Spring 2026

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:
Salford clinic team
The CrossFit fundraising team
The Wood family
The Crawford family
Hugo and Teddy
Ellis

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

AGSD-UK's Main Event on 4th-5th July is fast approaching and we'd love to see as many of you as possible there. If you've never attended an event before and are wondering whether this is for you, please be assured of the warmest of welcomes in a lovely venue that works hard to meet people's needs.

As well as updates on research and clinical developments, there's a variety of different activities to choose from, ranging from a support group session for younger people or a chance for some art-based creative relaxation, to the opportunity to interrogate a multi-disciplinary expert panel or explore reproductive health in GSDs. Most importantly there's plenty of time to share experiences and connect with others who understand life with a GSD.

There are financial support options if you need help to attend – so please don't hesitate, visit *here* to find out more.

The sessions are also designed to start addressing issues raised by your feedback to our community survey and listening exercise, which has shaped AGSD-UK's strategy for the next three years. You can see some headlines about our plans on pages 8-9 of this edition.

A wish for increased awareness and understanding of GSDs was a strong emerging theme, so a discussion about the challenges of talking to others about the condition and approaches that can help is featured on the Main Event programme. We're so grateful to those who have already helped boost awareness by sharing their stories for Rare Disease Day and International Pompe Day and these are featured on pages 14 & 20. We're looking forward to working with community members to do more to raise the profile of the condition in mainstream and social media over the coming months and beyond. We're also looking to expand our presence in GSD clinics so we can increase awareness of our support and services, particularly among people who are newly diagnosed. You can find out more on pages 12-13.

The need for more information about the help on offer was another area highlighted in your survey responses and on pages 26-27 of this edition there are details about grants that might be available for a range of needs. There's also important advice about benefit applications on page 35. We'd really appreciate your feedback to info@agsd.org.uk about other topics you'd like to see covered in Glisten or our information resources, as well your ideas for online sessions that are accessible for those unable to travel to in person events.

Your input is crucial to making sure our offer meets the needs of all community members. We look forward very much to hearing from you and having the chance to catch up with some of you very soon!

Val Buxton

The Main Event

AGSD UK
Association for Glycogen Storage Disease

Feedback from 2023 event

"...a great weekend of learning, meeting and generally having a good time."

"Great fun! Great accommodation and delicious food."

"This event was extremely informative so thank you!"

"It was superb. Team was great-very helpful. Would like to attend again."

"Fantastic weekend... Feel more supported."

"Good mix of scientific updates, patient support and fun activities"

4th - 5th July 2026

Burleigh Court, Loughborough *by popular demand!*

Whatever your GSD, join us for:

- Updates on latest research and treatment options
- Tips for living well
- New therapeutic activities to try
- Time to share experiences and make new friends
- Chance to hear from leading experts
- Fun for children and families

Click or scan here for more information and reservations.

FUNDRAISING ROUND-UP

Thank you to everyone who supports AGSD-UK -any donation, no matter the amount is hugely appreciated to help continue our vital support and services.

- Give wheel - **£2315.35** was raised for the Hugo 24 raffle
- Sioned Williams donated **£50** in lieu of sending Christmas cards
- Jess Sumner held a fundraising raffle/ tombola and raised **£352**
- Allen & Ros Russell raised **£252** at the camping meet for Mylo
- The Football Association Premier League Ltd donated **£250**
- Lavina Gibbs donated **£500**
- Harvey Gonzalez donated **£100**
- The Lancashire Foundation has made a generous donation of **£3,000.** This was part of the Tank family's incredible fundraising efforts in the name of their son Roman who has GSD3. www.justgiving.com/page/ravtank

Christmas decorations raise funds!

At the end of last year Frank and Irene Green raised **£636.50** from passers-by who enjoyed the spectacular display of Christmas decorations and lights at the front of their house in Altrincham, Cheshire, and donated in aid of AGSD-UK. Their adult son Ian has McArdle's. They raised a further **£355.50** from garage sales during the summer. And a further **£100** was sent direct to AGSD-UK from Oakwood Services, as one of their staff nominated Frank and Irene for an award from their charity fund.

So, in total this year they raised **£1092**. An excellent result, especially considering the bad weather over the holiday period. Our huge thanks go to Frank and Irene for their continued support and dedication, having put on the Christmas display and raised funds every year for 7 years now.

Ellis

Ellis is taking on an 8 day challenge in memory of his Mummy, Rosie. Rosie lived her whole life GSD3a and sadly left us in June 2025 exactly 1 week before her birthday. So, Ellis is cycling every day from the first of June until her birthday on the on the 8th, eight one-mile laps.

Due to his own Down Syndrome related complications, Ellis has hypermobility and therefore 1 mile loops are a very long way for him. He'll be fitting his bike rides in between school and extra curricular activities for 8 days in a row; rain, hail or shine!

He's already passed the £750 mark for fundraising for AGSD-UK and hopes to up his total as his challenge goes on.

www.justgiving.com/page/thedudecyclesformummy

The WODSquad take on The Hugo 24

On January 31st, a group of dedicated CrossFitters took on the 'Hugo 24', an extraordinary challenge of completing 24 workouts in 24 hours! Organised by Flo (our Chair of Trustees) alongside her son Hugo, who lives with GSD type 3b, the

WOD Squad from CrossFit Leamington came together for an unforgettable test of endurance, teamwork and resilience. Twenty committed CrossFitters completed every single workout, starting a new session on the hour, every hour, across the full 24-hour period. The event was generously supported by CrossFit Leamington, who opened their gym and created a fantastic environment for the challenge to take place. Across the day and night, the team pushed through fatigue, laughter, and the occasional low moment, creating amazing, often hilarious memories that will last a lifetime.

The support from friends, colleagues, and local businesses was truly incredible. Whether following the challenge live, donating, coming to cheer us on (there was so many of people, including our wonderful Teddy Molyneux and family from the GSD community) or delivering much-needed pizza and doughnuts in the early hours, every contribution made a difference. Thanks to this collective effort, the team raised over **£18,000** for AGSD UK through a JustGiving page and a successful raffle. We are so proud of everyone involved.

www.justgiving.com/team/hugoswodsquad

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.

OUR PLANS 2026-2029

AGSD-UK's new three year strategy is again built around priorities identified by community members.

These are our new strategy themes, based on the input we received:

BUILDING AWARENESS:

Examples of things you said...

"More knowledge among the general medical communities"

"It's depressing having to explain my diagnosis"

"More empathy when travelling on holiday or normal day to day living."

"Patience and belief from regular people"

SUPPORTING WELLBEING:

Examples of things you said...

"The benefits process was too exhausting and stressful to navigate and so for my own well being I stopped so we get no extra financial help."

"Being able to get... help with any funding applications to get funding towards anything that helps with daily living"

"More local get-togethers"

"A tailored exercise and diet programme"

"An adequate head support"

"...a more flexible wheelchair"

"...creating [videos] for each type of GSD would be really helpful"

COLLABORATING FOR CHANGE:

Examples of things you said...

"A continuous glucose monitor sensor"

"More funding and research into treatments or potential cures could lead to better management...and improved outcomes"

"News about research"

"Increased research and treatment options"

"Specific research breakthroughs to cure the condition ...genetic modification"

We now have new goals related to each theme:

BUILDING AWARENESS ← People affected will know about the support available, and encounter greater understanding from the public, professionals and decision-makers

SUPPORTING WELLBEING ← Everyone living with any GSD will have access to comprehensive support services and wellbeing programmes that are tailored to their needs

COLLABORATING FOR CHANGE ← Community members will benefit from research, treatments and service developments that are shaped by their input and driven through effective partnerships

Getting fit for the future

Our strong engaged community and financial stability will equip us for future challenges

AGSD UK
Association for Glycogen Storage Disease

Here are just some of the actions we have planned to help us meet these goals – you can find out more by visiting our website https://tinyurl.com/4jhvy6nr

Building Awareness *We will:*

- Work to secure an AGSD-UK clinic presence in every specialist centre
- Equip people with tools and support to talk about the condition to their family, friends, employers and the wider public
- Raise the profile of the condition and the support we offer through mainstream and social media.

Supporting wellbeing *We will:*

- Connect community members through our national events and in-person get-togethers in different parts of the UK
- Provide tailored online exercise programmes and nutrition clubs
- Develop new evidence-based print and video resources to help people live well with the condition, including those with the rarest GSDs.

Collaborating for change *We will:*

- Host regular professionals' meetings and education sessions, developing up-to-date information to meet the needs of this audience
- Extend our international reach and partnerships to help pool data and resources on behalf of the community
- Gather and deploy evidence to support the development and approval of new treatments and management strategies.

The McArdle's experience

Walking courses for 2026

Our two courses are booked, with around 25 people on each course – coming from UK, France, Germany, the Netherlands, USA and Canada, with a range of ages from 7 to 72. This year we have a young man with GSD 9d, which is very close to McArdle's. If you are interested, contact us for the latest situation in case someone has had to cancel: *type5@agsd.org.uk*

The two courses are: Young People's Course – Sat 1 to Fri 7 August, for ages up to approx. 25, those below age 18 to be accompanied by a parent/guardian. Adults' Course – Sat 8 to Fri 14 August, for those aged 18 and over.

The courses are in Pembrokeshire, the fabulous coastal National Park in the south-west tip of Wales. Our base will be at the comfortable and friendly National Trust centre on the Stackpole lakes on the south coast, from where we can do walks covering countryside, lakes, beaches, bays and cliff tops. A short drive away are features such as the Preseli Hills (for a peak!), a boat trip to Caldey Island, and a visit to Britain's smallest city – St. David's.

Course leader Bronte Thomas and assistant leader Eoghan Ross, together with team member Jan Golos, have just spent a week getting up-to-date on course training material and checking out some new routes. This will build on the experience we have of running courses in Pembrokeshire four times in the past.

A shorter format?

We are wondering whether in future years, we could offer something that newcomers might find a bit less daunting to start with, to build confidence for a full course another year. It might be organised as a long weekend and perhaps be offered at different locations around the country. These events would be at lower cost, and people might find one relatively close to them, meaning less travel. You may recall that David Thompson organised something similar, "Walking in Wiltshire", in June 2022. Newcomers and old-timers, please let us know what you think – *type5@agsd.org.uk*.

The Green Bridge of Wales, on the coast near our base. Celebrating achieving the summit of the Preseli Hills.

Exercise & GSD3: Is it important?

Living with GSD3 and finding difficulty in exercising, go hand in hand. The combination of muscle weakness, tiredness, possible breathlessness, aches and pains, all accompanied by the need to avoid a low blood sugar, makes even the thought of a gentle stroll in the nearby park a daunting prospect.

How to find that enthusiasm in the face of all those difficulties and possible poor advice?

Spanish scientists have shown us that exercise in GSD3 can be important. Children and adults took part in a 12-week exercise programme. Their progress was followed up and there were improvements in:

- Muscle strength
- Strength of the heart
- Lung capacity and less breathlessness
- A loss of fat
- Reduction in fatigue

The researchers concluded that exercise should be considered. If you want to exercise, do discuss this with your health care provider. You can find out more about the study here:

https://tinyurl.com/kw8b7x84

Advisor Presence in GSD Clinics

Elizabeth Davenport – Specialist care advisor

I've recently had the opportunity to attend multidisciplinary clinics at Salford Royal Hospital for adult patients with Pompe and other GSDs. It's been a fantastic experience, as it has allowed me to meet more members of our GSD community in person.

A big thank you to everyone who has come over to say hello, take a seat, and have a chat – it's been wonderful getting to know you all. I came prepared with our new leaflets and information resources, which were very well received by both the community and the metabolic team. This has been a great way to build connections and get to know the team at Salford better, helping us explore more ways we can support our community.

As my first clinic took place during Rare Disease Week, I was also invited to have a photo taken with the staff team, and AGSD-UK was

featured in the hospital newsletter and on social media – a lovely opportunity to raise awareness of our work.

I'm hoping to attend more clinics in the future, along with Zainib, our benefits and engagement and advisor, so we can continue strengthening these relationships and supporting you all.

Zainib Hussain – Benefits and engagement advisor

I have been working with the Pompe clinic at Queen Elizabeth Hospital Birmingham for over two years, collaborating closely with the specialist team and working with specialist nurses. More recently, I have also been supporting the hepatic clinic at Birmingham Women and Children's Hospital, working alongside specialist nurses to ensure children and families and patients receive the guidance and support they need.

In my role, I engage directly with patients and families in the waiting area, providing assistance to those referred by nurses who require additional support. I have supported many adults and parents, particularly those navigating the benefits system, helping them understand their options and achieve positive outcomes. Additionally, I've referred numerous clients to Elizabeth, who has successfully assisted with education health plans, university support, employment issues, and many other areas.

This collaborative work with specialist nurses and clinicians has fostered strong partnerships, enabling us to empower clients effectively. Together, we have achieved excellent results, helped more people, and received consistently positive feedback from patients and families. Our shared focus remains on supporting clients to access the resources and guidance they need to thrive.

'Having Zainib available for Pompe clinics has been very beneficial for our patients as well as staff. Zainib sits in the waiting room awaiting patients arriving in clinic where we introduce the patients to her and let them decide if they would like to engage.

Sometimes the patients are a little reluctant to speak, but with Zainib not pushing them and just offering general conversation and explaining her role within AGSD-UK, most patients tend to open up and realise that she can help with a lot more than they think. Aside from patients, staff knowing that Zainib is in clinic is also brilliant for our new patients which the consultants then inform the patient in their consultation and signpost them to her once their appointment is over.

I have too had some amazing support with Zainib outside of the clinic regarding patients and between us and what we both knew, we were able to support a patient and sign post that patient to appropriate service in a crisis. As a service and a charity, it is so important to have that relationship and Zainib is an amazing and knowledgeable person to support our service'.

Rare Disease Day 2026

This year for Rare Disease Day we put the spotlight on the night-time experiences of community members affected by GSDs. Faced with a range of challenges from night cramps to headaches to interrupted sleep and the anxiety of maintaining blood sugar levels, people affected by different GSDs talk here about what helps them get through the night.

Tharanikan Karan's story

"My son has GSD type 3a and is 4 years old. Each night, I gently check on him, guided by love and instinct, to make sure he is doing well and receiving the nourishment his body needs. If he needs more food, I am there without hesitation.

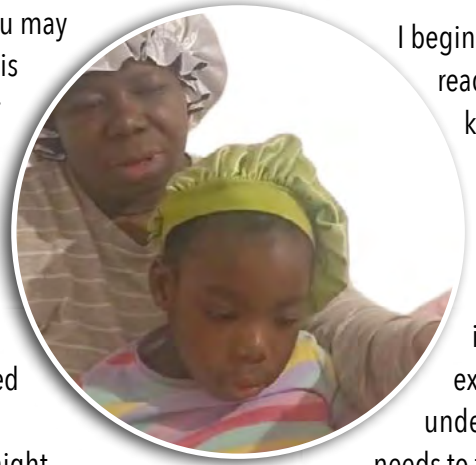
What once felt overwhelming has transformed into a steady, familiar routine that brings comfort and confidence. These quiet night time moments remind me of my strength and my deep devotion as a parent.

While the journey isn't always easy, it is filled with resilience, hope and so much love".

Gwen's story

"I was diagnosed with Pompe disease in 2015. A few years later, I began to notice that I was experiencing very painful headaches during the night. I knew this was one of the symptoms of Pompe disease, and I realised I needed help.

I spoke with my team at Queen Elizabeth Hospital, and it was decided that I needed to use a NIV (Non-Invasive Ventilation) machine. I have being using NIV since 2022. I wanted to share my experience with others in the community. I hope people know that if they are told they need NIV, there is hope. It can be life-changing, even if you may have heard others say it is uncomfortable, noisy, or that they do not want to wear a mask.



At first, I did struggle with the mask and found it uncomfortable. However, I knew I needed to persevere, and that is exactly what I did each night. I stayed positive because I knew it was necessary. My machine is quiet, and I use a half-face mask that sits under my nose.

I now sleep for around 8-9 hours most nights. What I really want people to take from this message is that if you are offered NIV, it is important to choose what works best for you. Your comfort matters. Getting 8-9 hours most nights can make a real difference.

I also take steps to make sure my bed is as comfortable as possible. I use a memory foam mattress and keep my bedroom calm, relaxing and comfortable".

Samuella's story

"My daughter has early-onset Pompe disease, and because of this, our evenings begin earlier than most families'. Bedtime is not a simple routine in our home; it is a carefully paced process built around her needs, comfort, and the strength she shows every day.

I begin getting Samuella ready for bed at 7pm, knowing that it takes time for her body and mind to fully settle. Rushing is never an option. Each step is intentional, shaped by experience and a deep understanding of what she needs to feel safe and calm.

Our routine starts with a warm shower. This moment helps her muscles relax after a day that often demands more from her than most people realise. Even everyday activities require extra effort, and by the evening her body is tired. After her shower, we brush her teeth slowly and gently, always working at her pace. These small, ordinary moments have taught us how important patience truly is.

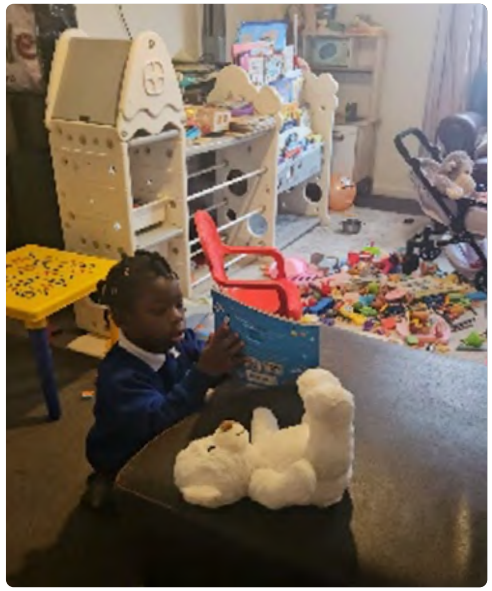
Once she is comfortable, we move into the quietest part of our evening: story time. We read her favourite books, using the familiarity of the stories and the sound of my voice to help her relax. These moments are about more than helping her fall asleep; they are about connection, reassurance, and creating a sense of peace before the night begins.

Night-time can be challenging. Samuella struggles with her breathing, and those hours are often filled with quiet vigilance. I listen closely, wake frequently, and remain constantly aware. While these moments bring worry and exhaustion, they also reflect the depth of love and responsibility that comes with caring for a child with complex medical needs.

As a mother, and as parents, this journey changed us profoundly. Samuella has made us stronger, more observant, and more resilient than we ever imagined possible. We have learned to notice the smallest changes, to trust our instincts, and to advocate fiercely for her needs. Through her, I have learned how to be better – not only as a parent, but as a person.

Despite the challenges she faces, Samuella continues to show incredible courage and determination. She is not defined by her diagnosis. She is a child full of strength, quiet bravery, and the ability to teach those around her what true resilience looks like.

Every evening routine, every sleepless night, and every small victory reminds us that while this journey is not one we chose, it is one that has shaped us with purpose, love, and an unwavering belief in our daughter's strength".



Amin's story

"When night falls, one person's world becomes quieter, but never truly still. As the lights go out and the house settles into silence, the soft, steady sound of a feeding pump takes over. For someone living with GSD 1A, this gentle hum is more than background noise–it is reassurance, routine, and a lifeline through the night.

Before sleeping, everything is carefully prepared. The pump is checked, the feed is measured, and the tube is secured. These small rituals bring comfort. There is always an awareness that blood glucose levels can drop quickly if something goes wrong, but that awareness has grown into confidence over time. Experience teaches what to look for, how to respond, and how to stay calm even when the unexpected happens.

Sometimes the pump alarms in the middle of the night. It might be a blockage, a disconnection, or an empty feed. Waking suddenly can be unsettling, but it is followed by action, not panic. A quick adjustment, a reset, and the steady flow resumes. Each moment reinforces resilience and trust in the routines that have been built night after night.

Sleep is often interrupted. Blood glucose checks, quiet movements, and the glow of a small screen become familiar companions in the dark. While rest may come in fragments, there is also a sense of safety in these moments. Every check is a reminder that health is being protected, even while the world sleeps.

Finding the correct feed rate is a delicate balance. Too much or too little can affect how the body feels by morning. Over time, this balance becomes clearer. The body gives signals, patterns emerge, and adjustments are made with care. What once felt overwhelming slowly becomes manageable.

Movement during sleep can sometimes cause the tube to shift. When that happens, it is met with patience rather than frustration. The tube is repositioned, the feed continues, and sleep returns. Daily attention to hygiene around the gastrostomy site also becomes part of the routine, helping to prevent infection and maintain long-term wellbeing.

Beyond the physical challenges, the night can bring quiet anxiety. There is always a lingering thought–what if something happens while I'm asleep? Yet with each night that passes safely, that fear softens. Confidence grows, built on consistency, preparation, and self-awareness.

Independence at night can feel limited, especially when support from carers is still needed. But this support is not a weakness–it is a bridge. It allows strength to grow, skills to develop, and independence to slowly expand over time.

Living with GSD 1A through the night is not easy, but it is powerful. Each night completed is an achievement. Each morning is a victory. This is the story of one person, facing the dark hours with courage, adapting to challenges, and waking up every day stronger than the night before."

Amanda's GSD9a Story

25 The number of feeds/meals the children need to eat between 7am and 1:30am

36 The number of balanced sugar free, low complex carb, high protein meals/snacks we provide to our children per day. Between 5am and 1am.

1 The number of our children genetically diagnosed and in receipt of specialist health care.

15 The minimum number of finger pricks and glucose checks per day.

3 The number of our children affected.

5 The maximum number of hours we get between meals, milk, and snacks overnight

10 The number of years this condition has dominated our lives

1 The number of people confirmed type 9a in Scotland, our eldest son Chance

Hypoglycaemia – Pale, lethargic, eyes rolling, unable to speak, shaking, sweating.
Ketosis – Vomit, headache, sore tummy, gagging, loss of appetite.
Enlarged liver – Distended abdomen (big tummy).
Other issues - Low iron, low vitamin D and low muscle tone.

All of these can be corrected with treatment. All caregivers can achieve this for their loved ones.

Jodi's GSD3 Story

Every night while most people are going to sleep my body is preparing for a fight. Because of my condition, glycogen storage disease type 3a, my body can't store and release energy the way it is supposed to.

That means while I sleep, my blood sugar can drop to dangerous levels. So, I don't get the choice to just rest. I have to connect myself to a feeding pump. This tube is not just medical equipment. It is how I survive the night. It slowly delivers nutrition while I sleep keeping my blood sugar stable and helping prevent serious complications like seizures, coma, muscle breakdown and even death.

Without it something as simple as sleeping could become life threatening. It's a constant reminder that my body works differently. But it's also the reason I wake up the next morning, this is my normal.

When my blood sugars get low, I get dizzy, shaky and my brain feels cloudy, sometimes I even talk backwards. Panic creeps in because I know my body is running out of fuel. Sometimes I can feel it start to get low, but there are times when it drops suddenly with no warning, and I pray that I don't pass out before I make it to the kitchen.

Low blood sugar can lead to muscle breakdown, when my muscles don't get energy, my body starts to break them down. It feels like a deep crushing soreness that won't go away, like as if I have been hit by a train or a tsunami, like a trailing horse times ten all throughout my body. Even small movements hurt, even to be touched is unbearable. It's not just pain, it's a warning, my body is literally using itself to survive.

That's why keeping my blood sugar stable with tube feeds, isn't a choice, it is survival.

This is my GSD journey

These are the words of Jodi Bateson. Jodi comes from Michigan in the States and been vlogging the story of her life with GSD. She has kindly allowed us to reproduce the latest vlog for our magazine. She wants to spread awareness and help people to not feel as alone as she has, so many times.

International Pompe Day

The goal of International Pompe Day, 15th of April, is to foster international awareness of Pompe Disease, a life-changing inherited condition.

The parents of little Arla from the Wirral are keen to help raise awareness of infant onset Pompe and worked with AGSD-UK Pompe advocate Claire Wright in the run up to International Pompe Day to gain media coverage, with a digital reach of 7.9 million people.

The family first became concerned about Arla when she was repeatedly becoming unwell with chest issues. She was very weak; her parents were worried she was not feeding properly and was losing a significant amount of weight.

Arla's Mum, Emmie Wood said;

"We knew something wasn't right, so we kept taking her back to the doctors. Eventually, our GP referred her to the hospital, where further tests were carried out. During these tests, they discovered that her heart was enlarged, which was a huge shock for us. Following this, genetic testing was done, and that's how we finally received the diagnosis of Infantile Onset Pompe Disease (IOPD). While it was devastating news, it also gave us some answers after a very worrying and uncertain time."

Daily life for the Wood family requires constant adjustment, resilience, and planning. Much of their routine revolves around managing medical needs – appointments, treatments, and monitoring Arla's health.

Emmie went on to say; "Emotionally, it can be very challenging. There are moments of anxiety and exhaustion, but we try to focus on the positives and carry on day by day."

Emmie said, "From the moment of diagnosis, AGSD-UK provided clear, reliable information that helped us understand Pompe Disease and what to expect.

"They helped us apply for grants during Covid when things were hard with money as Arla was an inpatient in Manchester hospital and I was at home with three other children. They have been amazing and helped us along the way with anything we have needed. They have really been there for us."

AGSD-UK commented:
"Families affected by Pompe can face huge challenges and it's vital they get the early support they need. We want to see consistent signposting at diagnosis to organisations like AGSD-UK, so no family misses out on the range advocacy and support services we offer.

"People affected by Pompe and other glycogen storage disorders tell us just how isolating life with this rare and a little-known condition can feel. We're here to provide a listening ear, connect people to others who understand their experiences and make sure all-round help and information is available to enable them to live positively.

"While pushing for greater awareness, along with continued research into better treatments and ultimately a cure, we want to ensure families living with Pompe feel supported, informed, and empowered from the very beginning of their journey."

Welcome to Dr Chiara Pizzamiglio

We are very pleased to offer a warm welcome Dr Chiara Pizzamiglio, Consultant Neurologist, who became the new clinical lead for the NHS highly specialised service for McArdle disease and related disorders in March this year.

Dr Pizzamiglio trained in medicine and neurology at the University of Milan and Novara (Italy). During her neurology residency, she developed a strong interest in neuromuscular disorders and subsequently moved to London where she completed her training with an attachment to the MRC Centre for Neuromuscular Diseases, National Hospital for Neurology and Neurosurgery, where she worked within the McArdle Disease and Related Disorders service under the supervision of Professor Ros Quinlivan.

She was appointed as a Consultant Neurologist for the NHS Highly Specialised Service for Mitochondrial Diseases in 2024, and in 2026 also to the McArdle Disease and Related Disorders Service.

Alongside her clinical work, Dr Pizzamiglio has been actively involved in research on McArdle disease, contributing to multiple publications since 2019.

Dr Pizzamiglio has kindly agreed to present at our **Main Event**, in Loughborough on 4th/5th July, providing an opportunity for many McArdle's people to get to know her.

We look forward to many years of liaising closely with Dr Pizzamiglio, to continue the great work of the McArdle's clinic in supporting people diagnosed with this ultra rare condition.

Dr Pizzamiglio has contributed to the following publications relating to McArdle's:

Clinical impact of supervised resistance training in patients with McArdle disease: a case series.

https://tinyurl.com/2vy7n7be

The McArdle assessment of severity tool (MAST): validation of an innovative new tool to measure the clinical severity of patients with McArdle disease.

https://tinyurl.com/3bc6vksp

The McArdle disease physical activity paradox and insights into exercise adaptation: a case study on running with McArdle disease.

https://tinyurl.com/2s446ypj

Photos: Dr Chiara Pizzamiglio
On a fund-raising Park Run, third from right

Natural history of McArdle disease: a single centre study of a cohort of 220 patients. 2021

https://tinyurl.com/5y4x4hnr

Phenotype and genotype of 197 British patients with McArdle disease: An observational single-centre study.

https://tinyurl.com/mptzzdsz

Frequency of coronary artery disease in people with McArdle disease.

https://tinyurl.com/4nx7d7jb

Is the expression of muscle glycogen phosphorylase tissue-specific? New perspectives on McArdle disease.

https://tinyurl.com/4sr2mw9d

Natural history of McArdle disease in a cohort of 220 patients. 2019

https://tinyurl.com/y789pnkc

In future, as part of my role in the Glisten editorial team, I'm now going to be coordinating content for Glisten that relates specifically to McArdle disease. So, if there's an article or information you would like to contribute or topics that you would like to see included, please email me.

In the next issue I'd like to include an article about cycling and e-bikes as I've found with the advent of e-bikes, it's a great way to not only stay fit and active but also participate in an activity with non-McArdles friends. If you are a keen cyclist with experience to share, please send it by email marked for my attention to **info@agsd.org.uk.**

Regulatory progress for Ultragenyx DTX401 gene therapy for GSD Ia

In February, ***Ultragenyx*** announced that the U.S. Food and Drug Administration granted Priority Review for the Biologics License Application for DTX401, an investigational gene therapy for GSDIa, and set a decision date of August 23, 2026. Priority Review does not mean approval, and FDA will evaluate the totality of the data before making a decision. Ultragenyx shared a social graphic *here* explaining this news and outlining the next steps in the FDA review process.

They will continue to share updates with you throughout this process as they are able.

Beam-301 gene therapy trial for GSDIa

BEAM-301 aims to correct the most common disease-causing mutation, R83C, in patients with glycogen storage disease type Ia (GSDIa). BEAM-301 has the potential to normalise blood glucose in these patients without continuous supplementation and improve key metabolic parameters. BEAM-301 is currently being evaluated in an open-label Phase 1/2 dose-exploration trial in patients with GSDIa. Dosing is complete in the first cohort and enrolment has been initiated in the second cohort. Beam expects to report initial clinical data in 2026.

Grants that can help community members

AGSD-UK specialist care advisor Elizabeth Davenport describes how she can help community members access grants.

I can help people in the GSD community find and apply for grants that can make everyday life easier. I spend time researching different charities and funding opportunities across the UK so I can point people in the right direction and help them understand what they may be eligible for. For many people, the application process can feel overwhelming, so I try to break it down and support them through it.

Grants can offer valuable support in many areas of daily life. This may include help accessing workplace adjustments, mobility aids, or funding for equipment that promotes independent living. Support may also be available to assist with essential household costs, such as white goods and other necessary bills and items. In addition, grants can help provide sensory equipment, technology, and opportunities for children and young people to take part in activities that might otherwise be out of reach.

What motivates me most is seeing the impact these grants can have. Something that might seem small to others–like funding for a piece of equipment or a sensory item–can transform someone's quality of life. By sharing information and helping with applications, I hope to make funding opportunities more accessible and ensure people in the GSD community know they're not alone and that support is available.

These are some of the UK grants and funding sources I signpost people to, however this list is not exhaustive:

- Access to Work – Government funding that helps disabled people stay in work or start a job by covering things like equipment, support workers, or travel.
- Cost of living support – www.gov.uk/cost-of-living/bills-housing-health. Find out what support is available to help with bills and costs in England. This includes support with utility bills, support from your local council and Council Tax Reduction.
- British Gas Energy Trust – has now introduced a White Goods Fund to help support clients in England and Wales experiencing financial hardship.
- Barchester's Charitable Foundation – Provides small grants for mobility aids, wheelchairs, and other equipment for disabled adults.
- Willow Foundation – Bespoke special breaks, days and treats for young adults.
- Independence at Home – Helps fund essential household items and equipment that allow people to live independently.
- Florence Nightingale Aid in Sickness Trust – Provides grants for medical and disability equipment.
- Disabled Facilities Grant – Local authority grant for home adaptations like ramps, stairlifts, or accessible bathrooms.
- Glasspool Charity Trust – Glasspool is a UK charity that provides small grants, through support organisations, to people in financial hardship for essential everyday needs.
- Family Fund – One of the largest UK charities giving grants to families raising disabled or seriously ill children.
- Cash for Kids - Support children from birth up to and including 18 years of age who are affected by poverty, illness, or who have additional needs.
- Wooden Spoon Charity – Funds equipment, facilities, and opportunities for disadvantaged or disabled children.
- Turn2us - Find out what help may be available to you through benefits, grants and other sources of support.

For more information or support, contact our AGSD-UK specialist care advisor – elizabethdavenport@agsd.org.uk

Gina's McArdle's Journey

Website information led to a diagnosis

Hello, I am Gina Ortiz, age 27, from the USA.

For 6 months before getting diagnosed I had been reading and learning a lot about McArdle's, mainly from the IamGSD and AGSD-UK websites. I related to every single personal story on the sites, and that encouraged me to bring my symptoms to a doctor for the first time.

I feel both happy and bittersweet now to have McArdle's confirmed. Happy because I now have an answer, but bittersweet because it took over 20 years, and a lot of emotional (and physical) pain to get to this point.

I was born on a US Army base in 1998 and lived on military bases my whole life. Looking back, I can clearly see all the barriers that made my condition overlooked and hard to identify. The military life in itself is its own isolated world. Physically you are isolated on a fortified base and socially you are isolated to your immediate family and other military families. You move to a different base every few years and you go to military hospitals.

I think doctors on military bases are not trained to recognise genetic conditions in soldiers or their family members, or to think about genetic conditions being a possibility. Back In 2015 I went to the ER because I hurt my arm at school and it became stiff. I am pretty sure I was having a muscle contracture and/or rhabdomyolysis. But the military doctors only gave me fluids and observed me overnight before sending me home the next day. Also, I don't think children born to military parents have newborn genetic testing done either. At least not in 1998, but maybe (hopefully) it is different now.

I feel mostly normal again since hurting myself last August but I'm still having some soreness in my legs and chronic fatigue. I've been reading about the ketogenic diet in relation to McArdle's and I think I may have been unknowingly doing this diet before I even knew what McArdle's was. I think I may need to experiment with different diets though. The problem is I don't feel hungry in the mornings, and my first meal usually is not until the late afternoon. I am also trying to work on my mental health and self-esteem after feeling very negative about myself for so long. I know this will take some time.

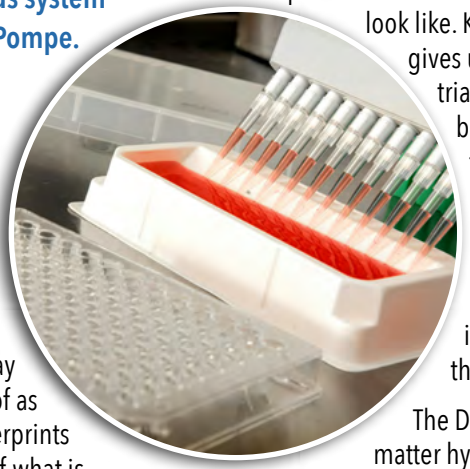
I joined the McArdle's Facebook group last year and I am enjoying talking with McArdle's people. I am looking forward to learning how to live better with McArdle's and prevent injuries going forward. Thanks so much to IamGSD and AGSD-UK for the information which helped me get on the path to diagnosis.

Photos: Gina Ortiz

Gina on a hike in Washington State, USA.

New biomarker could guide future brain-targeted therapies for Pompe disease

Researchers at Duke University School of Medicine in America are investigating a potential biomarker to better detect and monitor central nervous system (CNS) involvement in Pompe.



Biomarkers are biological markers in the body, such as molecules or processes, which can be measured or observed. They are used to help to find out if there are changes happening in the body and why this may be. They can be thought of as "biological clues" or fingerprints that provide a snapshot of what is happening in the body at a given moment. Biomarkers are essential for developing new treatments, enabling personalised medicine, and enabling faster, more accurate diagnoses than relying on symptoms alone.

Current Pompe enzyme replacement therapies remain the standard of care for treatment, but they do not cross the blood-brain barrier. This leaves Pompe central nervous system symptoms unaddressed as patients get older.

The team at Duke evaluated something called a glial fibrillary acidic protein (GFAP), which is a biomarker associated with astrocyte activity in the brain. Astrocytes are star-shaped glial cells in the central nervous system that help form the structure of the brain.

Kristen Hagarty-Waite, PhD, postdoctoral associate and co-author, has been focusing on the more severe end of the disease spectrum to determine what the biomarkers look like. Kristen has said, "This gives us an idea of a clinical trial endpoint for blood brain barrier crossing treatments which are currently being developed, but we need to create the infrastructure to show industry how important this is."

The Duke team found white matter hyperintensity – bright spots that show up in an MRI that signal damage to the white matter of the brain. "We are seeing increases in white matter hyperintensity over time in some," Hagarty-Waite said. "So, figuring out how GFAP correlates and changes over time helps us build a picture of what can happen.

Findings suggest this new biomarker may more accurately reflect the degree of neurological involvement and could serve as a valuable tool for identifying children at risk for developing CNS disease. It could also be useful in tracking disease progression and response to emerging therapies.

https://tinyurl.com/5avhtkye

The Generation Study now testing newborns for more GSDs

GSD types 6 and 9 have been added to other GSDs and rare conditions being tested for as part of The Generation Study.

The study is looking at the utility and feasibility of using genome sequencing to screen newborn babies for rare conditions, in order to deliver more timely diagnoses and access to NHS care and treatment.

The Genomics England study, run in partnership with the NHS, is now operating in 79 hospitals across England and has so far involved more than 66,000 participants.

In latest developments the study will now include testing for the PYGL gene associated with GSD6 and the PHKA2 and PHKG2 genes associated with GSD9.

This is in addition to the G6PC1 and SLC37A4 genes associated with GSDI, the GAA gene associated with GSD2 (Pompe) and the AGL gene associated with GSD3.

You can find out more about the study here: Generation Study.

Retirement of Dr Richard Godfrey

We are very pleased to acknowledge the contributions to the understanding of McArdle Disease made by Dr Richard Godfrey, who has recently retired from his involvement with the McArdle's Clinic. He is Senior Lecturer in Coaching and Performance at Brunel University in Uxbridge, west London. His interests extend to sports performance and increasingly to health-related aspects of exercise.

Richard previously served 12 years as a physiologist at the British Olympic Medical Centre (BOMC), the British Olympic Association's Department of Science and Medicine. During the last 7 years with the BOA he was Chief Physiologist of the BOMC and was involved in organising physiology service provision to elite sport. Richard has appeared in the press, and on radio and TV, on more than 40 occasions.

Richard became involved with McArdle's through John Buckley, who was the exercise physiologist assisting the UK McArdle's clinic when it was based in Oswestry, before it moved to London. It came about because back in 2003 Richard was doing some research on the role of lactate in the exercise induced human growth hormone response. It was useful to study some McArdle's people, because of our lack of lactate response to exercise. He came to the clinic and monitored people on the treadmill.

When the McArdle's clinic moved to UCLH in 2011 and changed from 4 clinics per year to about 25 clinics per year, it became practical for Richard to become regularly involved. He would oversee the exercise assessments of patients at the clinic, and over the years helped induct physiotherapists into their role. Even more importantly Richard became involved in McArdle research projects both with the McArdle's clinic and with Euromac. He contributed to a dozen or more projects and studies, cooperating with many leading McArdle's researchers from around the world. He has been involved with 16 papers and 12 conference abstracts (posters) on GSDs, managing to fit us in amongst the 100+ research papers to which he has contributed.

Rather tantalising is his work on investigating the possibility of a "Third Wind" in McArdle's. The full paper did not reach publication, so the subject remains an enticing prospect for future researchers to take up.

Richard suffered a heart attack in 2007. He undertook 60 weeks of high intensity interval work in the gym three times a week as an experiment to see if he could reduce the scar tissue from the heart attack. Cardiologists are adamant that scar tissue in the heart muscle is permanent and immutable. Clearly, that is incorrect as he has reduced his myocardial scar tissue by 55%. Richard's account made a significant contribution to medical science, published as an editorial piece in Experimental Physiology 2025; 110: 1017–1022.

Another major contribution which Richard and colleagues made to medical science arose from his work with blood lactate levels. This led to the development of a simple and very low cost pre-screening diagnostic tool for cancer. By making a diagnosis months ahead of conventional tests, it has the potential to save millions of lives around the world. It was published as Elevated Circulating Lactate Levels and Widespread Expression of its Cognate Receptor, Hydroxycarboxylic Acid Receptor 1 (HCAR1), in Ovarian Cancer in the Journal of Clinical Medicine 2023; 12: 217.

We thank Richard for his service to McArdle's and wish him a long and happy retirement.

Photos: Richard Godfrey, photo Brunel University
Supervising exercise assessment at the McArdle's clinic

AMICUS UPDATE

Amicus Therapeutics, a rare disease company active in the Pompe community, has been acquired and is now a wholly owned subsidiary of BioMarin Pharmaceutical Inc.

The companies say the purpose of bringing the organisations together is to strengthen support for people and families affected by Pompe and, over time, reach more people globally affected by rare conditions.

BioMarin is a global rare disease biotechnology company focused on delivering medicines for people living with rare genetically defined conditions. Like Amicus, BioMarin's origins can be traced back to a single family. The story began with a Dad whose son was diagnosed with a form of mucopolysaccharidosis (MPS). The family was told that the son wouldn't live to see his 10th birthday – a fate the family refused to accept.

BioMarin leadership emphasise that supporting the Pompe community is a priority and that they intend to continue–and build on–the relationships Amicus has established with advocacy organisations and members of the community. If you have questions about what this means, please speak with your healthcare team, or get in touch via info@agsd.org.uk

IS AI ALWAYS THE ANSWER?

AGSD-UK benefits and engagement advisor Zainib Hussain describes the pitfalls of using AI when making applications.

I have noticed that more clients are using tools such as ChatGPT to help with their Personal Independence Payment and Disability Living Allowance applications. Many are doing this because they feel unsure about how to explain their needs, or they worry that they may not be taken seriously if they do not use the "right" wording. This often comes from a place of anxiety and a genuine desire to be understood.

While this type of support can help with structuring answers and building confidence, there are occasions where the information provided does not fully reflect the client's individual circumstances or day-to-day experiences. At times, the wording can become more focused on meeting the scoring criteria, rather than describing the client's actual difficulties.

For example, a client may submit an application stating they are "unable to prepare a meal at all" because they have seen that this may score higher points. However, when discussing this in more detail, they may explain that they are able to prepare simple meals on some days, but struggle due to pain, fatigue, or safety concerns on others. While their difficulties are very real and significant, the original statement does not accurately reflect the full picture. This can lead to inconsistencies if the information is explored further during assessment.

These situations can unintentionally affect the outcome of a claim. As a result, some clients return for further support after receiving a decision that does not meet their expectations.

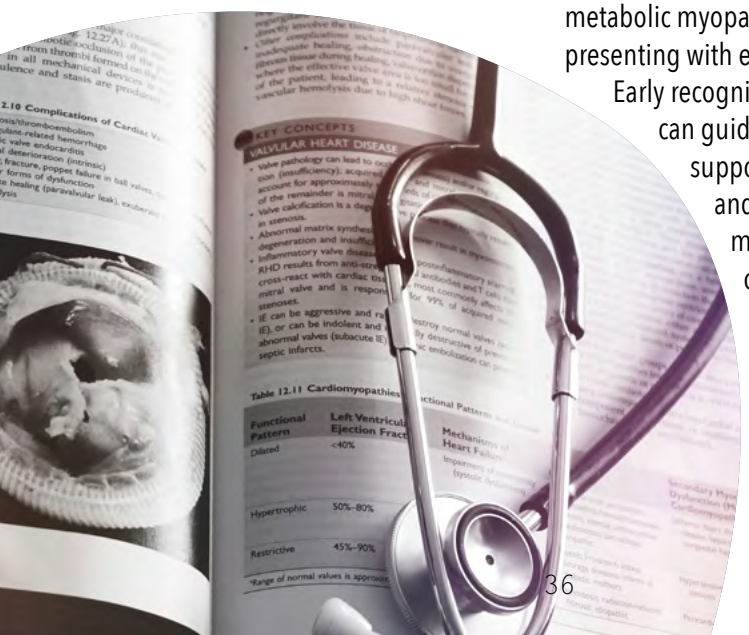
It is therefore important to gently support clients to describe their experiences in an honest, balanced, and personal way. Emphasising real-life examples, variability, and the impact of their condition can help ensure that their application is both accurate and reflective of their needs. Providing reassurance can help clients feel more confident in sharing their true experiences, which is key to achieving a fair and appropriate outcome.

This is one reason why charities like AGSD-UK exist, to help and support you with any questions and concerns you may have. We will work closely alongside you to navigate your PIP and DLA applications to get the result that is best for you. Contact one of the team today to find out how we can help you – info@agsd.org.uk.

How we monitor McArdle's research

Every week Google Scholar Alerts sends me a few notifications of new results for McArdle disease. Very often these are instances of McArdle's being mentioned in papers which are dealing with a much broader subject. However, at least once a month, sometimes once a week, there is something of direct interest with regard to McArdle's.

These mentions are very often of interest to the medical science community but have little or no relevance to the average McArdle's person. I try to keep an eye out for the few which might be directly of interest or even beneficial to us. Sometimes these papers are available under ***Open Access***, meaning that somebody has paid the publishers to allow the paper to be available to all, as opposed to being only accessible within a subscription-based journal. But for the majority of papers, we can only gather a few ideas of what the paper is about.



Here are three examples of ***Google Scholar Alerts*** followed up in recent weeks. These represent about 20% of the alerts received during that period, and even these are only of very peripheral relevance, the first being by far the most promising.

Exertional Rhabdomyolysis in a Young Male with Suspected McArdle Disease

An open access abstract published by ***Cureus***, March 2026. It could help raise awareness and improve the rate of diagnosis as the conclusion stated:

"This case highlights the importance of maintaining a high index of suspicion for metabolic myopathies in young adults presenting with exertional rhabdomyolysis. Early recognition of McArdle disease can guide management strategies, support safe exercise practices, and help prevent recurrent muscle injury and long-term complications."

Ventricular Tachycardia in a patient with McArdle Disease: Insights into a rare cardiomyopathy etiology with magnetic resonance imaging

Presented as a poster contribution at a medical conference and published in the Journal of the American College of Cardiology. This one is a bit academic but could be of interest to the medical professionals considering possible cardiac involvement in McArdle's.

"Our case highlights a novel cardiac manifestation of McArdle disease and suggests a possible need for cardiac MRI evaluation in these patients. Recognition of arrhythmic risk may alter management and broaden understanding of the possible cardiac involvement in metabolic myopathies."

Preimplantation Genetic Diagnosis, Testing or Treatment

This is an extract from the policy manual of the *Arkansas BlueCross and BlueShield Association*. The only reference to McArdle's is to the two most common mutations in the PYGM gene. However, it does raise the prospect of one day being able to routinely diagnose McArdle's either before or shortly after birth.

"Preimplantation genetic testing involves the analysis of biopsied cells as part of an assisted reproductive procedure. It is generally considered to be divided into 2 categories. Preimplantation genetic diagnosis is used to detect a specific inherited disorder… and aims to prevent the birth of affected children to couples at high-risk of transmitting a disorder. [It] may also involve testing for potential genetic abnormalities… for couples without a specific known inherited disorder."

Andrew Wakelin

PREGNANCY & GSDs

This charity loves welcoming newborns to the community, and that joy is even more when the mother has GSD. Carrying a pregnancy along with GSD can be a time fraught with worry, however there are many people, who with the right care and attention have successfully become pregnant and raised a healthy family.

Doctors, frequently, and with pride, will write in the medical literature about the successes they have had in helping women through their pregnancy. Many of their accounts will also detail the particular regime they used to help that one woman, usually from when the pregnancy is known about, to the months following the birth.

The following are just a few of the very many examples that have been written about.

German doctors have reported on the birth of a normal baby boy to a lady with GSD6. The baby was successfully breastfed.

Australian doctors describe a woman with GSD3 who went through pregnancy with no episodes of hypoglycaemia. They attributed this to careful metabolic management.

More doctors in Germany talk about the management of pregnancies in two women with GSD 1b, one of whom was taking empagliflozin (a drug taken by many with 1b to aid the immune system). Two healthy baby boys were born, both of whom were breastfed.

Of a cohort of 127 females with GSD5 (125) or GSD7 (2), a retrospective review found no particular problems had occurred in the antepartum/intrapartum or postpartum periods. Most had their babies before their diagnosis and had the same incidence of interventional delivery as the general population (16%). Anecdotal reports show improved activity tolerance during pregnancy.

The doctors in London have written about 15 successful pregnancies in 7 women with GSD3. They concluded that the overall outcome of pregnancy for the mother and child is good.

It seems that having a family and having GSD are not incompatible. If you are thinking of a pregnancy, do talk to your doctor and then come along to one of our meet ups or a conference to share your good news. Don't forget we are here to support you if needed.

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, National Lottery Community Fund, Mosawi Foundation, Sanofi, Ultragenyx and Vitaflo.

AGSD UK
Association for Glycogen Storage Disease

Upcoming events

- AGSD-UK Main Event 4-5 July 2026 Loughborough
- McArdle's walking courses: August 2026 Pembrokeshire
- Hepatic GSD updates with expert team from Gronigen, Netherlands 31st October 2026 online

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 2026/27

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