Autumn 2023

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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Phone: 0300 123 2790
Email: Info@agsd.org.uk
Charity Number :1132271

Thank you!

New fundraising opportunities

Know anyone who might be keen to take part in cycling or running events on behalf of AGSD-UK in 2024?

Please contact info@agsd.org.uk so we can get your place secured!

Plus let us know if you're interested in fun and simple activities to raise funds and awareness or have ideas you want to share...

Front Cover Images:

Photos from our Main Event in May this year

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 2023 edition of Glisten.

With this first edition of *Glisten* since our *Main Event* it's great to have the chance to look back on the moments captured by the lovely cover photos and on the wonderful feedback received.

Some of the highlights included the powerful stories from community members, Sioned Williams' brilliant harp recital, the lively percussion workshop, the relaxing art session and the hilarious raffle with great prizes –plus of course the presentations from leading experts on latest treatments and research. Most of all, people appreciated the chance to get together to share experiences with friends old and new and it was such a pleasure to meet so many of you there.

We're really grateful for the support of all our presenters, volunteers, staff and sponsors who helped make the weekend such a success.

We're already starting to plan next year's event and by popular demand are planning to return later in the year to the same venue in Loughborough, where they looked after us so well.

Meanwhile there are more chances to get together virtually over the next couple of months with our festive end of year event, living well socials and other presentations – see back cover for further details and just let us know what other activities you'd like to see included!

Val Buxton

Fundraising Round-up

Thank you to our members that have been busy fundraising over the past year

Rob Hill and his brother in law Steve ran the Bath Half this October and have raised over £5000.

In the summer of 2022, the family spent nearly 6 weeks in hospital with Oscar and he was ultimately diagnosed with glycogen storage disease, type 1A.

Oscar has a very meticulous feeding regime, with him having to be on continuous tube feeds for the majority of the day and night. As you can imagine having a toddler connected to tubing most of the time is incredibly challenging! He has had three operations one of which was a gastronomy meaning he no longer has the NG tube. He also now has a continuous glucose monitor which allows us to see his blood sugar via an app which is a huge help.

Soon after Oscar's diagnosis I knew I wanted to raise money to help the charities that have helped us and support people with a similar condition to Oscar. I signed up to do the Bath half marathon early in 2023 with the thought of spreading the training out throughout the year. However as getting to grips with Oscar's management took most of our time, I ended up cramming most of my training into the few months before the race.

The race itself went really well. My official time was 1:52:04 which was well under my 2 hour target. The atmosphere was incredible and definitely helped me

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FUNDRAISING ROUND-UP cont...

along. We have had amazing support. Currently between my brother in law (who also ran the bath half) and I, we have raised over **£5000!**

www.justgiving.com/fundraising/robert-hill30

www.justgiving.com/fundraising/tinnospage

June Bruce A big thank you to June who held a birthday fundraiser on Facebook and raised **£666**

Lee Jones has been raising money for the charity by dying his hair green!

London Marathon April 2023

You may be aware we had a number of runners participating in the London Marathon this year.

Huge thanks to Kirsty White, Dale and Lauren Esliger, Liam Coote and Lisa Nugent who collectively raised an impressive **£8766** for the charity!

THE MAIN EVENT

Our *Main Event* in May attracted over 100 people affected by a range of GSDs over the course of the weekend.

The programme was developed with people living with GSDs and was organised around our three goals:

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

The weekend was designed to include opportunities for socialising and sharing experiences with others, hearing about latest research and developments, trying out new therapeutic activities and talking about key topics relating to life with the condition.

The evaluation forms completed showed that:

- **98%** said the event enabled them to connect with others with similar experiences
- **98%** said it gave them the chance to gain tips about living with GSDs
- **96%** said it allowed them to become better informed about research and developments in the field of GSDs
- **95%** said it provided the opportunity to get information about support and services that are available
- **100%** said they had fun and enjoyed themselves.

The Main Event...Reviews

We were overwhelmed by the amount of positive feedback received - here are just a few typical examples:

Really loved the whole event...Content of the programmes was spot on. Nice to meet with Pompe people and with mixed group GSDs...Loved the drums and the art.

100% valuable. Well organised. Would love an event in this place again.

Spot on excellent. Love to come again. Loved the raffle experience. Well organised. 100%

Fabulous weekend! ...Loved the whole conference. Great speakers and workshops.

Great organisation.

Informative programme and social opportunity to connect with others.

Venue and food spot on.

Learned more about my disease today than I ever have before.

Our first one, loved every minute. Thank you! We'll be back next year. I've learned more today than when my son was diagnosed.

It was a well organised, very informative event. Learnt more today than in the previous 50+ years since initial diagnosis.

Patient stories were really brill- great to see their individual experiences. Thanks!

I thoroughly enjoyed the whole event. Good mix of informative, practical and what we needed. Great to meet other fellow McArdleites.

Absolutely excellent conference- enjoyed myself. Thankful for all the hard work. 10/10 (plus a nice little picture of a flower!)

It was great to hear from the experts!

Excellent venue and meeting. Thank you!

Very well organised and presented.

It was great to meet more people this time and get involved in more activities.

Many thanks.

A really important event that should continue.

The organisers did a great job.

Thank you.

Very good meeting. Well organised.

Lovely to meet and talk with everyone again. Wonderful update and insight into the gene therapy trials.

An enjoyable weekend and very productive. Thanks to the sponsors and organisers. The music programme (i.e. the harp concert) was one of the greatest highlights.

Great to listen to the guys at the start with their experiences.

Overall found the day very wonderful and looking forward to the next.

Good forum for networking. Good to be back in person!

It was a very positive weekend. Informative and fun.

As a grandparent of a child with GSD3b, it has been a lovely opportunity to meet others and gain greater understanding.

Understanding in both lived experience and the nature of progress of the disease.

I would give it 5 stars. The venue and atmosphere added a very positive dimension to the whole weekend.

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Three McArdle's courses in Wales 2023

Another successful year, this time in North Wales, with the largest contingent ever.

This year, the walking courses were again a great success, and in fact attracted the largest number of people ever with a grand total of 42! This was made up of the *Children & Parents* and the main course for adults. But also, for the first time ever, a *Future Leaders* course to help prepare people for leading such events in the years ahead. It was held under the auspices of *IamGSD*.

The *Children & Parents* event had 9 children, ranging in age from 13 to 20, plus 9 parents. They came from England, Australia, and for the first time from Poland. We have had siblings before, but this time there was another first in that we had twins, and they came all the way from Australia. That event overlapped with the main adult course, which extended for a full week and had a total of 8 course members from England, Wales and the US. The *Future Leaders* course had a total of 10 course members from: England, Scotland, the US, France, Germany and Canada. Each course had a "support team" of 5 people, 2 with McArdle's and 3 not.

Our base was at the historic Snowdon Lodge in Tremadog, Eryri (Snowdonia). We were very well looked after by the owners, Carl and Anya. We were very lucky to be given access to their training room, complete with audiovisual facilities. It was a bit of a trip to pick people up from the mainline station at Llandudno Junction, but Tremadog's position on the west side of the National Park was very helpful considering the bad weather that we had. We several times escaped further to the west, away from the mountains, to catch the better weather.

Last year Bronte Thomas was one of the two assistant leaders, along with Dan Chambers. This year, whilst the course and walks were planned by Andrew, the walks were led by Bronte. It was a tough year for her to start, with very bad weather and quite large groups. But she did very well, developed some new procedures and clearly enjoyed it.

Following here we have three comments on the courses. The impact has been incredibly positive, sometimes in very different and unexpected ways. One person, having walked with other McArdleites, has realised that he is so different that he has returned home to get his diagnosis checked.

Bronagh Ross

Mother of 16 and 19 year olds

It was only our second time joining the *Walking in Wales* course. I cannot thank enough Andrew Wakelin, Bronte Thomas, her whole family, Sioned Williams and Ali for making this such a life-changing week. The care, planning and support all week was exemplary! I felt just as taken care of as Eoghan and Caoimhe. The walks were well planned, with alternatives in place for the horrendous weather; the walk leadership was clear and thoughtful; the food incredible and the ambience homely, fun and supportive.

Caoimhe, Bronagh and Eoghan.

I feel Caoimhe and Eoghan's confidence has grown exponentially! Not just in relation to walking but in the company of this close group of McArdleites. Caoimhe is leaving for university with increased confidence that she will cope. As a mum it warmed my heart to see my children becoming much more extrovert than I have ever seen them. These weekends/weeks are life-changing and life-affirming.

Eoghan gets a harp lesson!

Thank you too to the other attendees for their support, low brow comedy and genuinely great company - Jane, Darcy, the twins Ned and Jamie, Andy, Jodie, Lucy, Feike, Tony, Alex, Willow, Sebastian, Maja, Katie, Alex 2, Tanya, Florian, Carole, Sarah and Annabelle. New friendships made, others deepened. Maybe I have forgotten someone as it was the largest group of McArdleites yet!!

The future looks bright with this new generation of leaders coming through and this is down to the incredible groundbreaker that is Andrew! From the bottom of my heart, thank you all for an amazing week – in Tremadog, North Wales.

These weekends/weeks are life-changing and life-affirming

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Three McArdle's courses cont...

Future Leaders Course

There will be a report of the *Future Leaders* course on the IamGSD website. The future looks bright with this new generation of leaders coming through.

Some of the Future Leaders from seven countries. Left to right: Alfred, Colin, Jen, Dirk, Michael, Rachel, Stacey, Frankie, David & Richard

Discussion over the art therapy project

Time for sightseeing at Conwy Castle

Crossing the causeway on Yr Wyddfa

Starting the top ridge on Cader Idris.

Jane Harris and Darcy Duggan

Parents of twins Ned and James

This week Darcy and I had a glimpse that McArdle's can give our children more than it takes. This was profound for us. We traveled from Australia to the walk in Wales with our 16 year old twin boys. It was a plan that took eight years to execute.

The boys got to be part of a group of 16 McArdleites. They got to be teens and young people together. They got to be with older McArdleites. They got to be cared for by 'aunties' and 'uncles' who either have McArdles, or love and care for a McArdleite. They got to climb mountains they (and we) would never have attempted and do physical things that were bold and audacious. They got to be with a bunch of McArdleites out the front of the pack at the end of a long days walk, with the non-M people trailing tiredly behind.

I know it's not possible for everyone to attend this week in Wales… it nearly wasn't for us (3 year drought + 3 year pandemic + weak Australian dollar).

Bronte Thomas's leadership in North Wales was excellent, as was Andrew's mentoring, and as was the assistance on the tracks from Eoghan. Hopefully it will be possible to bring Bronte to Oz to lead a southern hemisphere experience.

The four of us on the way up Snowdon.

An easier walk past Llyn Dinas and along the river into Beddgelert

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Bronte Thomas

Walk leader

The hardest but best and most rewarding week of my life. An extremely successful McArdle's Experience 2023 in Wales has come to an end. We climbed Cader Idris and Yr Wyddfa (Snowdon) whilst also walking miles every day. On the last day I managed to accumulate 30,000 steps and over 10 miles!

Leading some of the "children", in the clouds

Going from 1 in 100,000 to 1 of 16 is one of the most positively life changing things I've experienced. The connection within the community of those who have McArdle's is a bond I've rarely experienced before. I'm incredibly proud of everyone who attended this year, for the goals they've smashed and for all they've achieved.

I would like to say a massive thank you to my biggest inspiration, Andrew Wakelin. I will cherish and keep safe your walking poles for the rest of my life, and every mountain I climb I will take them along with me. In your steps, you've helped me grow into someone I can be proud of and I feel I've found my passion and calling in life thanks to your mentoring over the last 8 years. Giving me the opportunity to lead alongside you has positively impacted my future and I hope I've made you proud.

Now time to plan for next year in the Brecon Beacons, as well as get myself incredibly fit for a potential ***Walk over Wales*** in 2026

A good path and downhill to start a walk

The McArdle's Experience courses for 2024

For 2024 we will be returning to the ***Brecon Beacons National Park*** in South Wales. Riversides, waterfalls and relatively easy mountains will feature! We have the same accommodation which we used in 2018, a converted Meeting House and cottage near ***Hay on Wye***.

Having the children's event and the main course overlap has been very successful for the last two years, but we could not find suitable accommodation to do that for 2024. The children's event (extended to a week) will come first, followed by the main adult course (5 days) in the same accommodation.

We are actually renaming the children's event, the ***Under 21s*** course, and would still expect most to bring a parent (or two!). The cut-off age will be rather flexible to accommodate such things as affected siblings.

Full details will soon be on the website, perhaps by the time you read this. For either course, all you need is to be able to get into second-wind and then we take it from there.

Reserve the dates:

Under 21s course	Monday 29th July to Monday 5th August 2024
Adults course	Monday 5th to Friday 9th August 2024

McArdleites from 7 countries having a bit of fun around an ancient oak, the last time the course was in the Brecon Beacons

NICE UPDATE

NICE recommends cipaglucosidase alfa plus miglustat as a treatment option for late onset Pompe in adults

Over the summer the *National Institute for Health and Care Excellence* published final guidance recommending cipaglucosidase alfa (CIPA) plus miglustat (now known as *Pombiliti + Opfolda*) as an option for treating late-onset Pompe disease in adults.

The recommendation means that cipaglucosidase alfa becomes a third enzyme replacement therapy option alongside alglucosidase alfa (ALGLU) and the more recent avalglucosidase alfa (AVAL).

The decision follows a committee meeting in early May which reviewed evidence from clinical trials, the biotechnology company Amicus, and an expert clinician, as well as from other stakeholders. This included powerful testimony from AGSD-UK's nominated patient expert on their experience of the condition and of the therapy. AGSD-UK had previously submitted evidence based on survey responses and interviews with community members.

The committee determined that CIPA plus miglustat is indicated both for people who have had previous ERT treatment and for people who have not had ERT.

They refer to evidence from the clinical expert at the committee meeting who explained that data for CIPA plus miglustat and for AVAL suggested a more durable treatment effect than ALGLU so it was likely that newly diagnosed people would start on AVAL or CIPA plus miglustat. The expert indicated that clinicians would look at someone holistically when considering whether to switch treatment and suggested that people currently taking ALGLU would also be switched to AVAL or CIPA plus miglustat when response to ALGLU began to wane.

The availability of different treatment options for Pompe is extremely welcome, given the varying responses to therapies among people affected.

Full information on the NICE recommendation for CIPA plus miglustat is available on the NICE website:

https://tinyurl.com/457rjczp

NAVIGATING THE LABYRINTH

Reducing delays to a rare disease diagnosis

The *Specialised Healthcare Alliance* has recently produced a report setting out the long and challenging path to diagnosis faced by many people with rare conditions.

The report looks at the barriers encountered by those affected, along with opportunities to improve the experience and achieve timely and accurate diagnosis.

The findings set out in the report will come as little surprise to members of the GSD community.

They detail that on average rare disease diagnosis takes four years, involving an average of five different doctors and three misdiagnoses. Common experiences along the way include not being listened to by healthcare professionals -who often have limited knowledge of rare conditions, delays in accessing tests and obtaining results and a lack of information and emotional support throughout the process. These delays and the limited support provided can result in significant costs in terms of the physical, emotional, and financial wellbeing of those concerned, as well as wider social and economic costs.

The report looks at the policy opportunities to improve this situation offered by the UK Rare Disease Framework and sets out a number of recommendations to deliver rapid testing, improved professional awareness and better integrated support and service models for those awaiting diagnosis.

The full report is available at:

https://tinyurl.com/t8r8p3dx

See page 35 of this edition of Glisten for update on newborn screening research.

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MINDFULNESS

Following on from the last edition of Glisten and our focus on having good mental health to help with daily challenges and stresses, we move on to Mindfulness. This is a useful tool to use when we are feeling overwhelmed and worried about our reaction to stresses and triggers.

Mindfulness is maintaining a moment-by-moment awareness of our thoughts, feelings, and sensations. When we practise mindfulness, our thoughts tune into what we are sensing in the present moment rather than revisiting the past or imagining the future. Because we stay in that moment it can help us focus on our needs and stay calm.

So how do we practise staying in the moment, being mindful of our surroundings?

The mind website www.mind.org.uk/ makes the following suggestions:

If you want to try mindfulness, you don't need any special equipment for the following exercises:

- **Mindful moving or walking**
  While exercising, try focusing on the feeling of your body moving. If you go for a mindful walk, you might notice the breeze against your skin, the feeling of your feet or hands against different textures on the ground or nearby surfaces, and the different smells around you.
- **Body scan**
  This is where you move your attention slowly through different parts of your body. Start from the top of your head and move all the way down to the end of your toes. You could focus on feelings of warmth, tension, tingling or relaxation of various parts of your body.

- **Mindful colouring and drawing**
  Rather than trying to draw something, focus on the colours and the sensation of your pencil against the paper. You could use a mindfulness colouring book or download drawings on the internet.
- **Mindful eating**
  This involves paying attention to the taste, sight, and textures of what you eat. Try this when drinking a cup of tea or coffee for example. You could focus on the temperature, how the liquid feels on your tongue, how sweet it tastes or watch the steam that it gives off.
- **Mindful meditation**
  This involves sitting quietly to focus on your breathing, thoughts, sensations in your body or things you can sense around you. Try to bring your attention back to the present if your mind starts to wander. Many people also find that yoga helps them to concentrate on their breathing and focus on the present moment.

These examples are not the only ways you can practise mindfulness. So many activities can be done mindfully. Different things work for different people, so if you don't find one exercise useful, try another. You can also try adapting them to suit you and make them easier to fit in with your daily life, such as mindfully cooking dinner or folding laundry.

From November AGSD-UK will be running virtual ***well-being socials*** hosted by our care and welfare advisors Elizabeth and Zainib.

If you'd like to join a friendly ***Zoom*** get together to share coping strategies and try different techniques to stay calm and relaxed just contact info@agsd.org.uk or get in touch directly with Elizabeth or Zainib.

The first session will be on Thursday 30th November 7.00-8.00pm.

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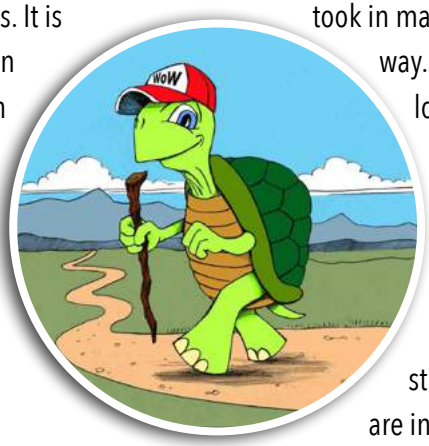
muscle GSD news

International courses

Following the ***Future Leaders*** course in Wales at the end of July, there are developing plans for courses around the world. In 2024 there will be another course in Germany run by the German support group SHG, this time inviting international participants. It is hoped to have a course in the US run by IamGSD in 2024 or 2025. There is even a suggestion that a course might be held in Australia in the next few years. And, of course, international visitors will as always be welcome at the AGSD-UK courses in the summer of 2024. Watch out for more details in due course on the IamGSD, SHG and AGSD-UK websites and Facebook groups.



Plans for a repeat of *Walk over Wales*

On the walking course this summer there was considerable enthusiasm for the idea of repeating the ***Walk over Wales*** (first held in 2010) in 2026. A small group of McArdleites walked from north to south across Wales, 210 miles, over 32 days and took in many mountains along the way. It raised awareness with lots of press coverage and even an item on the ITV 6pm news. It is obviously a major commitment to get that amount of time away from family and studies or work, but if you are interested in this long-term plan, please drop an email to Andrew Wakelin on type5@agsd.org.uk.

Online get-togethers over the winter

We are planning to again offer some online get-togethers over the winter months. We held a few of these last winter and they seemed to be appreciated. The idea is to have one per month (or two if there is demand), each with a particular topic, but also having at least half the time for open discussion. It is one way of people being able to meet others with McArdle's, which is so difficult with an ultra rare condition. Previously we held these on Saturdays at 4pm. If anyone feels there is a strong case for a different day/time slot, please email Andrew on type5@agsd.org.uk.

IamGSD news

New paper on children in Australia

A paper has just been published on the diagnosis and management of children in New South Wales, where they have a statewide centre.

The paper covers 8 paediatric cases and the children were aged 6 to 16. It is encouraging to see the finding:

> "Early diagnosis and intervention using serial exercise physiology assessments in McArdle patients can allow children prospectively to manage their condition effectively."

Adams L, Selvanathan A, Batten KJ, et al. Diagnosis and management of children with McArdle Syndrome (GSD V) in New South Wales. JIMD Reports. 2023; 64(5):327-336. doi:10.1002/jmd2.12389.

New patient stories coming

Watch out on the IamGSD website for a few more new ***Personal stories*** about McArdle's, and some of the other muscle GSDs. If you would like to tell yours, please read the guidance notes on the website and then get in touch. It is not just for McArdle's, but also all the other muscle GSDs covered by IamGSD.

Auto translate functionality on website

Probably the vast majority of people in the UK are comfortable with reading English, but if your first language is a different one, you can choose to read the IamGSD website in your first language. The site has been largely optimised to facilitate automated translation using the Google Chrome web browser. (Not all diagrams and publications are yet included.) It covers about 160 languages and there is a link on the top of every page to instructions on how to configure the automated translation. We understand that translation is now also facilitated in the Firefox browser, but IamGSD has no specific instructions for this as yet.

IAMGSD
International Association for Muscle Glycogen Storage Disease

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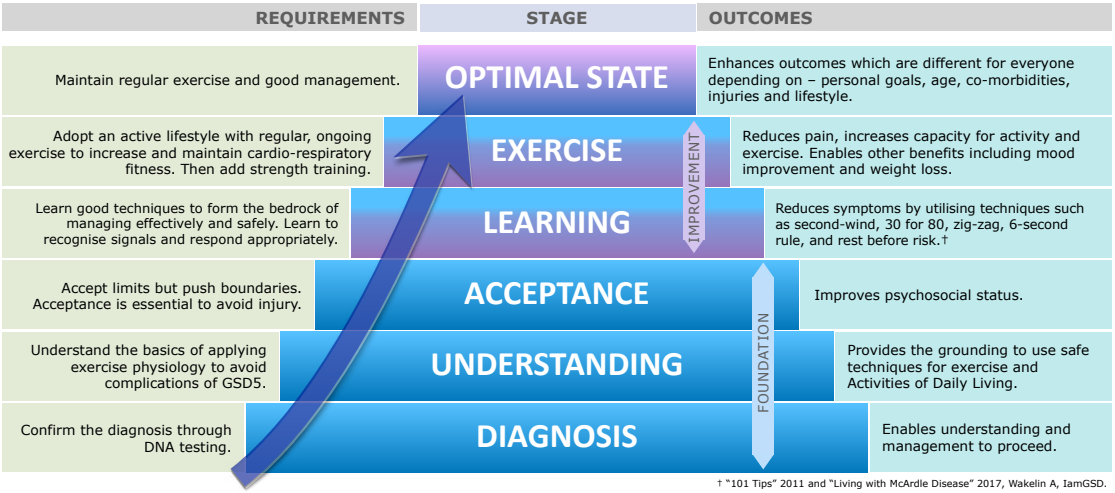
McArdle's Continuum of Care Model

A Continuum of Care model has been developed by an international team of expert clinicians and patient advocates.

This is a new practical tool for clinicians and McArdle patients. It is published in *Neuromuscular Disorders*, the journal of the *World Muscle Society*.

Led by IamGSD President Dr Stacey Reason and including Prof Ros Quinlivan and Andrew Wakelin from the UK, the team of 7 also included Salman Bhai (US), Andrea Martinuzzi (Italy), John Vissing (Denmark), and Nicol Voermans (The Netherlands). They identified areas of learning for patients to achieve an optimal state. The model outlines five pivotal steps (in order: diagnosis; understanding; acceptance; learning and exercise) to assist clinicians to determine patient-specific learning needs. This model serves as a translational tool to help optimise care for people with McArdle's.

The work on documenting this approach originated here in the UK. McArdle people may recall Andrew Wakelin presenting the *Peak Performance Pyramid* on these same lines. Now, using a formal process of six stages, this has been developed further by the expert team and agreed internationally.



† "101 Tips" 2011 and "Living with McArdle Disease" 2017, Wakelin A, IamGSD.

This new tool was presented by a member of the IamGSD Scientific Advisory Board, Dr Salman Bhai, at the conference of the World Muscle Society held in Charleston, USA, October 2023.

It is hoped that the great majority of people with McArdle's worldwide, who do not have expert centres such as we do in the UK, will be able to share this model with their clinician and other professional staff such physiotherapists. This should lead to an improvement in the number of McArdle's people achieving an optimal state.

The Continuum of Care paper is available from the Neuromuscular Disorders journal, with open access (i.e. free of charge). Go to the IamGSD website, Medical menu and choose *Managing muscle GSDs*.

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Pompe & me

Claire Wright

On the 23 September last year I woke up from an induced coma in an intensive care unit. I had pneumonia and a collapsed lung. The past 48 hours had been a nightmare for my husband and family who at times were not sure if I was going to pull through. For my part that morning I awoke very hungry and extremely relieved to find myself alive and in that setting where I was being so well cared for.

In the months leading up to my hospital admission, I had become progressively weak and had lost a large amount of weight. I had a couple of thankfully false cancer scares, was struggling with the menopause and really came to understand the term 'dead tired'. I had soldiered on, going to work each day but what on earth was making me feel so terrible?

Sixteen years earlier my older brother had finally been diagnosed with something called Pompe disease. He had gone through an extremely difficult time enduring test after test and biopsies before the cause was finally revealed. Back then there was no approved treatment, and he was lucky to get on the first trial of a drug called Myozyme. For that reason, I knew a little about Pompe, I also knew how rare it was. I am the youngest of three siblings, my elder brother is Pompe free, surely lightning could not strike twice?

On being admitted to A&E delirious and devoid of any other reason for why I was so ill I had apparently said to the doctors the words 'Pompe disease.' They of course had never heard of it as it was so rare but went off to investigate. This is where my Pompe journey began.

My first challenge in ICU was the ventilator that I now had to wear every night for at least six hours. I dreaded putting it on each night, it made me feel like I was drowning. I would stay awake as long as I could each night until I was exhausted and then the wonderful nurses would help me put in on and I would endure it until 6am. How was I going to cope with this forever? When was I going to get out of there? What was wrong with me? The questions went around and around inside my head. I decided early on that in order to not go completely mad I would need to take this situation in 'baby steps,' a day at a time. My first focus I decided was getting home.

Over the next few weeks, I received the most fantastic care and support from the nurses and doctors in ICU. As I got stronger the physio team came to help my rehabilitation, I would push myself so hard every day to walk a bit further with my frame. I wanted to go home from ICU and not go to another ward as the threat of COVID was still lurking. My bloods were seemingly being sent all over the country as the doctors struggled to find a diagnosis, day by day different diseases and ailments were ruled out as the results came back. It was so worrying and frustrating but as the results came back it was looking more likely that it was indeed Pompe.

The doctors told me that it would be at least 8 weeks before a final diagnosis could be confirmed but thankfully, I was now well enough to go home. I was elated. I was introduced to my fantastic thoracic nurse who brought me a Bipap ventilator and masks to go home with. By this time, I was keeping a diary, and, on the 5 October, I happily wrote the words 'Going home- do not forget the phone charger!'

The following months were a rollercoaster, anxiously awaiting my diagnosis, coming to terms with my disability, my nightly tussles with the ventilator and masks that would make my face so sore. I had decided to try and go back to work on a phased basis and at my brother's suggestion I contacted AGSD-UK. A decision that would provide so much practical support and reassurance in the coming months.

My Pompe diagnosis was not completely confirmed until I got my genetics report in January this year, by this time the news wasn't a shock, I was expecting it, but it hit home hard seeing it in black and white. Around this time, I had met my neuromuscular consultant and her wonderful team for the first time, I cried all the way through the meeting, which afterwards made me feel really foolish. It was early days I told myself, time will heal. I need to find a 'new me' living with Pompe.

My consultant arranged for me to have a new drug called Nexviazyme. I was so thankful, it had only been signed off by NICE the summer before, so I would have to wait for stocks to arrive. On 16 March I received my first infusion in hospital, I was one of the first 'treatment naïve' patients in the UK to receive it, I even made it into the hospital's internal magazine as a news story.

Four months on I am now receiving fortnightly infusions at home from a fantastic nurse. AGSD-UK have supported me so much over the last months. They helped me to successfully apply for Personal Independence Payment (PIP). This was one of the hardest things I ever had to write as it forced me to focus on the harsh reality of my disability and how badly it was affecting me daily.

I have so many thank yous to give for the care and support I've received over these tumultuous months. To the NHS for saving my life and continuing to care for me, to my husband and family for all their love and selfless support. To so many fantastic care professionals too numerous to thank here who continue to support me mentally and physically. My Pompe journey is only in its infancy but thanks to them I am looking forward to a positive future with Pompe and me.

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My Journey as a Single Mother of Two Beautiful Children

11 June 2023

Dear journal,

Today, I want to share my incredible journey as a single mother raising two extraordinary children. My son, who is now nine years old, was diagnosed with an extremely rare condition called Glycogen Storage Disease Type 1b when he was just a baby. Every day has presented its unique set of challenges. However, I am determined to navigate this path with resilience, love, and unwavering hope.

GSD1b is a condition where my son's liver cannot store or produce glucose. As a result, he requires feeding every two to three hours, and his nutritional needs demand constant attention and care. Watching over him and ensuring his health and safety has become a full-time responsibility, but I embrace it with all my heart. One of the most challenging aspects of GSD is my son's vulnerability to infections, which often leads to hospital visits. The fear and anxiety that come with these hospital stays can be overwhelming at times, but I draw strength from the unwavering support of my family and the exceptional care provided by his doctors. Their expertise and dedication have been instrumental in managing my son's condition and keeping him healthy and I am blessed to have such a strong support system.

As I wake up each morning, my first thoughts are always for my children. My son's condition requires careful attention to his dietary needs, constant monitoring of blood sugar levels, and regular visits to his medical team. The struggles of managing his health are demanding, both physically and emotionally, but witnessing his resilience and strength keeps me going.

I am also blessed to have a beautiful healthy daughter who is now 11 years old. She brings an incredible amount of joy and light into my life, serving as a reminder that even in the face of adversity, there is still so much beauty to be found. My daughter has also become an amazing source of support for her brother, standing by his side through the highs and lows of his condition.

This year, I took a significant step towards securing a brighter future for my children by starting university. Studying accounting and finance is not only an opportunity for personal growth but also a means to provide financial stability for my children. As I immerse myself in the world of numbers and calculations, I often find myself reflecting on the responsibility I carry as a single mother. My children's future is the driving force behind my commitment and perseverance.

Balancing my studies with the demands of my son's health requires careful planning and a strong support network. I have learned to lean on family, and community resources, cherishing every helping hand extended our way. The road may be tough, but I refuse to let it break my spirit. Instead, I choose to view every hurdle as an opportunity for personal growth and development.

There are moments of exhaustion and doubt when it feels like the weight of the world is on my shoulders. However, in those moments, I remind myself of the incredible love I have for my children and the strength that resides within me. Together, we face each day with determination, hope, and an unwavering belief in a brighter future.

Since registering with AGSD-UK two months ago, I have been fortunate to receive tremendous support from the organisation. The specialist care and welfare advisors Elizabeth Davenport and Zainib Hussain have been an incredible source of support in my life for the past two months, providing invaluable support and understanding as I navigate the challenges of my children's education and the worries that come along with it.

Zainib, has been my pillar of emotional and mental support. She has helped me navigate the challenges and uncertainties that often accompany GSD, providing a listening ear and valuable advice. She has created a safe space where I can openly share my anxieties about anything, knowing that she will provide understanding and support. Her ability to empathise and offer words of encouragement has been a lifeline for me, reminding me that I am not alone in my journey as a parent.

Elizabeth has been a constant source of guidance and assistance when it comes to my children's schooling. She understands the importance of education and the impact it can have on their future. Her dedication and commitment to their academic success has been a tremendous source of comfort and reassurance during this time.

Though the journey is far from over, I am filled with hope and determination. With the support of my family, the exceptional medical professionals, AGSD-UK, and their wonderful advisors Zainib and Elizabeth, I know that my children and I can face any obstacle that comes our way. Together, we will create a future filled with love, happiness, and success, where my children can thrive and fulfil their dreams.

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2.3 Peaks Climbed!

David Thompson reports on his training and preparations for climbing the UK's "Three Peaks" with McArdle's.

In the last Glisten, I wrote that I'd like to climb the UK's three peaks: Scotland – Ben Nevis, England – Scafell Pike and Wales – Snowdon (Yr Wyddfa). I asked anybody who might be interested to contact me.

Having attended the *Future Leaders* Walking Course in Snowdonia this July, it became clear to me that to lead such an event comes with significant responsibility. A key part is to undertake reconnaissance of the planned routes for each mountain, and to assess the time commitment and fitness demands that climbing these mountains would pose for somebody with McArdle disease.

I planned a trip for the week of 18-22 September with three friends, two were experienced mountain walkers and all three knew about my condition. A few weeks earlier the challenge might have been sunstroke, but in that week, the challenge was high winds and heavy rainfall!

In preparation I increased my attendance at the gym and from 12 August I adopted a vegan ketogenic diet. I'm vegan anyway but my previous experience of a high fat ketogenic diet had shown that it would increase my exercise capacity and allow me to shed some surplus weight which I was keen not to carry up these mountains.

In the couple of weeks leading up to our trip I started to change the focus of the fitness regime away from the gym and towards long hilly hikes. In the last week I completed three 10 mile hikes each, with >400m ascent (finding 400m was quite a challenge in my part of Wiltshire). Each of the walks started with a steep hill, but when I was strictly following a ketogenic diet (70% fat intake, 20% protein and 10% carbs) I seemed to be able to walk up it with minimal effort, without the need to wait for second-wind and with no sign of muscle cramping. However, at the 3 hour mark on each walk, and confronted by another big hill, I started to feel quite rough. I initially put this down to dehydration due to the hot weather. Confident of that assessment I ensured that I had electrolyte drinks with me for each of the mountains.

The first mountain was Snowdon on 18 September. Torrential rain and high winds whistling up the Llanberis Pass put the first day in doubt, but a quick call to Andrew Wakelin offered a solution by changing the planned route from the PyG Track to the Miners Track which would offer more shelter until the wind abated. We reached the summit in very high winds just as the clouds cleared for an amazing view out over to Anglesey and beyond. We descended down the PyG Track and completed the 874m of ascent and descent in 7 hours, only 1.5 hours more than the guide time for a non-McArdle person.

Snowdon summit – luckily the cloud lifted

Yes, we are headed there – Snowdon

Next day we drove up to the Lake District ready for the ascent of Scafell Pike. Wednesday brought more high winds and heavy rain. We had to wade across the flooded Lingmell Gill and then climb the mountain in wet boots. Visibility at the summit was less than 100m with wind gusting up to 110kph / 70mph, but on the way down the clouds cleared and we were offered a beautiful view across the Lake District and enjoyed the warmth of late afternoon sunshine. We completed the 906m of ascent and descent in 6.5 hours, 75 minutes longer than the guide time.

The ascent of both these mountains had gone well and I was pleased with the elevation

Peak 2 achieved - Scafell Pike

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that I achieved before I needed to rest. The ketogenic metabolism was clearly helping. The limit of my ability seemed to be my aerobic capacity with the need to stop being brought on by being out of breath rather than by muscle cramps. However, just as on the hikes the week before, at 3 hours in I started to feel rough even though I was fully hydrated. For Scafell Pike I had packed a glucose sports drink and an orange with me just in case it was caused by hypoglycaemia (a drop in blood sugar) and the intake of sugar certainly seemed to make a big difference. Interestingly, despite taking on board all that sugar I was still in Ketosis when I got back to our hostel.

Euphoria on descent from Scafell Pike

On the Thursday we drove from the Lake District to the Scottish Highlands in readiness for the ascent of Ben Nevis. The forecast was showing <100m visibility at the top, 1°C with high winds giving a -10°C windchill and a risk of heavy snow showers.

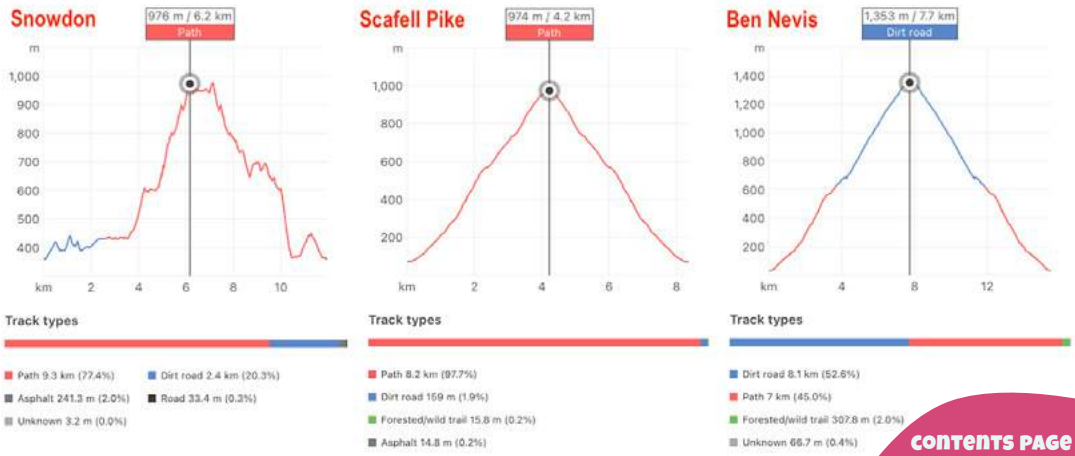
I was getting a bit concerned with the keto diet. As well as the hypoglycaemia, the high fat intake was dulling my appetite even when climbing high mountains and burning lots of calories. Before the trip I'd lost 9 kg (20 lbs) but I didn't want to lose any more. I decided to stop the keto diet that evening but that would have implications for the following day. I was awake all night with acute stomach cramps which didn't abate until I glugged a can of fizzy drink and then proceeded to bring up the previous evening's meal. I can only assume it was caused by the abrupt change in diet. I was in no fit shape to climb Ben Nevis the next day and I went back to bed as my friends set off for the mountain. Two to three hours later I pulled myself together, had some breakfast and set off after them. I texted them to say that I would meet them on the way down and I ended up meeting them at an elevation of 450m, about a third of the way up. Interestingly, for that ascent I was powered by glucose sports drinks rather than ketosis. I felt quite energised but the marked contrast was in having to stop in order to prevent muscle cramping rather than being out of breath. I've certainly come to the conclusion that mitigating the symptoms of McArdle disease with diet is a difficult path to navigate.

The extent of my ascent of Ben Nevis, at 450m

'Youth' hostel at Eskdale, Lake District

So that's how the trip ended, my friends (Sheni, Steve and Mark) climbing the 3 peaks and me climbing 2.3 peaks!

The week certainly had some challenges for me, but hanging out with friends I've known since university was a lot of fun. I'm hugely grateful for their patience, having to hang around in the wind and rain waiting for me to catch my breath. As far as I know, Ben Nevis is yet to be conquered by a McArdler. That is a challenge I have set for early summer next year. Anybody tempted to join me? I would say it is the least technically challenging of the mountains but with an ascent of 1325m, it is 50% more to climb than Scafell Pike, so not to be underestimated.



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People & Families Affected by GSDIa: Narrative Accounts

Eliza Kruger[1], Hayley de Freitas[2], Iris Ferrechia[3], Millie Gaydon[2], Andrew Lloyd[2]
[1]Ultragenyx Pharmaceutical Inc., Brisbane, CA, USA , [2]Acaster Lloyd Consulting, London, UK
[3]University of Connecticut, Farmington, CT, USA
Originally Presented at the International Glycogen Storage Disease Conference Annual Meeting, October 20–22, 2022

This study was initiated and funded by *Ultragenyx Pharmaceutical Inc.*

Ultragenyx conducted a study to capture narrative accounts of symptoms, impacts, and challenges experienced by individuals with GSDIa or the experiences of those caring for someone with GSDIa.

Nineteen study participants were recruited through the Association for Glycogen Storage Disease (AGSD) in the UK and The Children's Fund for Glycogen Storage Disease Research in the USA. 3 patients and 3 caregivers were from UK.

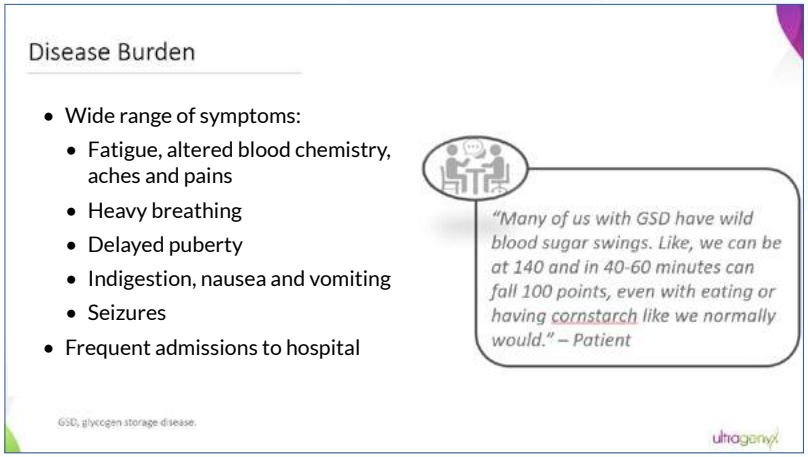
Participants were asked to write about their experiences of living with or caring for an individual with GSDIa. Participants were free to write their narratives as they wish, without responding to a specific set of questions.

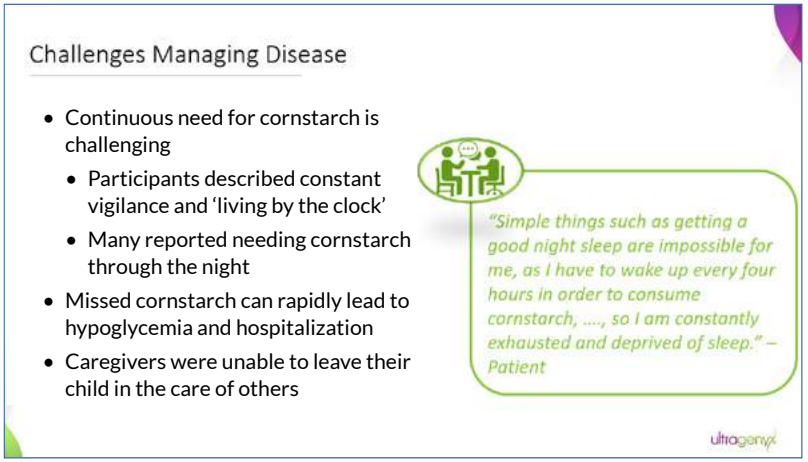
Narratives were then assessed to identify common themes across participants.

This study provided novel insights into the impact of GSDIa on individuals living with the condition and their caregivers. Participants reported a wide range of themes reflecting the broad and far-reaching impact GSDIa has on people's lives using an open-ended data collection format.

The results of this study are important to understand the impact of GSDIa on the lives of patients and their caregivers, primarily to add to current knowledge about experiences of GSDIa.

Thank you to the participants for sharing their experiences, and to the AGSD UK and the Children's Fund for Glycogen Storage Disease Research in the USA for their support.





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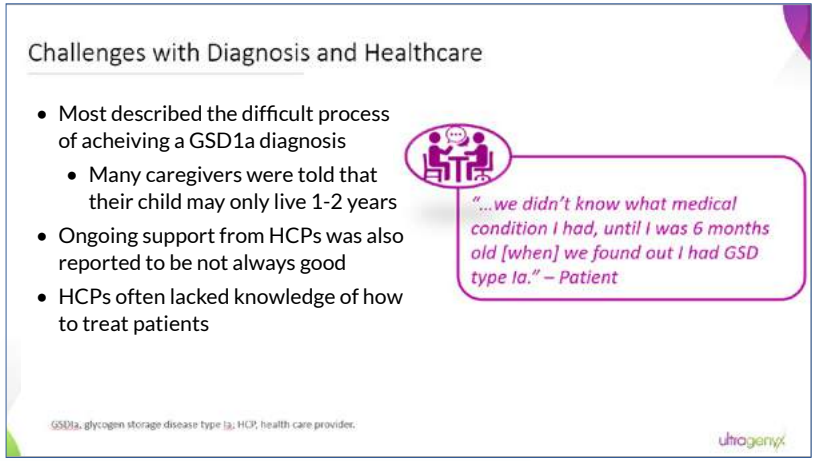

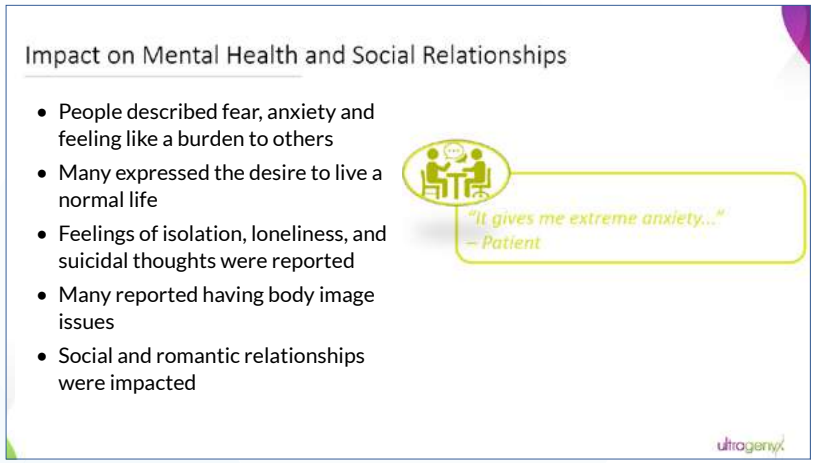

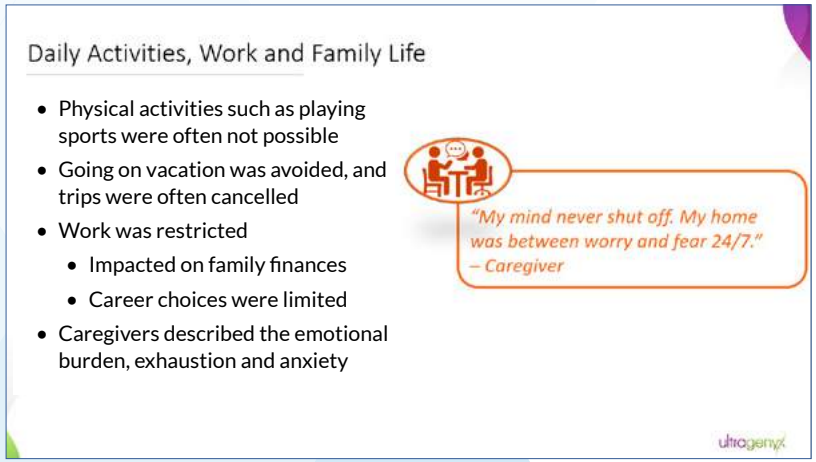

Date of prep: Jun 2023 Job code: UK-MRCM-DTX401-00145

GSDs on list of conditions for inclusion in Newborn Genomes Programme research study

Genomics England has published an initial list of conditions that will be screened for as part of their forthcoming *Generation Study*, which includes some GSDs.

The study will evaluate the utility and feasibility of screening newborns for a larger number of childhood-onset rare genetic conditions in the NHS, using whole genome sequencing.

Over 200 conditions caused by over 500 genes have been selected for screening as part of the research, which will sequence and analyse the genomes of 100,000 newborn babies in the UK.

The conditions have been selected on the basis of four criteria agreed through stakeholder engagement. These are:

A: There is strong evidence that the genetic variant(s) causes the condition and can be reliably detected.

B: A high proportion of individuals who have the genetic variant(s) would be expected to have symptoms that would have a debilitating impact on quality of life if left undiagnosed.

C: Early or pre-symptomatic intervention for the condition has been shown to lead to substantially improved outcomes in children, compared to intervention after the onset of symptoms.

D: Conditions screened for are only those for which the interventions are equitably accessible for all.

On the basis of these criteria the following GSDs will be included in the study:

- GSD 1a: gene G6PC1
- GSD1b and 1c: gene SLC37A4
- GSD 2 (Pompe): gene GAA
- GSD 3: gene AGL

The list of conditions may be subject to further change during the study, in response to emerging research and evidence. Genomics England plans to consult further on whether any additional conditions could potentially be looked for in the future as part of the research and will publicise how this will take place early in 2024.

The Generation Study could potentially pave the way for an expansion in newborn screening that might contribute to more timely diagnoses, better access to care and improved outcomes for babies and their families.

You can find out more at:

www.genomicsengland.co.uk/initiatives/newborns

Funding for new study on eating, movement & mental health in hepatic GSDs

We are delighted to announce that AGSD-UK have secured funding from The Mosawi Foundation to partner with the School of Sport, Exercise and Health Sciences at Loughborough University on a PhD project exploring eating and movement behaviours, mental health and glucose monitoring in individuals with inherited metabolic conditions that cause hypoglycaemia. AGSD-UK and Loughborough University will jointly fund the 3 year project which is due to start early in 2024.

This is an exciting opportunity to further our understanding of the psycho-social factors associated with hepatic GSDs. The research team, through this project, plan to produce evidence-based resources to better support the hepatic GSD community with healthy eating practices and specialist advice on physical activity behaviour and glucose control.

The interdisciplinary PhD, supervised by experts in their field, will explore the complexities of eating behaviours, social factors including mental health, physical activity behaviour and glucose monitoring that is involved in the management of inherited metabolic conditions that cause hypoglycaemia.

The PhD will involve 3 phases.

- Phase 1 will look to understand the prevalence of eating and mental health problems in those with inherited metabolic conditions that cause hypoglycaemia.
- Phase 2 will involve a study looking to explore eating and movement behaviours, mental health and blood glucose monitoring over time.
- Phase 3 will involve the co-design and evaluation of resources to support a wholistic approach to lifestyle behaviours (eg., healthy eating, movement) for better mental health of individuals with GSDs that cause hypoglycaemia.

It is our hope that this programme of work will advance our understanding of the eating behaviour and mental health experiences of individuals in our community and guide future directions on how best to provide them with appropriate support.

We are incredibly grateful to ***The Mosawi Foundation*** for their generosity in supporting this project.

You are likely to hear more about this project as it develops – we can't wait to share the journey with you and hope that many of you will be able to contribute to the findings and input into the development of the resources.

The supervisory team for the project include:

Dr Florence Kinnafick

(our Chair… better known as Flo Osborne, mum to Hugo who has GSD3b!)

Senior Lecturer in Exercise and Mental Health

Dr Dale Esliger

Reader in Digital Health

Dr Andy Kingsnorth

Lecturer in Digital Health with a special interest in physical activity behaviour and glucose monitoring

Professor Emma Haycraft

Professor of Psychology and Public Health with a special interest in childhood eating behaviours

The Mosawi Foundation
Ex Caritate Spes

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Exploring the role of exercise in glycogen storage diseases

Hello! I'm ***Claire Bordoli***, a dietitian and researcher at Nottingham Trent University. Alongside ***Dr. Philip Hennis***, we've had the privilege of meeting some of you at the AGSD-UK Conference and we'd like to update you on our ongoing research about exercise in GSDs.

Our initial findings

Through a study we conducted, we observed that individuals with GSDIIIa might experience challenges in maintaining fitness and strength. However, there's promising news: Exercise, both aerobic and strength training, can potentially benefit those with GSDs, enhancing their fitness, strength, and overall wellbeing.

Areas yet to explore

There's still much to understand about GSDs, especially regarding their long-term progression and the factors influencing this journey. Furthermore, we're interested in exploring the exercise routines and sentiments of individuals with GSDs.

Ongoing research details

In collaboration with Dr. Elaine Murphy from The Charles Dent Metabolic Unit in London, we're continuing our in-depth research. Participants visit Nottingham Trent University once a year to engage in a series of assessments, from quality-of-life questionnaires to fitness evaluations.

We also equip participants with an activity monitor to track their physical activity over a week.

We're pleased to note that a couple of participants with GSDIIIa have joined our study, and we're looking forward to welcoming more.

Online survey insights

Recently, we initiated an online survey for adults with GSDs, aiming to understand the factors influencing their exercise habits. Following the survey, we're preparing for follow-up discussions to gather more nuanced insights.

Future directions

Our research is progressing steadily. We're keen on enrolling more adults with GSDIIIa and are committed to this research journey for the foreseeable future. Once all data has been compiled, we aim to share our comprehensive findings with the community.

Funding

We are currently seeking funding to expand our research to include multiple sites across England.

Your support is invaluable

We genuinely appreciate the support and interest from the community. If you'd like to participate or have any queries, please reach out at claire.bordoli@ntu.ac.uk. Together, we can further our understanding of exercise's role in GSDs.

Survey opportunities

The following research opportunities are currently open to people living with GSDs.

If you're able to spare the time to take part in studies like this, it can really help increase understanding of GSDs and give people affected a voice. Your input could help shape the development of future treatments and management, along with the information and support available to the community.

Pompe survey

UK Pompe community members and carers are invited to take part in a survey in collaboration with a global pharmaceutical company. The survey explores:

- the impact of Pompe on you and your family and how this could be improved
- your experience and expectations of treatment and management of Pompe
- your experience of accessing information about Pompe.

To get involved just use the following link: https://tinyurl.com/26dty7hj -or if you'd prefer to answer questions via telephone or another format, please contact Pompepatientexperiencesurvey@wacomms.co.uk and this can be arranged.

The survey should take no longer than 15 minutes to complete and you will be reimbursed with a voucher to thank you for your time.

See more survey opportunities overleaf

Find the link to the Pompe survey here

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Survey opportunities cont...

Experiences of EAMS study:

Do you want to share your experience of the early access to medicines scheme (EAMS)?

Amicus Therapeutics is interested in getting your feedback on the scheme, and how you feel after completing the EAMS. You can share your responses by talking to a researcher from Core Human Factors in a one-on-one interview. Core Human Factors is a research company, working in partnership with Amicus to host interviews with study participants and will maintain your anonymity. The interview will take place via video conference or phone call and will take up to 45 minutes. If you participate, you will be reimbursed for your time as a thank-you. If you are interested in participating you can call/text: +1 267-705-6071 Email: join@corepn.com

GSD1a study

Adults with a confirmed diagnosis of GSD1a are invited to take part in a study to understand experiences of living with the condition, including symptoms and impact on daily life. This study consists of an interview over the phone or via video-conferencing, which will last up to 90 minutes. Participants will be compensated £150 after completing the interview. The study is being run by Opinion Health. Anyone interested should contact info@opinionhealth.com using 15332 in the subject line or use the following link to register: https://tinyurl.com/GSD1a

Find the link to the GSD1a study here

Pancreatitis study

Opinion Health is also looking for interview participants who may have been affected by acute pancreatitis. This will be a 60 minute online interview and participants will receive £185 for their time.

To find out more contact info@opinionhealth.com with 16106 in the subject line or register here: https://tinyurl.com/2sstjr73

You can find out more about Opinion Health at - www.opinionhealth.com

Find the link to the Pancreatitis study here

notes

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, Beam, Mosawi Foundation, Sanofi, Spark Therapeutics, Ultragenyx and Vitaflo.

Rob Hill and his brother in law, Steve

Upcoming events!

- **Living well online socials starting 30th November 2023**
- **Online festive event 9th December 2023**
- **AGM January 2024**
- **Parents' group starting 14th March 2024**

Details of these and other events in the coming months will sent out by email and on our website and social media - just contact **info@agsd.org.uk** for more information.

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AGSD UK
Association for Glycogen Storage Disease