as treatment continued. This improvement appeared to be dose-dependent. That is, those people receiving the highest doses of therapy, for the longest periods, experienced the greatest increases in BMD. The results of the analysis demonstrated that those treated with imiglucerase 60 units per kg every 2 weeks (in keeping with UK licensing recommendations³), achieved a normal bone density after approximately 8 years of treatment.

'Since imiglucerase increases bone density in people with Gaucher Disease, which may lead to a decreased risk of bone complications, achieving and maintaining a normal value might be considered an appropriate therapeutic goal for adults with type 1 Gaucher disease. In contrast to other tissues in the body, bone takes much longer to grow and the turnover of cells that make up bone is much longer. Therefore, it is reasonable to assume that improvements in bone pathology and associated symptoms, may take much longer than other manifestations of Gaucher disease.

'In the management of such patients, it would be important to perform an initial assessment of skeletal involvement by DXA and/or MRI² and annual monitoring of the effects of treatment on such parameters. As always, it is appropriate to tailor therapy on an individualised patient basis, with due consideration to all aspects of disease.'

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2. Weinreb NJ, Aggio MC, Andersson HC, et al. Gaucher Disease Type 1: Revised Recommendations on Evaluations and Monitoring in Adult Patients. *Seminars in Hematology*. 2004; 41(4 Suppl 5):15-22.
3. Cerezyme Summary of Product Characteristics

Chaperone Therapy - an update

In the last edition of Gaucher News, Amicus Therapeutics, of New Jersey, USA reported on a potential new method to treat patients with Gaucher disease. Dr. Richie Khanna led the experimental program and provides the following update.

During the recent American College of Medical Genetics meeting held on March 21-25 in Nashville, Tennessee, Amicus Therapeutics presented data from their preclinical studies of AT2101. 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 that lead to a deficiency in the lysosomal enzyme acid β -glucosidase (GCase). GCase is needed in the lysosome to break down a substrate known as glucocerebroside. GCase is manufactured in a part of the cell called the endoplasmic reticulum (ER). The ER contains a quality control system that only allows stably folded GCase to exit the ER and be sent to the lysosome to break down substrate. Unstable GCase is retained in the ER, resulting in a deficiency of lysosomal GCase and progressive accumulation of substrate.

The majority of individuals with Gaucher disease make GCase enzyme. However, most Gaucher

mutations (changes in the genetic material) may result in the production of misfolded, unstable GCase which is retained in the ER and is not sent to the lysosome to break down substrate. Amicus researchers are investigating the ability of AT2101 to selectively bind to and stabilize GCase. Once stabilized the GCase meets the ER quality control requirements, can exit the ER, and is transported to the lysosome where it is needed to break down substrate.

In order to better understand the effects of AT2101, scientists performed multiple experiments in mice that make human GCase with a mutation often found in Gaucher disease. These mice develop the following findings: decreased levels of GCase in various tissues, moderately increased spleen and liver weight, and elevated blood levels of IgG and chitin III (related to the human lab marker, chitotriosidase).

Among the key findings presented by Amicus researchers during ACMG:

- Oral administration of AT2101 to the Gaucher mice

demonstrated a significant dose-dependent increase in GCase levels in the brain, lung, spleen, and skin.

- Oral administration of AT2101 significantly decreased the liver and spleen weights in the Gaucher mice and also lowered plasma IgG and (mouse) chitin III levels.
- AT2101 interacts selectively with mutant GCase enzyme; activities of other measured lysosomal enzymes were not altered by AT2101 in any tissue examined from the Gaucher mice treated with AT2101.

Additional studies are being conducted to determine the effect of AT2101 in individuals with Gaucher disease.

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New Guide to Gaucher Disease Published

A new resource for patients with Gaucher has been produced by Sante Communications on behalf of Actelion Pharmaceuticals to help in understanding the disease and provide practical information on how to cope with it.

The information booklet, called 'Understanding Gaucher Disease', has been produced to act as a comprehensive guide to both patients and their carers. This booklet covers a wide range of topics including genetics and the

mechanism of the disease. There is a strong focus on pain management and the booklet includes a pain diary so that chronic pain can be self-monitored. It was supported by Actelion Pharmaceuticals and has been independently reviewed by the

Gaucher Association, specialist nurses and doctors.

Guidance includes information on specialist centres, diet and exercise tips, travel advice and financial support.

Copies of the booklet have been sent to all new patients contacting the Gaucher Association. If you would like a copy please contact the Gaucher Association or your specialist centre.

Nordic Gaucher Patients meet in Finland

On Saturday 28 and 29 April, 50 Gaucher patients and relatives from Norway, Denmark, Sweden and Finland met in Helsinki, Finland for the 2nd Nordic Gaucher Meeting. Tanya Collin-Histed represented the UK Gauchers Association and the EGA at the meeting. Anne-Grethe Lauridsen, Chairman of the Danish Gauchers Association reports on the week end;



Delegates at the Nordic Meeting

In 2005 Morbus Gaucher Föreningen Sweden (the Swedish Gauchers Association) celebrated their 30th anniversary with a conference in Gothenburg, Sweden. Gaucher patients from Norway, Denmark and Finland were invited to the meeting. It was a great success and it was decided to meet again in two years.

The Nordic countries have a very long tradition going back to the year 1397 of working close together. They have a common history and cultural heritage and the languages are linked.

Patients and their relatives met in Helsinki at a wonderful conference centre called Hanaholmen, situated on a small island surrounded by beautiful woodlands. Patients came from the very north of Norway - from Kirkenes, some came from the south of Denmark - from Vordingborg, some from the west of Sweden - from Borås and some from the east of Finland - from Kuopio. For some patients it was the first time they had met another Gaucher patient, others were experienced Gaucher patients and

met up with old friends.

The meeting started on Saturday after lunch with three very interesting speakers. The first speaker was Prof Ari Zimran of the Shaare Zedek Medical Centre in Israel who spoke about 'Treating Gaucher - in the past, today and in the future'. His talk took us through the history from Philippe Gaucher, who first described Gaucher in 1882 to the current licensed treatments for Gaucher disease. Finally he gave an overview of all of the clinical trials for possible new treatments for Gaucher disease.

It is interesting for Gaucher patients to look into the future and one must hope that these new drugs will address some of the unmet needs of Gaucher patients including the bone disease, the lungs and the neuronopathic form of the disease. Prof Ari Zimran spent the Saturday morning with us and many of the patients and their families took the opportunity to ask him questions and tell him about the situation in the various Nordic Countries.

The second speaker from University of Cambridge was Hanna Hyry (who is Finnish), who spoke about 'Efficiency versus Equity. Should society fund orphan drugs?' Indeed an interesting question for a Gaucher patient. Hanna gave us all some thing to think about and for sure we will remember her speech when we talk about this in the future.

Dr Markku Savolainen from University of Oulu, Finland was the last speaker of the day on the Saturday. He talk about Gaucher disease in general - the history, biochemical pathways involved in Gaucher disease, clinical follow-up of Gaucher patients and also about Gaucher disease in Finland, where there are nine patients. There is a National Gaucher Centre in Oulu, with experts in various fields of medicine (internal medicine, paediatrics, haematology, laboratory medicine, radiology). He also spoke about therapeutic goals.

On Sunday 29 April the Swedish and Danish associations held their annual general meetings. The five Finnish patients had time to talk to each other and identify the issues for Gaucher patients in Finland and the eight patients from Norway and their relatives established a board and held the first official meeting of the Norwegian Gaucher Association.

The final session allowed the four countries to talk about the future needs of Nordic Gaucher patients and how it is important to work together in the future. It was decided that a 3rd Nordic meeting will be held in 2009 in Oslo, Norway. The Chairmen of the Swedish, Danish and Norwegian associations and a representative from Finland committed to continue to work together in the future.

Bulgarian Gaucher Meeting

Tanya Collin-Histed, Executive Director of the Gauchers Association reports on the Bulgarian Gaucher Patients' meeting which took place on 11 and 12 May in Sofia;

'Dr Ashok Vellodi and I were invited by Vladimir Tomov, Chairman of the Bulgarian Gaucher Association to attend their association's annual patient meeting. To coincide with the meeting Dr Radka Tincheva, Head of the Department of clinical genetics at the University Pediatric Hospital, Sofia held a patient clinic and invited Dr Vellodi to be her guest at the clinic on the Saturday morning to examine the Gaucher patients.

In Memory

Vladimir Tomov, Chairman of the Bulgarian Gauchers Association opened the meeting on the Friday evening with a minutes silence in memory of Susan Lewis, co founder of the UK Gauchers Association and EGA who had died on Tuesday (8th May 2007). Susan had visited Bulgaria in 2000 and 2002 and was instrumental in securing ECAP aid for several Gaucher children in Bulgaria.

Presentations

'In addressing the meeting I spoke about the structure and workings of the UK Gauchers Association and the European Gaucher Alliance. Dr Ashok Vellodi made a presentation about paediatric Gaucher disease and its clinical management. His talk was followed by Prof Rumen Stefanov from the Bulgarian Rare Disease Organisation who reported on the 1st National Conference of Patients with Rare Disease in Bulgaria which took place on 14 April 2007. He told the audience that 50 representatives of patients with 16 rare diseases and five patient associations attended the conference. The conference

highlighted that although patients may suffer from different rare diseases, they all share the same experience and difficulties in their daily lives: lack of understanding from the community, lack of information, delay in getting the right diagnosis, restricted access to treatment. As a result of the conference the National Alliance of People with Rare Diseases was established which has sparked media attention. Vladimir Tomov was interviewed by the Bulgarian television which highlighted the importance of Patient associations and the need for a national strategy for rare diseases.

Future Directions

'Vladimir Tomov outlined the future aims of the Association highlighting the need for more patients and professionals working the field of Gaucher disease to become involved in the Association in order to be more effective and to improve information to patients and professionals. He is concerned to ensure that patients received treatment at the correct doses and to achieve permission to treat all adults over 18 years old. Following the evenings presentations an conference dinner was held.

'The following morning members of the patient association met to have their AGM and new members Rhadslova Tomov and Kristian aged 19 and 18 years respectively joined the committee to represent the children and young people with Gaucher disease.

Gaucher Clinic

At the same time as the AGM Dr Vellodi and I together with some families attended the Children's Hospital in Sofia to meet Dr Tincheva. In total Dr Vellodi and Dr Tincheva saw eight gaucher patients in the clinic including a young Bulgarian patient with Type 3 Gaucher disease. This patient has a number of serious complications related to Gaucher disease. Dr Vellodi has agreed to support Dr Tincheva to access additional treatment and medical care to seek to manage these complications and improve the patients quality of life.

'In the afternoon we enjoyed a walking tour of the Centre of Sofia and spent the evening sharing the experience of some families over dinner.

'Vladimir Tomov hailed the meeting as a great success. He said "the Bulgarian Gaucher Association have two new committee members, we have clear aims for the future and the sharing of information between Dr Vellodi and Dr Tincheva will help to improve the quality of life for the young Type III Gaucher patient."



Doctors and Gaucher patients outside the Children's Hospital in Sofia

GA-GCB, A New Enzyme Preparation Entering Phase III Trials

The last edition of Gauchers News (December 2006) reported on the clinical trials on GA-GCB developed by Shire Human Genetics being carried out at Shaare Zedek Medical Centre, Israel below Prof Ari Zimran and Dr Debbie Elstein provide an update on the trial;

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

'As we enter the third year of our experience in our clinic with GA-GCB, we are highly satisfied with this drug. The Phase I/II trial had 12 adult

patients with clinically significant anemia, thrombocytopenia, and spleen and liver enlargement who received 60U/kg GA-GCB every other week for nine months. Then in the extension study 10 patients, who continued treatment, were dose-reduced to 45U/kg, and again after a further 9 infusions, all patients were dose-reduced to 30U/kg.

'Infusion-related reactions and adverse events were mild to moderate at all dosages. Anti-GA-GCB antibodies were negative for all patients.

'The average change in hemoglobin from baseline to 30 months including through the reduction of dosage to 30U/kg was an increase of 18.7% in hemoglobin, an average increase of 154.0% in platelet counts, and average reductions in spleen and liver volumes at 24 months (latest time

point which MRIs were performed) of 70.9% and 26.9% respectively. Levels of the biomarkers chitotriosidase and CCL18, also decreased during the entire study.

'The results obtained are consistent with well-recognized therapeutic goals for Gaucher disease and were achieved within comparable time frames. These results also encouraged us to participate in Phase III clinical trials with GA-GCB. A multi-national trial is currently enrolling for children (above the age of 2 years) and adults with symptomatic type I disease who are naive to enzyme therapy, as well as a soon-to-be opened clinical trial involving a transition from the commercially available product to GA-GCB, again open to both children and adults who have been on enzyme therapy. The Royal Free Hospital in London under the auspices of Dr. Atul Mehta, will be among the sites that will be joining the trials.'

Additionally, these trials will be listed on www.clinicaltrials.gov.

New Book on Gaucher Disease is Reviewed

Dr Patrick Deegan, Consultant in General Internal Medicine and Metabolic Diseases at Addenbrooke's Hospital reviews the new book 'Gaucher Disease' edited by Prof Ari Zimran and Prof Tony Futerman for the Gauchers Association;

'This is a comprehensive and encyclopaedic review of Gaucher disease, from basic or "pure" science - through clinical science and practice - to the societal and ethical issues involved in treating a very rare disease.

'In 1998 a volume of Ballieres Clinical Haematology was devoted to Gaucher disease, but the book under review is not only more up to date, but much broader in scope. The editors have delivered articles from almost all of the important researchers, clinicians and interested parties in this area of medicine.

'In reading this book, several broad points struck the reviewer. Although

Gaucher disease is rare, the amount and quality of research published on this condition is extraordinary. The eventually successful attempt to find treatments for Gaucher disease has led to an intense and revealing focus on the biology of this condition, which in many ways acts as a model or prototype for other rare, "orphan", diseases.

'This book also reflects the diversity of the Gaucher "community"; basic scientists, clinical scientists, practising clinicians, the pharmaceutical industry and patients. The editors have chosen wisely, I believe, to allow this diversity to prosper in the pages of the book. The

subject matter of the chapters often overlaps, allowing different opinions to be heard. This is most relevant in the chapters that discuss the controversy relating to the ethics and costs of treatment. This section should be read as a whole and no one chapter should be regarded as definitive. The reader will find a range of opinion expressed, often in personal and polemical terms, but when taken together the section is an accurate reflection of the breadth of the debate.

'This book may be seen as a metaphorical home for the Gaucher community. All those interested in Gaucher disease, from whatever point of view, will find much within its pages that is stimulating, challenging and instructive.'

CRC Press, 2006, Hardback, 544 pages