



NEWS FROM HQ

Welcome to our latest news bulletin, I hope you enjoy catching up on our news and events that have been happening over the summer weeks.

We are very excited to introduce our new Patient & Family Support Worker, Claire Bickley. I know she has already been in touch and met some members since starting with us. Please do get in touch with Claire direct in the office if there is anything she may be able to help you with; she would love to hear from you.

I hope you enjoy reading about the project work former trustee Maddie Collin will be undertaking over the next 12 months. I am sure you will be given the opportunity to get involved with these projects and meetings, but please do get in touch with Maddie to find out more.

We have been inundated with support over the summer weeks, not only with our annual London to Cambridge bike ride, but our amazing members also have been busy organising their own fundraising events from Tough Mudders to getting involved with Jeans 4 Genes.



Sarah Allard

I hope you were all able to celebrate International Gaucher Day. If not don't worry because it is extended for the whole month, why not choose your own day with family, friends or your work place; please do share any photos or stories I would love to hear from you.

Finally don't forget to keep up to date on all the latest news by following us on Facebook and Twitter.

Sarah
Operations Manager



- Find us on:
- Facebook
- Twitter
- www.gaucher.org.uk



WELCOME TO CLAIRE BICKLEY - NEW PATIENT AND FAMILY SUPPORT WORKER

We are delighted to introduce our new Patient and Family Support Worker, Claire Bickley to you. Claire has been with us for a few weeks now and has been busy getting in touch and meeting some members.

If there is anything you need help or information on then please do get in touch with Claire, she would like to hear from you.



Claire Bickley

Hello, I would like to introduce myself as the new Patient and Family Support Worker at the Gauchers Association.

I have a background in family support both in the UK and New Zealand, working for the past 6 years with seriously ill children and their families in London.

I am here to offer a wide range of emotional and practical support to Gaucher patients and their families. This could be support with benefit and housing applications, contacting local community services, applying for grants, organising home care, attending appointments and liaising with schools and colleges. I am also here as a friendly ear for members to talk through any concerns and worries.

My main days of work will be Tuesdays, Wednesdays and Thursdays and I am contactable by phone on 01453 549 231 or 07795 192 311, or via email on claire@gaucher.org.uk. I am happy to talk on the phone or to meet you at the hospital, your home or a local coffee shop.

I look forward to meeting some of you at our nGD Conference in November, or at the specialist clinics which I will be attending next month in Great Ormond Street Hospital and Birmingham Children's Hospital. Please do get in touch, I look forward to hearing from you.

MADDIE COLLIN TAKES UP ROLE AS PROJECTS COORDINATOR

Hello everyone, I want to introduce myself and tell you a little bit about the project work I will be undertaking over the next twelve months. As many of you will know, I have been involved in the work of the Association since 2015 when I joined the board of trustees. Since then, I have represented fellow Gaucher patients at meetings; travelled abroad to share my story with physicians, pharmaceutical company representatives and other stakeholders. Now, having just completed my postgraduate degree, I have stepped down from the board and joined the Gauchers Association team as their Project Coordinator. I am really looking forward to this journey and meeting many of you along the way.

During this time, I will be re-writing and redesigning the Type 2 information booklet; running a patient led research project - collecting the views of patients and family members to decide what research in Gaucher disease they feel is most important to carry out at this present time; organising education days for type 1 patients; and producing a film to demonstrate the work of the Association.

I am sure you will hear more about these projects in the coming months, however if you would like more information then please contact me by email maddie@gaucher.org.uk



Maddie Collin

PUBLISHED PAPER ON WEARABLE TECHNOLOGY

RESEARCH

Open Access



Measuring disease activity and patient experience remotely using wearable technology and a mobile phone app: outcomes from a pilot study in Gaucher disease

Aimee Donald¹, Huayin Cizer², Niamh Finnegan¹, Tanya Collin-Histed³, Derrilyn A. Hughes¹ and Elin Haf Davies^{1*}

Abstract

Background: Gaucher disease is an inherited lysosomal storage disorder of which there are three subtypes. Type 1 disease has no neurological involvement and is treatable with enzyme replacement therapy. Type 2 disease results in infant death and type 3 disease is a heterogeneous disorder characterised by progressive neurological decline throughout childhood and adult life. Endeavours to find a therapy to modify neurological disease are limited by a lack of meaningful clinical outcome measures which are acceptable to patients.

Results: We present results from a pilot study utilising wearable technology to monitor physical activity as a surrogate of disease activity/severity paired with a mobile phone app allowing patients to complete self-reported outcome measures in the real world as opposed to the hospital environment. We demonstrate feasibility of the approach and highlight areas for development with this study of 21 patients, both children and adults.

Conclusions: We illustrate, where patients engage in the methodology, a rich dataset is obtainable and useful for proactive clinical care and for clinical trial outcome development.

Keywords: Gaucher, Wearable technology, Mobile health

In December 2015, we reported on work being done by the Association in partnership with Dr Aimee Donald, Paediatric Clinical Research Fellow, Manchester Children's Hospital and Dr Elin Haf Davies on a pilot study for the use of wearable technology and mobile phones apps to assess the impact of nGD on daily living, to learn more about the condition, to encourage research and to develop management and interventions to support patients.

We are delighted to announce the published paper which reports on the outcomes from this pilot study is available online. To read this paper please visit our website at www.gaucher.org.uk/news

Thank you to everyone who took part in this study as well as the dedicated team who worked so hard to take this to the next level and we will keep our members up to date on this research study through our future news bulletins, website and social media pages.

DR AIMEE DONALD GRADUATES

The Gauchers Association is very pleased to announce and congratulate Dr Aimee Donald on her graduation. Aimee works at Manchester Childrens Hospital She has a specialist interest in Paediatric Neurodegenerative disease and a PhD in neuronopathic Gaucher disease. Much of Aimee's work is a result of the UK GAUCHERITE study – a longitudinal observational study of the UK cohort of patients.



Dr Aimee Donald

Aimee writes: 'Naturally I'm delighted to have completed a thesis which was all about neuronopathic Gaucher disease - my work has explored everything from the genetics of the disease to how the disease affects people over time and then started to look at some of the new data we have from things like wearable technology and eye movements. It's definitely only an introduction to the work I will do throughout my career - I have lots more plans! I feel so privileged to have spent time learning from the Gaucher patient community and to have met so many wonderful people during my work'.

INTERNATIONAL GAUCHER DAY 2019

International Gaucher Day (IGD) was launched in 2014 by the International Gaucher Alliance (IGA) and is celebrated annually on 1st October to raise awareness of Gaucher disease.

Following the success of the 2018 'Rare Stars' campaign, they continued the theme this year, but with a more personal focus on those unseen rare stars who have shown support to Gaucher patients during their journey. This might have been a parent, a sibling, a friend, an advocate or a clinician (doctor, nurse, etc.)

The UK Gauchers Association, in full support of this campaign, nominated their own 'Rare Star' for 2019: our former Chief Executive and a parent of a Gaucher patient, Tanya Collin-Histed in recognition of her continued dedication to advocate on behalf of all Gaucher patients throughout the world.

We posted a celebratory pack to each of our members, including a copy of the official poster, factsheet about Gaucher disease the IGD and temporary tattoo. We would love you to send us photos of how you have used these to be able to share in our next news bulletin as well as social media. Feel free to email sarah@gaucher.org.uk

The Association celebrated IGD a little early this year at their recent AGM meeting, by all wearing their tattoos. We were delighted to share so many photos on social media throughout the day.



Tanya Collin-Histed
with daughter Maddie

Whilst her initial involvement with the UK Gaucher Association was because of Maddie's diagnosis there is no doubt that Tanya is the embodiment of the turning of private pain to public gain. Tanya is fearless and tireless in championing the cause of the Gaucher patient both in the UK and throughout the world – whether giving personal support given to patients and their families, often in distressing circumstances or challenging healthcare providers and industry to secure access to treatment. Working with clinicians, scientists companies or governmental bodies Tanya's approach is professional and focused always keeping the ultimate goal of the best interest of patients in her sights. To so many Tanya is already a star giving comfort and hope, advice and leadership. There is no doubt that because of Tanya the world is already a better place for Gaucher Patients and she hasn't finished yet! Tanya isn't a rare star – she is a rare superstar!

2019 LONDON TO CAMBRIDGE BIKE RIDE RAISES OVER £12,800

Once again we were blessed with near perfect conditions for our annual London to Cambridge bike ride on Sunday 15th September.

This was our 8th charity 60 mile bike ride; it is always wonderful to welcome previous riders as well as many new faces this year. We were delighted to welcome Team MP Moran, a local building firm who each year participate in a charity event and very kindly chose The Gauchers Association this year.

Over 72 registered riders gathered at Victoria Park, Finchley, London to be welcomed by the Association's Chairman Dan Brown before being sent on their way.

Riders enjoyed our three refreshment stops along the route, enabling them to take a short well-earned break to refuel. Dan Brown hastily made his way from the start to Cambridge so he could congratulate riders with Sarah Allard from the office at the finish line with a finisher's medal.

It was wonderful to welcome many family members at the finish who were excited to cheer their riders home, particularly Joy Moore and her family who patiently waited to welcome their riders who all took part in memory of Keith Moore, Gaucher patient and long term supporter of the Association who sadly passed away earlier this year.

We must say a huge thank you to each of our volunteers, without whom we could not run this vital fundraising event. We were very kindly supported by volunteers who helped to set our riders off in the morning, to marshals along the route to ensure everyone went the right way and our volunteers who helped at our refreshment stops. A special thank you to June Morris who once again was our 'Star Baker' at the last water stop; her cakes were the talking point in Cambridge, many riders say this is a highlight of the ride!

Further thanks must go to Chris, Stuart and Simon from CSS who planned and signed the route and were there on the day as support for the cyclists. Janos the mechanic was very kindly driven by Malcolm, who was not too much in demand on the day and kept all riders on the road to the finish.

Our final thanks must go to everybody who took part on the day; feedback from those involved in the day has been excellent. This event has raised over £12,800 for the Association which was a tremendous effort.



THE WELLS BOYS RAISE OVER £3,750



Thank you so much to the Wells family for their amazing fundraising efforts and congratulations to everyone who took part in the North West Tough Mudder. Mum Rebecca writes:

'Having planned a small family event, on the 7th September we took a team of over 100 runners to complete the North West Tough Mudder. The adults ran either the 8 mile or 5k muddy obstacle course. They risked electric shocks and swimming through ice, yes the mud was the easy bit! The children ran a slightly kinder, one mile course and had an amazing time

We have raised over £22,500 for six charities who have helped our family through the loss of our son James and following William's diagnosis with Gaucher disease. Many of Will's class mates refused to give their sponsorship to anyone other than 'William's charity!!' Another friend gave a talk to his class about William and Gaucher disease resulting in many of his classmates donating their pocket money. It was a tough challenge for everyone, but the weather was kind and we had an amazing day. We were very grateful to have the chance to give something back to the wonderful Gauchers Association'.

MEMBERS FUNDRAISING



A big thank you to **Jackson**, his class mates and parents who supported Jeans 4 Genes this year raising a fantastic **£389**



Thank you to all our supporters who have recently sent us donations and who pay a monthly standing order in the last month we have received **£4,123**.

Gift Aid - we have reclaimed over **£2,448** on all online donations so far in 2019.

We have received over **£1,057** through **Facebook** donations so far this year.

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 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 and leaflets.



We are delighted to have secured 5 charity places for the Vitality Big Half Marathon on 1st March 2020 in London.

Take on this iconic half marathon in March 2020 and you'll enjoy a stunning 13.1-mile course, starting at Tower Bridge and finishing at the Cutty Sark in Greenwich where you'll be able to enjoy free access to The Vitality Big Festival and relax with your family and friends and celebrate your achievement.

General entry for this event has now sold out, but you can secure your place by choosing to run for us. We will provide each runner with a running vest and can help with setting up an online fundraising page.

If you are interested or know a keen runner that would like to take part whilst supporting the Gauchers Association please get in touch with Sarah in the office on sarah@gaucher.org.uk or telephone 01453 549231 - she would love to hear from you!

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making donations go further

Gift Aid is an income tax relief designed to benefit charities and Community Amateur Sports Clubs (CASCs). If you're a UK taxpayer, Gift Aid increases the value of your charity donations by 25%, because the charity can reclaim the basic rate of tax on your gift – at no extra cost to you.

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Supporting patients and families raising awareness and promoting research