Autumn 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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Charity Number :1132271

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 a direct debt is a great way to pay 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:

Roman Tank, The Pompe get-together, Kerrie and Dom, The McArdle's experience, Amanda Holthaus daughter

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.

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Message from the Chief Executive

With the end of the year fast approaching, this can be a time to reflect, plan and make some new resolutions.

Reflections can sometimes be painful, but they can help us acknowledge issues we might need help with, think about ways forward and identify things to celebrate and build on.

For some community members looking back on this year, money worries have added to day-to-day challenges and have been heightened by threatened welfare changes. Some planned welfare reforms have now been influenced by people affected speaking out about their impact and you can find an update on page 9.

With more potential changes on the horizon it's important to have a voice, particularly in the review of Personal Independence Payment that will help shape rules on future eligibility. And if you've been putting off applying for benefits because the process can seem daunting, make this the time you ensure you get all the financial help you're entitled to. Just contact info@agsd.org.uk so one of our expert advisors can support you every step of the way.

This year our community survey has highlighted the many other stressors people face, from family relationships or housing problems to concerns about school or employment. Our advisors can help with practical support and advocacy on a whole range of issues and are ready with emotional support as well, so please don't wait to get in touch.

Anyone who ever feels stressed and anxious can take the chance to join our online anxiety workshop on 23rd November (see page 13). You can be sure of a warm and relaxing Sunday afternoon welcome, along with useful tips to help you cope.

Many parents of children affected by GSD will recognise the anxiety powerfully expressed in stories from Amanda and Laurence reproduced on pages 14 and 36 of this Glisten edition. But they will also relate to their deep appreciation of every positive life brings and the way they draw on these to give them strength.

Like so many community members Amanda and Laurence deploy this strength for the benefit of others affected, getting involved in advocacy and sharing their stories to offer hope and raise awareness of the condition. Others are motivated by their experiences to become determined fundraisers for the cause, like those featured in our fundraising section. We're hugely grateful to everyone around the world who contributes in any way to improving life with GSD.

When you reflect on this year we hope you draw strength from every pleasure and achievement, while taking time to think about where you could benefit from support and resolving to get this. Whatever your resolutions for 2026, AGSD-UK wishes you all good things and, thanks to all your efforts, will continue to be here whenever needed.

Val Buxton

Fundraising Round-up

Thank you to all the community members who work so hard to fundraise for AGSD-UK.

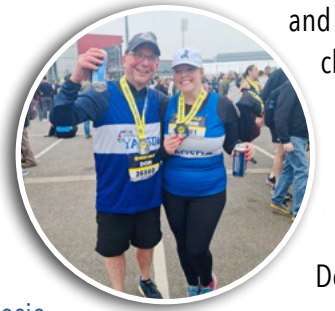
- **£130** was raised in donations in memory of Rosie Thompson.
- Alexandra Fraser donated **£100** after raising money on International Pompe Day.
- Trevor Ford donated **£100**

Running for Roman

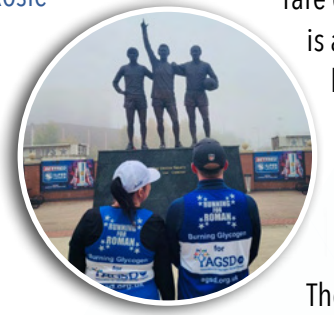
Kerrie Shakeshaft

On Sunday 12th October 2025, my running buddy, Dom and I took part in the Manchester half marathon. The city is close to my heart as I was born there. Upon signing up, we made the decision that we would be *Running for Roman* my best friends' 2 3/4 year old, who has the ultra rare disease, GSD3.

We set out on our mission to raise the profile of the charity and much needed funds, to continue the amazing work AGSD-UK is already doing for Roman and all the people affected by glycogen storage disease. I am delighted to say we raised over £1000 and by proudly wearing our charity running vests, more than 140,000 spectators who lined the 13.1 mile route, now also know about the charity.



Despite living with this ultra-rare disease, our little Roman is a happy boy who is living life to the full and with the supportive network of family, friends, the charity and medical professionals, he will continue to do so!



There is a well known phrase in the world of runners, "*We don't sweat, we sparkle!*" But now we are part of the AGSD UK community, this has changed to, "*We don't sweat, we glisten!*" Along with Rav, Leanne and Arya Tank, Roman's beautiful family, we will continue to raise funds for this exceptional cause.

After Kerrie and Dom's fantastic efforts, the total of the Tank family's fundraising page has reached £7503 + £1699.50 gift aid. https://www.justgiving.com/page/ravtank

Fundraising Update Continued

Hugo's WOD Squad

Hugo's WOD (workout of the day) *Squad* is a group of avid Cross-Fitters from *CrossFit Leamington*, who will be taking on 24 workouts over a 24 hour stint. Yep, that's one workout per hour, on the hour, every hour, for 24 hours! The event will take place at the CrossFit Leamington Box on Jan 31st kicking off at midday and will finish with the last workout, named 'Hugo' at 11am on Sunday 1st Feb.

Hugo is *Flo Kinnafick's* 6 year old son and has GSD 3b. Flo, the AGSD-UK chair, is keen to raise as much money as possible running up to the event and during the 24 hours. Please feel free to follow her Instagram page @flo_kinnafick where she'll be keeping everyone updated on the progress of the fundraising and how the 24 hours of workout goes!

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

Ayr Classic Motorcycle Club

Ayr Classic Motorcycle Club raised **£350** for AGSD-UK at their annual classic motorcycle show in June. Thanks to Andy Chalmers, AGSD-UK was a chosen charity for the event.

Club Chairman Ricky Lieper (left) & Andrew Chalmers (right)

"I was privileged to be able to nominate AGSD-UK as my chosen charity for funds raised at this year's Ayr Classic Motorcycle Club show.

AGSD-UK has helped me since my diagnosis with McArdle's almost 12 years ago, so it's a charity that's close to my heart.

Myself and my wife Laura ran the helmet park for the day where we stored people's helmets for a donation to AGSD-UK. Unfortunately typical Scottish weather meant a very wet day and we didn't have as many helmets as we would have liked!"

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.

Festive Fundraising Ideas

Christmas is just around the corner and it can be a fantastic opportunity to raise vital funds for AGSD-UK, as well as increasing awareness of GSD and how it can affect daily life.

Here are some simple awareness and fundraising activities that you could organise within your neighbourhoods, social groups, at home or in your workplace.

- Festive bake-off competition
- Sending AGSD-UK season's greetings cards; contact info@agsd.org.uk to order, in return for donation
- Asking for donations instead of cards
- Pledging to do a fundraising challenge in the New Year
- Santa themed quiz night
- Festive singing and collection
- Anything else you can think of!

Need some help?

If you need any help with your awareness and fundraising, for example with publicity, prizes, materials or just general support, then please get in touch with us at info@agsd.org.uk

Let us know what you're doing!

We'd love to know if you have something planned! Please get in touch so that we can share how you are supporting the GSD community in raising funds for vital services.

All proceeds raised on behalf of AGSD-UK will help make sure anyone affected can continue to receive crucial practical help and support from our information and specialist advisory services.

Please direct all fundraising to our Just Giving Page www.justgiving.com/agsd or use the QR code.

AGSD-UK cards are available this year

GSD Type 3 Workshop in Denver 2025

Clinicians, researchers and families from all over the world met up on the last day of the annual AGSD-US conference to discuss the future care and treatment for those affected by GSD3.

Notre Dame University in association with AGSD-US, the Reisenhauer family and the GSD Type 3 Foundation, hosted a day of talks, presentations and personal experiences. These were accompanied by thoughts on the current state of affairs, problem solving and future action.

The day started with deeply moving presentations from family members and people with GSD3. All areas of life were covered, with tears, feelings of anxiety, frustration, panic and the sense of a small bit of control being shared. There were tales of dashing to hospital (accompanied by helpful traffic cops), the fear of handing your child over to the care of someone else on their first day of school and the difficulties of navigating the more common ills of life while living with GSD, accompanied by the feeling of being made to feel ridiculous.

Following on, there were insightful presentations from professionals covering areas such unmet medical needs from both the patient's and the professional's point of view, updates on the natural history of GSD3 and the role of a dietician. The audience then heard about the role of fats in a diet and general patient nutrition and exercise. The morning ended with an international panel of speakers discussing clinical care.

The afternoon centred on research with an update on gene therapy, biomarkers and mRNA therapies. Finally, the day rounded off with a general discussion of the next steps to be taken.

This was a long overdue meeting but was a vital step forward in finding a more hopeful future for everyone affected by GSD3.

Trustee Ailsa Arthur with Cathy, Dave and Cayla Reisenauer

Welfare Reform Updates

July saw a series of amendments to the government's welfare reform proposals before these were voted into law. The amendments to the government bill came in response to concerns from people affected and disability charities about the potential hardship the proposals would cause.

Concessions made included reversing some planned cuts to Universal Credit and protecting current claimants of Personal Independence Payment (PIP) from stricter eligibility rules. The government also conceded it would not change PIP rules until it had time to consider the findings of a review. The amendments to the bill means that any future changes to PIP will not be introduced before autumn 2026, after the full review of PIP has been completed.

The updates below reflect the state of play following introduction of the Universal Credit Act:

- The Pathways to Work reform, announced in March 2025, promises intensive employment support for sick and disabled people: 1,000 work coaches will be deployed to help around 65,000 claimants with health related benefits.
- The same reform pledges that people with severe, life long health conditions who will never be able to work will have their income protection improved and will not face repeated assessments.
- The government has committed to uprating some benefit amounts in line with inflation or above. The standard allowance for Universal Credit will go up more than inflation every year until 2029 / 2030. This means that single claimants who are aged 25 or over will receive £725 a year more by 2029/2030.
- However for the health related element of Universal Credit, existing claimants will have their rate frozen at the current level (around £97/week) until at least 2029/30.
- For new claimants the health element of Universal Credit will be cut in half and frozen from April 2026 at a reduced rate of around £50/week.
- On PIP, the government's previous proposal that claimants must score 4 or more points in at least one daily living activity in addition to the usual criteria, to qualify for the daily living component, is now part of a wider PIP review.

In light of the impact of the changes to the health related elements of Universal Credit and potential for further announcements on welfare in the November budget, disability organisations are continuing to challenge the government around its plans for disability benefits, as well as pressing to play a full part in shaping and contributing to the PIP review process.

For more background on the welfare changes see:

GOV.UK
Scope
Carers UK

For any queries and individual support with an application or appeal don't hesitate to contact ***info@agsd.org.uk*** and an advisor will be in touch - we're here to help!

The McArdle's Experience 2025

Support team member, *Harriet Thomas-Bush* reports on the learning, socialising, and fitness building on the McArdle's walking courses.

In August, members of the *GSD5 McArdle's* community once again took over Wales for our annual walking courses. The 2025 contingent was particularly international, with eight nationalities being represented, ranging from England, Wales, Denmark, France, Germany, USA, and Ireland to Australia. This year, we were in the *Eryri (Snowdonia) National Park* staying at the amazing *Snowdon Lodge in Tremadog*, with the terrific support of Carl and Anya. We were blessed with largely beautiful weather for both the Under 21s course, and the Adults course. It was a full-on experience with around 275 person-days of attendance across members of both courses and the support team.

Everyone conquered a variety of different terrains, including forest paths, the beach, and mountains. The course leaders demonstrated different techniques for such terrains, as well as for everyday life, and tips for supporting your mind and body through many thousands of steps.

This year the leadership team saw *Eoghan Ross* stepping up in the position of assistant leader alongside course leader *Bronte Thomas*. Having two different experiences of McArdle's disease, as well as the experience of *Andrew Wakelin*, provided such a full picture of the range of symptoms and challenges, as well as excellent discussion points.

We celebrated two birthdays, visited castles, the beach, waterfalls, all whilst learning incredibly valuable lessons from our course leaders and from each other, whether on a walk or by the firepit in the evening. There were even matching T-shirts for everyone, and of course a lot of excellent ice cream.

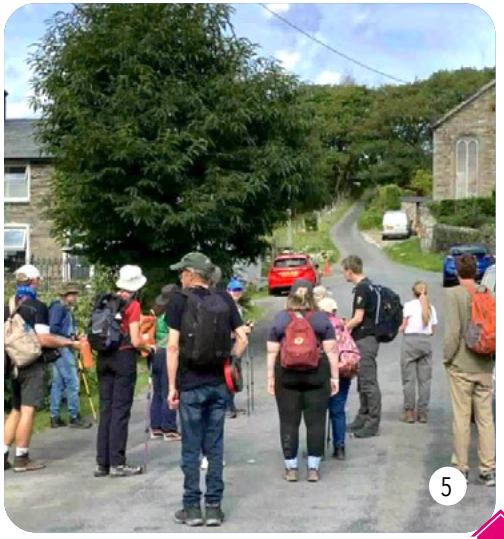
A special thanks to our team leaders for keeping everyone safe and for providing such incredible insight. We truly do thank you for your dedication! Thank you also to the support team, for cooking, driving, and ensuring everyone had an excellent time. A special thanks to the *Ogwen Valley Mountain Rescue Team* who were kind enough to show us around their HQ and equipment, and give us a talk on staying safe in the mountains. We are very grateful to you for being on stand-by every day of the year, and regularly saving lives.

If you have McArdle disease, we would love to see you in 2026 when we take over the Pembrokeshire coast!

A note to those who were on the children's course – the puzzle thieves send their sincerest apologies for causing such chaos. We haven't laughed that much in ages. We are ALL not guilty!

"I loved the walking course again this year! We did a variety of things from having a social day in Caernarfon and exploring the castle, to walking up part of Yr Wyddfa (Snowdon)! I even learned some new things this year about my McArdle's (this was my fourth year)! So if you are thinking about coming next year, whether its your 1st year or your 16th year, you should definitely come along!" – Sarah, age 19.

Images: 1, Hans from Germany on his second course. 2,Taking a break from walking on soft sand. 3,Course Leader Bronte Thomas, 4, Assistant Leader Eoghan Ross, 5, Briefing how to tackle a steep slope.



Save the Date! McArdle's Walking Courses 2026

Courses & Dates: We will have provision for beginners and for more advanced walkers, including those who have been on the course before. If you are uncertain as to which course is best for you, please communicate with us: type5@agsd.org.uk.

Young People's Course – Saturday 1 to Friday 7 August.

For those aged up to approx. 25. *Those below age 18 to be accompanied by a parent or guardian.*

Adults Course – Saturday 8 to Friday 14 August. *For those aged 18 and over.*

Venue: The walking courses will be held in Pembrokeshire, a coastal National Park in the south-west tip of Wales. We have enjoyed courses there four times in past years from 2012.

Accommodation: We will again be staying at the comfortable and friendly National Trust centre at Stackpole on the south coast. From the door we can do walks taking in: countryside, lakes, beaches, bays and cliff tops. And a relatively short drive away are features such as the *Preseli Hills*, a boat trip to *Caldey Island*, and a visit to Britain's smallest city – *St. David's*.

Leaders: ***Bronte Thomas*** and ***Eoghan Ross***, with backup from ***Andrew Wakelin*** – all with experience of this venue, accommodation and walk routes.

More details and the 2026 booking form to be updated on the web page very soon

A walk from our base in Pembrokeshire, taking in lakes, coast, cliffs, bays and beaches. Oh, and ice-creams!

Anxiety Workshop

Many people today experience symptoms of anxiety -even if they don't always recognise it or talk openly about it. It can show up in different ways, like trouble sleeping, racing thoughts, difficulty concentrating, irritability, or even physical symptoms like a fast heartbeat or stomach issues. Daily stress, uncertainty, and pressure can all add up, and it's more common than ever for people to feel overwhelmed. Talking about it is an important first step; bringing awareness and tools to cope with anxiety can really make a difference.

You're invited to attend a supportive and practical workshop designed to help you cope with anxiety in a healthy and effective way. Whether you experience occasional stress or ongoing anxious thoughts, this session will provide helpful tools, techniques, and a safe space to learn and connect with others who understand. Together, we'll explore strategies to manage anxiety, build resilience, and improve overall well-being. Join us and take a positive step toward feeling more calm, confident, and in control.

Would you like to better understand how anxiety affects you – and learn simple tools to manage it?

Join our supportive lazy Sunday afternoon workshop on the **23rd of November** from 2.00-3.00pm, where we will:

- Explore how anxiety impacts the brain and body.
- Learn practical techniques to manage everyday anxiety and improve your quality of life.
- (Optional) Enjoy a calming 20-minute guided relaxation session. Simply find a private, comfortable space – a chair or your bed – and have a blanket handy to wrap yourself in.

This group will take place on ***Zoom*** and is a safe, welcoming space for learning, sharing, and finding moments of calm.

If you are interested, please get in touch – we would love to have you join us.

Email elizabeth.davenport@agsd.org.uk for more details.

Since I thought I'd lose you

Amanda Holthaus

"Since I thought I'd lose you, I am not the same. In both good ways, and in more complicated ways, I am changed as a parent.

I am more afraid now. The fear that shook our lives has crept into every corner of my mind and takes the stage from time to time. Like I don't trust the ground I stand on to hold me up, I am on guard. Hyper-vigilant. Unsettled. Every lab result will hitch my breath. I will anticipate every appointment with nervousness. I will think about your future with heavy uncertainty, and worry.

I am also more grateful now. The small things feel so big. Every milestone feels like a gift instead of an expectation. I know how much harder your body has to work towards these and it makes me so proud. The deep appreciation I have to experience every day with you makes me feel so isolated from the parent I was before. I was I then. I took so much for granted and nothing could have made me see that like the raw fear of losing you. I appreciate every day through a new lens.

I have been humbled. The picture I had in my head about what life would be, looks different now. There are considerations, accommodations, and limitations, and these are hard and frustrating at times. But they've also allowed me to see another side of things, and I needed to see that side. I feel empathy and connection to others who I never could have understood before. It's much easier to accept now that you really shouldn't sweat the small stuff, and life doesn't go our way, but I've learned to appreciate it as it is.

Relatability is so rare. We are now members of this small club of people who share a struggle. And while I appreciate that tribe so much, I feel isolated at times from the majority. I can't make them understand how we've changed, how we worry, or this unique thought process. They'll never understand and while I am happy they won't, it makes the parenting journey feel a bit lonely too.

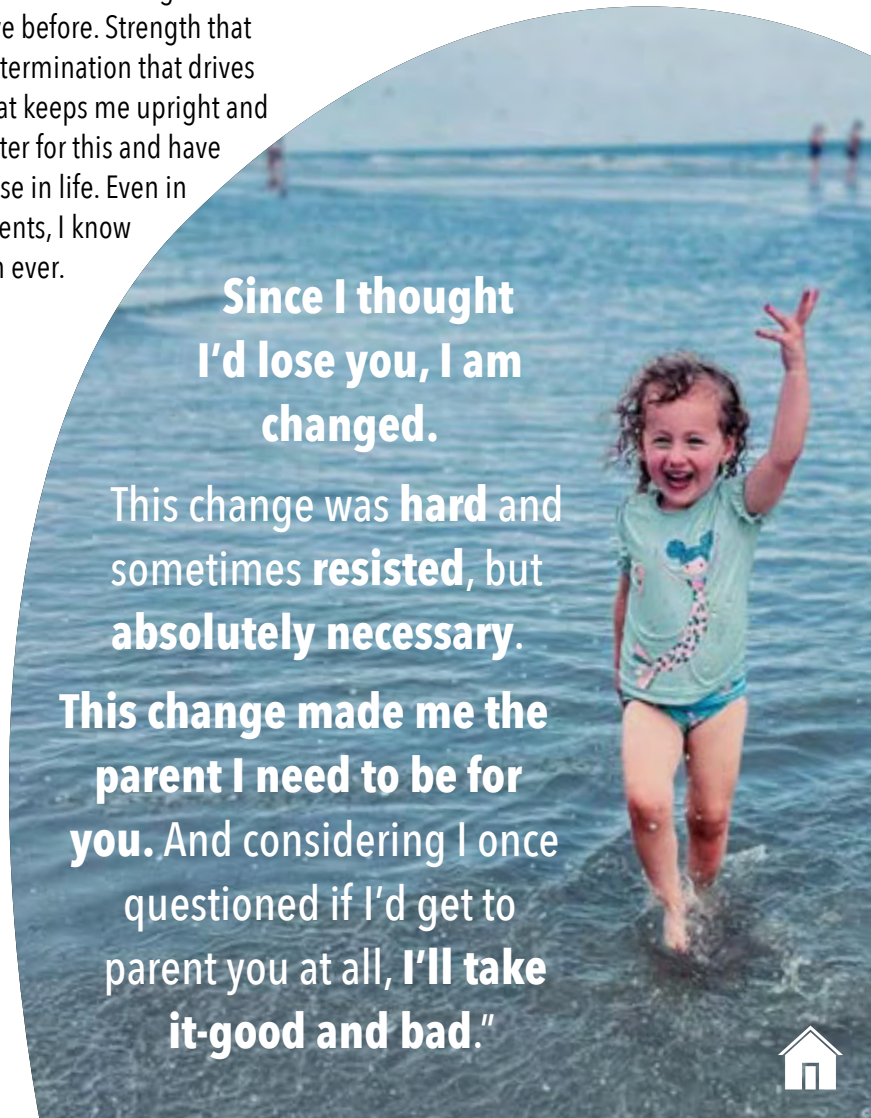
Our priorities have shifted. Finding ways for you to lead a normal life, while also understanding that there may be limitations have been key for me. I will not put physical abilities and physical strength on an achievement pedestal. Maybe sports are out of the question, but at least you can attend school. Maybe you won't dance ballet or earn a black belt, but at least you can walk.

Your bedroom walls may not be lined with trophies, but I know in my heart that your achievements will be no less on their own scale. And I am proud of everything you do nonetheless.

I am stronger now. I never thought I wasn't strong before you, but after you does not compare. Thinking I'd lose you completely depleted me, and then it fuelled me. It ignited something in me that I did not have before. Strength that surprised me, determination that drives me, and hope that keeps me upright and onward. I am better for this and have additional purpose in life. Even in my weaker moments, I know I'm stronger than ever.

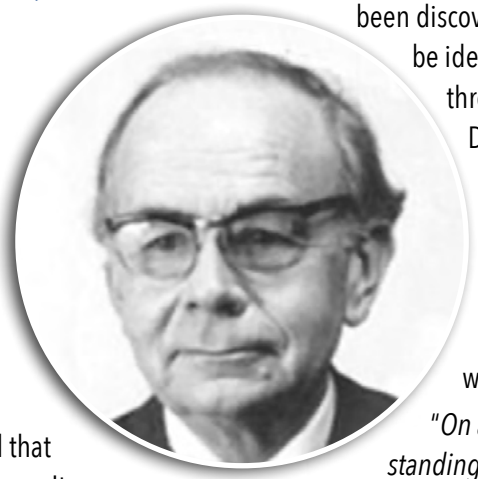
Since I thought I'd lose you, I am changed. This change was hard and sometimes resisted, but absolutely necessary. This change made me the parent I need to be for you. And considering I once questioned if I'd get to parent you at all, I'll take it-good and bad."

https://pompeperspectives.com/



75TH ANNIVERSARY OF MCARDLE DISEASE

Andrew Wakelin comments on 2026 being the 75th anniversary of the publication of the original paper describing what became known as McArdle disease (and later GSDV or GSD5).



Dr Brian McArdle (1911 – 2002) reported on a case of a man with pain on exercise, followed by weakness and stiffness. His investigations led him to describe a *Myopathy due to a defect in muscle glycogen breakdown* and that became the title of his paper. It was presented over 22 pages in the journal ***Clinical Science*** published in February 1951.

Research in London

Dr McArdle was at the time working at the Clinical Research Unit of Guy's Hospital in London and cooperated with several colleagues who undertook chemical estimations. He was able to describe what was the first disorder characterised by a gross failure of the breakdown of muscle glycogen to lactic acid.

Dr Brian McArdle (1911 – 2002)
Title page of the 1951 paper

At that time there was, of course, no genetic testing possible and the PYGM gene had not been identified. The myophosphorylase enzyme had not been discovered, so could not be identified as deficient through muscle biopsy. Dr McArdle had to rely on much less direct testing, and this extract from the paper gives a flavour of the sort of evidence that was able to be collated.

"On another occasion his standing pulse rate rose from 78 to 152 as the result of running up 90 steps in 65 seconds, falling to 84 five minutes later. The pulse rate of a normal subject climbing the stairs in the same time rose 22 beats and fell below the initial value within a minute."

Expansion of knowledge

In the following years other researchers joined in and investigated the metabolic process which was failing, so the whole process is now more clearly understood. This gives a better base for research looking for a treatment or even a cure.

We are very lucky that clinicians and researchers have taken an interest in McArdle's, and we probably get rather more attention than numbers as small as ours would normally attract.

The man studied in the original paper was aged 30, as was I in 1980 when I was finally diagnosed using many of the same tests, plus a muscle biopsy. They thought that I was probably case number 50 in the world. So that was roughly 50 cases discovered over a period of about 30 years. In the most recent 30 years, from 1995 to 2025, I estimate that about 3000 cases have been identified worldwide. I don't suppose the prevalence of McArdle's has changed, but thank goodness both awareness and medical science have moved on very considerably.

Coming years very promising

With global cooperation between patient groups such as AGSD-UK, clinicians, researchers and international organisations such as Euromac and IamGSD, the future is promising. We are managing to lower the average age of diagnosis (the target is age 10), and management approaches, treatments and even a genetic cure are all possible. We are very grateful to Dr Brian McArdle for having got this started!

MYOPATHY DUE TO A DEFECT IN MUSCLE GLYCOGEN BREAKDOWN.

By B. McARDLE.*

(From the Clinical Research Unit, Guy's Hospital.)

(Received March 23, 1950.)

THE energy for the contraction of muscle is largely derived from the oxidation and breakdown of glycogen, a process involving many interlinked reactions and dependent on the functional integrity of numerous enzyme systems. It is surprising, therefore, that only a few diseases are known in which a serious disturbance of muscle function is directly attributable to derangement of the carbohydrate metabolism of the muscle. This paper records what is believed to be a hitherto undescribed disorder which is characterised by a gross failure of the breakdown in muscle of glycogen to lactic acid.

The patient, George W., aged 30 years, was admitted to Guy's Hospital on 19.9.47, under the care of Dr. A. C. Hampson, who on clinical grounds diagnosed a disorder of muscle metabolism and very kindly referred him to me for further investigation. For as long as the patient could remember, light exercise of any muscle had always led to pain in the muscle and, if the exercise were continued, to weakness and stiffness. For example walking a few hundred yards, particularly if fast or uphill, provoked pain in the calves and thighs, and lifting heavy weights resulted in pain in the arms. Even chewing sometimes gave rise to pain in the masseters. The pain, at first dull and aching, increased with continued exercise, while the muscles became progressively stiffer and weaker. Usually all the symptoms rapidly disappeared on resting, but when he continued the exercise not only did the symptoms increase in severity, but they persisted longer when he was finally forced to rest. If he was gripping a heavy object, he might eventually have to drop it owing to the pain and weakness, but his fingers remained in the flexed position and it might be five to ten minutes before he could voluntarily extend the fingers fully. He had found that frequent passive extension of the fingers, though stiff and painful, shortened recovery.

* Work undertaken on behalf of the Medical Research Council.

I wish to thank Mrs. B. Evans and Miss L. C. Carson who performed many of the chemical estimations.

An afternoon at the Museum

AGSD-UK recently held a social afternoon for anyone affected by a hepatic GSD at Salford Museum. There was a lot of laughter, learning and tales of life with GSD.

Vitaflo were kind enough to come and share some of their recipes along with delicious samples. Their food is suitable for people with a hepatic GSD: brownies, chocolate pudding and pizza were all on the menu. The recipes can be found on their website (https://www.gsdandme.co.uk/recipes)

If there's a get-together in your area, or you're nearby when we hold our next one, please do come and join us - it will be a lot of fun.

"We had a really enjoyable day, meeting everyone, walking around the museum and eating the food.

My father particularly enjoyed talking to other parents." ***Esther***

"Last month's AGSD-UK hepatic meet-up truly reminded me how powerful connection can be. Being surrounded by people who understand the journey - from the challenges to the small victories - felt both comforting and uplifting.

It wasn't just about sharing medical stories; it was about sharing strength, laughter, and hope. Every conversation reminded me that we're not alone in this, and that together, we can turn awareness into action and fear into courage.

I left feeling inspired, supported, and deeply grateful for our AGSD community. Here's to more moments of understanding, healing, and togetherness." ***Maryam***

"I am the big sister of a very brave boy who lives with GSD1b. The AGSD-UK get-together in Salford was such a special day for our whole family.

It was really inspiring to see so many people come together to support each other. I loved meeting other siblings and families who understand what life is like with GSD. It made me feel like we're part of a bigger community, like we're not going through it alone.

Watching my brother share his story made me so proud of him, and it reminded me how strong and amazing he really is. I hope there are more events like this in the future, because they help us connect, learn, and feel supported.

Thank you for making it such a positive experience." ***Imaan***

"It was lovely to see some familiar faces at the get-together. And it was great to hear some updates on how everyone is doing. We tried new recipes and shared ideas. Thank you for supporting us again AGSD-UK." ***Cat***

"The AGSD-UK get-together in Salford was such a worthwhile experience for me and my family. It really highlighted the importance of connection, community, and open communication. It was so encouraging to meet others with shared experiences, to feel understood, and to know we're not alone on this journey. These kinds of gatherings create a real sense of belonging and support, something that's truly invaluable." ***Iqra***

"I really enjoyed the AGSD-UK get-together in Salford. It was amazing to meet other people who understand what it's like living with GSD.

I felt proud to share my story, and I hope it helped others feel like they're not alone. It was great to see how everyone supports each other, like one big team. I also liked making new friends and hearing other people's stories too.

I think these meet-ups are really important, and I hope we can have more of them in the future!

Thanks for having me!" ***Mohid***

Pompe Get-together

We had a wonderful Pompe get-together in June 2025 in Reading, and it was truly a day to remember. The venue was fantastic – spacious, welcoming, and perfect for families to relax, connect, and share their stories. Everyone had a great time and left feeling uplifted.

What made the day so special was the sense of understanding and belonging. For many families living with Pompe, it can often feel isolating – constantly having to explain the condition to others who may not fully understand. But at this event, no explanations were needed. Everyone "got it." There was comfort, empathy, and strength in simply being together.

The feedback was overwhelmingly positive. Families shared how empowering it felt to meet others who share their journey, to exchange experiences, and to know they are not alone. These gatherings remind us that while Pompe may be rare, the community's resilience, compassion, and support for one another are truly extraordinary.

Tony's Story

My name is Tony Powell. I discovered I had Pompe after a relatively short diagnostic journey around eleven years ago. I had realised there was something 'odd/wrong' when I was working away and was getting leg cramps rushing around on a large site, which seemed unusual, following it up led to the diagnosis. My diagnosis was relatively quick, helped by health cover from work allowing me to accelerate my appointments and therefore diagnosis.

I've always referred to my infusions as 'stabbings', not a reflection on the nurses who have been universally wonderful, and I am extremely grateful I get my infusions at home. I have carried on working and colleagues also call my infusion days 'stabbings'. I reduced my hours to 4 days a week shortly after I reached 60 to better manage the fatigue that is inevitable with Pompe whilst trying to maintain a normal-ish life. I will be finishing work at the end of 2026, a balance between accepting the relentless progression of Pompe and enjoying my job. I was lucky to have fallen into a job I have enjoyed, including some unique experiences, but it will be 40 years by the time I stop and it is becoming frustrating to work around the progression of Pompe.

I suspect some of the progression is probably in my head, I have got increasingly bothered by the prospect of falling over, although I've done it often. But on a couple of occasions I have discovered that I can be stranded until I get help or find something I can pull on to stand up again.

AGSD-UK and other Pompe support links online have been a great help in understanding that you're not alone and others are wobbling along similar paths, sharing solutions to similar issues that I trip over, occasionally literally!

My personal motto since diagnosis is *'look up, not down'*, although clearly I need to do the latter when walking!

The Transition From DLA to PIP

AGSD-UK benefits advisor ***Zainib Hussain*** explains that the transition from Disability Living Allowance (DLA) to Personal Independence Payment (PIP) presents several challenges that families should be aware of. The criteria for DLA and PIP are different, and understanding these differences is crucial.

From my experience, parents often manage all aspects of the DLA application themselves. When it comes to PIP, parents still handle the forms, but there is an additional assessment process. During the PIP assessment, the assessor will ask the claimant (the child or adult) questions directly rather than relying solely on the parent's input. Therefore, it's essential for families to understand that the claimant must provide honest and accurate information about their disability or condition. Being clear and transparent is vital, as the assessor cannot award the correct level of support if the responses are incomplete or understated.

It's also important for parents to be aware of how the support they provide is described. For example, assistance that parents give in daily tasks - such as preparing meals, supervising activities, or supporting with health needs - needs to be clearly reflected on the PIP form. Every element of care, from daily living support to additional supervision for health issues, should be accurately documented to demonstrate the level of support required.

By understanding the differences between DLA and PIP and providing clear, honest information, families can help ensure the claimant receives the appropriate support.

Guidance for parents

Moving from DLA to PIP can be confusing because the rules are different. Here's what parents need to know:

- Assessment process: unlike DLA, PIP involves an assessment where the assessor will ask the claimant (child or adult) questions directly. Parents can't speak for them.
- Honesty is key: the claimant must answer honestly and clearly about their disability. Avoid downplaying difficulties - accurate answers lead to the right level of support.
- Documenting support: parents should describe all the help they provide, such as:
  - → Daily living tasks like preparing meals and drinks and managing routines.
  - → Health-related support, for instance administering medication, managing symptoms, and monitoring for safety.
  - → Supervision, including being present to prevent accidents or respond to emergencies, particularly for children or adults with complex needs.
  - → Emotional support, for example providing reassurance, encouragement, and guidance to help the claimant cope with daily challenges.
  - → Practical guidance, potentially involving teaching skills, helping with communication, or assisting with social and educational activities.
- Be detailed: even small daily support matters. The PIP form should show how much help is really needed.

Don't hesitate to contact ***info@agsd.org.uk*** to get expert help with making a claim - it can make all the difference:

"Your efforts played a crucial part in helping my son overcome his reservations about acknowledging his illness. I'm thrilled to share that thanks to your support and guidance, my son has now been awarded PIP"

By understanding these points and including all practical, supervisory, and emotional support, families give assessors a full picture of needs and greatly increase the likelihood of the correct PIP award.

Retirement of Professor Ros Quinlivan

Professor Ros Quinlivan is due to retire from her role as lead of the NHS Highly Specialised Service for McArdle Disease and Related Disorders in December 2025.

We would like to wish her all the very best for her retirement, after over three decades of dedicated service in the interests of people with neuromuscular diseases, in particular McArdle disease and other muscle GSDs.

Ros Quinlivan obtained a BSc in psychology then studied medicine at *University College London* and did post-graduate training in paediatrics at London teaching hospitals. She was the first clinical research fellow in neuromuscular disease at *Guy's Hospital*, working across paediatric and adult specialties. She was awarded an MD by University College London for her research into the cardiomyopathy of Duchenne and Becker Muscular Dystrophy. She was appointed a consultant in 1995 in Gobowen, Oswestry, where she was director of the *Wolfson Centre For Inherited Neuromuscular Disease*. Whilst there she led an embryonic McArdle's service on just four afternoons per year, based at the *Robert Jones and Agnes Hunt Hospital* in Gobowen, where she had worked with Professor RHT Edwards on some of the very earliest cases of McArdle's to be recognised.

From those humble beginnings, her service gained recognition from AGSD-UK, which provided support and even some early funding.

Moving to University College London Hospitals in 2010, she led the transition service for adolescents with neuromuscular disease at *Great Ormond Street Hospital* and the *National Hospital for Neurology and Neurosurgery*, part of University College London Hospitals, at Queen Square. She established the national McArdle disease service in 2011 at the *Queen Square Centre for Neuromuscular Diseases*. The service is located in the National Hospital for Neurology and Neurosurgery and other institutes around Queen Square.

Her work and that of her multidisciplinary team has been the envy of the McArdle's world and has been of huge benefit to those with McArdle disease. It has provided many people not only with their original diagnosis but with management strategies for coping with the disease and reducing its impact on their daily lives. It has developed into the world class service it is today, all the time led by Professor Quinlivan.

In addition to her clinical work, Professor Quinlivan has led and supported very many research projects and has published numerous papers on the subject of the McArdle disease.

She has presented on McArdle's at professional conferences and patient group meetings around the world. She was an instigator and founding board member of the *Euromac Registry*, a member of the *AGSD-UK Medical Advisory Board*, and of the *International Association for Muscle Glycogen Storage Disease (IamGSD) Scientific Advisory Board*. She has also cooperated in the development of strategies to help patients manage their condition, and has cooperated in the drafting of a great deal of literature and videos to assist patients.

We wish Professor Quinlivan a long and happy retirement and thank her for everything she has done for us over many years.

We will be welcoming *Dr Chiara Pizzamiglio* as the new consultant neurologist who will lead the service from March 2026. She has previously worked with the clinic, including undertaking research towards her doctorate. In the interim the consultant will be *Dr Matt Parton*.

Professor Quinlivan visiting the "Walk over Wales" fundraising and awareness-raising event in 2010, with Stacey Reason and Andrew Wakelin.

Photo credit: University College London Hospital

Pompe Disease Monitoring Workshop

Article written by Amicus Therapeutics who organised and funded the meeting.

This October, thirty healthcare professionals from around the UK joined together for a two day workshop about monitoring in late-onset Pompe, funded by *Amicus Therapeutics*.

Attended by doctors, physiotherapists and nurses from across the multi-disciplinary team, the workshop aimed to share experiences and feedback about improved monitoring practices employed over the last year in late-onset Pompe, with a view to implementing a consistent approach to monitoring.

The October 2025 workshop followed an initial Amicus-funded ***Pompe Disease Monitoring Masterclass*** held in October 2024, where initial areas for improvements in monitoring were discussed. Ideas for adjustments to patient reported outcome measures (PROMs), respiratory monitoring and physical assessments were initially debated in order to improve the patient-relevance of monitoring practices, adjust for the real-world setting, increase the number of touch points and enhance the holistic care that patients receive from different healthcare professionals in the multi-disciplinary team.

In parallel, centres in the UK have independently been working on consensus guidelines which reflect these proposed adjustments. Those guidelines are expected to be published in the coming months.

The recent workshop built on the work from prior sessions, focusing on the following areas:

- Optimised use of patient-reported outcome measures and some fresh research with the patient community to explore views on completing these, including the relevance and simplicity of different tools
- The importance of respiratory monitoring and the role of respiratory physios in aiding monitoring assessments. There was also a demonstration on the utility of ventilator data for monitoring and discussion of patient views on accessing this data
- Updates from physios who are now embedded within the specialist Pompe service at some UK centres, including the virtual exercise classes they have been running
- Implementation of the ***North Star Assessment*** for limb girdle type muscular dystrophies ***(NSAD)*** in routine practice for late onset Pompe to better assess muscle function. The metabolic centres shared their experiences with trying the assessments with their patients and seeing how well they had correlated with understanding functional ability in daily life activities. They also reflected how they had acted as a helpful conversation catalyst.

The recent workshop featured patient representatives who presented community feedback and provided input into the topics covered, exploring with healthcare professionals how to optimise the available tools which support patients and families in preparing for their appointments.

The main take-away was seeing how fruitful it can be to work together as a clinical community with patient representatives in understanding how monitoring should look in the current context and how it can be tailored to individuals' needs and priorities. Some of the clinicians and physiotherapists in attendance commented on the phenomenal shift in practice in only a year, demonstrating the desire from all to improve how people living with Pompe are monitored. Further work will be carried out to embed the new monitoring approaches in all centres to bring consistency across the UK.

Dr Patrick Deegan, consultant metabolic physician at *Cambridge University Hospitals NHS Foundation Trust* commented *"It is very rewarding (and possibly even unusual) to see that a small number of efforts made last year to encourage UK centres to collect monitoring data in a more complete and consistent way, has led already to improvements in how we work and collaborate with each other. For me, another important take home message is that collecting this data is not only useful for research, but it can have a direct impact on the life of the individual patient, through better communication with healthcare professionals about real-life problems and thus better advice about adaptations and support. I look forward to further progress in showing that these efforts lead to better care for patients."*

For those in the UK living with late onset Pompe, monitoring visits may look different as these new approaches are being trialled. If you have feedback to provide from your own experience, please do speak to your specialist healthcare team.

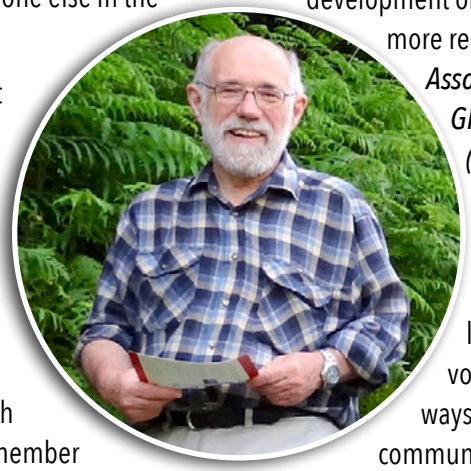
NP-NN-GB-00011025 NOVEMBER 2025

Spreading the McArdle's workload

Could you lend a hand?

Andrew Wakelin

After 21 years in the volunteer post of McArdle's Coordinator for AGSD-UK, and at age 76, I am really trying to take more of a backseat and start heading towards handing over to someone else in the not too distant future.



It was in 1999, almost 20 years since my diagnosis, that I finally found ***Dr Ros Quinlivan*** in Gobowen, who was then reserving just two afternoons a year to see people with McArdle disease. I remember her wanting to tell me about "*second wind*", but I was sitting there telling her about it having discovered it for myself, but not knowing its name or understanding the mechanics of it. I soon found myself helping to support the embryonic McArdle's clinic. It was at the clinic that I met ***Ann Phillips***, then the President of AGSD-UK and ***Nick Owston***, the McArdle's Coordinator. Unfortunately, Nick died very unexpectedly in 2003 and in 2004 I took over as McArdle's Coordinator.

More international focus

The role of coordinator has been very developmental, just as McArdle's knowledge and the clinic have both also been developing at a fast rate. Then add to that the development of the ***Euromac Registry*** and more recently the ***International Association for Muscle Glycogen Storage Disease (IamGSD)***. It has been all but impossible to write a "job description".

We now have a long list of people who volunteer in many different ways to help the McArdle's community. Quite a lot of work, such as the publications, have moved from being initiated by AGSD-UK to become the responsibility of ***IamGSD*** with an international remit. With relatively small numbers of people affected by McArdle's, it is highly valuable to coordinate a lot of work internationally, rather than each country doing entirely its own thing.

A huge team

We have a huge team of volunteers who are already helping with so many aspects. We can't name everybody, especially without their permission, but I would like to give a special mention to some, who are public about their involvement. One of the longest standing is ***Margaret Carter***, who handles the vetting and admission of new members to the main ***Facebook*** group. There are another dozen dedicated Admins on this and the other more specialist Facebook groups. Then there is ***Stacey Reason*** who is the President of IamGSD and, of course, the other trustees who you can read about on the IamGSD website. ***Bronte Thomas*** now leads the annual walking courses, together with ***Eoghan Ross***, assistant leader. ***Rachel Thomas*** leads the support team for the walking courses, and serves on the AGSD-UK Board of Trustees. A team of half a dozen form the ***Patient Liaison Panel*** for the McArdle's clinic. ***Sioned Williams*** is probably our highest grossing fundraiser over the years. ***David Thompson*** now coordinates McArdle's content for ***Glisten*** magazine. There are probably another dozen or more who help out on a very ad-hoc basis. My apologies if I've missed anybody who really should have had a mention.

Areas of work

The coordinator's role can now be much more focused on the direct needs of the McArdle's community here in the UK. We have around 360 of us diagnosed, that's including our friends in Ireland. I shall try to set out the key areas which are involved:

- Trying to make contact with all newly diagnosed people here in the UK, to offer them information and support, and to send out copies of our various publications.
- Answering questions from McArdle's people and, with permission, putting them in touch with each other.
- Liaising with the McArdle's clinic in London, and with clinicians in Wales, Scotland, Northern Ireland and Ireland who take an interest in McArdle's.
- Shaping the McArdle's programme of AGSD-UK's "Main Event".
- Liaising with AGSD-UK staff and volunteers on specialist subjects such as publicity, fundraising, events, Glisten magazine and the website.
- Liaising with researchers, clinicians and organisations internationally, especially Euromac and IamGSD.

Contact us to explore the possibilities

Do you have a special expertise or interest which is relevant to our activities? Could you spare some time to get integrated into our McArdle's support network and lend your support in one of the areas listed above, or possibly in another area?

For a no obligation initial chat please contact me on type5@agsd.org.uk; or our CEO Val Buxton on val.buxton@agsd.org.uk.

Drug Development & Resources for GSD1a

Some useful links and information on GSD1a have recently been shared.

In the USA, the Children's Fund for Glycogen Storage Disease Research recently hosted a drug development meeting on GSD1a. The meeting included powerful stories from people affected, as well as input from experts on current management and investigational therapies and discussion panels with community members.

A recording is available on YouTube on the following link:

https://www.youtube.com/watch?v=VtXD7dQ70Ug

Children's Fund for Glycogen Storage Disease Research Youtube recording

The organisation has also shared an update for community members in the USA from Ultragenyx, on their US Biologics Licence Application for DTX401 investigational gene therapy for people with GSD1a. You can find more information here:

https://www.instagram.com/curegsd/p/DNyWcLc3MNW/

Ultragenyx has committed to keeping the community updated throughout the process.

Ultragenyx have also produced a very helpful new web resource for a US audience, with input from people affected, to promote understanding of GSD1a:

https://www.understandinggsdia.com/

Ultragenyx new web resource

Duke the Talented Hypo Alert Dog

Duke looks after Emilee Rivera. Emilee has GSD 9 and Duke's amazing talent is to warn Emilee before her blood sugar drops too far.

Duke attended the American AGSD conference at Denver this year. He flew there with his family and, as a medical alert dog, he was allowed a seat on the plane, rather than a place in the hold.

At the conference he was busy. He not only helped to present a talk on medical alert dogs, but he took it upon himself to warn many of the participants that their blood sugars were dropping. Just before lunch was a particularly hectic time.

Most dogs can detect minute chemical changes in our bodies. They may recognise a drop in blood glucose and the onset of a hypoglycaemic attack before you notice anything is wrong. Training to be an alert dog usually takes over a year.

Duke is a much loved member of the Rivera family. He watches over Emilee and has helped her through difficult times. And on occasions he has been faster than a continuous glucose monitor in alerting Emilee to a hypoglycaemic episode.

Emilee loved attending the AGSD US conference. She came home with more confidence and independence and is counting down the days until the next one.

The Challenge of McArdle's & Cerebral Palsy

Lorenzo Lynch's Story

Bronte Thomas and Andrew Wakelin introduce Lorenzo and how he got on with the walking course this summer.

Lorenzo is 15, lives in Mid Sussex and has been diagnosed with McArdle disease, but that is just a tiny bit of his story because he also has cerebral palsy. When his mother Nada got in touch, it was the first time that we had heard of somebody with both these conditions and it obviously gives Lorenzo a much greater challenge than McArdle's alone. When Nada told us that Lorenzo was interested in attending the walking course in North Wales with his carer, Thomas, and would need a ground floor level-access bedroom, we wondered how Lorenzo would manage.

It was late 2024 that Lorenzo received his McArdle's diagnosis, at the age of 14. Although McArdle signs normally start to be seen around the ages of three or four, Lorenzo's McArdle's symptoms had largely been masked by his movement problems from his cerebral palsy. But then he was twice hospitalised with episodes of rhabdomyolysis and this obviously needed greater investigation. He was diagnosed via genetic testing with a gene panel. He did very little exercise following his second hospital admission in October and although he got the diagnosis before Christmas it took a long time for both him and his family to process the information, and health anxiety was a big issue. He started doing some exercise and twice did the 12 minute walk test with his physiotherapist, but could only walk very slowly due to his cerebral palsy. He has since been referred to the McArdle's clinic.

Nada wanted Lorenzo to attend the 2025 walking course in Eryri (Snowdonia) for the social aspects and the peer support, but also to gain knowledge about how to manage McArdle's. She was not sure how much actual walking he would be able to do, but felt it would be good for him to learn to use walking poles. We agreed it would be good to build his confidence and to meet others with McArdle's. Nada reported that Lorenzo had walked about 3 km one Saturday, stopping for breaks but not sitting down. She felt he could definitely do more. If necessary, he could turn back with his carer and wheelchair and she could collect them in the car.

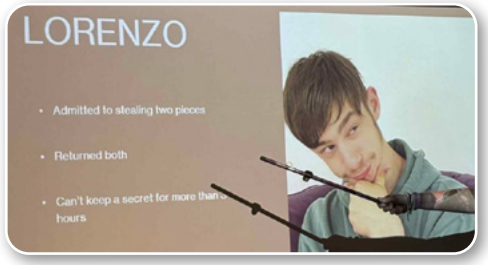
When Andrew saw them arrive in their van at our base in Tremadog with not just a wheelchair, but also an electric wheelchair being unloaded via a hoist, he wondered how far the course would meet Lorenzo's needs. He need not have worried.

Lorenzo has contractions in many muscles, but particularly in his legs, arms and neck. He wears braces on his legs. Despite all this, he was determined to join the course.

At the start of the course, Lorenzo was anxious about how much he could achieve, and did not like the thought of walking for miles in a large group. After two days, he had completely outdone himself by walking miles, and actually enjoying it! The best thing was that Lorenzo's personality started to shine, he was becoming more confident and was a star of the group, adding important insight, sassy remarks and making everyone laugh. Lorenzo even managed challenges such as walking to the lower lakes of *Yr Wyddfa (Snowdon*), and walking on a long stretch of sand. He embarked on varied walks that challenged and pushed him further. Occasionally he would make use of one of the pickup points, along with others who felt that they had done enough.

We will leave the last words to Lorenzo:

"I highly recommend the Wales walking trip, it was the highlight of my summer. I got to know people from all over the world, and hear their stories of McArdle's. Thank you to Andrew, Bronte and Eoghan for organising and leading the course. I will be back next year."



Exposed – jigsaw piece thief!

Muscle GSD Research Update

The Metric Group

McArdle disease Education, Training and Research International Collaboration

Dr Sam Torrens shares details of a new McArdle disease research and collaboration forum called the _METRIC Group_. Dr Torrens is an applied physiology researcher in metabolic exercise physiology and biochemistry at _Queensland University of Technology_ in Australia.

Established in early 2025, the ***McArdle disease Education,Training and Research International Collaboration*** known as the METRIC Group is an international, multidisciplinary network of medical and health professionals with a shared interest and expertise in the treatment and management of McArdle's. Due to the rarity of the condition, clinicians with the necessary knowledge and experience to appropriately advise individuals with McArdle disease can be equally rare - making collaboration essential.

The METRIC Group holds regular online meetings and operates as a multidisciplinary healthcare team. These meetings serve as a platform for members to educate and train one another in specific areas of expertise related to McArdle disease, share the latest international research findings, and foster global collaborations. The group's overarching aim is to improve the quality of treatment for individuals living with McArdle disease, enhance the number and quality of research projects conducted internationally, and ensure that the outcomes of these projects are rapidly disseminated to both the global medical community and affected individuals.

The group includes professionals from neurology, genetics, physiology, rehabilitation medicine, physiotherapy, occupational therapy, biochemistry, pharmacy, nutrition, and research. The group also includes two individuals living with McArdle disease to ensure the patient perspective informs discussions and initiatives.

Be sure to look out for future METRIC Group initiatives and participate in the latest research projects. Together we can make a real difference.

Medical and health professionals interested in joining METRIC Group can contact ***Sam Torrens*** *at sam.torrens@mater.org.au.*

Please note: METRIC is a ***professional network*** *and is* ***not intended for patients****.*

Experiences of living with GSD5 (McArdle) disease

Challenges and strategies. A qualitative study in the Netherlands

Disability and Rehabilitation 2025, Vol. 47, no. 14, 3649–3656 s

This was a qualitative study conducted during 2022 and 2023 with interviews of 13 individuals with McArdle disease in order to explore their life experiences. It concluded that individuals with McArdle disease often experience many difficulties prior to their diagnosis and that they often devised their own coping strategies.

The research found that diagnosis provided explanations for the symptoms that participants experienced, helped mitigate harsh responses from others and contributed to a more positive self-image.

"Although participants had difficulty coping with the variability of daily life, together with understanding the limits of their bodies, finding balance with activities and rest, learning about the disease, and engaging in a rehabilitation program, individuals with GSD5 can experience improvements in daily functioning."

The research recognises the value of educating patients with McArdle disease about the condition and suitable management strategies such as the second wind, implementing the 6 second rule and understanding the physical limitations whilst staying as active as possible. It also identified the psychological stress experienced prior to diagnosis that arises from self-doubt and questioning whether your symptoms are real and recognises the value of offering psychological support to patients.

https://tinyurl.com/5fzjppa7

FROM DIAGNOSIS TO ADVOCACY

Laurence Letreguilly shares her family's journey with GSD IV

When my partner Remi and I welcomed our son Marius into the world in December of 2019, we couldn't have imagined how our lives would change. In just a few years, I went from receiving a shocking GSD IV diagnosis for my son Marius, through a journey of medical mistrust, to becoming an international advocate for all patients and families impacted by a GSD diagnosis.

After a normal pregnancy, Marius' birth was unexpectedly traumatic. I underwent an emergency C-section due to insufficient amniotic fluid, and Marius was immediately taken away by the care team because he was having difficulty breathing. The next few days were filled with challenges for Marius and the gut-wrenching feeling of not knowing what was wrong. Instead of being congratulated for welcoming a beautiful baby into the world, Remi and I were receiving information that was worrying and the doctors didn't have answers. Our son's body position was not normal and it appeared that his hips were dislocated (which we learned later that they weren't). Even after we brought Marius home, he struggled to gain weight, and as first-time parents, we had nothing to compare this journey with. The uncertainty continued and so did the endless visits with the doctors.

Marius' doctors referred us to a geneticist who ordered the review of a small panel of genes. The test results came back with no specific answers, so whole genome sequencing was the next investigative step. The results came back, and my son received the GSD IV diagnosis at the age of two. Our questions shifted to, "What does this mean for Maruis' future? Is his life at risk? How do we help him?"

GSD IV is an ultra-rare genetic disorder caused by changes in the GBE1 gene and is inherited in an autosomal recessive manner. I've learned that it is a disease spectrum, with early-onset and adult-onset presentations which 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 anywhere between infancy and adolescence and causes a range of liver, neurologic, muscle, and heart manifestations. The adult-onset form of GSD IV (or Adult Polyglucosan Body Disease) presents as early as in the mid-30s with primarily neurologic and muscle manifestations.

The first specialist we saw in Paris told us that Marius would need a liver transplant; we were devastated. However, it seemed like the doctor lacked familiarity with the disease and hadn't seen his prior medical charts; so, we did not have confidence in working with her. Consequently, we got another specialist involved.

Eventually, we found a more knowledgeable team in Bordeaux. There, we learned that Marius did not need a transplant. With this news, we could focus on managing the condition and helping Marius thrive. We now have a deep understanding that parents of children with rare diseases have to become parent advocates as well as experts themselves.

I found that it takes time as a parent to come to terms with a rare disease diagnosis. It took Remi and me a full year after learning the diagnosis to start to reach out to the GSD community.

When I first contacted the French GSD advocacy organisation ***L'Association Francophone des Glycogénoses*** (AFG) and attended a family gathering, there were no families who shared our diagnosis, but through AFG and a private GSD IV Facebook Group we connected with two families in France. Through AFG, I also learned about the APBD Research Foundation's 2024 Focus Group for APBD and Early-onset GSD IV. Being a part of this international group has led to even more connections and opportunities, which is so important in such a rare condition.

I'm now one of AFG's Board of Directors and because GSD IV is ultra-rare, one of

the most important things I can do as a mother is to share my son's journey and make sure that others see him as a child and not just a medical case. My advocacy helps drive research, provide support, and ensure that no GSD family walks this journey alone.

Marius is five and a half years old now and is obsessed with sports. He doesn't walk, but that doesn't stop him. Inspired by his love for rugby, he created his own version of the game called 'Sitting Rugby.' With the help of the French muscular dystrophy association and other organisations, Marius has met renowned international rugby players like ***Antoine Dupont, Thibaud Flament,*** and ***Maxime Lamothe***.

These experiences have been extremely motivating for Marius, allowing him to continue being active, which is the number one thing that helps him progress.

Marius's journey is a powerful reminder that living with a disability is an invitation to find creative ways to adapt the things you love to fit your world.

Marius' joy, determination, and resilience shine through in everything he does. Whether he's swimming, playing, or creating his own games, Marius fills each day with laughter.

If there is one thing I want anyone reading our story to know it is that my son is wonderful.

TAKE AWAYS FROM THIS EDITION

- Don't miss out on help from expert AGSD-UK advisors on benefit applications and any other challenges you're facing - we're here to help
- Join our lazy Sunday afternoon anxiety workshop in November
- Take part in some festive fundraising or take on a challenge for the New Year

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

AGSD UK
Association for Glycogen Storage Disease

Upcoming events

- Anxiety workshop: 23 November, online
- AGSD-UK AGM: 18 January, online
- McArdle's walking courses: August 2026, Pembrokeshire

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 2025/26

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Association for Glycogen Storage Disease