

# Gauchers NEWS

April 2017

## The Gauchers Association celebrate their 25th Anniversary

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## CHAIRMAN'S CHAT

### WELCOME

Dear Friends

Welcome to the April 2017 edition of Gauchers News. It has been a while since our last newsletter in June but hopefully you have been keeping up to date with all the Association's news on our website.

As many of you will know, the Association celebrated its 25th Anniversary in November last year and whilst it was a moment to reflect on the many achievements and successes of the past 25 years it is also important to recognise that there remain many challenges for our community.

As a patient group we are extremely lucky to benefit currently from fully funded treatments and home care and the expertise provided by the NHS Specialist Centres. Gaucher patients throughout the UK receive the highest level of care from some of the world's leading specialists in Lysosomal Storage Diseases. However, we must never be complacent.

You will read on page 4 about a review process that the NHS and NICE have completed in relation to proposed changes in the way that new, potential treatments for rare diseases are appraised. The Association, together with numerous other patient groups, took part in the consultation process for this review. Unfortunately the conclusions of the review have extremely negative consequences for the rare disease community. Whilst the current treatments for Gaucher disease will not be affected, future treatments for patients with unmet needs that might otherwise become available most certainly will be. The way in which treatments for rare diseases will be appraised moving forward make it highly unlikely that any new treatments will be approved for NHS funding.

Please look out for further updates on the NHS review on our website. We may well be reaching out to you all to help support the cause!

Dan



It has always been one of the Association's key objectives to ensure that those with a rare disease are not discriminated against simply because there are fewer of them. This is a mission we, together with our colleagues within the rare disease community, will continue to fight and we will be doing everything we can to influence the relevant decision makers within the NHS and government to amend the conclusions of the review.

Making sure we can continue to represent patients' interests and advocate on their behalf is just one of the many functions that the Association now performs and as a small charity we rely heavily on the many generous donations we receive through a variety of channels in order to maintain our services. We celebrated our 25th Anniversary in November by hosting a dinner at the Royal College of Physicians in Regents Park. It was a truly fitting venue and a wonderful evening. We marked the occasion with a souvenir brochure which, together with a raffle and other donations received on the night, generated over £100,000. This will be vital in ensuring that the Association can continue with all of its good work.

I would like to express the Board's sincere thanks to everyone who helped make the Anniversary celebration such a success. A particular thanks has to go to Sarah, our Information and Charity Officer, for all of her efforts in organising such a fantastic event.

## NICE UPDATE

### NICE and NHS England slaps those suffering from ultra-rare diseases in the face

**In late October 2016 NHS England and NICE launched a 12-week consultation on changes to the arrangements for evaluating and funding drugs and other health technologies appraised through NICE's Highly Specialised Technologies (HST) programmes.**

This is a clear message from NICE and NHS England published on 15th March, that they are unwilling to continue to pay for innovation treatments for rare diseases to the extent that they do now.

In summary, these changes coming into force from April 1 this year, will see the introduction of a £100,000 Quality-adjusted life year (QALY) threshold for medicines evaluated via NICE's Highly Specialised Technologies (HST) programme, which assesses treatments for ultra-rare diseases. This threshold will effectively stop the flow of new medicines reaching patients with ultra-rare and complex diseases. Many treatments for ultra-rare conditions that are currently funded by NHS England have costs per QALY of more than £500,000 including the three medicines that have been approved by NICE's HST process to date. It is

widely acknowledged that QALY thresholds are not appropriate for evaluating medicines for ultra-rare diseases, due to the small patient populations and often limited data.

The Gauchers Association, Members of other patient groups, and the Pharmaceutical Companies will work tirelessly to put pressure on The UK government who now need to act at lightning speed and reverse this initial NICE & NHS England policy not just in the context of patients with ultra-rare diseases but also that of the life sciences industry who will see no incentive to investing in the UK market if their innovative medicines and technologies have no prospect of reaching the patient.

You can read the full announcement from NICE and NHS England in the NICE board papers released on 15 March 2017 on our website

**We will use our website and Facebook page to keep our patient community informed. If any of our Members or their family Members would like to talk about this further, please call the Gauchers Association on: 01453 549231**

## NICE GIVE A MINDED 'NO'

### NICE give a minded 'NO' to Genzyme new oral therapy for GD 1 adult patients

Members will be aware from reports in the Gauchers News over the past two years that the National Institute of Health and Care Excellence (NICE) Highly Specialised Technologies Appraisal Committee is currently undergoing a technical appraisal of Eliglustat (trade name Cerdelga) which if positive will enable English patients to get funding from NHS England to get access to this new oral therapy for adult type 1 patients if clinically suitable.

On 14 March, NICE published the outcome of their appraisal and recommended that Eliglustat is not recommended within its marketing authorisation for treating type 1 Gaucher disease, that is, for long-term treatment in adults who are cytochrome P450 2D6 poor, intermediate or extensive metabolisers.

Patient and Clinical Experts will reply to this recommendation and appear at the second evaluation committee on 19th April to make another case for the treatment to be funded.

In summary, the committee found that, although they recognised that Eliglustat is likely to have a significant impact on people's lives beyond its direct health benefits, it had not been given enough justification for the high cost per person of Eliglustat.

It is important to note that though this is not yet final guidance and may be subject to change and after considering these comments, the committee will prepare the final evaluation determination (FED).

You can read the full Evaluation Consultation report released on 14 March 2017 on our website at [www.gaucher.org.uk](http://www.gaucher.org.uk)

**We will use our website and Facebook page to keep our patient community informed. If any of our Members or their Family Members would like to talk about this further, please call the Gauchers Association on: 01453 549231**



### EUROPEAN GAUCHER ALLIANCE (EGA) NEW TEAM MEMBER

I just wanted to write a brief note introducing myself as the EGA new boy, and to say how very pleased I am to be joining you in 2017. I am Chris Hilldrup and I have been lucky enough to have been selected to try and fill Jo's shoes whilst she takes maternity leave. I come to you with a varied background having been an Outdoor Pursuits Instructor, commercial property surveyor and

renovator and most recently a Detective within the British Police Service. However I would say that of all the hats that I have worn in my time I am at my happiest when trying to help others. Perhaps a cliché, but my driving force is attempting to make a difference either improving a situation or helping others achieve their goals.

I am a family man with a passion for the great outdoors. I share common concerns for the world that we will leave our children and it is for that reason that I am relishing the opportunity to contribute towards the Gaucher cause.



## PERSONAL STORY

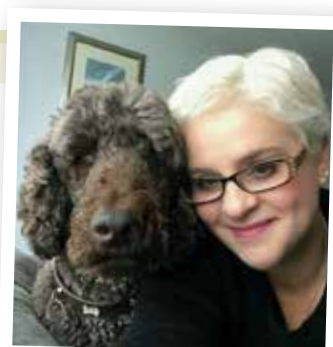
# MY DIAGNOSIS JOURNEY

**Type I Gaucher patient Adele, tells her diagnosis journey and the impact on her and her family life:**

I was diagnosed in 1991 at the age of 24 and a new mother, after a routine check with my local GP he sent me to see a gastroenterologist who informed me I had an enlarged liver and spleen. I went on to have a liver biopsy and this was when I got the diagnosis of Gaucher disease.

By the time I got referred to Addenbrooke's hospital lysosomal disorder unit I was in the early stages of another pregnancy. After months of seeing doctors whose first words were always "I have never heard of that before", it was a joy to be welcomed to the Gaucher clinic and finally be treated like somebody normal. We met Liz Morris (specialist nurse) my very first time visiting and continue to see her each time we go.

I had my little boy in October 1994 and our family was complete, but after two pregnancies I was quite unwell due to the Gaucher symptoms. Professor Cox was keen for me to start enzyme replacement therapy, however I wanted to wait for Cerezyme to become available, as at the time Ceredase was the only alternative. Unfortunately I had to go on ERT sooner than expected and started having twice weekly infusions at my local hospital. Being time consuming and impractical it was arranged for me to have home infusions. Nurses came with a truck load of equipment and taught both myself and my husband how to set up the infusions. My husband Andrew turned out to be a natural and for the next 16 years he did my infusions at home twice a week and after changing to Cerezyme as soon as it was available we never looked back.



Adele and Willow

The enzyme was amazing and I gradually felt much better than I had ever felt in my life before. We worked our life around the infusions, always making sure that we did them at the time when we wouldn't have unexpected visitors, as I was never comfortable with people seeing me attached to a drip.

In 2011 at a routine Gaucher appointment I was asked to consider taking part in a trial for a capsule (Eliglustat) as a different type of treatment for Gaucher disease. Even though it would mean the long trips to Addenbrooke's every three months and lots of extra tests, I was immediately interested. This is what I always wanted to "pop a pill" like everybody else. Right from the start I loved taking Eliglustat, I have had no side effects and the ease of taking a pill twice a day compared to the rigmarole of infusing is a joy.

For three years we continued with the regular appointments for the trial. We got to know Cambridge quite well, especially the "Wok & Grill" my husband's favourite eating place!

Presently I am still taking Eliglustat and after having a hip replacement last August I can honestly say that I have never felt better, which allows me to do all the things I have always wanted to do. Mostly this involves daily long walks with my dog.

## NEW CONSULTANT

# DR CHON YEW TAN

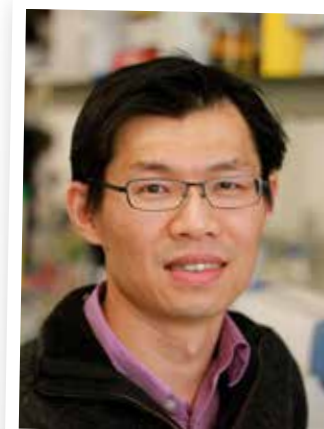
**NEWLY APPOINTED CONSULTANT AT ADDENBROOKE'S HOSPITAL, CAMBRIDGE UNIVERSITY HOSPITALS NHS FOUNDATION TRUST**

Dr Chong Yew Tan has recently been appointed as Consultant Physician with an interest in Metabolic diseases in Cambridge and is sharing this work and the clinical activities of the Lysosomal Disorders Unit with its Clinical Director, Dr Patrick Deegan and other physicians including Dr Paul Flynn and Professor Cox.

Dr Tan, who is married with two children, graduated in Medicine with Honours and numerous undergraduate prizes from Trinity College, Dublin. After junior training posts and higher professional qualifications, Dr Tan completed his Specialist Training in General Internal Medicine as well as Diabetes Mellitus & Endocrinology a few years ago.

Chong Yew came to Cambridge in 2006 and later joined the Wellcome Trust Clinician Research Training programme as a Clinical Research Fellow - in 2012 completing his PhD studies in energy metabolism with Prof Tony Vidal-Puig.

After appointment as Clinical Lecturer in the University department of Medicine with Professor Cox, Chong Yew further developed his interests in Metabolic diseases and Lysosomal disorders. Throughout this period, he has taken a leading role in the national **Gaucherite** project bringing much needed computer skills into the development of the bespoke, relational database that holds (anonymously) the clinical, imaging and laboratory information on all the participants.



Dr Chong Yew Tan MB MRCP(UK) PhD

Prof Cox writes: 'It is wonderful to be able to welcome Dr Tan, a brilliant young clinician into this field. Chong Yew brings not only his clear-thinking intellect, but unique skills and experience from other clinical specialties. As those in the community will appreciate, while many metabolic conditions are very rare, those like Chong Yew, with the required expertise and enthusiasm to treat patients with challenging disorders, like Gaucher disease, are even rarer...'

# UK PATIENT AND FAMILY SUPPORT WORKER



Helen Whitehead

### BENEFITS AND APPEALS

I have been contacted over the last few months by many people experiencing difficulties when moving onto the newer benefits such as PIP (Personal Independence Payment) and ESA (Employment Support Allowance). Both of these benefits require people to have a face-to-face assessment where you gain points for being able to do certain tasks and have to score a certain number of points to be eligible.

If it comes back that you are not eligible all is not lost. The first step is to ask for a Mandatory Reconsideration – this is just asking them to relook at your case. For this

you can provide extra evidence such as specialist letters, a diary, and statements from carers etc. But it is very important to get this back to them in time within 1 calendar month. If this is still a no you can take this to appeal, again within 1 calendar month.

Helen writes "I went with a type 1 Gaucher member to their appeal last November, it was successful, they went from receiving 0 points to enough points to be eligible for the benefit. Together the member and I went through all the paperwork with a fine toothcomb and contacted the relevant people for more evidence to submit to the appeal. The thought of an appeal with a judge is quite daunting, but the experience wasn't. On the panel were a judge and a Dr, both in normal clothing – no wigs! They sat one side of a desk and we sat the other. They asked the Gaucher patient

lots of questions about what they can and can't do and they both listened and asked relevant questions. We were then asked to leave the room whilst they discussed the case and after a few minutes were invited back in where their decision was explained."

Statistics show that around 60% of appeals are successful (for PIP and ESA). Also it is better to attend the hearing in person, rather than opt to have it done on paper. You can take someone with you into the appeal, it could be someone who knows you well or it can be a representative, for example me from the Gauchers Association or someone from the Citizens Advice Bureau.



### HOW CAN I HELP?

I can support you through any stage of the process, from completing the initial form, through the mandatory reconsideration process and appeal.

It can be helpful to be able to meet face-to-face to go over all the details and make sure that you have the evidence is required and get you fully prepared. If this affects you, please get in touch.

If you have any unmet support needs, I work Monday to Thursday. To get in touch you can call me at the Gauchers Association on 01453 549231, call or text me on my mobile on 07795 192311, email me [helen@gaucher.org.uk](mailto:helen@gaucher.org.uk) or find me on Facebook by searching for 'Helen Whitehead' and the Gaucher logo. If you would rather meet in person we can arrange this too either at your home or a coffee shop.

### THINKING AHEAD TO THE SUMMER

Hopefully your travels will go without a hitch. But what would happen if you fell ill or got injured in a foreign country?

You may need to pay to get treated or flown home. Travel insurance gives you peace of mind. You must disclose all your health conditions including Gaucher Disease.

We have a list of providers that our members have recommended who will insure people with Gaucher Disease. Please contact us if you would like a copy.

### EMPOWERMENT PROJECT UPDATE

The empowerment project is aimed at young people and adults with type 3 Gaucher Disease.

We meet as a group around 3 times a year, supported by a Consultant and Clinical Nurse Specialist. We started our last meeting at an Italian restaurant in London, with 12 of us catching up with before heading to the theatre to see 'Matilda the Musical'. This was a fantastic show, with great music, talented young actors, with everyone saying what a great experience it was to see a West-End show. The group then headed to a hotel to prepare for a full day involvement in the Wearable Technology project (please see more about this on pg 21). We all met bright and early the next morning and headed to the Royal Free Hospital where the group participated in a variety of activities, such as eye test, walking test, smell test and received the device (looks like a watch but takes measurements of steps) and shown how to use the app.





## RESEARCH

### Making Gene Therapy a reality for patients with neuronopathic Gaucher Disease...

Since our last brief update in the June 2016 edition of Gauchers News the Gauchers Association and Drs Simon Waddington and Ahad Rahim and their team at University College London (UCL), along with Dr Elin Haf Davies submitted an application to the European Commission for a 2020 Horizon bid to take our Gene Therapy forward into a clinical trial, entitled RightingGaucher.

Horizon 2020 is a pot of money provided by the European Commission for research and innovation projects from 2014 – 2020 to produce world-class science, remove barriers to innovation and makes it easier for the public and private sectors to work together in delivering innovation.

Unfortunately, we were not successful but have identified others avenues of funding which we intend to pursue.

One of the key next steps for this work to continue is to work with the European Medicines Agency (EMA) to gain advice on the pre-clinical work we still need to undertake and on designing the clinical trial protocol. On 7th March the Gene Therapy Team (pictured below) supported by Dr Anupam Chakrapani & Dr Julien Baruteau, from Great Ormond Street Hospital attended a Scientific Advisory Working Party meeting at the EMA to discuss these issues. We anticipate feedback from the EMA in April.

Ongoing reports on the progress of this work will be reported on the Association's Facebook page and website.

Rapsodi

See page 20



Gaucherite

See page 14/15

## Clinical Trial ALERT!!!

Genzyme's oral therapy in Combination with Cerezyme in Adult Patients with Gaucher Disease Type 3 (LEAP)

In the June 2016 edition of Gauchers News we reported on the impending clinical trial for adults (18 years and older) with Type 3 Gaucher disease with Genzyme's small molecule GZ/SAR402671. We are delighted to report that this 52-week two-part study phase 2 clinical trial is now recruiting with sites open in the UK and US.

Recruitment onto this trial has been challenging for our nGD patients in the UK as despite this trial being for Type 3 patients there was a mandatory eligibility requirement of lung function (the ability of the lungs to transfer gas from inhaled air to the red blood cells in pulmonary capillaries) and that had excluded all the UK GD 3 community, meaning that with three sites open in the UK 0 patients have been enrolled in this study.

However following communication with the Gauchers Association and the Principle Investigators demonstrating that there were no eligible patients in the UK, Genzyme have removed this mandatory criteria from the trial, the UK Centre's will now invite those patients that wish to be considered for this trial to visit their Centre for assessment.

Below is an outline of what the study objectives are and further details, including the eligibility and exclusion criteria can be found online at; <http://bit.ly/2ns323X>

### The Primary Objective of the study:

#### Part 1:

- Evaluate central nervous system (CNS) biomarkers in adult Gaucher disease (GD) type 3 (GD3) patients that distinguish GD3 from Gaucher disease type 1 (GD1).
- Screen adult GD3 patients who qualify for treatment with GZ/SAR402671 in Part 2.

#### Part 2:

- Evaluate the safety and tolerability of GZ/SAR402671 in adult GD3 patients.
- Evaluate the change in cerebrospinal fluid (CSF) central nervous system (CNS) biomarkers from adult GD3 patients receiving GZ/SAR402671.

### And Secondary Objectives:

- Evaluate the pharmacokinetics of GZ/SAR402671 in adult GD3 patients.
- Explore the efficacy of GZ/SAR402671 in infiltrative lung disease (IDL) in adult GD3 patients.
- Explore the efficacy of GZ/SAR402671 in systemic disease in adult GD3 patients.
- Explore the efficacy of GZ/SAR402671 in neurological function and on exploratory CSF biomarkers in adult GD3 patients.

**PLEASE NOTE:** To be eligible for this study all participants **MUST** be on Cerezyme for at least 6 months prior to enrollment, at a stable monthly dose and must continue at the same monthly dose during the study.

## INVOLVING MEMBERS

# HEMOCARE PATIENT INVOLVEMENT MEETING

### Gaucher member attends a Hemocare Patient Involvement meeting

Gaucher member Ralph Wood attended a hemocare patient involvement meeting hosted by Healthcare At Home on 29th September 2016. The Association is very aware that many of our members wish to support us in offering their patient expertise by attending meetings as representatives of the Gaucher community.

Ralph writes 'The meeting was attended by various patients of Healthcare At Home (HAH), Company Representatives and a facilitating organisation for running the meeting.

We were given an update on some of the initiatives currently underway by HAH, and some feedback on changes that were made since the last Patient Involvement meeting held in the spring 2015. Amongst the changes and successes given was the expansion of HAH to looking after newly discharged patients from Hospital, and handling a recent drug shortage without patients being aware of the problems.

The point was made to HAH, that such successes and changes need to be communicated to their clients (i.e. Hospitals and patients), and an easy method of informing patients would be a flyer in with a patient's medication or given to them by their nurse on a visit.

We also made the point that HAH have three separate groups of patients i.e.

those receiving drug delivery only, those receiving nurse support doing an infusion, and those being supported post-discharge from Hospital. Each have different needs and concerns about HAH and as such need separate patient involvement meetings (say every 6 months).

We also made the point that any systems that HAH design that have any impact on patients or the nurses should have patient involvement at the design stage. This came about when we were told that the new 'Nurses visit scheduling system' trialled in London was designed and implemented without any patient input.

We also talked about communication into HAH, and it was raised that nurses make notes on their tablet whilst on visits, but this appears to be ignored and the patient is later asked by HAH HQ for the same information (e.g. drug stores). It was also mentioned that patients should have an ability to 'shop online' for items (ancillary items such as needles and syringes) for their next delivery rather than rely on telling someone in the HAH office and having them interpret the request.

Overall I was under the impression that the senior HAH officials in attendance took note of the comments made by patients and welcomed the feedback. We were also all in agreement that HAH do provide a vital service and that their nurses are star players'

**The Association is very grateful and send thanks to Ralph who attended this meeting. If you would be interested in being involved with patient involvement meetings please contact Sarah on [sarah@gaucher.org.uk](mailto:sarah@gaucher.org.uk) or 01453 549231**

## INVOLVING MEMBERS

# SCOTTISH INHERITED METABOLIC DISEASE EXPERT MEETING

### The work of Inherited Metabolic Disease (IMD) Expert Review Group in Scotland

Gaucher patient and member, Dame Anne Begg attended two meetings in Scotland as a patient expert and representative of the Association during 2016; Anne writes:



At the moment, people with very rare conditions who live in Scotland sometimes have their clinical needs overseen by a consultant

and clinical team elsewhere in the UK. This is the case for most of those with Lysosomal Storage Disorders (LSD), including Gaucher disease, who require Enzyme Replacement Therapy (ERT). At the moment Scottish Gaucher residents have part or all of their care overseen by specialist services in Salford (Manchester), London or Cambridge. The numbers of patients involved is so small it has been sensible to ensure Scottish residents have had access to clinical teams with the detailed specialist knowledge of their ultra-rare condition.

While Gaucher disease is one of the Lysosomal Storage Disorders it is also an Inherited Metabolic Disease (IMD). While the numbers of patients with the individual conditions are very small in Scotland, when taken together with other IMDs the

numbers suggest there is a need for a more coherent approach to how IMD patients are supported across the whole of Scotland.

As a result, an Expert Review Group has been set up by NHS Scotland to look at the provision and delivery of services for those with an Inherited Metabolic Disease. The group has been meeting throughout 2016 including a meeting of patient's representatives in Glasgow.

The discussion has revolved around the shape of IMD services across Scotland, the number of centres there should be and whether adult and paediatric services should remain separate or should come together in a single centre in either Glasgow or Edinburgh. While final decisions have not yet been made it looks as though the group will decide on a single core centre for both children and adults with satellites in other geographic areas across Scotland.

However, there is recognition that there may need to be some separate arrangements for LSD patients in Scotland particularly those on ERT. There will continue to be a need for a cost sharing scheme for ultra-orphan drugs.

The Report from the Expert Review Group has yet to be finalised and published and will be available on our website

The Association is very grateful and send thanks to Dame Anne Begg who attended this meeting



# UPDATE ON GAUCHERITE



Professor Timothy Cox

Following on from our last edition of *Gauchers News*, Timothy M Cox, Frances M Platt from the Universities of Cambridge and Oxford give an update below on the Gaucherite project

Many readers will know that THE NATIONAL GAUCHERITE PROJECT (Gaucher disease Investigative Therapy Evaluation) is an on-going wide-scale observational 'cohort' study of Gaucher disease in the UK. This project has been principally funded as the first and, so far, only genetic disorder included in the Medical Research Council's burgeoning Stratified Medicine research portfolio.

We are exploring the outcomes of treatments and quality-of-life benefits with multivariate disease stratification to define distinct types of disease behaviour. This approach is now believed to be one of the best means of tailoring treatments in particular patients with particular therapies. It also should lead to greater understanding and accurate prediction of disease behaviour and truly inform decisions about treatment.

Our participants, (aged 5 years and over) with Gaucher disease, attend the eight National Specialist Centres for the advanced treatment and monitoring of Gaucher disease. As of December 2016, and now in its last year of funding from the MRC, 209 patients including 17 children have enrolled and this is a wonderful testament to the engagement of the whole community – who already contribute hugely to research efforts across the UK – and abroad.

**GAUCHERITE** involves the collection, assembly and collation of detailed clinical and pathological information. This includes historical and present outcomes from serial DEXA and other radiological imaging, as well as haematological, biochemical and genetic data in the ultra-deep characterization of UK patients recorded with all medication. A single bespoke Biobank holding deep-frozen plasma, sera, frozen blood cells and DNA – and other diverse clinical material – is also curated for later investigation in a manner to be agreed in consensus by the Programme.

The project includes two special research strands for paediatric and adult patients: the osseous (involving bone problems and the skeleton), including (multiple myeloma); and the neurological strand, involving Parkinsonism and also many ill-understood clinical aspects of the neurological effects of Gaucher disease (so-called 'type 3') affecting adults and children.

This is now a time for taking stock of all the (fully anonymized) information we have

collected in a special dedicated and easy to use data resource designed especially for **GAUCHERITE** to provide information for public release. With this in mind, the next step will be a formal interim analysis – funding for which is being currently sought from one of our supporting companies who have signed collaborative agreements with the MRC.

In the light of the findings and the critical importance of the future outcomes of the programme, we are in no doubt that the project will have lasting value in directing future therapeutic initiatives for the benefit of all patients. To this effect, we are looking to the future to maintain the database (in effect curated by Dr CY Tan), and the services of our excellent clinical manager (Sister Kathy Page) and Clinical Research Fellows (Drs Simona D'Amore – bone – and Aimée Donald – neurological) as well as the resource of blood cells and other material in the Biobank in Oxford – under the curation of Dr Kerri Wallom, who works with Professor Frances Platt, co-Principal Investigator in Oxford. Everyone at every level – service directors, nurses, doctors, secretaries, clerks, database managers and handlers, radiologists, scientists, has done a fantastic job – despite the terrible pressures currently in the NHS.

It is striking that we have had the fullest engagement – at our research away day and at the management consortium meetings – from first-rate and senior representatives of the biopharmaceutical industry. The presence of Actelion, Sanofi Genzyme and Shire at consortium meetings has been a model of collaboration and disinterested scientific behaviour at every level; our industrial collaborators have given constructive assistance freely with an excellent following at each of many meetings up and down the country. So far little money has changed hands – but know-how and ideas have been generously shared to the huge benefit of the work! We are indebted to these colleagues for their collegial support.

While we have some way to go, massive strides have been made: at least 80% of the UK patient population is signed up with nearly all the children, too. I am pleased to say that we have managed recently to find some funds to maintain continuity through key clinical research staff – if not yet for complete 'future-proofing'. This has come from the Biomedical Research Centres of the National Institute for Health Research in the NHS and needs to be expanded.

Finally, Mr Jeremy Manuel, Tanya Collin-Histed and the officers of The Gauchers Association, your members and all patients up and down the country in all the centres have, done wonders in supporting this work. We will report back again with concrete information soon; but, there is really good cause for genuine celebration and of credible hope!"

*(on behalf of all investigators in the UK GAUCHERITE programme – Royal Free Hospital, London; Royal Manchester Children's hospital; Salford Hospital, Manchester; Great Ormond Hospital for Children, London; Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery, London; Queen Elizabeth hospital, Birmingham, Children's hospital, Birmingham, Addenbrooke's hospital, Cambridge).*



## 25TH ANNIVERSARY CELEBRATION

On the evening of Saturday 5th November 2016 around 150 guests gathered at the Royal College of Physicians, Regents Park, London to celebrate the momentous milestone of the Association's 25th Anniversary.

The Associations Directors were delighted to welcome friends old and new, including, members, clinicians, scientists and industry representatives, who had all purchased tickets at £25; many of whom had travelled long distances to attend this celebration.

The evening started with a reception held in the Dorchester library, containing a collection of rare books and manuscripts including Caxton's Recuyell of the Historyes of Troye, 1473, which is the first book ever printed in the English Language. Whilst guests mingled and caught up with old friends they were entertained by a Jazz quartet from The Royal Academy of Music throughout the evening.

**'Thank you so very much for inviting us to the Twenty-fifth Anniversary. It was a brilliant celebration and for your guests seemed to run impeccably! The mark of professionalism and sheer effort.'**

At 8pm guests were invited to make their way the Osler and Long room for dinner and was presented with a copy of a souvenir brochure. Once seated Dan Brown, Association Chairman officially welcomed everyone and gave a floral presentation to Tanya Collin-Histed, Sarah Allard, Helen Whitehead and Claire Lightfoot in recognition of



**'Just wanted to say how much we enjoyed the anniversary dinner last Saturday. It was great to catch up with people we had not seen for a long time. A delightful evening.'**

their continued dedication to the Association.

Dinner was then served and the room was filled with conversation as guests reminisced and shared stories of the past 25 years. Guests were once again entertained by the Jazz quartet along with Jack and Leon, who performed close up magic, involving playing cards and even a gold fish!

After the main course, new Association Director and Gaucher type III patient Madeline Collin took the microphone and gave a very personal dedication on how the Association has supported her and her family and her wish to give something back. Jeremy Manuel, Association Director introduced Professor Timothy Cox from Cambridge University who gave a toast to the Association on reaching such an important milestone.

Throughout the evening a fundraising raffle was held, by asking guests to sign their name on a sticky to a £20. At the end of the evening two were drawn from the bucket and congratulations go to Miriam Davis who won a view from The Shard and dinner for two at The Savoy Grill and Camilla Sandars who won afternoon tea for two at The House of Lords kindly donated by friend of the Association Lord Palmer. At total of £1,700 was raised on the night.

The evening was a huge success raising over £106,000 to enable the Association to continue with their important work by producing our souvenir brochure, thanks go to Dan Brown, Jeremy Manuel, Andrew Bloom, Sue Noe and Sarah Allard for making this possible. Special thanks go to all of the souvenir brochure sponsors, Jamie Brown of Two Boys for designing the brochure and of course everyone who was able to attend for making it such a wonderful and memorable occasion.





'We certainly had a great time celebrating the 25th anniversary of the Gauchers Association'

'A big thank you to you and the team for a truly wonderful dinner event in London'



'Thank you for all the hard work you put in to make this such a lovely memorable occasion.'





## RESEARCH

# NEWS FROM THE GBA PARKINSON'S RESEARCH TEAM



Have you enrolled yet onto the RAPSODI online study?

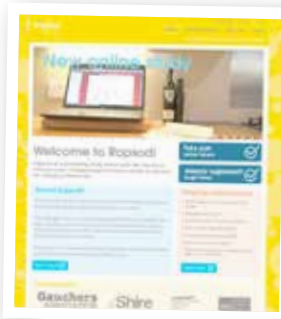
Ambroxol clinical trial begins at the Royal Free and University College London.

The research team at the Royal Free Hospital and University College London, led by Professors Anthony Schapira, Atul Mehta and Derralynn Hughes have over the last 6 years been investigating variation in the GBA gene and what it can tell us about Parkinson's. People with Gaucher and some of their family members carry the GBA gene and there is a slighter higher chance of developing Parkinson's in later life if you carry this genetic alteration, but

this is still a very low risk and most people do not develop Parkinson's. The research team is currently observing associated patterns of movement, memory, sleep, mood and sense of smell using an online study portal. The aim is to intervene much earlier before the movement symptoms associated with Parkinson's ever develop and to protect the nerve cells in the brain.

With the generosity of time and help of people with Gaucher their research has now led to a phase II drug trial of Ambroxol, a drug traditionally used as a respiratory medication which has been showing very promising neuroprotective qualities.

Recruitment started last year in 2016 on the RAPSODI study. So far 82 people with Gaucher disease and their families have done the online portal, mainly from the Lysosomal Storage Disorders Unit at the Royal Free in London. You can visit the study portal to see more about the research [www.rapsodi.org.uk](http://www.rapsodi.org.uk) People are now being invited to take part from the other centres around the country and the team will be visiting Cambridge, Salford and Birmingham in the next few months. The study takes about 45 minutes to do on your computer or laptop at home and then some postal tests.



### HOW TO GET INVOLVED

If you would like to take part or learn more please contact Sarah Cable (clinical research nurse) [rapsodi@ucl.ac.uk](mailto:rapsodi@ucl.ac.uk) 07753 982 063

## WEARABLE TECHNOLOGY STUDY

# A NEUROLOGICAL DISEASE SEVERITY ASSESSMENT



In July 2016 we reported an update on the research work being done by the Association in partnership with Dr Aimee Donald, Paediatric Clinical Research Fellow, Manchester Children's Hospital and Dr Elin Haf Davis, aparito on a pilot study for the use of wearable technology to assess the impact of nGD on daily living, to learn more about the condition, to encourage research and to develop management and interventions to support patients. Dr Aimee Donald writes an update:

'As some of you will know I've been working with the Gauchers Association and Aparito to bring a project, using a range of technologies and research tools, into action for assessing disease severity in patients with Gaucher Disease. We are finally recruiting into this study and learning a lot!

The purpose of the study was to see if there are more creative ways to assess disease severity and activity which are less intrusive and more acceptable to patients; while teaching us more about the disease process. To do this we are giving patients activity monitors, much like 'fit-bits', to wear so we can see if how active they are reflects how severe their disease is. The wearable monitors are linked by Bluetooth to an app on their mobile phones which patients can interact with; they can use the app to tell us about things that have happened to them, symptoms they've had and how they feel. We're comparing this technology with assessments patients usually have in clinic; walking tests that sometimes they find too difficult to complete.

### What's involved?

When patients first attend for this study we're also asking them to try out a technology called 'Eye See Cam'; these fancy goggles measure their eye movements which we know is a difficulty experienced by patients with type 3 Gaucher Disease; we wonder if we could

measure these difficulties more accurately than the techniques we currently use.

We are also asking patients to answer some questionnaires about their sleep and to play a game assessing their smell. There is another study running simultaneously looking at the links between Gaucher Disease and Parkinson's Disease (RAPSODI); so if you were recruited to both studies we wouldn't ask you to repeat the same tests! This part of our study is aimed at children in particular who wouldn't be eligible for the Rapsodi study.

### How to sign up?

Everything in the study is quite fun and so far we've recruited seven UK patients with Type 3 Gaucher Disease; we're really keen to recruit more – especially those with Type 3 (including children). We are now able to recruit patients who attend the Royal Free, Salford Royal, Royal Manchester Children's, Great Ormond Street and The National/ University College London, Hospitals. We soon hope to be able to recruit also in Cambridge. If you haven't been approached and would like to be involved please email [aimee.donald@cmft.nhs.uk](mailto:aimee.donald@cmft.nhs.uk) – I look forward to hearing from you!

You can also contact Helen Whitehead, The Association's Patient and Family Support Worker if you would like more information on [Helen@gaucher.org.uk](mailto:Helen@gaucher.org.uk) or 01453 549231

## NEURONOPATHIC

# UNDERSTANDING NEURONOPATHIC GAUCHER DISEASE

Understanding the UK Neuronopathic Gaucher Disease Patients Community and their needs

In the UK, we have approximately 30 patients with a diagnosis of Type 3 Gaucher disease, ranging from 2 years to mid-50's. With the introduction of Enzyme Replacement Therapy (ERT) those patients who would not have survived childhood are living longer into their 20's and beyond and are faced with new clinical and social challenges and over the next few years we hope to see several new treatment possibilities coming to trial.

Therefore, on 18th January, the Gauchers Association in collaboration with Dr Derralyann Hughes, Royal Free Hospital, London organised an educational day on neuronopathic Gaucher disease (nGD). The aim of the day was to bring together UK treating Consultants (paediatric and adult), Clinical Nurse Specialists and Psychologists treating nGD patients from the 9 Centres of Excellence in England and Wales with a view to;

1. Identify current challenges in patient care, using case studies
2. Share current research areas/interests/clinical trials
3. Identify outstanding areas of research
4. Review current clinical guidelines for clinical care in England

A full report of this day will be available on our website from April and we will send an email to all our members and a link via Facebook once it has been uploaded.



## RARE DISEASE DAY

# 28 FEBRUARY 2017

28 February 2017 marked the tenth international Rare Disease Day coordinated by EURORDIS.



On and around this day hundreds of patient organisations from countries and regions all over the world held awareness-raising activities based on this year's theme of research. Research is key. It brings hope to the millions of people living with a rare disease across the world and their families.

Here in Dursley the team all donned their raising awareness t-shirts and took part in the Dursley

annual pancake race. Congratulations to Chris for coming second overall!

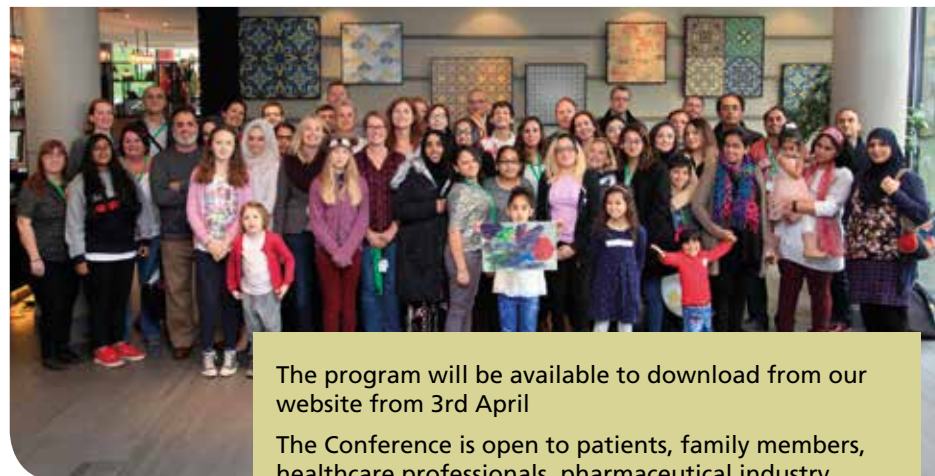
To find out more visit [www.rarediseaseday.org](http://www.rarediseaseday.org)

## NEURONOPATHIC

# 8TH NEURONOPATHIC GAUCHER DISEASE MEETING

We are pleased to announce the 8th nGD family conference will take place on 17th-20th November 2017 at Centre parcs, Woburn, Bedfordshire. This event 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, including topics such as updates on the latest research, potential new therapies, genetic modifiers, education, benefits and some very heartfelt, personal stories told by patients, families and many people directly affected by Gaucher disease along with a full children's activity program for the under 16's with our fantastic volunteers, all of whom are fully DBS checked



The program will be available to download from our website from 3rd April

The Conference is open to patients, family members, healthcare professionals, pharmaceutical industry and Homecare representatives.

THE COST IS AS FOLLOWS:-

- Family (up to two adults and three children) £150
- Single Family member £50
- Gaucher Patient (over the age of 18) £50
- Pharmaceutical and Homecare representatives (for Saturday only) £150

To book your place please call 01453 549231 or email Sarah on [Sarah@gaucher.org.uk](mailto:Sarah@gaucher.org.uk)





## AWARDS

# THE 10TH ALAN GORDON MEMORIAL AWARD

**We are now inviting nominations for the 10th Alan Gordon Memorial Award to be sent to Sarah Allard, Information & Charity Officer of the Gauchers Association, by the 30th April 2017. We will announce the recipient of the Award in the next edition of Gauchers News, on our website and Facebook page.**

The Alan Gordon Memorial Award can be given to an individual or group of individuals who in the opinion of the Award Committee –

- Has made a significant contribution to Gaucher disease, its treatment or management
- Has made a significant contribution to the treatment of Gaucher disease or Gaucher patients or their families
- Is particularly worthy of recognition by this Award, either through research or personal involvement or, in specific cases, to individuals or their families having special needs.

The Alan Gordon Memorial Fund was created in memory of the late Alan Gordon who died in December 1995. Alan was a founding member of the Gauchers Association and its first Treasurer, elected in 1991. His contribution during our formative years helped to shape the Association, both through his wise counsel as an Honorary Officer, and as a member of the Executive Committee.

### Previous recipients of the Alan Gordon Award are -

**Dr PramMistry**, formally of the Royal Free Hospital and now Chief of Paediatric Hepatology and Gastroenterology at Yale University.

**Dr Terry Butters and Dr Fran Platt** of the Glycobiology Institute, Oxford.

**Tanya Collin-Histed** for her work with neuronopathic Gaucher disease.

**Professor Ari Zimran and Dr Debbie Elstein** of the Gaucher Clinic at the Shaare Zedek Medical Center in Israel.

**Professor Hans Aerts** of the Amsterdam Medical Centre.

**Professor Carla Hollak** of the Amsterdam Medical Centre.

**The Lysosomal Storage Disorder Clinical Nurse Specialist Team** – Great Ormond Street Hospital, London

**Dr Ashok Vellodi** – Great Ormond Street Hospital, London

If you feel someone, whether a doctor, researcher, professional or layman, or a particular group of people warrants recognition, please send your nomination stating why you consider your choice deserves the Award, to Sarah Allard, The Gauchers Association, 8 Silver Street, Dursley, Gloucestershire, GL11 4ND or by e-mail to [sarah@gaucher.org.uk](mailto:sarah@gaucher.org.uk).

The closing date for nominations is 30th April 2017

## RESEARCH

# MODERN APPROACH TO PATIENT RESEARCH



**New insights about living with Gaucher disease is gained through the use of mobile technology.**

Every person living with Gaucher disease has their own unique story to tell. This is why Sanofi Genzyme has been working with Redline, a patient-centric consultancy, on a new mobile phone-based research project that aims to uncover insights about what it's

like for people living with Type 1 Gaucher disease. Sanofi Genzyme has been committed to supporting the Gaucher community for over 20 years and is motivated to improving the understanding of how Gaucher disease impacts the everyday life of patients and their families.

One of the main objectives of the project is to understand the challenges and aspirations of patients at key points of their lives. This includes investigating how the impact of Gaucher disease varies at different life stages such as: studying at university, coping with the demands of a career, juggling the requirements of a busy family life, right through to getting the most out of retirement. Another important area includes identifying the unmet needs of patients with the aim of exploring how things can be improved further in the future.

But this isn't just another patient survey. What makes this project unique is that it utilises mobile-phone technology to help get to the heart of patient needs. Using an app, it enables the patients to respond to questions about their life living with Gaucher disease in their own environment, without the pressure of needing to give feedback on the spot. Instead, patients respond at a time and in a way, that suits them so they feel comfortable to reveal what is most important to them.

We've already collected some fascinating feedback that enhances our understanding of the impact of Gaucher disease on modern life. As the project nears completion, we are in the process of compiling all the findings and we look forward to sharing these findings with the Gaucher community in the near future.

Our thanks go to the Gauchers Association who helped with recruitment of all the participants and to all those who took part.

# EUROPEAN GAUCHER PATIENT GROUPS HIGHLIGHT UN-MET NEED IN GAUCHER DISEASE

On 15th December 2016 a meeting was held in Amsterdam with several European Gaucher patient groups in attendance; a full write up below:

## Report from Sanofi Genzyme organised meeting with European Gaucher Patient Groups

Despite the continuing advances in treatment for Gaucher disease, patients and their families face many issues throughout their lifetime. The role of patient groups in providing support and advice is an important part of their treatment journey. A recent Sanofi-Genzyme-hosted meeting of Gaucher patient group leaders from across Europe highlighted some of the many unmet needs – both medical or personal – for patients and their families.

The biology of Gaucher disease is now well understood, but its natural history has changed with the advent of treatment. As Gaucher patients' life expectancy increases, diseases associated with later life are becoming more evident. Some cancers, particularly multiple myeloma, have been shown to be more common in Gaucher patients and age-related diseases such as Parkinson's or osteoporosis are becoming increasingly common in Gaucher populations. Despite this increasing knowledge about Gaucher disease there remain scientific areas still to be explored, for example why patients with the same mutation differ in their clinical presentation.

Do patients understand the increased risks and is enough being done to ensure these

risks are managed? Across Europe many Gaucher patients are either unaware or not prepared to acknowledge the increased risks to their long-term health. The role of the patient organisations in educating their members about the risks without adding extra stress and worry for the patient is becoming an important part of their activities. Whether this education is delivered by webinar, patient leaflets or as part of the continuing pastoral care will depend on the individual country approach, to ensure that their community can receive the best possible advice and support.

Within Gaucher, patients with type 3 disease (GD3) have specific needs that are yet to be addressed. The onset of seizures (and their effect on prognosis) and the tremors, fatigue and balance issues that accompany the disease all have a significant impact on patient self-confidence and social life activities and should be further explored using real world data. Anxiety about the future is a constant burden for patients and their families. Mental health problems such as depression and anxiety remain major issues and should be an integral part of the clinical management. Although the small number of GD3 patients makes specific outreach

and support activities more difficult, it is important to identify specific opportunities that will help raise the profile of the GD3 community and their requirements.

Outside of medical research and education, European patient groups remain the key emotional and supportive link for the Gaucher communities in their countries and are working to deal with new problems. An example is the increasing number of patients diagnosed in childhood but surviving to the point where they need to move to adult care. This is bringing new challenges for healthcare systems and transitioning to adult treatment centres can be challenging, as occasionally there is insufficient handover between the respective physicians.

Patients suffering from rare diseases are known to have more mental health issues than the general population and, although clinicians realise the need to integrate mental health into the overall treatment plan, funding issues are causing delays and a lack of specialist care. Even when available, general mental health services are often not tailored to address the specific needs of Gaucher patients.

Again, patient groups are addressing these issues by building more effective communities for their patients. At their heart is the emotional support they provide and the benefits that provides for mental health. Regular group meetings allow both patients and their families to talk about the disease, how it affects their lives and to share their experiences – processes that are

proven to reduce anxiety, depression, and other mental health issues.

All the patient groups agreed the medical care for Gaucher patients has improved enormously over the past decade, but there remain many unmet needs for the patients they represent. Access to treatment remains a major issue in some European countries and more research is needed into understanding the correlation between Gaucher and other diseases, the effect of nutrition, and the use of food supplements such as vitamin D for bone health or vitamin B complex for neuronal nutrition.

Whatever the future of Gaucher care, patient groups across Europe are continuing to care for their patients through increasingly effective education, support and the sharing of best practices. As knowledge about likely comorbidities becomes more available, Gaucher patient groups will also need to work closely with those from other disease areas to ensure the information and advice they provide remains current and beneficial to their communities.

in many research studies ranging from; longitudinal surveillance studies, registries and early and late phase clinical trials of new drugs. We pride ourselves for being an active research centre that has recruited the first international participants onto registries and even first national participants on to complex clinical drugs trials. BCH is an accessible research centre for local, national but also international patients.



# GAUCHER MASTERCLASS IN SOUTH AFRICA

On Tuesday 17th October, Tanya Colin-Histed of the EGA, Prof Chris Hendriksz, Dr Greg Pastores and Dr Hylton Sevitz ran a MASTERCLASS in Cape Town South Africa. This was joint venture between the EGA and the UK Gauchers Association who provided funding through the Susan Lewis Memorial Fund to support many of the attending doctors.

**Tanya writes; The Gaucher Masterclass attracted physicians from all over Africa including Cameroon, Botswana, Sudan, Rwanda, Malawi, South Africa and Zimbabwe. Rare Disease Botswana were also represented by their cofounder and mother of two children with a rare disease and Aimee-Kate Bosch, a Gaucher patient and a member of the Gaucher and LSD Society of South Africa.**



Delegates at Gaucher Masterclass South Africa.

The idea for this masterclass came from a) the EGA's work in educating doctors and b) following several applications to the EGA from several different countries in Africa for help in getting access to ERT through charitable access programmes as Governments in these countries do not provide the treatment through public health programmes, although a few patients in South Africa are now receiving ERT from the Government, these are in the minority. As these doctors are only treating one or

two patients it is important to a) improve their knowledge of the disease for diagnosis and clinical management and b) to develop a sustainable network for ongoing support and guidance.

Prof Chris Hendriksz led the masterclass and donated his time to develop the programme and identify the speakers. Chris, a doctor from one of the LSD treatment centers in the UK, South African born and his wife Flo have set up

a company that are making educational videos to support the development of knowledge and clinical practice guidance for metabolic conditions and they videoed the masterclass to provide ongoing tutorials for the participants that they can tap back into.

The day focused on areas such as an overview of the disease and its pathology, highlighting its clinical signs and symptoms, a description of the different sub groups of the disease and the challenges of diagnosis. One of the morning sessions led by Dr Karen Fieggen from XXXX who highlighted the challenges of diagnosis in an environment where many children and adults had other clinical/health challenges such as TB, HIV and malnutrition. Topics such as co morbidities, the use of biomarkers and different types of therapeutic options were discussed, often

with lots of questions during the sessions which stimulated a lot of very valuable input and awareness of local challenges.

In the afternoon, the focus was on supporting the patients and families, the development and sharing of clinical guidelines and the importance of building networks in countries. I finished the day with a presentation of the value of working with the patient organization outlining my story as a mother, which is often at the heart of many rare disease patient groups, the work of the UK Gauchers Association as an example of how to support patients and their families and then the strategic role of the EGA.

The Feedback from the masterclass has been very positive, both on the day and through the feedback forms we have received back from the participants.

**The Susan Lewis Memorial Award provided bursaries for a number of doctors to attend this meeting please read their reports below:**



Ayman A. Hussein

**Ayman A. Hussein writes** 'Thank you for sponsoring my attendance at the 2016 Gaucher Masterclass held in Cape Town, South Africa. My name is Ayman Hussein, I am from Sudan, and I am a medical doctor and a registrar in training in Medical Genetics at the University of Cape Town. I found the course very useful not only because of the excellent content in the presentations but also because of the opportunity to meet and interact with other course attendees and the organizers. As well as being a useful update on the science and

clinical management of Gaucher, it was an opportunity to learn more widely around practical considerations and social, ethical and legal issues that are relevant to it'.

**Eda Selebatso writes** 'My name is Eda Selebatso. I am a mother of two children with rare conditions. I Co-founded the Botswana Organisation for Rare Diseases (BORDIS) with my husband in 2015. I am the chairperson of BORDIS. To meet the demands of leading BORDIS I have to pursue personal development in areas that address the needs of rare disease patients. That



Eda Selebatso



includes knowing about conditions which is why I was very interested in this class. I got invaluable networks and information which I immediately used when I got back home. I already have been in touch with Prof Hendricksz on some of our patients to find better ways to proceed and that has been wonderful. I also now have doctors with some ideas on management and know where to refer their patients. This for us in Botswana is hope that tomorrow can only be better'.

**Dr Dipesalema Joel writes** 'I am Dr Dipesalema Joel, a Consultant Paediatric Endocrinologist practicing at Princess Marina Hospital in Gaborone, Botswana. I am also a Senior Lecturer in the Department of Paediatrics and adolescent Health at the University of Botswana. Throughout my career ranging over a period of 14 years, I have worked with patients with rare diseases, in-born error of metabolism and endocrine disorders. As a clinician in Princess Marina Hospital

which is the main tertiary referral centre for Botswana, I work with these groups of patients on day today basis. As such it was quite resourceful for the European Gaucher Association to sponsor me to attend the Gaucher Master Class.

The Gaucher Master Classes were quite resourceful to me as I learned from the experts in the area as well as patients who lived with this disorder. I learned how to look after patients with Gaucher's disease. I would highly recommend this workshop for anybody who works with patients with rare diseases and I will certainly attend it again'.

Further Doctors who were funded by the Susan Lewis Memorial Award were Monkogoi Goepamang, Farai GW Chinhoyi from Botswana; Tapiwanashe Bwakura from Zimbabwe and Cedrik Ngongang from Cameroon. Unfortunately their reports have not been received in time to print this edition of Gaucher News.

Full details of the five areas of funding opportunities within the Susan Lewis Memorial Fund can be found on our website at [www.gaucher.org.uk](http://www.gaucher.org.uk) and also available as a PDF or hardcopy leaflet. Email Sarah in the office on [sarah@gaucher.org.uk](mailto:sarah@gaucher.org.uk) or call 01453 549231 and she will email or post you a copy.

## THE SUSAN LEWIS MEMORIAL AWARD

### DR ELAD SHEMESH



Elad Shamash

Since the last edition of Gauchers News, the Association has supported Dr Elad Shemesh, an Israeli medical doctor and a member of the Cochrane Collaborations' Genetic disorders group to attend and present at the prestigious in Seoul, October 2016.

**Elad writes –**  
'The Cochrane collaboration is a multinational nonprofit

organization of clinicians, statisticians and healthcare consumers working together to systematically review clinical trials in order to consolidate conclusions, mainly regarding efficacy and safety of drugs. These conclusions are then disseminated to inform healthcare professionals and patients about clinical effects of different treatment options. Briefly, the term 'Systematic review' refers to a complex mathematical procedure in which data from published studies are re-evaluated and compared from a new statistical viewpoint, alongside an assessment of possible biases in those studies (such process is generally named 'evidence-based medicine'). While the conduct of systematic reviews is now considered critical in consolidation of recommendations and guidelines of treatments in most of the common medical conditions, its use in rare disease has been limited.

In 2015, in collaboration with a group of experts in Gaucher disease - Professor Timothy Cox (Cambridge University), Dr. Patrick Deegan (Cambridge University), Professor Carla Hollak (Amsterdam medical

centre), Professor Neil Weinreb (Florida, USA), Professor Bruno Bembi and Dr. Laura Deroma (Udine, Italy), we published a systematic review on the treatment options for Gaucher disease. We have been working on two exciting follow-ups since then: An update of our analysis to include the two new eliglustat clinical trials that have recently been published- in order to inform the National health care system committee prior to publication of guidelines regarding its use (this update is expected to be completed and published in early 2017), and, on a set of suggestions regarding how treatment options in rare diseases should effectively be measured and how to enhance the applicability of systematic reviews in rare conditions.

Through the generous support of the UK Gaucher association via the Susan Lewis Memorial fund, I was able to attend this year's Cochrane collaboration conference (held in Seoul, October 2016) and share our ideas. My participation in the conference has also granted me the opportunity to learn new research techniques and to appreciate future trends in evidence-based medicine.

I would like to thank the UK Gaucher association for their commitment and continuous support in our research projects.

# THE EGA GOES TO PAKISTAN

On the 6th October 2010, the EGA received an email from the brother in law of Atif Qureshi, the father of Hijab who at the age of almost 2 years had been diagnosed in Pakistan with Gaucher disease. The email was a plea for help to get enzyme replacement therapy (ERT) for Hijab through charitable access as the Government in Pakistan did not provide this treatment. A few months

later with the support of Genzyme we were able to call Atif and give him the good news that Hijab had been accepted onto the Genzyme International Cerezyme Access Programme (ICAP). Atif later told me that the day I called him he was in the market with his Mum.



**This one plea for help was the start of the EGA's awareness of the suffering of the children of Pakistan who were dying of Gaucher disease because they did not have access to ERT.**

Over the next 6 years the EGA have worked alongside Atif and Prof Huma Cheema, a Gastroenterologists at the Children's Hospital in Lahore to support the development of clinical knowledge amongst her and her colleagues throughout Pakistan through video clinics and invitations to international meetings where she has presented on the situation and challenges in Pakistan to raise awareness and for her to develop a network of contacts with the key opinion leaders globally.

Through the EGA's work we have successfully managed to access ERT through Shire Charitable Access programme (CAP) in 2016 for a number of patients; are currently exploring the possibility of Orphazyme running a clinical trial for GD1 and 3 in naïve patients in the country with Arimoclomol and are looking at the possibility of an ERT and Bone Marrow Transplant (BMT) programme, whilst controversial ERT is not and will not in the

near future be available to all the untreated children in Pakistan and alternative solutions MUST be found as there are still many children remain untreated.

Tanya writes; Through the many meetings, video clinics and telephone calls I have longed to visit Pakistan to meet Atif, his family and the children and families in Pakistan to further understand their challenges and to use the voice of the EGA to talk to Government. So, after 6 years on Monday 7th November I flew to Pakistan with my dearest friend Dr Vellodi, a retired Paediatrician from Great Ormond Street Hospital to spend a few days at the children's hospital to meet families, speak to doctors and help in any way we can to improve the lives of the Gaucher patients and their families. I would also after 6 years meet Atif in person, it would be a very emotional trip in so many

Day one, after a delayed arrival, due to fog we dropped out bags at the hotel

and went straight to the hospital to meet Prof Huma Cheema and her team. I had the privilege to talk to lots of Gaucher families and I finally met Atif and his lovely family. Later Atif and I met with Genzyme's representative in Pakistan to identify the steps to set up a formal LSD Patient Group in Pakistan.

After a day in the clinic I had a late-night coffee meeting with Prof Huma Cheema, the Medical Director and the Health Secretary for Lahore Children's Hospital to raise the importance of the work being done by Prof Huma Cheema and her team focusing on her Fellowship Programme and the importance of access for treatment for patients. We also discussed their support on getting the necessary regulatory



approvals for a proposed clinical trial, agreement for support for a potential BMT/ERT programme and how to provide access to the ERT Eleyso into Pakistan.

Day two, an early morning start with a lecture by

Dr Vellodi on the management of LSDs and then I talked to the medical students, Fellows and Faculty Heads about the work that the EGA has been doing with Prof Huma Cheema to support the unmet needs of Pakistani children with Gaucher Disease. Afterwards we were invited to have tea with the Dean of the Hospital and then onto clinic to meet patients with Gaucher, MPS and Niemann - Pick. Lunch was off site at a restaurant with many heads of departments and the Dean and Medical Director of the hospital.

The late afternoon was spent at a private museum and then a spot of shopping with

a Prof Huma Cheema in Liberty Market. The day ended with dinner with Huma and her wonderful team overlooking Lahore.

Day three was my last day in Pakistan. Today at the hospital was an MPS clinic and Dr Vellodi saw many patients working alongside Dr Huma Cheema's team offering guidance and sharing knowledge of clinical management.

I spent the first part of my day with Atif talking about his plans to set up an LSD Society in Pakistan. I then had the privilege to go to his house to meet his family and we visited the famous Shallamar Gardens continuing to talk about the many challenges facing Pakistani patients and their families.

Then it was off to tea at the Chief Minister of Punjab's house to meet him and his wife to raise awareness of the work being done by Prof Huma Cheema, its value and the impact it has on the patients and families. We also talked about the need for further support for patients with resources and how he can support by making pathways smoother for importation of charitable treatment, approval for clinical trials and a financial commitment to support the costs involved in providing BMTs to patients.

It was a successful trip, we managed to do a lot of things in just a few days that has planted the seeds that now need attention from all of us to grow, to help the doctors, patients, families and Government to improve services such as family support, diagnostics, disease awareness. To build on the good clinical knowledge and to support access to treatment.





## FUNDRAISING

# MEMBERS FUNDRAISING

### Donations received from June to December 2016 totalling £ 8,419.29

Generous donations have been received from: Dr R Rees, Mrs H M Garbett, J Bray, M & J Picking, Mr & Mrs Moore, Susan and Ian Richardson, Oliver and Jones Associates and Royston & Lund Estate Agents.



Rebecca Troman-Brown and Trevor Brown made a generous donation and sent these words: - As part of celebrations on our wedding day on the 20th August 2016, we decided to give our guests money tokens to give to our chosen charities in lieu of traditional wedding favours. The Gauchers Association has always played an important part in my life, as my sister has the disease, so it was important for me to choose this as my charity, alongside one chosen by my husband. Throughout the reception, our guests came up and added their tokens to our charity jars and it really added a special part to our day. We are thrilled to be able to donate £150 to the Association, a charity close to our hearts.

Congratulations to **Mark Bardoe** who became a member of Club des Cingles du Mont-Ventoux or "The Madmen of Mont-Ventoux" by completing the challenge to climb this famous mountain by its three paved routes in one day. The climb was a total of 19000 feet over 43 miles and a fantastic total of £1342.96 raised.

Vitality London 10k Sarah and Helen from the Gaucher office, Ilana Manuel, along with Emily Lew, director, and her sister Katy flew the flag for the Association and ran the Vitality London 10k in July raising an amazing £5780.29.

We have 6 places available again in 2017, if you are interested please contact Claire for information on [admin@gaucher.org.uk](mailto:admin@gaucher.org.uk).



The Association relies on its members, their families and their friends who generously support the Association's work through subscriptions, donations and the organising of fundraising events.

We are, as always, extremely grateful for this support and would like to remind you that we have various fundraising resources available to help you such as balloons, t-shirts, running vests, thunder sticks, ponchos, stickers, posters, leaflets and a raffle licence.

We would like to thank all our members who generously donated additional funds with the payment of their annual subscription along with those who pay a regular monthly standing order.

### DONATE ONLINE

We rely on your generous support to enable us to continue to meet the needs of those suffering from Gaucher disease.

Donations can be made at [www.gaucher.org.uk](http://www.gaucher.org.uk)

If you would like to set up a regular standing order please contact Sarah on [sarah@gaucher.org.uk](mailto:sarah@gaucher.org.uk) or 01453 549231

### DONATE BY TEXT

You can also donate via text  
To donate now simply text  
GAUK01 £2 or GAUK01 £5  
or GAUK £10 to 70070

### *giftaid it*

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, it means that you'll actually be giving us at least £125.

Giving under Gift Aid means that so much more money can be raised at no extra cost to our donors. £1,451.67 was claimed during 2015

## FUNDRAISING

# LONDON MARATHON 2017

The marathon is one of our biggest and most important fundraising events of the year. Five runners completed the course in 2016 raising over £11,500 through the generous support of their friends and family as well as our members and we're hoping to do even better in 2017.

Everyone in the Gaucher community wishes our runners Mat Abramsky, Adam Conter, Dani Lucas, Max Bassadone and Daniel Rees the very best of luck with their training over the coming months. Please read their personal stories below and support them as much as you can.



**Adam Conter writes;** forever the optimist, I am a 33 year old Canadian more designed for hockey spectating than long distance running. With family and friends living in London, what better excuse to pay a visit to the commonwealth homeland than to prepare for the London Marathon. Training has begun for this young, slow, overweight father of 2 (under the age of 18 months), the arrival at the finish line will certainly be a triumph personally and a great accomplishment. Looking forward to training hard and raising money for a great cause, Adam is proud to be a member of the Gauchers Association fund raising team.

**Max Bassadone writes:** I am a 31 year old who is a keen short distance runner, 5-10km, and looking to test myself with a much(!) longer run. Aspirations of achieving sub 4 hours but will be happy to complete it and raise funds for a very good cause



**Mat Abramsky writes:** Having started my running career at the 2015 British 10k London Run, running for the Gauchers Association, I got the bug. With a brief hiatus whilst trying my hand in some triathlons in 2016, the 2017 London Marathon will be my most significant challenge yet...and I'm terrified! I work in property...which doesn't move, and certainly not quickly, so it's uncertain why I've taken up distance running. But I've dragged my cousin, Adam Conter, east of the Atlantic, and teamed up with my work colleagues Dan Rees and Max Bassadone, to help me across the finish line. I also hope raising some dosh for a fantastic charity like the Gauchers Association yields the karma-hydrates required for me to finish 26.2 miles. I'm proud to wear the Green and White for the Association once again.

**Daniel Rees writes;** I'm 28 years old and work with Dan Brown at Brockton Capital. I'm a keen cyclist, and not such a keen runner, so this is my first marathon. I've done a short number of charity events before normally on two wheels. I'm very excited about the day itself and look forward to raising plenty of money on the way for Gaucher. Thank you for the opportunity.



Dani Lucas has chosen to run the London marathon following on from her daughter being diagnosed with Gaucher disease.

**Dani writes:** I think that our diagnosis story will probably have similar features to other Gaucher families. It starts with unexplained symptoms and bemused doctors followed by wrong turns, changes of diagnosis, disbelief and stomach turning panic

In 2013 my husband, me and our 5 year old daughter Iris were living overseas in Lithuania. Following a minor illness it was noted by doctors that Iris's liver and spleen were enlarged. We were told it was probably a post viral reaction to a tummy bug. On a follow up visit some months later Iris's liver and spleen were still enlarged – although both organs were functioning normally. A medical anomaly that on our return to the UK was monitored by regular scans. Our paediatrician suggested that maybe Iris just had a large liver and spleen she had yet to grow into. In all other respects she was healthy – with all routine blood tests coming back normal. I don't think any of us – me, my husband, or the paediatrician, believed that the situation was normal – but with no other symptoms to investigate we were left with an unsatisfactory question mark.

In 2014 we moved back to the UK and Iris started a new school. Walking to school one day she complained of a pain in her ankle. Within weeks Iris's feet, ankles, knees, and elbows were swollen and Iris was finding it difficult to walk, sit, or stand.

As a child I suffered with Juvenile Idiopathic Arthritis (JIA), my niece and nephew both have JIA – so I was fully aware of the symptoms. Iris was quickly diagnosed with Polyarticular Juvenile Idiopathic Arthritis, (PJIA) and admitted to hospital to have the affected joints injected with steroids. Our Rheumatologist was keen to start immune suppressant drugs to treat the cause of the arthritis – however Iris needed to have a lumbar puncture first to rule out other diseases such as leukaemia. It was routine procedure –and we were reassured that with my family history it was almost certain that Iris had PJIA.

The evening after the procedure our

consultant called me at home. Now don't panic – but we have found the reason for Iris's enlarged liver and spleen. Her lumbar puncture showed signs of Gaucher cells. We think that she has Gaucher disease.

Then came many hospital appointments in Plymouth, Bristol and GOSH and many tearful consultations. What was Gaucher disease? Was Iris type 1 or 3? What was the treatment? Was there a cure? What was the long term prognosis?

Well, in time we knew that Iris had Gaucher Type 3, that there is treatment - which would be life-long, that there isn't a cure, and that we do not know how/when/if the neurological element of the disease will progress and what this will mean for Iris.

We were told that whilst arthritis can be a symptom of Gaucher Disease, in Iris's case her PJIA was completely unrelated. In a strange way we have PJIA to thank for the diagnosis of Gaucher Disease. I often wonder when Gaucher would have been diagnosed had PJIA not reared its head.

Iris started her fortnightly ERT at GOSH just before her 7th birthday in May 2015. We were eventually released for homecare treatment in October 2015. She also started immune-therapy for PJIA at the same time. I'd like to think that our diagnosis story finishes with an acceptance of the diagnosis and an uncertain future, but I think that's something that we work on day to day – hospital appointment to hospital appointment.

Iris is a happy, quirky, and unbelievably brave girl. She's overcome a lot in the last few years. Her ERT treatments with Jeanne from Healthcare at Home have become something she looks forward to - I told you she was quirky! She doesn't let Gaucher Disease, or her PJIA define who she is and I am incredibly proud of her.







## The Gauchers Association CHARITY BIKE RIDE 2017

SUNDAY 21st MAY 2017

We are very pleased to announce we will be repeating our charity bike ride after a year off with a brand NEW revised 60 mile CIRCULAR Route on

**SUNDAY 21st MAY 2017 - PLEASE SAVE THE DATE**

Start / finish Old Owen's Sports Ground, Potters Bar, EN6 4NE

**This event will be:**

- Brand NEW 60 mile CIRCULAR route
- FREE refreshments stops along the route
- Open to all cyclists
- Under 16's to be accompanied by an adult
- Fully marshalled route
- Parking at the venue

Limited number of places available so REGISTER NOW!

A fee of £25 will be payable on registration and there is a minimum £100 sponsorship pledge

If you require further information or have any questions please contact either Sarah or Claire on 01453 549231 or visit our website at [www.gaucher.org.uk](http://www.gaucher.org.uk) **We look forward to seeing you there!**

## The Gauchers ASSOCIATION

Fancy a  
Challenge?

Is 2017 the year that you do  
something awesome for  
The Gauchers Association?



**great north run**  
10th September

**2017**

**PRUDENTIAL RIDE LONDON**  
30th July

**great newham london SWIM**  
1st July

**SPORT WESTMINSTER BRITISH 10k**  
9th July

**BIRMINGHAM INTERNATIONAL MARATHON**  
15th October

Newcastle

Birmingham

London

Virgin

Let us know - get in touch!

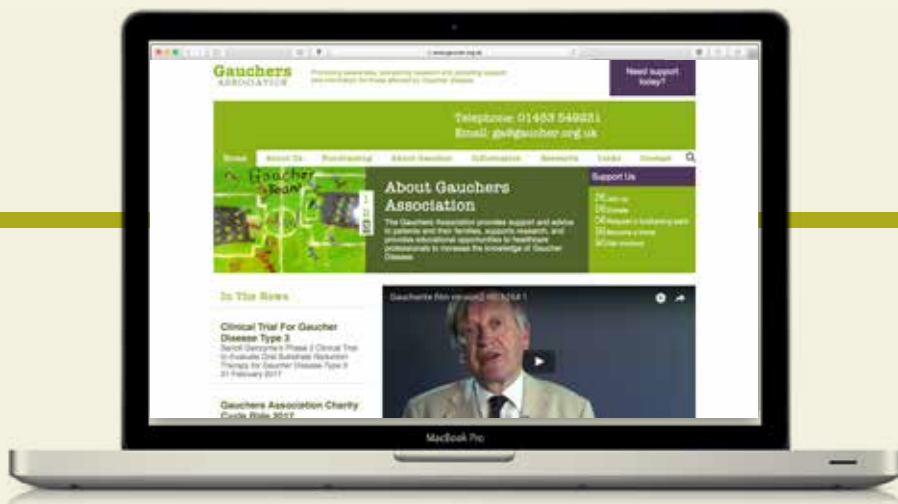
[www.gaucher.org.uk](http://www.gaucher.org.uk)

or contact Claire on 01453 549 231

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