Gauchers NEWS

July 2016

Raising Awareness of Gaucher Walk

Running the marathon for Gaucher

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ASSOCIATION NEWS

25TH ANNIVERSARY CELEBRATIONS

SAVE THE DATE - 5TH NOVEMBER 2016

This year is the 25th Anniversary of the Gauchers Association and to celebrate this momentous occasion we are planning a special celebration for all of our members, colleagues and friends.

On the evening of 5th November 2016 we will be hosting a party at the Royal College of Physicians in London and hope that as many people as possible who have been associated with us over the past 25 years will be able to attend.

TICKET PRICES ARE: £25 PER ADULT £10 UNDER 16'S
Numbers are limited, so please secure your place early and complete and return the enclosed booking form

We will be producing a souvenir celebratory brochure on the night in which there will be advertising, sponsorship and messages. We would however like to offer our friends and members the opportunity to include their personal greetings at a very special discounted price. Contact Sarah Allard for more details.

PLEASE COMPLETE AND RETURN THE ENCLOSED BOOKING FORM.

Full details for this event are available on our website - www.gaucher.org.uk - or please contact Sarah Allard on sarah@gaucher.org.uk or 01453 549231

CHAIRMAN'S CHAT

WELCOME

Dear Friends

Welcome to the new look June 2016 edition of Gauchers News. We've decided to freshen up the format which hopefully you will all enjoy. A big thank you to Sarah in the office for all her efforts in overseeing the changes and pulling the newsletter together as always.

I am writing this Chairman's Chat on the eve of the Brexit vote which also happens to be the week before the next two yearly meeting of the European Gaucher Alliance. It is ironic that whilst the UK is ensued in a debate over whether to remain part of the European Union, Tanya, Jeremy and I will join patient group representatives from 34 other countries to share experiences and collaborate on how best to learn from each other and help each other in our endeavours. The EGA continues to go from strength to strength with new member countries joining every year and is a fantastic example of the Gaucher community coming together. However the Brexit vote turns out (which by the time you are reading this we will all know!), the UK Association will, I'm sure, be a proud member of the EGA for many years to come. Look out for some reports from the EGA meeting in the next edition of the Gauchers News.

Moving on to matters at home I am delighted to say that Maddie Collin has been formally appointed as a member of the board of the Association. Maddie will be able to bring a different perspective to the board and in particular will be able to provide a voice for our younger members. A warm welcome to Maddie – we are delighted to have you on board (excuse the pun!).

As Maddie takes up her post it is with regret that Sue Noe has decided to resign from her post on the board after many, many years of unstinting service and dedication to the Gaucher community. Sue was one of only a handful of people who attended the very first meeting of the UK Association back in 1991 and has been involved ever since providing an immeasurable contribution to the activities of the Association and in supporting the wider Gaucher community. On behalf of the board and all of our members I would like to extend a huge, huge thank you to Sue for everything she has done over the past 25 years.

Finally, as mentioned above it is 25 years since the inception of the Association and we are hosting a special event on 5th November to celebrate. It promises to be a wonderful evening and we would love as many of you as possible to join us. Details of how to book your tickets are opposite.

Wishing you all an enjoyable summer and look forward to seeing you on November 5th!

Dan

PERSONAL STORY

FIRST STEPS TO STARDOM

I'm Kiren and I'm 15 years old with Gaucher type III and all I have ever wanted to be is a star. Recently I took the first steps in achieving my ultimate goal. Through my school, my music teacher Mr Hatton arranged a few visits to a local recording studio. I write my own songs and have a variety of different moments written down ready for the world to hear. My teacher plays a melody and I will sing and together will come up with the different song. Sometimes I even go to him with my own melody and ideas about how I want my songs to go.

Last week, I travelled from Kent up to Derby with my music teacher and my keyworker. When I arrived I met Mr John Stamp, the director of Esland Care and a producer friend of his called James. I was super excited the whole journey up and closer we all got, the more excited I got. I wasn't nervous at all but I did have butterflies and felt as though I would just burst. Once we arrived at the studio, I met another young man who would be the technician for the day. He showed me where I would be singing and what to do. I was super eager to start and just wanted to jump in straight away. They got my track ready for me to sing over and I jumped right in with both feet. It was an incredible start. I nailed the track and the whole team in the studio couldn't get the song out of their heads. Everyone was singing along with me while I was in the booth. It was amazing. I could see everyone dancing and mouthing the lyrics through the glass which made me feel incredible.

I was smiling so much that my jaw hurt. I went over the song a few more times and made some little changes to the melody and added in a backing vocal. The guys explained to me that they would cut out parts of the song and replace them with better versions from all the takes we had done. After the long drive up and the time in the studio we made our way back. Two weeks later and I have received the demo version of my track. I can't believe, I'm on my way to the top…….

EMPOWERMENT PROJECT

GAUCHER TYPE III EMPOWERMENT PROJECT

The Empowerment Project - a group of young people who meet 3 times a year – gives young people with Type 3 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).

VOLUNTARY WORK IN AN ADULT HOSPICE - BY IRMA SHAH

I have started Voluntary work in an adult hospice and am going to write about this in my article. I have worked with the elderly in the past few years in many places such as hospitals, day centres and extra care sheltered living.

In these few years I have learnt so much by working and caring for people with dementia, Alzheimer's and who are just reaching old age. I gained a lot of experience and felt like I want to and need to learn a bit more but was not sure how.

In 2015 we were very grateful and fortunate to have two speakers from an adult hospice and a children's hospice speaking at the nGD conference.

Listening to both speakers I took a great interest and thought about going to work with not just elderly but working with adults as a whole.

A hospice is a home providing care to the sick or life limiting illness (palliative care) such as cancer, motor neuron disease and many more.

With the help from Helen Whitehead, Patient and family support Worker I was invited to meet one of the Voluntary Work Coordinators 'at Duchess of Kent Sue Ryder in Reading to speak about my options and was shown around the hospice. After filling out all of my forms and providing my references. I started my training on 31st of May. I am looking very forward to working in the hospice and telling you all about it in the Future.

Helen Whitehead writes, our most recent trip was to Swindon designer outlet where they had a day of catching up with each other, sharing experiences of living with type III Gaucher disease and getting there from all over the country by public transport.

EMPOWERMENT PROJECT

OVERSEAS VISITS

In January we supported two members of the group, Sara Khan and Radhika Dhayatker, to attend Gaucher clinics in Bangalore and Chennai in India.

RADHIKA DHAYATKER WRITES...

In January 2016 I was privileged to accompany Tanya, Helen, Elin, Dr Vellodi and Sara to Bangalore and Chennai in India. I've never been to India before and it was my first time travelling abroad without my parents. It was an amazing experience and I am very grateful for this opportunity.

In Bangalore we attended the clinics at the Centre for Human Genetics where we met medical professionals who were very welcoming and friendly. We were guests at a conference attended by doctors, patients and family members. I had the opportunity to share my story in front of about 80 people whom I didn't know. Amongst the guest speakers at the conference was a mother who had two sons with Gaucher Disease type 1 and a father whose daughter had Pompe Disease. One of the special guests was a lady who was one of the richest women in India owning a biotechnology company. She took a picture with me and Sara and posted it on twitter. After the conference we went out for an evening meal with the doctors and staff. The next day we saw children who had Gaucher Disease and MPS, some received treatment and some didn't receive any treatment.

In Chennai the clinics were busier; some of the patients had travelled a long distance to get to the clinic. There were fewer Gaucher Disease patients than MPS patients. Again some were on treatment and some were not. Two sisters aged 9 and 13 travelled a long distance to get to the clinic, both had type 3 Gaucher Disease but only one was on treatment. Healthcare, treatment and medication in India can be hard to access and therefore some patients do not get the treatment they need.

Sara, Kiran & Radhika

I gained a lot of experience and learnt a lot from visiting clinics in India, we are privileged in the UK to have free healthcare and treatment which is widely available. I enjoyed the shopping and site seeing and I loved the food.

SARA KHAN WRITES...

In January of this year I was invited on a trip of a lifetime to India by the Gauchers Association, the trip would include visits to centers that treat both adults and children with genetic metabolic conditions e.g. Gaucher disease, MPS and Pompe disease. I knew that this was not going to be a holiday and that I was going to learn about what it is like living in a country where patients and their families struggle day in day out not having or receiving the correct treatment and or medicines that they desperately need and that it was not going to be easy.

In the UK we are very fortunate to have the NHS but in India there is no such thing to help people with things like costs. Treatment for Gaucher is extremely expensive and as I realised more and more in each clinic, both in Bangalore and Chennai, from every patient, the many difficult challenges they face in order to try and receive the correct drugs or even alternative medicines. I did find this extremely upsetting and on more than one occasion there were times where I found myself leaving the examination room because I had never before been exposed to such an emotional environment. On the other hand I was happy because I did meet some extremely inspirational people including all the doctors, medical specialists and staff who I personally give immense credit too as watching and observing them over the week overall was a real privilege.

When we were in Bangalore I was asked to do a presentation on my life as a patient living with Type III Gaucher disease. I talked about the time I was diagnosed aged 16 months, growing up through school and finally college. I talked in great detail about my successes in and out of school, I felt very comfortable and happy on stage as I knew I wasn't being judged by the audience. Amongst the audience was Kiran Mazumdar Shaw who is the chairperson of Biocon Limited a biotechnology company based in Bangalore. I now know that Kiran is an extremely important person in India having met many famous people like A-list Bollywood stars...I even got the opportunity to take an exciting selfie with Ms Shaw! In Chennai I took part in a wearable technology workshop with a group of patients who have type I Gaucher disease, this was to evaluate how each of them cope with daily activities. To do this we first weighed each of them then measured how many steps they took walking across the floor with a measuring tape, we also used the measuring tape to measure their leg length. All the information we recorded on sheets and would eventually be recorded on the wristbands provided, which over the next few weeks will record the patients activity and be sent back to the UK along with surveys.

The hotels we stayed in both in Bangalore and Chennai were very nice, the staff were exceptionally polite, helpful and hospitable towards me at all times which made the trip even more enjoyable. The weather was amazing not too hot or too cold just pleasant throughout day and night. Transport was very efficient in and around India from the plane journey, the train journey to the hired car and rickshaws. I particularly enjoyed the visit to the sari shops and the temple it was lovely to experience a different culture.

I would definitely do the whole thing all over again

PATIENT AND FAMILY SUPPORT WORKER

Helen Whitehead - PFSW

Helen Whitehead

Over the last few months I have attended Gaucher clinics at Birmingham Children's Hospital and Great Ormond Street Hospital. It is great to catch up with patients I have met previously and new patients alike and to give information about the work the Gauchers Association does.

I have continued to support members with benefits applications – such as PIP (Personal independence payment), ESA (Employment Support Allowance) and DLA (Disability Living Allowance). I have recently attended a training session on Universal Credit (UC) a new benefit that is taking the place of various other benefits. I can support you through the process, 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 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 home or a coffee shop.

What is ESA?

ESA stands for Employment and Support Allowance and if you are ill or disabled will give you financial support if you are unable to work and personalised help so you can work if you are able to.

The quickest way to claim is by phoning the DWP (Department for Work and Pensions) free on 0800 0556688. Or print out the ESA1 form (www.gov.uk/employment-support-allowance/how-to-claim) and send or take to your nearest Jobcentre Plus Office.

You will then be invited to a work capability assessment to decide whether you qualify.

Travelling abroad this 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. We have a list of providers that our members have recommended who will insure people with Gaucher disease. Please contact me if you would like a copy.

ALISON WILSON GAUCHER IN IRELAND

Alison and one of the Northern Ireland Nurse specialists (Nicky Cluskey)

The last few months have been a whirlwind of organisation ahead of my maternity leave – by the time you are reading this article I will already have left my desk (I can hardly believe how fast the time has gone in!).

In the last few months I have spoken to most of our members across the Island of Ireland and lots of hours have been spent supporting families to complete forms, write care plans and meet with all sorts of different agencies to communicate needs. It has been a hectic few months!

You will all have received a letter encouraging you to make contact with the Team in the Gauchers Association Office if you need support in my absence. Please do get in touch on 01453 549231!

In my absence my mobile number will be diverted to the MPS Society – this is the other charity I work with and it supports another similar group of conditions. If you happen to get through to this number they will be more than happy to provide you with the contact details you need to get hold of a member of staff from the Gauchers Association

Thank you! I want to take this opportunity to thank all of our members and contacts across the Island of Ireland for all your well wishes over the last few months. I hope to return to work in the spring of 2017 but will be in the office to catch up with the medical team a few times between now and then.

Editor's note: We are pleased to announce Cora May Wilson was born on Thursday 23 June 2016, weighing in at 9lb 5oz. We send our very best wishes to the family

MEMBERS SURVEY - HELP US TO SUPPORT YOU!

Please look out for our online survey coming to you soon! This is an important project for us to help develop and improve our understanding of the needs from Gaucher patients and their families. Your feedback is important.

RESEARCH

Orphazyme – interested in the GD3 community

The Gauchers Association have been working with a small biotech company; Orphazyme from Denmark who are working in the field of LSD with small molecule therapies.

CEO Tanya Collin-Histed and Professor Tim Cox from Addenbrooke's Hospital, Cambridge are working with Orphazyme around the design of a trial and identifying possible trial sites around the world. For more information on Orphazyme please visit their website; http://orphazyme.com/

Rapsodi

See page 13

Gene Therapy Update

Over the last 7 years the Gauchers Association has been working with and has provided research funding to Drs Simon Waddington and Ahad Rahim and their team at University College London (UCL) to look at the potential of bringing Gene Therapy to the Gaucher community with a focus on Type 2 and Type 3 where there are huge unmet needs.

In February 2015 the Association received Orphan Designation for Gene Therapy for Gaucher disease and we are delighted to report that we have successfully submitted our report to the European Medicines Agency (EMA) on the progress that we have made over the last 12 months and what our plans are going forward.

The Gauchers Association continue to financially support Giulia Massaro, a PhD student for 3 years (2014 – 2017) through the UCL Impact scheme for a project focusing on investigating the efficacy and safety of minimally invasive intravenously administered gene therapy.

More information on both of these projects will be reported in the December edition of the Gauchers News on our progress.

Volunteers needed

The SHP-GCB-402 for Gaucher Disease is looking for 40 people to take part in a clinical research study. To be eligible you must:

- Be between 16 and 65 (inclusive)
- Have type 1 Gaucher disease
- Not have received enzyme or substrate replacement therapy in the past year
- Not have received any osteoporosis-specific treatment in the past year

Additional criteria will be assessed at an initial screening visit. For further information, please contact Derralynn Hughes at the Royal Free Hospital or visit www.ShireTrials.com

Gaucherite

See page 15

Genzyme's GD3 Oral Therapy – Clinical Trial Update

At our 7th nGD Family conference in Manchester in November last year we were extremely grateful to Dr Derralynn Hughes and Dr Anupam Chakrapani who led a small closed session for the patients and parents of our GD3 community to inform them of a clinical trial that would be starting in 2016 and that 3 of our adult centres in the UK would be designated as trial sites.

It was extremely important to be able to have these discussions with the patients and their family members so that they could ask questions and then take that information away with them to decide if they wanted to put themselves forward for the trial once it opened in mid-2016.

This phase II trial will be opened to adult patients aged 18-40 years old with a confirmed diagnosis for Type 3 Gaucher disease. The trial drug is an oral therapy which will see patients take the new oral trial drug for 6 months in addition to their ERT, with a view to then continuing for the remainder of the trial, 52 weeks in total with the oral drug ONLY which is believed to cross the blood brain barrier.

More information will be reported in the December edition of the Gauchers News on our progress.

CONNECTIONS BETWEEN GAUCHER & PARKINSON'S

Lucy Collins, a PhD candidate working alongside Professor Timothy Cox and Professor Roger Barker gives her final update on her studies

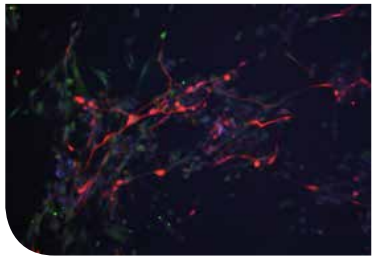
We have come to the end of our research study Investigating the connections between Gaucher and Parkinson's Disease and we would like to sincerely thank everyone for participating.

We would like to inform you of the progress we have made and the main outcomes so far in the study. The aim of this study is to better understand the association between Gaucher disease and Parkinson's disease. We are trying to better understand why the two diseases sometimes occur together in families, and we believe that by the exploring these two disorders closely we can get an understanding of how Parkinson's disease might be caused.

The study objectives are as follows:

1. To assess memory and thinking in people with Gaucher disease and relatives of people with Gaucher disease.
2. To create person specific neurons from skin cells.

What we have been doing with your skin biopsies samples:



We have been working on developing a new personal cell model to better understand the underlying cause of Gaucher and Parkinson's. This is done by creating nerve tissue from cells taken from a small skin sample. These cells are 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 shown is an example of neurons (in red) made from a skin sample. We worked on generating a high number of neurons and found that growing them at low oxygen conditions gave higher number of neurons. 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.

One of these experts is Dr. Janelle Drouin-Oulette a researcher at the University of Lund in Sweden. She is using sophisticated techniques to better convert these skin cells into neurons.

Aims of the memory and thinking tests. In parallel to our cell work we looked at memory and thinking in people with Gaucher and/or Parkinson's disease. We wished to see if there are any patterns or signatures of differences in how problems are thought through, solved and then remembered that may help us identify the apparent link between these two conditions. We found that there was large variability in people in these tests. These tests will also feature in a new upcoming study called Gaucherite (see page 15).

If you would like any further information about the study or to take part please contact Lucy on lmc58@cam.ac.uk

Thank you kindly to everyone who has helped us with our study.

Please help us with a pioneering new research study that aims to find answers into the link between Gaucher and Parkinson's.

Recruitment begins on a new internet based study that has been designed and developed with help of people with Gaucher and their families.

Over the past six years researchers led by the neurology and haematology teams at the Royal Free Hospital and University College London have been investigating a link between people who carry the GBA gene, which causes Gaucher disease and Parkinson's disease. 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 researchers have been able to study what happens in cell pathways of the GBA gene as a result of the immense generosity of people with Gaucher disease who attend the Lysosomal Disorders Unit at the Royal Free hospital. By giving their time and help we have been able to make important discoveries about how the GBA gene is linked to Parkinson's and most importantly how we might be able to stop this happening.

Understanding what changes happen in some people many years before Parkinson's develops and why other people in the same family, who also carry the GBA gene and don't develop the condition, will help us predict who is at risk of developing Parkinson's so in the future we can give treatments to stop this occurring.

The next step is a new online research study called **Rapsodi** that has been developed with the help of people with Gaucher and their families and can all be done from home.

The study is being led by Professors Anthony Schapira, Atul Mehta and Dr Derrylynn Hughes at the Royal Free Hospital, London. They are now working with other lysosomal units throughout the UK. These are based at: Addenbrooks hospital, Cambridge, Queen Elizabeth hospital, Birmingham, Salford Royal Infirmary, the Royal Manchester Children's hospital, the National hospital for Neurology and Neurosurgery, London and Great Ormond Street Children's hospital, London.

The discovery linking the GBA gene to Parkinson's is probably one of the most important breakthroughs in Parkinson's in recent years and opens up new and promising avenues for research into how we may target and develop powerful new treatments for everyone at risk. But none of this research is possible without the time and help of people who carry the GBA gene and the researchers are immensely grateful to your participation

IF YOU WOULD LIKE TO GET INVOLVED

The study takes about 45 minutes to an hour to do each year via an online portal you can log onto and then some postal tests to do at home. The study team is on hand to take calls and talk through any questions. **If you would like to get involved or learn more please: Dr Stephen Mullin or Sarah Cable (Clinical Research Nurse) rapsodi@ucl.ac.uk 07753 982 063**

NICE UPDATE

Gauchers Association Patient Experts go to NICE to support the appraisal for Genzyme new oral Therapy for Type 1

In December 2015's edition of Gauchers News, we reported that Genzyme had re-engaged with the National Institute of Health and Care Excellence (NICE) new Highly Specialised Technologies Appraisal Committee to schedule a technical appraisal of 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.

As part of the NICE appraisal the Gauchers Association were invited to submit a patient statement on behalf of the UK Gaucher community on this new treatment and we are extremely grateful to all of our members who supported us in making this possible. The Association were also asked to nominate a number of patient experts from the Gaucher community who had experience with the new treatment either through the clinical trial or on compassionate grounds and we were delighted to put forward three nominations for this. NICE have now selected two of the three patient experts who have now submitted their own patient statements and who will attend the formal appraisal committee meeting on 20th July in Manchester. We are extremely grateful to all three of these patients' experts for their support in this appraisal as it is a big commitment and we hope that leading up to and on the day we will be able to support them both in this event.

Looking ahead, the first committee meeting will be held on the 20th July and a decision will be made by NICE based on all the evidence submitted prior to and on the day. NICE will then either approve the treatment or ask for additional evidence and a second appraisal date is set for 21st September if needed.

This is a very important time for Association and we are confident that we will provide a strong and fair case to NICE to help them make a decision regarding access to Cerdelga for the English Gaucher community.

More information on this appraisal will be reported on the Association's Facebook and website and in the December edition of the Gauchers News.

Editor's note:
The Association have been informed by NICE that the first appraisal meeting due to take place in Manchester on 20th July has been rescheduled and will now take place on 21st September with a second date of 22nd November.

GAUCHERITE 3 YEAR STUDY

Calling ALL Gaucher Patients - Are you enrolled onto GAUCHERITE?

GAUCHERITE is a three year study that aims to better understand the UK population of patients with Gaucher disease with a view to developing new and better treatments. One aspect of the research will focus on bone disease and another will focus on neurological disease (brain involvement).

To date the study has successfully recruited 130 patients from centres around the country, the target for the study is to recruit 250 by late April to allow a period of 6 months to analyse all the data, the study funding ends in September 2017. Patients are being invited to enrol at all eight specialist Lysosomal Storage Disorder centres around the country.

How do I enrol?

The clinical team at the treating centres are sending out letters to all Type I & Type III patients a few weeks before they are due in clinic, this introduces the study and includes a patient information sheet which outlines what the study is about, what data they want to collect and other important information in order for you to make an informed decision about whether or not you would like to take part in the study.

When you attend your clinic appointment either your clinician and/or a member of the research team will go through the study forms with you and answer any of the questions that you may have. You can then make a decision and if you decide to take part in the study you can sign the form, the patient information leaflet can be taken home with you as a record of what the study is about.

I haven't heard from my centre?

If you would like to take part in the study but have not heard from your treating centre then please contact your clinical nurse specialist and ask them to send you the introduction letter and patient information form. Then next time you are in clinic you will be able to go through the forms with them and sign up to the study if you decide that you want to.

Creating a UK Gaucher database

One of the exciting things about this study is that a national patient database of all UK Gaucher patients will be developed which will be a valuable tool for further research into this community to address unmet needs.

RAISING AWARENESS
COTSWOLD WALK

Over £1,100 was raised at the Gauchers Association Raising Awareness Day

Sarah Allard, Information & Charity Officer writes 'On a sunny Sunday morning we welcomed 60 walkers of all ages to Dursley, Gloucestershire who eagerly collected their t-shirts and maps and set off to start a beautiful 5 mile circular walk, taking in part of the Cotswold Way. There were a few heart pumping climbs, but well worth it for the stunning views of the Cotswolds. The children were all given a trail card to collect stamps along the way to be rewarded at the end with a bag of well-earned sweets. With lots of chatter along the way and the opportunity to meet other walkers who all asked questions about the charity; the day proved to be a great success; the sun shone throughout which really complimented the array of bluebells in flower.

Whilst we were all out walking, Claire Lightfoot was very busy back at HQ preparing a full afternoon tea, which everybody fully appreciated and devoured upon their return, along with having the opportunity to learn about Gaucher disease and the important work we do through reading our A2 posters on display and chatting to CEO Tanya Collin-Histed and Patient & Family Support Worker Helen Whitehead. We were delighted to welcome a newly diagnosed Gaucher type III family who decided to make a long journey to be a part of our day. This was hugely important to them as it gave them the opportunity to talk and ask lots of questions to another type III patient, Tanya Collin-Histed and Helen Whitehead. They were the last family to leave in the afternoon saying they had a really enjoyed the day.

The Association would like to thank everybody who helped make the day such a success and raise over £1,100.

Our thanks also go to Sanofi Genzyme who supported this event through an unrestricted grant.

BIRMINGHAM CHILDREN'S HOSPITAL METABOLIC TEAM

The Metabolic Team at Birmingham Children's Hospital cares for the whole spectrum of Inherited Metabolic Disorders. The IMD team consists of Consultants, trainee Doctors, Clinical Nurse Specialists, Dietitians, Biochemists and administrative staff. Birmingham Children's Hospital is one of the designated Paediatric Centres for children with Lysosomal Storage Disorders.

For patients with a Lysosomal Storage Disorder, the team has designated Psychologists, Physiotherapist, Speech and Language Therapists. There is also a close working relationship with Neurology, Cardiology, Orthopaedics, Neurosurgery, Opthalmology, Respiratory, Dental & Stem Cell Transplant teams for patients with an LSD and these specialities participate in the MDT clinics and patient assessments, as required. As a result, these specialities have a wealth of experience and expertise to assist with the management of our patients.

At BCH we are fortunate to be able to offer multidisciplinary team (MDT) clinics. These enable patients to be seen in clinics on a single day by a number of specialists. The appointments are coordinated by the IMD Coordinator and helps to reduce multiple hospital visits. There are currently 12 designated LSD clinics scheduled throughout the year, to which the Patient Support Societies are invited. Currently BCH are building the first Paediatric Rare Disease Centre which will improve on the facilities offered to families travelling to BCH for MDT clinics. The Rare Disease Centre has been purposely designed to meet the needs of all patients accessing the facility including full accessibility for wheelchair users, a family kitchen, multi sensory rooms, youth room, 10 outpatient clinic rooms to allow full MDT clinics and an open waiting area that will allow families to network and meet with support groups.

The metabolic team offers 24/7 advice and support for their LSD patients, as required and the Clinical Nurse Specialists are available to provide support and advice during working hours. The Team meet weekly to discuss patients as a whole team, to ensure a multidisciplinary approach is taken. All Consultants are actively involved with all patients across the entire range of LSDs and patients and families get to know all the metabolic consultants over the course of their child's care.

The Metabolic Team have a good palliative care service and work well with Community teams to support patients requiring palliative support. The team also have a well established Transition service and link with University Hospital Birmingham and Addenbrookes Hospital to offer joint Transition Clinics for patients with an LSD. Young people are seen in a transition clinic at BCH between the age of 14 and 16 years and then are supported at an adult clinic, by the BCH team attending their appointment at the adult hospital.

Birmingham Children's Hospital is involved 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.

BIRMINGHAM CHILDREN'S HOSPITAL METABOLIC TEAM

CONSULTANTS:
Dr Saikat Santra, Dr Suresh Vijay, Dr Julian Raiman

CLINICAL NURSE SPECIALISTS:
Rachel Gould, Louise Simmons, Catherine Stewart, Elaine Salmons, Kirsty Darling

ADMIN STAFF:
Jenny Beardmore, Anna Hughes

IMD COORDINATOR:
Theresa Stokes

RESEARCH STAFF:
Alice Stewart, Jitendra Sheinmar

LABORATORY:
Tim Hutchin, Hayley Sherrod-Cole

ALLIED HEALTH PROFESSIONALS:
Emma Scobie – Speech & Language Therapist, Liz Wright – Physiotherapist, Rosie Jones – Dietitian, Shauna Kearney (Psychologist)

CONSULTANTS WITH INTEREST:
Alison James & Victoria Clarke (Dentists), Dr Evangeline Wassmer (Neurology), Mr Joe Abbot (Opthalmology); Dr Sarah Lawson (Stem Cell Transplant), Mr Guirish Solanki & Mr Desi Rodrigues (Neurosurgery), Shauna Kearney (Neuropsychology), Dr Ashish Chikermane (Cardiology).

GENZYME PATIENT ADVISORY MEETING

Contributing to Patient Needs and Developing Patient Support Programmes

On Monday 25th April Madeline Collin, a young adult with Type 3 Gaucher disease and a Member of the UK Gauchers Association's Board of Directors travelled to Amsterdam with her Mother Tanya Collin-Histed, Chief Executive of the UK Gauchers Association at the invite of Genzyme Europe to participate in a Patient Advisory Board meeting in Holland.

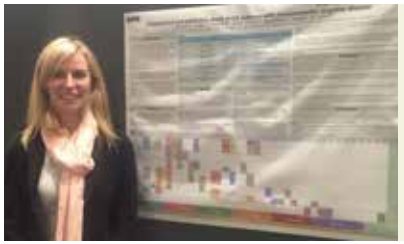
The purpose of the meeting was to bring together patient advocates from around Europe (representatives attended from Sweden, UK, Slovenia, Denmark, Greece and Serbia) to talk about unmet needs within the patient community with a focus on Type 3 Gaucher disease, bone disease and the links with Parkinson's and cancer. The format of the meeting was to hear personal stories from patients who have experience in these areas and then to have a discussion about what current support was available to patients around these topics and what the outstanding needs were.

Maddie writes; participating in this meeting was extremely beneficial to me, it allowed me to tell my own story of growing up with type 3 Gaucher disease, the challenges I have had to face but also to share how it has made me a stronger person, that I haven't let it define who I am but have used it to help others, to give them hope and to make a good life for myself. It's always hard to talk about the past and to focus on the fact that I have this condition but I know that it can help others and in making a decision to join the UK Gauchers Association Board of Directors I am confident that I can help others who are on the same journey as me whether they are just starting and are parents or are teenagers or young adults, living with such a rare disease can be very lonely.

Tanya writes; my remit for this meeting was to talk about the work of the Gauchers Association in the UK supporting patients and their families with type 3 Gaucher disease. Unlike many other European countries we have a largish (in rare disease terms) GD3 community with to date 29 patients in the UK and have recognised for many years that they have huge unmet needs which we can help support them with. After giving a formal presentation I then chaired a session asking a number of questions to stimulate a discussion amongst the group around non-medical needs of patients, how can patients influence research, how can we bring the GD3 community closer.

Needless to say I was extremely proud of Maddie and it was great to expose her to the wider Gaucher community, hopefully she will continue to be a great advocate for young people and also for patients with Type 3 Gaucher disease.

THE SUSAN LEWIS MEMORIAL AWARD



Since the last edition of Gauchers News, the Association has supported Dr Aimee Donald, Paediatric Clinical Research Fellow at Manchester Centre for Genomic Medicine to attend the WORLD Symposium present her work on neuronopathic Gaucher Disease.

Aimee Donald writes... The WORLD Symposium is a conference which brings together clinicians and scientists from all over the world to learn and share knowledge on rare diseases involving lysosomes. This year the symposium was held in San Diego and I was fortunate enough to attend and share my work on neuronopathic Gaucher Disease with the generous support of the UK Gaucher Association.

I currently work as the Paediatric Clinical Research Fellow with Gaucherite, a study which many of you will have been invited to participate in. Gaucherite is a study looking to understand more about how Gaucher Disease affects patients in the UK so that in the future we can develop more targeted and specific treatments for each patient as an individual. I am particularly interested in patients who have neuronopathic Gaucher disease and so I undertook a small piece of work examining the disease experience of a group of children and young adults who have sadly died as a result of the disease over the last ten years. Fortunately, not many patients have died during this period, however that means that generalisations about the disease can't be made on the basis of information gathered on such a small group. Reviewing their case-notes however highlighted the timing of disease progression for each patient; I can now use this information to investigate some of these findings in more detail. Also, by presenting this work at an international meeting, I was able to discuss our experience in the UK with doctors from other parts of the world who in some cases had similar experiences. By collaborating in our research we will learn a lot more about the disease and hopefully be able to generate new approaches to treatment in the future.

While at the conference I learnt more about the underlying scientific basis of treatment for Gaucher disease and also had the opportunity to learn about other lysosomal storage disorders which may inform our understanding of Gaucher Disease. I had a wonderful trip and would like to thank the Gaucher Association for enabling me to attend.

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

MY ANNUAL TRIP TO INDIA

Tanya Collin-Histed, Chief Executive of the Gauchers Association reports on her recent trip to India

Since 2011 I have been extremely privileged to travel to India on four occasions to visit Gaucher clinics in Chennai, Delhi, Mumbai and Bangalore. Each year I have the opportunity to meet new families whose children have been diagnosed and to meet up with children and their families who I have seen in clinic before. These trips are always emotional as in some cases it has given me the opportunity to see children grow up and be well on treatment, but in many cases see those not able to access treatment deteriorate and in many cases sadly pass away.

This year I was lucky enough to travel to Bangalore and Chennai to attend clinics with Dr Ashok Vellodi, Dr Elin haf Davies, Helen Whitehead, Radhika Dhayatker and Sara Khan as part of the Association Empowerment project (see pages 6&7).

On this trip three cases stood out to me that I would like to share with you:

Patient A

This young girl is now 9 years old, I first met her in 2013 when I visited Bangalore. She has a condition called Saposin C which is like Gaucher disease but cannot be treated with ERT but could be treated with SRT i.e. Zavesca or Genzyme new oral therapy Cerdelga. When I met her in 2013 she was very ill, her bone disease was very severe and she could hardly walk. Sadly despite a lot of efforts by the treating doctor and me we were unable to get her Zavesca manufactured by Actelion as they did not have a charitable access programme in place.

I was very shocked to see her once again this January as I did not believe that she was still alive, however she was very fragile and had just experienced spontaneous factures in her legs. It's hard to not take these images away with you and after talking to her doctor who had tried very hard to get some sort of treatment for this young girl, I promised that I would try my best to help in anyway possible.

A few weeks later, armed with this girl's photo I attended the WORLD conference in San Diego and showed this photo and recounted this girls' story to senior persons in Genzyme who I have had the opportunity to work very closely with over the years and respect very much. No promises were made but after a lot of hard work I am delighted to say that finally this young girl will be given the opportunity to receive treatment on a compassionate basis by Genzyme.

Patient B

This little bundle of joy is now 5 years old and I first met her in 2013 as a very sick child in her mother's arms, she had a huge liver and spleen and was very distressed, at the time there was no treatment available for her and her doctor wanted mum to agree to a splenectomy to save her life, however mum was scared and wanted to wait for ERT to be available. During the clinic session Dr Vellodi asked me if I would share my personal story with this mum and tell her about Maddie and how Maddie had had a splenectomy and how important it was to do this. A few days later this little girl had surgery and on my return to the UK I worked with the child's doctor, the Indian patient group and Genzyme and later on that year she started treatment through Genzyme charitable access programme, her treatment was later on provided through an employment insurance programme through the State where she lives.

Seeing her in clinic this January was incredibly emotional, she's beautiful and growing well just like any other 5 year old little girl. Sadly when I was in clinic I became aware that due to her father's employment status her treatment was going to be stopped, however after talking with her doctor and talking to Genzyme the family have been informed that she will continue to receive her treatment.

Patient C

A little boy aged just 3 years came into the clinic in Chennai with his parents, they were very young too. This child had been seen in a local hospital in December and told that he had a condition but to come along to the clinic in Chennai and see the doctor and they would help treat him. He had Type 3 Gaucher disease and mum and dad did not know this or what this meant for their son. They did not know there was no access to treatment for their child and mum stood in the room and silently cried. The team at Chennai are used to dealing with these difficult cases, manging these devastating diseases with symptomatic care where there is no access to ERT.

Finally Tanya writes, for me these trips to India have a number of purposes, they provide me with a chance to learn about the natural history of the disease without treatment, to understand how patients in countries like India where there is limited access to ERT are looked after by extremely dedicated doctors and in my role as CEO of the European Gaucher Alliance (EGA) to raise awareness of the remaining unmet needs of Gaucher patients globally.

WEARABLE TECHNOLOGY AND DISEASE SPECIFIC APPS FOR nGD

In December 2015's edition of Gauchers News, we reported on work being done by the Association in partnership with Dr Aimee Donald, Paediatric Clinical Research Fellow, Manchester Children's Hospital and Dr Elin Haf Davies on a pilot study for the use of wearable technology and mobile phones apps 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.

Throughout this project a group of type 3 young adults have been working closely with the Aimee, the Association and Elin to develop the app to ensure that it collects data that is important to them as well as ensuring an app design that is easy to use. In Manchester at the 7th nGD Family Conference Aimee, Elin and James Skinner from aparito (the company co-founded by Elin Haf Davies that has been developing the software) met with the girls to show them the watches and phone apps and ask for their feedback, this was then collated, analysed and changes made.

Several months later in early February the girls met up again with Aimee and James to discuss the changes and also to measure how long it took the girls to complete some of the questionnaires on the phone app and then on paper as a comparison.

I am delighted to report that ethics has now been approved for this project through Central Manchester University Hospitals NHS Trust and we intend to identify a date over the next few months to enrol the young Type 3 patients, aged 13 years and above in this study.

We hope to present some of the data from this study at several International Conferences in 2017.

This project has been made possible by a patient support grant from Genzyme, a Sanofi Company and Shire.

DISEASE SPECIFIC APPS FOR nGD

Patient engagement

aparito has worked with the GA to develop a disease specific app

nGD family conference:

- Presented concept
- Workshop with paper mockups
- Feedback and suggestions session

Design and prototype:

- Built simple prototypes
- Incorporated feedback and suggestions

nGD empowerment group:

- Tested tech demos
- Compared with paper based tests

RESULTS

Lessons from user testing with nGD empowerment group

- The app represented a 47% time saving on paper PROS
- 100% of GA testers would rather use the app

NEW APP FEATURES FOR nGD

7 new PROs:

- Global self worth
- CHU 9D
- Perceived stress
- Rosenberg self worth
- PedSQL MFS (multidimensional Fatigue Scale)

NEW APP FEATURES FOR nGD

7 new events including:

- Sleep
- Bone pain
- Breathing problem
- Tremor
- Menstruation
- Missed school/work
- Other illness

LSD COLLABORATIVE S4RD

Meet Laura – Students 4 Rare Diseases (S4RD) new employee

'If You Hear Hooves, it May be a Zebra…A Medical Student Empowerment Project' is a joint project between the UK LSD Patient Collaborative and a Committee of Medical Students originally from Barts London School of Medicine and Dentistry, now qualified doctors working as F2's in hospitals around the country.

Following the success of this project the UK LSD Collaborative secured funding from Biomarin, Genzyme, Shire and Ultragenx to fund a post to continue to develop rare disease societies throughout medical schools in the UK. I am delighted to report that Laura Curran was appointed for 15 hours a week to be the projects new Educational Co-ordinator. Laura is based at MPS House in Amersham and is managed by the UK LSD Patient Collaborative.

Laura writes; "Hi I'm Laura and I joined Students 4 Rare Diseases (S4RD) in March as Educational Co-ordinator, working on a part-time basis.

My main responsibility is to raise awareness of the organisation and promote the work of the S4RD amongst both our current medical school societies and within non-member medical schools. I am tackling this by utilising social media to try and enter their sphere and by contacting the schools directly to help us advertise our events and who we are. I am hoping to use social media as a place we can share important educational information provided by the LSD Collaborative and other reliable medical sources on the individual rare diseases, to our following medical student's.

One of my aims is to update our current website and make it more multifunctional where medical school societies can log in and obtain lecture notes from events they could not attend and share them amongst their fellow students. And, also upload lecture notes from their own events so that they can be shared equally. It would also be a great place to store information on event speakers and other useful information.

As well as being here to support the S4RD committee I am also a single point of contact for the students in the medical societies and potential medical schools. It is important that I am here to support them in any way I can, and also as medical students move up through the ranks, I want to nurture and develop relationships with the society leaders and the rest of the society so we always have other students in line to take over.

I have really enjoyed my first few months and am really excited about assisting the committee in progressing the fantastic work that has already been done. This is a completely different role to what I have done previously, and a new post for the committee, hopefully I will be able to use my previous experience in making this role work for the committee!"

Contact Laura, Students 4 Rare Diseases (S4RD
Phone: 01494 764 788 **Visit:** www.students4rarediseases.org
Working hours: Tues 12.30-14.00. Wed 11.00-7.00. Thurs 12.30-17.00.

RARE DISEASE DAY 2016

As you may already know, Monday 29th February saw the ninth International Rare Disease Day. Each year sees hundreds of patient organisations from around the world get involved to raise awareness of rare diseases, like Gaucher disease and put them in the spotlight.

Eurordis, the organisation behind Rare Disease Day reported 2016 was bigger and better than ever before. Events were held in 85 countries (all 28 EU countries) including some new comers to Rare Disease Day in the form of Andorra, Aruba, Indonesia, Libya, Mauritius, Moldova, Tanzania, Tunisia, Uganda and Zimbabwe proving just how this important day is growing year by year.

The theme for 2016 was The Patient Voice' recognising the crucial role that patients play in voicing their needs and instigating change that improves their lives and the lives of their families and carers.

The Rare Disease Day 2016 slogan 'Join us in making the voice of rare diseases heard' appeals to a wider audience, those that are not living with or directly affected by a rare disease, to join the rare disease community in making known the impact of rare diseases.

The Gauchers Association got involved by running their own Social Media campaign throughout the day and invited our members to include their own words or pictures to help others understand what it is like to live with a Rare Disease. I would like to say a BIG thank you to everybody who got involved; I am pleased to say we had over 2,000 views on our Facebook postings.

As well as running our Social Media Campaign, on Monday 29th February Sarah Allard attended celebrations hosted by Sanofi Genzyme and gave a presentation on the importance of raising patient voices for those affected by Gaucher disease.

Vivienne, Toni, Peter & Sarah

To celebrate Rare Disease Day, each year The Independent contains a supplement by Media Planet that is dedicated to rare diseases, featuring everything from rare disease medicine commissioning and research to patient stories. For 2016 the Gauchers Association were fortunate enough to be given the opportunity to feature an advert within the supplement, helping to raise awareness for Gaucher disease; our thanks to Shire for supporting this initiative.

LONDON MARATHON

Five runners raise over £11,500 for the Gauchers Association

More than 39,000 runners took part in the 36th edition of the Virgin London Marathon on Sunday 24th April, including the one millionth runner crossing the finishing line. Calum Phoenix, Matthew Gillan, Giulia Massero, Nick Goldstein and Julia Teper all ran for the Gauchers Association, this event being one of our biggest fundraising events and forms a significant part of the income we need to continue to support the Association's activities and initiatives. Our runners' stories:


Calum Phoenix


Nick Goldstein


Giulia Massaro


Julia Teper

Calum Phoenix writes; well wow, what can I say! after all the build-up, the long cold nights running throughout winter, the early morning quick light jogs before my day at work, the long weekend runs to build up my distance and of course the well enjoyed 'let's do absolutely no exercise today' days .

Of all the preparation that I put into the London Marathon, nothing could have actually prepared me for the feeling of hitting the wall on 22 miles but even more so nothing could have prepared me for the elation of actually crossing the finish line. The spectators, the drums, the whistles, the bands, the 'GO ON CALUM, KEEP IT GOING' all helped me to keep pushing on, to help remember why I was running the marathon even when your mind starts to drift after 26 or so miles. I can't thank the people of London enough for making it one of the most memorable days of my life, something I will never forget. To anyone thinking of running a marathon, do what I did and if you get an opportunity, just say YES! You won't forget it!!!!!"

Nick Goldstein writes; it was a huge honour to run the marathon for the Gauchers Association. I'm delighted to have raised over £2500 for such a worthy cause. The atmosphere is truly incredible and it was inspiring to be part of the event. To anyone considering a marathon next year I cannot recommend it highly enough. Raising money for charity whilst putting your body through an immense test is the perfect cocktail for an inspiring experience. At about 35km it felt like the final 7km would be too much. However, the thought of letting down those that had sponsored me and donated so generously is what got me, and I'm sure many others, to the end.

Giulia Massaro writes; when I first mentioned to Tanya that I always wanted to run a marathon, I underestimated the effort, tears and sweat it would take (literally!). Two years later there I am, jumping on the early tube on a cold Sunday morning. Destination: Greenwich. Waiting for me there was a colorful river of people in running shoes. The atmosphere was great, from the starting line to the very last mile: the spectators cheered and everyone appreciated the immense determination of the runners, even when exhaustion caused them to start walking. I saw thousands of people putting themselves on the line and fight for the vest they wore. It doesn't matter for who we were running for, how long it would take, if we received a medal or not: we were there for different reasons, but our common goal was to help.

Every day I contribute to the Gaucher cause with my scientific research, but this time I was proud and honored to give my personal support to the Association. Thank you for giving me this exciting opportunity and thank you all for your generous help!

Matthew Gillan writes; 24th April at last, race day. My wife wakes me up at 6am with the same worried look on her face she's had for the last few weeks. Still asking me if I'm sure I want to compete. I remind her of the last 8 months winter training 4 days a week life's been on hold preparing for this day. Half an hour after the race starts I cross the start line and off we go AMAZING . Thousands of people as far as you can see. The first ten miles fly past, 12 miles in, Tower Bridge WOW I'll never forget crossing it GOOSEBUMPS. The last six miles were PAIN. Reading people's stories on their vest got me through. The best in the human spirit on show.

I will always carry the people involved with Gaucher in my heart for the opportunity

And will endeavour to spread awareness about this little known disease wherever I go

Julia Teper writes; this year I was honoured to have had the opportunity to run the London Marathon for The Gauchers Association.

Despite this being my third London Marathon the amazing atmosphere and excitement at the start line was no different to when I first ran it nearly 10 years ago.

Thankfully despite a light scattering of snow first thing the weather conditions were great and the crowds were as enthusiastic as ever. I was really lucky to have my husband and daughters and some lovely friends come and support me on the day which really gave me a boost when I was feeling exhausted!

The run for me was very hard as I wanted to achieve a personal best. There were moments when I just wanted to stop running and join the walking wounded around me towards the end but I kept going and achieved a time of 4 hours and 52 minutes which was what I was hoping for!

The lovely Emily from The Gauchers Association greeted me at the finish line and I was delighted to share with her that I had raised over £2500!

Marathon day ended for me with quarter pounder meal, long bath and a big sense of achievement!

The Association would like to thank all its members, friends and their supporters who helped to raise £11,552. A very special thank you also goes to our five runners who endured the 26.2mile course to help us.

We are delighted to announce we have FIVE Golden Bond places available for 2017, please contact Sarah on 01453 549231 or email sarah@gaucher.org.uk if you feel inspired to take part.

MEMBERS FUNDRAISING

Donations received from November – May 2016 totalling £ 4,914

Generous donations have been received from:
Ellen Solomons, E G Ansell, S Buzzard, Keith & Joy Moore, Akshantha Shetty, Lee Baker, Angela & Ben, Dean Turner, Eve Hatton and Lewis Saunders.

Julie Howells kindly donated £360 in connection with a St David's Day service held in her local Carmel Baptist Church. After the service the congregation enjoyed lunch together of Calve (Welsh soup), Barabrith and Welsh cakes, with each member paying a small fee.

The Royal Naval Club and Royal Albert Yacht Club very kindly donated £120 following an after dinner talk on ocean rowing by Elin Haf Davies

Beaumaris Rowing Club raised £320 followed by once again a talk by Elin Haf Davies about her rowing / sailing life.

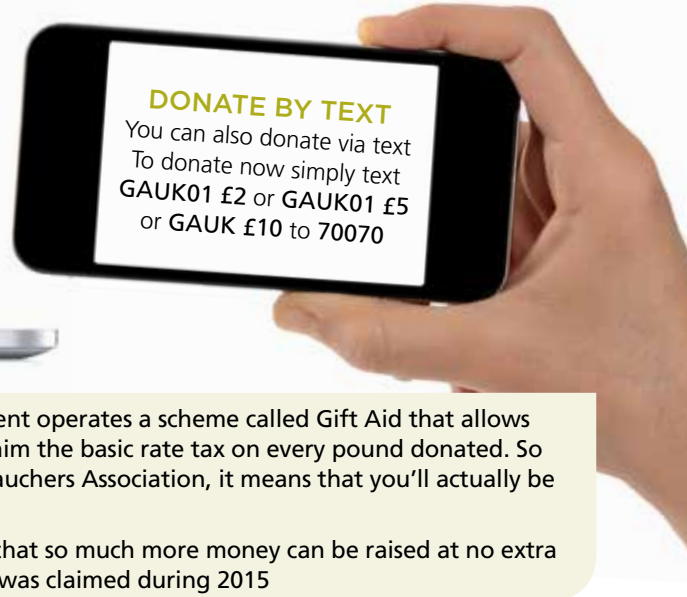
Congratulations to Stephen Mullin who recently completed the 18 mile trail run in Devon called the Hartland Hartbreaker and raised £398

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.

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



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

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