



April 2018

## In memory of Pauline Simpkins



Pauline Simpkins

**Sadly Pauline passed away on 21st December 2017, her family have kindly written a few words in her memory below:**

*Pauline Simpkins, our wonderful Mum who sadly passed away on 21 December would have been so happy that so much money was collected and donated to the Gauchers Association in her memory.*

*Pauline was married to Don for 65 years and was Mum to 5 girls, 3 of which have Gaucher disease. She was also Grand Mum of 9 and Great Grand Mum of 10 and was truly loved by them all.*

*Pauline and her 3 daughters were at the very first meeting in 1990 and were*

*absolutely overwhelmed to meet so many other people who suffered or had relatives who suffered from Gaucher disease. She was eternally grateful that her girls were at last going to be looked after by a team of experts at Addenbrooke's hospital.*

*Our Mum used to love coming along to the monthly executive meetings and the highlight for her was the Anniversary event on the steam boat on the Thames and the opening of Tower Bridge to allow the boat through. She never stopped talking about this wonderful evening.*

*Pauline is sadly and fondly missed by all of her family and friends.*

The Gauchers Association would like to express their thanks to the family for writing this and for the donation of £397 kindly donated in memory of Pauline

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## General Data Protection Regulation

**GDPR** - By now you should have received our information and forms in the post. Please do send your completed consent forms to us to enable us to continue to send you all the latest information. Without your **explicit** consent we will be unable to contact you after **May 2018** and your personal details will be deleted from our database.

If you have not received our information in the post please contact us on [admin@gaucher.org.uk](mailto:admin@gaucher.org.uk) or download direct from our website [www.gaucher.org.uk](http://www.gaucher.org.uk)

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## Update on GAUCHERITE Project



Professor Timothy  
Cox

We kindly thank **Professor Timothy Cox** from University of Cambridge who has written an update on the GAUCHERITE project.

THE NATIONAL GAUCHERITE PROJECT (Gaucher disease Investigative Therapy Evaluation) is an ongoing wide-scale observational 'cohort' study of Gaucher disease in the UK. This project has been principally funded as the first and, so far, only genetic disorder included in the Medical Research Council's burgeoning Stratified Medicine research portfolio.

In the late summer of 2017, we appointed the services of a Biostatistician (Dr Khadijeh Taiyari) for one year. Dr Taiyari is working closely with the research team on the analysis of the massive amount of clinical, laboratory and radiological data the team have collected over the past three years. This analysis is at the heart of our main aim: that is, to obtain sufficient information to find the best treatment for all patients, according to their particular characteristics.

These are exciting times for us and already there are strong indications that through this project we will gain unforeseen and clinically important insights into Gaucher disease; GAUCHERITE will also serve as a permanent platform on which future clinical research projects can be based. There is a massive amount of information categorized and we are only now starting to investigate it. For those who follow the world of informatics: several petabytes! We are exceptionally fortunate that Dr CY Tan, Consultant Metabolic Physician in Cambridge, has such a deep knowledge of these matters; Chong Yew is effectively curating and managing the Gaucherite database, to which he brings immense dedication.

One heartening and immediate benefit of the work has been the recruitment of outstanding young people to work on Gaucher disease independent of financial support from industrial sponsors. Naturally, it is invidious to mention names, but of note, we have three very brilliant and utterly dedicated young women who have been crucial to our success, and who will undoubtedly 'make a difference' in the field: Simona D'Amore and Aimée Donald (the two clinical research fellows respectively working on the bone and the neurological aspects) have already made important discoveries and observations and both will be working on Gaucher disease (amongst other matters) for years to come. The other person, Sooyoung Rogers (née Lee), our Data Manager, has done absolutely first-rate work and will be continuing beyond her original contract until the end of February to complete the 'data cleaning' and other aspects – including a study on those patients who are no longer with us and who were treated too late in their disease to secure all the benefits needed to restore their health. While such information is thankfully increasingly difficult to obtain (and could easily be lost as the NHS goes digital), these data are of immense importance once and for all to capture - and to analyse in depth. As an aside, we are all delighted that not only did Sooyoung get married during this period but she was able to re-enter the University of Nottingham Medical School as a clinical student with just a couple of years to go before she qualifies as a doctor.

Overall, the recruitment process represents a huge achievement: it required an astonishing commitment by all who enrolled and helped us – especially the UK Gauchers' Association. This support, your support, has allowed us to meet key milestones, including the enrolment of 250 patients – a very tough call indeed! We are astonished by the interest and personal commitment by so many people to enrolment – despite the many burdensome consequences of so doing! Patients have willingly signed up, and more than that, so many continue to complete the study documents. All of the eight specialist centres have enthusiastically participated and engaged at every level.

As we approached our recruitment milestone, we have, for now at least, closed recruitment to the study. However, we are happy to report that the Ethics Committee have recently approved our request to extend the study for a further 2 years.

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## GAUCHERITE continued

In the background to all this activity, since our last update, the Medical Research Council demanded that we develop a Data Management plan, which will help with the future proofing of this study. This, we completed, under almost unsustainable pressures, together with our Data Access Policy. The documents were promptly accepted by the Council. Such a policy agreement is essential for us to establish the ground rules for sharing the data with approved investigators in the public domain and support legitimate collaborative work generally in the long term. With the benefit of hindsight, they also put us in a safer position in directing what research might be considered useful or justified at least from the patients who have provided this data.

We are always encouraged by enquiries from patients about the outcomes of this study: here we are pleased to report that information for public release will be released by the end of 2018 on the outcomes of phase one of this study.

Finally, apart from the Gaucher Community itself in the UK, we would like to thank Mr Jeremy Manuel, Tanya Collin-Histed and the officers of The Gauchers Association, for their immense contribution and constant support as well as advice.

Tim Cox, Kathy Page

(on behalf of all investigators in the UK GAUCHERITE programme – Royal Free Hospital, London; Royal Manchester Children's Hospital; Salford Hospital, Manchester; Great Ormond Hospital for Children, London; Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery, London; Queen Elizabeth Hospital, Birmingham, Children's Hospital, Birmingham, Addenbrooke's Hospital, University of Cambridge, University of Oxford).

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## Rapsodi new release



Rapsodi is a pioneering study which uses an online platform to find new ways of diagnosing Parkinson's earlier and to develop life changing treatments.

The GBA gene, which is carried by people with Gaucher disease, has been providing some unique insights into why people might develop Parkinson's disease. Research has shown that we may be able to intervene many years before people develop Parkinson's and can therefore stop it in its tracks.

In 2017, we halted recruitment to refurbish the online portal and in January we launched it with a brand new look!

The study is based at the Institute of Neurology of UCL, one of the leading institutions in neurological research worldwide

If you have Gaucher Disease, Parkinson disease or if you are a relative of a person that fits in one of these two categories you can take part in the study

All the study is done on your PC, so you can help our research comfortably from home. It takes only one hour!

A team of researchers are ready to answer all your questions and can help you with any issues at any time

If you want to find out more and to take part in the study visit [rapsodistudy.com](http://rapsodistudy.com)

The RAPSODI team

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## Members Fundraising



This is one of our biggest and most important fundraising events of the year. Five runners completed the course in 2017 and raised over £14,700 through generous support of our members as well as the runners friends and family.

We wish our runners **Paul Gillard; Chris Frost (Frosty); Lucy Fairall; Matias Ricciardi** and **Matthew Furniss** the very best of luck with their training over the next few weeks in the build up to this iconic 26.2 mile course!

## London to Cambridge Cycle Ride 2018 Sunday 16th September

**London to Cambridge charity bike ride is back!!** Why not grease up your chain and join us for this fun day out of fundraising, raising awareness and keeping fit! If taking part is not your thing then why not get involved on the day by volunteering as a marshal or at one of our water stops. All help will be much appreciated.

Online registration is now open! Visit our website at [www.gaucher.org.uk](http://www.gaucher.org.uk) Sign up early as numbers are limited



**Glen Moran**

Children at Harting School chose to support two charities this year – The Gauchers Association and Pets As Therapy. One of the past pupils had Gaucher disease hence the suggestion, which came from the children. Their first fundraising idea was to wear odd socks which has raised about £94. They have some other events 'up their sleeves' and are keen to run a sponsored silence next term – clearly something that the teachers would enjoy too!

Congratulations and thank you to **Glen Moran** who completed the Milton Keynes half marathon earlier this month in memory of his daughter Emily. Glen has raised over £845 for the Association and neuronopathic research.

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 would like to thank all our members who generously donated additional funds with the payment of their annual subscription along with those who pay a regular monthly standing order.

If you would like to set up a regular standing order please contact Sarah on [Sarah@gaucher.org.uk](mailto:Sarah@gaucher.org.uk) or 01453 549231

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