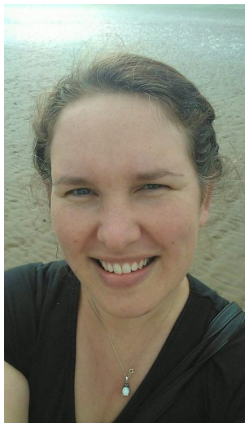
July 2018

Advocacy - Patient & Family Support Worker



Helen Whitehead

You have been contacting me with a range of different support needs; benefits are still high up.

I have now attended 4 court tribunals and all have been successful getting what they are entitled to.

Please do get in touch if you would like support through the process and the earlier you get in touch the better.

I have also supported people with school issues, attended clinics and helped get specialist equipment

I am busy planning our Family Fun Weekend on 6th/7th October 2018; including a visit to West Midlands Safari Park and an overnight stay at a nearby hotel. This will be open to **all** families with a child with Gaucher disease and booking forms will be sent out soon.

Find us on:

- FACEBOOK
- TWITTER
- GAUCHER.ORG.UK

IT'S TIME TO RENEW YOUR MEMBERSHIP!

If you have any unmet support needs, do get in touch; either call me at the Gauchers Association on 01453 549231 or 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 your home or a coffee shop.

Meeting the needs of our older community

At the start of July I will be undertaking a year-long research project looking into the needs of people with Gaucher disease aged 50 and over. I will be doing this along side my Patient and Family Support role. If you are aged 50 plus expect to hear from me about this over the coming months.

Travel Insurance

With Summer here you may be planning a trip away. Have you thought about travel insurance? If not please look at our travel insurance guide, available to download from our website or email me for a hard copy. We also have information sheets about different benefits, disabled students allowance and help with health costs; again these are all available from our website at

www.gaucher.org.uk

London to Cambridge Cycle Ride 2018
Sunday 16th September

London to Cambridge 60 mile charity bike ride is back we would love to see you there!

Full details and online registration on our website at www.gaucher.org.uk

Update on Rapsodi

The Rapsodi study started in 2012 and is a pioneering study that looks to find new ways of diagnosing Parkinson's earlier to develop life changing treatments. The GBA gene, which is carried by people with Gaucher disease and has been providing some unique insights into why people might develop Parkinson's disease. Please read their latest update:

A thank you from us

We wanted to reach out firstly to all our participants and thank you for taking the time to participate in the Rapsodi GD study.

Whether you've been involved in the study from the beginning, or have recently registered to take part, we hope you will continue to keep in touch with the study team over the coming months.

Also, don't forget to mention the study to your relatives—the more family member we can involve in the study the better!

'*Thank you for taking part, every bit of information gets us closer to understanding how to prevent Parkinson's developing and progressing*' - **Professor Anthony Schapira**

Our findings so far

The Rapsodi GD study has now been ongoing in various forms since 2012. We previously published the results of the study in two papers (2012 & 2015).

These publications provided a major contribution to our current understanding of the relationship between Parkinson's disease and mutations in the GBA gene.

We are also writing a paper that will be submitted within the next year, so we are hoping to reach our ambitious recruitment target before then.

Ambroxol Trial

Thanks to the data we have collected so far, we were able to design a clinical trial on a drug called Ambroxol, an active ingredient in many over-the-counter medications that can be used to treat respiratory disease.

Recent studies have indicated that Ambroxol may help to reduce the build up of alpha-synuclein, a toxic protein which accumulates in neurons and is a defining aspect of Parkinson's.

The trial recently closed and we hope to show that Ambroxol can delay the development of Parkinson's disease in people that carry the GBA gene.

The Future of Rapsodi

We will soon be working in partnership with Addenbrooke's hospital in Cambridge to expand the study even further, and hope to be working with other UK centres very soon

How you can involve your relatives

If you think anyone in your family might be able to spare some time to take part, we would be delighted to involve them in the study.

Please contact Soraya (s.rahall@ucl.ac.uk) or Marco (m.toffoli@ucl.ac.uk) if you think any of your relatives would like to take part in the study.

We can also be reached by telephone on 020 77940 500 (extension 31588)

Clinical Trials update Sanofi Genzyme

Larisa Petrakova-Stone, from Sanofi Genzyme has provided the latest information on their two clinical trials; **LEAP** and **ELIKIDS**:

LEAP - Adult patients with Gaucher disease type 3; (Venglustat) in combination with Cerezyme

This is a global study; with sites in the UK being **The Royal Free Hospital London, Salford Royal Hospital Manchester** and **Addenbrooke's Cambridge.**

The approximate completion date for this study is 2021 with a global estimated enrolment of 10 participants. Current global recruitment has been significantly delayed compared to the original timelines but are ongoing to meet the global market.

Part 1 of the primary objective of the study is to evaluate the central nervous system (CNS) biomarkers in adult Gaucher disease type 3 (GD3) patients that distinguish GD3 from Gaucher disease type 1 (GD1). Also, to screen adult GD3 patients who qualify for treatment with venglustat.

Part 2 of the primary objective is to evaluate the safety and tolerability of venglustat in adult GD3 patients, and also to evaluate the change in cerebrospinal fluid (CSF) central nervous system (CNS) biomarkers in this population.

The total duration in part 1 for GD1 patients is 45 days, while GD3 patients, the total duration is up to 168 weeks, including 2 part-treatment of 52 weeks and 104 weeks respectively.

ELIKIDS - Safety and efficacy of Eliglustat with or without Imiglucerase in paediatric patients with Gaucher disease (GD) type 1 and type 3

This is a global clinical trial that is now open and enrolment planned; the UK expects to open 3 study sites (**London, Birmingham** and **Manchester**) later this year and recruitment in the UK is anticipated to start in 2019.

There are two drugs being investigated in the population; Eliglustat (GZ385660) and Imiglucerase (GZ437843) and the approximate study completion date is 2023.

Recruitment will initially be to Cohort 1, which will be GD1 and GD3 patients at pre-specified therapeutic goals. Patients in Cohort 1 will receive eliglustat alone. The study will recruit later to Cohort 2 (GD! and GD3 patients with persistent disease despite adequite / optimal ERT for at least 3 years), these patients will receive eliglustat in addition to imigluerase.

The primary objective of this study is to evaluate the safety of eliglustat in paediatric patients (2-18 years old)

The secondary objective is to evaluate the efficacy of eliglustat and quality of life in paediatric patients (2-18 years old).

Further details including recruitment can be found at the clinical trials.gov website: https://www.clinicaltrials.gov

International — Botswana event - FYMCA Medical 1-3 June 2018

Following on from our announcement in April that Chief Executive, ***Tanya Collin-Histed*** will be moving to a full-time role for the EGA, she attended and presented at this meeting; please read her report below:

'I was invited to be part of the faculty for a meeting in Botswana of doctors from all over Africa; 36 doctors attended from 14 different countries, including: Ghana, Nigeria, Sudan, Botswana, Mauritius, Ethiopia, Kenya, Malawi and Lesotho.

The model for this meeting was to educate doctors on rare metabolic conditions, looking at how to diagnose and treat patients with what is available in their country.

Cont'd

International — Botswana event - FYMCA Medical 1-3 June 2018

The meeting was videoed and will be uploaded to a platform where all the delegates can access. There will also be an exam, and after a year, a second meeting will be held to focus on specific issues that have been highlighted by the delegates, i.e. diet, pain relief, psychology.

This meeting was organised by FYMCA Medical, a for-profit organisation with a focus on improving rare disease education and services for doctors, patients and patient organisations in the developing world.

Throughout the three-day meeting, the doctors attending highlighted previous patients that they are now thinking may have a metabolic disorder.

I made a presentation at the meeting about the role and value of patient organisations and spoke about the role of a national patient organisation and also the work of the EGA.

Prior to the meeting, a pre-meeting questionnaire was sent out to everyone and I had the opportunity to include a few questions regarding patients, patient organisations, diagnostics and registries, etc. which will help the EGA to better understand the situation in these African countries.

I will now write to all the delegates who attended the meeting to identify potential African patients/family members that could attend the patient huddle at the September's

RAREX meeting in Johannesburg where we will support 27 participants through a grant from Care Beyond Diagnosis, a US non-profit organisation the EGA is working closely with.'

Dan Harding

Members Fundraising

Congratulations to **Dan Harding** on completing a10K run on 10th June 2018; Dan very kindly stepped into the 'running trainers' of wife **Angela** who had signed up for this event to run in memory of her dad who had Gaucher disease and would have been celebrating his birthday. Collectively they have raised **£250**

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. Please do get in touch!

Thank you to everybody who has registered to support us when shopping with Amazon. This is very easy to do if you haven't registered, just visit www.smile.amazon.co.uk when shopping online, choose to support The Gauchers Association and for every purchase you make Amazon will make a donation to us without costing you a penny! Please support us if you can

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