

## **Directors Report 2014 to 2015**

### **Office Move**

In May 2015 the Gauchers Association and European Gaucher Alliance moved offices. The new offices provide larger office space to the organisations in Dursley.

### **Advocacy Service**

#### **a) Patient & Family Support Worker**

The Association provides a dedicated Patient & Family Support Worker that provides support and advice to meet the non-medical needs of the Gaucher community in England, Scotland and Wales. Type of support provided includes:

- Telephone Helpline
- Advice on Welfare/Disability Benefits
- Advice and support on Educational issues
- Advice on Housing and equipment
- Projects – supporting and developing projects to support families, young people and young adults with Gaucher disease.
- Employment advice, Curriculum Vitae support, work experience placements
- Attending clinic appointments with patients at their request and hospital specific Gaucher clinics
- Attending hospital Gaucher clinics to meet patients and family members and talk to them about the Association

#### ***New Patient Day***

The Association arranged a fun day at West Midlands Safari Park in 2014 for all the new families who hadn't yet met each other (as well as meeting members of the Association). Four families with newly diagnosed children attended the day which gave them the opportunity to get to know each other and make friends.

#### **b) All Ireland Advocacy Worker**

Through a partnership agreement with the Society for Mucopolysaccharide Diseases, the Gauchers Association provides our Irish members and their families an advocacy service to act as a support network, to bring about more public awareness and to promote and support research. Alison Wilson, a genetic counsellor based at Belfast Hospital in Northern Ireland, has been working as an advocacy worker for the MPS Society since 2011 and in 2013 her role was extended to cover patients and families with Gaucher disease.

### **New Staff**

The Association appointed a new administration assistant in April 2015, Claire Lightfoot will support Sarah Allard enabling her to take on more responsibility and become more involved in other Association projects.

### **National Highly Specialised LSD Service**

#### ***Gauchers Centres and National Specialised Services***

We would like to thank all of the clinicians, the Clinical Nurse Specialists and the teams at each of the centres for their continued advice and support to the Association and their dedication to Gaucher patients. The national service provides our members with the very

best treatment. The adult centres work with the paediatric centres in helping patient transition from childhood to adulthood.

### ***LSD Expert Advisory Group (EAG)***

As patient representative, Tanya Collin-Histed sits on the EAG and attends bi-annual meetings with representatives from each of the LSD centres in England (and in Wales as an observer) and the NHS England Commissioning staff to discuss clinical management, guidelines, research, treatment options and other issues associated with the National Highly Specialised LSD Service.

In addition, along with other members of the UK LSD Patient Collaboration Tanya Collin-Histed attends meetings at the LSD Centres to discuss service provision and patient care for LSD patients and their families.

### ***Developing Clinical Guidelines***

Tanya Collin-Histed was invited to join the working group that was charged with developing a protocol for the use of high cost drugs in the treatment of Gaucher. This working group is being established as part of the Metabolic Disorders Clinical Reference Group (CRG) Quality, Innovation, Productivity and Prevention (QIPP) programme for 2015/16 looking at the use of high cost drugs in the treatment of Lysosomal Storage Disorders (LSD) and Metabolic diseases. The aim of the programme and working groups is to ensure that all high cost drugs treatments for Metabolic and LSDs are used in the most clinically and cost effective way.

### **National Homecare Framework for LSDs Service**

As the patient representative for the UK LSD patient Collaborative, Tanya Collin-Histed sits on the national project team for the development of a national homecare tender for the 8 Lysosomal Storage Disorders (LSDs) specialised centres in England.

The existing homecare contract which commenced on 1st October 2013 came to an end in September 2015. Tanya attends the six monthly homecare review meetings with all the providers on behalf of the LSD Patient Collaborative and for the last 18 months has been a member of the taskforce to draw up the new Homecare Contract which commenced on 1<sup>st</sup> October 2015.

### **Wales**

Tanya Collin-Histed, the Association's Chief Executive is a member of the Welsh Governments Clinical Evidence Evaluation Group (CEEG) for rare Diseases in Wales. The group meets quarterly and advises on how treatments for rare diseases are commissioned and managed in Wales.

### **Gaucher disease in Europe and other parts of the World**

The Gauchers Association is one of the founding members of the European Gaucher Alliance (EGA), a collaboration of patient representative groups around Europe and the World which now has 42 members (representing 41 countries). Tanya Collin Histed was appointed as the Chief Executive Officer of the EGA in June 2014 and Jeremy Manuel, one of the Association's directors, is the Chairman of the EGA. The EGA is operated on a day to

day basis from the Association's headquarters in the UK. Tanya Collin-Histed splits her 37.5 hour working week between the two organisations.

### **Neuronopathic Gaucher Disease Type 3**

#### ***Developing Patient Related Outcomes (PROs)***

The Association have been working closely alongside Genzyme to develop a study to interview Type III young adults with Gaucher disease in the UK to learn which symptoms of the disease are the most common, and how these symptoms typically affect the lives of patients who live with the disease. It is hoped that by having a better understanding this may help improve how the patient's experience is accounted for in clinical trials that measure the effect of disease treatments. We hope that it also may help patients and their families by highlighting the consequences of the disease that most need attention, possibly leading to new programmes and support services.

#### ***Quantifying the Burden of Disease for Patients & Families with nGD***

Tanya Collin-Histed, attended a small patient advisory board meeting in Boston, USA hosted by Genzyme Sanofi to discuss the burden of disease for patients and their families with Type III Gaucher Disease, Tanya was one of the 4 patient representatives at the meeting who were specifically invited to the meeting because of their personal and professional experiences with Type III.

Each of the patient advocates told their personal story and spoke about their work in Type III and the challenges and issues that face the Gaucher community. This meeting was organised by Genzyme to support their commitment to further understand the challenges of Type III Gaucher patients and their families and explore how they could possibly demonstrate a functionality that improved and had a meaning to the patient when planning a clinical trial

#### ***Genzyme GD III Clinical Trial***

Tanya Collin-Histed has been working with Genzyme Global to understand their approach and provide encouragement and guidance on a clinical trial that will seek to address the unmet needs of Type III for Gaucher patients.

#### ***Wearable Technology***

In partnership with Dr Elin Haf Davies and Dr Aimee Donald, Paediatric Clinical Research Fellow and the Gaucherite Consortium, the Association have raised the funds through unrestricted grants from Shire and Genzyme to conduct a pilot study for the use of wearable technology and mobile phone applications to assess the impact of nGD on daily living. This study will help to learn more about the condition, to encourage more research into these problems and to develop management and interventions to support patients.

#### ***Type 2 booklet***

A new Type II Gaucher disease booklet was produced by Helen Whitehead, the Association's Patient & Family Support Worker and Dr Anupam Chakrapani, a Paediatric

Consultant at Great Ormond Street Hospital. The booklet is available as a hard copy and also a PDF.

### **Research & Development training**

EUPATI, European Patients' Academy on Therapeutics Innovation launched its first 15 month course involving e-learning and face-to-face events. Tanya Collin-Histed was one of just 50 to be selected to undertake this course in Europe, upon completion; she will have the knowledge to make meaningful contributions to patient empowerment and advocacy and to contribute to the broader dialogue on patient involvement in medicines research and development across Europe. The course runs from October 2014 to December 2015 and is a commitment of at least 250 hours of e-learning study and 8-10 days (including travel) to attend face-to face training sessions with the experts at EUPATI.

***Empowering Young Type III GD Patients to Shape the Future'***: 2014 saw the second year of our programme funded through the global patient advocacy leadership award (PALs).

Achievements of the projects from 1<sup>st</sup> July 2014 – 30<sup>th</sup> June 2015 have been;

#### ***Facebook***

Helen Whitehead set up a Facebook profile for work and a private group for the type III young adult group. This has been a success and has improved communication between the Association and our members

#### ***Type 3 Group meetings***

During this period the Type III young adults met at the Royal Free Hospital on four occasions to discuss various projects, such as;

- The 7<sup>th</sup> nGD family Conference to be held in November 2015
- Dr Derralynn Hughes to discuss the purpose of research to educate the young adult for participation in future possible trials/studies
- The buddying project

All of these meetings were held on a week end and attended by Dr Derralynn Hughes and Clinical Nurse Specialist Niamh Finnegan from the Royal Free Hospital, London

#### ***New Funding to continue the Empowerment project***

The remaining funding for this project from the original PALs grant will see the project through to the end of January 2015, further funding will then be required to sustain this work. Helen Whitehead, Patient & Family Support Worker (PFSW) has taken over the day to day management of this project with the girl and is working with Great Ormond Street Hospital to apply for future funding.

#### ***Overseas Visits and Ambassador Project***

In January 2015 Irma Shah travelled to India with CEO Tanya Collin-Histed, Clinical Nurse Specialist Niamh Finnegan and Drs. Ashok Vellodi and Alex Broomfield to clinics in Chennai and Delhi. As part of this visit Irma recorded her experiences on a video diary that can then

be used for future projects i.e. funding applications, upload onto our website, and share with the other Type III young adults.

In May 2015 Irma Shah and Nadia Fattouki travelled to Jordan with Dr Ashok Vellodi and Helen Whitehead to visit a clinic in Amman and to meet untreated patients and their families.

In July 2015, Irma Shah, one of the young nGD adults went with Dan Brown, the Association's Chairman and one of our young Type 1 patients to Berlin to participate in Shire's Ambassador Programme. This initiative is about Gaucher patients telling their personal story to inspire others and improve awareness of the challenges of living with the condition.

Helen Whitehead supported Irma Shah in attending a Shire event in Prague in July, where Irma met young doctors and also other Gaucher patients from Italy and presented her personal story.

Elin haf Davies, an independent consultant who has a background in nGD and works closely with the Association asked the Association to contribute to a book she is helping to write about the Empowerment project to highlight the importance of involving children in health and research.

A poster of the Empowerment Project was accepted for the WORLD conference in Florida, United States in February 2015, the Association and the young adults were named as co-authors and this will form part of their portfolios.

### **Fundraising & Awareness**

The Association held its fifth charity cycle ride in September 2013. The 60 mile cycle was from London to Cambridge linking two of the original Gaucher centres. The cycle ride was coordinated by Sarah Allard, the Association's Charity and Information Officer, with support from Alan Rosen, Jeremy Manuel, Liz Manuel and a group of their friends who all gave their time for free. The event attracted more than 100 riders and raised over £35,000 for the Association.

The Association continues to use the 'Just Giving' and 'Virgin Money' sites, which enables members and supporters to donate funds online to the Association. Members are encouraged and supported to use this method for sponsorship for all fundraising activities including the London Marathon, annual subscriptions, the bike ride and individual member activities and is proving very successful.

The Association had five runners in the 2015 London marathon raising over £12,000 for the charity and for the second year we have secured three golden bond places in 'Ride London', a 100 mile bike ride challenge. In 2015 we have secured 5 places in the Great North Run and 5 places in the London 10K.

2015 saw the 5<sup>th</sup> Charity Golf day since 2005. This event was organised by the Association's Director Alan Rosen and his wife Sharon and in 2015 raised £5,500 for the Association.

Other member fundraising activities included; half marathons, Ironman, 101's, in memory, birthday celebrations, line dancing, mini marathons, golf competitions and donations from local organisations.

The Association is extremely grateful to all its members who continue to support the charity.

### **UK LSD Patient Collaborative Group**

The patient organisations for those affected by LSDs work collaboratively within the umbrella of the 'LSD Patient Organisation Collaborative' to work and lobby on behalf of LSD patients and their families in the UK. The group is made up of representatives from the Association of Glycogen Storage Disease, Batten Disease Family Association, the Gauchers Association, the Society for Mucopolysaccharide Diseases, the Niemann-Pick Disease Group UK, Save Babies (Krabbe) and CATS (Tay Sachs). As part of this collaboration, the Gauchers Association has undertaken to jointly promote and share understanding of their diseases to advance standards of care and to enhance the well-being of those affected.

The LSD Collaborative is now a recognised forum and provides the expert patient perspective to NHS England in respect of procurement of Enzyme Replacement Therapy and Home Care and the approval of new therapies coming to the market.

### ***Students for Rare Disease - If you hear hooves it may be a zebra.....***

Following the success for the Collaborative in 2013 when this project was awarded a global Patient Advocacy Leadership Award (PALs) by Genzyme, the project which is a partnership between a group of Medical Students at Barts and The London School of Medicine and Dentistry and the LSD Collaborative, seeks to educate future doctors about Rare Diseases, encouraging them to think outside the box and shortening the path to diagnosis. This in turn will enable the patient to be given appropriate clinical care at the earliest opportunity.

In October the Students 4 Rare Diseases won the 'Best Poster' at the International Society for Neonatal Screening (ISNS) Conference in Birmingham and funds were secured from Genzyme Sanofi, Shire Pharmaceuticals, Actelion Pharmaceuticals and Biomarin to launch a Rare Disease Society for medical students in Northern Ireland.

### **Mental Health**

Following on from the roundtable meeting that took place in March 2014 with 30 healthcare professionals representing all eight LSD centres in England and Wales, the LSD Collaborative have been awarded a grant from Genzyme, a Sanofi Company to take forward a number of the meeting outcomes. The grant will be used to fund a one year project to work with a group of young adults with Type III Gaucher disease and a number of other LSDs patients with Mucopolysaccharide Diseases I, II, IVA and VI to identify the emotional health and wellbeing issues they have experienced. This will be used to work with families of younger patients to overcome stigma and possible reluctance to seek help.

### **Research**

The Association has given a number of small grants to several research projects over the past few years. These projects have carried out important research and gone on to use the results to lever additional funds or identify further areas of research that need to be taken forward. In addition to giving grants, members of the Association support others as a

stakeholder in collaborative grant applications through time and links with patients and their families.

Through our Board member Jeremy Manuel the Association continues to be a partner, along with the eight specialist LSD centres in the UK in the '*Gaucherite*' project.

The project will examine at least 85 per cent of all UK Gaucher patients and 'stratify' them by the nature of their disease to allow therapy interventions to be targeted more effectively. They will also work closely with major industrial partners and The Gauchers Association.

### ***Three Year PhD Impact Studentship***

In 2014 the Gauchers Association in partnership with Ahad Rahim from University College London (UCL) successfully applied for an Impact Studentship award to continue the work that Ahad Rahim and Simon Waddington have been carrying out on Gene Therapy for Neuronopathic Gaucher Disease (nGD). The application is for a three year PhD post. The Association in partnership with the Javon Trust awarded a grant of £32,535 for the project, with the remainder of the funding being provided by UCL. The PhD studentship was awarded to Miss Giulia Massaro, who recently joined Dr. Rahim's laboratory and will be working in close collaboration with Dr. Waddington's group. This studentship will begin to investigate the viral vector administered to the mice via a minimally invasive intravenous route with two fundamental questions requiring answers:

1. Is this minimally invasive route of administration as effective in extending the lifespan of these mice and is the brain rescued from the lethal neurodegeneration?
2. Given the systemic nature of delivery, does this also address the visceral symptoms?

### ***Orphan Drug Application for Gene Therapy for the treatment of Gaucher Disease***

Following the positive results of the work of Simon Waddington and Ahad Rahim on the Intracerebral injections of nGD mice, the Association in partnership with Simon Waddington and Ahad Rahim and with the assistance of Dr Elin-Haf Davies are in the process of putting together an application for Orphan Drug Designation for Gene Therapy for the Treatment of Gaucher Disease. On February 12<sup>th</sup> 2015 the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA) granted the Association, as the sponsor, a positive opinion for orphan designation (EMA/OD/303/14) for adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene (Gene Therapy) as a new potential treatment for Gaucher disease.

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

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

The Association continues to support Andres Klein, a PhD student at Professor Tony Futerman's laboratory at the Weizmann Institute of Science in Israel. The Association are providing a second year's support to Andres to complete his PhD. Andres is undertaking a

two year PhD study into “Identification of novel pathways that modify severity and neuronal degeneration of Gaucher disease”. This funding ended in December 2014.

### ***RAPSIDO - Gaucher’s families helping find a cure for Parkinson’s***

The Association continues to support the research being undertaken at the Royal Free Hospital into the link between Gaucher disease and Parkinson’s by encouraging patients and their families to support the study through our newsletters and also direct mailings. Tanya Collin-Histed, Dan Brown and Jeremy Manuel attended a meeting with the project team from the Royal Free to advise on the patient information leaflet and how best to engage with patients and their families. Tanya Collin-Histed accompanied the project team to meet with the ethics committee to approve this project in London in June 2015 and is working closely with the patient involvement officer on the role out of this project in early 2016.

### ***Parkinson’s Study at Addenbrooke’s Hospital, Cambridge***

This study is designed to better understand the association between Gaucher disease and Parkinson’s disease. The study is interested in talking to family members of people with Gaucher and/or Parkinson’s disease patients to better understand why the two diseases sometimes occur together in families. By exploring these two disorders closely it is hoped to get a better understanding of how Parkinson’s might be caused. The Association has been involved in providing guidance on the patient information literature.

The Association publishes updates on all research funded by the Association and also other research projects undertaken in our national centres to support recruitment of patients and carriers in the Gauchers News and through our website and e-mail correspondence.

### **Susan Lewis Memorial Fund**

Susan Lewis was a founder of both the UK Gauchers Association and the EGA. Following her death in 2007 the Gauchers Association established the Susan Lewis Memorial Award to provide grants and bursaries to doctors and other healthcare professionals from developing countries (particularly Eastern Europe) to allow them to travel to the UK Centres of Excellence to undertake mentoring and educational programmes in the treatment and management of Gaucher Disease.

A number of awards were made in 2014 and 2015, these included;

Through the fund the Association sponsored Prof Timothy Cox to travel to Macedonia to attend the Macedonia Academy of Sciences and Arts (MASA) Conference on Rare Diseases on 15<sup>th</sup> November 2014; Dr Ashok Vellodi to travel to Jordan and, four leading scientists in the area of lysosomes and lysosome-related organelles (LROs) and their defects to attend an international conference.

All recipients of a Susan Lewis Memorial Award are required to write a piece for our bi annual newsletter to inform our members why they attended the meeting, what they learnt and how that can be translated back in the UK for patients and their families with Gaucher disease and other lysosomal storage disorders.



## **Alan Gordon Memorial Award**

The Alan Gordon Memorial Award is awarded every two years in recognition of a person(s) who has made a significant commitment/contribution to Gaucher Disease, its treatment and Gaucher patients and their families.

In February 2015, the AGM committee awarded the eighth Alan Gordon Memorial Award to Dr Ashok Vellodi from Great Ormond Street Hospital (GOSH) in recognition of his significant contribution to Gaucher patients and their families for over 20 years. The award was presented to Dr Vellodi at his retirement party attended by many of his patients and family members in March 2015 in London.

## **Treatment**

Gaucher patients continue to receive treatment paid for by the National Health Service. The cost of treatment remains high and with the development of treatments for other Lysosomal Storage Disorders we remain vigilant that all patients continue to receive the necessary drug as prescribed to them (see National Highly Specialised LSD Service Report)

## **New Treatments for Gaucher disease**

Genzyme's small molecule, Eliglustat for Type I adult patients received European marketing approval by the European Medicines Agency (EMA) in February 2015. The National Institute for Care and Excellence (NICE) will undertake a formal appraisal of the new treatment under the new NICE highly Specialised Technology appraisal process. The Association was involved in the initial scoping exercise of this treatment in January 2014 and undertook an in depth patient survey to support our patient evidence for the NICE appraisal which was due to take place in February 2015. However in late October 2014 Genzyme made the decision to put the appraisal on hold whilst they continue to develop their pricing model for Eliglustat in the UK which will ensure that patients are able to be prescribed a treatment based on their clinical needs rather than based on cost.

The timeline to restart the NICE appraisal process is not yet known however the Association will liaise closely with Genzyme and NICE and will communicate any updates through our website and Facebook page.

Genzyme's small molecule for Type III Gaucher disease – see report above

Gene Therapy for Gaucher disease – see report above

Velaglucerase alfa (marketed under the brand name VPRIV® by Shire) was approved as a recommended treatment for Gaucher disease for use within NHS Wales and has been routinely available in the NHS across Wales since the end of 2014. Prescription will be subject to clinical assessment in conjunction with patients on a case-by-case basis. The Gauchers Association supported this process by submitting a patient submission paper outlining the value of the treatment to the Gaucher community.

The Association maintains regular contact with all pharmaceutical companies involved with Gaucher disease and researchers to ensure that members are kept up to date with all developments and we relay to the companies the concerns and expectations of members.

## **Managing Information and Communication**

The Association continues to supply up-to-date information on the telephone, by mail and through our website. Our six monthly newsletter is sent out to over 1,300 families, doctors and other interested parties worldwide, and our extensive website is read all over the world.

### ***Shire Expert Meeting***

At Shire's spring expert summit in April 2015, Gauchers Association Chief Executive Tanya Collin-Histed was invited to make a presentation highlighting the Association's work in improving the quality of life of its members and Gaucher patient and families in the UK. At the summit an entire session was dedicated to highlighting the 'Quality of Life' of patients. The inclusion of this topic is often talked about but very little time is given to discuss these very important aspects at these professional meetings.

### ***International Gaucher Day***

The UK Gauchers Association joined many Associations' around the world to celebrate the first International Gaucher Day (IGD) on Saturday 26th July 2014. IGD is the brainchild of the European Gaucher Alliance (EGA) the aim of IGD is to raise international awareness of Gaucher disease and the slogan for the day is 'Rare but Not Alone.'

Here in the UK we asked our members to celebrate IGD by organising or hosting a coffee morning at home or at work with friends, family and colleagues. To help our members celebrate this very special day we also sent them a Bag for Life, goodies branded with the new IGD logo, stickers to give to friends and family and a poster to display at home or at work to help us to raise awareness of Gaucher disease. Our members raised £1,700 for the Association on the day

### **Database**

The Association's database that was installed in September 2013 is continuously updated has been updated through member's mail outs.

### **Website**

The Association's site is updated regularly. The site allows regular e-mail alerts to our members to ensure they are kept up to date with the latest news in the Gaucher field. Our online newsletter is now sent to over 450 people worldwide.

### **Facebook**

The Association posts news every Friday on new projects, research updates, current political issues etc. and has 247 followers.