6th nGD Family Conference Success

Type III adults take a bow after organising conference

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Welcome to the December 2013 edition of Gauchers News. In this edition you will read about the 6th nGD Family Conference. It was a landmark event for us as it was the first conference planned and organised (with a little help from Tanya and Sarah) by a group of very special young women.

Back in 2006, the wonderful nurses at Great Ormond Street started the Aunty Day Project which brought together a group of Type III girls aged 9-18 to build their confidence and social skills and to give them a secure and familiar environment in which they could share experiences with other children facing the same challenges. That same group of girls have now matured into young women and as they transition into a new stage in their lives the Association is delighted that they are participating in the Empowerment Project set up following the PALs Award granted to the Association by Genzyme earlier this year.

One of the initiatives of the Empowerment Project was for the Type III group to plan and host the nGD conference and I am sure that all who attended would agree that it was a fantastic success and all of the girls did an incredible job. I wanted to take this opportunity to specifically mention the personal stories that a number of the girls shared with everyone on the day which I'm sure no-one who attended will ever forget. It took a huge amount of courage for each of them to share some very difficult thoughts with a large number of people, many of whom were strangers, and each of them spoke with eloquence, emotion and a huge amount of maturity. However the most wonderful thing about each of their stories was the positive message they left everyone with and the fact that they are all achieving their own personal goals and looking to the future with huge optimism.

Everyone at the Association is extremely proud of all of the young women who are part of the Empowerment Project and we look forward to seeing and hearing about more of their achievements in the future.

As usual this edition of the newsletter is packed with articles on various topics so enjoy reading it and please do let us know if you have any feedback, suggestions or comments. They are always welcome.

Wishing you all season's greetings and a healthy and happy New Year.

Best wishes,

Dan
Jean Minty

21st March 1955 – 20th July 2013

Sadly Jean passed away on the 20th July 2013, she had recently suffered a water infection that went septic and had to be treated in Wigan Infirmary during which time she lost a lot of weight.

Jean was diagnosed with Gaucher disease 50 years ago at the age of eight. By nine she had had her spleen removed but she had difficulty walking and at the age of 11 she was admitted to Wrightington Hospital where she spent a long time on a correctional frame. This weakened her leg muscles and upon release she had to wear a calliper.

These setbacks did not stop Jean leading a full and active life. She joined the Guides and youth clubs and she acquired a passion for dancing and Northern Soul music which she enjoyed at the clubs and dances in her area.

In her later teens, she left home and travelled south to Plymouth, working and sharing a flat with her cousin Gillian. She moved on to Culrose Naval Base and spent some time working for NAAFI before moving on to work in a hotel in Guernsey where she met George, her husband-to-be.

In the late seventies Jean gave birth to her son Jay which led to complications with movement in her left leg. This resulted in her first hip replacement operation in the early 1980s which gave Jean a new lease of life and culminated with the birth of daughter Gemma in the mid-eighties.

At the turn of the 1990s Jean needed the other hip replacing which was again done at Wrightington by Professor Wroblenski and again was the catalyst for another lease of life. About this time Jean met a man who would change her life, Professor Timothy Cox.

Professor Cox found out more about her condition which gave Jean a greater understanding of what she could achieve. This gave Jean the determination to raise money for the Gauchers Association and at the first conference we attended she presented a cheque for £1000 to the Association.

Jean went on to learn how to drive, so she could get to her appointments at Addenbrooke’s but sadly she developed Parkinsonism and had to give up driving. She was transferred to Dr Dick at Hope Hospital but things did not improve. Jean didn’t let this get her down and she still wanted to get out and about.

Sadly her first hip replacement now needed a revision. The operation was done again at Wrightington by Professor Wroblenski and again it was a success and Jean’s lease of life returned. Unfortunately Jean’s Parkinsonism got worse and she returned to being cared for by Dr Lennox and Dr Barker at Addenbrooke’s but things did not dramatically improve so after a few slips and falls she was admitted to a nursing home.
During her time in the nursing home Jean still wanted to go shopping, out for dinner, to the cinema, visiting mummy and getting home as much as she could manage. During this period she had another hip revision but deterioration of her Parkinsonism curtailed her semi-active life and more time was spent in the home. Due to her lack of movement, extended ‘off periods’ of her Parkinsonism, loss of appetite and huge weight loss, when her last water infection was cured she didn’t have the strength to fight the lows of her Parkinsonism and this was what finally ended her courageous, caring life.

Jean is still sadly missed by family, friends and acquaintances.

Editor’s note: The Association would like to express their thanks to Jean’s husband George for writing this.

In Loving Memory of Laureenna

It is with deepest sadness that in August 2013 one of our Type III young women passed away. She was just 28 years old. Laureenna was a vibrant, determined young woman and a member of our Type III girls group and she was an inspiration to many of the younger Type III girls. She shared her story at the Association’s 2007 Family Conference and recently contributed to the transition booklet (see page 7). Laureenna will be sadly missed by all of us and our thoughts are with her family and friends, writes Tanya Collin-Histed, Chief Executive, Gauchers Association.

Dr Ashok Vellodi, Paediatric Consultant at Great Ormond Street Hospital was Laureenna’s consultant for many years, he writes:

‘I would like to thank the Association for inviting me to write a few words about Laureenna. Having looked after her from the age of two, I was able to observe, first hand, her brave and indomitable spirit battling with everything that came her way (and she had more than her fair share of problems), and she still came out on top where few would have succeeded. Her courage and fortitude inspired everyone who met her. It was a privilege to have had the opportunity of looking after her.’
Didn’t They Do Well!
Type III Girls’ Personal Stories

The Gauchers Association are delighted to include in this edition three of our younger members’ recent achievements. Below Nadia, Maddie and Radhika tell us how through their determination and hard work they are reaching goals in their journey into adulthood.

**Nadia Fattouki** writes, ‘Going to university far from home is exciting but yet a challenge, especially having to deal with my health. It is all about being an independent person and having Gaucher disease makes it a little bit of a struggle to keep up with everyone else at the same speed. It is not like school or college having special support teachers helping me out with my work and mobility needs. There is very good support at university as long as I know how to approach it. I may not be like everyone else at university, for example I am not a crazy party goer but that’s the great aspect of being at university because no one is the same. I make sure I look after my health whilst being away from home, by eating healthily to prevent bad cold and flu symptoms that can increase the risk of having an infection and keeping fit by walking. Overall I know it is not easy but I will keep learning.

**Maddie Collin** writes, ‘In August of this year I got a part time job as a care assistant in a nursing home looking after people with cancer, dementia and other long term illnesses. The job has been really good for me as it fits in with college and allows me to work flexibly so I don’t overdo it. I really enjoy looking after the people and have learned so much from my experiences. The job has its challenges like any other but is so rewarding. My Gaucher doesn’t impact on my abilities as a carer but I do get tired which is why the bank contract works so well for me. My colleagues are very understanding and because of this I don’t feel I have to explain about my condition much and I’m like any other member of the team.

**Radhika Dhayatker** writes, ‘I am 17 years old and I have Type III Gaucher disease. Having a rare medical condition usually becomes the main focus of someone’s life, however I am very ambitious and I’m currently studying for ‘A’ levels in Maths, Biology and Chemistry. Knowledge is power. I have completed Higher tier GCSEs achieving one A, four Bs, three Cs, one D and a Distinction. I could have done a bit better but I am very happy with my results. I accept it was challenging and now I want to go to university to achieve my goals and aspirations.'
Shaving for Awareness

Sarah-Jay Pickton was diagnosed with Type III Gaucher disease in 2011. So far she has raised £430 for the Gauchers Association by shaving her head! Please read her story –

‘I was diagnosed two years ago and I must say I had never heard of Gaucher disease. This is why I wanted to raise awareness and funding as everyone I talk to about the disease has never heard of it, including many doctors. This is a concern for me and fellow Gaucher patients.

There are a few people that are so dedicated to taking care of us; Dr Hughes and Linda Richfield from The Royal Free Hospital in London give up so much of their time to talk to us and take care of people with rare diseases. I have two wonderful nurses Debbie and Val who give me my weekly infusion and without funding and awareness, receiving my weekly medication wouldn't be possible.’

Frances, Sarah’s mum says, “Sarah has embraced her new look and calls it her ‘symbol of achievement’. She has had lots of people asking her why she cut all her hair off and it makes a good talking point in raising awareness for Gaucher disease. Sarah is really happy to have done this and would like to thank everyone who has been kind enough to donate so far and also to let everybody know that her fundraising page will be active until the end of January 2014 if you would like to support her.”

Please go to www.justgiving.com/Sarah-Jay-Pickton and give as much as you can to such a worthy cause. Thank you.

www.gaucher.org.uk

Type III Young Person’s New Transition Booklet

As part of the ‘Empowering Young Type III GD Patients to Shape the Future’ project (see page 8) the Type III young people have put together a new transition booklet. Maddie Collin who has been the lead on this project writes –

The idea for this booklet came from listening to everyone’s experiences with transitioning from paediatric to adult care and what they found challenging. Everyone played an important part in the making of this booklet, whether that be writing articles, finding out information about the different page topics or coming up with ideas for the layout. We hope that the booklet will help other Type III’s to have a positive experience with transitioning and that it will help them with any questions or problems they are having. The booklet is based on our experiences as patients.

The booklet is available as a hard copy or PDF, please contact Sarah on sarah@gaucher.org.uk if you would like a copy.
Empowering Type III Gaucher Patients

In June’s ‘Gauchers News’, we reported on our Global Patient Leadership (PALs) award for our project ‘Empowering Young Type III GD Patients to Shape the Future.’ Here is an update.

**Getting Together**
The girls, together with Tanya Collin-Histed and Niamh Finnegan, Clinical Nurse Specialist at Great Ormond Street, and supported by Dr Derralynn Hughes from the Royal Free met in June and September. The majority of the meeting was taken up with planning the 6th NGD Family Conference and the development of the Transition booklet that was launched at the Family Conference in November (see page 14/15 for a full report on the Family Conference).

At our June meeting we were joined by a new member, a young girl who wasn’t diagnosed with Type III Gaucher disease until recently and a diagnosis at this late stage in her teens came as an enormous shock to her and her family. Meeting the other girls and also for her mum to meet other mums in June was a great support which will hopefully continue through these meetings as well as the girls keeping in touch by text and Facebook!

**Going Forward**
After all their hard work planning for the nGD Family Conference the girls will have a few months off and meet again in the New Year to look at other projects. These projects include writing CV’s, interview skills, work experience placements, joining the Gauchers Association board, having a section on the Gauchers Association website and a possible trip to Israel for a few of them to experience international Gaucher projects.

‘Congratulations to the Gaucher association for the PALs award and to the girls for all of their tremendous hard work creativity putting together the family meeting. It is a privilege to work with them’

Dr Derralynn Hughes

*Editor’s note: these girls are a true inspiration, see page 6 of the newsletter to see what they have achieved.*
All Ireland Advocacy

The All Ireland Advocacy and Support Service is only 9 months old but we are delighted to have received several advocacy and support requests in these first few months.

The All Ireland Advocacy and Support Service provides support to individuals and families living with Gaucher Disease on the Island of Ireland. If you have any unmet support needs please get in touch with Alison on 0044 77862 58336 or 0044 28950 47779 or by email alison@gaucher.org.uk. If you would prefer to discuss your support needs in person, a home visit (or meeting in a cosy coffee shop) can be arranged.

Support comes in many forms – some people require written information to pass on to employers, insurance companies, schools or healthcare professionals; while others just need a chat and an opportunity to talk over their concerns.

Making Links and Raising Awareness – In these first 9 months we have had the opportunity to meet and liaise with pharmaceutical representatives and healthcare professionals across the Island of Ireland. We have also represented the Gauchers Association at Northern Ireland Rare Disease Partnership (NIRDP) meetings and at the Rare Disease UK (RDUK) management committee meetings. Making links and friends within the ‘Gaucher World’ is absolutely critical and we hope that people within the rare disease, metabolic and Gaucher Disease Communities are now aware of the Gauchers Association’s presence in Ireland.

How can you help spread the word? Please tell your homecare nurses, hospital doctors, school staff and any other professionals you come into contact with about our service. We have an excellent source of resources and can provide support to professionals as well as families.

What next? 2014 will bring with it a renewed focus on education. At the end of 2013 the Gauchers Association is being represented at a series of five ‘Genetic Road Show’ events for GPs, Public Health Nurses and Primary Care Doctors. These events were set up by the National Centre for Medical Genetics (Dublin). On 13th January and 28th February 2014 The Gauchers Association will be represented at Rare Disease events in Stormont Assembly Buildings (Belfast) and at an All Ireland Rare Disease Day event in Riddell Hall (Belfast). At these events Gauchers Association literature will be made available to patients, families, healthcare professionals and politicians. Raising awareness is key to improving services for those affected by rare diseases like Gauchers Disease.

Until Next Time... I look forward to updating you in our next edition with some photos of our events and education sessions. In the meantime, if you are planning any awareness or fundraising events in Ireland please do get in touch – we would be happy to support you.

www.gaucher.org.uk
Welsh Review of Orphan and Ultra-Orphan Drugs

The Welsh Government announced a review of their appraisal process for orphan and ultra-orphan medicines earlier this year. Mark Drakeford, Minister for Health and Social Services requested a group be established to review the appraisal of orphan and ultra-orphan medicines in Wales.

On August 27th 2013, three patient organisations were invited to present oral evidence to the review panel regarding access to funding for treatments, the experience of patients that had experienced delays from the local health boards for processing funding applications and the opportunity to share experiences (good and not so good) and identify systems that work well elsewhere and what makes this happen.

On behalf of the UK LSD Patient Collaborative, Tanya Collin-Histed, Chief Executive of the Gaucher Association attended the meeting and gave an oral presentation on the challenging experiences LSD patients have had over the past few years accessing Enzyme Replacement Therapy (ERT) and Substrate Reduction Therapy (SRT) compared to England where patients are able to access these treatments through the LSD highly specialised service, if they meet the eligibility criteria.

As a result of Tanya’s involvement in the meeting at the Welsh Assembly in August, she will continue to represent the Collaborative by providing advice and feedback on draft polices and being a member of implementing the Rare Disease Strategy for Wales.

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The LSD Patient Collaborative wins Patient Leadership Award (PALs)

We are delighted to announce that Lysosomal Storage Disorder (LSD) Patient Collaborative Group UK was the successful recipient of one of just eight Global Patient Leadership (PALs) awards from Genzyme. Out of 43 applications from 23 countries, the LSD Collaborative was the only UK organisation to receive an award for its ‘If You Hear Hooves it May Be a Zebra…….’ A Medical Student Empowerment Project.

The UK LSD Collaborative seeks to improve diagnosis and treatment of rare diseases
In the UK by educating and empowering the next generation of health care providers. Together, the organisations of the UK LSD Collaborative represent the Batten, Gaucher, Fabry, MPS, Niemann-Pick, Pompe and Krabbe disease patient communities.

Through the PALs project, the Collaborative will partner with 5th year medical students from Barts and the London School of Medicine and Dentistry, as well as the student-created London Society for Rare Diseases to support the development of rare disease societies across London's medical schools. Other activities include the development of a website, toolkit, and speaker's bureau.

Tanya Collin-Histed, Chief Executive of the Gauchers Association, said ‘We are delighted to have received this award. The Collaborative is a very strong group and we are recognised as a voice for LSD patients in the UK. It has enabled us to share resources and expertise amongst the groups whilst still retaining our own independence.’

Jeremy Manuel
Appointed To NICE Appraisal Committee

Former Gauchers Association Chairman and current Chairman of the European Gaucher Alliance, Jeremy Manuel has been appointed as a lay representative to the Highly Specialised Technologies Appraisal Committee of the National Institute of Health and Clinical Excellence (NICE).

In the reorganisation of the delivery of specialised services, NICE have taken on the responsibility for the assessment of treatments for highly specialised services which are likely to include the evaluation of treatments for rare conditions such as Lysosomal Storage Disorders. The appointment is for three years from 1st December 2013.

Jeremy said, ‘I am very pleased to have been appointed to this position by NICE who of course are well aware of my background in working on behalf of patients with a rare condition. I am looking forward to bringing my experience to the deliberations of the committee when it will be making decisions which will be of great importance to patients.’

Daniel Brown, Gauchers Association Chairman, said. ‘On behalf of the Board of Trustees and all of our members I would like to wish Jeremy congratulations on his appointment to the Highly Specialised Technologies Appraisal Committee for NICE. It is a testament to Jeremy’s standing within the rare disease community and the tireless work that he has put in over many years representing patients that he has been asked to undertake this role.’
Study Day in Scotland

On Thursday October 10th 2013, representatives of the LSD patient groups including the Gauchers Association’s Chief Executive Tanya Collin-Histed travelled to Scotland to attend a study day on LSDs at the Beardmore Hotel, Glasgow.

The objective of the regional meeting was to raise awareness amongst healthcare professionals who may contribute to the care of Scottish children and adults that have an LSD. The key aim was to educate non-specialist healthcare professionals working in secondary or tertiary care, about the presentation, diagnosis and long term management of LSDs with a view to increasing awareness and improving outcomes for patients and their families.

The meeting was attended by an intimate group of clinicians, company representatives and patient groups who listened to a number of short presentations on –

- Early Recognition & Diagnosis
- The Challenges of Diagnosis
- An Audit of the Diagnostic Period of Mucopolysaccharidoses (MPS) Compared to International Data
- The Practicalities of Homecare
- New Developments in Treatment and Providing Supportive Treatment

**Editor’s Note**

On the October 9th 2013, Health Secretary, Alex Neil announced that as a result of restructuring, the Scottish Medicines Consortium (SMC) needs to be ‘more flexible’ in the evaluation of new medicines. During the announcement he said ‘It is only right that Scottish patients have access to medicines that are clinically justified.’

The announcement also included the following:

- An extension of the £20 million Rare Medicines Drug Fund to 2016 • An improved patient support for engaging in the medicines approval process • An additional investment of £1 million to support SMC to make their work more transparent • The creation of a new peer approval system to allow clinicians to prescribe medicines that are not accepted for routine use by the SMC – replacing Individual Patient Treatment Requests (IPTRs) • The opportunity for the SMC to appraise new medicines which have not yet been submitted by a SMC announcement

This announcement provides patients and their families in Scotland with fairer access to treatment.
Three-year PhD Student Studies nGD

We are delighted to report success in securing an award through the Impact Studentship Scheme at University College London (UCL). The award will pay for a three-year PhD studentship working full time on developing neonatal administered gene therapy for neuropathic Gaucher disease (nGD).

In previous editions of the Gauchers News we have reported on the work of Simon Waddington and Ahad Rahim who first started looking into gene therapy in nGD in 2009 when the Gauchers Association awarded them a grant of £9,000 to purchase some viral vectors.

Ahad Rahim recently secured a UCL Excellence Fellowship and we are delighted to report that in securing this position he was able to position nGD as a top priority in his work from the very start at the School of Pharmacy. Ahad said ‘This will allow me to set the agenda within the Department but would also constitute a major long-term commitment to neuropathic Gaucher Disease research at UCL’.

The Impact Studentships Scheme being offered at UCL requires a match donation from charities and/or industry to fund PhD positions and we are delighted to report that the Gauchers Association together with the Javon Trust has been able to raise the necessary funds to support this three-year position. In addition the Association will work with Ahad and Simon to identify further funding to support annual consumable costs i.e. animal costs.

Ms Giulia Massaro, a highly motivated and committed student from Italy will take up this post in the next few months. The progress of this work will be reported regularly in future editions of Gauchers News.

Professor Ed Wraith Memorial Meeting

5th April 2014 The Point, Lancashire Cricket Club, Manchester

The Ed Wraith Memorial Meeting on Lysosomal Diseases is planned as an inclusive and educational event to bring together and educate clinicians, scientists, care providers, support groups and charities as well as affected families. It will be a half day conference, memorial and dinner, themed around the life and work of Professor Ed Wraith, who worked tirelessly and selflessly to improve the diagnosis, understanding and treatment of Lysosomal Storage Diseases. The meeting is designed to be accessible to all who knew Ed, therefore the registration fee is nominal. We urge you to book early for the best rates by visiting our website at www.gaucher.org.uk
6th nGD Family Conference

From Friday 8th to Sunday 10th November 2013, 81 patients, families, healthcare professionals, representatives from the pharmaceutical industry and Homecare providers converged on Reading in Berkshire to attend the 6th nGD Family Conference.

Over the weekend, we played host to 19 children and they were entertained by our wonderful volunteers with arts and crafts, ten-pin bowling, Laser Quest and soft play!

This conference was particularly special because it was organised in partnership with the ‘Type III Girls’ Irma, Maddie, Nadia, Saphia, Sara, Sarah, and Radhika. Irma Shah opened the meeting and Nadia Fattouki chaired the morning session with Sara Khan as the afternoon Chair.

The meeting programme was designed by the girls with a focus on personal stories. Maddie, Saphia and Radhika spoke about growing up with Gaucher’s disease and the challenges they have faced and overcome. Their messages were very positive and many a tear was shed at their honesty. Radhika’s father Kishor and Sara’s mother Sheena spoke openly about the struggle with diagnosis, feeling alone and how meeting other parents had helped them through difficult times.

A selection of other topics that were presented over the course of the weekend included –

- Genetics and Pregnancy by Sister Elizabeth Morris
- Drug Trials and Strategies for Treatment by Dr Derralynn Hughes
- How ERT works by Dr Suresh Vijay
- Gene Therapy for nGD by Simon Waddington & Ahad Rahim

The last talk of the main programme on Saturday was by Dr Ashok Vellodi who spoke passionately about his personal journey looking after children and young adults with Type III Gaucher disease.
On the Sunday whilst the children enjoyed their activities, the families had a closed session looking at topics such as the changes in the benefits system and how the provision of the statement of special educational needs system is changing.

Amongst the families that attended the meeting were three new UK families, two from Sweden, one from Bulgaria, two from Italy and two from Denmark.

Feedback from those that attended was extremely positive and the clear message from the weekend was ‘even though having Type III Gaucher disease presents many challenges to the individual and the family, with treatment, good clinical management, friendship and a good support network it is possible to achieve your goals and aspirations.

‘I just wanted to say how impressed I was by the meeting this weekend. I was so proud of all the young team with Type III Gaucher disease who took the time to organise and present at the meeting. The information given was factual and relevant. I also thought it took great bravery and tenacity to stand in front of a room full of people and relate their personal stories. Girls, you are awe-inspiring, I hope you take the praise and feedback that will be given, and strive to continue onwards with your life’s journey.’

Jean Mercer, Clinical Nurse Specialist, Manchester Children’s Hospital

‘Please could you pass on my congratulations to all the girls for doing such a tremendous job with the organising and delivery of such a superb conference. I have to say it was the best one I have ever been to. The personal stories were incredibly moving and inspiring. I hope the rest of the weekend went well and the girls celebrated their success.’

Claire Smith, Genzyme

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Gauchers NEWS
Susan Lewis Memorial Award

The Susan Lewis Memorial Award was established in 2007 to provide bursaries to doctors and other healthcare professionals from developing countries to travel to the UK Centres of Excellence to undertake mentoring and educational programmes. The remit of the Award has now been extended to support a wider range of educational initiatives and encourage the education of doctors generally in the field of Gaucher disease.

During 2013, the Association supported Dr Elena Pavlova and Dr Katherine Aitchison to attend meetings throughout Europe. Here are their reports:

Dr Elena Pavlova

I am a MD/PhD graduate and a research fellow in the Department of Medicine at the University of Cambridge, where I have been studying Gaucher disease with Professor Cox for several years.

I started my research and clinical work in Gaucher disease in the Moscow Research Centre for Children’s Health where together with Russian colleagues we looked after children with Gaucher disease. I was fortunate to meet Susan Lewis and other members of the Gauchers Association to whom I am grateful for their gracious support of my research at Cambridge.

My present study focuses on studying hematological cancers in Gaucher disease. This study aims to understand why fatal lymphoma and myeloma complicate Gaucher disease. It promises to reveal more about the general mechanisms of lymphoma.

Many thanks for the award. I participated in the Graduate Course ‘Lysosomes and Lysosomal Diseases’ which was held for graduate students and postdoctoral scientists. The course was a part of the 19th European Study Group on Lysosomal Diseases Workshop. The agenda was extremely fascinating and covered various aspects of lysosomal diseases. It was a great opportunity to hear the most recent findings and new directions in the LSD field.

It is fascinating how research into LSDs may contribute to our better understanding of fundamental biological processes and many other pathological conditions suggesting new prospective studies for research. This was a special experience for me and a wonderful chance to meet researchers with overlapping interests.

I am grateful to the Gauchers Association for granting me the Susan Lewis Memorial Award to support my attendance at this magnificent international meeting.
Dr Katherine Aitchison reports on the European Society of Gene and Cell Therapy conference:

I am a PhD student at the University College London Institute of Child Health and for the last three years I have been working on developing gene therapy for the treatment of Type I Gaucher disease. Gene therapy works in the same way as bone marrow transplants but instead of using cells from a second person, the transplant consists of the patient’s own cells which have been genetically corrected so that they contain a healthy copy of the glucocerebrosidase gene. This reduces the risk that the transplanted cells will be rejected or cause graft-versus-host diseases, as can happen with standard bone marrow transplantation. The genetic correction would be permanent as we would target the stem cells which are the foundation of the blood system and all cells which develop from these stem cells will carry the functional gene.

This year I was lucky enough to obtain a grant from the Susan Lewis Memorial Fund which allowed me to travel to Madrid for the annual European Society of Gene and Cell Therapy (ESGCT) meeting attended by almost a thousand scientists and clinicians. Over the four days I attended talks by some of the world leaders in the field of gene therapy and I heard some very encouraging data regarding clinical trials and pre-clinical work which demonstrated how far the field has come in the last few years. I was chosen to present a poster at the conference in order to communicate the work that I am involved with and I was fortunate enough to speak to members of other groups also working on gene therapy for Gaucher disease. This allowed me to see the strategies they are employing, discuss their progress and try to find solutions to problems common between the groups. In addition to Gaucher disease, several presentations focused on gene therapy for other lysosomal storage disorders (LSDs) and because of the similarities between many of the LSDs it is possible to apply what they have learnt from their work to my own project and thereby develop my own work. It was also very interesting to get an idea of the wider gene and cell therapy field and see some of the incredibly innovative technologies being developed around the world and the novel methods for treating disease which are being investigated. It’s a very exciting time to be working in the field of gene therapy and seeing the promise of targeted treatment become a reality with many successful clinical trials.

I would like to thank the trustees of the Susan Lewis Memorial Fund for giving me the chance to attend this meeting and benefit from hearing the many impressive speakers.

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. Email Sarah in the office at sarah@gaucher.org.uk or call 01453 549231.
Buckingham Palace Garden Party

In April 2013, Don Tendell took a call from Sarah in the office asking him if he’d like to go to one of the Queen’s annual Garden Parties at Buckingham Palace. This is his story:

I took a quick look in my diary and seeing the date free I made a management decision and said yes. When I told Maureen I had a thousand questions thrown at me, none of which I could answer! I am sure all readers will guess the most important question she asked – what dress should she wear? Naturally she had nothing suitable to receive the Queen so a shopping trip was hastily planned.

The household then returned to its normal state for a few weeks. On Tuesday 30 April 2013 we received an envelope postmarked Buckingham Palace and I must admit, opening the envelope was exciting. Inside was the invitation –

*The Lord Chamberlain is commanded by Her Majesty to invite Mr and Mrs Donald Tendell*  
*To a Garden Party at Buckingham Palace on Thursday 6th June 2013 from 4pm to 6pm*

With it came instructions for the day including parking, identification, no cameras, and dress code. For me there was no real problem, a choice between a grey or blue suit, both of which I have, but for Maureen (and me) the sartorial nightmare began!

At this point I wish to send my appreciation to Maureen’s friends for providing the support needed during this stressful period. For those of you who do not know us, we have been on a significant number of cruises where the dress code is formal, smart casual and casual and to my eyes the wardrobes are full of suitable ladies attire. To Maureen, the wardrobes are empty and the Garden Party is a very good reason for a new dress and hat. Fortunately we live in an area near Windsor that is familiar with Royal occasions so the local dress shops are very helpful in providing guidance as to what one should wear to such an occasion.

The day arrived and it was the most perfect summer’s day you could wish for. Maureen had found a dress and hat she liked, the car was washed and with the sat-
nav set for The Mall, off we went giving ourselves plenty of time for the London traffic. There were no problems on the journey and as we arrived in The Mall the police were on hand to guide us straight away into a parking place.

We arrived a little early so we walked to the Palace through St James Park as a number of others were doing. At the gate there was a small queue that moved quite quickly so showing our security documents we soon found ourselves walking across the parade ground into the Palace itself. Guided through the Palace and catching just glimpses of the surrounding paintings and décor, we arrived on the West Terrace overlooking the gardens.

To say the lawn and gardens are magnificent is an understatement, they are huge and in front of us on the lawn were three main tents; the Royal Tea Tent, the Diplomatic Tea Tent and the Main Tea Tent for guests such as ourselves. We walked down the terrace steps onto the lawn and as we passed the main Tea Tent we decided to take the opportunity of sitting at a table with two free chairs.

We noticed that two lines of guests formed from the Palace Garden Entrance to the Royal Tea Tent and at precisely four o’clock the bands stuck-up the national anthem and from there Her Majesty the Queen together with His Royal Highness the Duke of Edinburgh and the rest of the Royal party made their way to the Royal Tea Tent.

So the Party began. It was very informal, guests moved around the gardens freely and came from all walks of life, religious orders, diplomatic staff, military, police, civil servants, local government offices, sports and of course those representing charities. Unfortunately we did not have the opportunity to meet any of the Royal party but we did enjoy the sandwiches and cakes of which there appeared to be an endless supply. I think Maureen even had an ice cream! Throughout the afternoon the military bands played a variety of music to entertain guests.

After tea Maureen and I decided to walk around the wonderful gardens and lake. It was during this walk that we noticed the request of no cameras was not being complied with we made our way back to the main lawn as the party drew to a close and joined the other guests in the two lines that formed to applaud the Royal party on their return to the Palace. Although I managed to see Her Majesty, the Duke of Edinburgh and others, unfortunately, Maureen (at 4ft 10 ins in heels) saw nothing. We learned later that 8,000 people were in attendance but with the size of the grounds I would never have thought so.

It was now time to leave. We made our way back through the Palace, across the courtyard, through the gates and down the Mall to the car for the journey home. It was a wonderful day and experience to be remembered with perfect weather adding to the spectacle.

Maureen and I pass on our appreciation to the Gauchers Association for making this possible.
4th London to Cambridge Bike Ride

Yet again, it was another great day out, writes Alan Rosen:

I am delighted to report that our 4th Gauchers Association Charity Bike Ride from London to Cambridge was held on Sunday 8th September and was an overwhelming success. Not only did we have a record number of over 120 enthusiastic cyclists sign up for the ride all, (but one) finished and most arrived before the rain!

The cyclists were sent off just after 8am following some inspiring words by Lord Palmer of Childs Hill, a long-time supporter of the Association, who together with Lady Palmer chatted to riders before the start. The route was, as in previous years, from the playing fields of UCS School in West Hampstead finishing some 60 miles later in the grounds of Addenbrooke's Hospital Cambridge, where the riders were greeted with warm enthusiasm by Professor Cox.

It was a beautiful day for cycling and riders commented on the enjoyable route through the Hertfordshire and Cambridgeshire countryside. Of course the most pleasurable parts of the ride are the refreshment stops, all were manned by our enthusiastic and welcoming volunteers. We’d like to give special thanks to Davina Ridley for allowing us to take over her front garden and as in previous years, the team from Cambridge provided a scrumptious array of homemade cakes devoured with delight by the riders. Some said that not only were these cakes a highlight of the ride but the reason for doing it!
The feedback we received was really positive. Everyone thought it was a superb day and the organisation was excellent. Janos the mechanic who was on hand throughout the day to deal with any mechanical problems, (the odd puncture, but that was it), and the team of marshals fulfilled their roles brilliantly, ensuring the cyclists followed the route correctly. In fact all the volunteers from those managing the start, the marshals, the refreshment stops and the team at Addenbrooke's at the finish did a most magnificent job. Without these dedicated people the ride would not be possible.

New this year for those arriving in Cambridge early was the trip into the city to see the sights and return to Addenbrooke’s before starting the ride home. This little extra was much enjoyed by those who joined, even if some sightseeing was limited to the hospitality of a Cambridge hostelry!

Of course the most important part of any charity fundraising event is to raise much needed funds and we are delighted to report we raised almost £40,000. This money is vital to enable the Gauchers Association to carry out its vital work for patients.

Special thanks go to Shire and Genzyme, our industry Platinum sponsors who generously supported our ride. Thank you also to Amicus who kindly sponsored Janos, our mechanic.

We have already fixed Sunday 7th September 2014 for next year’s ride. We look forward to even more people joining us next year, the more the merrier so please start training and put the date in your diary.

In addition, we were delighted and honoured that David Brodie chose to celebrate his 60th birthday by bringing his family and friends to join the ride. David describes his day:

As my birthday approached, I thought long and hard about how to mark the event. I have loved cycling for as long as I can remember, and so I was keen to involve family...
and friends in some sort of cycle event. I had recently been diagnosed with Parkinson’s and so I also wanted to do something to tackle this disease.

Jeremy Manuel and Sarah Allard of the Gauchers Association came up with a wonderful suggestion. We could invite our guests to join the ride, with any funds they raised being earmarked for the research being conducted at the Royal Free Hospital into the link between Gaucher disease and Parkinsonism (1) which is exploring why there are ten times as many carriers of Gaucher disease among those with Parkinson’s than would be expected by chance. This work may in turn lead to new insights into Parkinson’s and, in the longer term, perhaps contribute to a cure.

And so it was that 17 of us joined the other riders at the UCS playing fields at 8am on Sunday 8th September. ‘Team Brodie’ included my wife and two children, and thirteen friends who wanted to join the fun and make a difference. Some of us had been cycling for years while others were relative novices. Some were kitted out in lycra, others in civvies. Two of them shared a tandem and were actually the first of our team to finish, spurred on no doubt by the thought of tea and buns while they waited for the rest of us. I did not mind spending longer on the road – the ride passes through some stunning countryside.

What we all shared was an amazing day, magnificently organised and marshalled, and ending with a wonderful birthday cake supplied by The Gauchers Association, which we enjoyed on the coach back to London before a welcome doze.

With the help of others who attended my birthday party the previous day, our riders raised over £7,000 which I know will be well spent. In addition, our participation encouraged another Gauchers Association supporter to ask their friends to do the same to mark a key wedding anniversary. These things have a way of snowballing, and I am glad we helped play a small part in the important work being done by a wonderful charity.

I am deeply grateful to the Gauchers Association for offering me a very special way to celebrate my 60th birthday.
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. If you would like to set up a standing order to the Association, please contact Sarah by email at sarah@gaucher.org.uk or call 01453 549231.

Donations Received Between May 2013 – October 2013 Totals £17,686

Generous donations have been received from: Charities Trust; William Brake Charitable Trust; Manuel Swaden Solicitors; Province of Hertfordshire Dyrham Park Lodge; Thomson Reuters; Mr & Mrs Golker; Mrs D Hood

In Memory

The family of Jean Minty who passed away on 20th July (see page x) kindly donated £600 in her memory.

Friends and family kindly collected £504.75 in lieu of flowers at the recent funeral of Mrs Doreen Joyce Owen of Anglesey, North Wales.

The late Miss Edna Pearson, who sadly passed away in April 2013, left a legacy of £1,000 to the Gauchers Association.

Well done to Calum Phoenix for completing the Great North Run in memory of his older brother, Liam Phoenix and raising £650.

A charity poker game raised £130 in memory of Dave Cooke.

Mrs Galton, Mrs Yates & Mrs Brenchley kindly donated £1,000 on behalf of their brother, the late Martyn Rogers in accordance with his wishes.

Mr Jeff Hammerschlag kindly donated £500 through the New West End Synagogue and their Kol Nidre Appeal.

Keith & Joy Moore very kindly collected £25 from a charity box in their local ironmongery store.

Professor Atul Mehta kindly donated £300 following on from a haematology teaching session he was involved in with The Society of Chiropodists and Podiatrists.

The Eleanor Lily Foundation very kindly donated £6,000 towards Neuronopathic research.

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

Donate by Text

You can also donate via text – Text GAUKo1 £2 / GAUKo1£5 / GAUK £10 to 70070 to donate now!

Thank you so much!
Virgin London Marathon 2014

Once again, the Association has successfully secured five Golden Bond places for the 2014 Virgin London Marathon on Sunday 13th April 2014. There will be over 34,600 runners taking part in the 33rd London Marathon passing more than 700,000 spectators along the route from Blackheath to The Mall.

This is one of our biggest and most important fundraising events of the year. Six runners completed the course in 2013 and raised nearly £14,000 through generous support of our members as well as the runners’ friends and family and we’re hoping to blow that number out of the water in 2014!

We wish our runners John Cowan, Andy McIntyre, Phil Dawson & Alison Brimacombe the very best of luck with their training over the next few months. Please read their stories below and support them as much as you can.

We still have one Golden Bond place available so if you’re up for the challenge of a lifetime and feel inspired to run, or you know someone who was unsuccessful in the ballot, please call Sarah in the office on 01453 549231 or email sarah@gaucher.org.uk.

John Cowan writes: My name is John Cowan. I am a 29 year old, slightly balding father of two from Leeds. I have decide to run this year as I have a longstanding commitment to my mother Susan who is a Gaucher sufferer to help raise funds for a worthy cause. I have been aware of the Association and the important role they play in representing and supporting Gaucher sufferers and their families for many years and thought that it was long overdue that I helped show my support in raising both funds and awareness. I was diagnosed with testicular cancer two years ago and now that I am fully recovered I am trying to invest more of my time in positive and worthwhile pursuits. The experience of training for and running in the London Marathon been a massive one and it will also help me supress the middle age spread for the time being, which can’t be a bad thing either!

Andy McIntyre writes: I live in London and work for a property development company. My friend and colleague Dan has Type 1 Gaucher disease and I get to witness on a daily basis how the right treatment
lets him live life to the full. Despite having completed a few triathlons over the years I have never run more than 10km, so this will be a real test of my endurance. I’ve heard so many amazing stories about the day itself, I can’t wait to run down the finishing straight on The Mall where my wife Caroline and son Jake will be cheering me on. Just the small matter of some serious training first!

Phil Dawson writes: I’ve been with Genzyme for just over five years and am responsible for disease awareness as well as being responsible for working with the adult Centre at Salford Royal and Paediatric Centre at St Mary’s. Gaucher is obviously a big part of my working life and hearing of the struggles patients and their families go through to get a diagnosis and then to get proper care is a huge motivation to keep going when times get tough.

I’ve been running to keep fit for a few years but 2013 was the first time I’ve ever entered a formal race – I did a 10k in April and a seven-miler in September so the full 26 miles is a huge step up! My wife recently completed the York marathon which was a big inspiration. My three girls are also hugely excited that daddy might be on telly!

Thanks to the Gauchers Association for this fantastic, but scary, opportunity and thanks to all my friends and colleagues who are going to sponsor me (hopefully!)

Alison Brimacombe writes: I have been working for Genzyme as a Clinical Field Specialist for just over two years. I help to increase disease awareness within East London, Kent and Surrey and I am thrilled to be running my first marathon for such a great cause!

Remember, we have one coveted Golden Bond place still available!

If you, a friend, colleague, neighbour, Twitter follower or Facebook friend are up for the challenge, call us on 01453 549231 or email sarah@gaucher.org.uk!
Fancy Dress Party Fundraiser

Ruth Newton describes how she wanted to celebrate a milestone birthday and remember Louis Duke with friends and family:

**This year I turned 50! How did that happen? I decided I wanted to have a fancy dress party and instead of choosing one theme like most people would, I chose three of my favourite things – giraffes, angels and musicals.**

I also decided to try to raise some money for The Gauchers Association as I worked as a teaching assistant at North Kidlington Primary School looking after Louis Duke who we lost to Gaucher in 2009. He was the cheekiest, most loving, happiest little boy I have ever met and my life was and is richer because of him. I organised to have a photo booth at my party so people could have a party memory and raise some money for Gauchers.

We set up an area for my brother-in-law to take the photos for us; everyone was more than happy to take part and we raised £150 for the Association which was great. Louis’ mum and auntie Laura came dressed as the 118 guys as myself and Jayne (Louis’ other teaching assistant) had for a school dress-up and it brought a tear to my eye.

Turning 50 was just fine and it was made even better knowing we raised money for such a worthwhile cause.
Middle East and North Africa Meeting

The Association’s Chief Executive, Tanya Collin-Histed was invited by Genzyme to give the plenary talk at their Middle East and North Africa (MENA) Lysosomal Storage Disease Forum in Dubai on 4th October. Her talk was about her personal journey from a parent to professional advocate.

The meeting brought together over 100 rare disease experts and physicians from different specialties across the Middle East & North Africa, including Tunisia, Algeria, Saudi Arabia, Jordan, India, Pakistan, Algeria and Iran.

The objectives of the meeting were to support the rare disease community in MENA with an educational event that included scientific content and to foster networking amongst this group of healthcare professionals. The target audience was geneticists, paediatricians, neurologists, neuromuscular and metabolic disease specialists, endocrinologists and haematologists.

The meeting which ran over one and a half days addressed the four rare genetic diseases (Pompe, MPS I, Gaucher and Fabry) and each session stimulated questions regarding diagnosis, clinical practice and access to treatment. Each of the presentations gave the audience the opportunity to vote on specific clinical questions to increase awareness and provide educational opportunities.

Access to Enzyme Replacement Therapy (ERT) for patients in this region is very varied, some countries are able to provide ERT through Government programmes, and in other countries some patients receive treatment through charitable access programmes i.e. Genzyme’s International Charitable Access Programme (ICAP). In some countries access is only possible for nationals of that country.

Members of the audience had previously met and are in the process of developing clinical guidelines for LSDs which they hope will be available in 2014 and support other healthcare professionals involved in treating LSD patients in MENA as well as supporting the on-going challenges we have regarding access to treatment.

The key challenges for this region are diagnostics, genetic counselling, raising awareness of these diseases and also the lack of and need for patient support.

Tanya’s presence highlighted the importance of working with patients and how patient groups can work in partnership with clinicians and the Ministry of Health to support the development of patient services and robust health infrastructures.

www.gaucher.org.uk
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Maybe you work for a company that would like to place an advert?

Please contact us by end of April 2014.

If you would like to be included in the next issue and would like further details and a booking form please contact Sarah on sarah@gaucher.org.uk or 01543 549231