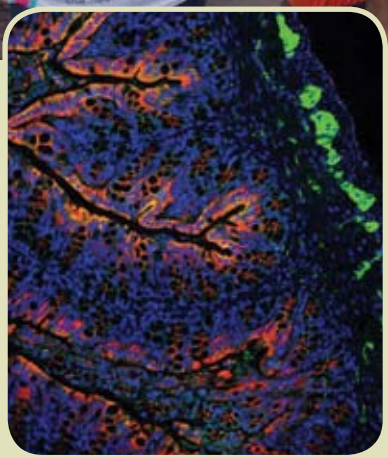
GauchersNEWS

June 2015 Gauchers ASSOCIATION

London Marathon runners raise over £12,000



Gene Therapy for Gaucher Disease

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LYSOSOMAL STORAGE DISORDER CENTRES

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Front page top photo: Atul Mehta & Dan Brown
Bottom photo: Intestine cells of mice that have received an intravenous injection of AAV

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Chairman's Chat

Dear Friends,

Welcome to the June 2015 edition of Gauchers News.

Since the inception of the Gauchers Association almost 25 years ago we have always tried to be at the forefront of the rare disease community, representing the interests of our members and looking to facilitate development around the treatment and care for Gaucher patients. We continue to strive to achieve these ambitions and in this edition of Gauchers News you will read about another significant step that has been taken in this area.

In previous editions we have reported on the progress that has been made by Dr Ahad Rahim and Dr Simon Waddington and their team at University College London (UCL) in their research studies in to gene therapy for Gaucher disease. The Association has been involved as part of this project from a very early stage providing the initial grant to Ahad and Simon which allowed them to begin their research.

The UCL team have made significant progress in their research and following proof of concept being achieved in the mouse model for acute Type II GD as well as extensive discussion between the UCL team and the Association, it was agreed that the Association would apply, as sponsor, for a positive opinion from the European Medicines Agency for orphan designation of the gene therapy as a new potential treatment for Gaucher disease.

I am delighted to say that earlier this year we found out that the application was successful. This allows us to start the process of achieving regulatory clearance for a potential clinical study into gene therapy. You can read more detail on pages 4, 5 and 6.

This is a small step on a very long journey to hopefully finding a new therapy for Gaucher disease but one we are extremely proud of and credit must go to Tanya, our Chief Executive, for leading the way and yet again demonstrating her unerring determination to help the lives of patients and their families. The Association would like to express its gratitude to Dr Elin Haf-Davies a close allies of the UK Gaucher community who has been integral in supporting the Association to take this project forward utilising her knowledge of the regulatory process and disease burden of patients and families. We look forward to providing further updates on this exciting project on our website and in future editions of Gauchers News.

Before I sign off I would like to mention the fantastic efforts of our runners in the London Marathon who raised over £12,000 for the Association – an incredible achievement. We still have places for the London 10K in July and of course our annual London to Cambridge bike ride in September so sign up soon!

On behalf of everyone at the Association, I wish you all a great summer.

Dan

Gauchers Association granted Orphan Designation for Gene Therapy

The UK Gauchers Association is delighted to announce that on February 12th 2015 the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA) granted us, as the sponsor, a positive opinion for orphan designation (EMA/OD/303/14) for adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene (Gene Therapy) as a new potential treatment for Gaucher disease.

The regulatory world in Europe can seem overwhelming since it requires careful navigation between the National Competent Authority (NCA) in each Member State and the European Medicines Agency (EMA).

To benefit from the EU regulation on orphan medicinal products for rare diseases, it is necessary to follow a centralised procedure at the EMA. This allows for the designation of orphan medicinal products and puts in place incentives for their research, marketing and development. Traditionally, it has been considered that only pharmaceutical companies can engage in this procedure with the regulators, however this is actually open to individuals, academic and clinical institutions as well as patient groups.

Tanya Collin-Histed, Chief Executive of the Gauchers Association, said 'whilst existing enzyme replacement and substrate reduction therapies are effective in treating many areas of Gaucher disease there remains significant unmet needs and challenges. No treatment is available to treat the neurological aspects of the disease which can range from eye movement and auditory processing to myoclonic seizure and premature death. Gene therapy could potentially address this by providing a cure for Gaucher disease. Although this is a first step in the long and complex process of bringing a treatment to market, we are extremely excited to receive this opinion, and to initiate the procedures that allows for communicating directly with the regulators.'

What is Gene Therapy, How Does it Work and What Does it Potentially Mean for Patients with Gaucher Disease? Simon Waddington and Dr Ahad Rahim writes;

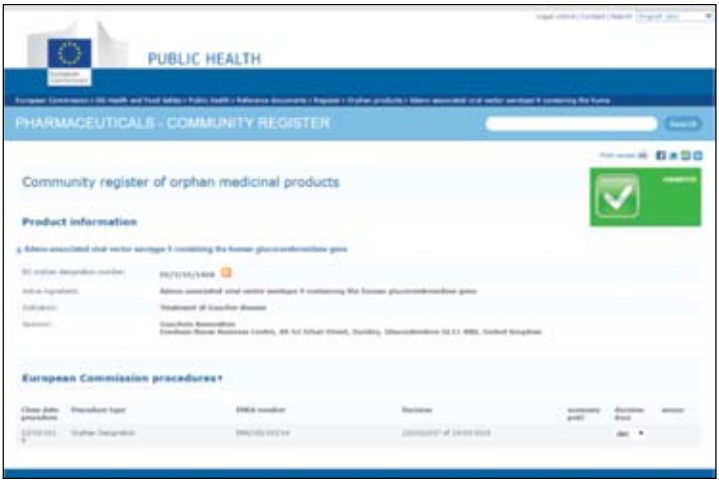
Nearly every cell in our body contains a nucleus, each containing an entire, identical blueprint of our entire body. This blueprint is encoded into long strings of DNA which are bundled up tightly into chromosomes so that they fit in the nucleus. The DNA code is subdivided into genes – each containing the code for an individual protein. Every cell contains the same number of genes (about 24,000) but actually contains duplicate copies of each gene – one from our father, and one from our mother. When we inherit these genes, sometimes from our parents, the gene copying process can introduce an error (a 'mutation')

which is so serious that the gene now codes for a protein that can't function. In many cases this is OK – the copy from our other parent allows us to make sufficient amounts of that protein. However, sometimes if we inherit broken copies from both parents, no functional protein can be made. This is what happens with neuronopathic Gaucher disease (nGD); the patient has inherited two broken copies of the gene encoding Glucocerebrosidase.

The most elegant solution would be to repair the mutation in one or even both copies in every cell in the body. The technology to achieve such efficiency is still in its infancy, however it may be sufficient if we are able to deliver working copies of the genes to a proportion of the right cells. The state-of-the-art tools with which we can achieve this are the basis of a whole field of technology known as gene therapy – delivering genetic material to cure diseases. Nature has provided us with an excellent vehicle to deliver genetic material – the virus – and it has been possible to take viruses which infect humans but do no harm, and create synthetic versions (known as 'vectors') which are designed only to deliver working copies of genes.

We have been working to develop a gene therapy treatment for nGD. To do this, we have been using a mouse model created by Professor Stefan Karlsson in Sweden where affected mice perish before 15 days of age. We injected a vector, known as AAV9, to deliver working copies of the human glucocerebrosidase gene to neurons in the brain of these mice on their day of birth; most of the treated mice lived more than ten times longer than expected and were healthy enough to breed. Given the encouraging results with mice, we are now looking at how this treatment might be given to babies diagnosed with this disease.

A session will be held at the nGD family conference on Sunday 15th November to update patients and families on this exciting development and will enable families to meet with the researchers and ask questions. We will keep you updated on our progress as we take each step forward on this long and exciting road by posting updates on our website and Facebook page and in future editions of Gauchers News.



Intravenous Gene Therapy for the treatment of nGD

In the December newsletter we reported on how our preclinical gene therapy studies are providing invaluable insights into the neuropathic forms of Gaucher disease (nGD) and supporting continued development of novel therapies. On this basis, Giulia Massaro, a PhD student supervised by Dr Ahad Rahim (UCL School of Pharmacy) and Dr Simon Waddington (UCL Institute for Women's Health) was funded by the Javon Trust, the Gauchers Association and the UCL Impact scheme for a project focusing on investigating the efficacy and safety of minimally invasive intravenously administered gene therapy.

In our study, we use a mouse model of acute Type II nGD that develops the severe neurological symptoms and does not survive beyond 14 days of age.

Giulia's preliminary findings show that administration of a viral vector carrying the functional GBA gene to a small number of new-born mice through a single intravenous injection can effectively rescue them from premature death. Rather than dying at 14 days, treated mice were still alive two months after the injection with the viral vector. Giulia has closely monitored the animals by weighing them, performing a set of behavioural tests and undertaking pathological analysis of the tissues.

Preliminary data are showing an improvement in the neuropathology. On-going experiments will confirm these promising results and they will hopefully show that gene therapy also has a positive effect in the visceral pathology. This approach would be highly desirable as a single administration of gene therapy could potentially treat both the brain and the visceral disease associated with nGD and provide a systemic treatment in these mice.

Giulia will now treat a larger number of animals and conduct further experiments to validate the data we obtained so far and collect further samples for analysis in collaboration with researchers at Great Ormond Street Hospital and the UCL Institute of Child Health. Excitingly, Giulia's work on nGD was shortlisted for the prestigious Fairbairn Award (named in memory of Lez Fairbairn, a scientist at the Cancer Research UK Paterson Institute who died suddenly in 2005 aged just 46) at this year's British Society for Gene and Cell Therapy Annual Conference 2015 in Glasgow, Scotland where she will present her data to the gene therapy scientific community.

We will report on developments in the next issue of Gauchers News.

Rob Marshall – A personal story

My name is Rob Marshall. I'm 35 and I have Type I Gaucher disease. I was diagnosed in early 2012 when I was 32 and prior to this, I had no idea of the condition or even knew anyone with a rare genetic disorder. The diagnosis came as a shock, but also as a relief to finally have a diagnosis following years of discussions with my GP and other medical professionals.

Rob completing the 2014 London to Cambridge bike ride

On receiving the news, my key concern was 'how could I continue with all that life demands whilst having ERT on a regular basis.' I've always been a keen outdoors enthusiast and I'm regularly out on the mountain bike, hiking and climbing and more recently have taken up road cycling which I thoroughly enjoy.

The Gauchers Association and the team at the Royal Free Hospital have provided a great level of support and information to me over the past three years and in 2014 I decided to take part in the annual 'London to Cambridge' cycle ride to raise funds for such a worthy cause. This was the first organized cycling event I had taken part in and I have to say it was one of the most enjoyable rides I have ever been on. It was very well organized by a very welcoming and friendly team with some brilliant cake stops thrown in along the way for good measure! What I really liked was the fact that there were so many different people taking part, from keen cyclists to those who just turned up and rode to Cambridge. The route was very scenic and easy to follow and the sense of achievement at the finish line was great.

It's a great way to support a great charity and raise awareness of the condition. It would be great to see more people on the ride this year on Sunday 6th September so come and join us and give it a go!

Rob with his son Jack

We've moved!

Our new address is:
8 Silver Street, Dursley
Gloucestershire GL11 4ND

Our telephone number and email address have remained the same:
01453 549231
ga@gaucher.org.uk

Like us on Facebook

"like" us on facebook

*Don't forget to visit and like our Facebook page at **www.facebook.com/TheGauchersAssociation**. As well as keeping you up to date with news and events we hope to encourage greater awareness of Gaucher disease and the work of the Association.*

Alison Wilson – Gaucher in Ireland

Alison Wilson

It's been a while since I last updated you on how our work has been progressing in Ireland so please read below for a long-overdue progress report! As you know, the Gauchers Association has had a presence in Ireland for a while and I have had some contact from patients and families who have needed support and to whom I have gladly been able to help but this invitation is open to everyone who could use some support, advice or further information about Gaucher disease.

Please don't hesitate to contact me if there's anything at all I can do for you. You can call me on 028 950 47779 or 07786 258336 or email at alison@gaucher.org.uk.

As well as providing individual support to families here in Ireland, I have also represented people with Gaucher disease in a number of forums including the Northern Ireland Rare Disease Partnership and The Rare Disease UK Management Committee. I feel that it's important to have a voice at these forums because it ensures that everyone in the wider rare disease community understands and is aware of the needs of Gaucher patients and their families.

In the last few months I was delighted to attend the Gaucher Leadership Forum in Berlin and also a Metabolic Study Day in Dublin. Both meetings provided an excellent opportunity to learn more about Gaucher disease and I could also network with specialists working in the field. I'm still on a steep learning curve and I try to grab every possible opportunity to learn more about Gaucher disease and how it impacts those who live with it. I am now making plans to pass on this knowledge by organising a study day in Belfast that will look at the diagnosis and management of a range of Lysosomal Storage Disorders (including Gaucher disease). We are delighted to have secured funding through the Gauchers Association to support Gaucher families in Ireland (both north and south of the border) to attend this study day and to meet each other to eat, chat and socialise. I will be in touch with all our known families in Ireland with details of this event nearer the time but if you know of anyone who you think would like to join us, please pass my details onto them.

When tackling a condition like Gaucher, knowledge is power! I would encourage all those reading this to seek out opportunities to learn more about Gaucher and to talk to people who are living with it, both as patients and their friends and families. My experience is that those who are more informed and who have support from others in similar situations often find difficult times much easier to manage.

Again, if there's anything at all I can help you with, please ask!

Patient & Family Support Worker report

Helen Whitehead

I've been in my role for over a year now and it has been great to have met so many of you around the UK and to have spoken to so many of you on the phone, via email or on Facebook.

As always, if you do have any unmet support needs, you can call me at the Gauchers Association from Monday to Thursday on 01453 549231; you can call or text me on my mobile on 07795 192311 or alternatively you can email me at helen@gaucher.org.uk. You can also find me on Facebook by searching 'Helen Whitehead' and you'll know it's me because you'll see the Gauchers Association logo as my profile pic. If you'd rather meet in person for a coffee and a chat, I can arrange to come and see you at home.

The type of support I can offer varies, from helping people with benefits applications including PIP (Personal Independence Payment) and ESA (Employment and Support Allowance). Where these applications are concerned, my advice is to get in touch early as the whole process takes weeks! Even if you just would like someone to check through the (lengthy) form I am happy to help.

I have supported patients with their hospital visits; accessing home adaptations; liaising with homecare teams; appeals for schools; helping to complete college applications and attended Gaucher clinics at Addenbrooke's and Birmingham Children's Hospital.

Irma Shah and Emma Rooney

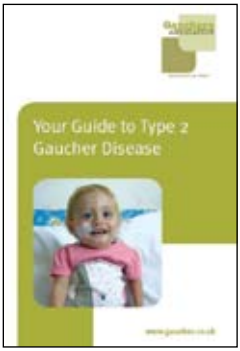
In November 2014 I accompanied Irma Shah, as part of her role as a Patient Ambassador with Shire, to their headquarters in Switzerland. Irma spoke about her story and I was able to give an overview of my role as Patient and Support Worker to everyone at Shire.

In December I visited Healthcare at Home in Burton-on-Trent and in March I visited BUPA in Harlow. I was able to have a look around their facilities and the dispensary so I could see how the delivery system works. It was interesting to see how the homecare systems work and to build links within both companies.

Helen Whitehead attending Genyme's Rare Disease Day

In February this year I attended an event at Genzyme's offices in Oxford for 'Rare Disease Day' where I was able to give the staff at Genzyme an insight into my role in patient advocacy.

An updated Type II Gaucher disease booklet is now available, both printed and as a PDF and I worked alongside Dr Annupam Chakrapani, Paediatric Consultant at Great Ormond Street Hospital to create the booklet. If you'd like a copy, please contact me.



That's all for now but please, if you have any issues you would like to discuss, or you just need a friendly ear, please don't hesitate to contact me. I'll look forward to hearing from you soon!

Welcome to Claire Lightfoot

The Gauchers Association would like to welcome Claire Lightfoot to the team!

"Hi, I'm Claire and I've just joined the team at the Gauchers Association as an Administrative Assistant and I'll be working part-time on Tuesdays and Wednesdays.

I've previously worked in both the banking and retail industries but now I'm really looking forward to the challenges of my new job and I'm excited to learn more about Gaucher disease and to get involved with the great projects and fundraising activities coming up this year! I hope you are too and I look forward to meeting a lot of you over the course of my time with the Association."

Type III GD: The Empowerment Project

The Empowerment Project gives young people with Type III Gaucher disease a voice and will empower them to shape their own futures and the futures of those living with Type III Gaucher disease.

They will get the opportunity to develop their own individual skills and to also be part of a group looking to improve the lives of all patients with Type III GD. This year we are looking for further funding to enable this project to continue over the coming years. The group is co-ordinated by Helen Whitehead (the Patient and Family Support Worker) and supported by Association Chief Executive Tanya Collin-Histed, Niamh Finnegan (Clinical Nurse Specialist) and Dr Derralynn Hughes (Adult Consultant) and we meet three times a year. We now have our own Facebook group which helps with communication as we are based all over the country.

7th nGD Conference

At our meeting in March we continued to plan the 7th nGD Conference to be held 13th–15th November in Manchester (see page 14 for more details). We have chosen the venue, designed the programme of speakers, had input into the children's programme and many of the group have chosen to speak at the event. The fact that they are volunteering to speak shows how much their confidence has grown!

The girls and Niamh during a meeting

Clinical Trials

The group were interested in finding out more about clinical trials as it's possible they might be asked to participate in the future; to learn the importance and understanding the process and what their commitment would involve. Over the last couple of meetings we have started looking at clinical trials and how they are devised. Dr Derralynn Hughes writes:

'During the Empowerment Project the Type III girls have been exploring the role of clinical trials in drug development. During the process of bringing a drug from a scientific concept to a medicine which can be prescribed, a number of phases of clinical trials must be performed. In our discussions we have explored how a clinical trial might be designed to answer an important question around whether the drug works, is safe and in some case which of a number of drugs may be most beneficial.

We used the example of an imaginary clinical trial exploring the difference between fully caffeinated and decaffeinated coffee. We designed entry criteria for the study

worked through the type of assessments which may be performed, the objectives and the possible end points. We discussed how a similar process occurs in the design of a drug trial. We have subsequently also explored how a patient-reported outcome might be designed and how this might be helpful in assessing the effects of a new drug.'

Overseas Clinics

We are supporting members of the group to attend overseas Gaucher clinics alongside professionals from the UK. Irma Shah attended clinics in India. Here is her report:

'In January 2015 I had the amazing opportunity to visit India with Dr Ashok Vellodi, Dr Alex Broomfield (Royal Manchester Children's Hospital), Niamh Finnegan (Clinical Nurse Specialist) and Tanya Collin-Histed (Gauchers Association) to attend Gaucher and MPS clinics.

We spent two days in Chennai and attended one Gaucher clinic and one MPS clinic. It was a great experience and I have learned a lot from being part of these clinics. I have Type III Gaucher myself and attend clinics. However, the clinics in India are very different to the ones we have in the UK. For example, in the UK they are very private and patient confidentiality is very important whereas in India there are more people in the room other than the doctor, patient and parents.

Irma Shah and Shriya from Delhi

We travelled to Delhi and visited AIIMS Hospital where the clinics were being held. Again, the clinics were very different to the UK. Patients and families sat in one big room and waited to be seen by Dr Vellodi and Dr Broomfield.

Irma Shah and Tanya Collin-Histed during a clinic

After this trip I have realised that I am fortunate to receive Enzyme Replacement Therapy and to have access to home care. Patients in India have to travel a very long way to have treatment or attend clinics. I had an opportunity to speak with some families in India and talk about the challenges they face and for them to ask me questions. I hope to carry on meeting with families from different countries and hearing about their experiences.

I really enjoyed India and we also had some time to see the sights and explore when we were out of clinics. Travelling overseas has made me more independent and confident than I was before.

7th nGD European Family Conference

We are pleased to announce that the 7th nGD European Family Conference will take place on November 13–15th 2015 at the Park Inn Hotel in Manchester and is open to patients, families, healthcare professionals, the pharmaceutical industry and Homecare representatives.

As always, the conference will be packed with some important and interesting presentations by many of our stakeholders, including topics such as Genetics; the latest research and information on the on-going clinical trials; looking to the future and living with Type III Gaucher disease; education, including primary, secondary and higher education as well as the role of the patient and the family support workers.

As well as these great presentations and talks from some of the world's leading experts in Gaucher disease, you will also hear some very heartfelt, personal stories told by patients, families and many people directly affected by Gaucher disease.

We also include a full children's programme of information and entertainment to the under 16s with our amazing volunteers, all of whom carry full DBS checks.

Costs are as follows:

Family (up to two adults and three children) **£150**

Single family member **£50**

Gaucher patient (over 18) **£25**

Pharmaceutical & Homecare representatives (Saturday only) **£150**

As always, places are filling up quickly so to reserve your space and for all the information you need, please call Sarah in the office on **01453 549231** or email **sarah@gaucher.org.uk**.

International Gaucher Day

Following on from the successful first International Gaucher Day (IGD) last year we are pleased to announce plans for this year's event are well underway and will take place on 1st October 2016. 'Rare but not alone' will remain the official slogan of IGD.

IGD is the brainchild of the European Gaucher Alliance (EGA) based on an idea from the participants at Go with Gaucher in 2012 (a project working with young Gaucher patients).

The EGA have decided to change the date for this year to avoid summer holidays, thus dovetailing the start of Gaucher Awareness month in USA.

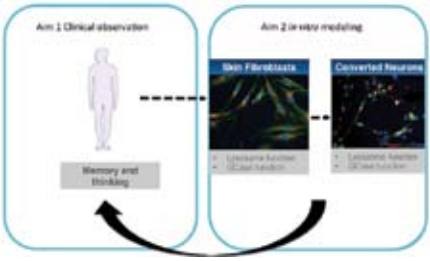
Once again the UK Gauchers Association will be participating in this event by asking our members to celebrate IGD and will be sending packs, please look out for these! During your celebrations, don't forget to take pictures and send them to us, these will be shared in the December edition of Gauchers News and the EGA website.

Further information about this event will be posted to our website and Facebook page, please visit www.gaucher.org.uk.

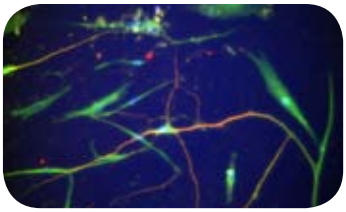
2014 celebrations

Investigating connections between Gaucher and Parkinson's

After the report in the last newsletter, Lucy Collins, a PhD candidate working alongside Professor Timothy Cox at Addenbrooke's updates on her study on the associations between Gaucher and Parkinson's. She writes:



'I would like to sincerely thank everyone for their participation and for your patience with our assessments and support for our research. Some have taken part in our memory and thinking tests and some in our skin biopsy tests. We'd like to inform you of our progress and the main outcomes so far in both parts of the study whose aim is to better understand the association between Gaucher and Parkinson's.



Skin sample

What have we been doing with your skin samples?
We have been working on developing a new personal cell model to better understand the underlying cause of Gaucher and Parkinson's. This is done by creating nerve tissue from cells taken from a small skin sample. These cells are valuable as we can use them in the test tube to investigate conditions that occur in the brain; we can also test various drugs for potential benefit using this patient-specific model. The image shown on the left is an example of a neuron (in red) made from a skin sample. This research is fairly new and we have been making attempts along with experts in these techniques to produce these nerve cells from skin. One expert is Dr. Tim Sargeant working in Adelaide, Australia. He intends to use these cells to further our understanding of lysosomal biology in the development of Parkinson's disease.

Aims of the memory and thinking tests
In parallel with our cell work we are looking at memory and thinking in people. We wish to see if there are patterns or signatures of differences in how problems are thought through, solved and then remembered. These tests are ongoing in clinic and similar tests will feature in a new study called Gaucherite (see page 21).

If you would like more information about the study or to take part, please contact me on lmc58@cam.ac.uk. '

Parkinson's research update from the Royal Free

We know that Gaucher patients and carriers of the Gaucher gene are at increased risk of developing Parkinson's in later life. There are excellent treatments for the symptoms of Parkinson's, but the aim of the UCL-Royal Free group is to develop a treatment to slow the development of Parkinson's and possibly prevent it.

Thanks to all for their time and samples. Your help and support are essential and have been invaluable in allowing us to progress. Below is a summary of our research.

Ongoing Studies

Potential Treatment for Gaucher-associated Parkinson's
Research – mostly based on skin cells – shows that the drug ABX reverses accumulation of substances that lead to Parkinson's in certain cells. This has gone so well that we soon hope to begin an exploratory drug trial to test whether the changes 'in the test tube' can be replicated in patients suffering from Gaucher-associated Parkinson's.

Identification of the Earliest Signs and Symptoms of Gaucher-associated Parkinson's
Over the last six years we have assessed and taken samples from Gaucher patients and their families to look for early symptoms and chemical markers of Parkinson's. We have a much better understanding of the course of Gaucher-associated Parkinson's and our ultimate aim is to identify (and treat) the earliest stages of Parkinson's in those carrying the Gaucher gene before it progresses.

Future Plans

RAPSODI GD
This is the first step towards identifying patients for a major drug trial to prevent Gaucher-associated Parkinson's. We will recruit over 1,000 Gaucher patients and family members, assessing them yearly using an internet portal which detects the early signs of Parkinson's. We will recruit more participants to understand more accurately which patients develop Parkinson's and, crucially, why. We envisage that the 'high risk' individuals we identify will be invited to participate in a future drug trial.

Exciting Times Ahead
Everything we have achieved couldn't have been done without your help. We hope we can count on your continued support as we pursue our ambition towards discovering a drug to stop Gaucher-associated Parkinson's.

Dr Ashok Vellodi recieves 8th Alan Gordon Memorial Award

In February 2015, the AGM committee awarded the eighth Alan Gordon Memorial Award to Dr Ashok Vellodi from Great Ormond Street Hospital (GOSH) in recognition of his significant contribution to Gaucher patients and their families for over 20 years.

In February 1994 Dr Vellodi joined Great Ormond Street Hospital London as a Consultant Paediatrician in the Metabolic Unit. Since then he has been a lead clinician in developing the hospital as one of the initial four Gaucher Centres of Excellence in England designated by the Department of Health in 1997.

Dr Vellodi developed a special interest in neuronopathic Gaucher disease (nGD) and was the Chairman of the 'Task Force on Neuronopathic Gaucher Disease, European Working Group on Gaucher Disease' which published the 'Management of Neuronopathic Gaucher Disease: A European Consensus' in the Journal of Inherited Metabolic Disease in 2001 which was revised in 2009.

Dr Vellodi with Dhillon family

As a member of Genzyme's European Cerezyme Access Programme (ECAP) Medical Advisory Board, he was instrumental in bringing treatment to many children and adults with Gaucher disease in Eastern Europe where their Governments didn't have reimbursement programmes for the treatment of the disease.

He not only used his experience and expertise in the UK but he has travelled overseas making presentations on Gaucher disease, lecturing trainee doctors, supporting doctors in clinics to develop their knowledge of the disease and highlighting the challenges of patients and doctors in countries like India and Pakistan where there is little or no reimbursement for treatment.

Dr Vellodi studied medicine in India and makes bi-annual visits back to work alongside the dedicated physicians who look after the Gaucher patients, many of whom are very sick. He supports the physicians in developing their knowledge of the disease and uses his experience of how to best manage the manifestations, particularly without access to Enzyme Replacement Therapy (ERT).

Personal Tribute from Tanya Collin-Histed

Gauchers Association Chief Executive has known Dr Vellodi since 1996. Her daughter Maddie was diagnosed with Type III Gaucher disease when she was 17-months old and he has been her consultant for 18 years. Here is her personal tribute:

'I first met Ashok in January 1996. He was sat in the corner of a room in the oncology department at Great Ormond Street Hospital whilst Maddie's dad and I were told that she didn't have cancer but probably a rare disease called Gaucher. We were also told that Dr Vellodi was an expert in the condition. I remember it like it was yesterday. It was a day that changed our lives forever.

Over the past 19 years Ashok has become a friend and has supported me not only as a parent, but also as a vital source of inspiration and encouragement to become a patient advocate that has enabled me help support families and patients over the years and work alongside doctors, researchers, other patient's advocates and pharmaceutical companies to improve the lives of Gaucher patients all over the world.

I have been extremely privileged to travel around the world with Ashok seeing patients and families who have looked to him for answers. He has such compassion and respect for the patients and doctors in the countries that face many challenges, including Pakistan, India, Jordan, Bulgaria, Greece and Serbia and I see him at his best in these situations.

Jeremy Manuel, Irma Shah presenting Dr Ashok Vellodi with his award

It was almost impossible to pick one or even two events or achievements to share to demonstrate what his contribution to the UK and global Gaucher community has achieved since there are just so many. However for me, the two key events are firstly that he is the UK and global lead on the clinical management of GD 3, providing advice to doctors and patients all over the world and secondly, his commitment to his work in the field of humanitarian aid with an emphasis on Eastern Europe, India and Pakistan.

I would also like to say that as a parent of a sick child, life has had huge challenges but he has always been there for his patients. They have been his only focus and I know that he has had to fight many battles for all of them and for this we thank him and will be forever grateful.'

Cont'd overleaf

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Retirement and Saying Goodbye

Sadly, Dr Vellodi retired from clinical practice at Great Ormond Street Hospital in February this year, however he will continue his research role at the hospital until February 2016.

The Gauchers Association and many of the families whom Dr Vellodi has looked after over the years gave him a surprise party in March to thank him in person for his compassion, kindness and dedication to them. On behalf of the UK and Global Gaucher community we would like to wish Dr Vellodi a happy and relaxing retirement.

Dr Vellodi writes; 'To describe a journey of 21 years in a few sentences is never easy; even less so when I think of where the road has taken me. I have been privileged to have got to know many members of the Gaucher community in the UK and abroad. There have been some outstanding clinicians and scientists. However above all I have got to know many wonderful families; I have never ceased to be amazed at their resilience and fortitude through some very difficult times. I have been privileged to have been able to share something of this with them. Throughout it all we have worked together, we have shared good times and bad. I hope that I have been able to repay, in some small measure, the faith that families have had in me. I was very touched to see so many families who had given up their time to be there that evening and I am very grateful to the Association for having organised it. It was a very emotional evening for me. To see children whom one has looked after for nearly two decades, now grown up into confident young adults, and their smiling parents; no paediatrician could ask for more.

I was quite overwhelmed to have been presented with the Alan Gordon Memorial Award. I would like to dedicate it to the families I have worked with over the years. I have many memories to cherish and hope to be able to keep in touch with as many of you as possible. And of course, I will always be available to serve this community.'

Gaucherite study:

Neurological Disease – What does Gaucher Disease do to the brain?

Dr Aimee Donald

We are pleased to announce that the Gaucherite Study has started recruiting patients! This study aims to better understand the UK population of patients with Gaucher disease with a view to developing new and better treatments. One aspect of the research will focus on bone disease and another will focus on neurological disease (brain involvement).

In February this year Dr Aimee Donald, a Paediatrician working in Manchester, UK joined the Gaucherite Research Group as the Paediatric Clinical Research Fellow. Aimee has an interest in neurological disorders in children and will use this interest to focus on neuronopathic Gaucher disease (nGD) and the neurological features seen in some Type I patients, in particular Parkinson's disease.

Aimee will meet and examine all patients enrolled in the study (including adults) as well as asking them to answer a series of questionnaires. The aim is to understand the differences and similarities between patients which might help us to understand why and how Gaucher disease can affect the brain. Some of the questionnaires will look at how the disease affects patients' quality of life and day to day functioning while others will look at whether it affects mood, memory, thinking and sleep.

Aimee explains why this work is so important: 'both children and adults with different types of Gaucher disease are affected in different ways. At the moment, we have an idea of which children will have difficulties affecting their brain function but the extent of those difficulties is never clear at the time of diagnosis. We also don't have any good, effective, treatments for this aspect of the disease. If we can understand the differences between patients with different types of neurological disease we might be able to predict disease progression and develop new treatments.

We also know that there are aspects of life which patients struggle with which haven't been described in research before. If we can identify these features we can look at ways to improve how we manage them; for example, difficulties with thinking or fatigue. We are beginning to learn about the relationship between Gaucher disease and Parkinson's disease which is another significant area of research. Parkinson's disease affects a small number of patients with Type I Gaucher disease and I hope to be able to predict more accurately which patients this will affect and therefore allow earlier intervention in this disease process.'

If you are interested in taking part or would like further information about this study, please speak to your clinical team at your specialist centre.

Patients & representatives get expert R&D training

The European Patients' Academy on Therapeutic Innovation, or EUPATI, is funded by the Innovative Medicines Initiative, Europe's largest public-private initiative which aims to speed up the development of better and safer medicines for patients.

Its fundamental goal is to provide scientifically reliable, objective, comprehensive information to patients on medicines research and development. Another key focus of their incredible work is to increase the capacities and capabilities of well-informed patients and patient organisations so they can be more effective advocates and advisors on current research into medicines, including but not exclusively in clinical trials, with regulatory authorities and in ethics committees.

Out of over 300 pan-European applications, we are delighted to say that our Chief Executive Tanya Collin-Histed was one of just 50 to be selected to undertake a 14-month course involving e-learning and face-to-face events. Upon completion, she will have the knowledge to make meaningful contributions to patient empowerment and advocacy and to contribute to the broader dialogue on patient involvement in medicines research and development across Europe.

The course runs from October 2014 to December 2015 and is a commitment of at least 250 hours of e-learning study and 8-10 days (including travel) to attend face-to-face training sessions with the experts at EUPATI.

Tanya writes: "Although challenging and a huge time commitment, through this learning I have increased my knowledge on what goes into medicines research and the complex development process. I have already been able to use it in my role, understanding terminology and the numerous stages of development and regulatory challenges and it has improved my confidence in dealing with the many stakeholders I encounter in my role as Chief Executive of the Gauchers Association and the European Gaucher Alliance (EGA)."

Shire expert meeting

Shire's spring expert summit entitled 'Gaucher Disease – Driving Excellence in Patient Care' was held on 17–18 April 2015 at the Novotel Amsterdam City Hotel in Amsterdam, The Netherlands. It was chaired by Professors Ari Zimran and Hans Aerts and attracted over 115 physicians, researchers and healthcare professionals from 28 countries.

Gauchers Association Chief Executive Tanya Collin-Histed was invited to make a presentation highlighting the Association's work in improving the quality of life of its members and Gaucher patient and families in the UK. She writes:

At the summit an entire session was dedicated to highlighting the 'Quality of Life' of patients. The inclusion of this topic is often talked about but very little time is given to discuss these very important aspects at these professional meetings. In my presentation, I outlined the work of the UK Gauchers Association and how improving the quality of life of our members is the core of our Association and underpins and shapes everything we do.

Emma Rooney, a young woman with Type I Gaucher disease talked about her own personal experiences and gave a powerful message of the impact of diagnosis, how it affected her family, her dreams and how it has helped shape who she is today.

Dr. Gregory Pastores, Associate Professor of Neurology and Paediatrics at the NYU School of Medicine in New York gave a physician's perspective on assessing quality of life in clinical practice and how important it is to recognise the whole person.

Anat Oz, a nurse from Israel spoke about the value of providing home therapy to Gaucher patients and how this can improve the quality of life for both patients and their families.

Dr. Debbie Elstein from Shaare Zedek Medical Centre in Israel described her work with other international physicians on developing and validating a new quality of life tool in GD which is sponsored by Shire. I [Tanya Collin-Histed] am the CEO of the European Gaucher Alliance (EGA) and the patient representative on this working group. Currently all quality of life assessments that are carried out as part of on-going clinical management or within clinical trials use generic quality of life questionnaires and they don't capture the important aspects of the impact of Gaucher disease on patients. They are usually brief snapshots of the last seven days and therefore do not often give a true picture of the burden of disease and/or treatment on patients.

We will continue to update members in future editions of Gauchers News on the roll-out and use of this new quality of life tool for Gaucher disease.

The Susan Lewis Memorial Award

The Susan Lewis Memorial Award was created to provide bursaries to doctors and healthcare professionals from developing countries to travel to the UK Centres of Excellence for mentoring and educational programmes.

Following a review, it was extended to support a wider range of initiatives and to encourage the education of doctors in Gaucher disease. Since the last Gauchers News, the Association has supported clinical nurse specialist Niamh Finnegan and Paediatrician Dr Alex Broomfield to travel to India to attend two Gaucher clinics and present at a metabolic meeting along with Professor Timothy Cox at the 3rd Rare Disease South Eastern European Meeting in Macedonia.

Tanya, Irma and Niamh

Niamh Finnegan writes; 'I was delighted to be invited by Dr Vellodi to visit India in January to attend clinics in Chennai and New Delhi and to a meeting with the India Lysosomal Storage Disorders Support Society (LSDSS) as well as patients and families. I successfully applied to the Susan Lewis Fund for support for my trip and I am grateful to the Association for approval and for allowing me to participate in an inspiring and amazing experience.

On Monday we had a clinic at Mediscan. Dr Sujata and her team gave us a warm welcome and as this was my second visit I was delighted to meet some of the staff I had met three years earlier.

Monday was the Gaucher clinic and we saw many patients and families with Gaucher disease. Some had travelled hundreds of miles and their dedication to their children and desperation for advice and treatment was inspiring and humbling.

On Tuesday we saw mainly MPS I, II and VI patients in the multi-disciplinary clinic. Many were not on ERT and desperately needed help and advice. It was an honour to meet them and hear their stories but it was also heart-breaking.

Wednesday we flew to Delhi and went to AIIMS for a clinic. Here we saw a mixture of patients with LSDs including Gaucher and MPS and children with other metabolic conditions. Many families attended the clinic, held in the boardroom.

We then met with families and the LSDSS and I spoke about how we give ERT in the UK focusing on home infusions. Families and professionals were interested from a financial saving standpoint and also to make treatment more accessible. It was a wonderful experience to meet so many families.

Tanya, Irma and I also had the amazing opportunity of visiting the Taj Mahal. We also had a tour of Delhi and saw all the famous landmarks. Even the memories of the 'life flashing before my eyes' auto rickshaw journeys which left me traumatised at the time now make me smile!

I will never forget the experience and I am very grateful to Dr Vellodi, the Association and the Susan Lewis fund for enabling me to go.'

Dr Alex Broomfield writes; 'Given my personal connections with the Indian sub-continent – my father was born in Nowshera, I spent three years in Delhi as a teenager and did voluntary work in Himachal Pradesh – I needed little incentive to jump at the chance of helping in clinics there. Given this was possibly my last opportunity to work clinically with Dr Vellodi, a true pioneer in the treatment of Paediatric Gaucher, the chance was too good to pass up. I am deeply grateful to for the support from the Association for enabling me to do so.

The first day clinic in Chennai encapsulated and included everything I had hoped for from this trip. In addition to the childhood flashbacks of the rickshaw drivers, once at the clinic the majority of the patients had Type III Gaucher disease, indeed were mainly homozygous for the L4444P mutation. Dr Sujata and her team look after patients on ERT and those who have not had the opportunity as sadly, application for compassionate use programs is still limited. It was sobering for me, whose practice has fallen in these years of increasing therapeutic options to see the severity of some of the visceral manifestations and a stark reminder of the importance of a nationally-funded treatment program. It was clear that the opportunity to discuss with Dr Vellodi and to meet Irma and Tanya was deeply inspirational to the families; some of whom have weekly trips of up to two days on a train to receive their infusions.

The rest of the trip passed at breakneck speed, a clinic split between myself and Dr Vellodi due to the number of patients on day two was again a testament to the hard work of all the specialties involved in the care of the patients. While the drive of Manjit Singh of the LSDSS and the inventiveness, intelligence and humanity of the staff at AIIMS in Delhi made a deep impression, my fondest memory comes from the evening on the last day back in the centre of Delhi. While drinking saffron-flavoured tea, I observed Dr Vellodi's the bartering skills, whose knowledge of pashminas was surprisingly extensive! I end by taking this opportunity to thank the rest of the group for helping make this such a memorable trip and as I settle back into work in Manchester, I thank both Ashok and Niamh for the encouragement and advice they have given me over the years. I hope this and what I have seen on this trip will stand me in good stead for the future.'

3rd Rare Disease South Eastern European Meeting in Macedonia

The 3rd Rare Disease South Eastern European Meeting, organized by the Macedonian Academy for Science and Art was held on November 15th 2014 in Skopje, Macedonia. This meeting is dedicated to bringing news from the academic community in EU to Macedonia.

The Association of citizens for rare diseases 'Life With Challenges' is supporting this congress as the cooperation between the patient and the academic community is very important for progress in providing a better quality of life for patients with rare diseases.

We see such meetings as a chance to network and strengthen cooperation with the academic community, medical professionals, industry, patients and government institutions.

In the area of Gaucher disease there were two lectures, one commercial - on the subject of Eliglustat from Genzyme, and one on management of Gaucher disease from Professor Timothy Cox. As a patient organization we appreciated the opportunity to have such a distinguished guest as Professor Cox, as we are aware of the need to raise awareness on disease management, especially for a rare disease such as Gaucher, of which 10 patients in Macedonia are affected.

'Life With Challenges' used this opportunity to have a Gaucher family dinner, and all of us together (families, doctors, pharmaceutical representatives and academics) had a nice evening with Professor Cox who talked to everybody and tried to answer all our questions.

Professor Cox writes; 'It was an inspiration to see what Ms Vesna Aleksovska and just a few colleagues have achieved already in Macedonia - not only for Gaucher patients (special to Vesna's heart) but applying this experience for the benefit of patients with other rare conditions in need. It is not always easy to live in this part of the Balkans.

By sheer persistence and much strategic planning within the rare disease initiatives of the EU, Vesna and her colleagues have created links at the highest political level to promote equitable access to treatment and proper care.

Vesna, a key member of the European Gaucher Alliance, had the foresight to ensure the engagement of local physicians in the best standards of international practice; this is not easy when the healthcare is not an obvious source of personal advantage for cash-strapped doctors with other diverting priorities.

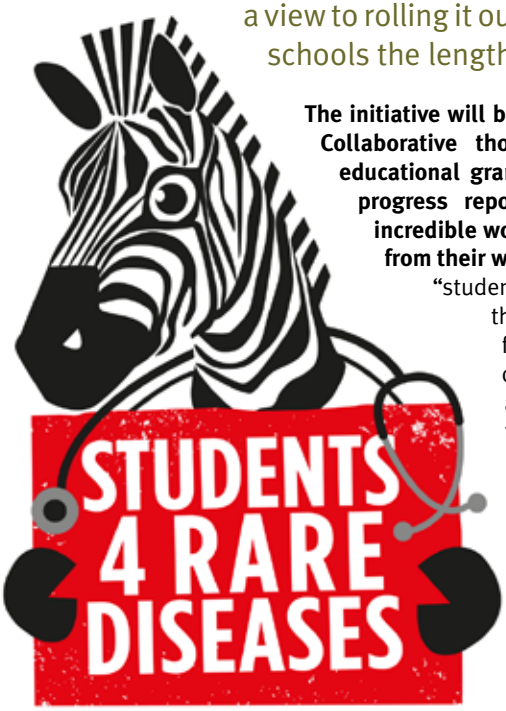
That all ten known Macedonian Gaucher patients can flourish with sustained delivery of enzyme therapy is little short of a miracle. They and their families were very interesting to meet over dinner; they all appeared to be in such good health and Vesna herself is now expecting her first baby!

We should not forget that this happy outcome is a tribute to many people working within, and alongside Genzyme in the former Soviet states. Over more than a decade, the Emergency Cerezyme Access Programme (in response to a fierce challenge led by the late Susan Lewis with the EGA) is a tribute to what can be achieved through genuine partnerships between companies and the communities of patients they serve.

I thank the Association, and in particular, the Susan Lewis Memorial Fund, for supporting this visit. Macedonia is a small and diverse country, but even this short visit provided a big education. It left a lasting impression of the power of humanity and of those who never give up, but also, just what selfless work it takes to achieve anything worthwhile for those in need.

If you Hear Hooves it may be a Zebra...

Continuing on from the success of the 'If You Hear Hooves, It May Be A Zebra...' project, the guys at Students 4 Rare Diseases (www.students4rarediseases.org) are employing a Development Officer to support the committee to take the project forward with a view to rolling it out to a whole host of medical schools the length and breadth of the UK.



The initiative will be supported by the UK LSD Patient Collaborative though support, advice and some educational grants and we look forward to future progress reports. If you aren't aware of the incredible work these guys do, this is an extract from their website:

"students4rarediseases is a committee that connects rare disease societies from different medical schools in order to share speaker information and advertise events. We originally founded Barts and The London Society for Rare Diseases in 2011 and expanded the project in the form of S4RD in 2013 so that we can help students in other medical schools to get involved. Our main aim is to create a network in order to pool resources and information about rare diseases."

Visit the UK LSD Patient website at

www.lsdcollaborative.org.uk

How the Association works alongside the NHS on behalf of Gaucher patients

Gaucher patients in England are managed at one of eight centres of excellence. The Gauchers Association works closely with all of the centres and attends the regular Expert Advisory Group (EAG) meetings chaired by Edmund Jessop, of the Specialised Commissioning Team NHS England to support the development of the service and to ensure that the patient voice is always heard.

These meetings, held two or three times a year, bring together physicians from all eight centres and members of other patient organisations from the various LSD charities. The focus of the meetings is to discuss the management of the LSD Service, the treatment options, research, treatment protocols and future service developments. Over the past six months there has been a lot of work going on and Tanya Collin-Histed, the Association's Chief Executive reports on the work that she has been involved in:

NHS England consultation on how Specialised Services are commissioned

In England the NHS is responsible for commissioning treatments for patients. Under new arrangements which came into force in April 2013, in order to get the NHS England to pay for a new medicine, (which is not subject to NICE) it has to be developed through a number of different pathways and committees. Finally, a group called CPAG make the final decision before it is rubber stamped for financial approval and then made available to patients.

In October 2014, a 10 year old boy with Morquio syndrome (MPS IV) challenged NHS England with High Court legal action challenging the scorecard system used for deciding which lifesaving treatments to commission, highlighting that there is no published policy explaining its use and that there had been no public consultation over its use. As a result of this challenge NHS England launched a 90-day consultation about how it will prioritise which specialised services and treatments to invest in. Once the consultation closes, then there will be a further 90 days public consultation before CPAG can make a decision. Meanwhile there is no interim process for funding much needed treatments and patients remain uncertain about if and when they will be able to access these treatments.

The Gauchers Association alongside many other stakeholders have made a formal response to the consultation on behalf of its members.

Developing Disease Protocols

This NHS initiative will seek to develop disease protocols to ensure that all high cost drug treatments relevant to that disease or drug are used in the most clinically and cost effective way. It will also look to grow the clinical evidence base for the use of high cost drugs in the treatment of these conditions.

Gaucher disease was one of the first diseases to go through this process. Membership of the group that pulled together these protocols which included paediatric and adult protocols for ERT and SRT as well as being broken down by Type I and III, consisted of an NHS Commissioner, an adult and paediatric doctor, a senior pharmacist and Tanya Collin-Histed of the Gaucher Association.

Once the protocol has been finalised the treating clinician completes a protocol for each patient that will be uploaded onto a new NHS system acting as an audit for the service. Clinicians will be required to regularly update the patient's protocol.

Patient/Charter Agreements

Members of the UK LSD Patient Collaborative will be working with NHS England over the coming months to develop these agreements which will be issued to all new and existing patients. It will explain what a patient can expect from the treating centre and the patient's responsibility in return for receiving clinical management and treatment of their condition. A letter will be sent to all patients prior to their hospital appointment informing them they will be issued with this agreement enabling them to bring along representation/support if they wish.

Homecare Contract

Patients with an LSD who receives ERT are supported by a national homecare service, allowing them to receive their infusions at home. The service enables patients to become independent infusers through training or to be semi or fully supported by a trained paediatric or adult nurse. This is managed through each of the eight hospital trusts who work with the appointed homecare company to meet the needs of their patient cohort.

The first contract (2012–2015) attracted four homecare companies onto the framework; one company – Medco – withdrew in 2013 and another – Central – were unsuccessful in recruiting patients onto their service. This left two – Healthcare at Homecare and BUPA Homecare – to service the needs of LSD patients throughout England.

The current contract ends in October 2015 and over the last year representatives from each of the eight treatment centres and Tanya Collin-Histed as a representative of the UK LSD Patient Collaborative have been working alongside staff from the NHS's Commercials Medicines Unit to develop a new contract to take the service forward. It is anticipated that a number of new companies may tender to be part of the LSD Homecare contract which will drive up quality and choice for patients.

The contract will describe the level of service that patients should receive from their homecare company and set out guidelines and working practises that the companies should adhere to that ensures safety and quality to patients.

Gaucher Leadership Forum

The Gaucher Leadership Forum (GLF) takes place annually and brings together clinicians and researchers from all over the world to discuss outstanding needs and challenges in the Gaucher community. This year's meeting, entitled *'New Clinical Science of Gaucher Disease'* took place in Berlin on the 27th and 28th March and attracted 223 participants from 49 countries.

Tanya Collin-Histed writes: There were a number of important topics on the two-day agenda, including:

Scientific Steering Committee and speakers

Professor Tony Futerman of the Weizmann Institute of Science in Israel spoke about Potential Therapeutic Targets for nGD and how neuronal death occurs in nGD is not known. What is known is that cell death can occur due to inflammation and by altering the pathway using a mouse model exhibiting many of the features of the neuronopathic human disease which can cause this results in dramatically improved visceral and neurological disease in the mouse model of Gaucher disease. This suggests that these might act as potential therapeutic targets.

Ricardo Feldman of the University of Maryland: School of Medicine in Baltimore, USA talked about what we might learn from induced pluripotent stem cell models of Gaucher disease. Pluripotent stem cells are reprogrammed fibroblasts from patients and studying these cells provides researchers with an opportunity to explain the molecular mechanisms that underlie the pathophysiology of Gaucher disease. The information that emerges is that normal glucocerebrosidase may be involved in many fundamental cellular processes, including developmental pathways, and maintaining a functioning pool of lysosomes.

Professor Claus Niederau from Germany gave an overview of modern management of Gaucher disease. Today there are three (two in Europe) licensed enzyme preparations and two substrate inhibitors available with other potential therapies on the horizon. Treatment of visceral and haematological complications has proven to be very successful with ERT and SRT although there are still many unmet challenges for patients. New treatment challenges will include which patients to treat with ERT and which to treat with SRT, and to identify those patients who would benefit from a switch in therapy.

Professor Huma Cheema from the Children's Hospital & Institute of Child Health in Lahore, Pakistan gave a passionate presentation on unmet needs of children with Gaucher disease in her country. She talked about the significant burden of genetic

diseases, including Gaucher disease, owing to a high level of consanguineous or same-caste marriages. She also spoke about the lack of awareness and the non-availability of routine testing (until 2013) which has been overcome by a huge multi-pronged media awareness campaign to paediatricians and family physicians, the government and the media. A national database for LSD was established in 2013 since prior to 2006, most patients in Pakistan died without diagnosis or referral in this resource-constrained environment.

Patients that present at the children's hospital are severe with significant organomegaly, failure to thrive, requirement for transfusions and repeat infections with bone pain. Tragically, only 12% of the patients in Pakistan are on ERT (the majority through Genzyme ICAP), and for most, a cure is only a dream. The government and private donors have approved therapy for the first time in three patients but the challenges facing this patient population are huge and questions relating to the definition of rarity and the pricing of orphan drugs as well as the impact on the healthcare system need to be addressed.

Dr Seng Cheng from Genzyme, a Sanofi company talked about emerging therapies for lysosomal storage disorders; 'Enzyme Replacement Therapy does not address CNS involvement in lysosomal storage disorders, therefore efforts to address this limitation have been initiated to develop alternative therapies, and a concept that is gaining interest is substrate reduction therapy. This therapeutic approach transverses the blood-brain barrier and may be a candidate to treat the neuropathic manifestations. Genzyme's pre-clinical data in mouse models of neuronopathic Gaucher disease using a small molecule drug are supportive of initiating clinical testing in patients with Type III GD as well as late on-set Tay-Sachs disease.'

Workshops

On the Saturday, we held four workshops and each one was presented twice so that each participant could attend at least two. They were entitled:

- Spectrum of Clinical Manifestations in Gaucher Disease
- Gaucher Disease Type III: Addressing Patients' and Families' Concerns
- Unmet Needs in Assessment of Bone Disease in Gaucher
- Gaucher Disease and Cancer: Mechanisms and Management

In addition to the workshops, Gauchers Association Young Board Member Maddie Collin made a video for the Type III workshop to illustrate the unseen challenges of living day-to-day with Type III GD, highlighting what future potential treatment should be targeting to improve the quality of life for patients.

European Gaucher Alliance

The EGA board were invited to attend the GLF and during the weekend they met with Genzyme's senior management to discuss patient access to Cerdelga, Genzyme's new small molecule which is now licensed in both Europe and the USA.

London Marathon runners raise over £12,000

On a damp and chilly Sunday morning in late April, more than 38,000 runners set off for the 35th London Marathon, including our very own Atul Mehta, Avani Mehta, Lisa Robson, Lizzie Strong and Claudia Cataldo who all ran for the Gauchers Association. This event is one of our biggest fundraisers and forms a significant part of the income we need to continue to support the Association's activities and initiatives. Please read our runners' stories below:

Lisa Robson

Lisa Robson writes: 'I feel privileged to have run the London Marathon for Gauchers. I lost my precious baby sister Emma, aged two and a half to the disease and 19 years have now past and I feel comfort from doing her memory so proud.

The marathon has to be the most challenging thing I've ever done. I was so emotional and pain really set in at 19 miles but I knew it was only temporary. It was an amazing experience, memories made I'll never forget. Thank you to everyone for their support and generosity. '

Atul & Avani Mehta

Father and daughter Atul and Avani Mehta write: 'when we heard last summer that we had two places to run the London Marathon to raise funds for the Association, our initial feeling was one of excitement tinged with nerves. The chores of training during the dark early winter mornings began to bite around Christmas time but as the days became longer and warmer, the training runs became longer – and surprisingly easier.

When it finally arrived, the day was perfect – cold, cloudy and dry. After some wise words of last minute advice from [Association Chairman] Dan Brown – he and [Association Chief Executive] Tanya have both conquered the challenge – we were off!

No more leafy streets of Hampstead and open spaces of Regents Park – we were in the depths of East London, cheered on by London Cockneys, London Irish and London Welsh. The crowds were truly amazing and soon we realised that the Poles, Greeks, Bangladeshis, West – and East – Indians were also out in supportive force. The halfway mark was the welcoming site of Tower Bridge; and then the sight of familiar family faces was magic! My wife Kokila and her family have supported us through the whole journey.

We decided to link our fundraising between the Gauchers Association and Myeloma UK. The work recently published by Prof Timothy Cox's team in Cambridge, looking at the Gaucher gene and changes of myeloma or similar cancers in mice and the positive impact that treatment has on reducing the risk is vitally important and we will report on this work in the next Gauchers News. Also, we have had a huge amount of support in terms of donations to our Just Giving page (www.justgiving.com/teams/mehtamarathon) from friends and well-wishers, yes but also huge support from patients.

A very big thank you to you all!'

Lizzie Strong and Claudia Cataldo write: 'Marathon day arrived and we were both very anxious and nervous for what the day had in store. Now, the whole experience just seems a very surreal blur. Both of us were carrying small injuries due to a long and intense training schedule but with times of 4.38 and 5.13 we made it over the finish line.

We were both lucky enough to have our friends and family showing their support every few miles along the route with the rest of the crowds who were amazing and kept us going.

Training for the marathon was without a doubt the toughest challenge we've ever had to do, but actually running the marathon was an incredible experience and it made all those tough training runs worthwhile.

The London Marathon is a massive event known internationally and it was an honour to run and raise such awareness for a good cause like the Gauchers Association and we would like to thank all of those who donated.'

Lizzie Strong & Claudia Cataldo with supporters

The Association would like to thank all its members, friends and their supporters who helped to raise £12,313. A very special thank you also goes to our five runners who endured the 26.2mile course to help us.

We are delighted to announce we only have three Golden Bond places still available for 2016. Please contact Sarah on 01453 549231 or email sarah@gaucher.org.uk if you feel inspired to take part for such an amazing cause. It may well be the best thing you'll ever do!

Upcoming fundraising events

We have a number of great fundraising events coming up throughout 2015. If you are interested in taking part in any of them, please call or email Sarah in the office on **01453 549231** or **sarah@gauchers.org.uk** and she will give you all the information you need to know!

July – We still have places available for the London 10k run on Sunday 12th July. This iconic event is celebrating its 15th anniversary with a new route taking in even more of London's iconic landmarks. There's more information in the article in this edition of Gauchers News.

August – Good luck to our cyclists who are taking part in the Prudential Ride London-Surrey 100 on Sunday 2nd August. Sarah Allard (aka 'Sarah in the office') along with her husband Neil and Rob Marshall will be taking to the closed roads through London and out into Surrey's stunning countryside. With leg-testing climbs and a route made famous by the world's best cyclists at the London 2012 Olympics, it's a truly spectacular event for all involved. Please support them if you can.

September – The Sixth London to Cambridge Bike Ride on Sunday 6th September. Join us for this great day of cycling, a scenic 60 mile route from the Royal Free in Hampstead finishing at Addenbrooke's Hospital in Cambridge. Mechanical support and four refreshments stops are provided free of charge, including some of the most delicious cakes you will ever eat! Read Rob's story on page 7 of this edition of Gauchers News for inspiration!

September – We will be wishing good luck to our runners taking part in the Great North Run. Wendy & Lee Burbidge, Susan Quintano, Lisa Robson and Dylan Gould will be taking part in the world's leading half-marathon from Newcastle-upon-Tyne to South Shields along with 57,000 other runners. Please show them your support!

Member's fundraising

As a hardworking charity, we rely on our members, their families and their friends who generously and unwaveringly support our work through subscriptions, donations and the organising of fundraising events. We are, as always, extremely grateful for this support and we would like to thank everyone who supports us via standing orders and to those who kindly donated additional funds with their annual subscriptions.

If you would like to set up a regular standing order please contact Sarah in the office on **sarah@gaucher.org.uk** or **01453 549231** and as a reminder, we have lots of great fundraising resources for you including as balloons, t-shirts, running vests, thunder sticks, ponchos, stickers, posters, leaflets and a raffle licence.

Donations received from November 2014–April 2015 totalling £2,596

We have received some very generous donations from the following people:

AT Morris, Susan & John Reizenstein, Kim Stewart, Michael Cavendish, AJ White, Mr D A Bernstein, Akshantha Shetty, Joseph Kent, Trust Inheritance Ltd, Ian Morrison Charitable Fund, Mr C McFadzean, Mr & Mrs Smalley, Mr & Mrs Shetty, Mr & Mrs Beecham, Judy & Paul DeWinter Charitable Trust, The Weinstein Foundation, Stephen Buzzard, Mrs P Aminoff, G4S Utility.

Lesley Gardner from the Dursley 100 Club kindly donated £100 from their recent raffle.

Debbie Frenkel kindly donated £100 in celebration of Sarah Manuel's 21st birthday.

Keith & Joy Moore very kindly donated £100; £20 from the local DIY shop collection box and the rest was donations from friends not sending Christmas cards!

Donate online

We rely on your generosity to enable us to continue to meet the needs of those suffering from Gaucher disease. Donations can be made online at **www.gaucher.org.uk.**

Donate by text

You can also donate via text – Text **GAUK01 £2** / **GAUK01£5** / **GAUK £10** to **70070** to donate now!

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Gift Aid your donations

The Government operates a scheme called Gift Aid that allows charities to claim the basic rate tax on every pound donated. So if you donated £100 to the Gauchers Association, the Government add on 25% (or £25 in this example) so we actually get £125 – and it doesn't cost you a penny more!

Thank you!

Gauchers ASSOCIATION

Gauchers Bike Ride

London—Cambridge on Sunday 6th September 2015

Back by popular demand

Start: 8am University College School Playing Fields, Farm Avenue, **London** NW2 2BS

Finish: Addenbrooke's Hospital, **Cambridge**

- Scenic 60 mile ride
- Open to all cyclists
- Fully marshalled route
- FREE refreshment stops along the route
- Under 16's to be accompanied by an adult
- Easy return by coach or train

Limited number of places available so register NOW!

Either go online at: www.gaucher.org.uk/fundraising/cycle_ride_2015

or contact Sarah on 01453 549231 / email: sarah@gaucher.org.uk

A fee of £25 will be payable on registration and there is a minimum £100 sponsorship pledge. Coaches will be laid on to return cyclists to London at £25 per person (including bike)

Find us on Facebook

www.facebook.com/TheGauchersAssociation

In aid of the UK Gauchers Association, which supports UK sufferers of this disease. People with Gaucher disease lack sufficient activity levels of an enzyme called glucocerebrosidase. This enzyme helps the body break down worn out cells and as a result of the enzyme deficiency, a fatty substance called glucocerebroside accumulates in the spleen, liver, bone marrow and sometimes in the central nervous system. For more information go to www.gaucher.org.uk