Spring 2025

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

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*Thank you!*

## Strength in numbers!

Members are a crucial part of AGSD-UK. They help us increase our voice and have an important say in the way the organisation runs.

**Join or renew your membership today.**

It costs just £15 a year and benefits include the option to receive hard copies of Glisten magazine and a small AGSD-UK gift, plus discounts off future events. Setting up an annual subscription is a great way to pay or make a contribution and helps us manage our funds effectively.

Click or visit **www.agsd.org.uk/help-us-help/register-or-join/** to find out more

Front cover images:
From top to bottom:
McArdle's walking courses
Liam Coote post marathon
The Main Event 2024
Liam & supporter, post marathon

AGSD-UK recognises that not everyone is online and we don't want anyone to be excluded from the information they need. If we mention a website or include a link in this edition of Glisten that you can't access, please phone **0300 123 2790** so we can print and post it to you.

If you currently receive a hard copy of Glisten but would prefer to have a PDF version emailed to you, please get in touch at **info@agsd.org.uk** and we can save your preferences.

# contents

# Message from the Chief Executive

This year is the final one of AGSD-UK's current strategy, built around the things community members said would make most difference to their lives with the condition.

Over the past three years we've been focusing on making progress under the three strategy themes:

- Support across the community
- Help with living well
- Better treatments and service access

To facilitate community support we've developed our offer of online community groups, local gatherings and our popular ***Main Event***. You can find reviews on pages 8 and 25 of this edition.

To equip people to live well with GSD we've set up our regular ***Living well socials***, developed new information resources alongside our magazine, and provided crucial advocacy and support through our care and benefits advisors.

And to help ensure progress with research, the development of new treatments and service standards we've worked together with professionals, researchers, treatment industry partners and rare condition charities to promote engagement in GSD research, building evidence to support service improvement, access to effective treatments and better tailored studies that reflect community needs. There are details about current studies you can get involved in on pages 16 and 31.

As we move forward with the development of our next strategy it's crucial your views and priorities are front and centre. We're so grateful to those of you who've already contributed your experiences and perspectives by filling in our community survey, sharing the impact of the condition and the things that matter most.

Priorities emerging so far include a wish to see more local gatherings and we hope very much you'll take the chance to sign up for our local Pompe and hepatic GSD get-togethers which we move each year around the UK, plus the McArdle's walking courses - see pages 18 and 25 for more information.

The importance of support with benefits and access to equipment and adaptations has also come through, underlining the challenges faced by some community members. Please remember our expert advisors are always here to help, especially in these uncertain times - just contact **info@agsd.org.uk** to be put in touch and see page 22 for the latest on proposed changes to the benefits system.

Another area respondents have been highlighting is the need for increased understanding of GSDs from the public, professionals and friends and family. The efforts of community members who've recently been raising awareness by sharing their stories on social media are really appreciated and you can read more on page 32.

To make sure your views and experiences help shape AGSD-UK's priorities for the next three years please submit your survey response via the following link: **https://forms.gle/1QeRxKL56cE43NBh8**

Thank you!

*Val Buxton*



# Fundraising Round-up

**Thank you to our members who have been fundraising for AGSD-UK.**

In 2023, *Liam Coote* ran the London Marathon for AGSD-UK in memory of his friend Louise. He was back at it again, this time running in the Manchester marathon on the 27th of April this year.

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

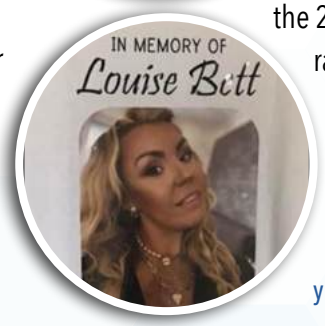
*I wanted to run in the Manchester Marathon 2025 in memory of Louise and I hope to raise as much awareness and money as I can to support this fantastic charity.*

https://www.justgiving.com/page/liam-coote

Liam has currently raised **£980** plus **£232** in Gift Aid

*Rav and Leanne Tank* are continuing to raise awareness and fundraise for AGSD-UK where they can. So far, they have raised **£6448** plus **£1454.50** gift aid.

https://www.justgiving.com/page/ravtank




*Tina Courtney* ran the 2025 London Marathon to raise money for AGSD-UK who support her two great nephews and their families. She has raised **£580.**

https://tinyurl.com/ybvwjwdb

*Allen & Ros Russell* collected **£900** for AGSD-UK at their camping meets. Their grandson *Mylo Moore* has GSD.

**£122** was raised in memory of *Ronald Tomkiss* whose great grandson has GSD.

# Fundraising Update Continued

*Susan Smith* raised **£570** in donations collected at the funeral of her son, *Mark Adam Tugby*.

*Lavinia Gibbs*, who has two children affected by McArdle's, donated **£500** in lieu of sending Christmas cards.

*Frank and Irene Green* raised **£1103.30** with their Christmas decorations which they do every year to fundraise for AGSD-UK.

At our ***Main Event*** this year, the raffle raised **£885.66.**

**Huge thanks to you 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.**




# Sioned Williams 'It Takes Guts'

In April we were treated to an evening of sublime music and engaging stories from renowned harpist ***Sioned Williams*** in the lovely setting of the National Liberal Club in Whitehall. Sioned worked tirelessly to stage and promote the event with Kettner Concerts in order to raise awareness of McArdle's and other GSDs and generate funds for AGSD UK.

We're extremely grateful to Sioned and husband Ali for all their dedication and hard work and privileged that Sioned shared her talents on behalf of the GSD community.

*"I was delighted to see a full-house at my harp recital. The evening was a story-telling of the 'ups and downs' of life in the fast lane as a musician with a disability. I exhibited items from my childhood, spoke, shared anecdotes, both highs and lows, and performed diverse solos, showcasing the versatility possible on the concert pedal harp. Through the music and stories, the audience were rapt with attention and were visibly moved when Florence Kinnafick, AGSD-UK Chair, spoke eloquently about our charity and the reality of living with very rare disabilities. I am deeply grateful to those who came to support from within AGSD-UK as well as friends and families who travelled such long distances to cheer me on. Thank you! I raised just over £4,000 in total for AGSD-UK, though I do believe that sharing the message was as important as any pot of money. The more we share, the more we can hope for in the future."*

https://www.justgiving.com/fundraising/sioned-williams28


# The Main Event...Reviews

**It was wonderful to see so many community members at November's *Main Event,* and to receive outstanding feedback about the weekend. The event was full to capacity with people affected by a range of GSDs and the evaluation showed that right across the board people felt it enabled them to:**

- Connect with others with similar experiences
- Gain tips about living with GSDs
- Become better informed about research and developments in the field of GSDs
- Get information about support and services that are available
- Have fun and enjoy themselves

*Here are just some of the comments received:*

It was a very valuable experience coming to this event with my family as not only did we meet people who are dealing with the same thing as us but also with professionals who mirror the work they do with our centre.

**It was superb. Team was great, very helpful. Would like to attend again.**

Big, big thank you to a fantastic team who put this event together. After a five year break it was good to return and reconnect with familiar faces and meet new ones.

This ***Main Event*** has made a significant impact by providing important resources, support, education for individual families affected ...

**Loved it - it was the best!**

Good mix of scientific updates, patent support and fun activities.

Great fun! Great accommodation and delicious food. This was my first time coming to a conference and I really enjoyed it.

At each conference I have been to there has always been something new to learn.

**Loved the music therapy on Sunday.**

**Thank you for another great conference!**

**Raffle was fab!**

A really important event that should continue.

Heartfelt thanks for an incredibly well-organised and impactful conference... The programme itself was so well curated, delivering both depth and relevance that kept us all engaged throughout the weekend...



**I really enjoyed seeing other people with GSDs.**

**Very good meeting. Well organised.**

We were very impressed with the organisation and the agenda.

Thank you for organising and managing such an impactful Main Event for AGSD-UK. Your hard work, attention to detail and dedication made the event run smoothly and successfully. The sessions were insightful. We truly appreciated it.

Thank you so much for looking after us. Your care and attention meant a lot. We're truly grateful for everything you do to keep us comfortable and supported. Thank you.

**Nice to catch up with old and new friends.**

**Loved the music session.**

**Excellent weekend - informative and fun. Thank you.**

Fantastic weekend. Thank you. Feel more supported.

Many thanks again to everybody involved with organising a great weekend of learning, meeting and generally having a good time.

This was our first time at the conference. Everyone was really helpful and supportive. Thank you so much.

# The Main Event 2024: A Researcher's Perspective

We're very grateful to the expert clinicians and researchers who gave their time to be involved in our *Main Event*.

Dr Sam Torrens, an applied physiology researcher specialising in metabolic exercise physiology and biochemistry at *Queensland University of Technology* shares his impressions from the perspective of a researcher into McArdle disease.

*I had the pleasure of presenting at the 2024 AGSD-UK event in Loughborough, an incredible opportunity for both personal and professional growth. As a researcher focused on rare metabolic diseases, particularly McArdle disease, the conference provided a valuable platform to promote my research and share it with people who could truly benefit. It was a unique occasion where individuals with GSDs, clinicians, and researchers came together, learning from one another, fostering collaborative networks, and discovering the latest advancements in GSD research.*

*The event was expertly run and well-organised, with every detail carefully thought out to ensure a seamless experience for all attendees. Accommodation, meals, and even entertainment were all included, which not only made the event highly educational but also enjoyable. It was a perfect balance of learning, networking, and relaxation, creating an atmosphere that encouraged deeper engagement with the topics at hand.*

*I would very much encourage other researchers and clinicians with a background or interest in GSDs to submit their abstracts and take the opportunity to present at future conferences. GSD research and healthcare is incredibly interdisciplinary, spanning fields such as neurology, biochemistry, physiology, genetics, physical therapy, nephrology, hepatology, and psychology, among others. The diversity of expertise brought to the table only enriched the discussions and learning throughout the event.*

*The conference had a strong sense of community spirit, teamwork, and altruism, which contributed to a motivating and inspiring atmosphere. It was a reminder of how important it is to work together as a community, and I truly look forward to being part of this event again in the future.*



# Managing Ketotic Hypoglycaemia in GSD

My name is Catherine and I am sharing my story of how my children and I all manage ketotic hypoglycaemia.

Growing up, until the age of 4, I didn't sleep through the night without having milk. I was constantly eating, much more than any of my friends but because I was an only child, my parents just thought I needed more food because I was growing. My dad was a good cook so there was always food ready to snack on.

I played lots of sports as I child and my first word was 'ball'. But I took a particular interest in tennis. I loved it and went from club standard to national standard very fast at a young age. By the age of 15, I had a world ranking, and I was training full time at a tennis academy. This was the age at which endurance fitness training began and the age healthy diets were recommended to help performance and become leaner. When I started a diet plan which was three meals a day, I started to feel shaky and weak by mid-morning and after endurance training, I would wake up multiple times during the night needing to eat. It was at this point, I started suffering from fatigue and aching muscles during long tennis matches. I started trying to eat more and more but couldn't reach the energy demands needed to maintain the sport at such a high level. It was this and also injuries that ended my short career as a tennis pro aged 17 but I was 1,208 in the world in women's WTA rankings, so I was proud of my achievements and to be honest I just wanted to be well. Because I still wanted to be part of the sport, I started tennis coaching but long days of this led to the same feeling of fatigue, weakness and even a couple of episodes of passing out during exercise. I had no idea what was happening, but I had a feeling it was low blood sugar. So, at age 20, I bought a glucose monitor. I took the readings to the doctor who said everything was normal but it kept happening. I didn't give up, I did my research and read about causes of hypoglycaemia. I managed to see an endocrinologist who did a liver ultrasound that showed mild enlargement of my liver and I sent these results to Salford Inherited Metabolic Unit. I then got referred and Dr

Hendrikz put a CGMS on me. It was there that we could see my blood glucose was dropping very low overnight and during exercise. I was tested for GSD and the results showed possible carrier status but were inconclusive, however I was still managed like I had GSD. Over the next few years, I continued to try to be involved in sports because it was such a big part of my life but I couldn't manage, my muscles were becoming much weaker.

I was then lucky enough to have my two wonderful children, Niall now 7 and Pearl, 5. It was when Pearl was age 2 after a bout of gastroenteritis that I noticed the same symptoms in her that I see in myself. Pearl was already under the Royal Manchester Children's Hospital due to her faltering in weight with a milk allergy and terrible reflux as a baby, so her paediatrician arranged an overnight fast. Pearl dropped during the fast and ketotic hypoglycemia was diagnosed. This led to the recommendation that Niall, my son, be tested on a glucose meter and he also had low blood sugar with very high ketone levels. They were both started on cornstarch therapy and have been under endocrinology and metabolics at RMCH since.

They both seemed to be doing well but in the last couple of years Niall has started having more hypos and much more fatigue, whereas Pearl continued to improve. It was then that I decided to come to the GSD Main Event. I wanted to listen to the talks to see if I could prevent Niall from getting these lows and help his energy levels. It was here that I got to speak with Dr Weinstein who I already knew lots about because of reading about his incredible work within GSD. He mentioned GSD 9a as a cause of our hypos/ symptoms. I've since given this information to our fantastic consultants at RMCH and they have recently tested Niall for this. He is now under a dietician and prescribed glycoside. He has also had fantastic input from an occupational therapist who has created a pacing plan for Niall at school.

The Main Event meant an incredible amount to me. I had always been on my own not understanding why food was a lifeline for me unlike anybody else, who just ate for pleasure. Nobody understood how I felt if I missed a meal or the exhaustion I felt after exercise. I met others just like myself and I listened to their stories which were similar to my own. I heard other CGMS monitors beeping with low glucose alarms just like my own, and my children played with others in the crèche who also needed extra snacks or feeds. I now feel like I belong somewhere, and I've been given emotional and social support by the GSD community since the event which has helped our whole family.

In 2020, I studied for a degree in prosthetics and orthotics. This is an unusual course that involves the biomechanics of movement with health. I wanted to use the medical knowledge I had gained through being an advocate for myself on my journey to start a new career helping others with medical conditions. I am now an orthotist in my local hospital and I help people walk with splints, braces and devices. When a patient comes into my clinic room needing help, I understand what it means to be in less pain, and how important it is to maintain their independence. And whilst my job still does cause fatigue and muscle pain towards the end of the week, I really enjoy it. It gives me a sense of purpose to help others and often puts life in perspective. In recent years, I have learned how to pace myself and since gaining this skill, I've achieved as much, if not more than anyone without a medical condition could achieve. In my experience if you manage a metabolic condition every day, you become a fantastic planner and decision maker because you do this day in day out to keep yourself well. This is something others can struggle with for their entire lives.

Whilst we await the results of Niall's tests which will take some time, we are going from strength the strength as a family now we have a multi disciplinary team looking after us at RMCH and at Salford Inherited Metabolic Unit. I feel comfort knowing we are already on the correct treatment plan even if the cause of our hypoglycaemia isn't found, but on the other hand, I feel like having a firm diagnosis can help our family plan for the future and help scientific research create cures for these rare conditions.

To all the GSD families or families with similar metabolic conditions, this isn't easy, but it makes us who we are. Listen to your body and work at your own pace and it doesn't matter how fast you go, forward is forward.

Looking forward to seeing you all at the next event!

# Get switched on

**The following information from *National Grid* may be particularly helpful for community members who rely on electricity to refrigerate medication or to power equipment. It includes how to join the *Priority Services Register* for extra support.**

With named storms passing through more regularly, it's good to be prepared for potential power cuts. Whatever the conditions, National Grid is here to keep the lights on and support you 24/7.

**Switched on advice**

Visit the ***Switched On Hub*** and you'll find lots of useful help and information about what to do if the power goes down. Plus, safety advice should you come across fallen power lines, or if your home is flooded. **https://tinyurl.com/2p9rxn5r**

**Our Switched On Hub also provides information on:**

- Fuel poverty support
- Our guarantees to you
- How to contact us

Sometimes we may need to visit customers. Our ***Arrangements for Access to Premises*** statement explains what you can expect from us when that happens.

https://tinyurl.com/mrs6jt2d

**Switch on to the Priority Services Register**

If you are elderly, have a disability, are chronically ill or rely on electricity to power medical equipment, you're eligible to receive extra support during a power cut by joining our ***Priority Services Register***.

It's free to join, simply call us on **0800 096 3080**. You can learn more about the PSR in this ***Special Services Statement***.

https://tinyurl.com/bdzae68r

***Switched On Hub*** ***Arrangments for Access to Premises*** ***Special Services Statement***



# GSD Travel International: An Idea Worth Considering

***Iris Ferrecchia* – AGSD US**

In May 2022 at the annual meeting of the ***Scandinavian Association for Glycogen Storage Disease, Markus Landgren***, MD suggested that it would be wonderful if young people with GSD could travel with more peace of mind as other young people do all across Europe. ***Sabine Schepler*** asked me if I would like to help her make this idea become a reality.

Since that day our advisory board has grown to include parents and young people from the Netherlands, Denmark, Austria, Germany, and the United States.

The board has consulted web designers, data security experts, liability experts, and the like. Zoom meetings have been held with interested parents and their children to assess the kinds of information they would need before travelling all resulting in the following suggested approach:

1. The individual with GSD, who wishes to travel would contact the GSD Association in their country (if they are participating) to confirm their eligibility. Diagnosis and note of stable health by their physician would be required.
2. Young people between the ages of 18 and 28 would be eligible to participate alone or with friends, which may add a level of safety.
3. The *GSD traveller* would be connected to an *advisor* in the area where they wish to travel.
4. The advisor in the destination location would provide information such as their experience with hospitals in the area, where to buy medical and dietary supplies, etc.
5. The advisor would outline the limits of their involvement. They may only want to be a consultant or they could choose to support during a medical emergency.
6. The travel would be at the traveller's own risk.

This idea has been presented at the German and Swedish associations as well as online in the United States with many people expressing interest.

If you are interested in participating in such an endeavour, please contact Iris Ferrecchia at *WhatsApp* 1-508-596-6846.

Erasmus MC
University Medical Center Rotterdam

# IPA / ERASMUS MC **POMPE SURVEY**

A worldwide investigation into the clinical condition of late onset Pompe patients

## What is the Pompe Survey?

*The Pompe Survey monitors the effects of Pompe disease on patients' lives, and how these may change with treatment. It collects this information directly from patients. Each year, patients from different countries fill in questions regarding physical problems, quality of life and treatment. The International Pompe Association (IPA) and the Erasmus MC maintain the survey, which started in 2002.*

## Why do we need the Pompe Survey?

*The information collected in the Pompe Survey can be used to compare the effects of new treatments, from the patient perspective. It also helps to inform physicians about the (changing) needs of patients. The international set-up provides the large participant pool needed for such evaluation.*

## Participating in the Pompe Survey

### Who can participate?

*All individuals aged 16 and over, with a confirmed diagnosis of late-onset Pompe disease, can participate. Children will also be able to participate soon.*

### Survey completion time

*Participating means to fill in a online or paper questionnaire of about 30-45 minutes every year.*

### Survey in different languages

*The questionnaire is available in English, French, German, Spanish, Italian, and Dutch.*

## GET IN TOUCH

**Erasmus MC**
pompe.survey@erasmusmc.nl
www.pompesurvey.com

**International Pompe Association**
info@worldpompe.org
https://worldpompe.org

Scan to participate

# Pompe Survey

Are you 16 or older and living with late-onset Pompe disease? Make your voice heard and help advance patient-centered research by participating in the *Pompe Survey*!

The *IPA/Erasmus MC Pompe Survey* is an annual online questionnaire that collects data on the personal experiences of individuals living with Pompe disease across a wide range of areas, including physical symptoms, quality of life, and current treatments. Since 2002, the survey has aimed to document the evolving needs of people with Pompe disease and assess the effects of treatment from the patient's perspective.

Research on patient-reported outcomes is essential to ensuring that care and treatments prioritise patient wellbeing. It also helps capture experiences that may not be fully reflected in clinical testing. As second-generation enzyme replacement therapy (ERT) becomes more widely available, the lived experiences of patients around the world are more valuable than ever.

Data from the *Pompe Survey* contributes to scientific research focused on assessing the impacts of treatment on Pompe disease and identifying potential unmet needs. Previous studies using this data have shed light on several important topics, including the natural course of the disease, the effects of ERT on survival, the impact of Pompe disease and ERT on daily life and quality of life, and highlighted fatigue as a key issue in late-onset Pompe disease.

In addition to supporting research, a future goal of the survey is to offer participants personalised feedback, enabling them to track their own results over time and monitor changes in their responses.

The *Pompe Survey* team welcomes voices from across the globe and is especially interested in hearing from individuals living in the UK. Participation is entirely voluntary and takes approximately 30–45 minutes each year.

Want to learn more or sign up to participate?

Scan the QR code on the opposite page or email **pompe.survey@erasmusmc.nl**

Web links:

Natural course of the disease
https://tinyurl.com/5n8amxbz

The effects of ERT on survival
https://tinyurl.com/3uyjnhej

The impact of Pompe disease and ERT on daily life and quality of life
https://tinyurl.com/yc6vdkv4

Significance of fatigue
https://tinyurl.com/mvh2rnj6


# McArdle's Walking Courses in Wales 2025

*Bronte Thomas*

We will be running our walking courses again for 2025, in beautiful Snowdonia (Yr Wydra). There will be a children and parents' course from 1st - 8th August, which can also accommodate younger adults aged 18-24, while our adults' course for those aged 25 and over will run from 9th-15th August.

We aim to add in some exciting walks and visits for both returners and those new to the course and to Wales We also hope to run some different choices of walks throughout the courses.

Regardless of your experience with the condition, or with hiking, it's an amazing opportunity to meet and share others affected and enjoy the great outdoors in a safe and comfortable environment, where everyone's journey matters. The only requirement is that you are able to get into second wind, and we can help you with the rest.

**To book, for the adults' course use this link:**

https://tinyurl.com/47r54zxy **and for the young persons' and children and parents' course, use this link:** https://tinyurl.com/4839r3ex

To discuss the costs or any other questions or issues, please contact Andrew Wakelin AGSD-UK McArdle's coordinator at type5@agsd.org.uk.

Here are some testimonials from those diagnosed with McArdle's who have been on previous walking courses:

"I really enjoy the cooking and the amazing walks that we do on the walking course. I really like the activities we do after the walks like bracelet making, playing games and paddling in a river! The friends that you make are incredible! I would definitely recommend."
*Sarah, 17, UK*

"Going to the walking course was the first time I met people with McArdle's and it represented for me the opportunity to exchange experiences, tips and every day situations with people like me. I really recommend, the atmosphere is nothing but kind and loving! Will definitely do it again."
*Elisa, 18, France*

"We learnt a lot about different aspects of McArdle's and were able to hold serious and sometimes funny discussions which would really never happen elsewhere. We met wonderful people from all over the globe, and we'll be connected now for ever. Thank you everyone who made this event so special. Already looking forward to 2025 in Snowdonia!" *Sioned, 71, UK*

# NAVIGATING SCHOOL LIFE WITH GSD

**My name is Anosha. I am 19 years old, and in my second year of university.**

To come this far in my life there have been many ups and downs along the way, however I have been taught that whenever you feel there is darkness there is always light at the end of the tunnel.

It all began a month before my third birthday. I started nursery specifically for special needs which my health visitor at the time, helped my mum get me in to. This was the best option for me as it allowed my mother to go back to work. I stayed in that nursery until I started primary school and whilst I was in that nursery I remember having a good time and making friends. However, I did have one instance where my naso-gastic tube was pulled out by another child, which was not a pleasant experience!

In the early years of my school life I was making good friends. However, as I was growing up in school my friends were getting harder and harder to keep which took a massive toll on my mental health. Throughout primary school I had a good experience with my teachers, especially my key worker who used to sit with me during lunch time. She would ensure I had eaten, as well as administering my feed when it was time during the school day. She supported me through my primary school years, and I am very grateful for her. Academically I was pretty good with English and moderately good at science however I was really struggling with maths so much so that I had intervention because I was finding it incredibly hard. The intervention lessons started as I was getting towards my SATS years, and the school also gave me extra time during my SATS exams.

During the summer holidays before starting year 7 at high school, my baby sister was born, which meant that it was one of the greatest summers of my life. I did however have my mini button changed under general anaesthetic as it had got stuck! After this my parents gave me my first phone. I was so excited, I felt grown up even though I was only 11!



When I started high school, I was really excited but nervous about making friends. Years 7 through 10 were great for me. I remember I had a good friendship group, good classes and decent teachers despite the fact I was still struggling. Unfortunately, just before lockdown I had a really bad argument with my friend group which caused us to stop being friends for good.

Lockdown was not a good time in my life. I was struggling mentally with the loss of my friends, being home all the time and not having anything to do. I did try to pick up some hobbies like scrapbooking, journaling and playing video games like Minecraft. When online classes started, I wasn't really paying 100% attention to them as I was unsure about whether we were going to go back to school.

After lockdown had eased and we went back to school, I received my GCSE results! I passed all my exams which meant that I could go to my choice of sixth form. I chose BTECH Health and Social Care.

When I stared 6th form, I was really excited for a change in my academic life as I chose a subject that I knew I could flourish in. Those two years were amazing, I bonded with my class and I was doing well in my subject. However, my health was up and down. I was having migraines every other week. I was sleeping for hours when I came home. This carried on throughout the two years. When it came to me applying for university, my health really played a part in my choices. I went to a couple of open days and it was good to be able to get feel for the journey from home to uni. I applied to UCL, Brunel, University of West London, London Metropolitan and London Southbank. I really wanted to get into either UCL or London Southbank. One day I went out with my auntie and had a bad migraine -so much so that I couldn't speak properly as if I was having a stroke. This situation really affected my choices. I received my BTECH results and was so happy as I achieved a D* D* D*. These top grades meant that I got all five my choices.

I talked to my family members, teachers and friends to ask them what university I should choose to go to as I really wanted to go to my first choice UCL. We decided to go for my local one which was the University of West London. I was gutted but it was a good decision due to my health.

I am now studying education studies at UWL and so far, so good and I'm looking forward to the rest of my course!

# Changes to the Benefits System

**New government proposals on welfare reform are contained in the recently published Green Paper: *Pathways to work: reforming benefits and support to get Britain working.***

**Liz Kendall** the Work and Pensions Minister has said the Green Paper reforms would help those who can work get back into work, while those who cannot remain protected. However we know the plans have caused anxiety among some community members.

The following ***Disability Rights UK*** summary explains the proposed reforms. The Government is consulting on some of the proposals while others will be voted on in parliament in June. There will be no changes to benefit entitlements until at least 2026.

You can find out more about the potential impact and ways to have your say by visiting **https://tinyurl.com/3azjkxht**

And remember our specialist advisors are here to help you with any queries or benefits applications. Just contact **info@agsd.org.uk**

*The Work Capability Assessment* (WCA) will cease in 2028. Instead, extra financial support for health conditions in *Universal Credit* will be assessed via the *Personal Independence Payment (PIP)* assessment and be based on the impact of disability on daily living, not on capacity to work.

Under this new system, financial support from PIP (non-means tested) and the health element of Universal Credit (means-tested) will both be non-work related.

However, the Government will also be legislating to guarantee that work in and of itself will not lead to someone being called for a reassessment or award review.

## Universal Credit

The Health Element top-up will be denied to under-22s to prevent a direct path from education to benefits. The top-up will also be cut for new claimants and frozen for existing ones.

The top rate of Universal Credit for the most disabled will be cut, but the standard rate will rise above inflation for the first time, reaching a £775 annual increase by 2029-30.

## Personal Independence Payment

The Green Paper proposes that PIP be **focussed more on those with higher needs** but will remain non-means tested.

However, there will be a shift to face to face assessment ***while ensuring we continue to meet the needs of our people who are claiming, who may require a different method of assessment (e.g., due to the need for a reasonable adjustment)***.

A new PIP eligibility requirement is proposed to ensure that only those who score a minimum of 4 points in at least one daily living activity will be eligible for the daily living component of PIP. This requirement would need to be met in addition to the existing PIP eligibility criteria.

This means that people judged to have lower needs only in the daily living activities (scoring 3 or less for each activity) will no longer be eligible for the daily living component of PIP. People with a higher level of functional need in at least one activity will still receive PIP.

This change will be introduced through primary legislation. It will apply to new claims and for existing people who claim, future eligibility will be decided at their next award review.

## A new contribution-based Unemployment Insurance benefit

The Green Paper asks views on replacing New Style Employment and Support Allowance (NS ESA) and New Style Job Seekers Allowance with one new Unemployment Insurance benefit. The rate of financial support would be set at the current higher rate of NS ESA. The benefit would not require a health assessment and would be based on an individual's National Insurance record as is currently the case. Those with long term health conditions, would also be able to claim other relevant benefits where eligible.

Disability Rights will be publishing further information and views on the Green Paper's proposed reforms.https://www.disabilityrightsuk.org/

*Pathways to Work: Reforming Benefits and Support to Get Britain Working Green Paper* is available at gov.uk (**https://tinyurl.com/9cwwwykp**)

# Global APBD Community Chat

**AGSD-UK are partnering with the APBD Research Foundation (https://www.apbdrf.org/) and Alex - The Leukodystrophy Charity (https://alextlc.org/) on a special on-line chat to engage the adult polyglucosan body disease community around the world.**

The chat event allows for globally dispersed people affected by APBD to connect with each other and join a growing community of advocates who are committed to helping each other through their medical journey and supporting more research possibilities.

To register for free, click here:

https://tinyurl.com/5dhuevw7

This chat offers people affected by APBD a safe and understanding space to:

- Connect with each other
- Share their common experiences, as well as the less common symptoms
- Hear from Sarah Williams about her APBD journey and advocacy efforts
- Discuss ways to navigate unexpected challenges
- Find out more about resources offered by the patient advocacy and research organisations

*"Having APBD can be lonely, frustrating, and frightening. Until recently, I knew no one else with APBD, apart from my brother. I am so grateful for this special program to connect patients from around the world."*

Sarah Williams, UK-based APBD patient and advocate.

*"The APBD Research Foundation's work is driven by APBD patients and families who take bold steps to shift from living in isolation to reaching out to others, to building a community, and joining forces for finding treatments and cures. I look forward to our conversations on June 25."*

Harriet Saxe: member of the foundation's board of directors and volunteer moderator of the patient chats.




# Online Groups & Community Get-Togethers

You can be sure of a warm welcome at our upcoming online group sessions and local meet-ups!

Take the chance to share experiences, get some mutual support and try out new therapeutic techniques by joining in one of our friendly online groups or coming along to one of our local get-togethers.

Upcoming sessions include our regular Living Well socials and men's groups plus a local Pompe gathering over afternoon tea in Reading and a meet-up for anyone affected by hepatic GSDs in the North West.

For more details of local get togethers and Zoom links for online sessions just email

info@agsd.org.uk

*"...the time went too quickly. An hour later, we'd made new friends, re-acquainted ourselves with others, and we know there's always someone there for us, and that we are well supported."*

*"We really enjoyed meeting the other families and sharing our experiences - it makes us feel that we are all on the same journey."*

*"Living with such a rare condition can be tough, especially when people don't always understand what we go through, so connecting with others who do was incredibly comforting."*

*Just...to express my appreciation for you arranging the 'Men with GSD' virtual group. I found the meeting to be very informative, listening to the stories of others in the group - their symptoms, their diagnosis, life's challenges...I look forward to attending more group sessions...*

*The group has made me feel stronger because I feel supported by others who understand what I'm going through. When others feel down, there is always somebody who will listen. If I have any questions there is always somebody to reach out to.*

## Forthcoming Dates

**Pompe local get-together:** Saturday 7th June 2.30pm **Reading**

**Living Well Social:** Thursday 19th June **online**

**Men's Group:** Wednesday 9th July 7.00pm **online**

**Hepatic local get-together:** Saturday 20th September **North West**

# Muscle GSD's Research Update

Dr Sam Torrens is an Applied Physiology Researcher in Metabolic Exercise Physiology and Biochemistry at ***Queensland University of Technology***. Sam has recently had the following research paper published.

## McArdle disease and carbohydrate ingestion before exercise: Timing on exercise tolerance, clinical relevance, and application to real world settings

Sam L. Torrens, Evelyn Parr, Craig Mcnulty, Lynda Ross, Helen L. MacLaughlin, Liza Phillips, Robert A. Robergs. Sports Medicine and Health Science, Available online 3 March 2025

This research considers the optimum timing for ingesting sucrose (carbohydrate) prior to exercise and its limitations in terms of the practical benefit it may bring to those with McArdle disease.

### Background to the research:

Carbohydrate ingestion before exercise is a common management method advised for individuals with McArdle disease. Current Clinical Practice Guidelines advise the ingestion of 37g of carbohydrate 5-10 minutes before exercise for significant improvements in exercise tolerance. This is based on the theory that increasing blood glucose availability to the active muscle during the early stages of exercise will partially offset the blockage in muscle glycogenolysis that severely limits glycolytic activity in McArdle disease.

### Main Outcomes:

- Improvements in exercise tolerance (as measured by heart rate and rating of perceived exertion) were greater when the time between ingestion of carbohydrate and the initiation of exercise was increased from 5-minutes before exercise to 25-minutes before exercise. This is because increasing the time between ingesting carbohydrate and initiating exercise allowed more of the ingested carbohydrate to enter the blood stream and become available to the exercising muscle.
- Blood glucose levels were significantly higher at the beginning of exercise for carbohydrate ingested 25-minutes before exercise compared to ingestion 5-minutes before exercise.
- Because the average exercise tolerance (i.e., ability to produce power as a unit of physical work) is so low during research studies involving individuals with McArdle disease (<50 Watts), the application of findings to real world settings is limited as most activities of daily living (i.e. walking on a flat surface or walking upstairs) far exceed the power outputs achieved by participants in research trials. This therefore raises questions regarding the practical application of carbohydrate ingestion before exercise for IWMD, even when statistically significant results are reported in research findings.

### Practical implications:

- Waiting for a longer period after the ingestion of carbohydrate (25-30 mins) before you start exercise is better than the currently advised 5-10 minutes.
- There may be very little perceived benefit from ingesting carbohydrate before exercise in real world settings for individuals with McArdle disease due to the very controlled conditions in which lab experiments take place.

Sam wanted to emphasise that the conclusions of the paper do not advocate for carbohydrate ingestion before exercise, but they do point to a more appropriate timing for carbohydrate before exercise if people want to try it. This is of course with that caveat that it should not be used regularly (due to the potential for weight gain), and instead the best current management method available is safely prescribed exercise to increase the individual's aerobic capacity.

Another take away point from this research which may have relevance to those with McArdle disease who have higher exercise capacities (e.g., runners, cyclists) and regularly train to increase this capacity is the finding that:

*"….despite IWMD having no access to muscle glycogen stores, the participants of the current trials still displayed a high relative contribution to total energy expenditure from carbohydrate…. While participants in the current trial exercised at what would be considered a low absolute intensity, clinicians and exercise professionals working with IWMD should be aware of the potential for exercise-induced hypoglycaemia, particularly for individuals with a greater exercise capacity working at higher absolute exercise intensities and/or prolonged exercise periods."*

https://tinyurl.com/24rmyhvj

# Continuous Glucose Monitors and GSDs

## What is the current state of play and will they become commonplace for people with a hepatic GSD?

Continuous glucose monitors or CGMs are trending. These little white discs are everywhere. They can be spotted on the arms of celebrities, politicians and people following diets.

AGSD-UK is often asked for help or advice concerning CGMs. This article tries to answer a few of those questions, highlight the path to their future and look at the work that goes on behind the scenes.

A CGM is a device that allows you to check your blood sugar level at any time and with ease. Should people with a hepatic GSD be wearing one?

Currently CGMs are only available on the NHS to people with type 1 diabetes and some people with type 2 diabetes. They are not licensed for use by people with GSD.

A few people with a hepatic GSD have been given a CGM. These are usually given in unique circumstances, where the medical need of an individual is exceptional. To obtain one a doctor will make an individual funding request (IFR) application. This is a lengthy process and a doctor's request can be refused.

## Behind the scenes - how a drug or device achieves a licence:

The pathway for any drug, device or treatment, before it is available through the NHS, is roughly, as follows and routine CGM use for people with a hepatic GSD will have to follow this route:

1. A drug or device will go through studies and clinical trials.
2. In the UK, the Medicines and Healthcare products Regulatory Agency (MHRA) will assess the trials to see if there is robust evidence that the product is safe and effective. If passed, a licence will be issued.
3. After the product has been licensed, other organisations will need to approve the drug or device before it is made available through public funding. They will look at how well it works and cost effectiveness.

## These organisations are:

- NICE | *The National Institute for Health and Care Excellence in England*
- SMC | *Scottish Medicines Consortium*
- AWMSG | *All Wales Medicines Strategy Group*
- DHSPSS | *Dept of Health, Social Services and Public Safety in Northern Ireland*.



Following approval by these bodies the treatment should be available on the NHS. Even after this process, access may occasionally be affected by local policy.

Currently CGMs for GSD are still being studied to assess their effectiveness. There have been several papers written. The quotes from this selection of papers suggest positive indications for CGMs when used in a monitored and supported fashion.

AGSD-UK is keen to see CGMs made available through the NHS where there is evidence to support their use and individuals could benefit, working with community members health professionals and researchers to support this outcome.

## Exerpts from some the studies on CGMs and GSD

1. **Role of continuous glucose monitoring in the management of glycogen storage disorders** (Mrudu Herbert et al. 2018)

Research carried out at Duke University concludes:

*'This study revealed that in addition to overnight hypoglycaemia, CGMS can uncover previously undetected, subclinical, low glucose levels during daytime hours. Additionally, the CGMS detected daytime and overnight hyperglycaemia, an often overlooked concern in liver GSDs. The CGMS with concurrent dietary adjustments made by a metabolic dietician improved metabolic parameters and stabilized blood glucose levels.'*

https://tinyurl.com/mw42edfz

2. **A retrospective in-depth analysis of continuous glucose monitoring datasets for patients with hepatic glycogen storage disease: Recommended outcome parameters for glucose management.** (Fabian Peeks et al. 2021)

The authors of this study in the Netherlands state:

*'We conclude that in-depth CGM analysis can be a powerful tool to assess glucose management and optimize treatment in individual hepatic GSD patients.'*

https://tinyurl.com/mucx987t

3. **The use of continuous glucose monitoring in the practical management of glycogen storage disorders.** (Fiona A White, Simon A Jones. 2011)

This team from the Willink in Manchester describe CGMs as a reliable and valid tool for assessing blood sugar management in 'real life' settings, unlike hospital management.

https://tinyurl.com/5fyj82zn

4. **Personalized management of hepatic glycogen storage disorders: The role of continuous glucose monitoring.** (Ambika Gupta et al. November 2024)

*'CGM is an efficacious adjunct in the personalized management of hepatic GSD patients, in the Asian Indian population. The study also underscores the need for long-term follow-up to determine the role of glycemic management in growth, general well-being, and metabolic control in the GSD subtypes.'*

https://tinyurl.com/b9c473wc

5. **Glycemic control and complications in glycogen storage disease type I: Results from the Swiss registry.** (Nathalie Kaiser et al. 2019)

*'The quality of glucose control is related to the presence of typical long-term complications in GSDI. Many patients experience episodes of asymptomatic low blood glucose. Regular assessment of glucose control is an essential element to evaluate the quality of treatment, and increasing the frequency of glucose self-monitoring remains an important goal of patient education and motivation. CGM devices may support patients to optimize dietary therapy in everyday life.'*

https://tinyurl.com/2r449baj

6. **Continuous glucose monitoring metrics in people with liver glycogen storage disease and idiopathic ketotic hypoglycemia: a single centre, retrospective, observational study.** (Overduin et al. 2024)

In a study at Groningen in the Netherlands of 47 people with GSD, the authors comment that-

*'Several studies have shown the potential of continuous glucose monitoring (CGM) for people with liver GSDs, reducing the duration of hypoglycaemic episodes and unveiling unrecognized/asymptomatic hypoglycaemia'*

This observation led in part to their study and their recommendations, illustrated by the following quote-

*'Despite dietary treatment, most liver GSD cohorts do not achieve CGM metrics comparable to healthy individuals. International recommendations on the use of CGM and clinical targets for CGM metrics in liver GSD patients are warranted, both for patient care and clinical trials.'*

https://tinyurl.com/mr3zpvd6



Loughborough University

AGSD UK
Association for Glycogen Storage Disease

# HOW DO HEPATIC GSDS AFFECT WELLBEING?

## Survey for people with a hepatic GSD and their carers

**What is the study about?**

- We want to find out more about the mental, social and physical wellbeing of people with a hepatic GSD and their carers, to identify areas where there could be more support
- There will be two follow-up surveys, one month and one year after the first
- The first survey will take 15-25 minutes to complete, the second will take 5-10 minutes, and the third will take 15-25 minutes

**Why take part?**

- Taking part will improve our understanding of living with a hepatic GSD and may contribute to better support in the future.
- Participants that take part in all surveys will be entered into a prize draw for one of three £50 vouchers

**Who can take part?**

- Adults (18+) with a hepatic GSD, types 0, 1a, 1b, 3a, 3b, 3c, 3d, 6, 9a/PHKA2, 9b/PHKB and 9c/PHKG2)
- Carers of someone with a hepatic GSD
- Carers can also choose to invite their adolescent (12-17) with a hepatic GSD to take part

**TAKE PART:**

bit.ly/GSD-Wellbeing

Contact the researchers:
a.potter@lboro.ac.uk
f.e.kinnafick@lboro.ac.uk

# Community Stories From Rare Disease Day & International Pompe Day

**February 28th 2025, was Rare Disease Day and our GSD community members took the chance to share what they wished people knew about living with the condition. Here are just a few of the powerful personal stories that were shared.**

Hear from Katie and Harry, whose son Teddy is affected by GSD3a:

*"Rare Disease Day is an opportunity to share our story, raise awareness, and remind families like ours that we're not alone. Greater research, understanding, and support can shape a better future for children like Teddy.*

*GSD3a is part of Teddy's life, but it doesn't define who he is. He is strong, resilient, and full of potential. With the right care and awareness, he, and others like him, can continue to thrive.*

### Teddy's Journey with GSD3a

This is Teddy. He's four years old, full of fun, with a love for cars, cats, Lego, trains, playing with his cousins and having fun with his friends. Teddy has a rare genetic condition called Glycogen Storage Disease Type 3a (GSD3a), which affects how his body stores and releases energy, making everyday life more challenging.

GSD3a is a metabolic disorder that prevents the body from breaking down glycogen into glucose - the fuel our bodies need. This affects his liver and muscles, leading to low blood sugar, muscle weakness, and a strict diet that must be carefully managed around the clock.

Looking at him, you might not realise he has a rare disease. But behind the scenes, his care is constant - watching the clock, managing his diet, tracking carbs, and ensuring he eats the right foods at the right times. If Teddy's blood sugar drops, he is at risk of hypoglycemic seizures, which are life-threatening for him. He cannot fast like most people, so he must consume food every three hours, either orally or via his G-tube. He also undergoes 'corn starch therapy' which is drinking a mixture of cornflour and water several times throughout the day to help maintain his blood sugar levels. At night, he relies on a feeding pump to keep his blood sugar stable.

His muscles are also affected. Activities like going on walks, climbing, running, or even standing up for periods of time can be harder for him, and he tires more easily. But Teddy enjoys life every day, and we hope his condition will never hold him back.

Despite these challenges, he is thriving and has now started school, which he loves. He's figuring things out, making friends, laughing, and having fun. He has great confidence and talks about the things he loves, and even about his time spent in the hospital. For him, hospital admissions feel normal; they are all he knows. His resilience is inspiring.

Living with GSD3a isn't just about medical management, it's about raising awareness and building a supportive community. Many people, including medical professionals, have never heard of GSD, making daily life even more complex. Explaining his condition to teachers, caregivers, and doctors is a constant effort.

That's why Rare Disease Day is so important. It's a chance to share Teddy's story, raise awareness, and remind families like ours that we're not alone. Research, understanding, and support can change the future for children like Teddy.

Teddy may have GSD3a, but it doesn't define him. He is strong, brave, and capable of so much more than his condition. With the right care and awareness, he, and others like him–can continue to thrive.

### Maryam talks about The Silent Battle: Living with GSD Type 1a

Imagine living in a world where your body doesn't have the energy to function like everyone else's. A world where food isn't just a comfort, but a lifeline- one missed meal could send you into a crisis. This is the reality of Glycogen Storage Disease Type 1a (GSD1a), a rare genetic condition that has shaped every aspect of my life.

Most people don't think twice about going hours without eating, but for someone like me, that isn't an option. My body lacks the enzyme needed to release stored glucose, which means my blood sugar can drop dangerously low within hours. The simplest things- sleeping through the night, forgetting to eat, or getting sick- can turn into life-threatening situations. I live on a constant schedule, tied to strict feeding routines and medical interventions just to survive.

Growing up, I struggled to explain my condition. When I was younger, all I wanted was to be like the other kids- to join sleepovers without fear, to play

outside without the exhaustion creeping in, to eat what I wanted without worrying about my blood sugar crashing. But GSD 1a doesn't give you those choices. I had to learn responsibility early, always carrying emergency snacks, knowing the warning signs of a hypoglycemic episode, and navigating a world that didn't understand what it meant to live with an invisible illness.

The hardest part isn't just the physical toll it's the isolation. How do you explain to people that food, something so simple for them, can be your greatest fear and your greatest need at the same time? That feeling of being different, of watching others live freely while you calculate every bite, every hour, every risk.

But despite the struggles, GSD1a has made me resilient. It has taught me the power of advocacy, of speaking up for those who live with the rare conditions that the world often forgets. I've had to fight for proper care, for understanding, for awareness. Every hospital visit, every emergency, every sleepless night has shaped me into someone wo refuses to be defined by an illness.

This *Rare Disease Day*, I want people to know: *We are not fragile - we are fighters. Our journeys may be different, but our strength is unmatched. If you've never heard of GSD1a before, let this be your reminder that rare diseases deserve attention, support, and most importantly, compassion.*

*To those living with GSD, to the parents staying up at night ensuring their child's blood sugar stays stable, to the friends and family who stand by our side- you are not alone. Together, we can make the world understand that rare isn't just a label. It's a story of courage, perseverance, and hope*

*Let's raise awareness, not just today, but every day. Because our stories matter.*



## Jeanne shares her experience of living with McArdle's:

*"What I would like people to know about living with this rare condition.*

Firstly - this condition it's not an obvious problem, it's invisible to anyone other than the person it affects, but this changes once it's more painful and disabling symptoms become apparent. McArdle sufferers don't know what normal energy levels are, we manage our everyday life, and it becomes second nature to assess everything we do. We never know when we have 'done too much' until it's too late and the muscles' reaction to the stress put upon them is always a painful and often debilitating experience, so we learn from experience what to be wary of.

Before diagnosis we are floundering and wondering why we can't kneel or bend, carry shopping and even struggle with food prep. We can't understand why we can't walk anywhere without the need to stop - until diagnosis, we aren't aware of the 'second-wind' thing! We know that we get a second wind and are always grateful to get it, as prior to this 'kicking in' - we hit a wall, this is not only a dreadful feeling, but it also heightens our vulnerability - there's nothing worse than being away from home and hardly being able to put one foot in front if the other. It's stressful and embarrassing, especially when young, trying to keep up with 'normal' people while walking in everyday situations (I'm not talking about hiking or walking marathons, just trying to get to the shop, bus stop or train). Being mobile and staying safe is a difficult challenge - our legs feel like lead and our hearts race, the feeling we have is best described running on empty and is hard to describe. It is a condition that I was diagnosed with in the early 1970's via a muscle biopsy.

Although this condition affects me it impacts on my whole family - they understand the need to manage this condition as much as I do, and they are there to step in and take care of me when I get it wrong and lose the ability to be mobile, have use of hands, arms or legs, even fingers don't like to be left out and often rebel against carrying plates! So, this is a small insight into living life with a rare genetic condition that can't be treated or cured, it can only be managed. Some days more successfully than others."

**Hear from Olivia, whose daughter is affected by Pompe**

**Hear from Mohid, who talks about his experience with GSD1b**

April 15th was International Pompe Day, and community members got involved in raising awareness on social media about what a typical day with Pompe might look like and what they find most helpful.

### Doris shares her reflections on everyday life: My Life in the Slow Lane

What is my day living with Pompe muscular dystrophy like? The headliner words that come immediately to my mind when I was asked to ponder this question for International Pompe Day 2025: snail mail, slow coach, go slow.

Self-pity wells up in me during those moments of self- reflection. My diagnosis is Late Onset Pompe. It is visceral when I think of how much my boundless ideas-generating mind has to be whittled back into the only format my body can cope with: 60% of physical effort. However, this

sound clinical advice to let the Borg Rating of Perceived Effort rule my daily activities helps me to navigate fatigue and accept my physical limitations.

The effect of these limitations is felt in all areas of what makes me love my life: family, work, hobbies/leisure time. My percentage of perceived effort is not static and over the years it is sliding down, down, down. My better days and weeks are when I stick to 50%. Accommodating my life within those parameters has forced me to give up some areas, like paid work.

I struggle to communicate adequately how this 'slow life' is an essential and integral element of my Personal Independence Payment (PIP) allowance. My Pompe brings to the fore the tricky application of PIP's eligibility criteria of "timely, repeatedly and safely" when it comes to assessing a daily activity. The Borg scale, with its focus on perceived effort allows you to listen to your body. It allows subjectivity and at the same time integrates this subjectivity into a clinical framework. I find this sliding scale builds routes to self-compassion and self-care between my body and my mind.

Feeling that I am living a 'reduced' life is a tough ask for someone who rather enjoyed giving 110%! Alas, that was decades ago but I do remember the glowing feeling it can give. After all, slow can be viewed positively too, right? There are whole movements out there: slow food and slow holidays come to mind. They have a focus on prioritising a relaxed, mindful approach. A lot of enjoyment can come from savouring experiences and connecting with your activities.

I tell myself that 'slow is ok and better for your Pomp body' in many ways and often. For example, when sitting on my perching seat in my throughfloor lift with the journey upstairs taking ages. I am immensely grateful for Occupational Therapists who have the capacity to take the long, slow view and help me eliminate the dreaded activity of climbing stairs out of my life.

### Claire writes about the difference exercise makes for her:

There are many daily challenges of living with Pompe Disease.

I enjoy participating in my weekly online physiotherapy exercise classes, which have continued to aid my mobility, improve my fitness, and improve my overall quality of life. In addition to the physical benefits, I really feel more positive mentally after the classes.

It is valuable to meet others with Pompe and hear about their experiences whilst exercising with them. It feels good to be doing something constructive to try and help lessen the progression of my illness.

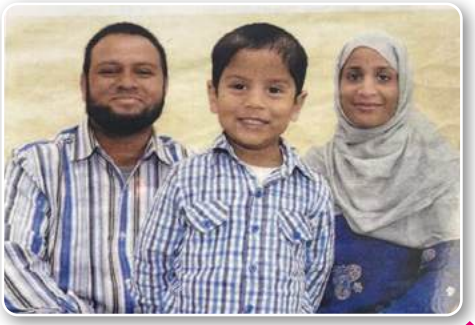
### Hear from Quasim about his experiences:

I'm Qasim and I live in Portsmouth. I'll be turning 18 this July. I'm currently studying Business Studies at college and hope to continue my education further in the future.

I was diagnosed with Pompe disease just a few days after I was born. My first infusion was at only 7 days old at ***Great Ormond Street Hospital*** (GOSH). I was one of the very few children in the UK diagnosed with infantile-onset Pompe disease and was the first child in the country to start ***Myozyme*** infusions from birth.

I don't remember much from those early years until I was around four, but my mum often shares stories about those tough early days. She and my dad have both shown so much strength and love through everything. From the long hospital stays to the emotional rollercoaster of the unknown, they never gave up. Their strength has always inspired me to keep going, even when things felt difficult.

I've recently passed my driving test, and I feel amazing! It means so much to finally



be independent – especially since my parents have been my "taxi service" for everything over the years. It feels like a big step forward, not just for me, but for them too.

I also want to say a massive thank you to AGSD-UK for all the support they've given me and my mum over the years. They've always been there, offering help and guidance, and it really means a lot to us.

Thanks to the incredible care from the metabolic team at GOSH, I've come such a long way, and I'm now at the final stages of transitioning into adult care.

I always try my best to get involved in everyday activities. I love swimming – it's something I really enjoy. I also love football, but unfortunately, due to wearing leg braces, I've had to give it up, which has been hard.

But despite the challenges, I keep going and stay positive. I'm so grateful for how far I've come, and I wouldn't be here without the strength, sacrifices, and endless love of my parents. They've been my biggest supporters, and I hope to make them proud as I move into the next chapter of my life.

**Thomas shares his reflections on everyday life:**

Pompe disease is a rare genetic disorder caused by the deficiency of the enzyme acid alpha-glucosidase, leading to the accumulation of glycogen in the body's cells, particularly affecting the muscle and heart tissues.

Living with Pompe disease has profoundly impacted my life, as I navigate the challenges of muscle weakness and fatigue that can hinder daily activities and limit my independence. The emotional toll of managing a chronic condition is significant, often accompanied by feelings of uncertainty about the future.

However, I have also found strength in the support of my wife, family, healthcare team, and the Pompe community, which has fostered resilience and a deeper



appreciation for the small victories in life. Through ongoing treatment and therapy, I strive to maintain my quality of life, embracing each day with fight, determination and my faith in God. *Romans 8:18*

I consider that our present sufferings are not worth comparing with the glory that will be revealed in us.

**To check out all the fantastic *Rare Disease Day* and *International Pompe Day* posts or to stay up to date with what is happening within the GSD community and any upcoming events, please visit and follow our Facebook and Instagram page:**

https://www.facebook.com/AGSDUK/

https://www.instagram.com/agsd_uk?igsh=enN6aXIONjB0ampv

# My Family's Journey with GSD IV

Adult Polyglucosan Body Disease, the late-onset form of GSD IV and Andersen Disease, the early-onset form of GSD IV share the same underlying root cause – changes in the GBE1 gene which causes deficiency of a protein called glycogen branching enzyme (GBE).

Deficiency of this protein leads to dysfunctional glycogen storage and metabolism, and the build-up of harmful substances called polyglucosan bodies in different tissues.

Given that these ultra-rare conditions are essentially different manifestations of the same biochemical problem, there is a powerful argument for our APBD community and the broader GSD IV community to work together to identify patients and families, build power and strength in numbers, and advance research and therapies in development together.

*APBD Research Foundation*

**Amber Robertson worked with the APBD Research Foundation in the USA to share her GSD IV journey to increase the profile and understanding of the condition and help more than 25,000 individuals affected globally. To learn more about the APBD Research Foundation, visit apbdrf.org.**

Even before Mia was born, I could sense that something about her was different. With my other children, I felt constant movement during pregnancy, but Mia stayed curled up in one spot and hardly moved at all. When I mentioned this to my obstetrician, he assured me that it was normal. But I was sure that there was something more going on.

As soon as Mia was born, there were signs that she could have a medical condition. She had a skin tag on her ear, tightness in her neck on one side, and a broken growth plate below her femur. Mia stayed in the neonatal intensive care unit (NICU) for a few days because she was struggling to breathe. These symptoms led the doctors to test her for a genetic condition called *Turner Syndrome.* We waited three agonizing months for the test results, only to hear that they were negative.



My husband and I were relieved at first, but we quickly realized Mia's symptoms were still unexplained. She wouldn't startle, couldn't raise her arms above her head, and didn't pass her hearing test for her right ear. When we brought these concerns to her pediatrician, he reassured us and promised to reevaluate her at two years of age. But my intuition told me that if we waited, we'd miss something critical.

Doctors are taught: "When you hear hoofbeats, think of horses, not zebras." It's a way of reminding them to consider common explanations first rather than spend time searching for rare conditions. But what if your child is a zebra?

Before Mia was 5 months old, I absolutely knew that if she was going to get the care she needed, I'd have to be her fiercest advocate. I left my job of 11 years to focus solely on her care. My entire life shifted from being a working mother into a full-time caregiver. When I voiced my concerns to Mia's orthopedic surgeon, I finally felt heard. She agreed that Mia's case warranted further investigation and referred us to specialists in neurology and genetics.

When we saw the neurology team, they recognized Mia's very low muscle tone and admitted her to the hospital for failure to thrive. An MRI revealed an underdeveloped cerebellum and a thin corpus callosum, which can cause motor and speech delays. From there, we were fast-tracked to the genetics team. They ran additional genetic tests to look for what could be causing Mia's symptoms. After four torturous weeks of waiting, we finally had an answer: Glycogen Storage Disease Type IV.

GSD IV is an ultra-rare condition caused by changes in the GBE1 gene and is inherited in an autosomal recessive manner. It is a disease spectrum – with early-onset and late-onset symptom presentations – that occur when the body has low activity of a specific protein needed to make glycogen, a stored form of energy. The abnormal glycogen does not have the correct shape, causing it to build up into clumps called polyglucosan bodies. The early-onset (or "pediatric form") of GSD IV presents at different stages from in utero to adolescence, causing liver, neurologic, muscle, and heart manifestations. The adult-onset form of GSD IV – referred

to as adult polyglucosan body disease (APBD) – presents as early as in the mid-30s with primarily neurologic and muscle manifestations.

Even with a clear diagnosis, the challenges didn't end there. Getting the equipment Mia needed, like her feeding chair, CPAP machine, electric wheelchair, and scoliosis brace has been a battle. I've had to research everything myself, seeking out the right devices and asking for them. One of the most difficult obstacles we have had to deal with is our limited access to resources. The distances we travel each week – four hours each way to the hospital in Sacramento and three hours round-trip for occupational therapy in Redding – have drained us both financially and emotionally. For example, finding a physical therapist with expertise in these metabolic disorders was especially challenging. For now, Mia receives physical therapy via virtual appointments; this is a challenge because the therapist can't physically hold her.

The unknowns are the hardest to bear. Although I have learned a lot in the last two years about GSD IV, I still have questions, and I'm realising that even the experts don't have all the answers. No one can tell us if her liver will be affected, if she will ever walk, or her expected lifespan. Most of what I've learned about Mia's condition comes from other families affected by it. Families who, like us, are becoming the experts on GSD IV themselves.

For Mia's sake, I've started advocating in our community, pushing for wheelchair accessibility in our schools. They've already modified the kindergarten playground, and I'm working with the schools to get the bigger playgrounds accommodated. I want the schools to be ready for Mia by the time she gets there. She deserves to feel included, not defined by her differences.

If I've learned one thing, it's that being Mia's mother means being her voice and her constant support. I've gained a depth of medical knowledge I never thought possible and discovered a strength I never realised I had. For anyone else on a similar journey, my advice is to speak up. Don't be afraid to advocate for your child, even if it feels uncomfortable or intimidating. Our children need us to fight for them, and we parents can't be silent.



# TAKE AWAYS FROM THIS EDITION

- Get involved in the Pompe global survey or Loughborough research on hepatic GSDs
- Register for upcoming local hepatic and Pompe get-togethers and the McArdle's walking courses
- Join our online *Living Well Social*, men's group or APBD global chat
- Fill out the community survey to help shape AGSD-UK's priorities

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, The D'Oyly Carte Charitable Trust, Beam, National Lottery Community Fund, Mosawi Foundation, Sanofi, Ultragenyx and Vitaflo.

Amicus Therapeutics

The D'Oyly Carte Charitable Trust

Beam Therapeutics

The Mosawi Foundation
Ex Caritate Spes

sanofi

Community Fund

ultragenyx pharmaceutical

Vitaflo Innovation in Nutrition

AGSD UK
Association for Glycogen Storage Disease


## Upcoming events

- Pompe local get-together 7th June 2.30 | Reading
- Next Living Well Social 19th June | Online
- Adult polyglucosan body disease international chat 25th June | Online
- McArdle's walking courses 1st-8th Aug | Wales
- Hepatic get-together 20th Sept | North West

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 2025

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