August 2020

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

Can you help us to help others?

See inside how you can fundraise for us and be inspired

Association for Glycogen Storage Disease UK
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Phone: 0300 123 2790
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Charity Number :1132271
August 2020

Thank you!

This issue is dedicated to everyone who has raised money for us this year, and in the past.

Thank you so much!

An important question...

Could you help with fundraising?

We have had varying amounts raised for us this year 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:
Lynne Sharman selling Covid-19 face masks
Chance Craig (GSD9a)

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 Chairman

Welcome to Glisten, looking back to our last edition in February it's clear that we are living in a very different world.

Most importantly, to our knowledge, all members affected by GSD and the professionals we know and care about, are well, with very few cases of Covid-19 reported from our wider community.

In March the Evelina Family Day was our first casualty, the event being postponed to 2021. This was swiftly followed by cancellation of the Pompe Social, the GSD3 CATS team get-together, the Pompe Support Team training weekend and International Pompe Day meetings. We negotiated hard but these late cancellations meant we unavoidably lost almost £1000.

As a small charity we are light of foot and able to respond rapidly to changing circumstances. Following some quickly convened meetings, we determined that our reaction to Covid-19 would focus on becoming a centre for quality information, responding to urgent needs of all members; offering practical support and sticking to the published guidelines and facts, not speculation. Trustees agreed a quick issue grant, £50 payments to those in urgent need which could be paid within 48 hours. All the time we, like everyone else, were coping with our own fears, anxieties and practical issues, such as switching to working from home and supporting family members.

It has been an extremely busy few months, with enquiries during the first two months increasing by more than 40%. Our website has seen greatly increased traffic, especially to the Covid-19 information area. We have issued many updates and alerts about webinars and Government guidance and we have provided clarification when some of the published information has been confusing and conflicting. This has resulted in some very good feedback from Specialist centres and wider industry.

Our GSD Co-ordinators have been busy arranging Zoom meetings to help keep people informed and special thanks to Gemma Seyfang who, in spite of having to postpone her own wedding, has run many Zoom sessions, including a famous fun quiz. Becky Walton, a hairdressing trainer also provided a Zoom showing cutting and styling tips for those brave enough to have a go at home. Many appreciated the British Inherited Metabolic Disease Group (BIMDG) webinars (they are available for watching on our COVID-19 web page).

Yes, there were serious issues, principally queries about shielding letters, and this affected our GSD community in particular. Some were not told to shield but should have been and some to shield unnecessarily. We have taken up this issue within the NHS and contributed our feedback to several Covid-19 surveys.

Families

Our families merit a special mention. They are caring for sick children, managing complex diets, coping with disrupted home-care and fearful of attending vital hospital appointments. All this, whilst also home-schooling and home-based working; life has been very arduous. We understand and we applaud you all.

We need your help

Right now, our greatest need is for help with fundraising for the charity. We are not able to apply for most of the Covid-19 related funding streams because they are specific to certain localities and we operate nationally. One simple action I would recommend you all to consider is linking up your Amazon account with AGSD-UK. This costs nothing and 0.5% of any purchases you make via Amazon are passed to the charity. If you go to www.smile.amazon.co.uk you simply choose the charity you wish to support, we are listed under our full name 'Association for Glycogen Storage disease (UK) Limited'. With so many people shopping on-line now and many of you already using Amazon, this could provide a continuous income stream for almost no effort – please have a look.

Conference 2020

Obviously, a multi-GSD community conference is not advisable and so we have decided to cancel the 2020 conference at Wyboston Lakes. We will be in touch with an alternative offering, watch this space for alerts about Autumn plans. We are looking into holding a conference-style event as soon as practically possible, in 2021, subject to health alerts and safety guidance concerning Covid-19.

With all my best wishes

Nick Jones

Acting-Chair AGSD-UK August 2020

Campaign for Newborn Screening

We have always been supporters of the Campaign for New Born Screening.

Pat Roberts was a long-time champion and has now joined forces with another charity called ArchAngel MLD. Together they are providing a refreshed and dynamic movement. AGSD-UK is working alongside them on this important project.

Together we are strong!

Georgina Morton, of ArchAngel MLD Trust, gave us this report:

ArchAngel joined forces with ***Nickie Aiken MP*** to call for urgent review and expansion of the UK Newborn Screening programme, including the addition of all appropriate metabolic disorders, to align with other high-income countries.

The campaign was endorsed by key rare disease organisations and clinicians, as well as over 70 cross-party MPs, and successfully secured a dedicated debate with Health Secretary ***Matt Hancock***. Despite the intervention of Covid-19, we continue to prepare a robust argument to present at the appropriate time.

The voice of the patient community is vital to this and we urge all supporters to sign the petition.

AGSD-UK has a campaigning area on the website www.agsd.org.uk/about-us/reports-and-more/our-campaigns/. Here, we have gathered four stories of parents' experiences in the USA, where Pompe disease is on the NBS panel in many States.

Georgina Morton, Chair of ArchAngel MLD Trust with her family

During June we promoted this heavily by email and on social media.

You will also find a link to the UK petition - please sign and support if you can **www.nickieaiken.org.uk/Archangel**

Good Governance

Policy Updates & Safeguarding

In June 2020 some of our policies were overdue for review, update and re-confirmation by Trustees. We did a mini-review and ensured that a new list was posted on our website here: www.agsd.org.uk/about-us/reports-and-more/our-policies/

This is crucial so that everyone knows we have good governance and that we are transparent in our work.

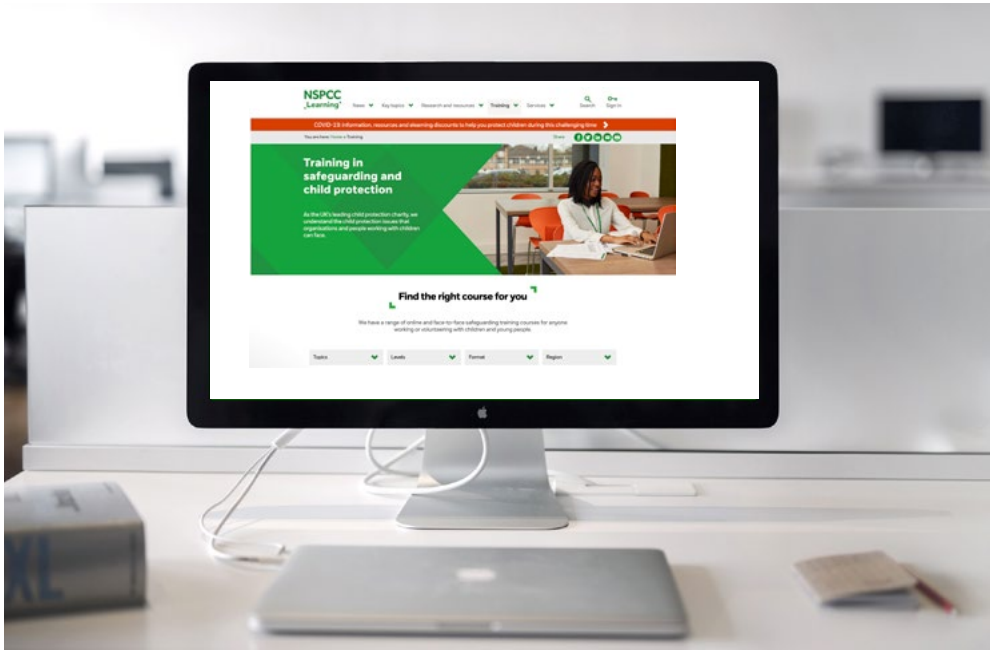
Jane Lewthwaite is Safeguarding Officer and Ailsa Arthur, Safeguarding Lead and both have updated training.

We recommend, as useful for anyone at all, this short online mini-course from NSPCC. It costs just £25 and takes a few hours at most.

It can be found here- www.learning.nspcc.org.uk/training/introduction-safeguarding-child-protection

All our staff, Trustees and volunteers have up-to-date DBS checks.

NSPCC Learning



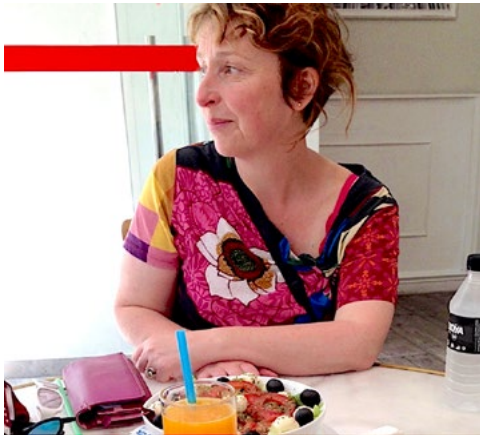
new Trustees

New Trustees

We welcome two new co-opted Trustees. The AGM will take place later in the year.

Sarah Chikhani has joined the board of trustees and will bring her knowledge and expertise in finance to act as our treasurer. Added to this, she has been a pharmacist for the last ten years, working in both the public and the private sector. Her skills in these areas will be a great benefit to the AGSD-UK. She enjoys running and exploring the countryside with her dog.

All at the AGSD-UK offer her a warm welcome and look forward to working with her.



Rachel Thomas Hello, I would like to introduce myself. I am Rachel and have just joined AGSD-UK as a trustee. My daughter was diagnosed with McArdle disease, GSD5, about 6 years ago.

We have been to Wales on the walking courses for the last 5 years and met so many amazing people from all over the world who have so many stories to tell. The journey to get Bronte diagnosed was a long and painful one, as it was for many. I am in contact with many of the McArdles that we have been privileged to meet and talk to several McArdle mums every day via the internet.

I have become very interested in finding a way to support our teens and young adults through diagnosis and then on into adulthood. Diagnosis at a young age is obviously an immense advantage but for many it's the start of a very difficult journey and I hope to be able to contribute to the discussion on how we may be able to improve this.

AGSD-UK on Social Media

Lisa Massimo took over our social media management in February 2020 and we say a massive thank you to her for transforming our Facebook page.

Throughout the Covid-19 pandemic, the Facebook page has allowed for the latest updates regarding shielding and other issues to be available in one place, as soon as they are announced, as well as providing an online community of people going through the same thing together.

There are now dedicated online meeting groups for those with GSD1, GSD2, and GSD3, as well as regular quizzes in the GSD2 Group.

Relevant web seminars are also available, for example Medics for Rare Diseases, which educated junior doctors on the issues surrounding a rare disease, such as GSD.

The latest events and fundraising updates are also available, to keep up to date with everything that is going on at AGSD-UK.

Thank you again Lisa for helping build this online community and for all of the support and signposting offered during this difficult time.

The Facebook Page can be found here:
www.facebook.com/AGSDUK

GSDscreen – our YouTube channel

The AGSD-UK YouTube channel has recently been given a bit of a makeover and we now have playlists for several of the GSDs, and a playlist of patients' stories.

It is now easier to find and navigate. Lisa Massimo recently added her video explaining how to set up a "Give as You Live" account, but many of the other videos are older and had been rather hidden away.

Take a look… you may be quite surprised what is there! If you have suggestions for videos or channels which could usefully be added to GSDscreen, do please let us know. In particular we are looking for a volunteer to coordinate the recording of many more patient stories, for all the GSDs. Contact us on:

info@agsd.org.uk

You can find our channel at:

www.youtube.com/GSDscreen

Supporting Wellbeing for All

Most, or perhaps all of us, experienced fluctuating emotions and difficult feelings over the last few months. There were times when I felt overwhelmed with anxiety and uncertainty. Then I regrouped, working through or around problems, often encouraged by other people being positive. It was good to listen and learn from everyone about how they were coping. Many turned to mental health support Apps, especially one called *Calm*.

We decided we would review some of them and then provide a resource for everyone affected by, or working with, GSD. It was important that we offered this to our close and valued professional community as well as members, patients, families.

Pompe Support Team, helped by Sue Davey, trialled some online courses and Apps and we decided to block buy places on course advertised on the next page.

So far, we have signed up over 50 people on the course.

www.agsd.org.uk/news/free-online-mindfulness-course/

It takes up to 25 hours to complete and you have access for six months so you can work through at your own pace.

You can sign up right to the end of 2020, just email info@agsd.org.uk for more information or to join.

Jane Lewthwaite

AGSD UK
Association for Glycogen Storage Disease

Living Positively

Work at your own pace, once started it is open to you for six months. A mix of videos, presentations & quizzes.

A gift for you from AGSD UK
Free, online
Mindfulness Course
for anyone affected by GSD

This course is being made available to patients, carers, family & professionals.

It takes about 25 hours and could help you in daily life, with relationships, coping with Covid-19, for your CV.

It is a broad skill suitable for all.

To join send an email to info@agsd.org.uk
You will receive an email from Forum Training with a link to login & password

FORUM TRAINING
Professional Development

The Basics of Mindfulness
Understanding Stress
Improving Resilience
Mindful Decision-Making
Focus, Performance & Communication

The course is titled Mindfulness in the Workplace but it is suitable for everyone even if they do not work; 'workplace' can be any team, family or group. Limited places.

A WORD ABOUT THE WEBSITE

Our new website continues to develop and is popular in the UK and around the world, delivering information and support to those affected by GSDs. Our regular Covid-19 coverage has been praised by clinicians and professional bodies.

Since it was established in early 2019, we have had over 108,000 page views, and are averaging over 4000 views per month. In the last year the largest number of referrals have come from search engines with over 18,000 referrals, and 3000 have come from Facebook.

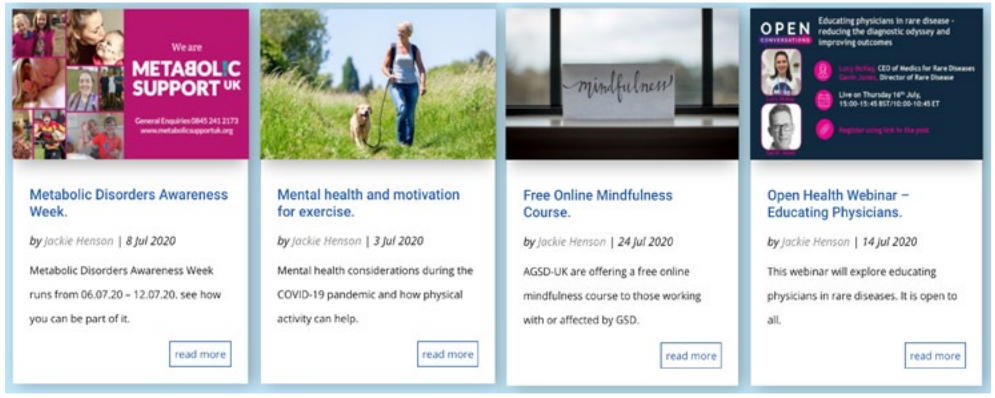
- Visit on a regular basis as we have a news story every week.
- Our news story on Dr David Weinstein's move has attracted well over 400 views.
- Our news story on McArdle's being mentioned in a Netflix documentary attracted over 1700 views from around the world.

We continue to build the information on each GSD. The most prevalent GSDs have each reached from 1500 to 5000 views in the last year.

We keep adding more personal stories from members and trying to ensure we have a good range for each GSD. Over the last year these personal stories have typically had 100 to 200 views.

- Our publications are available for browsing and reading online, jointly hosted with IamGSD.
- Our booklet "My GSD and Me!" has been opened online nearly 500 times.
- The booklet "Living with McArdle Disease", developed jointly with IamGSD, has been opened over 1000 times.

In the last year the site which users have most linked to has been IamGSD, closely followed by BIMDG.



HANDY GUIDE TO ORGANISATIONS

Confused as to what IamGSD and BIMDG stand for? We're here to help!

BIMDG British Inherited Metabolic Disease Group
Advancing the education, diagnosis, care & treatment of inherited metabolic disease

CAMBRIDGE RDN Cambridge Rare Disease Network
Addressing unmet needs of those living with rare conditions, their families

CQUIN Commissioning for Quality and Innovation - *Ensuring best practice is carried out within the NHS.*

ECRD European Conference on Rare Diseases - *Largest gathering in Europe for the rare disease community*

EMA European Medicines Agency - *Part of the EU, assesses and approves new treatments*

EUPATI The European Patients' Academy on Therapeutic Innovation
To improve the availability and quality of medical information

EURORDIS *Alliance of patient organisations representing 929 rare disease patients in 72 countries.*

Genetic Alliance UK *Charity for 200 patient organisations for people affected by genetic, rare and undiagnosed conditions.*

IAMGSD International Association for Muscle GSD
Patient-lead international group: McArdle's & muscle GSDs

IMI Innovative Medicines Initiative - *Aims to speed development of better, safer medicines*

IRDiRC International Rare Disease Research Consortium - *Aims to hasten diagnosis and new treatments*

Medics4Rare Diseases UK *Educating junior doctors in rare diseases so that they might diagnose them sooner.*

MetabERN European Reference Network for Hereditary Metabolic Disorders
Non-profit established by the EU.

METABOLIC SUPPORT UK *Charity committed to improving the lives of patients living with Inherited Metabolic Disorders and their families.*

NICE National Institute for Health and Care Excellence - *National Guidance to improve health & social care.*

NIHR National Institute for Health Research - *UK largest funder of health care research*

NORD National Organisation for Rare Disorders
US based. Services for patients &medical professionals, seeking to develop new diagnostics and treatments.

ORPHAN DRUG CONGRESS *Focuses on the most pressing challenges and opportunities to bring rare disease therapies to patients faster.*

ORPHANET *Reference source on rare diseases and orphan drugs. An access point for quality information concerning rare diseases.*

RDUK Rare Disease UK- *A National Campaign working to help implement the UK Strategy for Rare Diseases.*

Fundraiser Stories

THANK YOU EVERYONE!

We appreciate your support and love hearing all of your stories about what AGSD-UK means to you and your families. Here are just a few, thanks to all those not featured.

Take inspiration!

Sonia Worthy, You are worth a lot to us!

Sonia has so far raised **£150** with her club, The Inner Wheel. Pre-lockdown, a silent auction was planned, in aid of AGSD-UK, it's being rescheduled!

Sonia chose AGSD-UK because she has found the group an incredibly supportive place both pre- and post-diagnosis with GSD5. In Sonia's own words, being diagnosed with McArdle's was a largely positive experience, thanks to AGSD-UK, as she now has support and information that had previously been unavailable to her.

We wait to see what happens with the new silent auction and thank Sonia for her help too!

Kieron Stubbings, Super-charged!

Kieron was due to run the London Marathon in aid of AGSD-UK this year, but Covid-19 changed that! However, with his fiancé Gemma, they have managed to raise over **£3000,** with the total still growing. They have held many events, such as a Bingo night that raised over **£797** and an Indian Curry Night that raised over **£888**. They have also organised many auctions, and have more prizes still available, including a Mercedes Benz watch, a smart TV and an Ipad. All of the information about past fundraising and their upcoming projects, as well as running updates from Kieron can be found on their Facebook page and Virgin Money page, both linked in this article.

Kieron has chosen to donate to AGSD-UK because his fiancé Gemma has Pompe Disease, GSD 2. They are both massive supporters of the charity and we are as grateful for them as they are for us. Thank you so much to both of you and good luck with the marathon when the time comes Kieron!

Facebook: Kieron's London 2020 Marathon Fundraising Page

Virgin Money: https://uk.virginmoneygiving.com KieronStubbings

Lynne Sharman Covering up for Cash!

Lynne has raised **£120**, with the total still growing, making and selling face masks. Lynne sews as a hobby anyway, and when the Government encouraged us to begin wearing face masks, she decided to start making them from beautiful pieces of material that had been donated to her. She is currently selling the masks to her friends and family, as well as through her local hairdressers, where an accompanying leaflet on GSDs can also be found. Lynne is selling the masks at £2 each. We thank Lynne for her donation and her support for AGSD-UK!

Pictured is Lynne with a selection of masks she has made!

Dawn Fitzroy Slimming and Saving

Dawn is a Slimming World Consultant in Hindley, and every Saturday morning, she has a donation tin on her desk in aid of AGSD-UK.

She has raised in excess of **£500** in the name of her daughter, Becki Fitzroy who has GSD 3.

Thank you so much Dawn and all of your generous Slimming World members!

Lucie Morran's Marathon

Lucie completed the Heritage Marathon on the Suffolk Coast. She raised **£175** for the charity by running her first marathon! Lucie is a keen runner, and runs for Black Dog Running Club in Bungay. She signed up to the marathon to challenge her abilities whilst fundraising at the same time. Her childhood best friend's daughter has McArdle's disease, and she has seen first-hand how invaluable the support from AGSD-UK is, for not only those affected, but the entire family.

Thank you Lucie and we wish you luck on your next marathon!

Georgie Thompson's a High Flying Fundraiser

Thanks to Georgie who undertook a sky dive. She said,"AGSD-UK is a charity close to my heart".

Her bravery raised **£230.00**.

"AGSD-UK is a charity close to my heart"

More Fundraiser Stories

Simon Attewell Raised **£516** doing the 2.6 miles challenge.

Eileen Rice donated **£100** and said:

"I appreciate all the help, knowing that someone is there when I need to find out something or ask questions.

It is nice you understand and that I can lift the phone at any time if I am unsure about anything."

A special mention to GSD3 Co-ordinator **Sylvia Wilson & husband Ed**. Long-time volunteers and fundraisers, Sylvia continues to collect used stamps, trading them in for **£40** last year.

Nikki Christie's Bake and Take

With the help of everyone at Penns Primary School in Birmingham, Nikki Christie donated over **£300** to AGSD-UK, raised through a school bake sale and Jeans for Genes day.

Nikki herself has GSD3 and is grateful for all of the support she has received from AGSD-UK and so wanted to give back.

Thank you so much Nikki and all of the parents and children at Penns Primary School.

We hope you enjoyed your cakes!

Children! Go to page 53 for quizzes.

SELF RAISING?

Support from bread-maker Warburton's charity 'Family Matters'

We successfully applied to Warburton's Family Matters Community Grants for a small grant to help towards our Family Days.

They gave **£250.00** after voting by their own staff members.

We run these, usually on or near hospital sites, welcoming families, children and those affected by GSD. We have entertainment, lunch, usually a face-painter, and importantly, it's a chance for families to connect and have relaxed time to meet staff too.

Thank you Warburtons!

Conference Reports

Educating Physicians in Rare Diseases

Gemma Seyfang,
Pompe Support Team

Medics 4 Rare Diseases Webinar

Educating Physicians in rare disease
Reducing the diagnostic journey and improving outcomes.

Lucy McKay, CEO of Medics 4 Rare Diseases introduced the organisation. They support UK based junior doctors in diagnosing and treating rare diseases. Lucy has experience with patient lead organisations as her mother started one to aid her brother with his rare disease diagnosis; Lucy then went to medical school.

The aim of Medics 4 Rare Diseases is for rare diseases to be at least considered when presented with a group of atypical symptoms that do not apparently fit more common conditions. Doctors cannot be experts in 7000 different rare diseases, but can be aware that it is a possibility.

There are around 3.5 million people in the UK with rare diseases - this is the same amount of people who have asthma. So rare diseases are not as uncommon as once thought, and so need to factor more heavily in the diagnostic process.

The overall aim is to increase awareness of rare diseases by beginning with medical students, getting them to understand that they should suspect rare diseases or at least will come across them during their careers.

Q&A

Q: Who is it, on the diagnostic odyssey (journey), trying to target?

A: Doctors should be taking note of those with a rare disease as a patient knows more than the professional. Patients are EXPERTS in their DISEASE. It forms a kind of triangle:- the disease, the doctors and the patient groups. If the doctors work with the expert patients then the doctors will become another advocate.

Q: What should professionals be thinking about pre-diagnosis?

A: Lots of medical professionals look back to see when the symptoms started, a red flag; family history, clinical results, genetics. A list of symptoms is not a diagnosis. Professionals can remember this point, if you cannot help further; refer on!

- We don't want rare diseases to fall through the cracks, it's everyone's responsibility.
- Those with rare diseases need to remember they may not find someone in their GP surgery, their hospital, their area or even their country that knows about this rare disease they are presented with, but there will be the right person somewhere.
- There is hope: Keep searching.

And a final note

'Treat the Patient, not the Results'

EURORDIS ONLINE

Kempton Rees, IPA Community Advisory Board Member

When asked to represent AGSD-UK at the 10th ECRD conference, I jumped at the chance. After all, I'd never been to Sweden, let alone Stockholm. I had heard great things about it, and I quite fancied supping an extortionately-priced beer on the waterfront in the old town at the end of a long and interesting day.

That was before Covid-19.

The reality was somewhat less glamorous – sitting in my basement, staring at a screen with a tin of herring open beside me while Abba played quietly in the background. Welcome to the world of online conferencing!

The full programme was relayed on a virtual platform which, while new to me, was pretty intuitive. You can still access the platform, including all seminars, here: **https://onlinexperiences.com/scripts/Server.nxp**

The meat of the conference was of course the opportunity to take part in the live sessions.

They had begun with a welcome from HRH Crown Princess Victoria, followed by a plenary session in which a number of speakers spoke on various topics. There was much discussion of Covid-19 of course, with one speaker comparing it to a rare disease. There was general agreement that it highlighted the desirability of humanity to work together to eradicate illnesses.

Ana Rath, Director of Orphanet, highlighted the work of the 'Rare 2030' foresight study – a 2-year EU initiative which *"… gathers the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe".*

Details can be found here:

www.rare2030.eu

There was a choice of breakout sessions based on 6 themes:

1. The future of diagnosis: New hopes, promises and challenges;
2. Our values, our rights, our future: Shifting the paradigms towards inclusion;
3. Share, care, cure: Transforming care for rare diseases by 2030;
4. When therapies meet the needs: Enabling a patient-centric approach to therapeutic development;
5. Achieving the triple as by 2030 (sic): Accessible, available and affordable therapies for people living with rare diseases; and
6. The digital health revolution: Hype vs. reality.

I heard speakers on a diverse range of topics, including:

- The striking fact that 86% of GPs in England accept that the training they have received in rare diseases is 'poor', despite collectively representing a significant minority of their patients;
- The fact that historic diagnostic journeys for very young children have been far too long to enable relevant timely clinical decisions to be made – and the very impressive efforts being made by a team from Cambridge to bring relevant testing lead times down to days, rather than months;
- Ethical cost-benefit issues concerning the timing of imparting diagnoses – are people always ready for it?;
- Efforts being made to bring diagnostics and treatments to Aboriginal people living in remote places in Australia;
- Citizen-centred data sharing': proposals to provide individuals with full editing rights to their personal medical data, allowing people to access support on their terms; and
- A straw poll on the questions such as whether or not patients should be more involved in co-creating solutions for their needs (100% who responded said yes, which the moderator described as 'astounding' – presumably because he expected a handful of people to click on 'no' by mistake).

All in all, it was an interesting couple of days…

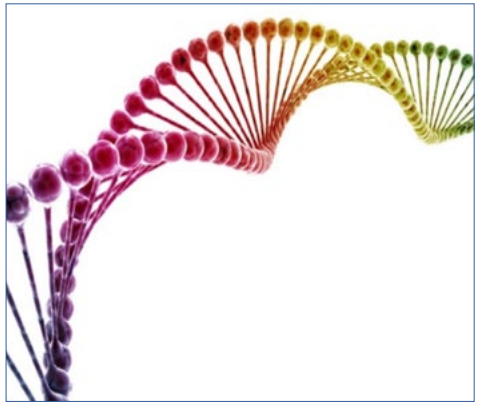
Are the rarest muscle GSDs really very rare?

Andrew Wakelin, McArdle's Coordinator

It is a fascinating question! AGSD-UK has a high profile in GSDs and is often referred to by people seeking diagnosis, by patients and by clinicians worldwide. In recent months we have been contacted about some of the very rare muscle GSDs, notably GSD 0b, 9d, 13 and 15, by people in Brazil Germany, Italy, Spain and the US. We always do what we can to assist.

The rarest muscle GSDs have only a handful of diagnosed cases each, for example GSD13 has just 6 known cases worldwide. (In comparison, there are around 4000 people globally with GSD5 McArdle's.) Both the Euromac Registry and IamGSD provide support for all these conditions, and AGSD-UK likewise.

With 0**b** and 9**d** it is vital to add the subtype letter, as 0 and 9 are more generally known as hepatic GSDs, but these two subtypes are muscle-specific. (Some of the 0 and 9 subtypes are liver-specific and some involve both liver and muscle.)



To what extent are the low patient numbers due to the difficulties of achieving a diagnosis?

Could we be at the same stage as the early days of McArdle's, with maybe many hundreds of cases yet remaining undiagnosed?

Constantly evolving genetic testing is leading to wider genetic panels, better tools for analysis, faster turnarounds and lower costs. In the coming decades maybe we will see these muscle GSDs overtake McArdle's in diagnosed numbers.

AGSD-UK maintains a record of people who contact us regarding these all ultra-rare GSDs so we can put people in touch, something which can be hugely supportive. It is very satisfying when they are spread worldwide. Much McArdle literature, particularly suggestions for practical ways to manage the condition, can be of great help to these people and we are pleased to share it with them.

The view from Scotland: GSD 9a

Amanda Crawford, Parent

My son Chance, now four, became the first person diagnosed with GSD 9a in Scotland in October 2018. When he was born, he refused to take milk for the first six hours and then when he did start feeding, he would cry in pain, be very sick, and then feed again. This rotation was my new life. I queried his blood sugar with the emergency doctors, but nothing ever came of it.

In spite of the projectile vomiting and stomach soreness, he was a bright, interactive and happy baby and never cried without reason.

His other symptoms included sweats, high temperatures and loose, acidic bowel movements, beginning at birth and ending with his eventual treatment. I was advised these were all caused by "undetectable viral infections".

Before the age of one, he was rushed to hospital on at least eight separate occasions and was seen by countless health visitors, GPs, and paediatricians.

Around eight months old his tummy became noticeably big, but health professionals dismissed my concerns.

Chance rarely slept, but would play in his cot happily. When he did sleep he'd wake up to five times a night and I would go in with milk for him. He was ten months old before he began weaning. I took all of these signs that something was wrong to doctors but my concerns fell on deaf ears.

Finally, following another referral for suspected allergies, bloods were taken at twenty-two months old and our rollercoaster diagnostic journey began.

We had another baby boy, CJ, and by the time of diagnosis I was pregnant with a daughter. It was an incredibly difficult time. We were lucky to be diagnosed within eight months, but our clinical team did not believe that all Chance's symptoms were related to the genetic condition and it was a further, frightening five months of deterioration before Chance was appropriately assessed and treated.

Since seeing a GSD specialist at London's Evelina Children's Hospital in April 2019, Chance has been fitted with a mic-key button feeding tube and started a strict two-hourly feeding plan incorporating food, small amounts of carbs and higher volumes of protein, daytime corn-starch and two overnight feeds. We never give him anything sugary, before treatment he lived off coco pops and toast! But this doesn't stop him raiding cupboards for chocolate, his only weakness.

Since beginning treatment, all of his "viral" symptoms have subsided: he sweats only if he has had something sugary or too many carbs; his eczema is under control; and most importantly his blood sugars normalised within twelve weeks of starting treatment. We now only test his sugar levels and ketones if he becomes symptomatic and his ability to eat and try new foods has improved dramatically.

Chance understands his body and condition which helps teach him independence. I understand from specialists that Chance is severely affected and can only last around one hour between meals without treatment. Feeding around the clock whilst caring for three small children brings new challenges, and is indeed tough, but we are very proud of how Chance has handled everything.

I am forever grateful to the specialists who have supported my son and our family through the past year.

Planning Ahead

Jane Lewthwaite
Specialist Care Advisor

Perspectives on making healthcare choices ahead of time

Covid-19 brought huge changes, and some adjustments, for many.

For older people and those with existing health problems linked to increased risks, there was fear too, at times. Subject to their range of symptoms and disease progression, some AGSD-UK members had shielding letters, some did not, others received a letter and were then advised it had been incorrectly sent. It was confusing, although the second Metabolic Webinar clarified a lot. It nudged some people to focus on what they wanted if they became ill.

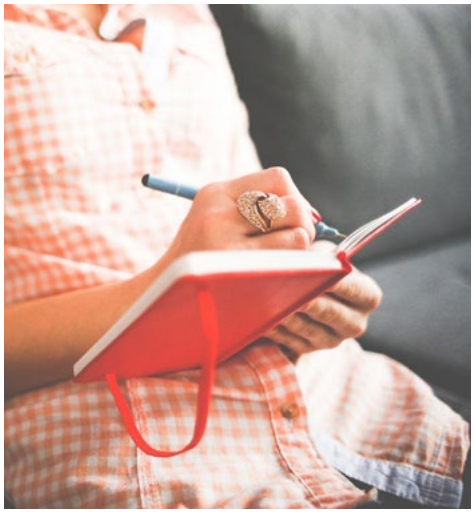
Planning ahead: A few practicalities

- Keep contact names and numbers for your healthcare professionals handy, on view, and ensure family, friends and carers have them too.
- If you have a risk alert card for your health condition, ensure it is accessible and that family or carers know where it is.
- Hold on to explanatory booklets and ensure they are to hand.
- Obtain and store extra supplies of medications, feeds and equipment.

Record your views now, for the future.

Making practical steps toward planning ahead helps some people to feel more in control. It can reduce anxiety to know your opinions are recorded. It helps loved ones and professionals to deal more confidently in difficult situations.

Advance Care Planning is a way to think ahead, to describe what's important to you and to ensure other people know your wishes for the future. Thinking ahead, discussing with others and writing things down means that your wishes are known and respected, and you're more likely to receive the kind of care you want in the place you choose if you become unwell, or if you could no longer speak for yourself.



An Advance Statement is a written statement that sets down your preferences, wishes, beliefs and values regarding your future care. The aim is to provide a guide to anyone who might have to make decisions in your best interest if you have lost the capacity to make decisions or to communicate them.

Advanced Decision Making is also called a 'living will'. It allows you to refuse treatment, even if this might lead to your death. An advance decision is legally binding which means that those caring for you must follow your instructions.

www.nhs.uk/conditions/end-of-life-care

ReSPECT (Recommended Summary Plan for Emergency Care and Treatment) The Resuscitation Council has designed a range of information for professionals, patients, parents and carers all available on their website.

Respecting Wishes

Rowena Johnston's Story

Rowena has Pompe disease; this is her experience of planning ahead:

I received the 'shielding' letter in mid-March and Dr Hiwot, my specialist at QE Hospital Birmingham phoned me, he was clear that I must shield completely from this awful virus. Five years ago, my body rejected completely any form of ERT, so I now am unable to have a fortnightly, 'boost to my enzyme level'. I am now in a situation that I have very little spare energy and even a simple cold can make me very unwell for two/three weeks.

My GP contacted me later and started to discuss filling in a ReSpect form. These were introduced in 2019, but the Covid-19 situation has seen GP's being asked to suggest that more of their patients, with underlying health problems make their wishes known, should they become seriously ill. These forms are not just for the current problems but are a statement of wishes to cover any kind of ill health situation. They are not legally binding, nor a guarantee that you'll have access to a full range of treatments.

Their forms were intended to be completed after discussion, both with the GP or health care professional and the patient's family, if they wish. They were originally available online and then the GP could download a copy to be completed when the patient attended the surgery, or was with the healthcare professional at a hospital appointment. This, of course, cannot now happen, so a paper copy was sent to me to complete after the GP had discussed it with me over the phone.

If you go to **www.resus.org.uk/respect** you will find all the documents and information there for you to download as you require, to help anyone start to consider these things. It is intended to put down clearly, in medical language, details of your considered wishes, that they may not be able to ask you at the time, or you may not be in a position to express.

Actually filling in the form was tricky because it is online and I had trouble using the format. Also, you cannot make any mistakes or corrections. All phone numbers and information must be presented clearly and correctly. I can see the sense in this, as any alterations may not have been made by the patient, which could alter their stated wishes. It has to be absolutely clear what your wishes are, so that there is no misinterpretation by a Health Care Professional who may only have seconds to assess it. It is not a guarantee that you will get treatment but that those having to make decisions know what you would prefer to happen. I have stated that my ultimate wish, at my age, is to survive even if that means reduced quality of life.

"I feel these forms are vitally important and am pleased that I have been asked to complete one. Ultimately, it may be worth all the frustration!"

Benefits Advice

Here is our view about GSD and claiming disability benefits such as PIP and DLA.

Since *Jane Lewthwaite* started as Care Advisor, she has had plenty of chance to work with members/patients with GSD. She says,

"GSD is often not well understood and can be difficult to explain in the simple terms needed for benefit claims.

Claims Assessors need to know only certain key facts about how people cope in daily life with a very particular set of tasks. It takes knowledge and experience to word claims clearly and frankly.

Most of the time patients cope daily and feel they are 'moaning' if they write about how truly difficult life can be for them.

I find people are not getting their point across and so having an external, neutral helper is most important".

Julie Jackson

Your benefit questions can be answered.

We recruited a part-time benefits advisor, *Julie Jackson*. She has 30 years' experience in the Citizens Advice Bureaux.

Initially, she helped with some overdue first appeals for PIP (called Mandatory Reconsideration).

She has completed PIP and DLA applications, as well as eight appeals. She has also completed two full benefit reviews for families, advised on ESA applications and she has also secured further help from a specialised cancer help charity for one family.

Do you have any questions about eligibility for benefits?

Julie has time to help you, email

info@agsd.org.uk

The Doctor Will Zoom You Now

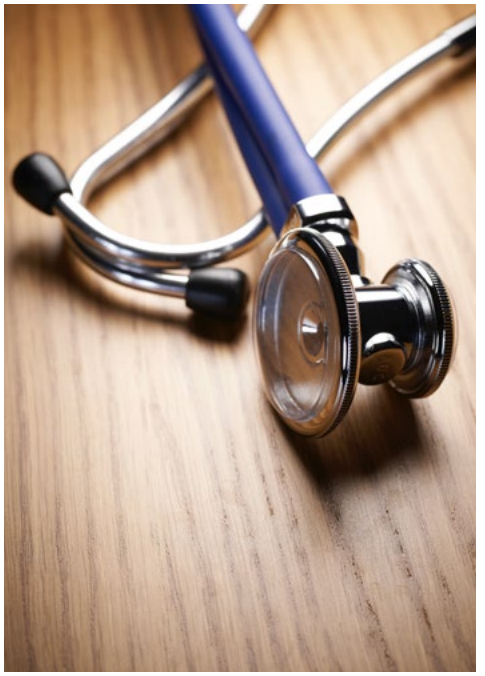
Matthew Arthur

The ongoing COVID-19 pandemic has provoked a big discussion in the media and general public about the necessity of travelling to work and appointments to be there in person. With all of the technology available that allows for online communication and collaboration, many of us are arguing that we should continue working from home as much as possible once the pandemic is over.

There are many obvious benefits such as reduced CO2 emissions, less traffic on the roads, and time saved from travelling. However other people would like to return to normality as soon as possible and emphasise the value of real-life human interaction both for mental wellbeing and as the most efficient and natural form of communication.

This huge debate going on the world right now is relevant even to our small GSD community, and gives us the opportunity for our patients and consultants to discuss to how they'd like to proceed with online consultations in the future. An interesting study carried out by Traverse, National Voice, and Healthwatch England gives a small picture of attitudes and experiences towards online consultations in the wider healthcare community (link provided below).

https://www.nationalvoices.org.uk/publications/our-publications/dr-will-zoom-you-now-getting-most-out-virtual-health-and-care



GSD as a Teenager

The importance of community

Bronte Thomas-Bush

Having a Glycogen Storage disease as a teenager can be a lonely and isolating experience. Whether you are diagnosed from a young age, or later in life, it affects everyone similarly when it comes to mental health. It's so essential having a concrete support group around you, more importantly – one that understands.

My experience with McArdle disease, or GSD5, was a challenging one when I was first diagnosed at age fourteen. But, I found an amazing group of people who shared my experiences, and helped me come to an understanding of my condition. My world of glycogen storage disease became a lot bigger and filled with those who understood exactly how I felt. Being diagnosed as a teenager, whilst now advantageous, was at the time one of the scariest moments of my life. Now, I find sharing these experiences so valuable to guide the youngest members of our community through growing up with the disease. I have found a family and my journey with McArdle's has become so much easier because of it. I now know the future is bright for me, and I am supported and loved with my condition.

Note that IAMGSD has a campaign to get the average age of diagnosis for GSD5 down to below 10 years.

Bronte, second left, and Team McArdle's on the 2019 Walking Course

What's new in GSD?

Hepatic GSD update

Hepatic Guidelines

The Hepatic Guidelines meetings have been facilitated by the AGSD-UK for two years now. They consist of doctors, nurses, researchers and dieticians from all the specialist centres across the UK. The aim is to collaborate and share information with the intention of furthering knowledge and promoting research. Currently oral drugs are being trialled for GSD 1b in the UK following discussions at these meetings and research findings from Belgium and Holland.

The May meeting was postponed due to Covid-19 but another one is planned. We will keep you updated on any more progress.

Research summary

Ailsa Arthur, Trustee

Imbalanced cortisol concentrations in glycogen storage disease type I: evidence for a possible link between endocrine regulation and metabolic derangement. *Rossi A et al. April 2020*

This research was carried out in Italy to evaluate adrenal cortex hormone levels in GSD1 patients.

The results showed impaired levels of cortisol which were higher in GSD1 and lower in GSD1b.

This study has allowed the authors to hypothesize a potential new therapeutic target for GSD1.

Treating neutropenia and neutrophil dysfunction in glycogen storage disease IB with an SGLT2-inhibitor. *Wortmann SB et al April 2020*

This work carried out across Belgium and The Netherlands has led the researchers to repurpose the antidiabetic drug, empagliflozin. Clinically, symptoms of frequent infections, mucosal lesions and inflammatory bowel disease resolved with no symptomatic hypoglycaemia seen. There is now further work being carried out in the UK following the Hepatic Guideline meetings facilitated by the AGSD-UK.

Infectious and digestive complications in glycogen storage disease type Ib: Study of a French cohort.

Wicker C, et al. April 2020

Glycogenosis type1b (GSD1B) causes not only hypoglycemia but also infections and "Crohn's disease like" inflammatory bowel disease (IBD) that can significantly impair patient's quality of life.

We retrospectively evaluated infectious and digestive complications in 9 French patients (3 girls, 6 boys) diagnosed at 0.8 years on average, with a mean follow-up of 19.1 years.

Infections occurred earlier than IBD, at mean ages of 1.7 and 3.8 years, respectively. The average number of acute hospitalizations was 0.7 per year due to infections or digestive symptoms. Clinical presentations allowed separating patients into mild (n = 5) and severe (n = 4) intestinal involvement.

Patients in the severe group had more serious digestive symptoms but also earlier neutropenia with a tendency to a lower neutrophil count (NC) during follow-up, and a higher number of acute hospitalizations due to digestive symptoms and infections.

Treatments included G-CSF and cotrimoxazole (n = 7), 5-aminosalicylic acid (n = 2), and a polymeric solution enriched in the anti-inflammatory cytokine TGF-β (n = 4, "severe" group), and immunomodulatory treatment (n = 1).

In conclusion, infections and IBD are rare but severe complications in GSD1B. Neutropenia tended to be more prevalent in the severe IBD group than in the mild IBD group. Dietetic treatment with specific anti-inflammatory solutions seems particularly appropriate in these patients.

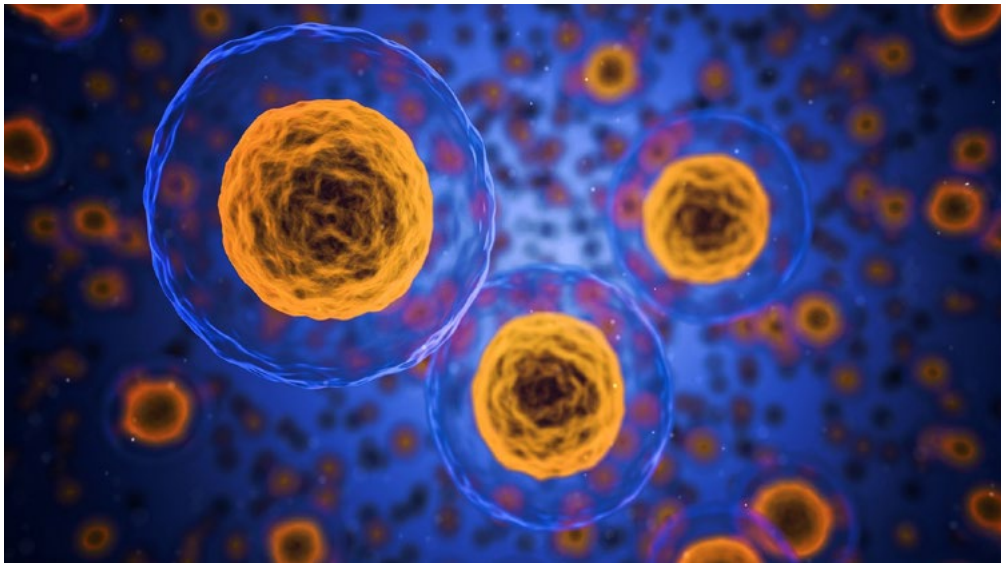
Glycogen storage disease type Ib: role of glucose-6-phosphate transporter (G6PT) in cell metabolism and function

Sim, Weinstein, Lee, Jun. November 2019

Summary and perspectives reprinted here

The biological role of G6PT in diverse cell types is being elucidated; G6PT mediated metabolism and intermediate metabolic products are critical for cell proliferation, differentiation, functions, and cell death. However, molecular mechanisms underlying the complex phenotypes observed in cells of GSD-Ib remain to be investigated. Moreover, considering that cells from different tissues or organs are confronted with a large variety of microenvironments and diverse metabolic fuels, it is possible that the role of G6PT may be sensitive to different microenvironments.

G6PT-deficient mouse model and cell lines are available, and their phenotypes should be characterized in detail, focusing particularly on metabolism and metabolic products. These studies may give valuable insights as to the development of novel therapies for metabolic and functional correction of cells, such as neutrophils and macrophages in GSD-1b.



Thank you and Adieu to Dr David Weinstein

We gathered messages of thanks and good wishes for a long-time friend and champion of Hepatic GSDs, David Weinstein who has moved to a new role. We created a special card, pictured top right.

He attended many conferences and was a focus of energy and knowledge.

He left his post at Connecticut Children's Centre at the end of July.

Photo of **Dr Weinstein** with **Bhuvan Prasad** at the AGSD-UK Conference, his family sent this message:

"We really admire all your dedication towards your work and providing brilliant support & guidance since 2008" from Bhuvan, Radha, Bhanu.

A few of the other messages sent to Dr Weinstein:

"Thank you for all you have done for us at the AGSD-UK. The time you have put in and the kindness you have shown to all our members will never be forgotten. Please keep in touch and all our best"

"Thank you, Dr Weinstein, for everything you have done for helping so many people. We all greatly appreciate it. I remember going to the conference in 2016 and meeting your team for the first time. Since then you have helped and changed my life so much and for that, I will always be grateful. Good luck and I can't thank you enough."

"Thank you for your dedication to Glycogen Storage Disease"

"You have given hope to many and thanks to you the future is brighter"

"Best wishes in all you do next"

Pompe Disease Update

Summary of Recent Research, Reports & News

Amanda Porter

Trial opens: Gene Transfer Study in Patients with Late Onset Pompe Disease

July 2020

Audentes is now enrolling patients in the Phase 1/2 clinical trial (FORTIS) to evaluate an investigational gene therapy product candidate in adults aged 18 years or older, ambulatory or non-ambulatory, with late-onset Pompe disease (LOPD).

Recruitment is currently taking place in the United States, with Germany and United Kingdom to follow.

This study will evaluate the safety and efficacy of an investigational gene replacement therapy, AT845, in adult subjects with LOPD. Subjects will receive a single dose of AT845 delivered via intravenous (IV) infusion.

Up to 2 nominal dose levels of AT845 are planned to be evaluated in FORTIS. A single AT845 administration via IV infusion is planned for each subject. The initial dosing cohort will receive a single dose at Dose 1 of AT845. The second dose cohort will receive a single dose at Dose 2 of AT845.

The core observation period will be completed by Week 48 for each subject. Subjects will be followed for a total of 5 years after administration of AT845.

A Phase 1 Study of the Safety of AAV8-LSPhGAA (ACTUS-101) in Late-Onset Pompe Disease (LOPD)

Dwight Koeberl, M.D., Ph.D
Duke University Medical Center, North Carolina, USA

The development of gene therapy has advanced to a point where reversal of the effects of Pompe disease can be foreseen. Pompe disease is a disease of the muscle resulting from acid alpha-glucosidase (GAA) deficiency in striated and smooth muscle*.

Despite the availability of enzyme replacement therapy (ERT) with recombinant human (rh) GAA, many patients have poor outcomes. The limitations of ERT have prompted the preclinical development of gene therapy for Pompe disease that more effectively corrects GAA deficiency in muscle.

Our strategy for gene therapy converts the liver to a depot for continuous secretion of GAA, accompanied by the receptor-mediated uptake of GAA in muscle. Furthermore, this liver-specific expression of GAA includes immune tolerance. Clinical translation of efficacious gene therapy will greatly advance treatment for Pompe disease by correcting GAA deficiency and suppressing immune responses against rhGAA.

*Striated muscle is composed of muscle fibres. Skeletal muscles are those that attach to your bones to allow your skeleton to move. Smooth muscle can be found around the hollow organs of your body, like your stomach and bladder. AP

High protein diet for Pompe disease? New trial opens

July 2020 Pompe disease weekly
Emily Malcolm, PhD

A small clinical trial at the University of Florida is recruiting 26 participants, aged 15 to 55, to test the effects of diet and exercise on Pompe disease progression. Patients will receive an individualised diet provided by a dietitian and an exercise plan created by a physiotherapist.

This trial is to gain evidence to support whether a high-protein diet, in combination with exercise, can reduce the symptoms of Pompe disease.

The aim of a high-protein diet is to reduce the amount of glycogen made by cells. By reducing the amount of carbohydrate intake, cells have less sugar available to store in the form of glycogen.

What is a high-protein diet?

A high-protein diet for Pompe disease consists of 25% - 30% protein, 30% -35% carbohydrates, and 40% - 45% fats. Protein from meat, fish, and eggs is better, in part because these foods are rich in alanine, an amino acid that plays a role in carbohydrate metabolism. Fats should include both omega-3 fatty acids (found in fish and supplements), and omega-6 fatty acids (found in olive oil, dried fruit, and cereals).

Carbohydrate consumption should be spread throughout the day and be in the form of whole grains, wholemeal pasta, and legumes, and not come from processed sugar found in sweets, fizzy drinks and desserts. Fruits are a good option because they also contain fibre, which can help with constipation a common symptom in Pompe disease.

Higher dosing of alglucosidase alfa improves outcomes in children with Pompe disease: a clinical study and review of the literature

Priya S. Kishnani MD et al. January 2020

Is Enzyme replacement therapy (ERT) with recombinant human acid-α glucosidase (rhGAA) at standard dose of 20 mg/kg every other week sufficient to halt the long-term progression of myopathy in Pompe disease?

A retrospective study was conducted on infantile-onset Pompe disease (IPD) and late-onset Pompe disease (LOPD) patients with onset before age 5 years, ≥12 months of treatment with standard dose ERT.

Eleven patients with IOPD or LOPD were treated with higher doses of up to 40 mg/kg weekly. There were improvements in gross motor function measure in 9/10 patients, in lingual strength in 6/6 patients, and in pulmonary function in 4/11.

Conclusion

Higher rhGAA doses are safe, improve

gross motor outcomes, lingual strength, pulmonary function measures, and biochemical markers in early-onset Pompe disease, and should be considered in patients with clinical and functional decline.

Respiratory Muscle Training in Late-Onset Pompe Disease: randomised controlled trial shows few changes

December 2019 Harrison Jones M.D., Ph.D.
Duke University Medical Center, North Carolina, USA

Persistent, progressive respiratory muscle weakness remains a primary obstacle to improving outcomes in patients with late-onset Pompe disease (LOPD). Respiratory muscle training (RMT) is an approach to providing resistance training to the inspiratory and expiratory muscles. 22 adults with LOPD and stable ERT were randomised to 12 weeks of RMT or sham-RMT.

The primary outcome was a change in maximum inspiratory pressure (MIP) in the treatment group and in the control group. Similarly, the change in maximum expiratory pressure (MEP) was similar in the treatment group and the controls. No statistically significant differences between groups were identified in other outcome measures including peak cough flow, measures of physical mobility, and diaphragm thickness and thickness ratio on ultrasound.

No significant differences between the groups were identified on exploratory measures from overnight sleep study or patient-reported outcomes of fatigue and sleep quality. However, patient-reported daytime sleepiness was significantly improved in the treatment group relative to control.

Why was this? There was greater than expected variability in responses for MIP and MEP in both treatment and control groups compared to the pilot data. Also, despite randomisation, subjects allocated to RMT were older, had been on ERT longer, and had greater respiratory muscle problems in comparison to those in the sham-RMT group.

Improved Muscle Function in a Phase I/2 Clinical Trial of Albuterol in Pompe disease – positive outcomes

March 2020 Dwight Koeberl, M.D. Ph.D
Duke University Medical Center, North Carolina, USA

For the albuterol group, forced vital capacity (FVC) in the supine (laid-down) position increased by 10 percent, and forced expiratory volume in 1 second increased by 8%; the 6-minute walk test increased by 25 meters; the Gross Motor Function Measure increased by 8% with the greatest increases in the Standing (18%) and Walking, Running, and Jumping (11%) subtests.

The placebo group demonstrated no significant increase in performance on any measures. These data support a potential benefit of extended-release albuterol as adjunctive therapy in carefully selected patients with LOPD based on ability to take albuterol on ERT.

Preclinical Studies for Oral –ERT of Pompe disease with a Tobacco Seed-Derived Recombinant Acid Maltase – ERT by tablet?

December 2019 Frank Martiniuk, PH.D.
JEM Group Inc.

The current approved enzyme replacement therapy (ERT) is via intravenous infusion of a recombinant human GAA (rhGAA) produced by CHO cells (Myozyme/Lymizyme, Sanofi-Genzyme) once every 2 weeks.

Although the current ERT has proven to be very efficient in rescuing cardiac abnormalities and extending life span in infants, the response in skeletal muscle is variable. In late-onset patients mild improvements in motor and respiratory functions have been achieved and the current ERT is unsatisfactory in the reversal of skeletal muscle pathology. Myozyme has been a wonderful first step, but it has revealed subtle aspects that must be addressed for successful treatment. Additional challenges for ERT include insufficient targeting/uptake of enzyme into disease-related tissues, poor tolerability due to ERT-mediated anaphylaxis, autophagic build-up, immunologic reactions and prohibitively high cost of lifelong. Hence, novel approaches for ERT are urgently needed.

Our objective is to develop an innovative and affordable approach for ERT via oral

administration (Oral-ERT) to maintain a sustained therapeutic level of enzyme on a daily basis to improve efficacy of treatment and quality of life for PD patients.

To this end, we hypothesized that tobacco produce rhGAA (tobrhGAA) can be ingested daily in a capsule that allows the maintenance of a therapeutic level of enzyme.

We have shown that torhGAA expressed in the seeds from transgenic tobacco plants is enzymatically active and can correct enzyme deficiency in GAA deficient cells and in vivo in disease-relevant tissues in GAA knockout (KO) mice-administered IP.

Long-term daily oral treatment showed increased muscle strength, tolerability, negligibility, antibody titers, decreased glycogen levels, increase GAA in tissues, long-term stability at room temperature, normalisation of spontaneous alternation/learning. There is support for proof-of-concept for Oral-ERT for Pompe disease.

Oral-ERT is an innovative approach that overcomes some of the challenges of Myozyme and might provide a more effective, safe and less expensive treatment.

Safety and efficacy Data using VAL-1221 in Patients with Late Onset Pompe Disease

October 2019 Priya Kishnani M.D.
Duke University Medical Centre, North Carolina, USA

Disease progression in Pompe disease, frequently associated with cytosolic accumulation of glycogen, is common despite conventional lysosomal-targeted enzyme replacement therapy. VAL-1221 is a fusion protein comprising the Fab portion of a cell-penetrating antibody utilising the nucleoside transporter ENT-2 to gain access to the cytosol, and recombinant human acid alpha glucosidase, which promotes lysosomal uptake via mannose-6-phosphate receptors. Thus, VAL-1221 targets both cytosolic and lysosomal glycogen.

In a 3-month controlled, dose-escalation study in 11 late-onset Pompe disease (LOPD) patients previously treated with Myozyme/Lumizyme at 20 mg/kg every 2 weeks for at least one year, we showed that VAL-1221 was safe and well-tolerated and that in comparison to Myozyme/Lumizyme, it provides largely dose-dependent improvements in motor, pulmonary and patient-reported outcomes, particularly in patients with no prior history of significant infusion associated reactions (IARs) while on Myozyme/Lumizyme.

Reprinted From Pompe Disease News

We chose this summary to highlight a very informative source of news for developments in Pompe disease.

Visit their website pompediseasenews.com

Behavioral, social & school functioning in children with Pompe disease

Aditi Korlimarla, Gail Spiridgliozzib, Mihaele Stenfanescua, Stephanie Austin, Priya Kishnani

Molecular Genetic and Metabolism Reports

Issue 25 Dec 2019.

Highlights

- Parents of children with Pompe disease completed standardized behavior checklists.
- Most children with Pompe exhibited age appropriate behavior and emotional functioning.
- Negative mood, learning, and attentional problems reported in infantile Pompe group.
- Negative mood and concerns about peer interactions reported in late-onset Pompe group.
- Most children with Pompe attend and succeed at school, with classroom accommodations.

IOPD: Consultation on Trial Design

Jane Lewthwaite

A pharmaceutical company approached AGSD-UK in May 2020 about getting together a small group of parents for a discussion about trial design for new treatments for Infantile Onset Pompe disease.

Jane gathered six families together and they participated in an online Zoom conference call, chewing over questions posed by the company and giving their opinions. All families benefited from the presentation and the chance to express their views.

Pompe Support Team Report

Telling you their own Covid-19 story

Pompe Support Team missed out on the annual training weekend in March, cancelled due to Covid-19. Also postponed was Ben's Stoke Social and two International Pompe Day meetings in April. PST sent out their newsletter as usual.

Gemma Seyfang ensured there were regular get-togethers on Zoom including hair-dressing tips from Becky Walton and a mental health support session from Dr Izzy Caller at Addenbrooke's Hospital.

"Although International Pompe Day (April 15) meetings were cancelled I wanted to be innovative and ensure we marked the day, so I invited everyone on Facebook across the world to send me their 'Isolation Inspiration' photos and comments or quotes of what they had been doing during lockdown. I wanted to show how we had stayed positive. I made a slide show link to video https://youtu.be/hRAaIz-VHR0 and released it on International Pompe Day. I also did a quiz that morning and we had a social, led by Ben, in the afternoon. I enjoyed hosting the quiz so much that I decided to do one for everyone weekly throughout the shielding period.

To be honest, I think being part of the Pompe Support Team has kept me positive and focused and it gave me something to enjoy and that held me together. I also really benefited from meditating at bedtime and my 13-year-old son Tyler even joined in"

Ben Parker

"Lockdown was a strange time, but playing with lots of clay and lots of zooming with friends and family helped break up the monotony.

Gemma's quizzes were great fun and it was nice to see old friends and see some new faces too.

Things are starting to happen again with me, my mental health booklet project is restarting and I'm going to get myself stuck into that over the coming months, (if you have contacted me, I will be in touch and if you want to help or have any ideas please get on touch with me on Facebook or to info@agsd.co.uk).

I'm hoping to plan a Stoke social for April next year, we will all deserve a drink or two by then, and I'm looking forward to seeing you all again soon."

Angela Biggs

"Covid-19 was a scary time because I had had a bad fall followed by surgery after breaking my leg. It was very difficult working out which members of my family were able to join me or take me to appointments, including x-rays.

My son is also in shielding as he is my carer so that was a problem. Having a second surgery recently was also hard. Hospital Covid-19 processes were odd, a nurse took my plastic gloves away from me and said that hand-washing was more useful.

After being in hospital for three weeks my hospital became a centre for treating Covid-19 so they 'facilitated' my return home rather speedily and was told I was going home and left within the hour. A friend came and did lockdown with us, that was really good".

Vicky Clarke

"It was nice having one to one time with my son, aged 11, who was home from school, and my husband who was off work most of the time, so I had all the support I needed and it was nice to live as a family for a while. For me, these are the positives. We had some exciting family news as well. I have not been in a shop since lockdown started, I drive my partner and stay in the car wearing a mask. Dr. Cole at Cardiff paused all ERT, including mine for six weeks but I restarted and everything is fine".

Donna Beresford

"Although not seeing my daughter for eight weeks was difficult, I actually enjoyed lockdown, pottering in the garden and I focused on eating healthily, protein packed recipes and not putting on weight. I was lucky because I am on a trial and it continued".

John Foxwell

"Covid-19 was fairly prevalent in my home area so I was very cautious.

Many people with Pompe disease struggle with incontinence so I re-drafted an alert card from the British Bowel and Bladder Foundation and it has been produced for this issue of Glisten.

The cards are now printed if you want one please email ***info@agsd.org.uk****"*

Semi-independent infusions

Jackie Hicklin's Story

Jackie has recently begun learning how to become semi-independent with infusions. How has she been getting on?

Jackie was completely dependent on a nurse for her infusions for 10 years. Early 2020 she was offered the opportunity to learn to be semi-independent. Her doctors explained how easy the process actually was and assured her that she would be fully supported in the training. She was due to begin this training a week before Covid-19 lockdown was enforced and so the process was rather delayed and extended. In total, because of lockdown restrictions, the training process took 3 months, to ensure that Jackie was doing everything safely and properly. She had fortnightly sessions with her care team, during routine infusion home visits.

Prior to becoming semi-independent, Jackie would spend a total of 5 hours with the nurse, once a fortnight. Now, that time has been cut down to 2 hours. The nurse cannulates but now Jackie sorts the pump and infusion herself. Jackie views the main advantages to being semi-independent as having a massive sense of pride that she is able to take care of herself. She also said it allows her to be more flexible with her time and allows her to have a slightly more normal life. Jackie also enjoyed the training and process of becoming more independent and would recommend it as something to think about to anyone who is curious.

Note: We understand independent infusions are not a viable step for everyone to take for many reasons, but felt it was important to celebrate how well Jackie is doing.

Congratulations Jackie

McArdle Disease Update

The latest from the world of McArdle's

Andrew Wakelin, McArdle's Coordinator

Moving My Muscles Month

In celebration of 10 years since the "Walk over Wales" (WoW), Andy Williams instigated Moving My Muscles Month (MMMM). It ran from 2 July to 2 August (as had WoW), and IamGSD promoted it to encourage McArdle people from around the world to make an effort to improve their aerobic fitness during the month. At present increasing aerobic fitness is the best treatment for McArdle disease. Some people walked, ran, swam, rowed, even used their treadmills and exercise bikes.

Covid-19 cancels walking courses

We hung on quite late hoping that the Covid-19 restrictions would be lifted enough to enable us to run the walking course and the Children & Parents event. However, finally we had to give in as the social distancing remained at 2m in Wales, and gatherings of people from multiple households in one accommodation was still not allowed. We usually have many participants from overseas and the travel restrictions were prohibitive for them. We also had feedback that people were finding it difficult to get time off work after all the disruption to their employers, and of course many people had financial constraints if they had not been able to work and not been furloughed. It is particularly sad for these annual events to be cancelled as this was their 10th anniversary year, but we are positively looking forward to getting back on track in 2021.

Video of German walking course

Alfred Ziegler and Monika Weingartz of the German GSD association organised their first walking course in September 2019 and I went over to lead walks and run tutorials.

Alfred has made a video from the many photos taken during the week and this is being made available on our YouTube channel GSDscreen.

Pictures from top: A drone view of the walking course group in 2019

Collator of Glisten, Harriet Thomas-Bush & Jeremy Michelson, GSD 7 on the Walking Course in Wales, 2019.

Happy faces on the first walking course in Germany.

Online consultations with McArdle clinic

Due to the Covid-19 pandemic the normal appointments at the national McArdle clinic in London had to be abandoned due to the risk of infection. Most appointments were then carried out remotely by phone or online video. They are hoping to return to normal clinics during the autumn, but of course this is subject to the course of the pandemic. For the future, the clinic is also investigating the possibility of an ongoing series of online group sessions, along the lines of a webinar each with a member of the clinic's multi-disciplinary team.

A number of research projects had to be suspended due to the pandemic. Hopefully these will resume in the not too distant future.

Euromac and CoRDS muscle GSD registries

Hopefully most people in the UK with McArdle disease and the even the more rare muscle GSDs will by now be included in the Euromac Registry. IamGSD wanted to get the large number of McArdle patients in the US included in the registry, but it was impossible due to the lack of clinicians with sufficient interest and commitment (Euromac is a a clinician-entered registry).

As an alternative, IamGSD has worked with Sanford Research to establish a patient-entered muscle GSD registry within the CoRDS platform. The collated data has been made as compatible as possible with the Euromac Registry in order to have as large as possible a resource for future research and trials. Details of both registries and links to them can be found on the IamGSD website. If you are in the UK but do not attend the McArdle clinic so are not on Euromac, please register with CoRDS.

Sydney & London paediatric series and survey

The metabolic department of Westmead Children's Hospital in Sydney, Australia, and the McArdle Clinic in London are collaborating to report on their joint cohort of paediatric cases. This will be the first such report and an important step forward.

IamGSD has an objective of reducing the average age of diagnosis of McArdle disease to below age 10. As part of their campaign to achieve that their President Stacey Reason is developing a survey of the experience of children with the disease and their path to diagnosis.

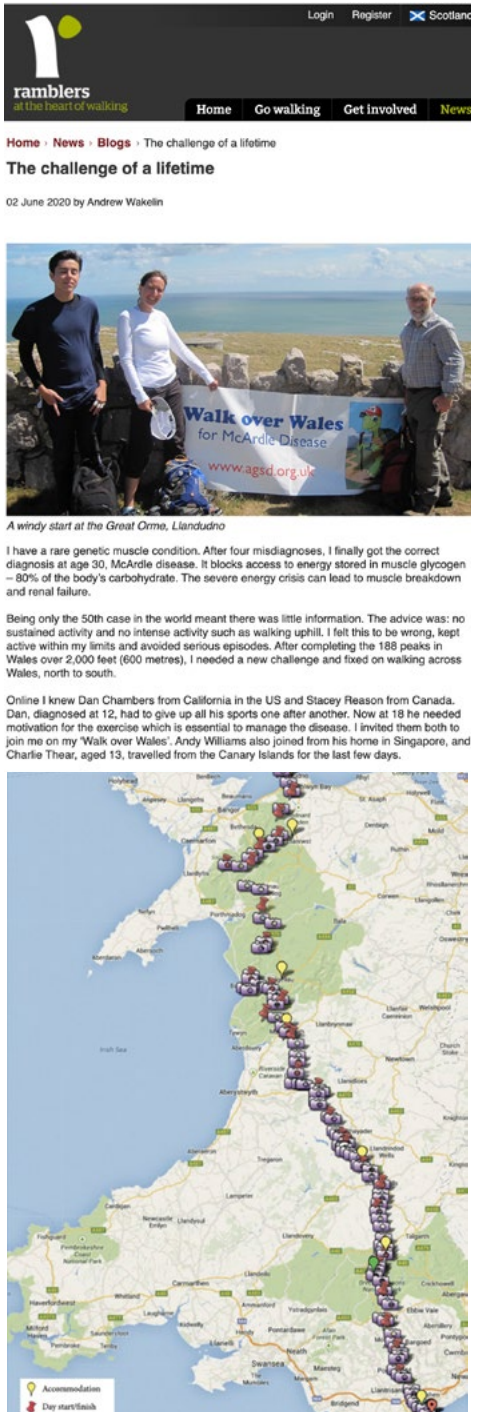
It is hoped that bringing these two initiatives together will help raise awareness in the medical profession of the signs and symptoms which could lead to a diagnosis of McArdle disease at a much younger age than has typically been the case in the past.

Awareness raising through the Ramblers Association

Under the heading "The Challenge of a Lifetime", I wrote the story of my McArdle's diagnosis, the "Walk over Wales" and how it had led to the inception of the Walking Courses. The story was featured in a newsletter sent to members of the Ramblers Association in June, with a link to the full story and a few photos on their web site. With Ramblers having over 100,000 members this may well have raised some awareness.

You can read the full story here:
tinyurl.com/y6rhdkaj

ramblers
at the heart of walking



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The challenge of a lifetime

02 June 2020 by Andrew Wakelin

A windy start at the Great Orme, Llandudno

I have a rare genetic muscle condition. After four misdiagnoses, I finally got the correct diagnosis at age 30, McArdle disease. It blocks access to energy stored in muscle glycogen – 80% of the body's carbohydrate. The severe energy crisis can lead to muscle breakdown and renal failure.

Being only the 50th case in the world meant there was little information. The advice was: no sustained activity and no intense activity such as walking uphill. I felt this to be wrong, kept active within my limits and avoided serious episodes. After completing the 188 peaks in Wales over 2,000 feet (600 metres), I needed a new challenge and fixed on walking across Wales, north to south.

Online I knew Dan Chambers from California in the US and Stacey Reason from Canada. Dan, diagnosed at 12, had to give up all his sports one after another. Now at 18 he needed motivation for the exercise which is essential to manage the disease. I invited them both to join me on my 'Walk over Wales'. Andy Williams also joined from his home in Singapore, and Charlie Thear, aged 13, travelled from the Canary Islands for the last few days.

Children's Section

Hope Travels is wonderfully organised by Julie Garfield in the UK & Mark Nicholson in the USA
Contact them on Facebook at Hope Travels Pompe Awareness
www.facebook.com/groups/1668559669887831

Hope Travels, a baby sloth, and **Cori the cat** are two mascots sharing fun for children and raising awareness of rare conditions.

PRIZES GALORE!

Good luck and HAPPY QUIZZING

Hope Travels is a friend to everyone with Pompe. On her journeys around the world she meets and greets, bringing friendship all the time. She shares a message of fun and staying strong in the face of Pompe. **Hope Travels** supports the campaign for New Born Screening.

summer quiz

If you or someone in your family has a GSD and you are under 18 years old, you can complete these quizzes, tear them off and send it to AGSD-UK.

Please include your name, address, your parent/adult's email address or phone number and send it to PO Box 1232, Bristol, BS48 9DD

CLOSING DATE 15TH SEPTEMBER 2020

Meet Cori the Cat...

Cori is a super hero. He travels around the world helping children and making them feel better. He is everyone's friend.

These are some countries Hope Travels has visited. Fill in the blanks.

1 C_l_m_ia
2 _m_ri_a
3 _anada
4 _h_llip_n_s
5 _n__on_si_
6 F_a_ce
7 S_ain
8 _er_any
9 _or_ ay
10 B_a_i_

Tie Breaker: complete the sentence below:

I am hopeful because.....

CORI'S CORNER BOOK QUIZ

1. Which author's first story for children was *The Gremlins*?
A) Roald Dahl ◯ C) JK Rowling ◯
B) Philip Pullman ◯ D) David Walliams ◯

2. In *James and the Giant Peach*, what provided the power for the peach when it floated through the air?
A) Balloons ◯ C) Seagulls ◯
B) Dreams ◯ D) Clouds ◯

3. Where is *Paddington Bear* from?
A) Quebec ◯ C) Madagascar ◯
B) Peru ◯ D) Scotland ◯

4. What is the *Gruffalo* scared of?
A) A snake ◯ C) A bear ◯
B) A mouse ◯ D) An ant ◯

5. What is the name of *Matilda's* teacher?
A) Miss Jam ◯ C) Miss Toast ◯
B) Miss Honey ◯ D) Miss Nutella ◯

6. In *The Cat in the Hat*, what type of pet does Sally have?
A) Cat ◯ C) Fish ◯
B) Dog ◯ D) Giraffe ◯

7. What kind of hat does Max wear in *Where the Wild Things Are*?
A) Crown ◯ C) Builders Hat ◯
B) Cowboy hat ◯ D) Top Hat ◯

8. Who is NOT a resident of the *Hundred Acre Wood in Winnie-the-Pooh*?
A) Owl ◯ C) Owl ◯
B) Badger ◯ D) Donkey ◯

Tie Breaker: complete the sentence below:

I like reading because.....

... carry on on another sheet of paper. You might win a prize!

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AGSD UK
Association for Glycogen Storage Disease

TAKE AWAY FROM THIS GLISTEN...

UPCOMING EVENT!

Our brilliant Finance Manager, and now also the Website News Coordinator, **Jackie Henson** is pictured with daughter **Rosie** as they prepare a sponsored Sleepover Camp in aid of AGSD-UK

- PLEASE help us with fundraising and donations
- SIGN the petition for new born screening
- CHECK out our social media and website. Do you want to get involved? Let us know!
- PHONE us for advice on completing benefit forms before you apply. We're here to help.
- CHILDREN, complete the quizzes & send them to us to win prizes!

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AGSD UK
Association for Glycogen Storage Disease