March 2022

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

We have had lots of the AGSD-UK community fundraising for us! See inside for more details

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Charity Number :1132271
March 2022

Thank you!

An important question...
Could you help with fundraising?

We have had varying amounts raised for us this year by supporters in a multitude of different ways; perhaps you could use some of their stories as inspiration.

Thank you again to everyone who supports us and we hope to continue helping you long into the future.

Front Cover Images:
Orin and Olivia Arthur, who have Infantile Onset Pompe Disease, during a trip to the beach.

Samuella and her father

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.

Contents

Message from the Chief Executive

Welcome to the spring 2022 edition of Glisten. Despite the challenging times, we hope this heralds brighter things ahead, with more chances to meet up and support each other. After our successful online *Winter Season* of events and presentations (see inside for details on how to catch up with these) we're hoping to be able to move forward with a face-to-face conference in the autumn, bringing the AGSD-UK community together again. See back page to register your interest now!

This edition also contains some green shoots in the form of the latest on promising research and developments, along with information on some of the aspects of living with a GSD you've told us you want to see more focus on, such as employment and education.

As we make plans for the coming year and beyond, it's really important everyone affected has the chance to have their say. There's still time to send your top three things that would improve life with the condition, to help shape AGSD-UK priorities. Just email me at val.buxton@agsd.org.uk.

We'd also like feedback about the things you want funders, policy makers, professionals, and the public to know about living with GSD – so we can make sure our messaging has the biggest possible impact.

Finally, here's a shout out for people who could give a little time be involved in phone or online groups to share ideas around fundraising, publications, or events planning. We'd aim to make this fun and interactive and keep the time commitment to a minimum!

Involvement from across the community is what makes AGSD-UK strong, effective, and relevant for people with any of the GSDs, so please do get in touch with your ideas or to find out more.

Looking forward very much to hearing from you.

Val Buxton

Fundraising Round-up

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

Sonia Worthy has raised £545

Having been diagnosed with McArdle's Disease in 2008 at the age of 60, I decided to organise a Silent Auction to raise money for AGSD. I have been to the AGSD Conference and taken part in one of Andrew's walking holidays, so decided to do some fundraising. I was the president of Clay Cross Inner Wheel Club, and a Silent Auction was a regular event on our calendar. It was originally planned for the first week of lockdown but obviously had to be postponed and all the goods stored in my spare room until October 2021. We have usually had about 50 people to these events and raised a considerable amount of money but unfortunately, we were down to 28 this time as people were Covid cautious. We did manage to raise £545 and those who came really enjoyed themselves as it was the first social event we had organised since Covid that wasn't online.

For people who have not taken part in a Silent Auction, goods are donated, and a bid sheet is placed by each item. The highest bid at the end of the auction receives the item. It is all good fun as people try to outbid one another.

Nikki Christie has raised £311.10

Nikki and Penns Primary School organised fundraising for AGSD-UK at their Christmas Carol Concert. The school held a festive afternoon where the pupils performed in a concert full of songs, poetry, readings and more! Thank you to Nikki and the school!

Frank and Irene Green have raised £602

Frank and Irene collected money with their Xmas decorations again this year

Surrey District Association of the Camping and Caravanning Club has raised £404.76

AGSD-UK is the chosen charity for the Surrey District Association and has been for a few years. One of their members, Mylo Moore has GSD3a, which is why it is close to their hearts. They have been fundraising throughout the year and have so far raised £404.76. This has been through coffee mornings, hot dogs at their last 'night at the proms' event and many small donations from other events they have hosted.

Condolences and a thank you to Sue Smith

We send our thoughts and condolences along with our thanks to Sue who collected £165 in donations for AGSD at her mother's funeral. We very much appreciate this donation in memory of Sue's mum.

If you are fundraising, email info@agsd.org.uk to ask for hard copies of our charity leaflet to help promote our cause and support your fundraising.

Remembering Louise Bett

AGSD-UK was very saddened to learn of the recent passing of Louise Bett. Louise was a long-time member of the charity and will be remembered for her determined and cheerful personality. Our deepest sympathies go to all her family and friends.

Our thanks go to everyone who has contributed to the JustGiving fund in her memory. This will be used to continue the essential work the charity does to lessen the load of life with glycogen storage disease.

Louise was born with GSD type 3 on the 14th February 1985. Although she lived with this disease all her life only a few would know as Louise did not want this to define her life or to be treated differently from others. She never let GSD get in the way of her goals or ambitions both at work and in play. Louise had many talents, none more so than her ability to make others feel special and good within their lives. Her generosity with both her finances and more importantly her time for others in helping with everyone else's struggles in life were well known to all who cared for her or even if they just knew her for a short time. She could put a smile on people's faces just walking into a room as she was so bright, bubbly, and funny, just full of life and it was so infectious that everyone who was lucky enough to know her would instantly be uplifted by her personality and zest for life.

Her ability to touch your heart was so special to everyone. Louise never let much stop her in her desire to live life to the full, be it special holidays or nights out on the town or giving up her time even when she may not have been feeling at her best. Louise's caring, loving, bubbly, optimistic nature was something that most of us can only aspire to. Louise will be in the in the hearts of her mum Liz and dad Jim for eternity, as well as in the hearts of all those friends who loved her deeply.

Our thanks go to *Media 10* who raised £2136 in memory of Louise.

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

Employment Rights

Elizabeth Davenport

Hello everyone! During my time with AGSD UK, a few people have contacted me regarding employment rights and holding down employment so I thought that I would share the following information. Firstly, it is important to highlight that if you have physical, learning, or sensory difficulties, you have additional rights at work. This is known as protected characteristics. This could include whether you can take time off work for appointments, work more flexible hours, or have adjustments made to your job or place of work.

Here's a summary of your legal rights at work and support to help you keep working.

- Your employer is not allowed to discriminate against you because you are disabled
- They must keep your job open for you and cannot pressure you to resign because you've become disabled
- Your employer must make reasonable adjustments to your place of work, the job you do, or the terms and conditions of your employment

So, what are reasonable adjustments at work?

Your employer must make 'reasonable adjustments' so that you are not disadvantaged at work. These could include the following:

- Adapting the workplace, for example by providing ramps
- Flexible working hours, time off for appointments or more breaks
- Giving you alternative duties or moving you to a more suitable workplace
- Providing specially adapted equipment, such as a computer, keyboard, telephone, chair, or desk
- Counting and recording disability related absence separately to sick leave
- Providing a support worker

Your employer will only be required to implement adjustments which would be 'reasonable' for them to implement. This includes the cost involved, the impact on others/their business, and whether the adjustments have a real prospect of being effective. Your employer is responsible for paying for the cost of the adjustment(s). The Government's Access to Work scheme may be able to contribute towards the cost of adjustments you need at work. More information can be found at: www.gov.uk/access-to-work.

I do understand that it can be stressful to speak to your employer and share personal information but please remember that the *Equality Act 2010* is in place to protect you along with charities, advocates, and union reps. If you would like more information, then please do email me and we can arrange a chat.

elizabeth.davenport@agsd.org.uk

Winter Season 2022

This January AGSD-UK held our second successful Winter Season of online talks, presentations, and events.

We ran our first Winter Season last year, when the pandemic meant we were unable to hold our face-to-face conference. This year we changed things up a little, showcasing the presentations and events over one month, with playlists themed around Videos for All, Pompe, McArdle's and the Hepatic GSDs. The new 'Videos for All' theme included introductions from our staff and volunteers as well as general topics such as a gene therapy talk from Dr Zoe Farrington, who works at Ultragenyx Pharmaceutical. The other themes included presentations from some of the top medical professionals in the field, such as Dr Charlotte Dawson (GSD2), Professor Ros Quinlivin (GSD5) and Joanna Gribben and Gemma Hack (Hepatic). These playlists also included videos from some of our members whose lives are affected by GSD.

Feedback has been overwhelmingly positive again this year and a huge thanks goes to the medical professionals and members who got involved and created the useful and engaging content, as well as to all those who watched and joined in. We always strive to provide you with up to date and interesting information as well as stories for members to relate to and hopefully gain something from. Special thanks also go to our fundraisers, donors and industry partners, many of whom support us each year.

Even though the Winter Season has now ended, the videos will remain on our YouTube channel *GSD Screen* for you to visit and watch at your leisure.

All details of our *Winter Season* can be found on the AGSD-UK website.

Vicki Lucass, Season Producer

Our viewing figures

Since we launched our *Winter Season* at the beginning of January, our YouTube channel GSD Screen, has had 2228 views, which is a significant increase for the channel, as well as 19 new subscribers. We have had views from all over the world including the UK, USA, Canada and Germany.

There have been people visiting our channel to watch presentations every day since the launch on January 3rd. Our most successful day was the 1st February where we had 128 views of our videos.

A Snapshot of the Season

We are incredibly grateful to all who have taken the time to record content for us. The presentations have been a great success so we would like to send a big thank you to our GSD2 Pompe contributors; Dr Charlotte Dawson, Antonio Ochoa Ferraro, Dr Jordi Diaz-Manera, Andrea Duckworth, Gemma Seyfang, Luke Fraser and Olivia Osabutey Ayor. Our GSD5 McArdle contributors; Professor Ros Quinlivin, Dr Stacey Reason, Wendy Newman, Andrew Wakelin, Andy Williams, David Thompson, Joff Brown and Sioned Williams. Our Hepatic contributors; Antonio Ochoa Ferraro, Gemma Hack, Jo Gribben, Ciara Harkins, Matthew Arthur and Sarah King. Our Videos for all contributors; AGSD-UK Staff Team, Dr Maninder Ahluwalia and Dr Zoe Farrington.

A small taster of the season….

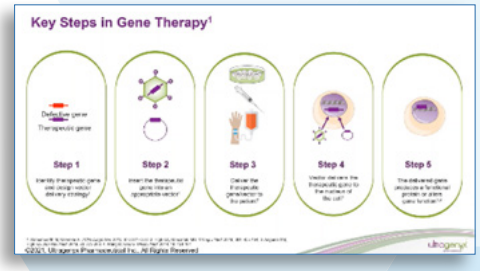
Here is just a small snapshot of a few of our informative presentations from the medical professionals and our lovely members who discuss their personal experiences with GSD as well as tips and advice. Visit our channel to watch these excellent videos.

Meet the AGSD-UK Team (Videos for all)

In this video we introduce our staff here at AGSD-UK. This includes CEO Val Buxton, Specialist Care Advisor Elizabeth Davenport, Benefits Advisor Zanib Hussain, Finance Officer Jackie Henson and our Charity Co-ordinator Vicki Lucass. If you would like to contact any of our staff members for support or assistance or would like more information, please email info@agsd.org.uk

Gene Therapy (Videos for all)

Dr Zoe Farrington from Ultragenyx Pharmaceutical present Gene Therapy. Ultragenyx is a biopharmaceutical company committed to bringing to patients novel products for the treatment of rare and ultra-rare diseases, with a focus on serious, debilitating genetic diseases.



New Treatments for Pompe Disease under EAMS (GSD2 Pompe)

In this presentation, Dr Charlotte Dawson and Antonio Ochoa Ferraro from *Queen Elizabeth Hospital* in Birmingham, talk to us about the two new treatments for Pompe under the *Early Access to Medicine Scheme*.

They discuss the research process that happens before treatment is available to patients, early access to Cipaglucosidase, treatment under EAMS, Cipaglucosidase alfa and Miglustat, practical aspects, Cipaglucosidase alfa compared with Myozome and trial outcomes.

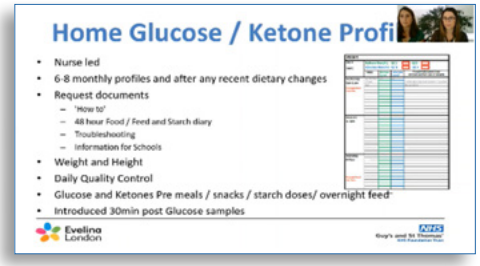
Travelling with Pompe Disease (GSD2 Pompe)

Andrea tells us about the experience she has gained over the years of travelling with Pompe Disease. She gives some tips and advice on how she doesn't let her GSD stop her from getting out and about and enjoying holidays and quality time with her family.

GSD at Evelina London Children's Hospital (Hepatic GSDs)

In their presentation, Jo Gribben and Gemma Hack talk to us about how the GSD service works at *Evelina London Children's Hospital*. To name a few things that are covered within this informative presentation, Jo and Gemma

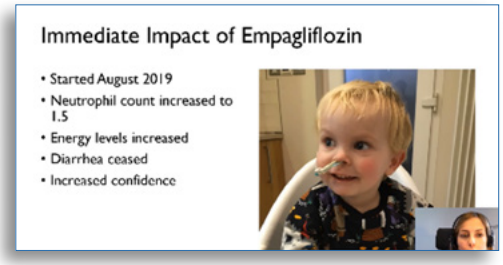
discuss the steps that happen from referral and on through the treatment process, including the various tests that will be undertaken, specific diet and nutrition and how patients are supported through their time at *Evelina London* until they are transferred to the adult metabolic unit.



Life Pre and Post Empagliflozin (Hepatic GSDs)

In this insightful presentation, Sarah discusses her family's experience with treatment for her son who has GSD1b.

Sarah talks to us about one of the main symptoms of GSD1b which is neutropenia and how it has affected her son James. She begins with his birth and the diagnosis process that they experienced and continues into early life. She covers the latest research as well as the treatment that James receives and the positive outcome he has experienced so far. She ends her presentation with long-term impact for James after having been on *Empagliflozin* for 2 years now.



Clinical Practice Guidelines for GSD5 & 7

Dr Stacey Reason, President of *IamGSD*, explains the concept of *Clinical Practice Guidelines* and how these have developed for GSD5 (McArdle Disease) and GSD7 (Tauri Disease). She introduces the international team of expert clinicians and researchers who were brought together to prepare the guidelines and explains the target audience and the extent of the contents. The guidelines are now published in *Neuromuscular Disorders (Journal of the World Muscle Society)* and have 'Open Access', so that anyone including patients, may download and refer to them free of charge.

Her passion for music, photography, and poetry - Sioned Williams (GSD5 McArdle)

Sioned Williams is an internationally renowned harpist who is passionate about her music, but here she speaks animatedly about some other interests too.

She speaks of her childhood experience with McArdle's symptoms, although not diagnosed for many years. An occurrence in 2017 led to

Sioned re-igniting her love of walking, and during her strolls, she started taking informal photos with her iPhone. Sharing images on *Facebook*, accompanied with short quotations, primarily from the great Persian poet *Rumi*, Sioned has quite a following for her new-found interest!

A Season of Events

This *Winter Season*, we ran some events for the AGSD-UK community. One of our volunteers Gemma Seyfang got herself hugely involved again in helping us provide fun events for all of our members affected by GSD. She held two quizzes, one for children and one for the adults. Thanks to Gemma, who also held the *Pompe Sunday Social*.

Thanks also go to our McArdle's co-ordinator, Andrew Wakelin for organising and putting in a lot of time and effort into the McArdle *Zoom* get-togethers and to Ailsa Arthur, one of our trustees for hosting the Hepatic coffee morning.

This year we also held a Pompe Live Event. Jacose Bell from Spark Therapeutics presented on the investigational gene therapy approaches for LOPD with an opportunity for a Q&A. The presentation was brilliant, informative, and well received by those who attended the event. Due to the nature of the content, we were unable to record her presentation.

If you would be interested in us running this Live event again, please email seasonproducer@agsd.org.uk

Thank you to Jacose for offering this opportunity for AGSD-UK.

How can I watch?!

If you haven't managed to watch our *Winter Season* presentations yet or are not sure how you can access them, here are a few tips. All of our videos are free to watch, any time you want!

Our YouTube Channel

https://www.youtube.com/GSDScreen

- Go to playlists
- Here you will find *Winter Season 2022* playlists
- Click on specific playlist you want to watch, and you will find a selection of presentations

Through our website

- Go to https://agsd.org.uk/
- Hover the mouse over 'how we help'
- Under events you will see *Winter Season 2022*. Click on this
- Here you will find all of the information on our *Winter Season*
- As you scroll down the page you will see the different playlists with a link that you can click on. This will take you directly to the playlist.

If you have any questions, have any feedback, or need any extra guidance on how to access our *Winter Season*, please email seasonproducer@agsd.org.uk

Pompe Research Invitation

Nicola Condon

I am a Master's Student studying Clinical Health Research at the University of Birmingham and also a Physiotherapist working at the Queen Elizabeth Hospital Birmingham. For my thesis I will be conducting a research study exploring physical activity and exercise for people living with Pompe disease and I would appreciate your help!

Study Title

Barriers and facilitators to Physical Activity and exercise in Late Onset Pompe Disease (LOPD): A qualitative study exploring patient perspectives and experiences

About the study

The purpose of this research study is to explore the perspectives and experiences of people with Pompe Disease when engaging in physical activity and exercise. To do this I am interviewing people with Pompe disease who are happy to talk about their experiences with exercise before and after their diagnosis and any challenges encountered when incorporating physical activity and/or exercise into your lives.

What does the study involve?

The study will involve an interview, which will likely take no longer than an hour. It can be completed over the phone or an on-line Zoom video call, whichever is easiest for you.

Who can take part?

Potential recruits to the study will be invited as follows:

- you are aged 18 years or over
- a UK resident
- and have been diagnosed with Pompe Disease.

I am interested in hearing your views and experiences, and keen to hear from all perspectives:

- Whatever your age or physical limitations
- Whether you see yourself as generally physically active or not
- Your experiences with exercise – good or bad / past and present
- If you wish to be more active what are the major barriers or possible solutions
- How important physical activity is to you as part of managing your condition
- How the national pandemic has impacted on physical activity

Consent to participate and Participant Information Leaflet

If you are interested in taking part in this study further information can be found on this URL link https://bham.onlinesurveys.ac.uk/exercise-and-pompe-disease-1.

Participants will provide electronic consent after reading the Participant Information Leaflet embedded in the survey.

Study Timeline

Recruitment period - February 2022- beginning of April 2022.

Ethical Approval

The study has been approved by the University of Birmingham's Science, Technology, Engineering and Mathematics Ethical Review Committee.

Plans for publication of results/ findings

We plan to present the findings of the study in national and international conferences and to publish them in a reputable peer-reviewed journal.

Contact and Further information:

If you want any further information about this study, please contact Nicola Condon via e-mail NJC645@student.bham.ac.uk

Thank you for your time and interest in this study.

Orin and Olivia

Hello everyone, we are the Arthurs, and we are parents to twins with Infantile Onset Pompe Disease.

This is the definition of our family, however it does not define us. Orin and Olivia were born in 2014 and went through a series of fortunate events which meant they were quickly diagnosed with IOPD.

Olivia spent her first year and a half in hospital between ICU and HDU and the day after her 1st birthday was given a tracheostomy and long-term ventilation. Many people assume our story is a sad hardship affair, nothing could be further from our reality!

For 18 months we were prepared for the worst with frequent visits to "the black sofa room" (for those not familiar, this is where you are taken off ward to give you really bad news), however our children are the strongest warriors I have ever encountered and continued to mystify everyone and got stronger each day. After over a year and a half in hospital (Olivia an inpatient and us living in Ronald McDonald house with Orin) we were finally home all together, and we promised each other we would focus every day on giving the twins their best life. Do not get me wrong in the back of our minds the reality of IOPD is always there, but it is not the daily focus. For the last 5 years we have had some of the most amazing highs and lows. Against all odds the twins have enjoyed visiting many countries travelling as far as Tenerife (I think we traumatised that airline forever with our ventilators and inflight chairs lol) and both attend mainstream school.

However, no matter how high you are flying you can come crashing to the ground in a split second. Olivia has a habit of becoming extremely ill in the blink of an eye and on more than a few occasions we have feared her journey was at an end. Then as if nothing had even happened to her, she is sitting up in bed watching ***Frozen***. I kid you not, I once spoke to a consultant in the hall, telling me after surgery to prepare for the worst as I watched her through the window over his shoulder swinging her teddy watching ***Frozen***. The surgeon himself was in disbelief of her strength and resilience.

With Orin it is so easy to forget he even has Pompe sometimes as he is doing so well. Orin has speech delay and walks on his tiptoes (which results in many tumbles if he doesn't keep his splints on). We were told that he was extremely unlikely to ever walk or talk.

Pompe has a massive impact on our lives but we have found a way to integrate it as just another little part of who we are. In my experience families with lifelong conditions seem to share a darker humour than most other people, as we frequently know the prognosis and to casual observers, we may appear deluded or in denial. I believe we look at the world differently and want to truly embrace life because we see its limits.

Life with the twins was going great. Their treatments were working well, both were in mainstream school (currently home schooled but will go back).

Two years ago, we were finally in a great place and planning a big adventure. ***Make a Wish*** had presented a trip of a lifetime to Disneyland to the twins and we had booked more adventures around that trip as Olivia has severe scoliosis and comfortable long travel seems less likely in the long term. Then we heard whispers of Covid hitting Italy. It appeared to be a more lethal version of RVS (this had left Olivia fighting hard in ICU for months and Orin on oxygen in HDU in the past). Therefore, we made the decision to impose our own strict lockdown (no one in or out, everything coming in was sterilised etc) until we could get the measure of this virus. Thankfully we had a metabolic team

who fully agreed with that status and supported us every step of the way. For parts of lockdown, we also had our amazing team of carers whom we employ directly though SDS and had adapted wonderfully and supportively to full PPE as part of everyday life (FFP3 masks, visors etc). This meant we could have support with the twins and make everyday routine so much easier to achieve. Unfortunately, there were many times when it wasn't safe to have them in due to the high-risk level and new strains emerging.

Our lockdown is coming to an end just now after two years of strict shielding. For many their experience of lockdown was difficult and affected their mental health quite badly. Our experience has been very different. Having complex children, we knew we were going to be in for the long haul and actively decided to view this as an opportunity rather than a sentence. We set up a full classroom (luckily for us mum is a teacher) and ran as normal a daily routine as possible. This involved wakeup, carers, breakfasts, school, family play time, dinner, more play time, bedtime. We are trained to do enzymes at home, so enzymes day is an all-day play day. We also continued to maintain a fully plant-based diet (no meat, eggs or dairy), so finding balance in a high protein diet when you cannot go to the shops for yourself definitely kept things interesting. Strangely all being together 24/7 only brought us closer together, and has given us such amazing, dedicated family time. To many people the idea of closing off from everything may seem like wasted years, however the kids have progressed beyond everyone's wildest dreams (educationally, medically, and physically) with such uninterrupted one to one time.

We have been lucky to live in an era of technology to make virtual appointments possible, allowing us to keep connected to all the medical teams. We were also fortunate enough to have Granny, Grandad and friends coming to the garden to chat to us from a distance.

When we first heard about Covid vaccines, the excitement was unbelievable but short lived. The initial announcement that there was no intention to vaccinate children under 12 was terrifying. This coupled with talk that the twins' complexities potentially meant they may never be vaccinated. For us this was not a viable option. With all the data suggesting it should be safe and our metabolic team agreeing the rewards outweighed the risks, we fought hard and loud to ensure we were not abandoned for simplicity or political reasons. Finally, just before Christmas 2021 the long-awaited call came, the twins would be vaccinated, the first in the UK under 12! This held as much fear as relief, it's never fun to go first and with limited information on the vaccine's potential impact on Pompe disease it made the decision process unenviable, but that's nothing new to parents of children with rare diseases. The twins (and us) have now been fully vaccinated and thankfully not a single side-effect to date. We decided to do a lot of interviews (TV, radio, papers) once the twins had their first dose as we were hearing from families who were not getting

this option and we hoped such public attention would help them get the same opportunity we had.

Finally, we have begun to plan to live our lives alongside Covid. We have planned a slow and steady exit in coordination with our metabolic team as we are aware of the twins weakened immune systems (as we have had no bugs in 2 years). However, big plans are in place and a summer of fun awaits. The twins are hoping ***Make A Wish*** can still do ***Disneyland*** this year and that they will see their Granny back in Ireland.

As for my wife and I, we will settle for five minutes peace and quiet.

Accelerating Medicines Partnership - Bespoke Gene Therapy Consortium

The *Accelerating Medicines Partnership® (AMP) Bespoke Gene Therapy Consortium* brings together partners from the public, private, and non-profit sectors, to foster development of gene therapies intended to treat rare genetic diseases, which affect populations too small for viable commercial development.

The Problem: With the current commercial drug development model, companies cannot recover the costs required to develop gene therapies to treat rare and ultra-rare genetic diseases, because these diseases affect relatively few patients.

The Solution: This project will create tools to streamline the gene therapy development process, aiming to reduce associated costs and encourage companies to pursue gene therapies for rare genetic diseases.

Building on the successful AMP model, this program focuses on generating a standard playbook for developing such gene therapies. This will be established and piloted using four-to-six clinical trial test cases and will include streamlined templates, master regulatory files, and uniform manufacturing processes to create a pathway toward the commercial viability and sustainability of gene therapies for very rare diseases. This approach could have substantial positive impacts on the larger gene therapy field, especially as the field moves into the era of genome editing.

Goals

- Make adeno-associated virus (AAV) technology more accessible to a broader range of diseases
- Accelerate the potential to streamline preclinical and product testing
- Facilitate scientific and regulatory advances that will ultimately benefit the entire field
- Bring gene therapies to all individuals in need sooner

Extract from Foundation for the National Institutes of Health Website.

See the link below to read more

https://fnih.org/our-programs/AMP/BGTC

Global Registry for People with GSD1b

We all know and understand that getting researchers and pharmaceutical companies involved in finding a cure for a rare disorder is a gigantic task. For an ultra-rare disease this is doubly difficult. One of the first critical steps to doing this is having a cohort of patients and carers for researchers and pharmaceutical companies to speak to.

CureGSD1b

www.curegsd1b.org is doing an incredible job of helping to create a contact list of all people with GSD1b globally to start this. They are collecting basic contact details and email addresses to join the *Rare X* data collection platform in February 2022. They currently have one of the most extensive contact lists of GSD1bs.

They have created a short questionnaire using Google forms to help capture some very basic demographics and contact information. This data will not be shared with anyone outside the GSD1b community. The data will help with the initial rollout of the patient registry, and give a more effective way to communicate with the GSD1b community about events, research updates, etc. By filling out the form you will be added to the *CureGSD1b* organization.

Please Register

GSD1b patients and the carers of people with GSD1b should register their details.

For more information and to register, visit www.curegsd1b.org and scroll down to find the link to join.

This questionnaire will take GSD1b patients and the carers of people with GSD1b less than one minute. It will help immensely to set up the foundations for finding cures and be a great resource of information and support for GSD1b patients and carers.

The *Cure GSD1b* website is available in 22 languages.

If you have any other questions or want to get involved more yourself, please email either Jamas LaFreniere jamas.sophieshopefoundation@gmail.com or Enrique L. Contreras Pulido e.contreras.pulido@gmail.com

CURE GSD1b

Education and Achievement with GSD1a

Maryam Ahmed

Without any explanation, I tend to shy away from anything that involves talking about my condition or anything that may make me emotionally vulnerable, as I tend to be an emotionally unavailable person. I think a lot of that usually comes from hiding feelings about how one lives with a rare genetic disorder, which has followed me through adulthood.

With education I'd like to be very honest, I have encountered various types of people/teachers/assistants etc. however my rare condition was never fully understood. I feel this is because I look 'normal'. I was often perceived as normal, but deep down I know I couldn't be. The extreme tiredness, fatigue and wear down from all my appointments, meant I was more tired and exhausted than the average child/teen. Also living with my condition was never easy because all I was trying to do was to NOT become hypoglycaemic and possibly go into a coma.

A hypoglycaemic attack made me unaware of my surroundings and made me lose any concentration within that moment, becoming fidgety and irritable, and confused. In that moment I would need an emergency dextrose drip which meant rushing into hospital no matter what time of the day or night it was. Hypoglycaemic attacks occur when your glucose (sugar) level is too low, your body doesn't have enough energy to carry out its activities, leaving you floppy and unable to function very well at all.

I wanted to become a midwife from a very young age because I had a love for babies and was fascinated with the career. Unfortunately, I couldn't pick the choices to go down that route due to my energy levels and health. So, I gave up on the idea, and resorted to business and ICT. This meant I got to sit down and study, but often I would fall asleep at the desk at school.

Although the teachers and schools understood I would have a low attendance rate throughout all years, I felt like it was never a priority on the school's behalf to give me the extra needs for education. Although, in year 10 and 11 my attendance had become extremely low, down to 28%. I remember being called to a meeting together with my primary carer, which is my mum. The school requested that I come into school 2 days a week and wouldn't have to do any work. My mum explained to them that at home, I would do nothing but sleep, so they made me a quiet room with cushions and blankets so I could sleep at school. This became more draining as I was getting fidgety that it wasn't my bed, which is what I knew to be a safe place.

As you can imagine, I didn't do very well in my GCSEs. I failed all my exams, despite being given extra time. I didn't understand much as I had missed way too much schooling. I knew I wouldn't pass, so the day I got my results I wasn't surprised.

I continued into college part-time to try and keep my mind off my health. However, it wasn't great. I was doing ICT and Business resits as I hadn't done great at school, and I wasn't particularly interested in these subjects. I tried to get into a course that would mean I became a midwife, but I was refused due to my health.

In my second year of college, I attended a normal health check appointment, but this turned into sour news straight away. I found out that my kidneys were rapidly failing, and they were at a very low percentage. This meant that I now had GSD1A and kidney failure. I was also told at this point that I only had a year to live if treatments weren't started asap. As you can imagine, this was very draining. I was fully supported by the metabolic team at Salford Royal, and the renal specialist at Preston.

I quit college not out of choice but because I started throwing up blood numerous times a day and became

extremely tired and unable to get out of bed. While undertaking dialysis I had become lost, lethargic, exhausted, and extremely tired. I had awful side effects such as extreme hair loss, coming out in clumps. I didn't want to see anyone who wasn't my carer (Mum), as I just wasn't the same person, the same smiling bubbly person I was known to be. Just because I had GSD1A, I never let it affect my smile, which is something I think is mistaken for 'being fine'.

I was fortunate to be able to get a liver and kidney transplant in 2018. I had lost all hope, and wanted to give up on life, as I saw no way out. It was at this point when that miraculous phone call arrived from Leeds to say we have found a match. It took me a long while to recover and to now figure out what "normal" was. I had never been "normal", but I now had to find out what that meant for me.

In 2019, I joined a foundation course to get my UCAS points, as the dream of being a midwife still existed. I managed to complete the part time course and was fortunate to get a place in Bolton University to study a midwifery degree in 2020. Due to the pandemic, I was advised to defer a year and to take time out as I was clinically vulnerable. I restarted in January 2022, as a student to become a midwife!

Once I qualify, I intend on going into genetic counselling, something I have discovered through fighting my own rare battle; to be there for others going through similar situations or even other conditions. I hope I can make this next dream come true! Despite my illness still having an effect on me, I aim to fulfil my dreams.

To be where I am now, I owe a massive thanks and a lifetime of appreciation to my mother who has cared for me throughout my life and has supported my dreams and goals. If it wasn't for my mother, I wouldn't be alive right now to write this.

Adaptations in the Home

Elizabeth Davenport

AGSD-UK Specialist Care Advisor

Adaptations in the home are there to make lives easier for everyone. I do hope that the following information about starting the process is useful and please do contact me if you have any questions.

Changes to your home can be small or big and subject to eligibility could possibly include:

- Fitting a lift, stair lift or a banister on the stairs
- Adding a bath lift, walk-in shower, or a rail you hold to pull yourself out of the bath (grab rail)
- Widening doorways
- Lowering kitchen worktops
- Adding an adapted bedroom
- Putting in an outdoor ramp or step rail
- Security, such as outside lights and intercom systems or household equipment and gadgets

How to get a home assessment?

Your local council offers a service that assesses your home and recommends changes to help. Either speak to your housing association or your occupational therapist for a free home assessment.

Do I pay for adaptations?

All local authorities have Disabled Facilities Grant (DFG) schemes. These are for adaptations that cost over £1,000. Adults and children can be eligible for a DFG, but they are means-tested for adults (based on your savings or income). Please remember that you cannot get a DFG for work that has already started.

Most local authorities also have a budget to pay for small changes to your home that cost under £1,000. These are not means-tested. Housing associations manage adaptations in different ways. Ask your housing association what their policy is. Some local charities can pay for some adaptations if you cannot get support from your local authority.

To discuss your options further, please contact your local authority. If you have any questions, then please let me know. Thanks, Elizabeth

elizabeth.
davenport@
agsd.org.uk

My Experience as a Mother with a Child who has Pompe Disease

Olivia Osabutey Ayor

As a mum expecting her first child, I was in full anticipation of having my baby in my hands safely on the due date. I took my antenatal appointments seriously and made sure I attended all of them. My ultrasound scan never showed any sign that my baby was going to have a problem.

On the 9th of March 2021 I had my baby, Samuella, through caesarean section and was told that she couldn't breathe properly. She was admitted to the neonatal unit and was in an incubator with oxygen for a few days. She was then later taken out of the incubator but still needed to be on oxygen in the unit until just before our discharge which was close to 3 weeks. She had been in the hospital from birth until this point. During her stay in the hospital a lot of tests were carried out.

We were told that our daughter's heart was enlarged, so they had to run more tests to find out the cause.

We were later referred to *Manchester Children's Hospital* for diagnosis. There we were told that Samuella had Infantile Onset Pompe Disease. I was really in shock and confused about what was going to happen to my beautiful daughter. I cried my heart out at the hospital and I was consoled by the doctor and the nurse, and they assured me of the treatment that is currently on offer to make her feel better and be able to live.

She was quickly admitted so she could start her treatment. At the hospital I spoke with a psychologist who counselled me few times. The nurses were always really helpful, and they kept consoling and assuring me that my daughter will get better. I decided to be strong for my daughter Samuella.

The nurses at the hospital introduced me to AGSD-UK who came in to help us get the financial support we needed. Zainib and Elizabeth from AGSD-UK introduced me to Disability Living Allowance (DLA). Zainib really did a great job by taking her time to take me through filling in the form. It was really hectic, but she was patient with me throughout.

Caring for a child with a health condition is not easy at all. I was aware that you have to be so careful when looking after them, so that you don't cause them any more problems or aggravate their condition. It is stressful but with time it becomes a routine, so you get used to it. Her way of feeding through the NG tube alone is very tiring but I have

a very supportive husband who assists me whenever he can since he is the only family I have here. A lot of support has been given to help encourage us, along with financial help like the DLA that comes in every month and the child benefit that comes every week.

I'm very grateful to God Almighty who knows his thoughts for us made it possible to come in to contact with amazing doctors, nurses, and support organisations who are there for the family when needed.

Thank you.

Olivia Osabutey Ayor

Give as you Live & Amazon Smile

Did you know that there is a free and super easy way to help AGSD-UK support patients and carers? ***Give as you Live*** is a project that exists to help raise money for charities, whenever you shop online. This is a great way to raise money for AGSD-UK which helps us support people with GSD.

How does it work? With each of your online purchases at thousands of online shops, these big brands such as Tesco, Sainsbury's, Argos, Screwfix and hundreds more, will give AGSD-UK a free percentage as donations for all the everyday online spending that you do. It costs you absolutely nothing and is an excellent way to help us!

Visit www.giveasyoulive.com to find out more. You can also visit our website (click the ***help us*** tab, then fundraise for us) to see our Social Media Manager, Lisa Massimo, show you how to set up an account.

Please tell your friends and family to sign up for free as it's a tremendous win-win situation for everyone. If you would like to get involved visit www.giveasyoulive.com/join/agsd-uk

Amazon also has their own project smile.amazon.co.uk

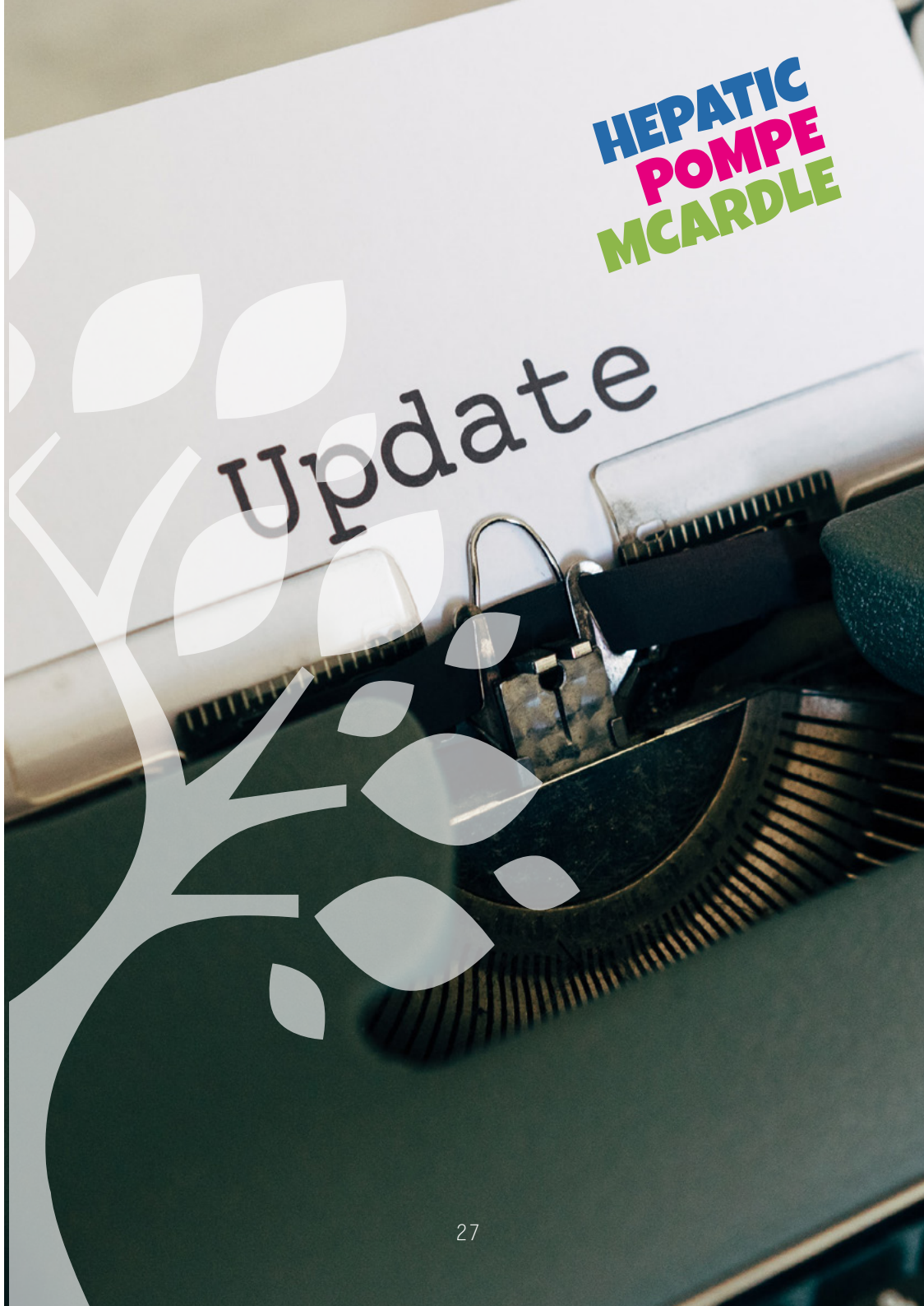
AmazonSmile is a separate portal from Amazon's normal site. It offers the same items, prices, and benefits, but Amazon's Foundation contributes 0.5% of the price of eligible purchases to AGSD-UK.

There is no cost to you or us to designate AGSD-UK as your charity to support whenever you purchase something. For Amazon users, this is a great way to help us out and know you are doing some good. Spend 5 minutes setting it up and from then on there is no extra work or cost to you, and AGSD-UK gains the benefit.

Whatever you decide, just know you will be helping to make a difference to people, and families affected by GSD and it's easy and free!

Give as you Live® Donate

amazonsmile
You shop. Amazon gives.



Hepatic Research & Articles

Ailsa Arthur

GSD3 The Search for Better Treatment

To date there is no cure for GSD3, and current treatments are mostly based on diet and management of symptoms.

There currently appear to be at least five groups around the world working on treatment and therapies for the disease. The AGSD-UK supports all these initiatives and actively encourages collaboration between researchers and patient associations to further the understanding of the needs of the people these treatments are being designed for.

Giuseppe Ronzitti and a French team (supported by Genethon, the "Association Française centre la Myopathie", the "Association Francophone des Glycogénoses", and the National Research Agency) are looking at options to deliver gene therapy. The AGL gene (affected in GSD3) is large, consequently presenting a challenge for the traditional method of delivery of gene therapy with an AAV vector (a virus that will carry the gene in the body). They have published a paper showing use of dual overlapping AAV vectors. With this method, they have demonstrated the possibility, in a mouse model, of correction of GSD3.

Duke University in the USA has had positive findings from an approach testing the delivery of gene therapy with a bacterial vector, ***Pullulanase*** in mice.

They showed that adeno-associated virus-mediated gene therapy with Pullulanase cleared abnormal glycogen accumulation in the liver and muscles and improved liver and muscle functions in GSD IIIa mice.

They concluded that their results suggest that Pullulanase is a therapeutic candidate for GSD III gene therapy with AAV.

Duke University has also recently published a retrospective longitudinal study and comprehensive review of adults with GSD3. They concluded that despite dietary changes, liver and muscle disease progression is evident in adults with 3a, muscle weakness being the major cause of illness. They also have a Facebook page for their GSD3 Clinical and Research Program.

Ultragenyx is looking at an approach using mRNA. Clinical trials have begun in the States and are scheduled to begin in Europe. They will last through to the end of 2022/2023. It is hoped to be effective at treating hypoglycaemia with the effect on the muscles yet to be determined.

Dr Rosella Franconi, in Italy, is part of a team that has been developing a synthetic gene, and is currently testing its effectiveness on an animal model. This has been financed jointly by the ENEA (The Italian National Agency for new Technologies) , the Italian GSD patient association (AIG) and IGEA SPA of Carpi (Modena).

Dicerna Pharmaceuticals have been looking at silencing the gene GYS2. This has been shown to prevent glycogen accumulation in the liver of a GSD3 mouse and reduces the fatty liver of a GSD1 mouse.

You can find all the links to read more on this research on our website. Visit www.agsd.org.uk, go to the GSDs tab, hover over 'Cori (GSD3)' and click on 'Research and Development'.

POMPE RESEARCH & ARTICLES

Luke Fraser

Gene Therapy Update

UK researchers reported the dosing of a patient at the *John Walton Muscular Dystrophy Research Centre*, run jointly by *Newcastle Upon Tyne Hospital* and *Newcastle University*, as part of the *FORTIS* clinical trial.

This marks the first adult patient in Europe to receive a neuromuscular gene therapy treatment.

The therapy - AT845 - delivers a healthy working copy of the acid alpha-glucosidase (GAA) gene directly to muscle cells. It has been developed by *Astellas Gene Therapies*.

It uses an adeno-associated virus called the AAV8 vector to deliver the working gene copy to target cells within skeletal and cardiac muscles.

Professor Jordi Diaz-Manera commented; "The enrollment of the first European patient in this Phase 1/2 gene therapy programme is a great achievement for the Pompe community and we are very pleased to contribute to innovative research in the pursuit of future therapies".

FORTIS is expected to fully conclude in January 2027.

Second Generation ERT Improves Lung Function and Mobility in LOPD

Long-term treatment with the second generation ERT avalglucosidase alfa has been shown to safely and effectively, improve lung function and walking ability in children and adults with LOPD, according to recently published data from the Phase 3 COMET trial.

These findings, along with positive long-term results from the Phase 2 Mini-COMET study in children and adolescents with IOPD, were presented by *Sanofi Genzyme* at the 2022 annual *WORLD Symposium*.

Lung function was measured via Forced Vital Capacity (FVC) and mobility via the 6-minute walk test. Both improved to some degree although the increase in lung function is not statistically significant.

A total of 95 participants completed the one-year treatment period and entered the study's open-label extension phase, where all will receive avalglucosidase alfa for 4.5 years.

Avalglucosidase alfa was designed to improve the GAA enzyme's delivery to lysosomes. Preclinical studies showed it has a 15 times higher cellular uptake than Myozyme and that it results in comparable glycogen clearance with a 5x lower dose.

Long-term ERT Use Can Help Older LOPD Patients

Long-term treatment with *Myozyme* results in variable but satisfactory motor and respiratory outcomes among older adults recently diagnosed with late-onset Pompe disease (LOPD), a study from Germany has concluded.

The research - *Long-Term Effects of Enzyme Replacement Therapy in an Elderly Cohort of Late-Onset Pompe Disease* - was published in *Neuromuscular Disorders*.

It included six LOPD patients diagnosed at a median age of 63 who had been on ERT for at least seven years.

Walking ability, muscle function, and lung capacity were measured at baseline and at regular intervals over the course of 8 to 12 years.

Findings showed that walking ability generally improved over time, with four of six showing an overall improvement.

Muscle strength and lung capacity as measured by measured by forced vital capacity (FVC) generally declined in the long term. However, these motor and lung function data may be difficult to interpret because little is known about how aging affects these aspects in people without LOPD.

The researcher's findings were similar to studies of *Myozyme* in younger patient groups, suggesting that starting ERT in LOPD patients after age 50 is both justified and beneficial.

LOPD Patients Show No Major Cognitive Deficits

Patients with LOPD do not show significant brain abnormalities or general cognitive impairment, in contrast with some patients with the classical infantile-onset form, a study has shown.

The research - *Is the Brain Involved in Patients with Late-Onset Pompe Disease?* was published in the *Journal of Inherited Metabolic Disease*.

Whether brain involvement is confined to classical infantile-onset Pompe disease (IOPD) remains controversial. While some studies have reported brain abnormalities in LOPD, others argue that such observations are age-related and similar to those seen in age-matched unaffected people.

To clarify, a team of researchers at *Erasmus University Medical Centre* in Rotterdam analysed the brain structure, vasculature, and cognitive function in 19 children and adults with LOPD, all younger than 60.

No brain lesions were found, whilst in terms of cognitive function. The team wrote most patients 'performed within the normal range on measures of general intelligence or processing speed - domains that are commonly affected in classic-infantile patients'.

Increased ERT Dosing May Help Infantile-Onset Pompe Patients

Patients with classic IOPD could benefit from increased dosing of *Myozyme* in terms of both survival and walking ability, according to a new study.

The research - *Effects of Alglucosidase Alfa Dosage on Survival and Walking Ability in Patients with Classic Infantile Pompe Disease* - was published in *The Lancet Child and Adolescent Health*.

A total of 124 patients were followed, receiving doses of either 20mg/kg per fortnight (standard) or per week (intermediate), or 40mg/kg per fortnight (intermediate) or per week (high). Some patients had their dosing either increased or decreased during the study period.

Among those patients who received ERT at a constant dosage, 16 out of 31 on the standard dosage, 12 out of 15 on an intermediate dosage, and 16 out of 18 on the high dosage were alive at the last follow-up.

Results showed a significant difference in the five-year survival rate between the standard and high dosages. Overall survival was significantly longer in those given the high dosage as compared with patients given the standard dosage. No such difference was found with the intermediate dosage.

For the group on variable dosage regimens, 20 of 28 treated with the increased dosage, 12 of 15 treated with variable dosages, and three of five treated with decreased dosage were alive at the last follow-up.

Survival was highest for patients started on the high dosage and lowest for participants started on the standard dosage, with a similar pattern observed when comparing survival for final dosages.

The authors concluded "on the basis of our results, we suggest that the current standard recommended dosage of Alglucosidase alfa in patients with classic infantile Pompe disease should be reconsidered."

Gene Therapy Potentially Better Than ERT for Pompe

Gene therapy given with chaperones was found to enhance the availability of GAA in a mouse model, according to a recently published study.

The treatment was shown to subsequently ease disease symptoms, with the mice showing significantly improved muscle strength compared with both untreated animals and those given standard ERT.

The study - *Hepatic Expression of GAA Results in Enhanced Enzyme Bioavailability in Mice and Non-Human Primates* - was published in the journal *Nature Communications*. It was conducted by a team including researchers from *Spark Therapeutics* and *Genethon*.

The mice were treated with the standard ERT dose of 20 mg/kg every two weeks for four months, or with a single injection of gene therapy at a low, intermediate, or high dose.

By the end of the study, the GAA enzyme was hardly detectable in the ERT-treated mice. However, GAA was still found in circulation in all mice treated with the gene therapy.

Mice treated with the two higher doses of gene therapy had higher levels of GAA in heart, skeletal muscles, brain and spinal cord compared with ERT-treated mice. Their

muscle strength significantly improved compared with ERT-treated and untreated animals.

The team found a dose-dependent reduction in glycogen content in the brain and spinal cord of mice treated with gene therapy compared with controls, unlike ERT-treated mice. Animals receiving gene therapy showed complete normalisation of glycogen levels in the heart, diaphragm, and arm muscles. Those on ERT showed such glycogen level reduction in the heart only.

The team also investigated the effect of combining chaperones with gene therapy. Two chaperones were tested, 1-Deoxynojirimycin, also called duvoglustat, and ambroxol. Both compounds had been reported to improve GAA activity.

Using the two chaperones in ERT-treated mice resulted in higher GAA activity in the blood compared with ERT alone.

Mice treated with gene therapy and chaperones had increased GAA in several muscle groups and in the spinal cord. Glycogen clearance also was improved in most muscles, in addition to the increased muscle strength shown.

Antioxidants Help ERT Efficacy in Pompe

Antioxidants can increase the effectiveness of ERT in combating oxidative stress - the damage in cells or tissues by reactive oxygen molecules, according to a study in cells and mice.

The findings also showed that enhancing autophagy - the destruction of damaged or redundant components within cells - reduces oxidative stress and increases ERT efficacy.

The study - ***Correction of Oxidative Stress Enhances Enzyme Replacement Therapy in Pompe Disease*** - was published in the journal ***EMBO Molecular Medicine***.

Researchers used a Pompe mouse model to measure oxidative stress within skeletal muscles, heart, and liver.

Results showed higher levels of reactive oxygen molecules and of oxidative damage in lipids within the muscles of these mice, in comparison with controls.

To investigate whether increased oxidative stress would affect GAA activity correction by ERT, the treatment was administered into Pompe fibroblasts. The team found that more oxidative stress in cells led to less efficient correction of GAA activity.

Using antioxidants was shown to reduce cellular stress and - when given in combination with ERT - to improve correction of GAA activity compared with ERT alone. The best-performing antioxidant was N-acetylcysteine.

The team concluded; "As antioxidants drugs are approved for human therapy and show in general good safety profiles, their use would represent a convenient and safe option as adjunctive treatments for Pompe".

The above summarises articles by the following authors: Marta Figueiredo PhD, Lindsey Shapiro PhD, Yedida Y Bogachkov PhD, Vanda Pinto PhD.

WALKING IN WILTSHIRE, JUNE 2022

Andrew Wakelin, McArdle's Coordinator

David Thompson has organised a three day weekend of ***Walking in Wiltshire*** over 17-19 June, and anyone with McArdle's is very welcome to join in for part or all of it. It is a chance to meet other people with McArdle's, to hear their stories and to swap notes. There will of course also be lots of learning points. The walks will be relatively easy and there will be plenty of pick-up points for those who wish to opt-out before others. Much of the walking will be riverside and canal-side. To keep things Covid-safe, even if the pandemic gains a bit of momentum again, the idea is to meet outdoors for the walks, and everyone book their own B&B accommodation (people living locally may just come each day from home) – rather than all being together 24 hours a day. David has made a video about the event which was included in our ***Winter Season***. You can find the video there in the McArdle's playlist. Please do drop a line to David if you are interested in joining us.

Youtube: www.youtube.com/user/GSDScreen
Email: walking.events@agsd.org.uk

CLINICAL PRACTICE GUIDELINES FOR McARDLE'S AND TARUI'S PUBLISHED!

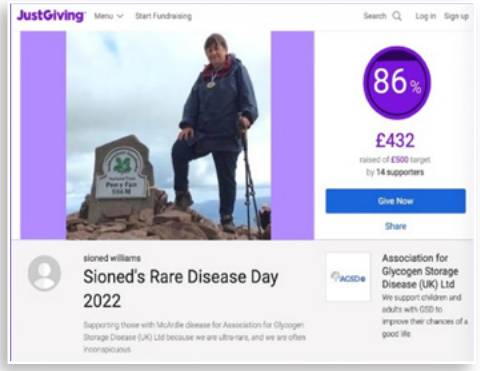
After about two years of intense effort by an international team of clinicians, researchers, and patient representatives, led by IamGSD President, Stacey Reason, we have finally achieved publication of ***Clinical Practice Guidelines for GSD5 (McArdle's) and GSD7 (Tarui's)***. It is a huge landmark as there had been two previous attempts over about the last 20 years and neither managed to complete the task. The paper of 13 pages appeared in ***Neuromuscular Disorders***, which is the official journal of the ***World Muscle Society***. The paper is accompanied by 18 pages of supplementary material linked to the paper, which are available for download online. The guidelines now contain a very considerable amount of consensus about diagnosis and management of these conditions. An apparently small but very important development is the change of terminology from "exercise intolerance" to "physical activity intolerance". This is already helping medical professionals to better understand the impact of these conditions on ADLs (activities of daily living).

IamGSD is very grateful to ***Reneo Pharmaceuticals*** for financial support enabling the paper to be available under ***Open Access***, so that both medical professionals and patients around the world may download the paper for guidance, free of charge. Immediately after publication we were made aware of patients who were able to take a printed copy of the guidelines to a medical appointment and feedback from the medical professionals was extremely positive.

When IamGSD was established one of its objectives was to "disseminate standards and best practice". Publication of the clinical practice guidelines is the defining step in this objective, but now we need to work on the dissemination so that all relevant clinicians are aware of these guidelines and adopt them.

Sioned's Rare Disease Day 2022

Rare Disease Day is on the last day of February every year and is celebrated all around the world. This year our own Sioned Williams made an impassioned video appeal on behalf of the McArdle's funds within AGSD-UK. In the YouTube video she not only 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 Principal Harpist with the *BBC Symphony Orchestra*. There are a number of serious and amusing anecdotes about achieving that career, with McArdle's.



The video is available on YouTube but has also been widely shared online in many different pages and groups. Sioned says she has gained so much from the expertise of the McArdle's clinic in London and the publications originated by AGSD-UK. She has joined the walking courses three times in her native Wales (in Snowdonia, Pembrokeshire and the Brecon Beacons). Her *JustGiving* page on behalf of AGSD-UK is still available for all those who would like to help to support our work.

Video: youtu.be/JfSXU2snaDc
JustGiving:
justgiving.com/sioned-williams28

Winter Season 2022 Success

In the *Winter Season 2022* we put out 10 videos in the McArdle's playlist, plus a very short introductory video and one presentation was split into two parts, so there were 12 in all. This compares to 21 videos in the previous *Winter Season*, which was actually run for three months instead of the one month this year. At the time of going to press this year's videos have had almost the same 1500 viewings which were achieved the previous season. So, for this season it is an average of about 125 viewings per video, the actual numbers ranging from about 60 to 200. It is interesting to compare this level of exposure with what is usually an attendance of about 15 people in the McArdle's group at an Annual Conference. Does this suggest that our efforts are better put in this direction even after the Covid pandemic is behind us?

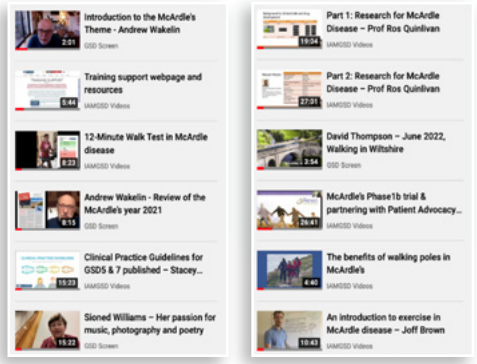
The videos were hosted between the *AGSD-UK YouTube* channel, and the *IamGSD* channel. They will nearly all remain available in the long term as only a few were date-specific. So, if you missed any, go back to the *McArdle's Winter Season 2022* playlist, and catch up with the ones that you have not seen.

Please give me feedback on type5@agsd.org.uk so that we know what you value and what you want in future. In case we do a *Winter Season* again next winter, do please also get in touch if you would like to make a presentation yourself.

McArdle's Zoom Get-Togethers

This season we had an online Get-Together each Sunday in January, so 5 in total. One was reserved for children and their parents, but the others were open to one and all. The message had really got out by the last Sunday, and we had a very good attendance.

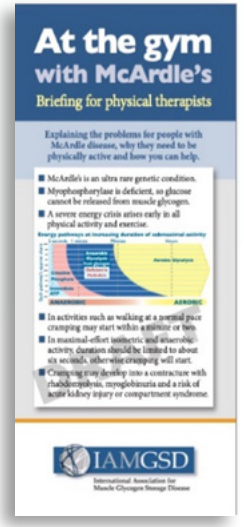
It is a great way of meeting a range of other people with McArdle's and swapping notes. We ran an additional one for children and parents in March. If there is the interest, we could organise one of these for adults each month, year round.



New IAMGSD Leaflet "At the Gym"

IamGSD already offers a successful range of leaflets, including *At Home*, *At Work* and *At School with McArdle's*. Now they are adding a new one *At the Gym with McArdle's*. It is intended for physiotherapists and personal trainers who are supporting people with McArdle disease in their efforts to exercise more and improve their condition. It is not a replacement for the expert guidance available from the McArdle's clinic in London, but for those in the UK who cannot attend the clinic, and of course for people in the many locations around the world where there is no expert support available.

The leaflet is the public facing part of a greater depth of support available on the *IamGSD* website on the *Medical* menu under *Training Support*. This offers details of aerobic and strength training and the method of doing the *12-Minute Walk Test* to confirm second wind and to monitor progress. There are also links to some useful videos explaining different aspects.



As we go to press, the leaflet is still in draft form. Feedback is invited and then any adjustments will be made before the final version is produced.

Personal Stories of McArdle's

Since the last edition of *Glisten* three more personal stories from people with McArdle's have been published on the IamGSD website. There might even be another one by the time you read this. These are really very varied stories with some interesting twists and turns. The people concerned are aged from 12 to 66, male and female. The stories are often entertaining, informative and even educational. At the very least they give you an understanding of how much we have in common, even though in some ways we are very different.

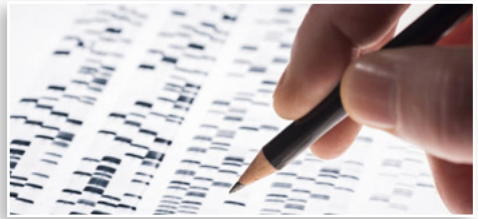
Feike Dirks
Netherlands
Age 66
GSD5 McArdle's
Read

Aidan Fernandez
USA
Age 12
GSD5 McArdle's
Read

Melanie Graves
USA
Age 54
GSD5 McArdle's
Read

Diagnosed at Age Zero

There's been a recent interesting development in the US. As background, a survey in 2000 found that almost 70% of people diagnosed with McArdle's had been first taken to the family doctor by the age of 5, and almost 90% by the age of 10, yet diagnosis wasn't achieved for an average of about 25 years. *IamGSD* has an objective of getting the average age of diagnosis to below age 10, as this is found to be highly beneficial.



The new development is that there has been an instance of a diagnosis being achieved at age 0! A young couple expecting a baby were concerned about possible genetic conditions that are prevalent in their community and so sought genetic testing. It was recommended that rather than testing individual genes the embryo should be tested with Whole Exome Sequencing. Unexpectedly, this came up with a diagnosis of McArdle disease and the couple contacted *IamGSD*.

We know of just one other diagnosis at an extremely young age. An expectant couple were interviewed in hospital about any genetic conditions in their family and were offered genetic testing through a research project. The testing was undertaken at the time of the usual newborn screening, and McArdle disease was found.

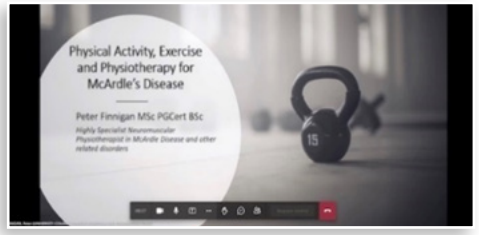
It is intriguing to think that maybe one day McArdle's could be included in routine newborn screening for all babies. Not that we would necessarily want it. No doubt there would have to be quite a lot of ethical debate before that might happen.

McArdle Clinic Webinars

Starting in autumn 2021 the McArdle's clinic has instituted webinars in place of the group "management clinics" which, pre-pandemic, were offered to interested patients and those with particular needs.

The second webinar was held on 2nd March 2022, on Exercise and Physiotherapy, presented by Peter Finnegan, Highly Specialist Neuromuscular Physiotherapist. It included about a 45 minute presentation and demonstration, followed by 30 minutes of a question-and-answer session.

The next webinar will be on diet and will be held on Wednesday 1st June at 10am. If you would be interested to attend, please contact Maria Patasin, Clinical Nurse Specialist on marialuvimin.patasin@nhs.net.



The McArdle's Experience
and Children & Parents Event

These events are back following two years of cancellations due to the Covid pandemic. We are offering both the adult course and the Children & Parents event. They are being run at the National Trust's Stackpole Centre in Pembrokeshire, which we have very successfully used several times before. SThey are based in two adjacent accommodation units and have the potential for two days of activity overlap, so that the children can meet adults with McArdle's.

The dates of the two courses will be:

The McArdle's Experience: 30 July - 6 Aug

Children & Parents event: 29 July to 2 Aug

Full details and a link for bookings will be on the website by the time this issue of Glisten is distributed.

We have three YouTube videos about our walking courses, on these two channels:

IamGSD Videos: *Walking with McArdle's* and *Living well with McArdle's*

GSD Screen: *Celebrating 10 years of the walking courses.*

Thank you for continued support from the Industry, Foundations and Trusts that help AGSD-UK. These grants allow us to work on projects, ensure our members at AGSD-UK have access to a high level of support and to run events and contribute to research in GSD.

Thank you for the continued support from Amicus, Astellas Audentes, Avro Bio, Edward Gostling Foundation, FMR Global Health, FYMCA, Sangamo, Sanofi, Spark Therapeutics, Ultragenyx and Vitaflo.

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ultragenyx pharmaceutical

Vitaflo Innovation in Nutrition UK

Take away from this Glisten...

- Share your top three things that would improve life with the condition, to help shape the future of AGSD-UK
- Let us know what you want funders, policy makers, professionals, and the public to know about living with a GSD
- Get in touch if you're interested in getting involved in discussions around fundraising, publications, or events
- Register your interest now for an Autumn conference, by contacting **info@agsd.org.uk**

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