Autumn 2024

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

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

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Thank you!

Strength in numbers!

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

Join or renew your membership today.

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

We're offering free membership until April for anyone who joins or renews their membership between now and the end of the year.

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

Front cover images:

Top: Hepatic get together

Middle left: Elle McNally and her boyfriend

Middle right: Roman Tank and his mum Leanne

Bottom left: McArdle's walking course

Bottom right: McArdle's walking course evening activities

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

Welcome to the Autumn 2024 edition of Glisten.

There's lots of evidence that getting involved is good for you. Involvement can bring benefits for your sense of wellbeing and purpose, whether from being part of something bigger and helping to make a difference for a cause you believe in, or through making connections with others and feeling like you're on the same side.

This edition of Glisten includes a number of ways to get involved.

There's an article about research participation, setting out how involvement can have an impact on the development of treatments and pave the way to improvements in care and quality of life for people affected by GSDs.

A piece about patient registries shows how these can also offer a way to support GSD research and developments, simply by allowing your data to be used anonymously.

Our forthcoming Main Event is a great opportunity to get involved with the GSD community, with the chance to build lasting, supportive relationships. We're really looking forward to seeing so many of you there. The programme includes a session on involvement with AGSD-UK for anyone interested, and in this edition two AGSD-UK volunteers reflect on their experience of volunteering and how rewarding they've found it.

Being a member is an important way to get involved with AGSD-UK, helping to shape what we do and raise our voice. You can find out more about this on the inside page.

There's also coverage of those involved with fundraising, like Sioned Williams and the Tank family, who make huge efforts to raise money for our crucial services. Plus there are some suggestions about festive fundraising activities you might like to try.

We're immensely grateful to everyone who gets involved, whether through fundraising, membership, research or volunteering. You make such an important contribution to the GSD community. We hope you feel proud of the difference you all make.

Val Buxton

Fundraising Round-up

Roman's Story

Since we first learned about our son Roman's condition, GSD III, and began to slowly understand it better, we felt it was important to share this information with our family and friends. We wanted to raise awareness of his condition in a way that would not only inform those close to us but also offer them an opportunity to support Roman and others like him. After doing some research, we found that partnering with a charity and raising awareness through fundraising would be a meaningful way to achieve this.

By creating a JustGiving page, we were able to tell Roman's story and provide a detailed explanation of his condition, how rare it is, and the challenges we face as a family. The platform made it incredibly easy for us to get started–within minutes, our page was live and we could include a bio about Roman, along with a direct link to the AGSD-UK website for more information. This allowed us to share not only his journey but also the larger picture of GSD III with those who wanted to learn more and offer their support.

The response has been nothing short of overwhelming. In such a short time, the amount of awareness we've raised, as well as the donations that have come in, have been far beyond what we expected. Seeing how many people have come together to support Roman has been heartwarming and uplifting for us. It has shown us the power of community and the difference that awareness can make.

We are incredibly proud of what has been achieved so far, but this is only the beginning. We plan to continue supporting AGSD-UK and raising awareness about GSD III. It's our hope that by sharing Roman's story and engaging with our community, we can contribute to greater understanding and support for those affected by this condition. We are committed to doing everything we can to help make a difference, both for Roman and for others living with GSD III.

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

So far, the Tank family have raised **£4453**

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Fundraising Update Continued

Tri-ing for Hugo and AGSD-UK

In the Spring edition of Glisten, we mentioned that our chair Flo Kinnafick and a team of family and friends took part in the *Outlaw Half Triathlon*. They all did fantastically and raised a total of **£2611**

https://www.justgiving.com/page/tri-ing4hugo

Winner Winner Chicken Dinner

£500 was donated from *Winner Winner Chicken Dinner Ltd*.

An Evening with Sioned Williams

Sioned Williams will present a programme of harp playing, life-stories and poignant and fun anecdotes, in a most prestigious series of events organised by *Kettner Concerts*.

Sioned is an international concert harpist who performs regularly worldwide, and many would not be aware of her disability. This event will be emphasising the abilities we all have despite the complex lives we experience living with our rare conditions.

Sioned has been offered this opportunity specifically to raise awareness of McArdle disease and other GSDs and to create a platform for donations. This will be done by inviting specific people who are well-known patrons of the arts or disabilities by welcoming the general public and by holding a charity auction, among other activities. AGSD-UK will be there to introduce and promote the cause. Sioned would welcome any help beforehand to make this a stand-out night with long-term impact.

Please contact Sioned on sionedwilliamsharp@mac.com or on 07810 648658 should you wish to be involved.

Some of you will have had the chance to enjoy Sioned's wonderful performance at last year's Main Event.

Festive Fundraising Ideas

AGSD UK
Association for Glycogen Storage Disease

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
- Asking for donations instead of Christmas cards
- Sending AGSD-UK Christmas cards; contact info@agsd.org.uk to order, in return for donation
- Christmassy community dog walk
- Christmas themed quiz night
- Festive songs or carol 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

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McArdleites tackle the Brecon Beacons

Harriet Thomas-Bush reports on the McArdle's walking courses for 2024.

Every year, a group of young people and adults with McArdle disease live together in Wales for a week to walk and learn from each other. We rotate location each time among the three National Parks of Wales. This year from 29th July to 9th August, McArdleites from around the world came together in Bannau Brycheiniog (the Brecon Beacons).

We kicked things off with an ***Under 21s*** course, with a lot of returners and a sprinkling of newcomers, the younger ones with parents. We met in the very comfortable Penyrwrlodd farmhouse just outside Hay-on-Wye. We had a week of learning, walking and fun, expertly led by Bronte Thomas and Eoghan Ross, with guidance from Andrew Wakelin.

Each day began with breakfast, a few learning points such as the *Rules of the Road* to protect everyone, and a briefing on the day's walk. Then we headed out on the routes, which became increasingly more challenging as the week went on. Easing in with a couple of miles on the flat, we progressed to a long walk under and around the waterfalls and a mountain, Hay Bluff. The week peaked with climbing Pen y Fan, the highest mountain in southern Britain and part of the Welsh ***Three Peaks Challenge***. We were incredibly lucky with the weather, which came as a shock to some – more suncream was definitely needed!

A happy group from the Under 21s Walking

In the evenings, we enjoyed dinner together as a group, lovingly prepared by Rachel Thomas and Harriet Thomas-Bush (me!), with much assistance from everyone else. We then had a presentation on various techniques to practise the next day. One evening there was a talk on all the IamGSD resources – website, publications and videos. Then everyone discussed living with McArdle's, particularly as a young person. After this, everyone relaxed over a jigsaw puzzle, friendship bracelet making, reading books together or a face mask! We also managed to fit in a tourist day in "book town" Hay-on-Wye, and a trip to the river for a paddle with an ice lolly.

A special thanks to Ali and Sioned who prepared the most incredible Persian banquet for us all to celebrate a birthday, after climbing Pen y Fan. Everything was amazing and we were so grateful.

I know we're all extremely proud of this amazing bunch of young people and everything they achieved in this week – you climbed mountains guys!!! The biggest thank you to Bronte, Eoghan and Andrew for your leadership and expertise.

After an emotional goodbye to the children and young people, we welcomed the adults, mostly new participants from around the world. This week was led by David Thompson with assistance from Andrew and followed a very similar routine of walks, briefings and dinners. A slightly shorter course, with less fortunate weather but nevertheless another mountain was climbed! Everyone takes a lot away from discussing their condition with people who understand it and taking advice from how others

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manage both day to day activities and interactions with medical professionals. This happens both on the walks and in the evenings.

Again, congratulations to all the adults on your successes this year and thank you to David for stepping in to lead everyone. Thank you to everyone who offered to drive us to our walks and thank you to everyone who helped us cook!

The biggest thanks go to Andrew Wakelin, without whom none of this would be possible. Thank you for creating the most wonderful environment that we all look forward to every year. We have a network of amazing friends and family because of you!

Next year we're off to Eryri (Snowdonia) to hopefully climb Yr Wyddfa (Snowdon) – weather permitting of course – with some trips to castles and a lot of ice cream thrown in. We hope you're able to join us!

A few people were heard to say it had been the best week of their lives, and one said the only thing wrong was that she had not come when she first heard of it 10 years ago! But we will leave the final words to one of our returning teens:

"I enjoyed the amazing walks that we went on and the cooking. I really liked the activities we did after the walks, like bracelet making, playing games and paddling in the river!" – Sarah

COURSES 2025

Plans are developing for two courses in Eryri (Snowdonia) **1st - 8th August** for Children & Parents plus young people, **9th -15th August** for the adult course.

Watch the website or Facebook group for details.

Register your interest at:
type5@agsd.org.uk

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Local Hepatic & Pompe Get-togethers

AGSD-UK holds get-togethers in different parts of the UK to give community members the chance to make connections with others affected in their region.

This year gatherings included a cosy afternoon tea by the fireside at a pub in Leeds for people affected by Pompe giving the opportunity to chat, share advice and have a good catch up. More recently families affected by hepatic GSDs came together to swap stories and share experiences over afternoon tea in Reading.

As shown here, feedback has been hugely positive, with people expressing how much they value the chance to connect in a supportive, informal setting.

Look out for more opportunities to get together in different parts of the UK next year. You'll be sure of a friendly welcome among people who understand.

"Thank you for organising the event on Saturday. The location and the hotel were really nice... The afternoon tea was great too. We really enjoyed meeting the other families and sharing our experiences - it makes us feel that we are all on the same journey. The girls feel like there are other people out there with the same condition who are sharing the same journey and the struggles they face."

"What a lovely afternoon, thank you for organising it. So nice to put names to faces."

"We had an amazing time and thoroughly enjoyed ourselves. It was such a valuable experience to meet other families facing similar health challenges, which made us feel much less isolated. Living with such a rare condition can be tough, especially when people don't always understand what we go through, so connecting with others who do was incredibly comforting."

Pompe & Exercise

By Elle McNally

I was first diagnosed with late onset Pompe in April 2020 at the age of 21, during the COVID-19 pandemic. At the time, I was living in a ground floor flat at university. I struggled with muscle stiffness, high fatigue and could not bathe or shower without assistance. In June 2020, I moved back to my hometown and into a house. The first few nights I was there, I had to physically crawl up the stairs as I couldn't manage to stand and climb steps.

In July 2020, I started ERT for the first time and within a month I noticed a change in my mobility and the pain started to lessen. By June 2023, the ERT I was on (Myozyme) had almost completely stopped having an effect on me and I was starting to struggle with increased pain and reduced mobility. In July 2023, I started a combination of new ERT (Nexviadyme) and the Queen Elizabeth Hospital Birminghams, virtual Pompe exercise class.

In the 6 months that followed, I felt myself getting stronger and being able to do things that my body has never allowed me to do (even before diagnosis) such as being able to tense my stomach muscles and feeling my core strengthen. In the last couple of months, I have been able to walk up the stairs while carrying items and even walk up the stairs completely unassisted. Considering how difficult and almost impossible this seemed to me four years ago, the knowledge that I have been able to strengthen my body after my Pompe diagnosis has been unbelievable.

The exercise class itself usually consists of some strengthening and cardio exercises, followed by some core and balance work. If there is ever an exercise that we might find too challenging to do, the class leader (who is a physiotherapist at QEHB) will work with us and advise different ways we can complete it so we are still able to get something out of the exercise, with the hope that one day the exercise can be completed as it is supposed to be.

One of the biggest benefits I find from doing this exercise class weekly is the amount of energy I find myself with afterwards. To go from feeling very fatigued more than half of the week, to find myself with energy to complete additional physical tasks has honestly made such a big difference on my life as I am able to get involved in more social situations and regularly babysit for my nephew. If anyone has been in a similar situation where fatigue is a very big struggle for them, I would seriously consider joining a Pompe exercise class if offered the opportunity. We use chairs and other aids for exercises when required so people with a range of mobility can join in and more virtual classes are being planned that can be done from your own home.

You can find out more about the Pompe exercise class from the following poster.

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Building healthier lives

NHS
University Hospitals Birmingham
NHS Foundation Trust

Evaluating the feasibility and acceptability of a virtual exercise group for patients with Late onset Pompe disease.

Nicola Condon, Rachel Curran, Trudy Pelton, Jade Desai-Proctor and Tarekegn Hiwot

Department of Inherited Metabolic Disorders, University Hospital Birmingham NHS Foundation Trust

Introduction and Aims

Late onset Pompe Disease (LOPD) is a rare, neuromuscular disorder, that causes progressive skeletal muscle weakness and respiratory muscle dysfunction (1).

Supported exercise training of low workload and sub-maximal aerobic training, has been shown as safe and effective in managing symptoms, restoring function and improving quality of life in patients with LOPD (2,3,4,5).

Patients attending the tertiary Inherited Metabolic centre at Birmingham reported difficulty maintaining exercise programmes due to lack of access to local physiotherapy services, reduced confidence and worry of causing harm.

Virtually delivered group exercise can promote social support and connection and be a motivator for active physical lifestyle (6). Group session delivered in this format could reduce known barriers such as distance and accessibility and enable peer support.

To evaluate feasibility and acceptability of delivering a virtual exercise group for patients with LOPD, delivered by neuromuscular physiotherapists from a specialist inherited metabolic disease centre.

Method

An evidence-based approach was used to develop online group exercise sessions, to include exercises that address common difficulties for people with LOPD.

Patients deemed suitable to attend the virtual exercise group were identified and invited to attend during their clinic appointment.

A virtual consultation was conducted by Physiotherapists prior to starting the class. An individual safety and risk assessment was completed, expectations and goals discussed and opportunity to trial class prior to joining.

Patients were advised to attend 6 weekly sessions with the option to continue further six weekly blocks.

The exercise sessions included aerobic, resistance, balance and flexibility training. Two 1-hour exercise groups were offered – seated or standing to accommodate for all levels of ability. No specialised exercise equipment was required.

Outcome measures were completed before and after six weekly blocks. These included: Fatigue severity score, and if able to complete - Timed 5 sit to stands, Timed up and go. Feedback from patients was received after a 6-week block of sessions

Results

A total of 27 LOPD patients were invited to attend the virtual exercise sessions. Figure 1 shows recruitment to the sessions between July 2023 and Augst 2024 and patient feedback for leaving group.

5 patients (4 female), with a mean age of 48years have attended for 6 sessions or more. Outcomes were recorded after each 6-week block of exercise and the graphs show a trend in improvement in outcome measures (Figures 2,3,4). Feedback was gathered from all patients (Figure 5).

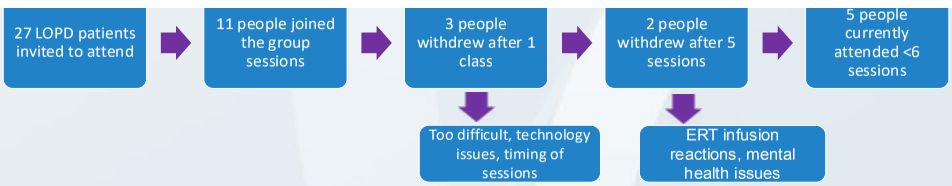
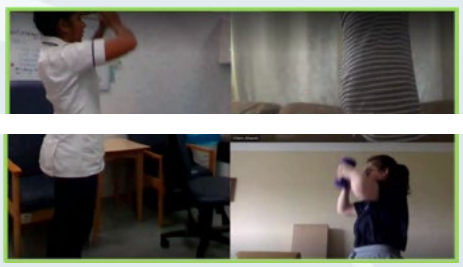


Figure 1. Patient enrollment in virtual exercise group



Picture 1. Standing exercise group

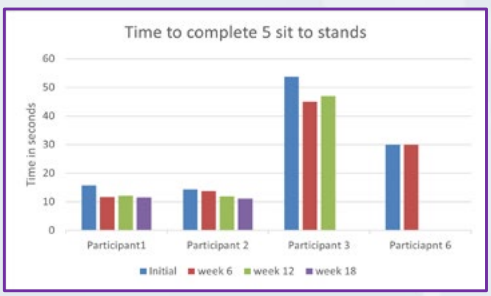


Figure 2. Time to complete 5 sit to stands

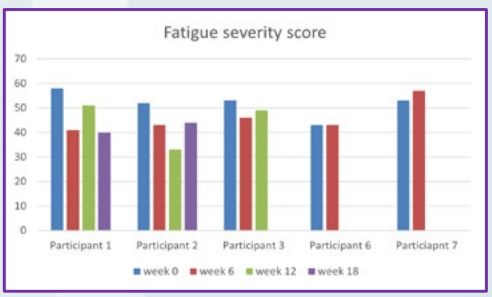


Figure 3. Fatigue severity scores

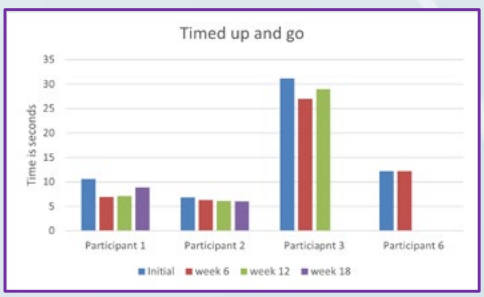


Figure 4. Timed up and Go

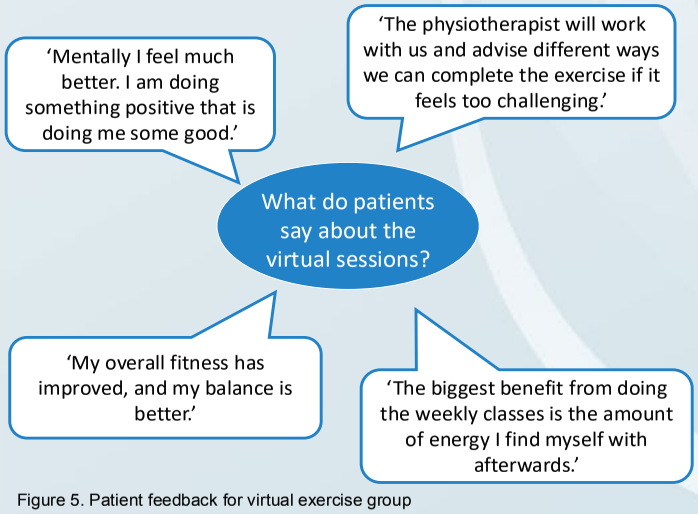


Figure 5. Patient feedback for virtual exercise group

Conclusion

Virtual group exercise sessions for people with LOPD in their own homes is feasible, well tolerated and can overcome barriers to exercise.

Live, physio-led sessions enables the physiotherapist to respond to individuals' need and adapt exercise in real time to different levels.

Improvements in fatigue scores and physical outcome measures were observed and patients reported improvements in functional status. The outcome measures will be evaluated in future programmes.

Although there is small sample size, this project demonstrates that virtual exercise sessions for patients with a rare neuromuscular diseases such as LOPD can be beneficial and safe.

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Budgeting for winter

Winter fuel payment is a one-off payment for helping with heating costs. The eligibility rules have changed and now you will only get this payment if you are of state pension age and claiming benefits such as pension credit.

You can normally get pension credit if you have reached state pension age, have limited income or savings and live in the UK. To get this year's winter fuel allowance you should apply by 21st December.

There are two parts to pension credit:

- Guarantee credit: You can claim guarantee credit if your weekly income is less than £218.15 if you're single or £332.95 if you're a couple. You may be able to claim guarantee credit if you're a carer, have a severe disability, have certain housing costs for example service charges or you're responsible for a child or young person who usually lives with you.
- Savings credit: this can only be claimed if you or your partner reached state pension age by the 6th of April 2016. This is now only available to existing recipients; the savings credit is for people who have modest savings.

When to apply for pension credit:

You can apply up to four months before you reach state pension age or anytime after you reach the pension age, however your application can only be backdated for three months.

To claim pension credit, you can either claim online at gov.uk or you can call the pension credit claim line on 0800 99 1234. They can fill in the application for you over the phone. Lines are open Monday to Friday 8:00 AM to 6:00 PM.

It will be helpful to have the following details to hand before you get started on your call:

- Your National Insurance number
- Your bank account details
- Information about your income, savings and investments
- Information about your pension, if you have one
- Breakdown of any housing costs, such as mortgage interest payments or service charges
- Your partner's details if you have a partner.

Pension credit can help with health costs. You can get free NHS dental treatment, help with the cost of glasses and transport to hospital. If you care for someone you might get an extra amount known as a carer's addition, which is worth up **£45.60** a week. If you have a disability, you may get an extra amount known as severe disability addition worth up to **£81.50** a week.

Useful links and advice can be found at **https://www.gov.uk/** or **www.ageuk.org.uk**

Can I claim pension credit?

To qualify for pension credit your weekly income must be below **£218.15** if you're single, and **£332.95** if you're a couple.

You or your partner must have reached the state pension age which is currently 66 for men and women in the UK.

You must live in England, Scotland or Wales. You can also claim pension credits if you have British citizenship and can prove you have resident pre-settled status from the EU settlement scheme or have indefinite leave to remain.

There is no savings limit for pension credit but if you have *more than £10,000 it affects the amount you receive.*

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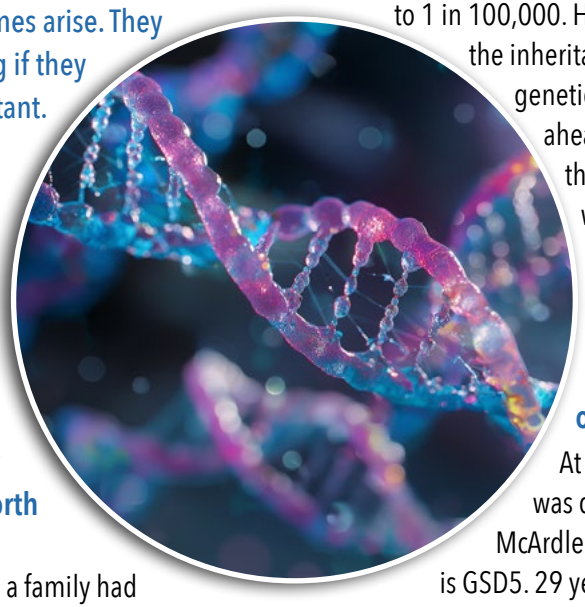
Stories of Diagnosis Being Corrected in McArdle's

It's surprising that on something as important as a diagnosis, very simple mistakes sometimes arise. They could be amusing if they weren't so important. McArdle's coordinator, Andrew Wakelin, describes some instances of mistakes in diagnoses he has helped to correct.



Too rare to be worth testing!

One young man in a family had been genetically confirmed with McArdle's. His sister had very similar symptoms and thought it would be very straightforward to get her diagnosis as well.

However, her doctor refused to undertake genetic testing. He claimed that with an incidence of about 1 in 100,000, the odds of a sibling being affected were too statistically remote to be worth considering.

The doctor clearly did not understand autosomal recessive inheritance. Once the parents in a family have been identified as carriers of causative mutations in the PYGM gene, then the risk of any child inheriting McArdle's is 1 in 4 - very different to 1 in 100,000. Having explained the inheritance pattern, the genetic testing went ahead. Sure enough, the young woman was confirmed as having McArdle's.

Man diagnosed with GSD5 turns out to have GSD6

At age eight, a man was diagnosed with McArdle's disease, which is GSD5. 29 years later, in discussing his symptoms in the McArdle's Facebook group, I thought his symptoms did not align well with McArdle's.

The man said that he had not had a muscle biopsy and the diagnosis was before genetic testing was available. I suggested he had a gene panel test for all the GSDs and also fatty acid oxidation disorders. It turned out that for all those years he had been misdiagnosed. He actually had Hers disease, which is GSD6. It has both muscle and liver involvement, and it was a great relief to the man after 29 years to finally have a better understanding of his condition and how to manage it.

The reason for the misdiagnosis is lost in the mists of time. Certainly there had been inadequate testing to support a diagnosis of McArdle's. But was that the full picture, or had somebody simply transposed a 6 with a 5?

Given diagnosis when only a carrier

A woman on the Facebook group appeared to be rather different to the typical McArdle disease person. Her symptoms did not fit well with those for McArdle's. However, she had genetic testing 10 years before and this was reported to her as confirming the diagnosis. I asked her to send me a copy of her report. They had indeed found two mutations in the PYGM gene. The first one was described as causing McArdle disease, and I was able to look up the particular mutation and confirm it. However, the second mutation was described as benign and I confirmed that it was also reported elsewhere as benign. So, although the first mutation was described as causing McArdle's, this would apply only if the second mutation was also capable of causing McArdle's. She was only a carrier rather than actually having the condition.

This misdiagnosis appears to again arise through a lack of understanding of genetic inheritance. Or maybe simple negligence in not completely reading the genetic report.

Misdiagnosed by Dr Brian McArdle!

A man was one of the very earliest cases diagnosed by Dr Brian McArdle himself, as was his sister, in the 1960s. Much later he discovered AGSD-UK and we talked. My comments about how different his symptoms were rather surprised him. The main difference was that his pain would come on after activity rather than during it. A few years later, he decided to contact the McArdle's clinic and they ordered a DNA test. We chatted as he waited in clinic, and I became even more convinced that the diagnosis was incorrect. It turned out that instead of McArdle's they had a condition called Periodic Paralysis.

The lesson is that if your symptoms seem very much at odds with what McArdle's people report, then it's well worth speaking to somebody who can assist you in getting your diagnosis

Mistakes of the past can often be corrected with the help of genetic testing.

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AGSD-UK can help you

When supporting people applying for welfare benefits, particularly disability living allowance (DLA) and personal independence payment (PIP), a recurring challenge is that many applicants are initially declined, later seeking support from AGSD-UK. A common issue is that people often haven't provided enough detailed information on their application forms. Typically, applicants will say something like, "I have this condition, and I have medical evidence to prove it," and then express confusion about why their claim was denied.

What many fail to realise is that the Department for Work and Pensions doesn't dispute the existence of the condition itself. Rather, what's crucial to the application process is explaining in detail how the condition impacts your daily life and what specific help or support you require from another person. It's not enough to simply state the diagnosis – you must clearly describe the challenges you face in everyday tasks, such as dressing, eating, mobility, or personal care, and how your condition makes it difficult for you to live independently.

For DLA claims, especially for children, it's important to highlight the extra care needed due to the child's condition. It's not just about the typical care a parent would provide but the additional, time-consuming, and often specialised support required because of the child's disability.

For instance, instead of focusing on what you normally do for a child, you need to explain how your responsibilities go beyond the norm-such as needing to monitor their health condition every few hours, helping with mobility aids, or managing medication routines.

Going through this process can be emotionally difficult for both the applicants and caregivers. The application forces people to stop and deeply reflect on the level of help they need or provide, which can be overwhelming. It's not uncommon for this to be the first time that someone fully acknowledges just how much support is necessary, and this realisation can be a difficult emotional journey. It also helps people come to terms with the reality of their condition, or the condition of the person they are caring for, as they are required to face the day-to-day challenges head-on and put them into words. This process, though tough, can also provide clarity and validation, helping both applicants and caregivers to understand the true impact of the condition and the extent of the support needed.

If you feel you need some help and support with PIP and/or DLA please get in contact with our dedicated benefits advisor, Zainib Hussain - **zainibhussain@agsd.org.uk**

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

Embracing Purpose: My Journey Volunteering with AGSD-UK

Maryam Ahmed

In a world that often feels chaotic and overwhelming, finding purpose can be a beacon of light guiding us through the darkness. For me, that purpose manifested through volunteering with AGSD-UK, an organisation dedicated to supporting individuals and families affected by glycogen storage disease.

My journey with GSD began with a diagnosis of type 1a, a condition that presented me with unique challenges and uncertainties. In the midst of my own struggles, I found great support through the resources and community provided by AGSD-UK. Their unwavering commitment to advocacy, education, and empowerment became a lifeline during my darkest days.

Inspired by the kindness and generosity I received, I felt compelled to pay it forward by volunteering with AGSD-UK. I wanted to be a source of hope and encouragement for others navigating similar journeys, just as others had been for me.

Volunteering with AGSD-UK can encompass a diverse range of tasks, each aimed at support, awareness, and progress. From organising fundraising events to getting people involved, every task plays a vital role in furthering the organisation's mission. Whether it's drafting articles for Glisten magazine or engaging with members through social media platforms, the tasks are as varied as they are rewarding.

One of the most appealing aspects of volunteering with AGSD-UK is the flexibility it offers. While the amount of time required varies depending on the different activities and personal availability, even dedicating a few hours a month can make a significant difference. It's about finding a balance that aligns with other commitments while still making a meaningful impact. The rewards of volunteering with AGSD-UK are amazing. It's the profound sense of fulfilment that comes from knowing you're contributing to a cause greater than yourself.

For anyone considering volunteering with AGSD-UK or any organisation for that matter, my advice is simple: take the leap. Embrace the opportunity to make a difference, no matter how small it may seem. Approach volunteering with an open heart and a willingness to learn, and you'll be amazed at the profound impact it can have, both on others and on yourself.

Getting involved with AGSD-UK

Community members make an incredibly valuable contribution to many different aspects of AGSD-UK's work, from being part of our Glisten editorial board to organising fundraising activities or helping with our governance.

Our volunteers undertake any necessary on-line training, and follow our policies and values, protecting our reputation and promoting our support and services. For some roles DBS checks may be needed.

In return volunteers can expect ongoing support, warm appreciation and a sense of fulfilment.

For more information about volunteering and ways to get involved, contact **info@agsd.org.uk**.

"AGSD-UK is a wonderful charity that has helped me in so many different aspects of my life. This is why I always want to give back anyway I can, whether it's with the charity themselves or other members. The most common thing I do with AGSD-UK is volunteer for their community events where I often give support, whether that's getting the venue ready, preparing talks or helping out at stalls. It doesn't take much of my time to volunteer. It's so simple and easy to do and everyone is so friendly and helpful!" Ciara Harkins

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20 TIPS FOR SKIING WITH McARDLE'S

IamGSD is working on some guidance for people with McArdle's (and maybe other muscle GSDs) wanting to go skiing. These are still in development, and it would be great to have feedback from anyone who already skis.

The following tips are mainly common sense, but they might not occur to you at the right time – so it is worth having a read.

1) **Tell your skiing friends** about your "different muscles" so they know to look out for you. Maybe try to do it on a reciprocal basis - ask if there is anything you should be aware of to support them. If you have the *101 Tips* book the energy/ATP reservoir diagram on page 54 is a very quick way to give a simple explanation that there is a problem with fuelling the muscles.

2) **Do not ski alone** in case you hurt yourself off-piste, or in an area with no cell phone coverage. Even walking in heavy ski boots is very hard, especially in fresh snow.

3) **Check your insurance cover** to make sure that it will cover you for rescue from the slopes, should you incur a contracture up there.

4) Wrap up warm! Cold muscles work more anaerobically, which we can't do. Consider hiring or buying heated ski clothing. Even getting dressed and putting on the skis can be quite an effort, so consider getting into second-wind before that.

5) **Get some help**. Can you get dropped off close to the lift, instead of having to walk to it? Can a friend help you carry the heavy equipment?

6) **Get into second-wind** in your legs, and preferably arms as well, before starting. And do not leave more than 15 minutes between being in second-wind and skiing, due to the risk of losing it again. That may mean getting into second-wind down in the resort and then topping it up immediately after getting off the top of the ski lift.

7) **Choose your lift** to avoid a drag lift that requires you to hold on with your arms and grip with your hands, even if it is very short. We need to use the ones with a seat, or the full cable car type.

8) **Choose your runs with care**. Some parts of the world use different colours, so be very careful to choose ones that suit you. Stay with the easiest runs until you are confident. If you accidentally find yourself on a run which is too hard for you, ask a guide for assistance.

9) **Consider the ketogenic diet.** Several people have reported skiing much more easily on it, but it is still developmental and might be difficult to adhere to in resort.

10) **Tell the instructor about your McArdle's** before the lesson. If they are not receptive, choose another group/instructor, or consider taking a private lesson. Things like walking sideways up hill is very intense for us, we have to do it very slowly, with rests.

11) **Don't push on in pain.** When on a run with a novice group, if you start to get pain when trying to keep up with the rest of the group you must stop. If the guide does not stop the group, it is probably best if you allow yourself to get left behind (with your friend).

12) **Downhill skiing** is probably the best option. Cross country skiing is very demanding of muscle effort, although the muscles are at least continuously moving. If you try it, take care to start on a trail which is short and mainly level. In downhill skiing the holding of a tensed position is the big issue.

13) **Keep the muscles moving** a bit every few seconds, don't just hold a fixed position. Maybe change the balance from side to side so alternate legs are taking the strain. Static (isometric) effort is the worst for us as it restricts the blood flow and we are very reliant on that to deliver energy to the muscles. If pain starts, stop for a rest.

14) **Consider hydration and food**. Take some water with you, maybe in a CamelBak so you can top up on the move. Carry an emergency snack which suits your diet plan, in case you need something and can't get what you want at the lift-top café.

15) **Ask a friend to stay with you** on the run, so that if you fall or have to stop there is someone who will see that, stop with you and help if needed.

16) **Plan for a break half way.** It might be good to arrange with friends that they will take a break about half way down a long run, just for a few minutes. But always listen to your muscles and don't be embarrassed to stop for a rest whenever you need, even if it is just before the end of the run.

17) **Take care getting up from a fall.** Do NOT try to get up whilst still in the skis. Lie there to rest for 30 seconds plus (count them, as it seems like eternity and there is a real danger of having a cramp if you get up too soon). Take off at least one ski, preferably two, then stand up and put them back on. This seems like a nuisance but the muscle strain of getting up, especially if you slip and have to make a 2nd or 3rd attempt, is likely to cause a contracture.

18) **Rest at the bottom of the run** for at least 30 seconds to recharge the ATP before moving off. And don't struggle to keep up with your friends without saying anything! Maybe opt out of the next run and catch up with them on the following one.

19) **Fixed contractures** – are you aware of what to do and when to seek medical attention? Just in case, you should carry one of the emergency cards in your pocket to remind you, and to show at hospital if you ever do need to seek medical attention. There is emergency guidance on the IamGSD website under the Medical menu.

20) **Enjoy!** The outdoors, the mountains, the views, the challenge, the excitement and the exercise!

If you have any feedback on these draft guidance notes, or comments about snowboarding with McArdle's, please email Andrew Wakelin on: **type5@agsd.org.uk**

WHY YOU SHOULD TAKE PART IN RESEARCH

If you are asked by your health practitioner to take part in research or a clinical trial, should you take part? The answer is simple. If you can, YES.

Do not think that you are too old, too ill or not important. The information that you will give provides the foundations of any developments in the future. Your experience is invaluable. Research is not just about blood results, enzymes, white coats and test tubes. Your experience of the condition is essential. The knowledge that comes from you is fundamental and based in the real world. Your input will give a true representation of the challenges and problems surrounding the disease. Children being born today will be hoping that by the time they reach adulthood there will be a cure. You can help them.
All medical knowledge is built up over the years and your participation will help future generations. This won't happen without the building blocks of knowledge that are being looked at today.

Who does the research and trials?

- Health practitioners who need the knowledge to improve the care they give.
- Researchers who want the knowledge to ensure the best treatment is given and to look for breakthroughs.
- Pharmaceutical companies who want the knowledge to know where to invest and take on the research.

The benefits of taking part in research and trials:

- Improved care and new treatments, now and for the future.
- A better understanding of the natural history and progression of the condition.
- Better knowledge of the basics of the disease can lead to planning for the future with timely care interventions, meaning fewer complications.
- Earlier and correct diagnosis.
- Any disease that has good basic, up to date data, with will be an attractive prospect for further researchers and investment from pharmaceutical companies.
- Cures and treatments can be accelerated.
- Information gathered where many patients have taken part can lead to new insights and new approaches to care and treatments.
- You may have an opportunity to learn more about your condition.
- Personal satisfaction from knowing that you are helping the community.

You have been asked to take part and are unsure?

- Do talk to the researchers. If you are not sure if the research is right for you, ask questions.
- Check if the research has any implications for your treatment decisions now or in the future and make sure you understand these.
- Ask about travel expenses and costs of overnight stays. They are often reimbursed.
- How long will the research take? Will there be a follow up? Will you need to go back?
- And if the request to take part in research comes from someone else other than your health practitioner, please do check with them first.
- And don't forget that AGSD-UK is here to help you with information.

Your participation can give hope. Be one of the building blocks.

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Catching up on conferences

AMDA/IPA International Conference

Kempton Rees

The Acid Maltase Deficiency Association (AMDA) was established in 1995 with the aims of funding research, raising public awareness, and advocating for the Pompe disease community. It has held joint patient and scientific conferences with the International Pompe Association every three years or so. I attended the latest conference in San Antonio, Texas in May this year.

The previous conference had been held in October 2019 – Covid having put paid to any such gatherings in the interim - so there was a lot to catch up on.

San Antonio is, by American standards, a quaint and quirky city, though its near neighbour, Austin, lays claim to being the quirkier with its proud slogan "Keep Austin Weird". San Antonio is best known as being the home of the Alamo, the missionary where Davy Crockett and his colleagues were overrun by Mexican troops in March 1836, a defeat which has become legendary.

There was little time for sight-seeing, however. The day before the conference started, I participated in community advisory board (CAB) meetings with two drug companies. CABs are a collective of patients and carers who meet with organisations involved in clinical research to help researchers better understand the needs of patients; what they want and expect from new therapies; and what they are willing to do to participate in research trials. They are subject to *Chatham House Rules*, which means that all participants can speak freely and use the information learned, but must otherwise treat everything that is said in meetings as confidential. So I cannot say more that that!

Luckily, I had remembered the lesson I'd learned from 2019. I wore multiple layers of clothing to shield me from the ice-box air-conditioning of the conference hotel (the temperature outside being in the 80s). Why do they think that's still necessary in this day and age?

The first day of the conference proper was taken up with workshops on mental health; the diagnosis of Late Onset Pompe Disease (LOPD) through new-born screening (which is progressively happening across individual states in the US); transitioning from paediatric to adult care; and exercise recommendations for LOPD.

This last workshop included a lively demonstration of various exercises that one can do, sitting and standing, by ***Keyuna "Coach K" Milam***. I'd strongly recommend checking out her YouTube video for Pompe patients which you can easily find by Googling her name and the word 'LOPD'.

That evening we were treated to a fantastic traditional Texan barbeque, rodeo show and line dance experience (which I chose to sit out!).

The second day was a full-on programme from 8am to 5:15pm with talks across an array of subjects including the effectiveness of different enzyme replacement therapies; new-born screening programmes; different techniques for the monitoring of patients (such as biomarkers, MRI and ultrasound); respiratory management; nutrition; exercise; speech impairment; and pain management. Key speakers presented research being carried out in numerous countries. They included our own ***Dr Mark Roberts***, who will be known to many of you.

The day ended with round-table discussions involving panels of clinicians, researchers and patient representatives.

The last day had a similarly early start. It began with a series of highly interesting (albeit often highly technical) short presentations from a number of people from universities and drug companies researching new therapies including gene therapies.

There followed reports on the IPA/Erasmus survey and Pompe registries before the conference closed with a look at exciting research into future potential therapies such as substrate inhibition (i.e. ways to limit glycogen production in muscle cells, rather than ways to remove it); gene-editing (not to be confused with gene therapy); and a talk on a new technique for producing mini muscle cells in the lab (given by the delightfully named ***Dr Pim Pijnappel*** from the Netherlands). We all left tired, but positive and looking forward to the next conference in a few years' time.

Presentations from the ***AMDA/IPA International Pompe Patient and Scientific Conference*** are available to view online at:

https://shorturl.at/B2h2A

APBD Scientific & Community Conference

In September the virtual ***APBD Scientific & Community Conference*** took place, bringing together more than 200 APBD/GSDIV community members from 15 counties.

Recordings of the sessions are available here:
https://shorturl.at/chBtU

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Pompe stories

I come from a South Asian background and was born with a rare health condition called Pompe disease. I received my diagnosis at the age of 14, after numerous hospital visits throughout my early years. I had a sense that something was amiss within me, so when the official diagnosis came, it wasn't a surprise, given that there is still no cure for this condition, and managing it is key.

In our culture, it's customary for parents to start looking for potential marriage partners for their children around the age of 18. However, my disability led them to believe that finding a spouse for me would be a formidable challenge. They thought that nobody would want to marry me because I might not be able to fulfil the traditional roles of a housewife or have children.

Interestingly, I had more freedom during my upbringing compared to my cousins, as my parents allowed me to drive, pursue my studies, and go out, while many of my cousins had stricter cultural constraints. Nevertheless, my disability was often seen as a reason for me to stay at home with my parents and never marry.

At the time, I was content with this arrangement, as I had no desire to marry one of my cousins, which appeared to be the only option available to me. However, as I grew older, my perspective shifted, and I longed to meet someone and build a life with them. I wasn't actively seeking a partner, but fate had other plans for me. I met someone through gaming, and we've now been together for over four years. I am genuinely happy.

I firmly believe that everyone deserves the opportunity to choose a life partner, regardless of their disability. Love knows no boundaries, and I am living proof that happiness and companionship are attainable for everyone, regardless of the challenges life may present.

I was diagnosed with Pompe disease in October 2016 after my younger brother passed away of it. I still remember the day clearly, my dad sitting in the genetics counsellors office and me and my siblings getting called in one by one to be told if we had the condition.

When me and my younger sister were told we had the disease we were obviously upset but I remember my dad being more upset and trying to hold back his tears. I just remember telling this counsellor I am getting married in two weeks and what happens now. I was told to tell my partner and that my details were being passed on to a consultant. But that was it. No support was offered not even from the counsellor. Thankfully my partner was very understanding and supportive and we tied the knot.

Now eight years and two kids later, thankfully we are going strong and adjusting together with my treatments and family dynamics. During this time, I have lost my two other brothers due to lung and heart problems unrelated to Pompe but the question of Pompe was still raised by the coroner due to its rarity and genetic testing. My older brother who was 33 when he passed away last year was featured in a Channel 5 documentary called *Cause of Death*. The coroner did bring up the family history of Pompe disease and questioned the accuracy of the genetic testing for Pompe as it is such a rare disease. I think this was done to raise the awareness of Pompe. You can watch the documentary on the Channel 5 app by logging in with an email address or through the link below.

https://shorturl.at/LgYI1

I would like to say that more awareness should be raised about organisations like AGSD-UK, especially by doctors, nurses and other healthcare staff. I felt as if I was not given any support when I was first diagnosed and felt completely alone and had no one to talk to. If I had known that AGSD-UK existed, I would have come to them in the first place.

Support organisations like AGSD-UK should be mentioned so people don't have to feel like they have no one to turn at their time of need.

I am grateful for AGSD-UK, Zainib and the lovely messages of support she has sent me. It makes me feel very supported and pushes me to live life to the fullest even with a rare disease like Pompe.

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Gene therapy research updates in GSDIa

Ultragenyx DTX401 in GSDIa

Ultragenyx have announced positive top line results from the ongoing ***Phase 3 GlucoGene*** study. The randomized, double-blind, placebo-controlled study has dosed 46 participants aged eight years and older with DTX401 or placebo.

Summary of the top-line announcement includes:

- The primary endpoint of the study was achieved, showing that treatment with DTX401 resulted in a statistically significant and clinically meaningful reduction in daily cornstarch intake compared with placebo at Week 48.
- All of the participants treated with DTX401 have been able to reduce their daily intake of cornstarch while maintaining their glucose control, and at Week 48 the mean percent reduction was 41.4% in the DTX401 group compared with 10.1% in the placebo group.
- The safety profile was acceptable and consistent with Phase 1/2 study results. Anticipated effects on the liver from AAV gene therapy were all non-serious and manageable with a prophylactic corticosteroid regimen.
- As with all experimental drugs, it is important to remember that at this time, the safety and efficacy of DTX401 have not yet been established.

What happens next?

- Continued participation by all patients enrolled in this study remains critical to its success. As indicated by the ***Phase 1/2*** study, reduction in cornstarch intake can continue over time and long term data will be essential as the company works to gain regulatory approval.
- The safety profile of DTX401, including AAV8 class effects and risks, will continue to be monitored in the study and through long-term follow-up.

Ultragenyx are preparing to hold discussions with global regulatory authorities to support a marketing application in 2025. Once a marketing application is submitted, regulatory agencies carefully review it to determine whether an experimental drug is considered of good quality, safe and effective, has a positive benefit/risk profile, and can be approved for use outside of a clinical trial.

Ultragenyx thanked the GSDIa community for championing this gene therapy program with the following message:

"From your early support to start the research years ago, to the patients and families participating in the DTX401 studies and the clinicians and researchers contributing to this research, we are humbled to work alongside you to rapidly move the development of DTX401 forward.

While we still have a lot of work ahead of us, we recognize that this is an important milestone for the community. We remain committed to sharing timely information as the program progresses."

BEAM-301 in GSDIa

Earlier this year, ***Beam Therapeutics*** put out a press release that reported on clearance by the ***US Food and Drug Administration*** (FDA) for their investigational new drug application for BEAM-301.

Now that the FDA has cleared their application, Beam is initiating activities to open enrolment in the United States for its ***Phase 1/2*** clinical trial investigating BEAM-301 in individuals who have GSDIa and at least one copy of the R83C mutation.

They are working to open their ***Phase 1/2*** trial in multiple US sites and dosing is expected to begin in early 2025.

Although the ***Phase 1/ 2*** trial will be US only, they hope the programme is successful, so that they may be able to expand outside the US for future studies.

You can read the press release below and a further update is expected in November.

https://shorturl.at/OG8ux

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The main event

9th - 10th November 2024
Burleigh Court, Loughborough

Whatever your GSD, join us for:

- Updates on latest research & treatment options
- Tips for living well
- New therapeutic activities to try
- Time to share experiences & make new friends
- Chance to hear from leading experts
- Fun for children and families
- For more information contact **info@agsd.org.uk**

PROGRAMME

SATURDAY 9TH NOVEMBER

AGSD UK Association for Glycogen Storage Disease

12.15 -1.00pm Arrival, event registration and informal welcome
1.00 - 1.45pm Lunch | FIFTY THE STREET
1.30pm Créche opens | MEETING SPACE 8
2.00pm Opening remarks and introductions | MEETING SPACE 14 & 16
2.05 - 2.50pm Our GSD stories | *Taylor Adam, Nikol Rainbank, Anosha & Yasmin Khan* | MEETING SPACE 14 & 16
3.00 - 3.45pm **Sessions with clinical experts:**

MEETING SPACE 14 & 16

Pompe
Chong-Yew Tan & Liz Morris
Metabolic consultant physician & Clinical nurse specialist Lysosomal Disorders Unit Addenbrooke's Hospital

MEETING SPACE 18

McArdle & other muscle
Maria Patasin
Clinical nurse specialist Queens Square Centre for Neuromuscular Disease

MEETING SPACE 17

Hepatic
Rebecca Halligan
Consultant in children's inherited metabolic diseases Evelina London Children's Hospital

3.45 - 4.00pm Tea break | BUSINESS LOUNGE
4.00 - 5.30pm **Optimising health with GSD:**

Pompe
Exercise, nutrition & nutritional supplements
Mark Tarnopolsky (Virtual)
Division head of neuromuscular and neurometabolic disorders, McMaster University Medical Center,

Exercise group experience
Nicola Condon
Clinical specialist physiotherapist Queen Elizabeth Hospital Birmingham

McArdle & other muscle
Physical training & gym experiences
Peter Finnigan
Specialist neuromuscular physiotherapist Queens Square Centre for Neuromuscular Disease

Bronte Thomas, Student support advisor and volunteer walk leader

Why exercise is the best treatment
Sam Torrens
School of Exercise and Nutrition Sciences Queensland University of Technology

Hepatic
Eating, physical activity & mental health
Research team
National Centre for Sport & Exercise Medicine
Loughborough University

Acceptability & efficacy of extended-release cornstarch
David Weinstein
Weinstein Rare Disease & Clinical Developmental Consulting

6.20pm Entertainment and drinks reception | *with the Extraordinary Mr Dudi* | CARNEY & SCOTT'S
7.20pm Dinner

SUNDAY 10TH NOVEMBER

THE MAIN EVENT

9.30am Music warm up session | *Ed Freitas* Echo Fox Events | MEETING SPACE 14 & 16
10.15 - 11am **Research updates with leading experts:**

MEETING SPACE 14 & 16

Pompe
Derralynn Hughes
Clinical director of research and innovation, Royal Free London NHS Foundation Trust

MEETING SPACE 18

McArdle & other muscle
Sam Torrens
Researcher and sessional academic School of Exercise and Nutrition Sciences Queensland University of Technology

MEETING SPACE 17

Hepatic
David Weinstein
Weinstein Rare Disease and Clinical Developmental Consulting

11.00 - 11.30am Coffee | BUSINESS LOUNGE
11.30 - 12.15pm **Concurrent living well sessions I**

OPTION A **Living aids and equipment** | *Paul Withington* Occupational therapist specialising in housing and equipment | MEETING SPACE 14 & 16
OPTION B **Parenting** | *Elizabeth Davenport* AGSD-UK specialist care advisor, *Zainib Hussain*, AGSD-UK benefits and engagement advisor | MEETING SPACE 17
OPTION C **Creative relaxation** | *Rachel Thomas* Artist and AGSD-UK Trustee | MEETING SPACE 18

12.30 - 1.15pm **Concurrent living well sessions II**

OPTION D **Getting involved with AGSD-UK** | *Andrew Wakelin*, volunteer coordinator for McArdle and other muscle AGSD-UK, *Ciara Harkins* AGSD-UK volunteer | MEETING SPACE 18
OPTION E **Living well social** | *Zainib Hussain*, AGSD-UK benefits and engagement advisor *Elizabeth Davenport* AGSD-UK specialist care advisor | MEETING SPACE 14 & 16
OPTION F **Men's health discussion group** | *Dave Hogan* Men's health support group facilitator for people impacted by rare conditions | MEETING SPACE 17

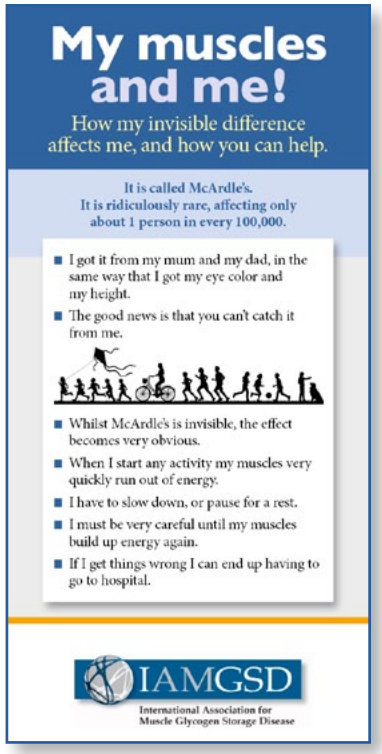
1.15pm Lunch and depart

Leaflet for McArdle children to share

IamGSD is launching a leaflet for young people to explain their invisible muscle problems to their friends, school mates, teammates, etc.

Aimed at 10 year olds and above, it avoids jargon and uses plain language. It should assist friends to understand what the young person deals with, and how they can help.

To comment on the draft or to pre-order printed copies, drop an email to Andrew Wakelin at: **type5@agsd.org.uk**



GSD1 & Hearing Loss

Researchers in Poland have noted that 1 in 10 people (from a sample of 40 patients) with GSD1a and GSD1b suffer some form of hearing loss. If you are concerned at all, do speak to your clinician.

Here is a link to the academic article

https://shorturl.at/5Cyns

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Patient Registries are there to save lives

Registries are not boring. Well, maybe just a little bit. But they are important. They exist to bring hope for the future and save lives.

What is a registry?

Registries are lists of data on patients, and this vital information brings insights to many people. Researchers, clinicians and pharmacologists to name but a few. Dedicated people use these lists to make patients' lives better and to find treatments.

This information will always be kept anonymous. It can be as simple as ages of people affected. Or, it could be more complex, covering someone's complete medical history and the changes that occur through time. The number of people can vary from just following a few patients within a clinician's practice, to the entire patient population within a country. Some registries are worldwide.

Registries exist to hold data that is specific to that condition, and they are used to improve diagnosis, increase knowledge, and promote care and treatment. They are essential in the world of rare diseases.

Who starts and holds registries?

Clinicians and clinical centres can hold registries. These generally only cover the country in which they started. EUROMAC is an exception in the GSD world. It is a Europe wide project, gathering information from clinical centres, for the main on people affected by McArdle's or type5.

CoRDS is an example of a non-profit research institution based at Sanford Research, USA and collects data on McArdle's. Patients can input the data themselves.

Pharmaceutical companies can hold registries, usually while conducting clinical trials on treatments in development. These are sometimes only used by that company.

Patient associations can start and hold registries. There are many advantages to these registries.

- They are independent and can aim to be free of commercial, academic or advocacy interests.
- They have an ability to provide a unique insight that can often be missed by clinicians and commercial companies.
- The international reach of rare disease patient associations, means that patient input will be of a high volume and diverse. Two qualities that are essential for planning, care and research.

Examples of registry successes

The INPDR (International Niemann Pick Disease Registry) has been involved in furthering research and the formation of current disease guidelines. They have ensured the patient voice is heard.

The Cystic Fibrosis Foundation Patient Registry has led to the development of therapies that have transformed cystic fibrosis from a deadly disease to a manageable condition.

EUROMAC (linked to clinical centres) has played a key role in training, education and the implementation of international clinical trials. The data has suggested that clinicians should pay attention to under-reported symptoms a fixed muscle weakness.

The International Pompe Registry (held by the pharmaceutical company, Sanofi) has helped to further understanding and promote breakthroughs in treatment.

The future for GSD

There are many GSD registries throughout the world, mainly held by clinical centres and pharmaceutical companies. They are usually small and ask different questions from the community. The different questions could be seen as an advantage as different aspects of a disease are observed. Sometimes this is a disadvantage as the data is disjointed or fractured. Accessibility is variable, depending on local policies and legislation.

The future could be one of international, independent collaboration where all data is accurate, well monitored, confidential and easily accessible. High quality, innovative and lifesaving research will be inspired and accelerated by the registry's involvement.

Policies will be transparent, promoting trust from the patient community and the patient experience of life with the disease will be an essential part of the data.

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Take aways from this edition

- Last chance to book your place at the Main Event here: **https://bit.ly/3U5TxFs**
- Can you help with Sioned's event? Contact **sionedwilliamsharp@mac.com**
- Interested in volunteering for AGSD-UK? Just contact **info@agsd.org.uk**
- Try some festive fundraising
- Look out for opportunities to take part in research or patient registries

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AGSD UK
Association for Glycogen Storage Disease

Roman Tank and his sister

Upcoming events

- The Main Event: 9-10 November, Loughborough
- Living well social: 19 December, online
- AGSD-UK AGM: 19 January, online
- McArdle's walking courses: August 2025, Snowdonia

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