Autumn 2022

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!

What's in a name?

Ever wondered why our magazine is called Glisten?

Do you love it as it is, or do you want to suggest a different name?

Would you prefer it to be changed to Glysten, to make clearer the connection with glycogen storage? Just email info@agsd.org.uk - and get in touch if you want to be involved in shaping the next edition by volunteering for our editorial board!

Thank you again to everyone who supports us.

Front Cover Images:
Theodore Molyneux with his CORI kitten!
Theodores' Auntie and Uncle, Jack Molyneux and Beth McGhee

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

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

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

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Message from the Chief Executive

Welcome to the autumn 2022 edition of Glisten.

Autumn is traditionally a time of transitions, but the sense of flux is starker this year, with new historical and political chapters in the UK. There are also new pressures to contend with, like the biting cost of living crisis, so this edition includes articles signposting important sources of support.

This a time of change for AGSD-UK too, as we move forward with new plans for 2022-25. These have been shaped by your ideas about what would have the most positive impact on life with the condition and they focus on three key themes:

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

You can find more details inside on how we aim to achieve the goals you've set and ways to get involved.

You'll also find more about the incredible efforts of fundraisers and other volunteers that are so crucial for delivering the progress the community wants to see.

While we cope with a changing world, face up to new challenges and chart new approaches to try to make the biggest possible impact, AGSD-UK's fundamental mission remains the same: to help people live positively with the condition.

We're determined to be a constant source of support for people affected and after all the disruption of Covid it's great to be able to welcome people back together, not only for virtual gatherings but also for valued opportunities to meet face to face and share experiences. Please look inside for information about different community get-togethers as well as how to register now for our full AGSD-UK event on 6th & 7th May 2023 in Loughborough.

We can't wait to see you there!

Val Buxton

Our plans 2022-25

AGSD UK
Association for Glycogen Storage Disease

Earlier in the year we asked you what would make the biggest difference to life with the condition. Your answers were grouped into three themes which we used as the framework for our 2022-25 plans. We set ourselves measurable goals under each of these themes as well as a detailed action plan, based on your responses.

Our first goal:

SUPPORT ACROSS THE COMMUNITY

Everyone affected will be connected to an active and supportive GSD community

Your responses strongly underlined the importance of support and connections across the community and we're working to achieve this by ensuring AGSD-UK is promoted in clinical settings, so everyone knows about the support we have to offer, running regular community events, growing and developing our team of volunteers and strengthening our social media presence.

Our second goal:

HELP WITH LIVING WELL

People with any GSD will be offered the information and activities they need to live well with the condition

Responses highlighted a range of areas where people wanted more information and opportunities to help them live well. We'll be tackling this by updating and broadening our offer of information and therapeutic activities, as well as developing and launching a self-management programme to help people live positively with the condition. We'll also be looking to roll out a new community support fund to help with access to therapy, respite, and equipment for those in greatest need.

Our third goal:

BETTER TREATMENTS AND SERVICE ACCESS

The community will experience improved management, treatment options and service access

Speeding access to diagnosis and better treatments and improving understanding among clinicians were the particular priorities people expressed here. We'll work towards this by strengthening evidence around the condition to influence policy, practice, and the roll out of new treatments and approaches, working with others to amplify our voice. We'll also be supporting professionals to collaborate and learn from each other, along with promoting research opportunities and understanding.

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Our underpinning goal:

Annual Report

STRONG AND EFFECTIVE ORGANISATION

AGSD-UK will grow sustainably, with robust governance, to increase our impact

In addition to our three main goals, we have a final underpinning goal about making sure we're a well-run and effective organisation, with the resources we need to deliver the support and services people want to see.

Having a clear strategy will help us generate income, monitor and demonstrate progress and focus all our activities and resources to make the biggest possible difference.

Involvement from across the community will be crucial in taking these plans forward. Please do get in touch with val.buxton@agsd.org.uk if you can spare just a little time to further shape these initiatives, lend a hand, for example with peer support or events, contribute fundraising ideas or activities or help steer the organisation as one of the board of trustees.

We're looking forward to working with you to achieve our shared goals for everyone affected by GSDs.

Planned activities to achieve our goals:

SUPPORT ACROSS THE COMMUNITY

- ✓ Take forward a systematic outreach programme to ensure all relevant clinical settings across the UK understand AGSD-UK's offer and signpost to us
- ✓ Develop a proactive engagement and support strategy for all those in touch with the charity
- ✓ Provide a training, support, recognition and succession programme for volunteers
- ✓ Deliver a programme of community events, on and off line, tailored to the needs of different groups, including children, teens, parents and others who are underserved
- ✓ Build sustained engagement through social media, scoping the development of a community support forum hosted on the AGSD-UK website

HELP WITH LIVING WELL

- ✓ Develop an on-line self- management programme, supporting people to live positively with any GSD
- ✓ Establish a community support fund, facilitating access to respite, therapy and equipment for those in immediate need
- ✓ Review and update all AGSD-UK information in line with best available evidence, working with people affected and clinical experts to address any gaps and ensure relevance, accessibility and inclusivity
- ✓ Expand our offer of appropriate therapeutic activities on and off-line

BETTER TREATMENTS AND SERVICE ACCESS

- ✓ Gather evidence on quality of life issues for people with all GSDs to underpin our influencing work and shape our information and support offer
- ✓ Bring together GSD professionals' networks to facilitate shared learning and quality improvement, coordinating meetings and educational activities and taking forward the development and auditing of consensus guidelines on priority topics
- ✓ Maximise the reach and impact of our advocacy services through close working with specialist teams and better integration with clinics on and offline, learning from service user feedback and providing relevant training
- ✓ Promote understanding of research and clinical trials along with opportunities to shape these and for participation among people affected
- ✓ Work with charity partners to influence relevant policy, focussing on opportunities to speed diagnosis and access to new treatments

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YOUR RIGHTS IN EDUCATION

We all want the best education experience for every student. Inclusive education means all students can access and fully participate in learning, supported by reasonable adjustments and teaching strategies tailored to meet their individual needs. It is against the law for a school or other education provider to treat disabled students unfavourably. An education provider has a duty to make 'reasonable adjustments' to make sure disabled students are not discriminated against. These changes could include providing extra support and aids (like specialist teachers or equipment). Schools must also make the buildings accessible for their disabled pupils as part of their overall planning duties.

Special Educational Needs and Disabilities (SEND)

All publicly funded pre-schools, nurseries, state schools and local authorities must try to identify and help assess children with special educational needs and disabilities (SEND). Your child may need an education, health and care (EHC) plan if they need more support than their school provides. Support needs could include 1:1 support in the classroom to scribe for a child or to make sure that the school is adapted, including a changing area if required. SEN support starts for children under five right up until or further education. Universities also support students so if you are considering higher education, please speak to your chosen university well in advance.

If you would like to discuss support then contact the SEN co-ordinator, or 'SENCO' in your child's school or nursery.

If your child is not in school, then contact your local authority. AGSD-UK is also available to support and work alongside your schools SEN co-ordinator. The Special Educational Needs and Disabilities Information Advice and Support Services (SENDIASS) is also a useful service. They offer information, advice and support for parents and carers of children and young people with special educational needs and disabilities (SEND). The service is free, impartial, and confidential. It is also a legal requirement that all local authorities have a SENDIASS service

For more information, please contact
Elizabeth Davenport, Specialist Care Advisor
AGSD UK
Email: Elizabeth.davenport@agsd.org.uk

FUNDRAISING ROUND-UP

Thank you to our members who have been busy fundraising over the past year. Want to fundraise on Facebook but not sure how? Contact info@agsd.org.uk and we can show you what to do!

13 Loch Challenge for AGSD-UK

£82.50 so far (Inc. Gift Aid)

Joe is Swimming Scotand's 13 largest lochs (12km-38km) for AGSD-UK and another charity.

I have been trying to put together a swimming challenge, ever since I completed my 70.3mile triathlon a few years ago for the Accord Hospice and the amazing care they gave my mum.

This time I wanted to add something else that is special to me. My closest friends in the world have two amazing kids, Orin and Olivia who have Pompe disease and as a family have shown me what it is to fight for what is important. After a chat with the kid's mum, Lyndsay, she recommended AGSD-UK as their charity of choice and told me the amazing things they do for others in the same situations.

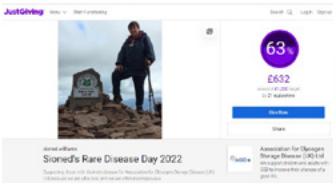
The original aim was to swim Loch Lomond last September, however in April, I found out I needed a new hip. All went well and I have been building up week by week and decided it was time to put plans back in to action.

After a lot of thought, we decided that Loch Lomond would be great to do and leading up to it we realised we could fit in a few other lochs as part of the training. This spiralled into finding out how many large lochs there are and that you can swim in them!! This led to the decision to swim the 13 largest lochs in Scotland which range from 12km to 38km which include Loch's Shin, Arkaig, Sheil, Katrina, Rannoch, Lochy, Ericht, Maree, Awe & Tay, Morar, Lomond and the famous Loch Ness!!

The challenges started early May at Loch Katrina (12km) and builds up from there. You can support Joe's challenge on his Just Giving Page https://tinyurl.com/445fsun5

Sioned's Rare Disease Day 2022

£727.92 (Inc. Gift Aid)



You may have seen in the last edition of this magazine, how Sioned Williams made an impassioned funding appeal. In her video she explains a lot about McArdle's, the difficulty of getting diagnosed and what support is available these days, but she also gives a fascinating insight into her high-profile career as an international musician, for 28 years as Principal Harpist with the BBC Symphony Orchestra. Sioned's video was very successful and will have gone a long way to help others, not only through the money raised but also the informative content she put together. Thank you Sioned for your continued support. You can watch the video here: https://youtu.be/JfSXU2snaDc

Penns School Fundraising £619.58 (Inc. Gift Aid)

Thank you to Nikki Christie and *Penns School* for continuing to fundraise for AGSD-UK

Thank you also to:

Yasmin Khan who donated **£240**

Brittany Brown who donated **£50**

Sally Robinson who donated **£2000** on behalf of Simon Gurney

Thank you to Louise Bett's family who donated the money raised from her funeral **£125**

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

The Great North Run

Jack Molyneux and Beth McGhee

Ever since we have been aware of Theodore aka Ted being diagnosed with GSD, Beth and I have been brainstorming ideas of how to help. These have varied from organising events to fundraising to raising awareness. Initially, a fundraiser seemed the best option and Beth, being very active and sporty, put herself forward for running the Great North Run.

As for me, I don't mix well with exercise. Golf is about as close as it gets to me doing exercise and I'm not even good at that. After much deliberation, I eventually decided to take the plunge and sign up for the ballot as well. A few weeks later, we discovered that we were successful, and the training began while we were living in Sydney.

From the outset Beth set herself a target of one hour 45 minutes and for myself, I set two hours. Over the past few months, we have been undertaking a fairly rigid training regime, but we did manage to slot in our wedding and honeymoon around it. We have progressed really well in the last few weeks, and we feel like we are in very good positions physically and mentally. Being much the more active of the two of us meant Beth was well on track with her target, meanwhile I was keeping my fingers crossed for mine.

The final couple of weeks of training were much more relaxed. We slowed down, preparing ourselves for the big day.

Beth produced a fantastic display in the race, finishing with a time of 1:42, beating her target time of 1:45. An excellent run despite being tripped by another runner at mile 10.

My race didn't quite go to plan. I managed to finish the race but well behind my target time. From mile 3, I was suffering with some pain with my knee, and it progressively got worse. I was aided by the fantastic people from St Johns Ambulance on the finish line and have been instructed to rest for 6-12 weeks. I was gutted but these things happen.

Despite all this, we thoroughly enjoyed the day. The local support is phenomenal. I was staggered by the number of people watching, cheering, applauding, offering sage advice or handing out sweets. You name it, they did it. Truly astonishing.

We are very pleased with our successes in completing the half marathon and the money and awareness that we have raised. No doubt we will get the bug again soon and will be looking for our next fundraiser. Thank you so much to Jack and Beth who have raised **£3274**.

https://www.justgiving.com/fundraising/superted2022

Samantha Morrison and her husband Pete also ran in the GNR for AGSD-UK. The charity has been a massive part of their lives and they have managed to raise an amazing **£863!**

https://tinyurl.com/3kbc3zmj

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Pompe get-together
Birmingham

July's AGSD-UK Pompe social was the first face-to face get-together for the community after so many months of being restricted to on-line meet-ups.

All those attending appreciated the chance to reconnect with old friends, meet new ones and to be around others facing similar challenges.

Here are some of their comments:

'I have enjoyed meeting up with old friends and new ones. It is invigorating to have things in common with other people and not feel an odd one out. There was much cheerfulness in the room it gave me a happy feeling, very welcome after the last few years. I realised how lonely the last few years have been and how we need more social meetings to enhance our quality of life'

'Meeting friends who are living through similar experiences. Being part of a very select group whose life's journey is so different from the mainstream'

'I have enjoyed meeting up with old and new friends'

'I have enjoyed meeting up face to face with my Pompe family. Having a chat and a laugh is good for Mental Health'

'Meeting everyone and now making friends'

'Enjoyed catching up after a long break'

'Seeing my tribe - being with those who understand me without any judgement'

'Getting out of the house, meeting friends and putting a face to people on Facebook'

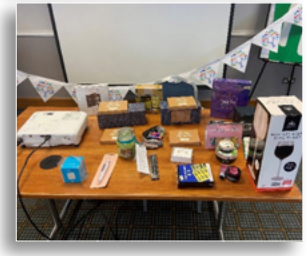
'Seeing everybody, relaxed agenda'

Funds from a raffle organised by Pompe Support Team member Gemma went to help Julie Garfield's efforts to send a sloth to everyone newly diagnosed with Pompe.

We know how much people in the community appreciate this so hopefully the raffle money can go a little way to support her in this.

'Today has been a fantastic, relaxing atmosphere. Great to have a catch up informally'

'No agenda, just being able to speak to others, with no medical science behind it. Lovely meeting up with people that we've met as it has been a while since we saw them'.



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Some top tips that got some of our lovely members through a tough last couple of years!

'The support from Pompe family'

'My strength- My Pompe community and meditation'

'Facebook, family, and friends'

'Keeping a positive mind and talking got me through the last few years. Focussing on the challenges to come and taking each day as it comes'

'Gemma's quizzes and coffee mornings. The cats and walks'

'Wine, lol. Positive attitude and good friends including Pompe family'

Please keep an eye out on our social media pages (Facebook, Instagram, and Twitter) for more information about future events for different sections of the GSD-community, both online and around the UK.

If you want to contact one of the AGSD-UK staff or would like to connect with the PST please email info@agsd.org.uk and we can link you to the right person.

PST Members from left to right: John Foxwell, Ben Parker, Vicky Clarke, Donna Beresford, Gemma Seyfang

AGSD-UK Staff from left to right: Val Buxton (CEO), Zainib Hussain (Benefits Advisor and Community Engagement), Elizabeth Davenport (Specialist Care Advisor), Vicki Lucass (Charity Coordinator)

You can help with McArdle's surveys

Two research projects this year have been seeking support from people with McArdle's to complete questionnaires to help direct their research.

The first is from the team at Radboud University in the Netherlands and you are invited to participate in a study about how individuals with McArdle disease learn to be physically active in daily life and in sports. The more information they can gather about the difficulties, the more people will benefit from this. The survey is anonymous and has nine sections. It will take approximately 20 minutes to complete.

Please note the survey closes soon.

Use this link:
https://tinyurl.com/radboudgsd5
Or email:
mcardledisease.neuro@radboudumc.nl

Radboud University Nijmegen

The second was from Queensland University of Technology in Australia. The team there are researching the effectiveness of current nutritional and exercise guidelines for individuals with McArdle disease.

The first phase has been gathering information from individuals with McArdle disease about the effectiveness of the current guidelines through a short, 10-minute survey.

The results of the survey will guide clinical trial research to commence at the end of 2022. The survey is closed now and QUT confirm the good news that they got enough responses to collect the data they needed.

QUT Queensland University of Technology

McArdle's Walking in Pembrokeshire

This year the walking courses were again in action after a two year gap due to the Covid pandemic. We were based at the ***National Trust Stackpole Centre*** in Pembrokeshire, south west Wales, where we have been before. It is an excellent setting in the ***Pembrokeshire Coast National Park***, next to some lakes and close to the coast with its beautiful beaches and cliffs. For the first time we had the Children & Parents event and the main walking course largely overlapping, and this proved to be very successful and welcomed by both groups.

Our first challenge was that there was a national rail strike on the day that half a dozen of us were due to travel by train down from London to Pembroke. We ended up organising a minibus taxi to collect them from Heathrow and bring them down to Stackpole.

There was an uptick in Covid cases just before the course and so we had plans in place for testing and procedures to follow should anyone test positive. As it happened one person did! Our planning paid off and they were able to largely isolate for a few days, yet still join us outdoors on the walks. Nobody else went down with Covid.

We had get-togethers each morning to discuss different aspects and go over some learning points. Every walk provided opportunities to try out techniques and to sort out issues. We enjoyed walks beside the lakes, along the coast past beaches and dramatic cliffs and even took a boat ride out to Caldey Island to explore that. We went out to St David's Head and did a coastal walk finishing at the lifeboat station. On one day we drove up to North Pembrokeshire to do a circular walk climbing the highest hill in the ***Preseli Mountains, Foel Cwmcerwyn*** at 1,759 feet.

In the evening we often had a talk or presentation by some of the more experienced members of the group, and Sioned Williams gave a short harp recital and talked about her world-class career despite her McArdle's. Of course, there was quite a lot of socialising and on two evenings we went out to a local pub for a meal.

We really lucked out with the most gorgeous weather, really warm and sunny yet there was often a light breeze or some scattered clouds. We just had a few spots of rain.

Across the two events the attendance peaked at 28 people, the most we have had together at any one time on previous courses. A lot was learnt, and many friendships were made. The feedback was excellent with everybody saying they want to come back next year!

Advance notice for next year

For 2023 we will be in Snowdonia, North Wales. The overlapping of the two events this year was a considerable success, so we will be doing that again next year.

The dates will be Friday 28 July to Tuesday 1 August for the Children & Parents event, and 28 July to 4 August for the main course. More details will be put on the website in the next few weeks.

Spotting the lighthouse at the highpoint of Caldey Island

Lucy celebrates making the highpoint

Lucy age 13 from England and Hans age 20 from Germany.

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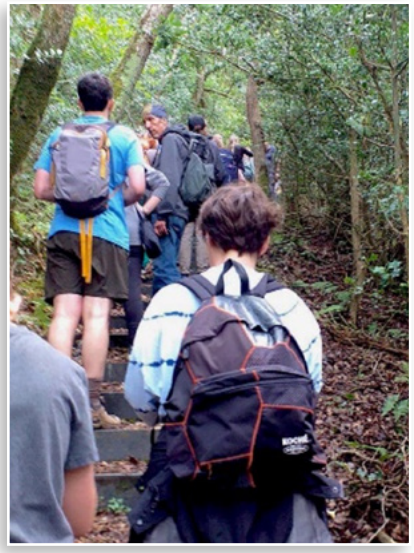
More pictures from the Pembrokeshire walking event



Caoimhe and Eoghan, siblings from Sheffield.

Walking in Wiltshire

The McArdle's weekend of ***Walking in Wiltshire*** over 17 to 19 June went very well, with a total of 8 people attending and most walking on each of the three days. Participants included one coming all the way from Sweden. Based in Bradford-on-Avon, the walks took advantage of river-side and canal-side routes so any uphill sections were relatively short and gentle.

Due to uncertainty about Covid when planning the event, the arrangement was to have everybody book their own accommodation and we would just meet outside for the walks (rather than staying in shared accommodation as we do on the walking courses). After each walk, we met in the evening for a chat and a

meal at various eateries, with outdoor seating. This all worked out well and we had no incidence of Covid.

Thanks to David Thompson for organising this and to all who supported it. ***IamGSD*** is considering drawing up a support package outlining how to organise such an event, in order to make it easier for people to organise these elsewhere around the world.

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The Rising Cost of Living and Where You Can Get Help

We know that for lots of people and their families, the rising cost of living is a real issue so I hope that the following information might be helpful.

Energy Cap: Keep yourself updated on the latest government plan to cap the cost of energy for British households for two years. *The Priority Services Register* is available to offer support if people are struggling to pay their bills or facing debt problems. Energy suppliers and network operators offer this free service to help people in vulnerable situations. Each keeps their own register, so simply contact your energy supplier or network operator. Suppliers must offer payment plans you can afford under Ofgem rules. This includes reviewing a plan you have agreed before. You can ask for 'emergency credit' if you use a prepay meter.

You are eligible if you:

- Are disabled or have a long-term medical condition
- Have reached your state pension age
- Recovering from an injury
- Have a mental health condition
- Are pregnant or have young children

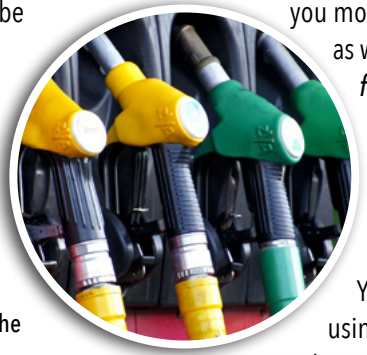
Finally, if you did not get the winter fuel payment that you felt you were entitled to and applied for, then make sure you report it via the *gov.uk* website too.

If you use an oxygen concentrator at home, you can get a rebate for the electricity it uses. The company that supplies your concentrator can make payments to your bank account. Call the Electricity Rebates (BOC Home Oxygen Service) Patient Service Centre on freephone 0800 136 603. Or the Electricity Refund (Air Liquide Healthcare) rebate team on freephone 0800 781 9939.

Childcare: Childcare can be a huge cost for many families and one that takes a considerable chunk of income. But the following might be helpful:

Tax-free childcare; up to £500 every three months for each child (up to £2,000 a year) or up to £1,000 (up to £4,000 a year) for a child with disabilities. 15 to 30 hours of free childcare; available to all three- to four-year-olds in England and Wales. For free childcare in Scotland, you need to make use of the *Funded Early Learning and Childcare* scheme. It is available to all 3–4-year-olds and some 2-year-olds. There are other schemes that will help you pay for childcare with your pre-tax salary.

Council tax: You may qualify for a reduction in your Council Tax bill if an adult or child in your household is disabled. To be eligible, one of the following must apply:



- The adult or child uses a wheelchair indoors.
- You have an extra bathroom or kitchen to meet the needs of the person.
- One of the other rooms is needed and mainly used by the person with disabilities.

Martin Lewis also suggests that you visit his website to check that you are in the correct tax band.

Benefits Check: You might also consider having a benefits check as your needs might have changed. Having the correct benefits may help to claim other reductions and rebates. Please contact AGSD-UK's Benefits Advisor and Engagement Officer, Zainib Hussain at zainib.hussain@agsd.org.uk.

Petrol Prices continue to rise; however, you can use apps and websites like *PetrolPrices* to find the cheapest station near you where you can fill up the tank. You can also save fuel and cut your costs by changing your driving habits. Driving smoothly in a high gear while sticking within the speed limit can knock 25% off your fuel bill, saving the average motorist £300 a year. Making sure your tyres are pumped up, turning off the air conditioning and losing extra weight by emptying the car boot of stuff you don't need, are also small changes that can make a big difference!

Water: If you are struggling to pay your water bills and you have not got a water meter, check whether having one installed will save you money. Many water companies, as well as the *Consumer Council for Water (CCWater)* have water usage calculators on their websites to help you work out how much you are likely to pay if you have a meter.

You can also look at ways of using less water, and you should make sure you do not have any water leaks that are making your bills higher than they should be.

If you are having problems paying a water bill, talk to your water company as soon as you can. You might be able to spread payments over a longer period through a payment plan. Water companies accept a variety of payment methods.

Many water companies run hardship schemes or fund independent charitable trusts which can help you pay your bills. You can find information about the different hardship schemes on the Consumer Council for Water website – or you can ask your water company. If you're on benefits, check if you're eligible for the *Watersure* scheme.

NHS Low Income Scheme may help with prescription costs, dental costs, eyecare costs, healthcare travel costs, wigs, and fabric support. Call 0300 330 1343.

Please contact me for more information

Elizabeth Davenport, Specialist Care Advisor AGSD UK Email: Elizabeth.davenport@agsd.org.uk

Gene therapy trials in hepatic GSDs

In June, AGSD-UK organized an online social and webinar with metabolic consultant Gisela Wilcox to provide hepatic community members with an update on Ultragenyx trial UX053. UX053 is an investigational mRNA therapy encoding glycogen debranching enzyme that is encapsulated in a lipid nanoparticle. mRNA medicines are instructions that direct cells in the body to make proteins like the one missing with GSDIII. The global study is IRB approved and aims to find out if UX053 is a safe and effective treatment for adults with GSD III.

Since then, community members have been keen for an update. We're told that the trial is currently enrolling patients in the USA, Canada, Italy and Spain. Sites in France will open shortly. A trial site is to still to be confirmed in the UK.

For more information please see:

https://tinyurl.com/5av4hs8s

Ultragenyx is also developing, DTX401, an investigational AAV8 gene therapy, designed to deliver stable expression and activity of G6Pase-a using a single intravenous infusion. Long term Phase 1/2 data demonstrate an acceptable safety profile and durability of response. The Phase 3 GlucoGene study is underway to evaluate the ability of DTX401 to reduce the use of cornstarch while maintaining or improving glucose control, as well as the therapy's impact on patients' quality of life. The study is fully enrolled, with patients participating from the USA, Spain and the Netherlands.

For more information please see:

https://tinyurl.com/44h4ev2k

Ultragenyx will be providing an update on its research programs during the IGSD (international glycogen storage disease conference). The online conference will be live starting on October 20th and will be available for viewing for multiple months afterwards. Registration is free.

More conference information, including registration can be found at www.igsd2022.com

Request for GSD1a survey volunteers

A company would like to interview people living with GSD1a and their care givers to understand the impact of GSD1a on their lives.

The information, which would be anonymized, would be used by a pharmaceutical company to develop materials to support people living with GSD1a.

The interview would be approximately 90 minutes. Interviewees will be compensated for their time if they consider this appropriate (approximately $150 USD).

The discussion will explore topics such as

- GSDIa diagnosis
- relationships with care teams
- sources of knowledge
- feeding routines
- lifestyle impacts
- disease burden
- understanding and sentiments toward gene therapy, etc.

If you are interested in participating, please contact ZS Associates at alexandra.bryant@zs.com.

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Research Round Up

A SYSTEMATIC REVIEW INVESTIGATING THE EFFECTIVENESS OF EXERCISE TRAINING IN GLYCOGEN STORAGE DISEASE

(Bordoli et al., 2022)

In this study the authors conducted searches within SCOPUS and MEDLINE to identify potential papers for inclusion to identify literature that has previously focused on the aspect of exercise and training in Pompe and McArdle patients. There are no curative treatments for GSDs, thus therapeutic options, such as exercise training, are aimed at improving QoL by alleviating signs and symptoms. In order to investigate the effectiveness of exercise training in adults with GSDs, they systematically reviewed the literature. Papers were independently assessed for inclusion and quality by two authors. We identified 23 studies which included aerobic training, strength training or respiratory muscle training in patients with McArdle's (n=41) and Pompe disease (n=139).

Results found that in McArdle disease, aerobic exercise training improved aerobic capacity (VO2 peak) by 14–111% with further benefits to functional capacity and well-being. Meanwhile, strength training increased muscle peak power by 100–151% and reduced disease severity. In Pompe disease, a combination of aerobic and strength training improved VO2 peak by 9–10%, muscle peak power by 64%, functional capacity and well-being. Furthermore, respiratory muscle training (RMT) improved respiratory muscular strength [maximum inspiratory pressure (MIP) increased by up to 65% and maximum expiratory pressure (MEP) by up to 70%], with additional benefits shown in aerobic capacity, functional capacity, and well-being.

The researchers concluded that the current evidence shows exercise training appears to be safe and effective in adults with McArdle disease or LOPD, with improvements observed in aerobic capacity, muscular strength, and functional capacity. The effect of RMT in Pompe, where sufficiently intense, was also found to be beneficial, with these improvements appearing to be maintained several months after training stopped. However, these findings are largely based on the limited quality of evidence available, largely derived from small uncontrolled intervention studies of short duration.

To read the full study:

https://tinyurl.com/5d4ady78

CHRONIC FATIGUE: WHEN TO SUSPECT AN INHERITED METABOLIC DISEASE

A.F. Tankeu & Christel Tran (2022)

Tiredness and fatigue are common symptoms and well known to people with a GSD but sometimes overlooked or at worst dismissed. This study comments that glycogen storage disease in particular leads to chronic fatigue. The tiredness is real!

https://pubmed.ncbi.nlm.nih.gov/34983209/

UNDERSTANDING THE ROLE OF SGLT2 INHIBITORS FOR GSD1b

Halligan, RK et al, Orphanet Journal of Rare Diseases. May 2022

This is a study that was discussed in part during meetings facilitated by AGSD-UK.

The conclusion states that empagliflozin is a favourable treatment option for neutropaenia and neutrophil dysfunction in GSD1b, bearing in mind the need for a gradual introduction because of the risk of hypoglycaemia. Improved wound healing in two patients meant that they were able to start PEG feeding. Anaemia resolved in 8 patients. An increase in height and improved blood results were also commented on.

Read the full study here:

https://tinyurl.com/3z6djpkv

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MACHU PICCHU WITH McARDLE'S!

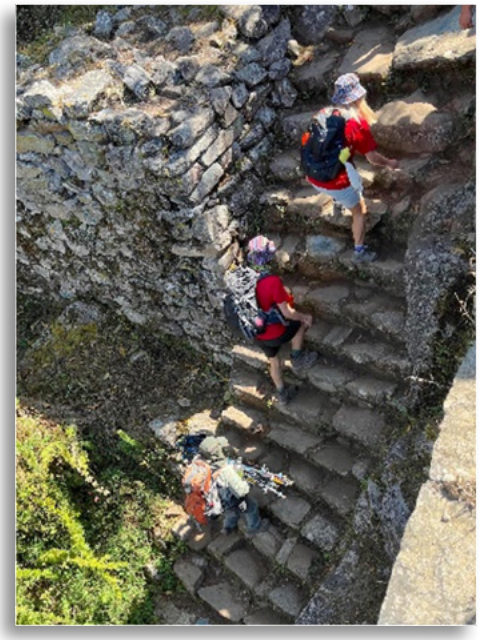
Deborah Corcoran

Diagnosed with McArdle disease at age 33, I never really got much fitter after that, until I had a heart attack in 2011 and realised, I had two options – give up or get fit.

Living in North Wales, I started walking and going up local hills until I found I could get into second-wind and there was no stopping me. I even climbed Snowdon. Then I walked the London Marathon and raised over £3000 for McArdle disease. "What next?" I thought. I always wanted to see Machu Picchu in Peru.

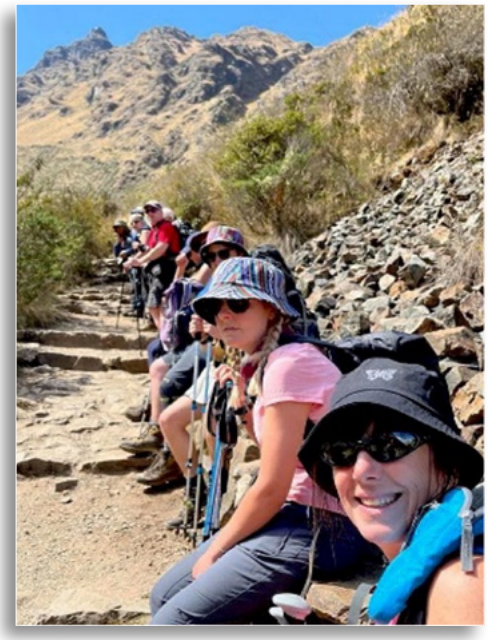
After a lot of planning and flying out to Peru my husband and I finally found ourselves walking along a river in between mountains, and we were off! I found this day OK. Although I was at the back for most of it, I did not feel ostracised. I spoke to our guides and the doctor about McArdle's. It was agreed that when we stopped for breaks or lunch, I was able to set off 10 minutes before the group. This worked so well and on day two when we climbed to the highest point at 4300m over Dead Women's Pass. I was the third person from our group to reach the summit. I was crying inside, tears of joy, tears of exhilaration, me with McArdle's made it to the top of the highest point ahead of 17 other people!

On day three our group was really separated with some people arriving a long time before others, myself and my husband just made it



before dark and a small group at the back finished in the dark with the help of our guides.

The final day was the trek to the *Sun Gate* and our destination of Machu Picchu. It was about a 2-hour trek from camp and not too difficult apart from the *Monkey Steps* which were terrifying for anyone. You could not walk up them, you had to clamber, and I kept having to stop as I was using my arm muscles as well as my legs and it was really hard. Once up them it was plain sailing, and we were an incredibly happy bunch arriving at the Sun Gate which overlooks Machu Picchu. We then walked downhill for around 45 minutes to arrive at Machu Picchu itself and had a lovely tour.



I am so glad I did this and feel really proud of myself. I know how to exercise with McArdle's, and I know my weak points and my positives. It was an amazing experience, and I loved every minute of it.

A full account of Deborah's trip to Machu Picchu will appear on the ***IamGSD*** *website in the personal stories section.*

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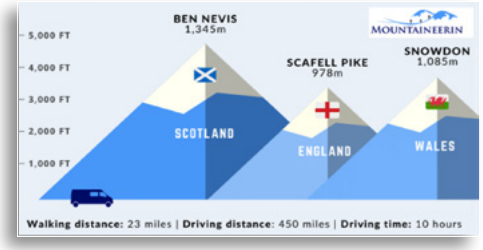
Are you up for the Three Peaks Challenge?

Several people who have been on the McArdle's walking course in Wales over its 12 years have asked about an additional challenge. Maybe others would also like such a challenge. We did once have a week when we attempted to do the 15 peaks in Snowdonia over 3000 feet, and despite a lot of bad weather we managed 9 of them.

Now David Thompson has suggested we should attempt the Three Peaks Challenge, that is climbing the highest peaks in Wales, England, and Scotland. They are Snowdon in North Wales at 3560 feet, Scafell Pike in the Lake District at 3163 feet, and Ben Nevis in the Grampians in Scotland at 4413 feet.



Whereas many people take on the challenge of climbing these three peaks within 24 hours, you will not be surprised to hear that we would not be doing that. To avoid injury people with McArdle's typically need to take about three times as long on uphill sections of walks. The provisional plan is to climb Snowdon on day 1, have a day for rest and transit, climb Scafell Pike on day 3, have another day for rest and transit, and finally climb Ben Nevis on day 5.

With travel to the start and home at the end, this will be a week-long adventure. On the most frequently used route the walking distance is 23 miles, and the amount of ascent is about 9800 feet. Be warned that these are iconic, popular mountains and they can be very crowded.

If this would be of interest to you, please drop a line to: walking.events@agsd.org.uk. If there is sufficient interest, we will start to make plans, possibly for 2023 – plenty of time to get training!

Graphic courtesy of: www.mountaineerin.com

The Main Event

AGSD UK
Association for Glycogen Storage Disease

6 -7th May 2023
Burleigh Court, Loughborough

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

- **Early bird registration available at discounted rate**
- **Register your interest now by emailing info@agsd.org.uk**

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How Benefits Have Helped Improve My Life

Maryam Ahmed

I approached AGSD-UK when I needed support with applying for welfare benefits due to my hepatic GSD. After being rejected for PIP, I was advised by my specialists to get into contact with the charity and take support. My health had deteriorated rapidly over time and my needs changed due to loss of sensation in all my lower limbs meaning I couldn't drive a normal vehicle. I required an adapted vehicle to fit my needs to help me become independent and allow me to be able to attend my appointments.

I was originally put in touch with Jane who helped me fill out my lengthy form. She helped ease my severe anxiety throughout every step of the application. We were confident this would go through, and they would change their decision. However, I started losing hope as Jane was stepping back from the charity into a consultancy role and my anxiety started getting worse again.

I was then advised that Zainib would step in and take over with my case, I was very reluctant at the start as I had been in touch with Jane from the beginning and I was going to withdraw my application with the extreme stress of an appeal. However, once Zainib got in touch with me and started getting information, she put me at ease and had full faith that we would win this appeal. She worked tireless hours trying to sort out the documentation and the evidence. We submitted the appeal and awaited a decision.

After a couple of months, I got an unexpected call from Zainib saying the appeal went through and got accepted.

This was the greatest news which couldn't have come at a better time. On this very day I remember coming home from the hospital stressed about how I would get to my future appointments, as it was becoming straining and tiring. This was further affecting not only my physical but my mental health too.

Throughout the waiting time for the decision both Jane and Zainib were very supportive and helpful, constantly checking in and making sure I was doing well and asking if there was anything they could do to help. It is incredible how people can affect our lives in such amazing ways. Zainib is from an Asian background and being Asian myself, I felt like she could relate to my issues while giving professional advice. I never imagined I would be so reliant on AGSD-UK.

Zainib encouraged me to become a volunteer and now I volunteer with AGSD- UK supporting other people who are struggling, helping with social media and contributing ideas for events and workshops. I'm really looking forward to meeting people at the hepatic GSD get-together very soon

If you need any help or advice with your benefits, please contact our community engagement and benefits advisor, Zainib Hussain at **zainib.hussain@agsd.org.uk**

Excitement of a Pompe Mother

Olivia Osabutey-Ayor

The idea that your child may not walk due to Pompe had me in a constant state of worry. But to God be the Glory I'm so excited to see my beautiful daughter taking her steps now.

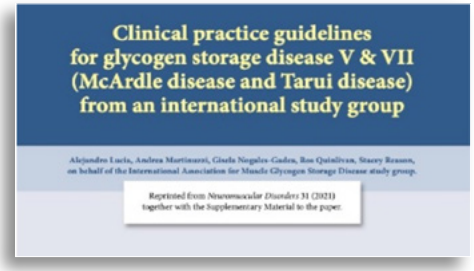
Her feet are getting stronger and firmer every day. I give thanks to God Almighty, the Doctors, the Nurses, and the medicines that everything has worked together for the good of my beautiful daughter Samuella. Not forgetting the amazing people with beautiful hearts who have been so helpful to me and my family in the claiming of my daughter's DLA and always checking on us.

The lovely Zainib Hussain and Elizabeth Davenport at AGSD-UK have been so supportive and caring. We're so grateful. Thank you.

Clinical Practice Guidelines

booklet for GSD5 and GSD7

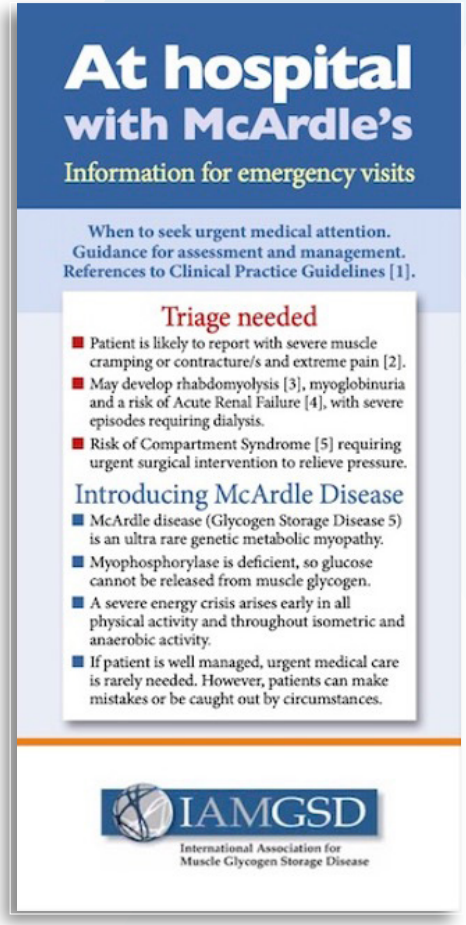
The Clinical Practice Guidelines for GSD5 (McArdle's) and GSD7 (Tarui's), published in Neuromuscular Disorders in December 2021, have now been produced in a booklet form, together with the Supplementary Material which was only available online via the journal. Published by IamGSD, this is now a valuable resource for doctors and over 130 copies have so far been distributed to clinicians, researchers, support groups and other interested parties around the world.


Clinical practice guidelines
for glycogen storage disease V & VII
(McArdle disease and Tarui disease)
from an international study group

When people with McArdle's and Tarui's need to consult with specialists for other conditions (maybe a rheumatologist, a cardiologist, etc), those specialists will usually not know about McArdle's or Tarui's. Taking a copy of this booklet to the appointment will help to provide the sort of information that they may need.

The booklet is available from the McArdle's Coordinator and is also available as a PDF which can be downloaded free of charge from the *IamGSD* website.

IamGSD Leaflet for Emergency Visits


At hospital
with McArdle's
Information for emergency visits

When to seek urgent medical attention.
Guidance for assessment and management.
References to Clinical Practice Guidelines [1].

Triage needed

- Patient is likely to report with severe muscle cramping or contracture/s and extreme pain [2].
- May develop rhabdomyolysis [3], myoglobinuria and a risk of Acute Renal Failure [4], with severe episodes requiring dialysis.
- Risk of Compartment Syndrome [5] requiring urgent surgical intervention to relieve pressure.

Introducing McArdle Disease

- McArdle disease (Glycogen Storage Disease 5) is an ultra rare genetic metabolic myopathy.
- Myophosphorylase is deficient, so glucose cannot be released from muscle glycogen.
- A severe energy crisis arises early in all physical activity and throughout isometric and anaerobic activity.
- If patient is well managed, urgent medical care is rarely needed. However, patients can make mistakes or be caught out by circumstances.

IAMGSD
International Association for
Muscle Glycogen Storage Disease

There is another new leaflet from *IamGSD* in their McArdle's series, this one is entitled *At Hospital*. It is intended for use when someone needs to attend hospital due to overdoing it, and perhaps having rhabdomyolysis. It can be quite difficult to know when medical attention is required, so there is a page aimed at the patient. It guides them on how to assess whether they need to attend hospital, and on what to do when they get there.

The inside is laid out like a traffic-light scheme with guidance for the doctor about what to assess, how to manage the episode, and when to discharge the patient. Then there are notes about particular complications and about lab results. It also highlights the two very important possible complications: acute renal failure and compartment syndrome. The hope is that the new leaflet will enable people with McArdle's to get triaged promptly on arrival and get the correct attention, at the same time making the decision path easier for the doctor. The leaflet was prepared with the assistance of an emergency doctor and *IamGSD's Scientific Advisory Board*.

We suggest that everyone with McArdle's has a copy of this leaflet at home, ready to pick up if they ever have an episode of rhabdomyolysis. You can request a copy from our McArdle's Coordinator, or you can download a PDF from the *IamGSD* website.

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Current management options for GSD1a

Last year a team of international experts from the Netherlands, France, Spain, Italy, the USA, Brazil and Canada put together an article *Glycogen Storage Disease Type 1a: Current Management options, Burden and Unmet Needs*

T.J. Derks et al Oct 2021

They looked at the importance of glycaemic /metabolic control and maintaining a balance with quality of life, independence, and anxiety. The importance of involvement of patients in setting out guidelines was emphasised, with quality of life an over-arching priority. It was refreshing to see this and an acknowledgement of the stress placed on parents and caregivers, was one of the factors acknowledged.

The experts concluded that oral glucose replacement therapy in the form of UCCS and strict control of nutritional treatment has significantly improved the prognosis of patients with GSD1a. However, achieving optimal glycaemic and metabolic control is challenging, and the risk of severe hypoglycaemia and chronic complications persists.

Both the disease itself and the need for meticulous adherence to dietary treatment have a significant physical, psychological, and psychosocial burden on patients and their parents/ caregivers. There is an unmet need for novel monitoring tools that measure outcomes that matter to patients. There is also a need for treatment strategies that address the underlying cause of disease, restore glucose homeostasis, prevent secondary metabolic perturbations and chronic complications, and improve quality of life.

Read the full study here:

https://tinyurl.com/4fnv7f5e

Observational studies in hepatic GSDs

Aerobic Capacity and Skeletal muscle Characteristics in Glycogen Storage Disease IIIa: an observational study

P.J. Hennis et al. Orphanet Journal of Rare Diseases. Jan 22

This research was carried out in the UK and supported and funded by AGSD-UK. Its purpose was to produce the expected values of strength and aerobic capacity in people with GSDIIIa. These could then be used as a reference. Resistance exercise was suggested as a useful activity. Patients and their cares could discuss this with their health practitioners.

The researchers commented:

Experts recognise that exercise is likely to be a useful tool for the assessment of functional status and as a treatment strategy to combat the pathophysiological consequences of GSD IIIa, such as myopathy, low bone mineral density and hypoglycaemia. However, they also state they are unable to provide strong guidance related to the prescription of regular exercise for patients due to the lack of information.

Read the study here:

https://tinyurl.com/37vu97p2

The potential of dietary therapy in patients with glycogen storage disease type IV

Terry D.J. Derks Epub Dec 21

In light of the success of dietary treatment for other hepatic forms of GSD, researchers initiated an observational study to assess the outcome of medical diets to limit the accumulation of glycogen in GSD1V.

The researchers looked at clinical, dietary, laboratory and imaging data. They comment that medical diets may have the potential to delay or prevent liver transplantation, improve growth and normalise serum aminotransferases.

Read the study here:

https://tinyurl.com/53x9sy94

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NICE Recommends Avalglucosidase alfa

The *National Institute for Health and Care Excellence* has published its recommendation of *Avalglucosidase alfa* (AVAL) as an option for treating Pompe in babies, children, young people, and adults, where AVAL is provided according to the commercial arrangement with the company.

The recommendation is the outcome a *NICE* technology appraisal process which carefully reviewed the clinical evidence available to date and submissions from key stakeholders.

Along with evidence submitted by *Sanofi Genzyme* and by clinical experts, the appraisal committee considered an AGSD-UK submission setting out survey responses from the Pompe community about the impact of the condition. The committee also heard from our nominated patient experts about the effect Pompe has on their lives and experience of using AVAL.

In their published summary of the benefits AVAL is expected to provide, *NICE* stated:

Avalglucosidase alfa (AVAL) is indicated for the long-term treatment of Pompe disease. AVAL is expected to provide benefits as a treatment option for IOPD and LOPD. Clinical experts explained that AVAL is the same enzyme as ALGLU but has a better delivery mechanism which should get more enzyme into muscle cells. Therefore, they expect AVAL to have a positive effect for people with Pompe disease and be a better option than ALGLU. People with Pompe disease are optimistic about future treatment with AVAL. One person who has had treatment with AVAL told of the positive effect it has had on their life. Since treatment with AVAL in the clinical trial, they no longer have mobility or breathing problems, and do not have to worry about not being able to do things that people without the disease may be able to do. The committee concluded that clinicians and people with Pompe disease would welcome an effective alternative to current treatment.

Pompe Support Team Member, *Gemma Seyfang* has been on the clinical trial for the last 5 and a half years and is excited that it has been so worthwhile, not only for her but for the wider Pompe community.

Gemma made an important contribution to the NICE process as one of our nominated patient experts and is delighted that fellow members of the community will now have access to the therapy.

Gemma's reaction on the day the recommendation was announced:

'This is a huge milestone for us. We have now made history as this is the first time NICE has approved a drug for Pompe Disease and by doing so we have put Pompe Disease on the map! Plus, the vials are double the size meaning less mixing time... let's celebrate!!'

Gemma Seyfang

'Sing it in the valley, shout it from the mountain top, Nexviazyme has been approved, and the hope will never stop!'

We're currently pulling together evidence for our submission to the NICE review of cipaglucosidase alfa with miglustat.

Email info@agsd.org.uk to tell us about your experience of the therapy.

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Reneo update on drug for McArdle's

Reneo Pharmaceuticals has provided an update on their Phase 1B trial of a drug code named REN001. This was being trialled in a number of conditions, including McArdle disease. The company stated: *We are pleased to see good safety and tolerability of REN001 in McArdle patients, as well as a substantial increase in fatty acid oxidation, a finding that is consistent with our prior clinical experience and encouraging as it relates to the ongoing program. We also saw a modest increase in walk distance but did not see a corresponding reduction in the common symptoms that McArdle patients experience*.

After 12 weeks of treatment, the 17 participants had showed an average increase of 38.5% in fat oxidation compared to baseline, but there were no major changes in the distance walked during the 12-minute walk test and no corresponding changes in heart rate, fatigue or pain. Given these findings Reneo does not plan to include McArdle's in their Phase 2 study but will continue to explore other diseases that are defined by the impairment of mitochondria to produce cellular energy. Reneo thanked the patients, families, and caregivers who made this study in McArdle disease possible. They continue to partner with IamGSD to further their advocacy and education mission.

Benefit of 5 years of enzyme replacement therapy in advanced LOPD

Maharaj et al., 2022

A study looked at the benefits of ERT on a patient that had been misdiagnosed for three decades with acute respiratory failure at presentation.

A 57 year old Caucasian Northern European non consanguineous female presented to the emergency room with a six month history of progressive generalized weakness, dysphagia, and increasing rest dyspnoea, orthopnoea, with marked deterioration two weeks prior to presentation. She reported being symptomatic of limb girdle weakness since her mid 20's and carried a diagnosis of "probable polymyositis" at age 26, after being assessed by neurology and a muscle biopsy performed. She received treatment with oral Cyclophosphamide and Prednisone for about a year. Over the last fifteen years, she was seen by four further neurologists with two further muscle biopsies performed within a year, because of a lack of a clear diagnosis.

In summary, the latter two biopsies, reported by the same neuropathologist, indicated no evidence of an inflammatory myopathy but was otherwise non diagnostic. Because Pompe was not considered by three of these clinicians in the differential diagnosis, acid maltase staining was not performed, and the diagnosis missed. The fourth neurologist assessed the patient with this presentation and the diagnosis of Pompe was considered.

Past medical history included hypertension, gastroesophageal reflux, treated hypothyroidism and a longstanding myopathy. Sensorineural hearing loss was diagnosed at age 38 and required hearing aids. She was not on a statin and there was no family history of muscle disease.

She was treated with enzyme replacement therapy (ERT) for over five years, after being diagnosed with late onset Pompe Disease (LOPD). She returned to independent living with the use of non-invasive ventilation at nights. ERT should be considered in the management of patients with advanced LOPD and the effects of ERT closely monitored.

You can read the full study here:

https://tinyurl.com/fr4hsxnm

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Pompe Research in Newcastle

At our online Pompe event in July, Newcastle Professor of Neuromuscular Disorders, and honorary consultant clinical geneticist ***Jordi Diaz-Manera*** gave an overview of some of the research his team is developing in Newcastle, along with developments in the Pompe field.

He mentioned the following research projects:

1. A research study with a new MRI sequence called carbon spectroscopy that is able to quantify glycogen in different organs. They are assessing if this tool is able to detect and quantify glycogen in the skeletal muscles of patients with Pompe and if there is a correlation between the amount of glycogen in the muscles and results of muscle function tests. They would like to use this tool as an outcome measure for future clinical trials. They are at this moment recruiting patients for this study. If you are interested in knowing more, you can read further about the study at:

   www.isrctn.com/ISRCTN11241741

And you can also contact the team at:

jordi.diaz-manera@newcastle.ac.uk
alejandro.gonzalez-chamorro@newcastle.ac.uk
steph.clutterbuck@newcastle.ac.uk

2. A research study that is implementing artificial intelligence to the analysis of muscle MRI aiming to accelerate the diagnosis process of patients with neuromuscular diseases. The group has already developed a first software, called Myo-Guide, that is able to predict a diagnosis from a list of 10 diseases assessing the MRIs. They are trying to increase the number of diseases that this software can work with to 25. For this they are collaborating with several groups around the world to obtain a large number of MRIs.

You can find more by contacting any of the following:

jordi.diaz-manera@newcastle.ac.uk
jose.verdu-diaz2@newcastle.ac.uk
alejandro.gonzalez-chamorro@newcastle.ac.uk
steph.clutterbuck@newcastle.ac.uk

3. The FORTIS gene therapy clinical trial sponsored by Astellas, is being run in several hospitals in USA and Europe. Newcastle in one of the centres in UK delivering the study. Although recruitment is not now active, they are looking for patients with late onset Pompe disease who are under a stable regimen of enzymatic replacement therapy.

For further details you can contact:

jordi.diaz-manera@newcastle.ac.uk
ellie.drummond@newcastle.ac.uk

Professor Diaz-Manera and the Newcastle University team at ***John Walton Muscular Dystrophy Research Centre*** have also been raising money for AGSD-UK. They not only ran in the ***Great North Run*** but also held a bake sale and raffle to raise money for the charity!

Thank you to the whole team for all their continued hard work!

THE John Walton MUSCULAR DYSTROPHY RESEARCH CENTRE

Newcastle University

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Small charities - why the work they do is invaluable!

Small charities do some great work, and of course, the AGSD-UK is one of them! How and why are we better than some of the larger charities and what challenges do we face?

We know our **community** because we **come from it** and **are part of it**. Trustees, volunteers, and some staff are **directly affected by GSD**. Consequently, we personally understand the challenges the charity tackles and are truly able to **represent the community we serve**.

We are specialists. We pride ourselves on our **knowledge and experience** of the **GSD world**. We are in touch with and are available to all people involved in this world, whether they be people affected by GSD, their family and friends, health professionals, or pharmacologists and researchers **looking for a better treatment**.

We **understand the impact** that we have by **monitoring it** and **personally witnessing** any improvements made to the lives of those we support. Because of the charity's in-depth and personal knowledge, corporate funders and individuals who donate can have faith that **any money given** will reach those **who need it most**.

As a small community we are able to support each other through **relationships** where people know and understand each other. **There is no need to be alone.** Members help each other with chat, swapping ideas, talking about their lives, and making friendships with a **true understanding**.

We are well positioned in the community and reach out to all affected by any type of GSD. **Because of this, people who would never ask for help come to us.** We are **experienced** in helping those who are hardest to reach.

We can be **agile, innovative, and responsive** to needs and changes, without the politics and bureaucracy of larger charities. Our small size means we **work as a team**, understanding and appreciating each other's skills.

We are there to support the community. This is invaluable and hard to put a price on. Something we know all our funders and fundraisers appreciate. **Yet.....**

The largest 4% of charities in the UK receive 82% of the income from charitable giving.

Help us to redress this balance!

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

AGSD UK
Association for Glycogen Storage Disease

Take away from this Glisten...

Theodore Molyneux and his Aunt and Uncle!

Upcoming events!

- McArdle and muscle Zoom get-together
  29th October 2022
- Pompe get-together in Swansea, December 2nd 2022
- AGSD-UK Main Event, Loughborough
  6th and 7th May 2023

Get in touch now with **info@agsd.org.uk** for joining details. And look out for more opportunities to come together in 2023.

Early bird registration is available now for the ***AGSD-UK Main Event*** in May next year at reduced cost - don't delay - contact us today!

Office contact details:

AGSD-UK, PO Box 699
SOUTHAMPTON, SO50 0QT
Phone 0300 123 2790

Email info@agsd.org.uk
www.agsd.org.uk

Charity number
1132271

Autumn
2022

Emma Grove Design 07990 520 807

AGSD UK
Association for Glycogen Storage Disease