Spring 2023

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

Thanks to all our inspirational fundraisers! Find out more about their efforts inside - and how you could get involved

Association for Glycogen Storage Disease UK
PO Box 699
SOUTHAMPTON
SO50 0QT
Phone: 0300 123 2790
Email: Info@agsd.org.uk
Charity Number :1132271

Thank you!

Fundraising challenge

Do you know someone keen to run or ride an event for AGSD-UK?

Have you got a great fundraising idea the whole community can take part in?

Contact info@agsd.org.uk for the chance to win a prize!

Front Cover Images:
Our London Marathon Runners:
Dale and Lauren Esliger
Liam Coote
Kirsty White

AGSD-UK recognises that not everyone is online and has access to a computer.

In this Glisten, if we mention a website or a link to information and you cannot use that to get the information you want, PLEASE PHONE 0300 123 2790

We do not want ANYONE to be excluded; we will print and post things to you.

contents

Message from the Chief Executive

Welcome to the Spring 2023 edition of Glisten.

The *Main Event* in Loughborough is fast approaching, and we can't wait to meet up in person with community members, professionals and partners.

The programme's been designed by people living with GSDs and we're aiming to include something useful and relevant for everyone, along with plenty of social time as well.

For those who can't be there we'll try to share as much information as possible in our next Glisten magazine.

The current edition includes some of the findings from recent surveys involving our readers. We know the volume of survey requests can feel overwhelming for a small community affected by such a rare condition, but the level of research interest in GSDs reflects the number of exciting potential prospects for new or improved treatments. Your input is vital in showing the impact of the condition and the urgency of new therapies, as well as helping to influence improvements in relevant policy and services. Thanks go to everyone who takes the time to get involved.

As always, the magazine features some heroic fundraising, with last month's marathon a real highlight. We're also including some of the excellent feedback received in the past year for our care and benefits advisor provision. We're constantly on the look-out for fundraising opportunities and new sources of support to enable us to continue delivering these vital services. If you have a fundraising idea, a relevant contact or know someone keen to take part in an event on behalf of AGSD-UK just get in touch with info@agsd.org.uk – we'd love to hear from you.

Val Buxton

Board and Volunteer Updates

Recent months have seen changes to our board and volunteer coordinator teams, with some welcome new additions, as well as others stepping back after contributing their valuable skills and time for many years.

AGSD-UK Chair

After generously bringing his business executive skills to the role of Chair, ***Nick Jones*** has stepped down from the position, which he had originally agreed to fill on an interim basis back in 2019. In addition to attending to his busy working life, in the near term Nick will continue to add his organisational experience and knowledge of McArdle's to the board of trustees and we're very grateful for all his input.

In the wake of Nick's decision, the board has appointed ***Florence Osborne*** as the new Chair. Flo became a member of AGSD-UK in 2020 when her son Hugo was diagnosed with GSD3b and joined the trustee board early in 2021. Flo is a Chartered Psychologist working as a Senior Lecturer in Exercise Psychology at Loughborough University. Her research interests focus on the psychology of exercise behaviour and how sport and exercise can support those with mental health issues. Flo loves all sport and has a passion for fundraising – often combining the two. Along with friends she completed the ***National 3 Peaks Challenge*** to raise money for AGSD-UK.

On taking up the position of Chair Flo said:

"AGSD-UK is a charity that is especially important to me and my family since Hugo's diagnosis. Before 2020, I had never heard of the condition. This is a big driver for my involvement. I want to help take forward our aims of improved treatment and better support for our strong and wonderful community. Having become involved as a trustee two years ago, I am delighted to have been nominated for the position of Chair and will endeavour

UPDATES FROM THE BOARD CONT...

*to work with our CEO and the other board members to continue the great work. I hope to meet many of you at the **Main Event** next month – please do come over and say hi!"*

New Trustees

Two new board members were approved at the AGM in January, with ***Charlotte Dawson*** and ***Kempton Rees*** joining the board.

Charlotte is a Consultant in Inherited Metabolic Disorders at Queen Elizabeth Hospital Birmingham and is Chair of AGSD-UK's hepatic professionals' group.

Kempton has a legal background and has been an active supporter of AGSD-UK, contributing to a range of working groups and Pompe research activities.

We are delighted they will be bringing their significant skills and experience to the board.

Volunteer coordinator changes

Over recent months *Gemma Seyfang*, *Jason McMillan* and *Sylvia Wilson* have all stepped away from their volunteer coordinator roles. They each have busy lives and we are extremely grateful for all the time they have given to AGSD-UK. They will be well known to many of you for the support they've provided and their involvement in research, events and activities.

We are very pleased that *Ben Parker* has now taken on the Pompe coordinator role while *Ailsa Arthur* will pick up the hepatic role. Both have been involved with AGSD-UK for many years and their commitment is really appreciated. Along with *Andrew Wakelin*, our dedicated coordinator for McArdle's and other muscle GSDs, they are very much looking forward to welcoming those of you able to come to the *Main Event*.

FUNDRAISING ROUND-UP

Thanks to everyone who has been busy fundraising for AGSD-UK this year.

Rob Hill is running in the Bath Half Marathon in October 2023 and has raised £215 so far. If you would like to support Rob or follow his journey this is the link to his Just Giving Page:

www.justgiving.com/fundraising/robert-hill30

You may remember that the last edition of Glisten featured runners taking part in September's Great North Run. They all did amazingly, raising both money and awareness of the condition and AGSD-UK.

Samantha Morrison raised **£1025**

Jack Molyneux and *Beth McGhee* raised **£3274**

The Newcastle University team from the John Walton Muscular Dystrophy Centre raised a total of **£1,134** through their sponsorship, bake sale and raffle

Lisa Nugent ran in the London Marathon 2022 raised a total **£569**

Huge thanks go to you to all.

If you want to fundraise for AGSD-UK and need any help or information, please email info@agsd.org.uk so that we can promote and support your fundraising.

THE LONDON MARATHON 2023

We had four incredible people taking part in this year's London Marathon. Here is a little on their training leading up to the event and why they wanted to run in support of AGSD-UK.

Lauren and Dale

*We were both part of a team of 8 friends who completed the **3 Peaks Challenge** (in 23 hrs) two years ago for AGSD-UK. We thoroughly enjoyed the challenge and also raising money for our favourite charity so when this opportunity came up, we could not say no! Our inspiration for the run and generally in life is our favourite 4 year old – **Hugo John Osborne** who was diagnosed with GSD type 3b almost 3 years ago. He is the happiest, smartest, and funniest little boy we know. We recognise how important AGSD-UK, and the work it does, has been to Hugo and his parents Flo and Tim. So, we are really pleased to have the opportunity to raise more funds for the charity.*

The past 3 months of training have flown by. We are both conquering our longest runs to date – 21 miles. While we both thought that our main focus would be improving our fitness, we spend much time researching, thinking and talking about the right clothing (no rubbing), trainers, food (before and during) and hydration. The hard work is now done, and we are really looking forward to the big day and to be running proud in our AGSD-UK vests.

Please find links below for Lauren and Dale's funding pages:

https://2023tcslondonmarathon.enthuse.com/pf/lauren-esliger

https://2023tcslondonmarathon.enthuse.com/pf/dale-esliger

Liam Coote

*In 2021 we lost our close friend **Louise Bett** who was born with Glycogen Storage Disease (GSD). Louise had been living with the rare disease for 36 years before her passing and had been a long time member of the charity.*

Running on behalf of Louise is something I really wanted to do for her and I'm grateful I'm getting the chance to do that while raising money and awareness for a charity close to her heart.

My marathon training has been going very well over the past few months. This is my first ever full marathon, and my training has seen me clock over 30 miles each week.

Living on the east coast of Fife, in Scotland my training has seen me bypass many beautiful and scenic routes. This has kept me going through the tough times and made me stronger for the race ahead.

Good luck to the rest of the runners!

Please find link below for Liam's fund raising page:

https://2023tcslondonmarathon.enthuse.com/pf/liam-coote

THE LONDON MARATHON 2023 CONT...

Kirsty White

I'm Kirsty, mum to two boys, Joshua (6) and Louis (2) and never did I think that I would be doing the ***London Marathon 2023****!*

The London Marathon was always something I watched every year with my Mum and sister, and I don't think Mum would believe I am about to run it!

I started running with the ***Couch to 5k*** *course in early 2018. Once I could run 5k, I joined the local running club and haven't looked back since. I am very lucky as I received my place for London through the ballot (third time lucky) and know how privileged I am to receive a place. Although it is a ballot spot, people asked if they could sponsor me and there was no doubt in my mind what charity I was going to raise money for.*

My Mum, Michelle McArthur had Pompe disease and although she passed in 2008, the charity did amazing things for her, and she was one of the first to trial the new drug. My Dad also raised money by completing the Vietnam bike ride for AGSD-UK back when I was a child.

Training for the marathon was great to start with but those very long runs are something else. It also didn't help living in the Fens with the wind to contend with! I'm lucky to have the support of my husband Andrew and lots of friends who have supported me on my journey.

This race is for my Mum, Michelle and I hope I do her proud.

www.justgiving.com/fundraising/kirsty-white31

Pompe Get Together in Wales

In December 2022 AGSD-UK held a Pompe get-together in Swansea, with Pompe Support Team volunteer ***John Foxwell*** and Benefits and Community Engagement Advisor ***Zainib Hussain*** hosting the event. It was great that community members in South Wales were able to catch up after so long. One of our members, ***Ken Rowe***, who attended the event, has been reflecting on the day.

The event was absolutely a delight and I met people who I already have a connection with. We all have Pompe - that's our connection. We don't need to say much but we understand the language of Pompe. It's so refreshing to have these get togethers. I really appreciate AGSD-UK who organise and pay for these events. It is a very isolating condition.

Trying to explain it to people is frustrating, for example sometimes I just don't want to explain to people who don't understand why I'm parked at a disability space. At the get-together it was great to share our experiences, knowledge, and common frustrations.

These face to face meet-ups have such a massive impact on our total wellbeing, especially after Covid. Thank you, AGSD-UK, and thank you for having staff like Zainib who is so easy to talk to and keeps in touch.

Please keep an eye out on the ***AGSD-UK Facebook*** and ***Instagram*** pages for information on more community events for different GSDs, both on-line and around the UK.

Whatever GSD you're affected by, our team of advisors and volunteer coordinators is always here for you - just contact info@agsd.org.uk and we can put you in touch with the appropriate team member.

AGSD-UK

Hepatic Get Together

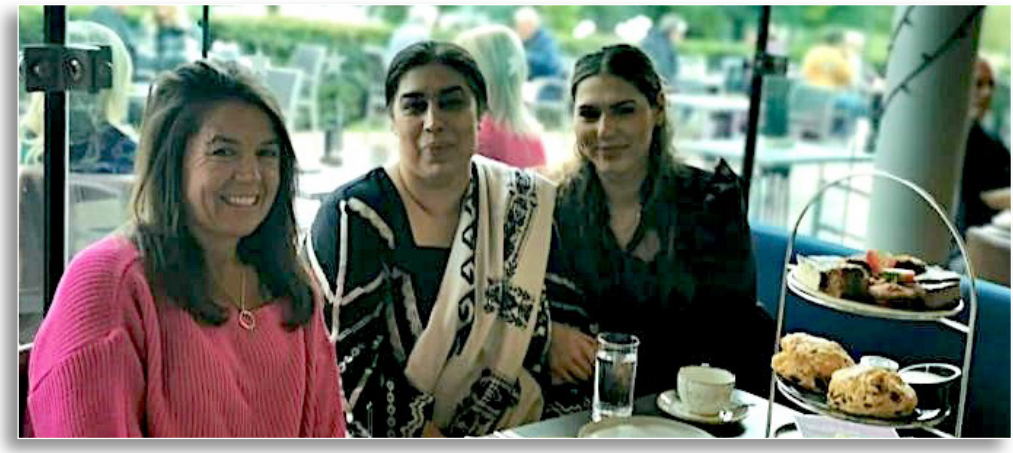
In October AGSD-UK held a get-together for people affected by hepatic GSDs in *Trentham Gardens*, Staffordshire. AGSD-UK volunteer *Maryam Ahmed* shares her thoughts on the day.

I bought my mother along with me, so that she can get a feel of the charity and the people behind it. The event took place in the afternoon - on a breezy but lovely day. The gardens were very spacious and nice and airy. It also had some shopping bits before the entrance to the gardens. I got there a little earlier and decided to have a lovely hot chocolate to warm up.

The event was well organised and catered for. Being a Muslim, the halal dietary requirements were met and being someone who has an allergy, I can confirm this was also well catered for and looked after by the team and the staff at *Trentham Gardens*.

I absolutely enjoyed the event. It was a pleasure to have the chance to meet face to face. Before the event I had spoken to people on the phone, but it's not the same as a face-to-face meeting.

After Covid, I was slightly nervous leaving my home. Having my mum accompany me gave me the ability to come and



be part of such a lovely team. It was wonderful to meet other people and to share our experiences and fears, common likes, and interests and most importantly a lot of information on GSDs. I am so glad that AGSD-UK has these events and get-togethers, as they really have an impact on our lives.

I look forward to future meetings with ASGD-UK and the team, and I strongly encourage people to come forward and share their experiences and thoughts and interests on different topics.

McArdle walking events in 2023

The McArdle's Experience

The main walking course is to be held in North Wales, based in *Tremadog*, from Friday 28 July to Friday 4 August. This is a new base for us, but of course we have been in North Wales many times, so we are very well prepared in terms of routes and activities. We still have a few places available, so if you've ever thought of joining us now is the time!

There are full details on the website, including links to several videos, so you can get a very good idea of what is involved. All you need is to be able to get into second-wind and then we take it from there.

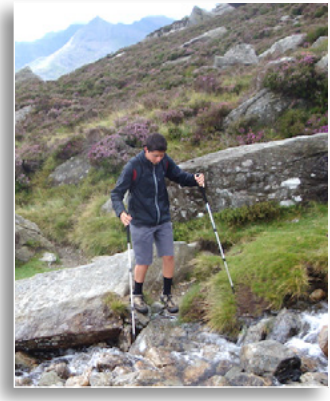
Children & Parents event

Last year, we had a partial overlap between the main course and the C&P event. It was very successful so this year we are fully overlapping. The dates are Friday 28 July to Tuesday 2 August. Some activities and walks will be combined, but for others, we will split into two groups. Again, full details are on the website.

Future Leaders training course

This is a brand new course to train leaders of future walking courses for people with McArdle disease and similar muscle glycogen storage diseases. It is going to happen the week before the main walking course – Friday 21 July to Friday 28 July. This is for participants who have attended the walking course in the past, and who wish to train towards becoming organisers or leaders of future walking courses around the world. We already have 13 people booked, but we are looking for more candidates to complete the course and receive a certificate. They will also receive a manual to help them run courses back in their own country in future. We have a short video about this new course and you can watch it here:

www.youtube.com/watch?v=ByA5SStMv-E



"Rest before risk" crossing a style.

Pausing for the fantastic view.

Using walking poles in crossing a stream.

Happy walking group on a summit.

Navigating a tricky section of riverside path.

Navigating a tricky section of riverside path.

A good path along the coast.

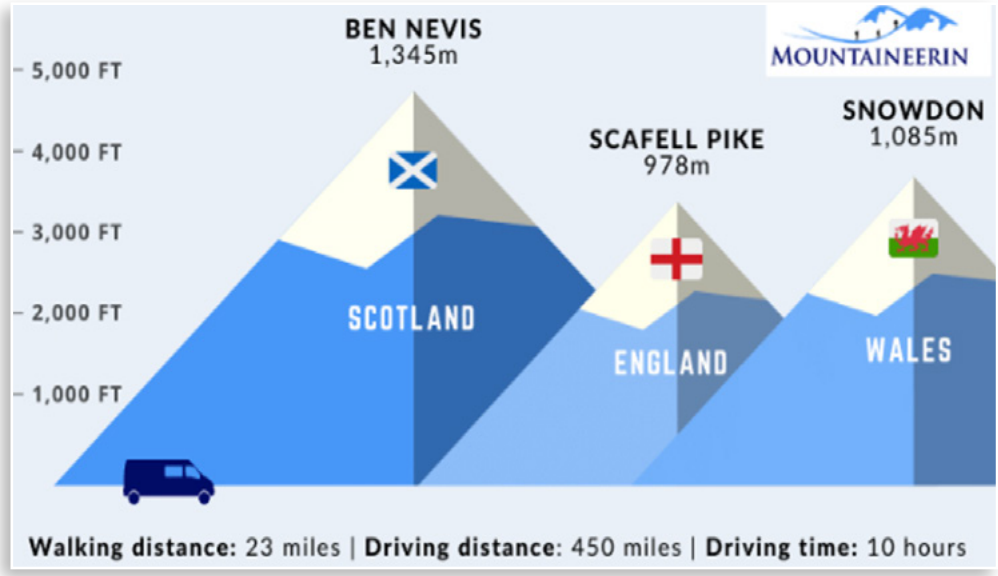
Are you up for the Three Peaks Challenge?

In the Autumn 2022 edition of Glisten, we published a short piece to sound out readers with McArdle's who might like to take on ***The Three Peaks Challenge***, albeit one spread over a week rather than 24 hours. The challenge is to climb the highest peaks in Wales, England, and Scotland. They are ***Snowdon*** in North Wales at 3560 feet, ***Scafell Pike*** in the Lake District at 3163 feet, and ***Ben Nevis*** in the Grampians in Scotland at 4413 feet.

I have received three expressions of interest. I'm hoping for a group size of about 6-8, so if you think you might be interested, please get in touch. I'm also planning to reach out to some people from overseas who've previously participated in Andrew Wakelin's ***Walking in Wales*** courses, so you may see some information posted on social media.

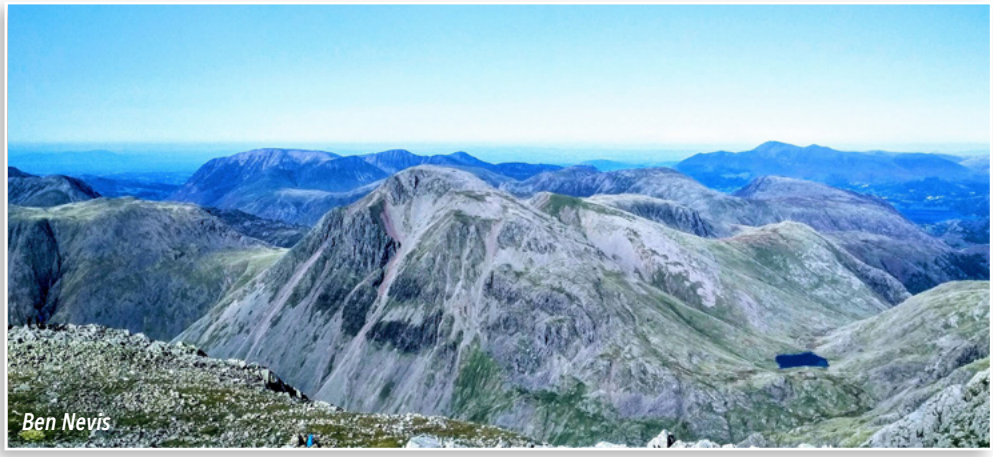
In terms of dates, I'm now thinking of May next year. That's probably best for avoiding midges in Scotland and should give us a chance to find a couple of extra people and optimise our fitness (I've re-joined the gym and reckon that will give me the chance to be as fit as I'll ever be by then).

David Thompson



Snowdon

Skafell Pike



Ben Nevis

Budget Announcements

The Spring Budget included a number of announcements relevant to the GSD community.

Most prominent was the announcement that the ***Work Capability Assessment*** would be abolished.

This assessment is used to determine the ability of people with health issues and disabilities to work. It has long been criticised by health charities for failing to capture work capability accurately and for the stress caused to those having to go through the assessment as part of their applications for ***Universal Credit*** and ***Employment and Support Allowance***.

Also announced was a new voluntary employment scheme called ***Universal Support***, aimed at encouraging people with health conditions and disabilities to seek work. Under the scheme there would be funding to match people to a suitable role and provide for their support needs, with 50,000 places a year to be made available once fully rolled out. Over £400 million was also committed to tackling major issues keeping people out of work, focusing on services for people with mental health issues, musculoskeletal conditions and cardiovascular disease.

By removing the Work Capability Assessment the Government plans to ensure that those who are able to can progress in or towards work, without the worry of being reassessed and losing their benefits. The reasoning behind the planned changes is set out in the ***Health and Disability*** white paper which was published alongside the Budget. This states:

We know that the health and disability benefits system can itself be a barrier to employment because it focuses on what people cannot do, instead of what they can. The current assessment process means you need to be found to have limited capability for work and limited capability to prepare for work to get additional income-related support for a disability or health condition.

This approach encourages people to see themselves as being unable to work and we know it can put people off from trying work or seeking employment support for fear of losing their benefits. We will

give people confidence that they will receive support, for as long as it is needed, regardless of whether they are working.

Whilst this may all sound like good news, concerns remain about how the Work Capability Assessment will be replaced. Its abolition leaves ***Personal Independence Payment*** assessment as the only mechanism for receipt of disability benefits. This is a concern as the assessment for PIP is also flawed and is intended to assess the extra costs faced by people with disabilities rather than their ability to work.

There are also worries at the suggestion that sanctions will be applied 'more effectively' to those judged capable of work but who 'chose not to seek it.' This raises concerns that more people may be forced into inappropriate work-related activity due to the threat of losing their financial support.

Health and disability rights charities will continue to press for improvements to try to ensure the system is fair. It's worth noting that as legislation is needed to implement these changes they are unlikely to happen for some time and may be overtaken by a change of government.

More positively, the budget confirmed the extension of the energy support scheme until the summer. It also announced £10m funding for the UK's ***Medicines and Healthcare Products Regulatory Agency***, in a welcome move to help fast-track access to innovative drugs and technologies.

Under a new approach from 2024, the agency would give near automatic sign off for treatments already approved by trusted regulators in other parts of the world, such as Europe, Japan, or the US, with the aim of achieving the world's quickest, simplest regulatory approval.

AGSD-UK is here for you!

AGSD-UK specialist care and benefits advisor services can help with a whole range of issues. Whether you need support with social care, education, housing, and home adaptations, navigating health services or applying for any type of benefit our advisors are here to listen, offer up-to-date information and advocate for you.

Here are just a few examples of the feedback we've received about the services over the past year:

Just wanted to send a quick email and say thank you really, for all that you've done and continue to do. I reached out to you in a moment of despair, although the outcome of my situation is uncertain.

I can say I feel much better after speaking to you. Living with a rare disease that impacts your life on a daily basis is already a struggle and having to prove that to a system who makes you feel like you're in the wrong, is a horrible feeling!

I was diagnosed with Pompe disease when I was 14 years old, and back then was made to believe that I may not be able to make it to my 30th birthday. I just turned 30, after everything that was thrown at me, I am still here, fighting!

I appreciate the incredible work that you do at AGSD-UK for people like myself. You give hope when it feels like there's none left and the strength to fight when it's so easy to just give up! The work that you guys do, going above and beyond to get justice for people that have been let down continuously is applaudable.

Thank you for making us feel that we are not alone. Thank you for your care and compassion.

I just wanted to take the time to email you to thank you for all your help and guidance.

As a result of your continuous support, I will now be receiving the enhanced mobility payment; this will help me greatly. Thank you once again.

I am writing this e-mail to thank this charity for all its support. Today I had a home visit from Zainib Hussain AGSD-UK Welfare Officer. She supported me with my PIP form. I was feeling overwhelmed and anxious to do this. I had no motivation whatsoever. It is hard enough to manage my Pompe as my heath has declined. You have to keep proving your illness to DWP.

Zainib got me to understand the form, and how to respond to each question...It has made me feel more at ease as the form is completed and I don't need to have it on my mind.

Thank you, AGSD-UK, for having a fabulous service.

I was recently diagnosed with Late Onset Pompe Disease, which has come as a huge shock to me. I am struggling daily to come to terms with both my physical and mental symptoms.

I desperately needed help and guidance and both Zainib and Elizabeth at AGSD-UK have been absolutely wonderful in supporting me at this really difficult time with both practical and emotional support about Pompe Disease.

In particular, I have recently needed to apply for Personal Independence Payment (PIP). I found the PIP form extremely hard to fill in not just because of its length but also my ability to talk about my symptoms and describe them accurately.

Zainib was absolutely fantastic in helping me to fill in the form and apply. I simply could not have done this without her and AGSD-UK.

Thank you all so much!

No matter what GSD your life is affected by, our team is here to help. Our specialist care advisor, ***Elizabeth Davenport*** and our benefits advisor ***Zainib Hussain*** have a wealth of knowledge and experience to offer. They can be contacted by emailing **elizabeth.davenport@agsd.org.uk** or **zainib.hussain@agsd.org.uk**

IamGSD website launches new pages

IamGSD has recently launched a number of new pages on their website. Perhaps the most important is "When to seek urgent medical care" and this can be found as the first item on the Medical menu. This page has a flowchart which gives guidance for you to make your decision as to whether you need to go to hospital for urgent, medical care. (See page 24)

Whilst muscle contractures and rhabdomyolysis can be very painful, the much more important risks are the possibility of acute renal failure or compartment syndrome. There is a secondary flowchart covering compartment syndrome. It is often quite a hard decision to make so this guidance with the flow chart and notes is a most welcome development. You can download both flowcharts and the notes as a double-sided A4 sheet to keep handy on your phone. This guidance has been checked over by IamGSD's Scientific Advisory Board. IamGSD estimates that across the 4000 to 5000 people currently diagnosed with McArdle's worldwide, about 20 times per day someone somewhere is having to grapple with this decision.

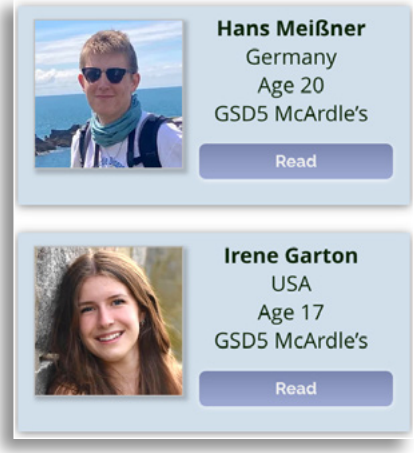
Another important new page is a sub page of the GSD5 McArdle's section, and it is called "Teaching each other". This page addresses nine areas which are sometimes the subject of misconceptions by medical professionals. Quite often they have only seen one or two people with McArdle's. It is understandably difficult for them to grasp the nuances of the condition, so IamGSD wants to alert you to these possible pieces of misinformation and give correct information to help you to address them with the medical professional concerned. Obviously, these issues will not arise with our McArdle's clinic in the UK, but some of them may do with your GP or with other specialists around the UK, who don't have the same in-depth experience with McArdle's.

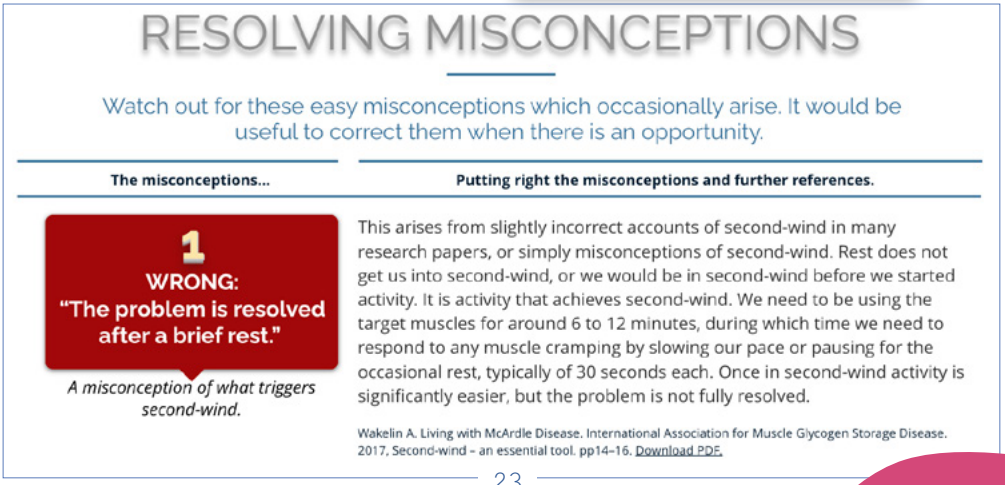
Here is an example of one piece of advice which is sometimes wrongly given to patients: "The problem is exercise, so don't exercise." This arises because the cardinal symptom of McArdle's is usually described as "exercise intolerance", although we are managing to gradually change this to "physical activity intolerance", which is a much more accurate description. In fact,

of course, it is essential for us to regularly exercise in order to maintain our aerobic fitness, which makes life with McArdle's so much easier.

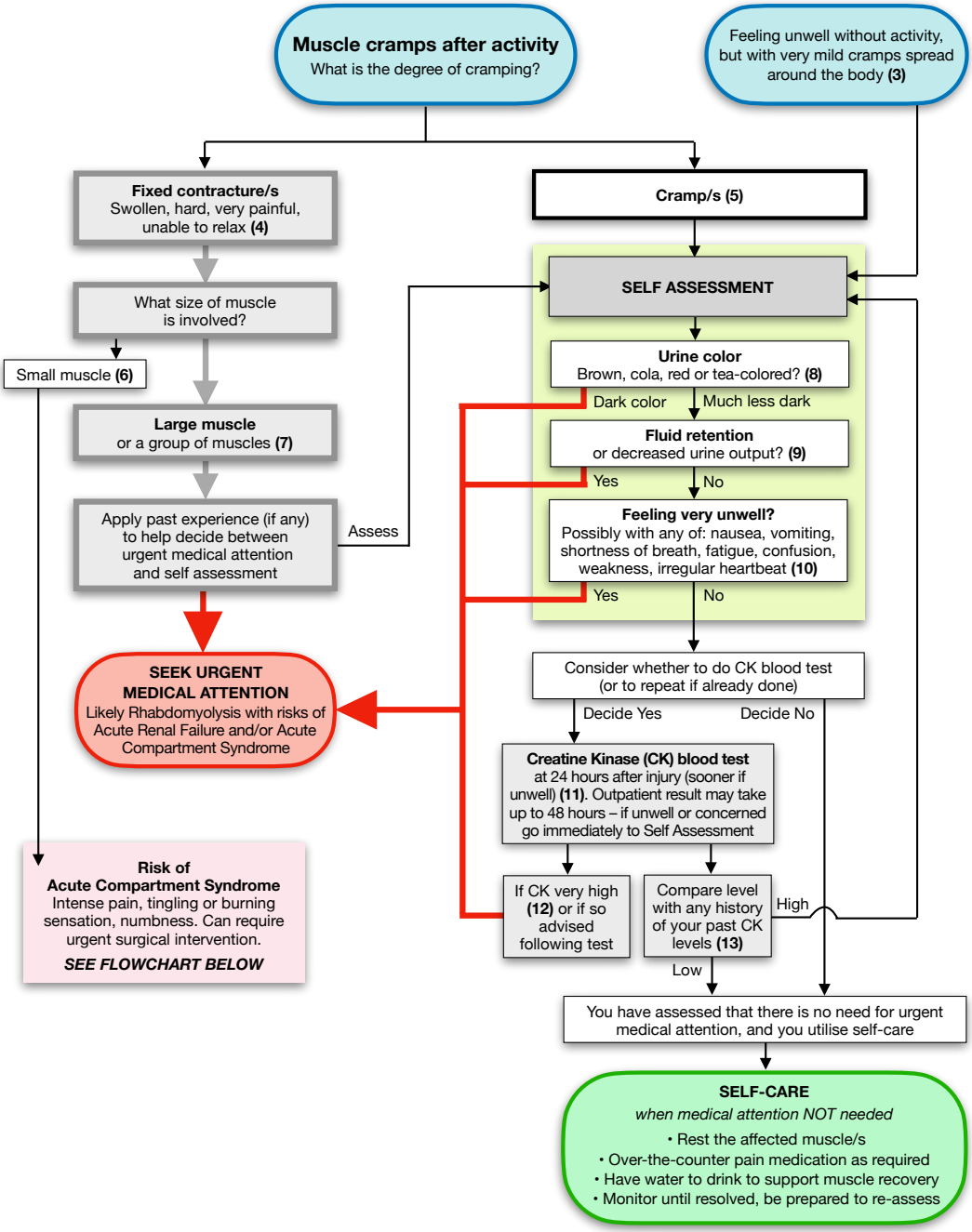
There is a new page called ***Designs for T-shirts and more***. IamGSD has made these designs available for free download so that you can take them to a local company to have your T-shirt, cap, mug, or whatever, made. The page is on the resources menu.

There are also a few new ***Personal stories*** from McArdle's people. One from ***Hans Meißner***, who attended the walking course in Wales last summer, one from an anonymous young man from the US and another from ***Irene Garten***, age 17, who has twice been over to Wales from the US to join the walking course. We have a few more personal stories in the pipeline, but if you would like to tell yours, please read the guidance notes on the website and then get in touch. It is not just for McArdle's, but also all the other muscle GSDs.





URGENT CARE DECISION FLOWCHART



Mental Health & GSD

ELIZABETH DAVENPORT

Having good mental health means that you can cope with daily challenges and stresses. For the next few editions of Glisten, we will look at ways to achieve a more relaxed and peaceful way to deal with triggers and challenges. I will also share information, coping strategies, and exercises on AGSD UK social media. For this edition I would like to look at anxiety.

What is anxiety?

Anxiety is usually a natural response to pressure, feeling afraid or threatened, which can show up in how we feel physically, mentally, and in how we behave. It's common to describe anxiety as a feeling of dread, fear, or unease, which can range from mild to severe.

Anxiety can become a problem if it starts to feel so intense or overwhelming that it starts to interfere with our daily life or affect our relationships. Anxiety can affect our mind, body, and behaviour.

Common symptoms of anxiety include:

- Feeling tired, restless, or irritable
- Feeling shaky, dizzy, or sweating more
- Being unable to concentrate or make decisions.
- Having trouble sleeping
- Worrying about the past or future or thinking something bad will happen
- Experiencing headaches, tummy aches or muscle pain
- Having a dry mouth
- Feeling pins and needles
- Noticing your heartbeat gets stronger, faster, or irregular, or you get short of breath when you start feeling anxious

If you can't tell if shortness of breath is from anxiety or if you're worried about any other symptoms, see a GP.

Ways to manage anxiety:

Try talking about your feelings to a friend, family member, health professional or counsellor.

Use calming breathing exercises. Taking time to concentrate and slowing down your breathing can help immediately if you are overwhelmed or triggered.

Mindfulness might also help. Find a space, even if it is just for a few moments, to concentrate on the present. Even just staring at an object can give our mind and

Mental Health & GSD cont...

body a moment to gather thoughts and emotions. I will discuss more about mindfulness in the next edition of Glisten. Try:

- Listening to a guided visualisation, meditating, or relaxing to calming sounds can give you the chance to reset your current feelings.
- Try activities such as running, walking, swimming, and yoga to help you relax. A bubble bath or finding some alone time to listen to your favourite music may also help.
- If sleeping is a problem, find a routine to help improve this. This could include no screens or mobiles in the bedroom. Eat less before bedtime and cut down on the caffeine.
- Try to eat healthily throughout the day.

We all have different ways to cope with stress and challenges and I would be very happy to look at options for you. Please email me and we can arrange a chat. I will also add some exercises and strategies on AGSD-UK social media.

Elizabeth.davenport@agsd.org.uk

Specialist Care Advisor

If you need urgent crisis support, please contact NHS 111, your own GP or go to your local A&E department if you require emergency support.

Useful Links

The links below might also be useful:

MIND

www.mind.org.uk

0300 123 3393 or legal line - 0300 466 6463

info@mind.org.uk

Local Mind services include befriending, counselling, crisis helplines, talking therapies, employment/training schemes, and drop-in centres.

Campaign Against Living Miserably (CALM)

www.thecalmzone.net

0800 58 58 58

CALM runs a free, confidential, and anonymous helpline as well as a webchat service, offering help, advice, and information to anyone who is struggling or in crisis.

YoungMinds

www.youngminds.org.uk

YoungMinds are a mental health charity for children, young people, and their parents, making sure all young people can get the mental health support they provide listening services, information, and support for anyone who needs to talk, including a web chat.

National charity helping people with Anxiety - Anxiety UK

www.anxietyuk.org.uk

Samaritans

www.samaritans.org

Night and day helpline: 166 123

The Mix

www.themix.org.uk

The Mix is a UK based charity that provides free, confidential support for young people under 25 via online, social, and mobile.

Befrienders

www.befrienders.org/

Worldwide directory of emotional support helplines.

You can also download a free PDF of our AGSD-UK Mental Wellbeing & Pompe Disease publication:

www.tinyurl.com/ypvybayf

Experiences of late onset Pompe

Few studies have investigated the experiences of people with late onset Pompe through getting a diagnosis, receiving treatment and managing their lives with the condition. Working with *Amicus and Pompe Support Network* we aimed to address this gap in early 2021 by conducting an online, in-depth, quantitative survey of people affected. The findings of the survey are set out below and highlight the urgency of developing and ensuring access to more effective treatments.

Our thanks go to all of you who took part.

Figure 1: Timeline of key events

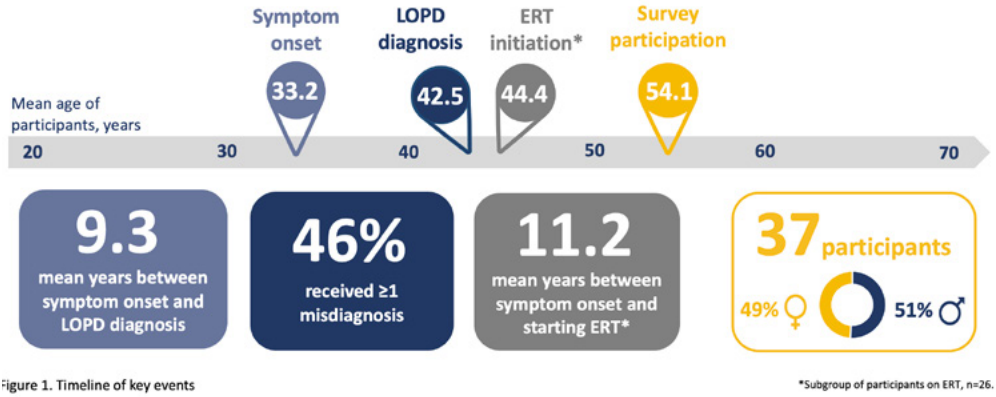


Figure 1. Timeline of key events

*Subgroup of participants on ERT, n=26.

- We surveyed 37 participants with an equal split of men and women whose average age at diagnosis was 42.5.
- Diagnosis came around 9 years after symptom onset, and notably nearly half of all participants received a prior misdiagnosis.
- A further 2 years went by before the start of treatment for most participants.

Figure 2a: Symptoms and help needed

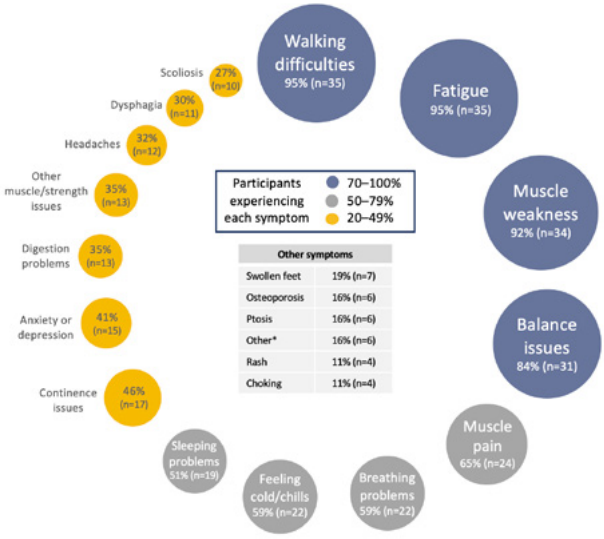
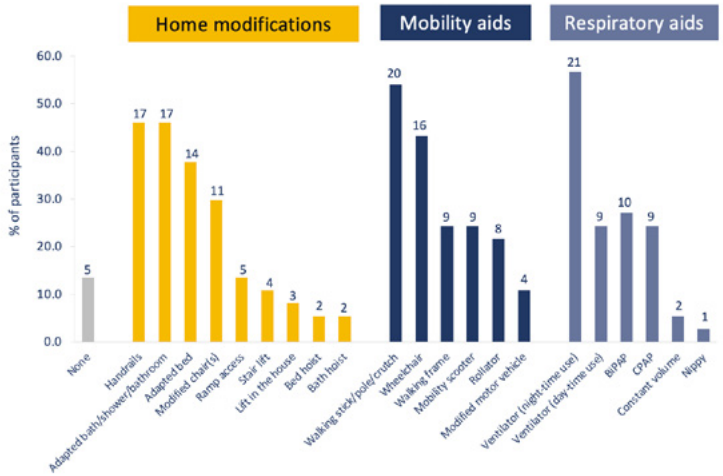


Figure 2a. LOPD-associated symptoms experienced in the last 3 months

*Other = speech difficulties (5%; n=2), nausea (3%; n=1), lazy eye (3%; n=1), pressure sore (3%; n=1), climbing hills/stairs (3%; n=1). Participants could select multiple answers. Total study population = 37.

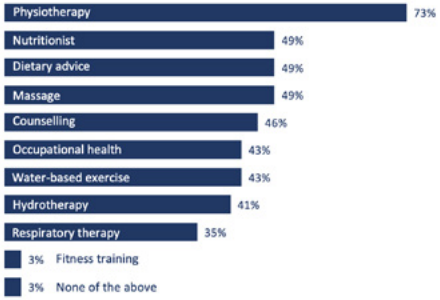
- The impact Pompe can have on day-to-day living is shown by the array of symptoms commonly experienced by participants.
- Over 90% of those surveyed had walking difficulties, muscle weakness or fatigue in the last 3 months.
- Over 80% needed assistance with day-to-day living, with the majority receiving help from family members.

Figure 3a: Physical aids currently used



- Looking at how people manage their condition, this figure shows the extent of home modifications and mobility and respiratory aids used by people living with LOPD.
- The most common home modifications were handrails and adapted bathrooms.
- Over half of people needed to use a walking stick or crutch, and a ventilator at night.

Figure 3b: Supporting therapies



- When asked which supporting therapies they would find useful to manage their condition, 73% of people answered physiotherapy, followed by consultation with a nutritionist, dietary therapy and massage.

Figure 3c,d,e: Current treatment and condition status since starting ERT

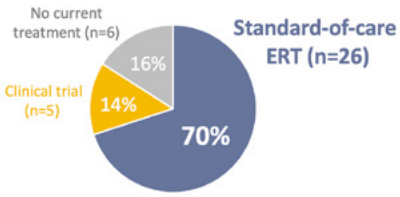


Figure 3c. Treatment currently received for LOPD
Total study population = 37.

Figure 3d. Length of time on ERT
Icons represent number of participants in each time range.
Subgroup of participants receiving ERT = 26.

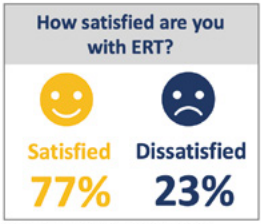


Figure 3e. Satisfaction with ERT
Subgroup of participants receiving ERT = 26.

- 26 of our participants were on enzyme-replacement therapy (or ERT) at the time of the survey – half of these for over 10 years.
- Unfortunately, 77% of people believed their condition to have deteriorated since starting ERT and this was most pronounced for people with the longest treatment duration.

Figure 3e, 3g: Current treatment and condition status since starting ERT

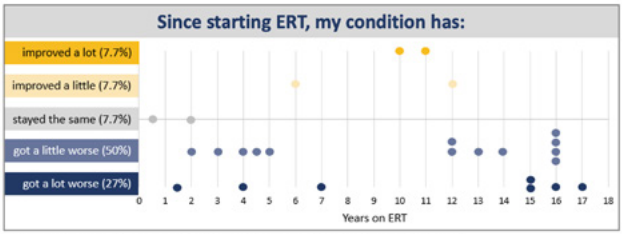


Figure 3e. Condition since starting ERT related to length of time on ERT
Subgroup of participants receiving ERT = 26.

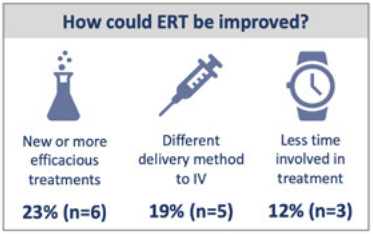


Figure 3g. Desired improvements to ERT
Subgroup of participants receiving ERT = 26.

- When asked what improvements to ERT they'd like to see, several mentioned the need for new, more effective treatments and some wanted an alternative to intravenous delivery.

Figure 4: Impact of COVID-19

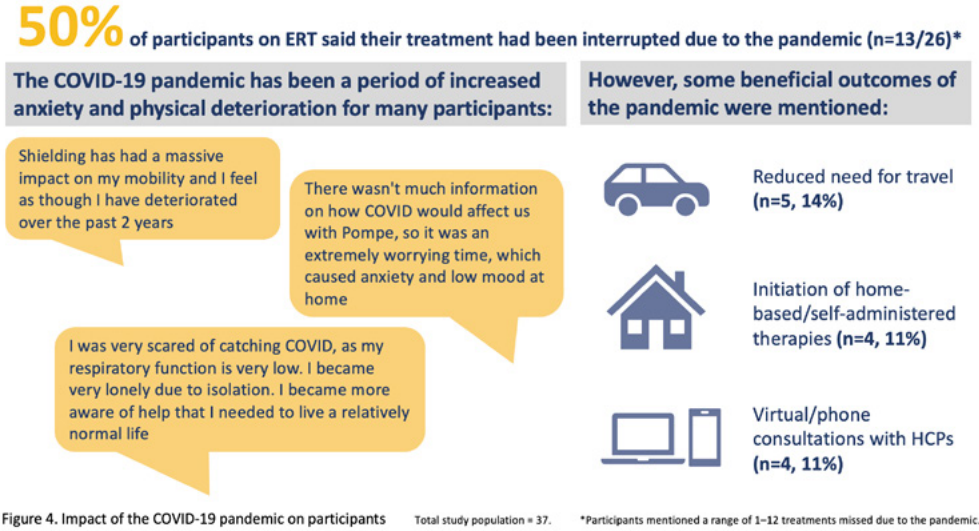


Figure 4. Impact of the COVID-19 pandemic on participants Total study population = 37. *Participants mentioned a range of 1–12 treatments missed due to the pandemic.

- Not surprisingly, the COVID-19 pandemic was a period of increased anxiety and physical deterioration for people living with LOPD - half of participants had their ERT disrupted due to the lockdowns.
- However, a few mentioned some benefits of the pandemic, including the reduced need for travel to and from appointments and the initiation of virtual consultations.
- Some people also learned to self-administer their treatment or received help from a carer rather than a nurse.

The results of this online survey revealed the substantial impact of living with LOPD, the unmet needs of people affected and in particular, the need for quicker diagnosis and development of more effective treatments.

We would like to thank the participants of this survey for their important contributions.

Instructional Video Series for McArdle's

You may be aware that both AGSD-UK and IamGSD have YouTube channels with videos about muscle GSDs, especially McArdle's. Some of these are professional presentations made for conferences, or for our *Winter Seasons* during the Covid pandemic, but many are quite informal, and some just a bit of fun.

IamGSD is planning to make a new series of more formal instructional videos on carefully chosen topics. These will be branded as a set and kept in their own designated playlist. They will form the foundation of helping people to understand and manage their McArdle's. The playlist is going to start with half a dozen videos, and we hope it will be extended in future months and years. It is expected that the first one to be released will be on the *Six second rule*. Watch out for it being launched with a news item on the website and a post in the McArdle's Disease Facebook group.

If you feel there is a particular subject which would be worthy of a short instructional video, do please drop a line to Andrew Wakelin on: andrew.wakelin@iamgsd.org

Uncooked Cornstarch Supplies

Access to uncooked cornstarch can be vital in enabling people with hepatic GSDs to maintain normal blood sugar, yet issues with prescribing, manufacturing and logistics are bringing real challenges for community members in securing the supplies they need.

Prompted by increasing concerns from specialist dieticians in our hepatic GSD professionals' group, AGSD-UK recently undertook a survey on uncooked cornstarch supply (not Glycosade).

So far 21 people who rely on uncooked cornstarch to manage their GSD or their parent/carer have replied. Two thirds of those responding to date reported problems getting supplies.

Issues raised include the variability in access to cornstarch on prescription, significant time spent, and distance travelled to secure the supply needed and the increasing costs and financial impact.

Here are just a few of the comments received:

"It can be difficult to find, as most shops don't stock it, just the big supermarkets, and then it's just luck if you can get it."

"I have had trouble buying cornflour from Tesco or there has been short supply. Also, the price has doubled in the last few months in the supermarket."

"Can't get it prescribed."

"I used to have to get a prescription give it to my family to take to a pharmacy 50 miles away, wait for the supplier to visit the cash and carry then collect. I now ask a family friend to buy from Cash and Carry."

"Expensive and not always available in the quantities required"

"I travel a distance to find a wholesaler that stocks it."

"The prices go up every time."

"As I am single mother it is very hard for me to buy it."

"Pharmacy was unable to source it through their suppliers."

Responding to the survey, ***Charlotte Ellerton*** Specialist Dietetic Practitioner in Metabolics said:

Uncooked cornstarch is vital for managing hepatic GSDs, and the increasing number of difficulties experienced by members of the GSD community in accessing this is a huge concern. We are very keen to listen to all the experiences of individuals and their families and carers so we can raise this with relevant groups and work together to ensure that everyone who needs uncooked cornstarch can obtain this.

AGSD-UK are still interested in hearing your experiences, good and bad – just fill in the form on the following link: https://form.jotform.com/230724085073351

Your feedback is extremely valuable as we continue to work together to try to improve access moving forwards. Thanks go to all those taking the time to respond.

Travelling with GSD

As summer is approaching, we thought that the following information might be useful, along with a few useful tips shared by the GSD community.

Where to start:

A little planning will make the trip easier from start to finish. When you are considering a break or holiday ask yourself the following questions:

- Where do you want to go? Find out how accessible your destination is. Online forums can help with this.
- Consider a specialist agency who can cover everything for you from booking your transfers to and from the hotel to whether the swimming pool has a hoist to which spas have wheelchair accessible treatment rooms.
- If travelling abroad check to make sure you have the best travel insurance cover. Remember that additional insurance to protect mobility aids may also be needed. Shop around for your insurance. A comparison website can show you the best deal depending on how much assistance you need. Ask the GSD community for their recommendations but do your own research to get the best package for your individual needs.
- Ask your GP if any immunisations are needed as particular health conditions

may need more. Your GP should also check if you are fit to fly.

- Check with holiday venues or airlines to see if you can get free or discounted space for your carer or PA.
- Checking in mobility aids: Your wheelchair will be stored with the luggage, and you will be given another to transport you around airports at either end and to and from your seat. If your personal wheelchair or mobility aid comes with specialist storage instructions or needs to be managed in a certain way, make sure the airline knows and you know where, when and how your mobility aid will be returned to you.
- Check that you have packed enough medication. If flying, pack meds in different bags in case any of your luggage goes missing. But also carry meds with you on board. Check to see if you need a letter to support carrying your meds on the plane. Do not forget any extras you may need to take with you such as compression socks, spare feeding tubes or masks.
- At the venue take an extension lead so you can plug your equipment into multiple sockets and have the power connected right next to where you need it.

Enjoy and relax…. If you have any tips that you would like to share, then please let us know.

New Leaflets From IamGSD

IamGSD has produced some new leaflets which are now available on-line and as PDFs from the IamGSD website, and as printed copies from Andrew Wakelin. There are now eight leaflets in total, as well as several books, all available through IamGSD.

The first leaflet is ***At peace with McArdle's***. This has ideas to help with emotional well-being and addresses issues such as:

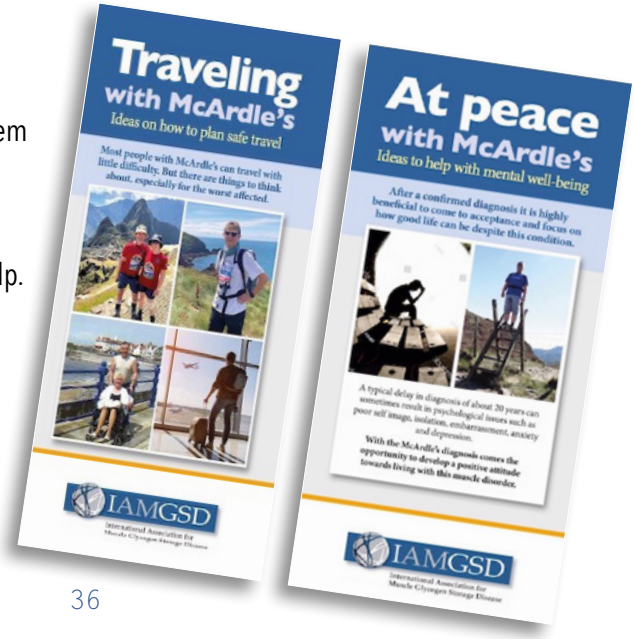
- accepting the diagnosis
- guidance on exercise
- learning about McArdle's
- putting things in perspective
- creating a support system
- feeling better
- looking ahead

Anxiety and depression can be a problem for some, so for those who have more serious or long-term issues there are suggestions about sources of further help.

The second leaflet is ***Traveling with McArdle's***. This title is pretty self-explanatory, and of course it has lots of tips about how to cope well with traveling (only one 'L' in American spelling!).

There are subjects such as: alone or in a group, at home or abroad, your luggage, your hotel, airports and stations, planes and trains, vacation activities, dangers to avoid, holiday insurance, plus a section for business travelers and one for language support.

The originally launched leaflets are: ***At home***, ***At school,*** and ***At work with McArdle's***, plus the more recent ***At the gym, At hospital;*** at the moment all remain available. Check out the ***IamGSD*** website, resources menu, publications page.



Every movement counts

The theme for this year's International Pompe Day was ***Every Movement Counts***. Here two people affected by Pompe describe how they keep moving.

'I like to go on walks (rolls) with my dogs, I have a Cavachon and a Labrador. My wheelchair has pretty good suspension so it can go over quite bad terrain such as woodland. So, I like to go out as much as I can with them and take them to different places. I also play ball with them and have taught them to put the ball within my reach.

After a long day sitting in my chair, I like to lay down and watch TV in bed, I have a rising bed which makes it easier to see the TV. My wife will then stretch out my hips and legs (shown by a physiotherapist).

Sometimes I will use a pedal machine on the bed which gives me good circulation and will hopefully keep muscle strength.

To relax I like to play the PlayStation or Nintendo. Then finish the night with a good book which usually sends me straight to sleep. (Person living with late onset Pompe)'.

'My daughter is starting a new adventure in her life; she is going to nursery. She loves playing with her friends and likes story time and sits on the carpet listening to the nursery nurse tell stories that make her laugh. She is excited and happy and loves meeting people.

Her favourite things to do are painting and singing. We have artwork all over the house and she gladly sings whenever and wherever possible.

She even sings for AGSD-UK staff down the telephone. (Toddler with infantile onset Pompe)'.

My placement in Northern Ireland

Ciara Harkins

I thoroughly enjoyed my time at university, I learnt a lot about myself personally and with GSD1a.

One of the biggest challenges I had faced, looking back, was my placement year. It was a long and tough time applying. I had always mentioned my condition but because of how much physical activity the job involved, I often got turned down. I was beginning to lose hope as I only had around a month before starting my final year, however a couple days later, I received a phone call asking if I wanted an interview for a potential placement.

Fast forward a week and I had been offerered the placement. The only catch was that it was in Northern Ireland. I was in two minds whether to take it or not as this was an opportunity of a lifetime. Moving to Northern Ireland where I only knew two people, was risky for someone like me! Without thinking too much about it, I accepted the offer.

The hardest part was yet to come, getting to my accommodation. I immediately spoke with my doctor regarding a travel letter as travelling with cornflour without an explanation of a professional is always interesting. I then booked my plane tickets and off I went to the airport. I said goodbye to my mum as I knew I wouldn't see her for a year due to Covid.

I was nervous in general but also about making sure I had all my medical supplies with me. I got through security a lot easier than I had thought. When travelling, I always take everything out of my bag to be transparent and had placed my doctor's letter with my bag going through the x-ray, along with keeping a copy with me.

The flight was only an hour, and luckily my friend met me at the airport to take me to my accommodation and briefly show me around.

I then started my job and had a year out in Northern Ireland. It was one of the best times of my life.

FLIGHT RIGHTS

I have a GSD that is progressive. The aches, pains and lack of mobility will take their toll as each year passes. Our dogs have noticed that the long rambles in the countryside have been reduced to shorter bouts of pottering in the local park. I feel their disappointment, but this is my life and I constantly appreciate the good things I am blessed with.

The long yearned for break in southern Europe at the end of our damp, grey winter was approaching, and I was very much looking forward to it.

I have written this piece to express my sadness at the way I was treated by one of our homegrown airlines who promise *'To Take More Care of You'*.

I had written in advance to ask for extra assistance, yet administration errors at their end had led to me walking, almost running many miles around the airport. I pleaded for help: they knew I needed help: but no one helped, so the plane was missed.

We bought extra tickets at our cost and flew out later that day.

The once '*World's Favourite Airline*' admitted liability and offered a derisory sum in credit as compensation. Many emails later we were offered the full amount to cover the cost of buying more tickets. However, this was only as a credit note that lasted a year. An unpleasant sting.

Does this compensate for the days I spent on holiday, not wanting to go out because my muscles ached too much? Does this make up for the fact that, for the first time ever, I gratefully sat in a wheelchair to make it out of the airport at the other end?

As I was writing this, I found a news item on the BBC website from December 2022 '*Disabled airport travellers given 'unacceptable support*'.

The Civil Aviation Authority noted in a report that many disabled and less mobile passengers missed summer flights at Heathrow due to poor accessibility performance.

I'm sharing this because we need to let people know about the situation. All publicity, no matter how small, will help to raise awareness.

A new campaign has been launched to press the government to give the Civil Aviation Authority powers to fine airlines and others in the airline industry if they fail to look after passengers with disabilities and their equipment.

The Rights On Flights campaign is calling for airlines and other industry members to be held accountable when they fail to provide adequate assistance, despite knowing in advance about passengers' needs. The campaign also calls for accountability when damage is caused to wheelchairs or essential mobility devices, or when passengers with disabilities are left on flights for a prolonged period once the flight has landed.

You can find out more about the campaign and how to get involved by visiting:

https://www.disabilityrightsuk.org/rights-flights

Personal stories from McArdle-ites around the world

There is an amazing community of people with McArdle's around the world, quite a few of whom have met each other at various international conferences and on the walking courses in Wales, Germany, and the US. And if not any of those, then they know each other through the McArdle disease Facebook group.

Here is a snapshot of what amazing things a few of them have been doing in recent months, overcoming, or finding ways around their physical limitations.

Christi Stevenson in the US found herself needing to help at work on a day when they would move furniture, etc. She very cleverly wore her **second-wind** T-shirt, to gently remind everybody that she was not going to be doing the same as them!

Siobhan McCann from Ireland, visited Rome and was proud to climb all the steps of the Basilica di Santa Maria.

"OMG, I made it to the top!". Martine Schinck from the Netherlands was travelling in Indonesia. Martine says "On this beautiful island called Padar the climb was more than 880 steps so I told my friend I wouldn't make it, but I started. Every time I thought 'Okay, stop here' I then decided to go a little further. And after 50 minutes I reached the top!"

A smiling McArdle's group on the second walking course, organised by Monika Weingartz (on the left) of the German equivalent of AGSD-UK, the Selbsthilfegruppe Glykogenose Deutschland (SHG for short!).

Penny Scott from Australia surprised herself by climbing five long flights of stairs to the top of the Pilliga Forrest Lookout Tower in the Timmallallie National Park, New South Wales. The park information warns that the tower sways in the wind! She says she had to stop for a rest on each landing, but she made it!

Third paper from the Euromac Registry

A third paper has been produced from the Euromac Registry, which many people in the UK have joined via the McArdle's Clinic. In the past, there has been a paper on how the registry was developed and constructed, and then there was another paper analysing the data. These papers remain available, but there is now a third paper, which is just about to be published, and probably will have been by the time this magazine reaches you.

This is the link to the Euromac website: https://www.euromacregistry.eu/

The latest paper analyses functional status and social participation, previous and current treatments (medication, supplements, diet, and rehabilitation) and any limitations of those registered with Euromac. The following standard questionnaires were used: ***Fatigue severity scale, WHO Disability Assessment Scale, health related quality of life*** and ***International Physical Activity Questionnaire.***

- Of 282 participants with confirmed muscle glycogenosis, 269 had GSD5
- Of them 196 (73%) completed all the questionnaires
- 180 (67%) were currently working. Previous medical treatments included pain relief (23%) and rehabilitation (60%)

- The carbohydrate-rich diet was reported to be beneficial for 68%, the low sucrose diet for 76% and the ketogenic diet for 88%
- Almost all participants (93%) reported difficulties climbing stairs
- The median FSS score was 5.22, indicating severe fatigue.

The Euromac Registry has provided insight into the functional and social status of participants with GSD5: most participants are socially active. Regular physical activity and different dietary approaches may alleviate fatigue and pain.

The full paper will be available to all through Open Access once it has been published in the journal. This is expected imminently. You will be able to access this paper, and can already access the first two papers, as downloadable PDFs, from the IamGSD web site – see the Research menu then Euromac papers.

Clinicians and researchers on McArdle's gather at a Euromac workshop in the Netherlands. Countries represented: Denmark, France, Germany, Italy, Netherlands, Spain, Sweden, UK, and USA.

Continuous glucose monitoring for children with hypoglycaemia: evidence in 2023

Worth C, Hoskyns L, Salomon-Estebanez M, Nutter PW, Harper S, Derks TGJ, Beardsall K, Banerjee I. C Front Endocrinol (Lausanne). Jan 2023

In this article the authors review developments in continuous glucose monitoring and its use in rare hypoglycaemic disorders.

They reflect that while there have been advances in device technology, accuracy at a given point remains low for children with non-diabetes hypoglycaemia.

They deduce that machine learning to prevent hypoglycaemia has not so far demonstrated sufficient predictive accuracy for it to be used in practice for this purpose.

Nevertheless they conclude that monitoring has led to a better understanding of hypoglycaemic disorders, helping with diagnosis and enabling informed changes in clinical management. The large amount of data drawn from continuous monitoring means there is better information about the incidence of hypoglycaemia and the data has also helped improve the design of clinical trials.

The evidence reviewed indicates that by supporting a more individual approach to prevention, with a focus on underlying behaviours, continuous glucose monitoring has resulted in a real world reduction in hypoglycaemia.

Having critically analysed the updated evidence for use of continuous monitoring in non-diabetic childhood hypoglycaemic disorders since 2020, the authors provide suggestions for its qualified use.

Find the link to the published evidence here

www.ncbi.nlm.nih.gov/pmc/articles/PMC9900115/

A novel drug trial for McArdle's

An American pharmaceutical company, *Edgewise Therapeutics*, is undertaking an early-stage trial of a drug in McArdle's and three muscular dystrophies: Becker, Duchenne, and Limb Girdle 2i.

The drug code name is EDG-5506 and it is described as a *"Novel Small Molecule to Protect Dystrophic Muscle"*.

Whilst McArdle muscle would not normally be labelled as dystrophic, the company is hopeful that the drug will be useful in this condition. The drug limits the extent to which the muscle can contract, the idea being to stop the muscle going as far as entering a contracture, and maybe even a serious cramp. This is a very preliminary trial and obviously we shall be keenly awaiting the results.

You can read about their plans on the link below, then follow: areas of focus, muscular-dystrophies.

Edgewise THERAPEUTICS

Find the link to these plans here

www.edgewisetx.com/

NICE Review Cipaglucosidase Alfa (with Miglustat) for Treating Pompe

Following its 2022 approval of Avalglucosidase alfa for treating infant and late onset Pompe, the National Institute for Health and Care Excellence (NICE) is currently moving forward with its appraisal of Cipaglucosidase alfa (with miglustat).

AGSD-UK submitted evidence to the initial NICE stakeholder consultation last autumn, drawing on findings from our 2021 survey of people affected. This showed the significant impact of Pompe and urgent need for a range of effective therapy options. We also responded to the recent technical engagement phase of the review. Our nominated patient expert with experience of the therapy has been selected to take part in the NICE committee meeting on 3rd May and we await the outcome with interest. This is expected to be published on 12th July.

AGSD-UK acknowledges the support of all the industry partners, trusts and foundations that help fund our work on behalf of the GSD community. Our thanks go to Amicus, Astellas Audentes, Avro Bio, Beam, Edward Gostling Foundation, Sanofi, Spark Therapeutics, Ultragenyx and Vitaflo.

Kirsty White in training for the London Marathon

Upcoming events!

- **The Main Event,** 20th - 21st May
- **McArdle Walking Courses**, 21st July - 4th August

Get in touch now with **info@agsd.org.uk** for joining details for any of these events and look out for more opportunities to come together in 2023

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Spring
2023

Emma Grove Design 07990 520 807

AGSD UK
Association for Glycogen Storage Disease