

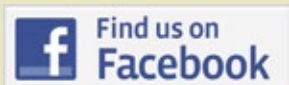
GauchersNEWS

December 2015

Gauchers ASSOCIATION



**Colour run raises
over £1,000 for
Gene Therapy**



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The Gauchers Association Limited
Registered Charity No. 1095657.
Registered in England & Wales No. 4468323.
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Front page top photo: Abdul Waheed and cricket
players. Bottom photo: Aura Cruz and family

Chairman's Chat

Dear Friends,

Welcome to the December 2015 edition of Gauchers News.

I wanted to start my Chairman's Chat by mentioning the 7th nGD Family Conference which was held in Manchester on 14th/15th November. It was fantastic to see so many familiar faces as well as some new ones. We truly are very lucky to have such a close, collaborative and supportive community and the conference was a wonderful example of that.



The weekend was carefully planned and organised by a very special group of young women who have all been participating in the Association's Empowerment Project that has been running now for a couple of years – Irma, Maddie, Nadia, Saphia, Radhika and Sara.

The Project has allowed them to become involved in a broad range of activities, aside from organising the past two nGD conferences, that have included the production of a new booklet for Type III young adults, work experience, trips abroad to visit Gaucher clinics in developing countries, public speaking and joining Association board meetings.

We have seen these young women mature and develop over the past few years and embrace everything that has been thrown at them. In summing up at the Conference I told them that I felt inspired by listening to their personal stories and watching their achievements and I came away from the Conference feeling extremely proud of the contribution they continue to make to the Association and the wider Gaucher community. I hope that this continues for many years to come and that they can lead the way for younger patients to follow in their footsteps.

Reviewing this edition of Gauchers News I have been blown away by the multiple fundraising efforts of our members, their families and friends over the past 6 months – there are 12 pages packed full of stories of how people have been raising money for the Association including an iron man (a special mention to Mark Histed here for his super human exertions!), the Marathon, the golf day, the Great North Run, the 5k Colour Run, Ride London, Tough Mudder, a cricket day and of course the Association's annual bike ride. A sincere thank you to everyone involved in fundraisings initiative held on behalf of the Association.

Finally, I can't sign off without plugging the Association's 25th anniversary next year! We are planning a special celebration on 5th November and would love as many of you as possible to join us so please save the date and look out for further details on our web site and in the June 2016 newsletter.

Wishing you all well over the festive season and a healthy and happy new year.

Dan

“Learning To Infuse Was The Best Thing I Did”

Personal stories are important to us. Not only do we appreciate the time it takes to put thoughts onto paper (or a computer screen these days), it gives us insight into what goes on in the lives of those living directly or indirectly with Gaucher disease. Here is one such story written by mum, Donna:



Tommy Whittaker

‘When we first got told Tommy had Gaucher disease in July 2013 we were devastated. We had no idea what it was or what was going to happen. We had never heard of it before and we quickly realised not a lot of people had.

We went home and hit the internet armed with nothing more than the name of an illness and the more we read the more worried we became. We barely understood anything we were reading but kept reading anyway.

Over the next few months we watched how Tommy took it all in his stride, accepted people prodding and poking him and the endless tests, not to mention the cannula that went in his hand every other week in hospital. Yet despite all this he never complained or made it difficult. It was then that we realised just how lucky we were. Yes, Tommy had this disease but watching him playing and going to nursery school you wouldn’t know anything was wrong with him and that alone makes us luckier than some.

Having infusions done in our home was definitely easier but Tommy didn’t seem to like a stranger coming in and I didn’t like him missing school so I asked if I could learn how to do it myself. It was the best thing I did. It is now just part of our everyday life, just something we do every other week after school. If we have a sudden change of plans on infusion day like a party for instance it’s not a problem, we just do it later or the following day and because Tommy is so used to me doing everything for him, he doesn’t ever moan or question it.

We have no idea of what the future holds for Tommy in terms of health but in all honesty even if he didn’t have Gaucher disease we still wouldn’t know.

I personally live everyday as it comes and thanks to Enzyme Replacement Therapy, Tommy is living his life, so far, a very happy little boy.

We treat Gaucher with Tommy the same way we treat our daughter’s colour blindness. It is just something about them that makes them who they are.’

Emily Rose Cavanagh

1996-2015



Miss her so much - - - - -
 "Softly the leaves of memory fall,
 gently we stoop to gather them all."
 my beautiful forever Angel - - -
 Emily Rose. x x x x Love Mum

Tanya Collin-Histed, Chief Executive, writes: Emily lost her battle and life to neuronopathic Gaucher disease at just 19 years old, she should have had her whole life ahead of her but it was taken from her cruelly on the 15th July, she died at home with all who loved her. My heart goes out to Emily's family; there are no words to express the sadness that Emily's loss has had on our small but close nGD community in the UK.

Patient and Family Support Worker update

Two years have passed since I started my role as Patient and Family Support Worker and it has been a real privilege to meet so many of you both individually and at events and meetings.



Helen Whitehead

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

I have information on claiming for benefits and can support you with the process such as claiming for PIP (Personal Independence Payment), ESA (Employment Support Allowance) and DLA (Disability Living Allowance). As you'll be aware, changes to legislation and rates change on a frequent basis and the application process can be a lengthy one so my advice is to get in touch as soon as possible to start the process.

In addition to helping to make financial claims, I can support patients with hospital visits and clinic appointments, liaise with schools, help access home adaptations, help with writing a CV and lots, lots more! Please do contact me if there's anything at all I can help you with. It's what I'm here for!

What is PIP?

PIP stands for Personal Independence Payment which is replacing DLA (Disability Living Allowance) for 16-64 year olds living in the UK. If you currently get DLA and are aged 16-64 you will be invited at some point to claim for PIP. All new applicants will claim for PIP. The payment is to help with some of the extra costs caused by long-term ill-health, a disability that affects your mobility or to do the activities associated with daily living (such as washing, dressing, cooking etc.) and you can claim this benefit regardless of your employment situation.

If you think you might be entitled to PIP, phone the DWP (Department for Work and Pensions) free on 0800 917222 and they will send you a 'How Your Condition Affects You' form. Once this is returned you will get invited for an assessment, normally face-to-face, with an independent healthcare professional. A decision is then made on your PIP claim. For more information please visit www.gov.uk/pip/overview.

If you need any help at all navigating PIP, please don't hesitate to contact me. There will be a section on benefits on the Gauchers website soon so look out for it.

Alison Wilson – Gaucher in Ireland

I hope this edition of the Gaucher News finds you well. This will be a relatively short update with a ‘call to action’ for all those living with Gaucher in Ireland.



Alison Wilson

In the past few months I have had the pleasure of supporting some of our Gaucher patients at clinic appointments and with a range of social welfare issues (including access to disability benefits and insurance).

It is always a pleasure to be able to point people in the right direction when they are feeling a bit ‘stuck’. What has become clear to me is that a good number of our Gaucher patients in Ireland have never met anyone else affected by the condition and very many feel they would like to have more information about their condition. This is something I would like to change. For starters, we are working alongside local clinicians to arrange a Gaucher Information Day in Dublin in the spring of 2016. This will be an excellent opportunity not only to meet others and learn about Gaucher disease but also to have your say in how services for people with Gaucher in Ireland are delivered.

Also, you can expect to receive an information pack and brief questionnaire by post in the first quarter of 2016. The aim of this will be to give you some information about what services are out there and also to determine how your condition is managed. And we want to understand a bit more about how many appointments you attend and who these appointments are with. We also want to hear your opinions on how your care could be better managed. This will hopefully be a springboard for developing a greater understanding of how to make the most of Gaucher disease management in Ireland.

If you are living in Ireland and have any unmet support needs or you need more information about any and all aspects of Gaucher disease, please do not hesitate to get in touch with me by phone on 028 950 47779 or 07786 258 336 or by email at alison@gaucher.org.uk. Best wishes to you all and I’ll look forward to updating you in the next edition of Gaucher News.

Season’s Greetings

The Gauchers Association would like to wish all our members, families, friends and collaborative partners Seasonal Greetings and to thank everybody for their support throughout 2015 and we look forward to lots of exciting and challenging projects in 2016 (see page 17).



The Great Ormond Street LSD Team

At Great Ormond Street Hospital (GOSH), patients with lysosomal storage disorders (LSD) are seen by a large multidisciplinary team (MDT). The patient's pathway to diagnosis often starts at their local hospital where the suspicion of LSD is raised. Blood and urine tests are sent to the Enzyme Laboratory at GOSH where a biochemical diagnosis of LSD is established. Close collaboration between the enzyme and molecular genetics labs allows genetic confirmation and future prenatal diagnosis.

Whilst investigations are underway the patient and their family are referred to and seen by one of the metabolic consultants and Clinical Nurse Specialist (CNS). Following initial assessment and depending upon the clinical need, children are referred to other specialities within GOSH, such as Speech and Language Therapy, Physiotherapy, Cardiology, Orthopaedics, Neurosurgery, Ophthalmology, Bone Marrow Transplant, Dental, Ear Nose and Throat and Respiratory/Sleep and Symptom Care. As a result, these specialities have a wealth of experience and expertise in the management of such rare conditions and in addition, we can coordinate these appointments to reduce multiple visits to the hospital.



The team at Great Ormond Street

The metabolic team has two wards – Rainforest Ward for long term patients and Kingfisher Ward which is a programmed investigation unit used for patients admitted for planned investigations and/or seeing multiple teams. Children are usually admitted to Kingfisher when they commence enzyme replacement therapy.

The metabolic team offers 24/7 advice and support for their LSD patients as required and the CNSs are available to provide support and advice during working hours. Due to both the complexity of the patients and the multidisciplinary approach that is taken, the whole team meets on a weekly basis to discuss patients.

Follow up of LSD patients are in consultant-run clinics and MDT clinics. The MDT clinics are disease-specific where we aim to see patients alongside the key specialities sharing their care and the patients are also seen by Speech and Language Therapy (SALT) and physiotherapy depending on clinical need. The team is well supported by both our social worker and clinical psychologist. Each consultant follows up patients from across the entire range of LSDs and in addition maintains a special interest in a smaller group of conditions in order to support up to date knowledge and research interest. The lead consultant in the team for Gaucher disease is **Dr Anupam Chakrapani**.

The metabolic team is also actively involved in research in LSDs, both at a basic science level and by running clinical trials in the Clinical Research Facility.

Consultants

Dr Lara Abulhoul
 Dr Anupam Chakrapani
 Dr Maureen Cleary
 Dr James Davison
 Dr Emma Footitt
 Prof Paul Gissen
 Dr Stephanie Grunewald

Clinical Nurse Specialists

Ingry Camero
 Sindi Mnkandla

Laboratory

Dr Clare Beesley
Clinical Scientist (Molecular Genetics)
 Derek Burke
Chief BMS, (Enzyme Lab)
 Katie Harvey
Clinical Scientist, (Enzyme Lab)

Allied Health Professionals

Noelle Brackett
Social Worker
 Dr Angela Simcox
Clinical Psychologist
 Gyani Swift
Speech and Language therapist
 Michelle Wood
Specialist Physiotherapist

Genzyme re-engage with NICE to bring Cerdelga to Gaucher Patients in England

In December 2014's edition of Gauchers News, we reported that Genzyme had made the decision to put a hold on the technical appraisal that was being carried out by the National Institute of Health and Care Excellence's (NICE) new Highly Specialised Technologies Appraisal Committee, whilst they continued to develop their pricing model for Eliglustat in the UK to ensure that patients are able to be prescribed a treatment based on their clinical needs rather than based on cost.

Eliglustat (marketed under the brand name Cerdelga™), Genzyme's new oral therapy for Type I Gaucher disease that was given marketing approval by the European Medicines Agency (EMA) in January this year.

We are delighted to announce that Genzyme have re engaged with NICE, updated information on the timings of the evaluation will be available on the NICE website shortly <https://www.nice.org.uk/guidance/indevelopment/gid-gaucherdiseasetype1eliglustat/id709> The Gauchers Association who took part in the original scoping exercise in January 2014 will play a key role in submitting patient evidence to the NICE committee. In preparation for the previous appraisal that was halted in late 2014, the Association undertook an in-depth survey with the Type 1 Gaucher community in England to seek the views of our patient community and we will repeat this to make a strong case for Cerdelga to be approved for NHS England funding so that patients who wish to switched to the new oral therapy and are clinically suitable will have that choice.

The Association will liaise closely with Genzyme and NICE and will communicate any updates through our website and Facebook page.

The Association is 25! Save the date...

Well not quite yet, but we are already planning our 25th anniversary celebrations in London, make sure you save the date – Saturday 5th November 2016!



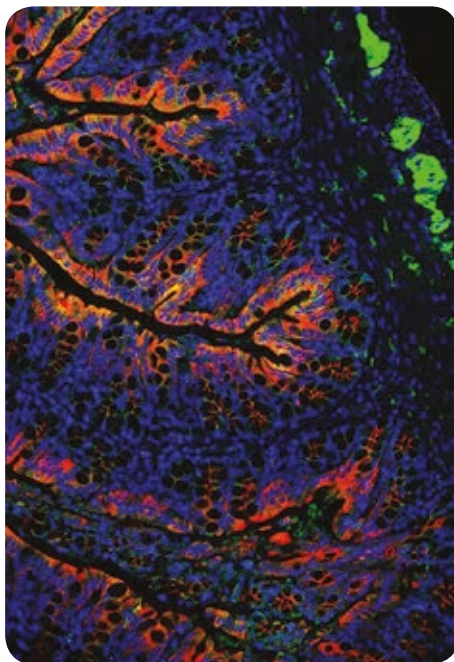
Gene therapy as a new potential treatment for Gaucher disease

In June's edition of Gauchers News we reported that earlier this year the Association were awarded orphan designation for adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene (Gene Therapy) as a new potential treatment for Gaucher disease from the European Medicines Agency (EMA). This first step allows us to start the process of achieving regulatory clearance for a potential clinical study into gene therapy.

Over the last six months, a small project team consisting of members of the Association, researchers Dr Ahad Rahim and Dr Simon Waddington and their team at University College London (UCL) and Elin haf Davies as an independent Consultant have been working behind the scenes to explore and move forward with the numerous complexities involved in such a project.

In a commitment to keeping our members up to date, a session was held at the nGD family conference on Sunday 15th November where our nGD patients and families listened to an update on the clinical work and were able to ask the researchers questions.

A more detailed report on our progress will be included in the June 2016 edition of Gauchers News.



New National Homecare Specification for LSD Services

‘As part of the national specialised service for lysosomal storage disorders, patients with an LSD are able to receive care that allows them to receive their fortnightly infusion in their own home. The first ever national framework (covering all eight LSD centres in England) ran from 2012 until it finished at the end of September 2015.

Representatives from each of the eight LSD centres, with representation from the UK LSD Patient Collaborative, have been working hard for the last 18 months to update the specification. Taking learnings and feedback from the last specification, the new framework began on the 1st October 2015. We are delighted to announce that a third company, Pharmaxo, will join existing providers Bupa and Healthcare at Home on the new framework, which will offer more choice to patients.

Patients and family members will hopefully see no major changes with the new homecare framework and we are hoping for a seamless transition and service as normal’, writes Tanya Collin-Histed, Chief Executive of the UK Gauchers Association.

Alison Davies, General Manager at Pharmaxo introduces the company: ‘Based in Corsham in Wiltshire, Pharmaxo has been offering innovative, high quality pharmacy services since 2010. Building upon this success we commenced homecare services in October 2013 providing home infusions. We have continued to develop our areas of expertise and provide homecare across a range of therapies including oncology, immunology and respiratory conditions, and now, LSD.

Pharmaxo, drawing on the experience of our sister organisation Bath ASU, develops services which are tailored to specific therapies. This enables us to focus on meeting the needs of the individual whilst maintaining quality and choice for all patients. By concentrating our attention on specific therapy areas we feel that we can invest in developing innovation in homecare which can deliver enhanced services and better value for patients and commissions.

Nursing services are delivered through our nurse provider, Ashfield Healthcare. With 15 years’ experience developing, designing, building and delivering clinical support programs they have the capacity and capability to develop LSD services with Pharmaxo, the NHS, patients and their families.

We are delighted to have been successful on the LSD Framework and are looking forward to working with the specialist centres in providing consistent high quality homecare services to patients and their families.’

How Do You Measure The Impact Of A New Treatment For Type III?

The current significant challenge to the development of any treatment for Type III Gaucher disease is the lack of clinically validated endpoints or biomarkers that reflect central nervous system disease (CNS). In this edition of Gauchers News there are a number of articles that are all part of the ongoing challenge to address this issue; including Genzyme's Clinical Trial for Type III Gaucher Disease – Meeting an Unmet Need on page 16 and Using Technology to Assess the Impact of nGD on Daily Living on page 14.

The Gauchers Association is partnering with Genzyme and Quintiles Advisory Services, a worldwide consulting firm, on a research study to better understand the experience of patients who have Type III Gaucher disease. The overall purpose of the study is to learn which symptoms of the disease are the most common, and how these symptoms typically affect the lives of patients who live with the disease. This understanding may help improve how the patient's experience is accounted for in clinical trials that measure the effect of disease treatments. It also may help patients and their families by highlighting the consequences of the disease that most need attention, possibly leading to new programmes and support services.

The study will collect information based on a one hour interview and will include adult, children and teenage Type III patients as well as carers. The interviews can be face-to-face, on Skype or at a patient meeting if feasible. Please note that the interview will be conducted by a third party and not Genzyme and any travelling expenses will be reimbursed.

All patients will be given a patient information sheet that will outline the purpose of the study, how it will be conducted and how and where the information collected will be stored and used. In addition they will be required to give their consent to take part in the study and all information will remain confidential and anonymous.

It is envisaged that the study will be multinational and involve patients that represent the spectrum of Type III Gaucher disease. Through the Global Gaucher network, patients will be contacted through their patient organisation to participate in the study, however if you do not have a patient organisation in your country but would like to participate, please do not hesitate to contact Tanya Collin-Histed at tanya@gaucher.org.uk.

Using Technology to assess the impact of nGD on daily living

The UK Gauchers Association in partnership with Dr Elin haf Davies and Dr Aimee Donald, Paediatric Clinical Research Fellow and the Gaucherite Consortium are planning to conduct a pilot study for the use of wearable technology and mobile phone applications to assess the impact of nGD on daily living, to learn more about the condition, to encourage more research into these problems and to develop management and interventions to support patients.

The study would commence in early 2016 and look to recruit 12 nGD patients to be monitored using the device and phone app for 12 months. The recruitment would be via the UK Gaucher Association with the Principle Investigators, Drs Donald and Davies from aparito, a company set up to 'combine disease specific mobile apps and wearable technology to empower patients and to make sure their doctors get the information they need when they need it'.

Study Description

Initially, 12 patients over the age of 14 with a diagnosis of nGD will be recruited and provided with a wristband and smartphone app. This technology uses a wearable wristband which will primarily collect data on movement and ambulation, along with details on sleep and with the potential to collect data on fine movements. The phone app will have a user interface capable of collecting data on the following aspects of health:

- **Medication** – details of medications prescribed and associated compliance
- **Health Care Usage** – option to record episodes of contact with health care professionals
- **Disease-specific quality of life outcomes** – questionnaires on health status for completion by patients and families
- **Events** – recording of falls, seizures, time off school, swallowing/choking events etc.

Baseline data will be required for all patients to give context to the subsequent data gathered; this data would be collected by Dr Aimee Donald from a combination of patient case notes (once consent has been given) and review at time of recruitment and includes:

- Genotype i.e. L444P/L444P
- Disease severity score
- Most recent haemoglobin concentration
- Patient demographics
- Walking test to assess selected spatial and temporal (time) parameters of gait

At the 7th nGD family Conference on 14th November, Aimee & Elin presented this pilot to the families (see pages 20-21 for report). They also demonstrated some of the apps with the young patients to get feedback on usability before the final design adjustments are made prior to the pilot starting in early 2016.

The Gauchers Association Family Fundraising Walk Save the date!

**The Gauchers Association Family Fundraising Walk
through the stunning Cotswolds takes place
on Sunday May 1st, 2016.**

**More details will follow but to register your interest,
contact Claire by email at admin@gaucher.org.uk
or on 01453 549231.**

A Clinical Trial for Type III Gaucher Disease – meeting an unmet need

Approximately 5% of Gaucher patients are classified as having Type III Gaucher disease, also known as chronic neuronopathic Gaucher disease. Despite enzyme replacement therapy (ERT) for Gaucher disease being developed over 21 years ago and now with 3 ERTs and 2 oral substrate reduction therapy's (SRT) on the market for Type 1 GD there has been little advance in developing a targeted treatment for GD3.



In 2004 Actelion sponsored a clinical trial into GD3 with ERT and Miglustat (Zavesca), however sadly this trial did not meet its end points and it was not taken into a phase 3 trial.

At the Gauchers Association 7th nGD Family Conference Dr Derralynn Hughes and Dr Anupam Chakrapani talked to the families and young adults with GD3 about a clinical trial in GD3 that is in the pipeline.

The small roundtable session enabled the families and patients to ask questions about the forthcoming trial.

Further details will be made available on our Facebook page and website as soon as the final details of the trial are confirmed.



New Projects & Opportunities for 2016

As we move into our 25th year of supporting Gaucher patients and their families in the UK, there is time to reflect on the challenges we have faced and overcome but we move into 2016 with a real sense of optimism and hope.

Here at the Association, we are ready to tackle those challenges head-on to ensure we continue to meet the needs of the UKs Gaucher community and we have identified a number of projects that:

- Will improve on the available information about Gaucher disease
- Will determine what you need from us and how we can best structure the Association to meet those ongoing needs
- Brings patients and families together

In addition to developing the Association to run for the next 25 years, we are initiating some exciting projects that we hope will lead the way into 2016 and beyond.

Information Booklet: Together with patients, family members, clinicians, nurses and representatives from the pharmaceutical and homecare companies we work with, we are creating two new information booklets for Type I and Type III Gaucher disease.

Member Survey: We are also undertaking a vitally important survey to explore our UK members' and patients' current understanding of the purpose, scope and value of the work of the Gauchers Association. The reasons for the survey are wide-ranging and will cover all of what we do but ultimately the results will tell us how we can look after everyone involved and meet not just immediate needs, but in the long-term.

A New Patient Day: Following on from the resounding success of 2014's Family Day where we took four families and Association members to West Midlands Safari Park so they could meet and get to know each other, talk about their experiences and make friends. There was even enough time to see the animals!

We're organising another one in 2016, most likely in April or May and it will be opened up to all new patients and families that have been diagnosed in the last two years. Invitations will be sent out via the treating centres but from us here at the Association. We decided to do it this way to maintain confidentiality.

We'll be posting more information on the website and on our Facebook page so keep an eye out for details. If next year's day is anything like last years, everyone coming will have a fantastic time!

Gaucher Type III Empowerment Project

The Empowerment Project – a group of young people who meet three times a year – gives young people with Type III Gaucher disease a voice and empowers them to shape their own futures and the futures of all those living with Type III Gaucher disease.

The group is co-ordinated by Helen Whitehead (our Patient and Family Support Worker) and supported by the Gauchers Association Chief Executive Tanya Collin-Histed, Niamh Finnegan (Clinical Nurse Specialist) and Dr Derralynn Hughes (Adult Consultant). We have been successful in a bid for a grant from Great Ormond Street Hospital and the Institute of Child Health for £8,750 which allows this fantastic – and vital – project to continue over the next two years.

Overseas Visits

We have continued to support members of the group to attend overseas Gaucher clinics alongside professionals from the UK. Irma Shah and Nadia Fattouki had the opportunity to attend clinics in Jordan and Taiwan.

Jordan

Irma writes; ‘this year I travelled to Jordan to find out about patients living with Gaucher disease. During our stay we visited a Member of Parliament and spoke to him about how Gaucher disease is managed in the UK by having access to treatment, patient and family support groups and charities. We also visited a private hospital and had a tour of the different departments. We then spent a full day meeting patients at a centre run by Dr Antaki (head of Jordanian Patient Group) to support children with disabilities. I think the patients felt very thankful to be able to attend these clinics as in Jordan there are no specialist healthcare teams to see them.’



Meeting Jordan Parliament

Nadia writes; 'I travelled to Amman, Jordan in June 2015. We visited a clinic to meet patients who have a storage disorder like me and also attended meetings with the Jordanian Health Minister and MPs. I felt a deep connection with the people in Jordan because I speak Arabic and seeing patients young and old was an emotional experience. I spoke with mothers who had limited knowledge of the condition and they weren't always given access to continuous treatment. The clinics are very different to the NHS clinics in the UK, which made me more appreciative of what I have in my own life. It was an emotional experience and the more time I spent in Jordan the more passionate I became to want to give something back and create change.'

We were lucky to visit different towns and villages in Jordan such as Jareh and Madaba which was a beautiful experience. We interacted with the locals, went to the small markets and shops as well as visiting Mount Nebo, a historical site dating back to the Roman Empire and the Babylonian period.'

Taiwan

Irma writes; 'I travelled to Taiwan with Tanya Collin-Histed, Dr Vellodi and Nadia Fattouki to meet patients and observe clinics for patients who have Gaucher and Pompe disease. We had a tour of the tropical gardens and then we were given a talk by Dr Chan and some patients. I then gave a talk to the group that was translated into Chinese. We did arts and crafts with patients and all the team which everyone enjoyed. We also visited a Buddhist temple, night markets and much more.'



Nadia & Irma meeting patients in Taiwan



Tanya, Irma & Nadia

Nadia writes; 'the flight from London to Taiwan was nine hours and we had to stop and change in Hong Kong. Arriving in Taiwan, It was hot, humid and very clean. Trying to immerse myself into a new culture was exciting and I enjoyed trying different Taiwanese foods such as sushi. We visited the city centre hospital and the labs and I enjoyed seeing the technology and science. The patients and medical staff have a different relationship; they value medical staff

highly and there is not a lot of focus on welfare. Patients are told what they need to know by the staff in a short amount of time, whereas in the UK there are a lot of different medical staff and more support to those who need it.

I met with other Type III Gaucher patients at an informal gathering and it was a chance for patients and family members to meet and interact with each other through engaging in activities such as garden walks, painting and sharing a small lunch. Families and patients all stood up one by one in front of everyone to share their stories and learn about the condition by observing a PowerPoint presentation. Not knowing any Mandarin I was fascinated to learn about the culture by trying out things I wouldn't have done before by speaking with locals, trying different foods and traveling around the city by public transport.'

7th nGD Family Conference

84 patients, families, healthcare professionals and representatives from the pharmaceutical industry attended the 7th nGD family conference in Manchester from Friday 13th to Sunday 15th November.

During the weekend 12 children were entertained by our fantastic volunteers with arts and crafts, and a day of fun in the snow at the Chill Factore (the UK's largest indoor snow centre).

The meeting programme once again was designed in partnership with the type III girls, Irma, Maddie, Nadia, Saphia, Radhika and Sara guided by Patient and Family Support Worker Helen Whitehead with a focus on current research projects, education, lessons learnt in nGD and wearable technology. At the end of the day the young adult patients had the opportunity to sit down and test some of the wearable technology alongside Elin haf Davies, James Skinner and Aimee Donald, their feedback will be used to make any necessary changes to the final version of the software that will be piloted in the New Year by the young adults to collect live data on their daily functioning.

Other topics presented included; genetics, palliative care, psychology and fantastic personal stories from patients and parents.

Saturday evening gave the families and professionals an opportunity to catch up with old friends and meet new ones with a meal together.

On the Sunday whilst the children enjoyed mosaics, decorating a pencil case or bag along with games and arts and crafts, the families had a closed

session looking at topics such as the Association's and UCL's Gene Therapy project and a forthcoming clinical trial for Type III Gaucher disease.

Families travelled from all over the UK as well as the USA, Sweden and Denmark to attend the meeting.

Feedback from those that attended was extremely positive and the clear message from the weekend was 'Together we are a stronger community'



Feedback

"The conference was both interesting and special. The girls did such an excellent job. I was extremely moved by several of the speakers and struck by their bravery and spirit."

Anna Rabbeth, volunteer in the Further Education group work

"I really enjoyed the time I spent at the conference and I learnt a lot. The patient stories were especially powerful and inspiring."

Sian Burgess, speaker on the Adult Hospice and Palliative Care

"It was such a great conference and we were very honoured to be asked to be involved with you all. Brilliantly organised and run. I thought the content of the programme was so good and

so inspiring to have the empowerment project with the young people so involved, especially for families with younger children to be able to see how well equipped these young people were at running their lives so well."

Sarah Cable, Rapsodi project

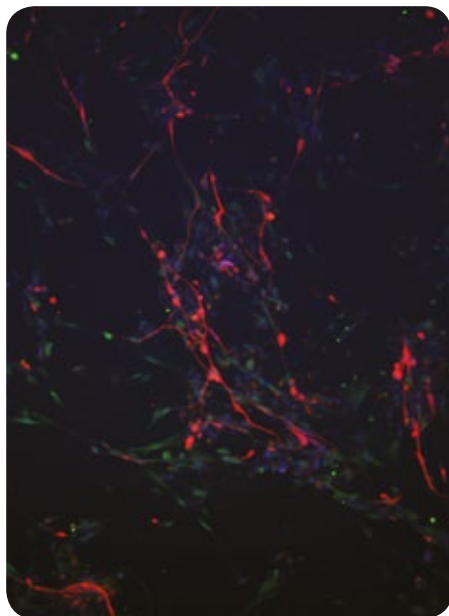
"Thank you so much for the opportunity to be invited to this fantastic meeting. Congratulations for the quality of the presentations and weren't the girls a knockout! Wow how they have blossomed and developed over the past couple of years, you all must be so proud. The real life presentations were so inspirational. It was also wonderful to meet old friends and make new ones."

Malcolm, Genzyme



Investigating the connections between Gaucher & Parkinson's

Following on from her update in the last edition of Gaucher News, Lucy Collins, a PhD candidate working alongside Professor Timothy Cox, Dr. Patrick Deegan and Professor Roger Barker updates us on her study on the associations between Gaucher and Parkinson's. She writes:



Firstly we would like to sincerely thank everyone for participating in our research study. We would like to thank you for your patience with our assessments and support for our research. We will be finishing up our visits for this study this year.

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 truly 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 is an example of neurons (in red) made from a skin sample. We are now working on trying to make a higher number of these neurons and

a pure population. We now know that growing these cells at lower oxygen conditions to mimic the brain's environment may help. This field of research is fairly new and we have been making many attempts along with experts in these techniques to produce these nerve cells from skin.

Gaucher families helping find a cure for Parkinson's



News of pioneering new internet based research study starting January 2016.

People with Gaucher and their families have been playing a crucial and leading role in new research to find a cure for Parkinson's. Patients at the Royal Free Hospital Lysosomal Storage Disorders Unit in London and at Addenbrooke's, Cambridge along with the Gauchers Association have been helping to develop an easy to use online study called RAPSODI to help with this research.

Why can Gaucher help find answers for Parkinson's?

The genetic alteration around the GBA gene that causes Gaucher disease is carried within some Gaucher families has been providing new and interesting clues into the development of Parkinson's in some people. 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. Nevertheless, this new discovery linking the GBA gene to Parkinson's is probably one of the most important discoveries in this area in recent years and opens up new and promising avenues for research into how we may target this Gaucher-Parkinson's (GD-PD) link and develop powerful new treatments for these diseases.

The research team wish to assess people with the GBA gene over several years. This is why the study has been developed to be mostly internet based, so it doesn't take up too much time and won't involve hospital visits. Instead you will be able to take part from home by visiting the study website on a yearly basis. It will take less than an hour to complete some online questionnaires, memory tests and simple keyboard dexterity exercises. The study plans to collect data already known to predict certain aspects of Parkinson's many years before any symptoms appear. Everybody will be treated anonymously because the overall aim is not to predict an individual's risk of developing Parkinson's but to look for patterns in the bigger community. But being able to predict Parkinson's earlier in the future will offer opportunities to help everyone more at risk of developing this condition.

The study is being led by Professors Anthony Schapira, Atul Mehta and Dr Derrylynn Hughes at the Royal Free Hospital in London and will be ready to join in January 2016, but if you are interested to learn more the team would love to hear from you. Please contact: Dr. Stephen Mullin or Sarah Cable (Clinical Research Nurse) rapsodi@ucl.ac.uk ph: 07753 982 063

Dr Aimee Donald in Sweden

The Susan Lewis Memorial Award has successfully provided bursaries to educate doctors and healthcare professionals in Gaucher disease since its creation in 2008.



*Aimee Donald, Bosse Johansson,
Prof. Timothy Cox & Dr Nicholas Smith*

Since the last edition of Gauchers News, the Association has supported Dr Aimee Donald, Paediatric Clinical Research Fellow at the Manchester Centre for Genomic Medicine to visit Sweden coinciding with her work alongside the Gaucherite research project.

Aimee writes: 'I have been working with the Gaucherite research team for six months and I was delighted to be invited by Professor Timothy Cox to join him on his invitation by the Morbus Gaucher Association to Sweden. The visit was an opportunity to learn more about the Norrbottnian form of Gaucher disease and to share ideas about service development in both of our countries. The Susan Lewis Memorial Award enabled this interesting, educational and enjoyable journey and I would like to thank the UK Gauchers Association for supporting me in this. I travelled with Professor Cox and Dr Nick Smith, a Paediatric Neurologist from Adelaide who is an expert in neuronopathic Gaucher disease. We arrived in Stockholm and went directly to the north of the country where we were met by our hosts, Bosse and Bo. We stayed at the Sunderby Hospital and had the opportunity to meet several patients and relatives. The following day we enjoyed a shared educational conference to celebrate 40 years of the Morbus Gaucher Association.

We proceeded north, eventually crossing into the Arctic Circle, stopping to meet patients and relatives along the way. The scenery was stunning; we spotted reindeer and learnt about the history of the country as we travelled. Bo and Bosse brought a family tree with them which explained the inheritance of Gaucher disease throughout the northern part of Sweden. It was fascinating to hear from families about their experience of the disease and how this has changed in the era of ERT.

We gradually made our way back to Stockholm, where scientists and clinicians from the Karolinska Institute joined us for further educational discussions. Professor Cox and I then spent time at Lund University with scientists and patients. This was one of the first opportunities I've had to meet so many patients with the neurological form of Gaucher disease and it has been invaluable in helping to shape my ideas about future research. The hospitality in Sweden was overwhelming and I felt like the trip inspired true excitement about the importance of improving our understanding of Gaucher disease.'

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 and also available as a PDF or hardcopy leaflet. Email Sarah in the office at sarah@gaucher.org.uk or call 01453 549231 and she will email or post you a copy.

Spotlight on Gaucher Film Contest – winners announced

The Spotlight on Gaucher Film Contest which was funded and organised by Shire Pharmaceuticals was launched in July, and encouraged those touched by Gaucher disease to share a short video about their experience with the condition.

The contest, which was supported by the European Gaucher Alliance (EGA), was open to all individuals impacted by or interested in Gaucher disease in any country to raise awareness of the condition and communicate a message of hope through shared experience.

The winners of the contest were announced on the occasion of International Gaucher Day (IGD), on 1 October. The winner, 'My Daughter' and runner up, 'Morena', both from Argentina, received an official contest certificate and were recognised as part of the IGD activities by the EGA. The winning videos were chosen by an external judging panel including representatives from the EGA, The Ma'aleh Film School, and the Gaucher disease medical and/patient community. Judging criteria focused on video creativity, storytelling and awareness raising. The Spotlight on Gaucher website received visits from 66 different countries and following the entry submission closure in September, a total of 13 entries were received from countries including Argentina, Columbia, Israel, Japan, Jordan, Russia, Rwanda and the United States. Spotlight on Gaucher helped to raise significant awareness of Gaucher disease with over 4,700 unique visits to the contest website and around 4,550 video views on the contest YouTube channel.

The contest was also supported by Shire's International Gaucher Disease Ambassadors, a group of motivated and inspirational individuals impacted by Gaucher disease, who share their personal stories to support further understanding of what it means to live with the disease. The Ambassadors created 'how-to' videos to support contest entries which were viewed over 960 times. For more information on the Spotlight on Gaucher film contest and to watch all the video entries, please visit: www.spotlightongaucher.com or www.youtube.com/spotlightongaucher. Thank you to all that supported the contest - stay tuned for further activities in 2016!

Rare Disease Day 2016



Rare Disease Day

29 February 2016 marks the ninth year that the international rare disease community celebrates Rare Disease Day. The Rare Disease Day 2016 slogan 'Join us in making the voice of rare disease heard' appeals to join the rare disease community in making known the impact of rare diseases. The Association will be celebrating this day through our website and Facebook page so would love to share your photos and stories. There will be lots of materials to download through Social Media to raise awareness and get involved. Keep a watch out for these!

International Gaucher Day

Thursday October 1st 2015 was the second International Gaucher Day (IGD).



IGD is the brainchild of the European Gaucher Alliance (EGA). It was celebrated in their 41 member countries through a series of very special events and is based on an idea from the participants at Go with Gaucher 2012, a seminar for young adult Gaucher patients held in Frankfurt, Germany. The aim of the day is to raise international awareness of Gaucher disease and the slogan for the day is 'Rare but Not Alone.'

Here in the UK we asked our members to celebrate IGD once again by organising their own events either at home or work with friends, family and colleagues. To help our members celebrate this very special day we also sent them a poster to display and a pen and pad branded with the new IGD logo, all to help raise awareness.

Here are a few stories of how people celebrated throughout the UK:



Judith Richards

Judith Richards, a Type I sufferer writes: 'I visited St Michaels Mount which is two miles away from my home in Penzance.

This photograph was taken on IGD, six days before my 60th birthday. I was diagnosed at age nine after many years of undiagnosed symptoms. I started having infusions 22 and a half years ago aged 37 at Addenbrooke's Hospital in Cambridge under the supervision of Professor Timothy Cox.

My regular outpatient appointments involve a round trip of 900 miles, but I feel thankful to the NHS and privileged to be able to make the trip and see such a wonderful and caring team at Addenbrooke's.' Judith kindly donated £20 to the Association.



Jeremy Manuel friends & family



Manuel Swaden Solicitors in London celebrated by sharing cakes in the office. Jeremy Manuel also hosted a dinner party for friends and family.

At the Gauchers office, Chief Executive **Tanya Collin-Histed** was busy making chocolate brownies for us all to enjoy. A rarity, just like Gaucher disease!

Rona Troman hosted a coffee and chat morning at her local church, Williton Methodist and kindly raised £30.

From all of us at the Association, we would like to send our most gracious thanks to all our members for their efforts in raising awareness and their very kind donations. Watch this space for IGD 2016!



Tanya Collin-Histed

Mark Histed – Iron Man

Mark Histed, husband of Association Chief Executive Tanya is at it again, this time as Iron Man! Here is his fantastic story:

A number of years ago I decided to do a fundraising challenge for a charity very close to my heart which involved me hiking up to the peak of Mount Kilimanjaro at 5,895 metres. Little did I know at that point that this would be the start of a trilogy of mad challenging events! When I completed the London Marathon two years ago – the second in the trilogy – and as I lay collapsed in a sweaty heap in Horse Guards Parade, a person (who shall remain unnamed) said ‘why don’t you do an Ironman’? My answer was short, contained no swear words and only had two letters in it. ‘No’.



So here I am two years later, one and a half stone lighter, older and supposedly wiser and I am the owner of a triathlon bike, far too much Lycra and a wet suit. During the very early hours of July 12th when you were all tucked up in bed, I was stood on the side of a large cold lake with a hundred and fifty other insane rubber-clad people, about to start a race that I knew would test my body and mental perseverance to its very limit.

For those of you who do not know what an Ironman is, it’s a 2.4 mile open water swim followed straight away by 112 miles on a not very comfortable racing bike followed by a full 26.2 mile marathon.



The swim is considered by most who attempt triathlons to be the most daunting element. When the starter gun fires all the friendliness disappears. There are hands, feet and elbows flailing around and you just hope you don’t get hit in the face. Thankfully my start was OK and I stayed out of the way. I settled into a nice rhythm and managed to take my mind off things by watching the fish on the bottom of the lake going about their normal daily business, oblivious to the commotion going on above them.

I exited the water after approximately 1hr 20 mins and started the jog to my bike. As soon as you stand upright all the blood drains down into your lower body and your next immediate challenge is to run in a straight line and not faint! Then comes the fun of getting out of a very tight wet suit as quickly as you can. Before I knew it I was clad in Lycra and was off on the bike having waved my family goodbye. Just to add to the fun, the wind had picked up and the first 28 miles were into a head wind, then because the wind wasn't challenging enough it was joined by torrential rain. I can assure you I was very glad to get off the saddle because over six hours and 112 miles does take its toll on one's posterior!



Tanya, Skye, Mark & Maddie

Then came the run. My legs were reluctant to say the least, and I looked as if I was suffering from constipation as I tried to jog out of transition. The sun had now come out in full force which may have been good for all the supporters but it wasn't so good for all us triathletes. The first half of the marathon wasn't too bad, but my body was now starting to suffer. I had voices in my head saying 'please stop', I had blisters on my blisters and my legs were cramping, but with the help of friends and family, some of whom actually ran and walked with me, I managed to keep going.

Then 13 hours and 5 minutes after I'd jumped into that cold lake I finally crossed the finish line. I have to say that it was the most amazing feeling when I finally knew I'd completed an Ironman distance triathlon. My body ached in places that I didn't know existed and did for a couple of days. Some have asked what's next. NOTHING is the stern response.

So why did I put myself through so much pain? The Gauchers Association has supported my step-daughter since her diagnosis with Gaucher disease nearly two decades ago. She is an inspiration in my life and is now a wonderful young lady who in spite of everything the disease has thrown at her, has refused to allow it to define who she is. She has surpassed all our expectations and I'm very proud to say that in September she started university, something my wife and I never dreamt would happen.

The charity continues to provide support and assistance to her and many other patients with the disease, as well as their families. It also helps the scientists, doctors and nurses that provide the essential support and clinical care needed.

The money I raised from this last mad challenge will help them continue to provide this essential support for many years to come. I'd like to express my gratitude to everyone who supported me on the day and also to those very kind people who sponsored me. Without your generous and kind support the charity would not be able to help the patients, their families and the scientists, doctors and nurses.'

Virgin London Marathon 2016

As always, we were pleased to announce we had five Golden Bond places for the 2016 Virgin London Marathon on Sunday 24th April 2016 but at the time of writing, four have been snapped up by eager runners!

There's one left so if you want it, it's yours! If you think a marathon is a bit too much like hard work, maybe you know someone who was unsuccessful in the ballot and who would like to raise money for the Gauchers Association? Let us know! Contact Sarah in the office on sarah@gaucher.org.uk or call 01453 549231.

The marathon is one of our biggest and most important fundraising events of the year. Five runners completed the course in 2015 and they raised over £12,000 through the generous support of their friends and family as well as our members and we're hoping to do even better in 2016. Everyone in the Gaucher community wishes our runners Matthew Gillan, Calum Phoenix, Giulia Massaro & Julia Teper 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.



Matthew Gillan

Matthew Gillan writes: 'I started running three years ago and have completed six half marathons, each time choosing a different charity to represent. Abdul Waheed is a friend from work and he was telling me about the great work you do and I will be very proud to wear your vest in London as well as raising as much money as possible. I also plan to spread awareness of the work you do. So far my training has begun slowly but I'm out most nights trying to add miles to the clock. The London Marathon will be a tough test but I'm very excited and hope to raise a significant amount of money to help raise awareness of the disease.'

Giulia Massaro writes: 'I moved to London two years ago to undertake a PhD at UCL in Dr. Rahim's group, where I am currently working on gene therapy for the treatment of nGD. When I first met the members of the Gauchers Association they said: 'This is our family.' Today, after having spent some time with them, I understand why. The Gauchers Association not only fund my research project but also give me a great opportunity to be part of and contribute to its community.'

In July I ran the British 10K London Run, raising money for the Association. Now I want to push my limits and participate in the London Marathon 2016. I am sure it will be good fun for a good cause!'



Giulia Massaro



Calum Phoenix

Calum Phoenix writes: 'Hi my name is Calum Phoenix, I'm 24 from Newcastle and will be running the London Marathon in memory of my brother Liam Phoenix who sadly passed from complications as a result of Gaucher aged 3.'

Julia Teper says: 'My name is Julia Teper and I'm a mum and personal trainer from Aldenham in Hertfordshire. I am delighted to be running the London Marathon in April on behalf of the Gauchers Association and am hoping to raise both money and awareness for this disease. Having a close

friend who is affected by Gaucher I feel very passionate about running the marathon for the Association. This will be my third London Marathon and hope to make it my best!'



Julia Teper

Charity Golf Day Raises £5,500

Although we haven't had one for three years, the Gaucher Association Charity Golf Day at the stunning Dyrham Park Country Club in Hertfordshire on May 20th was a resounding success!



We hosted 60 golfers of varying standard, from single-figure handicappers to decent club players and we played an Alliance Stableford, that is teams of four where each player plays their own ball throughout but scores are combined, and there were loads of great prizes, including one for the overall winners, the longest drive and 'nearest the pin' on the par three holes. After the golf, everyone made their way back to the clubhouse for a well-deserved buffet lunch.

We must say thank you to Hummingbird Motors who provided a Mitsubishi for us to give away to anyone who made a hole in one on the par three fourteenth hole and although one or two came incredibly close, we didn't find a home for the new car! We're hoping to next year so get to the driving range and start practicing! We were also delighted that Jason Isaacs (famous for his portrayal of Lucius Malfoy in the Harry Potter series) made up one of the fourballs with his brothers and father. It certainly created a buzz around the place! Best news of all though was that we raised £5,500 for the Association so a huge thank you to everyone involved in the day!

Great North Run raises over £1,400!

2015 was the first year that the Association secured five Golden Bond places for the iconic Great North Run.

The 13.1 mile course from the Tyne Bridge out to South Shields has been run since 1981 with over a million people crossing the finish line and is considered to be the world's greatest half-marathon.

From all of us at the Association, we'd like to say a huge 'thank you' to our five intrepid runners who pounded the streets of the North East on Sunday 13th September and below you can read their stories of the day.

Lisa Robson ran with **Susan Quintano**: 'The GNR was another challenging run for an amazing charity so close to my heart and I was raising money on behalf of my little sister Emma Hall. The amount of support I've received has been unbelievable and means so much. Raising money for Gaucher makes me feel so proud and I feel like I'm part of a team supporting each other. Memories last forever and it's good to remember.'



Dylan Gould

Dylan Gould writes: 'It was a pleasure to run The Great North Run 2015 for The Gauchers Association and raise £403 at the same time. The atmosphere was amazing amongst the runners and from the crowds that

seemed to line every metre of the course. The highlight for me was the seaside finish and the display from the Red Arrows although this was closely matched by getting a hug from my wife and daughter on the way round.

I was very pleased with my time of 1 hour 32 minutes after suffering a calf strain after only three miles. I was determined to finish and it just shows that anything is possible if you have belief and the desire to succeed. For anyone thinking of taking up running just go for it and get involved with fabulous events like this.'



Lisa Robson

Wendy & Lee Burbidge write: 'Lee and I completed the Great North Run for the Gauchers Association. It was our first time ever running a half-marathon and the experience was amazing with the fantastic sense of community amongst the 57,000 runners with so many charities represented. It gave us the opportunity to speak to people about the charity we were running for, both before the event raising our sponsorship and also whilst we were waiting to start. We finished in a time that we were both proud of, although we are definitely not a threat to the Mighty Mo!

Thank you for allowing us to represent you, it was our privilege and hopefully we will have the opportunity again in the future.'



Wendy & Lee Burbidge

Aura Cruz raises over £1,000

From all of us at the Association we'd like to say a big thank you to Aura Cruz for raising over £1,000 towards our continued work in gene therapy by participating in The Colour Run. Aura picks up the story:

'To raise money for gene therapy, I did my first fundraising event, a 5k colour run on Saturday August 15th 2015 with my family and friends and raised £457. After the colour run we organised a charity quiz night at The Famous Virgins and Castle pub in Kenilworth in Warwickshire which is owned by one of our family members, Laurie Howe. This helped us to raise another £370. On the night a local artist called Karen Pittaway auctioned one of her paintings for £250 and very kindly donated the money towards our fundraising efforts.

In addition, the fabulous Alexa Lauren Patisserie helped out by baking some rainbow cookies and raised £22. In total we raised £1099. It was a fantastic experience doing this with family and friends and it was for a great cause. I'd like to say thanks to everyone that took part in all the fundraising and donated money to the charity.'



London 10k runners raise over £1,400

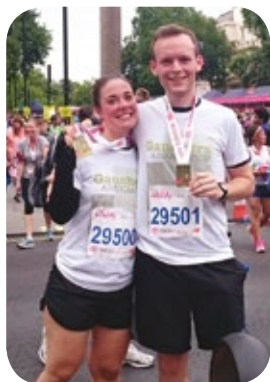
On Sunday July 12th, our six runners lined up to support the Association for the very first time on Piccadilly to take part in the British London 10k run.

Ilana, Sarah, Ben, Giulia, Michael & Matt all completed the course raising over £1,400 to support the Association's activities and initiatives, and their stories are here:

Sarah Manuel writes: 'On July 10th my cousin Ben Graham, my sister Ilana and I ran the London Vitality 10K pleased to be wearing our Gauchers Association running tops. The weather was warm and the sun was shining. There were approximately 25,000 runners and the route was lined with supporters. The clapping and cheering enhanced the atmosphere as we passed the London



Sarah, Ben & Ilana



Giulia & Michael

landmarks of Trafalgar Square, St James' Palace, and the Houses of Parliament on route to the finish in Whitehall. We were proud to be running for the Gauchers Association, a charity very close to our family and our hearts. We are delighted to have raised over £800 and want to thank everyone who kindly sponsored us.'

Giulia Massaro ran with **Michael Hughes** and as one, they said 'it was really nice, full of people and we had lots of fun on the day.'

Look out 2016 as 'Team Gaucher' from the Dursley office will be hitting the streets of London so keep an eye on our Facebook page for training updates!

The Association have secured six places for 2016 & 2017 for this growing event which takes runners around London's iconic landmarks for a one-off view of the city so if you're interested, please contact Sarah on the office.

Calum Phoenix – The Running Man

From all of us at the Association, we'd like to say a huge thank you to Calum Phoenix who raised over £700 by competing in three gruelling endurance events – the Great North Run, Tough Mudder and Torture Trails, an equally tough test of strength, stamina and mental determination! He writes:



'My brother Liam Thomas Phoenix sadly died from Gaucher disease before I was born and my fundraising started with Tough Mudder back in August as well as the Torture Trails 10K in September. The Great North Run was fantastic and I enjoyed every minute. I set myself a goal of getting under two hours and I managed it in 1h 59m!

As of Saturday 19th September I completed my third and final run to raise money for the Gauchers Association. The exhaustion completing these races is nothing compared the pain and trauma suffered by those who have lost someone to Gaucher disease as well as the effect it has on friends and families. To be honest, the three runs were an absolute joy and to top it off I raised £708 for a great cause.'

In memory of Tom Downes

Jacqui Downes kindly raised £50 through her Facebook status in memory of her late husband Tom who would have celebrated his 60th birthday this year, Jacqui writes;

Dear Tom,

You should be celebrating your 60th birthday today. I wonder what we would be doing if you were still here? A trip to Las Vegas or St Petersburg maybe?

Whatever it was I know we would fit in a meal with our lovely family; our daughters Angela Harding and Tracey Downes, our son in law Daniel Harding, Tracey's boyfriend John and our grandchildren Theo and Georgia.

Well, even though you are not here, we will all be going out this evening to your favourite Indian Restaurant in Warwick, to celebrate on your behalf, but most of all to celebrate having had you in our lives. Thank you for everything you did for us. You will stay in our hearts forever.

Happy 60th Birthday Tom! xxxxxx



The £50 donation represented the number of "likes" I received for the post.

The Prudential Ride London – Surrey100

On Sunday August 2nd 2015, our very own Sarah Allard and her husband Neil both completed the Prudential Ride London for the Gauchers Association along with 25,000 other keen cyclists. Here they share their stories of the day:



Neil & Sarah

Sarah: 'After months of training on the Cotswold Hills in the wind and rain, the weekend was finally here. At 5.30am we were dropped off and cycled the five miles to the start. The weather was a complete contrast to 2014, it really couldn't have been better. To be able to cycle in and out of London on closed roads and ignore traffic lights took some getting used to but was fantastic! The marshals and volunteers worked really hard to make the day run smoothly and the water and feed stations were plentiful. There was a huge spectrum of riders and bikes throughout the day; one chap on a Boris bike was asked just before tackling Leith Hill 'so, how many gears have you got?' to which he replied 'three, but only two are in working order!' Box Hill was absolutely stunning and the views from the top on such a clear day were well worth the push. The last 10 miles were a real effort but once I reached The Mall I knew I would finish with a smile on my face. We raised a grand total of £970 for the Association and I would like to thank all my friends and family who sponsored us for the ride. Thank you!'

Neil: 'Pulling up to the start line in the midst of the 500 or so other riders that comprised the 7:22am wave, the lethargy of a 4am alarm fell away to be replaced by the imminent challenge of the next 100 miles. With a blast of cheesy rock music and a countdown from 10, we were off. The first 15 miles disappeared under my wheels as red lights and box junctions and the right side of the road was joyfully ignored. Soon the outskirts of London sped by, through Richmond Park, past Hampton Court and over the Thames into the lanes of Surrey. The combined efforts of Newlands Corner, Leith Hill and Box Hill were forgotten in the high speed chase back into Dorking and Leatherhead and before I knew it the last kicker at Wimbledon was upon me and an out of the saddle effort, with the crowds cheering saw me over the brow and into the final 10 miles. Speeding between Whitehall and the Thames, Admiralty Arch came into view and in the blink of an eye I was on The Mall. After 100 miles I managed an attempt at a sprint, hitting 27mph over the line. It was a fantastic day and I've never seen so many bikes of all descriptions in one place. The training paid off, Sarah and I survived and I even have a medal and photo to prove it!'

The Gauchers Association have been successful in securing four places for the 2016 Ride London event on Sunday July 31st 2016. If you would like to support the Association by taking up this fantastic challenge, contact Sarah on sarah@gaucher.org.uk or call 01453 549231 – go on, give it a try!

The London To Cambridge Bike Ride

The Mayor of Barnet adorned with his official seal of office counted down the time and set the riders off for the 6th Gauchers Association London to Cambridge Bike Ride on Sunday, September 6th 2015 from the University College School playing fields in northwest London, close to the Royal Free Hospital.



It was gloriously sunny day but not too hot. It was perfect cycling weather and it stayed that way for the entire day.

The 60 mile ride followed the same route as it had in previous years with the welcoming finish from Liz Morris and her team at Addenbrooke's Hospital in Cambridge and Don Tendell awarding the medals at the finishing line. Over seventy cyclists took part and enjoyed fantastic opportunities

to 'refuel' at the four water and food stops sampling the breakfast croissants and buns kindly donated by Sherrards Patisserie at stop one and the amazing array of cakes 10 miles from the finish baked by June Morris and her team at stop four! Participants who have ridden before know to save room for such a special treat!

Thanks of course go to all who help the day run so smoothly, particularly the teams at the start and finish, the marshals en route and the volunteers at the refreshment stops.

Sarah must take credit for all the organisation of the ride (with the support of CSS) and the feedback from all those involved has been excellent, especially from those who took part for the first time. The event has raised almost £20,000 for the Association which was a tremendous effort. We are now in the process of planning our 7th Bike Ride and will let you know in due course when that will be. It's never too soon to start training!

2015 Cricket Day

The Gauchers Association Cricket Day raised over £7,500 and we'd like to say thank you to Abdul Waheed and his friends for organising such a fantastic day. This was the second Cricket Day after a short break of 12 years!

Abdul writes: 'Two rival (and very ageing) cricket teams, Rainbow CC and Pakistan CC once again put on their whites to contest the 2015 Gaucher Cup helping to raise £7,512 for much needed research work. With the average age of now 60+ (and a couple of players over 70) the match was keenly contested, with Rainbow coming out on top scoring 212 to Pakistan's 160 all out.

On a very sunny and hot September day, several hundred people with their families attended the match. There were activities for children such as face painting, a bouncy castle, Henna, cake stalls and plenty of food for everyone. The match ended with Dr Ahad Rahim from University College London giving a brief talk on Gaucher disease and the importance of both fundraising and of generating awareness in the community.

A special thank you to the management of Carrington Cricket Club for allowing us to use their facilities and also a very big thank you to all

the sponsors and individual contributions made on the day. We'd like to say a special thank you to the Sheriff of Nottingham for attending and giving great support on the day.'

Match Summary

Rainbow Cricket Club 212 runs

Pakistan Cricket Club 160 runs

Best Bowler: Manawar Sultan – 4 for 20

Best Batsman: Abdul Waheed – 68 runs

Best Fielder: Dr Ahad Rahim – an outstanding performance in the field



Abdul Waheed best batsman



Ahad Rahim best fielder



Manawar Sultan best bowler

Member's fundraising

The Association relies on its members, their families and their friends who generously support our 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 and if you'd like to set up a regular standing order, please contact Sarah by email at sarah@gaucher.org.uk or call **01453 549231**. We also extend our thanks to all those who have made anonymous donations online.

Donations received from May–October 2015 totalling £5,918

Generous donations have been received from **Manuel Swaden, Debbie Isaacs through the William Brake Charitable Trust, Ditcham Park School, Friars Lodge Benevolent Fund and Akshantha Shetty.**

Hazel Garbett raised £40 by opening her colourful garden to her friends and the public.



Sheena Hulme recently took part in the 10miles Great South Run and raised £730 for The Gauchers Association and the nGD fund.

Janet Thompson raised £57 by holding a cake sale in the staffroom at the school where she works. Thank you to Janet's family and neighbours who finished off the cakes and topped up the funds!

Jeff Hammerschlagg kindly donated £1,000 through the New West End Synagogue and their Kol Nidre Appeal.

Sylvia Beament of the Witham and District Lionesses



kindly raised £1,000 towards our neuronopathic Gaucher Disease research project by carrying out various fundraising events throughout the year including stalls, tombolas, quizzes and breakfasts; in memory of her granddaughter Lauren Goldsworthy who passed away from type II Gaucher disease aged 2 years.

Thank you so much!

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 online at **www.gaucher.org.uk** or through our Facebook page at **www.facebook.com/TheGauchersAssociation**.

Donate by text

Text **GAUK01 £2 / GAUK01£5 / GAUK £10** to **70070** to donate now!

giftaid it

Gift Aid it!

The Government operates a scheme called Gift Aid that allows charities to claim the basic rate tax on every pound donated, meaning if you are a UK tax payer and donate £100 to the Gauchers Association, the Government tops it up by £25 and we'll actually receive £125. Giving under Gift Aid means that so much more money can be raised at no extra cost to our donors.

How about planning a fundraising event in 2016?

For those of us who aren't quite so sporty there are still plenty of ways to raise funds for the Association as well as to increase awareness of Gaucher disease.

Really, anything goes! How about a bake sale, a coffee morning or a pamper evening? What about an old school cheese and wine, a silent auction or an open garden? The fundraising possibilities are literally endless! If you would like to plan an event we can send you a fundraising pack which will include balloons, posters, thunder sticks, ponchos and even a t-shirt for you to wear. We can also assist you in setting up online fundraising pages. Contact Claire by email at admin@gaucher.org.uk or on 01453 549231 and she will send you your pack and help with ideas! During your event, don't forget to take lots of pictures and send them to us for inclusion in the next edition of Gaucher News!

Sign Up To Giving A Bit

Another simple way to help raise money for the Association is to sign up to Giving A Bit. This is an online shopping site where your online shopping helps to raise funds for us and the best part is that it won't cost you a penny. Thousands of retailers are signed up to the scheme from Argos to Sainsbury's, Amazon to Zavvi and a typical shopping basket will generate a donation to the Association of 3%. Take a look at the website and sign up today. www.givingabit.com. Every little helps...

