August 2021

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

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

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August 2021

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, clockwise from top left:
Flo Osborne and her teammates completing the 3 peaks challenge, ***Phil Sharman*** running in the London Landmarks Half Marathon, ***Nikki Christie*** birthday fundraising with friends, ***Kieron Stubbings*** running in the London Landmarks Half Marathon, ***Nigel Walker*** during his training for the marathon, and ***Remi*** & his daughter ***Olani***

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.

*0300 numbers are charged as 01 & 02 numbers

Contents

Message from the Chairman

Despite the events of the last 18 months, life at the charity has been busier than ever. Enquiries and requests have risen by 40%. This has been offset, sadly, by the necessary lack of face to face contact. Nevertheless, staff and volunteers have risen to the challenge with additions in personnel and changes in roles.

We want to welcome to the charity, Elizabeth Davenport, who will be working alongside current staff as a freelance Specialist Care Advisor. Her profile is in the magazine for you to see. Elizabeth is very experienced and has the wonderful qualities of warmth and an ability to listen. We know she will serve our membership well.

Jackie Henson, a well-known and trusted face will continue her work as Finance Officer. Alongside that, she has taken on the extra duties of the website and database administration. She is looking forward to these extra responsibilities.

Vicki Lucass, who joined us last year and put together the immensely successful 'Winter Season' will be continuing in that that role. She stayed with us after the season finished, working alongside Jane Lewthwaite, on projects involving the Pompe community. She will be continuing in that role, as well as taking on next year's 'Winter Season'.

We want to mention Harriet Thomas-Bush who has also contributed by becoming involved in fundraising and editing. Thank you, Harriet. We look forward to working alongside you more.

For the second year running, there will be no face to face conference, but we are holding our breath, and hoping as soon as we are able, to arrange face to face family days.

Fundraising by members and volunteers has been exceptional. Our thanks to all of them, whose tales are told in the magazine. Relationships with our current industry partners, who support our work for the community remain close. In the past year we have also been fortunate in developing relationships with a wider diversity of newer companies. We have enjoyed working with all of our industry funders, old and new, all with the intent of serving and caring for the glycogen storage disease community.

Nick Jones Chair of Trustees

Trustees

Welcome to new Trustee, Florence Osborne.

Flo became a member of AGSD-UK in 2020 when her son Hugo was diagnosed with GSD 3b at 12 months old. She joined the trustee board early 2021.

Flo is a Chartered Psychologist working as a Senior Lecturer in Exercise Psychology at Loughborough University, School of Sport, Exercise and Health Sciences. Her research interests focus on the psychology of exercise behaviour and how sport and exercise can support those with mental health problems. She hopes to bring her academic and research expertise to support the GSD community in addressing important and relevant research questions.

She loves all sport and has a passion for fundraising – often combining the two. She, with friends, recently completed the *National 3 Peaks Challenge* to raise money for AGSD-UK.

We say goodbye to Trustee, Rob Seabourne.

After just over three years as a member of the Board of Trustees at AGSD-UK, I have taken the decision to step-back from my role, and from the charity.

This is not a decision I have taken lightly, but various personal and work related circumstances have culminated to dictate the decision. While my time spent at the charity has come with its challenges, particularly those we have faced within the last 18 months to 2 years, I can genuinely say it has been a huge honour and privilege to help navigate the charity through these times.

The people, the community and the culture that make up AGSD-UK are very special and are an enormous influence on the continued success the charity has and will continue to have. It is a brilliant charity, doing some excellent work for some truly special people. *Rob Seabourne*

Thank you to Rob for all his help, support, and contribution to the charity over the last few years. We wish Rob good luck in his future endeavours.

WHO ARE WE?

With so much change and uncertainty over the past year and a half we thought we would introduce you to some of the team that make up AGSD-UK and what they can do to help you.

Andrew Wakelin
GSD5 Volunteer & McArdle Disease Co-Ordinator

Finally getting a correct diagnosis at 30, Andrew had no help for 20 years until he found AGSD-UK in 1999. He helped develop the UK McArdle Clinic, became AGSD-UK's McArdle's coordinator in 2004 and Chairman from 2009 to 2014. He developed the association's governance, database, and current website.

He is on the board of ***Euromac***, is a founder of ***IamGSD*** and contributes the patient perspective to research projects. Andrew has produced many educational videos and his "101 Tips" book and other publications are in 8 languages.

In business, Andrew ran graphic design and software companies. Now well past retirement, he works hard to help McArdle people around the world improve their lives.

Andrew set paragliding world records in 1984 and was on the UK governing body. Despite McArdle's Andrew has climbed all 188 Welsh mountains over 2,000 feet, plus iconic mountains such as Kilimanjaro. He organised the ***Walk over Wales*** event in 2010 and has subsequently led annual walking courses for McArdle people from 18 countries.

Elizabeth Davenport
Specialist Care Advisor

Welcome to Elizabeth who will be working as a freelance part-time Care Advisor for this wonderful community and AGSD-UK staff team. Elizabeth started working within the charity sector in the 1990's. She has supported babies, children, young people and adults with complex disabilities and sensory impairments.

Her passion has always been to empower people to have the best quality of life and enjoy their local communities and services without comprising on choice and

experience. 10 years ago, she started to work with Niemann-Pick UK. Niemann-Pick Disease is a rare metabolic condition. Her role was to offer specialist non-clinical support, information, and advocacy, and is hoping to support the AGSD-UK community in a similar way. She looks forward to speaking to you all in the future and of course if you need anything then please get in touch with her.

Jackie Henson
Finance Officer

Jackie has been involved with AGSD-UK since 2013, initially as the bookkeeper but now she also assists with website updates, membership, general admin, and conference planning. She is a member of the Institute of Certified Bookkeepers (ICB) which helps to keep her current with bookkeeping requirements and legislation. Prior to bookkeeping Jackie was an engineer and had 10 years serving with the Royal Navy in various war zones, followed by 10 years at Motorola dealing with emergency services communications equipment. Outside of work Jackie is a volunteer Girl Guide leader for her local Guide unit and a taxi driver for her two dance-mad daughters.

Jane Lewthwaite
AGSD-UK Consultant and Pompe Advisor

Jane has been with AGSD-UK for well over five years. She started by developing the Care Advice Service for patients. She has developed the patient community voice and established links with Health Care Professionals, Specialist Centres, and charity networks too. Jane has provided input to research, trial design and forged strong links with Industry. She has been the connector and pivot for AGSD-UK in the last few years. Jane enabled the creation of informative materials; ***Pompedoo, 101 Tips for GSD3, Pompe Medical Overview, Falls Awarenes***s and ***Mental Health*** booklets.

Jane works on enhancing charity governance and policies including DBS checks, safeguarding, lone working and training for Trustees and Volunteers. She co-ordinates the Pompe Support Team and enables a raft of national and regional meetings on Pompe disease themes for patients and professionals. Jane curates the Glisten newsletter and has helped develop our website and database too.

Jane is our charity advisor and continues to invest in the UK Pompe community as well as providing oversight for other staff.

Julie Jackson
Benefits Advisor

Julie Jackson has 20 years' experience in the debt and benefit advice field and has been employed by Citizens Advice Bureau for 14 years. Before working for citizens advice Julie worked for the mental health charity Mind and this is where her passion and career journey started.

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

Lisa Massimo
Social Media Volunteer

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.

Lisa works with the AGSD-UK team to try and keep the GSD community informed on the latest research and what is happening within the community such as fundraising campaigns.

Thank you again Lisa for helping build this online community.

Vicki Lucass
Season Producer &
Charity Co-ordinator

Vicki is an ex Synchronised Swimmer who competed at an elite level for many years and went to the London 2012 Olympic Games. Since then, she has completed a degree in Sports Science and works as a Fitness Manager and Personal Trainer.

Vicki started working with AGSD-UK in November 2020 when she was brought in to work on the *Winter Season* of online presentations when we were unable to run the annual conference due to Covid-19. We hope to run a similar event in January 2022.

Vicki has since taken on a new role as Charity Co-ordinator which will see her get more involved in the charity by leading and organising projects for AGSD-UK.

New GSD1 Emergency Cards & Leaflets

We are currently in the process of creating some GSD1 Emergency Cards and Leaflets. The cards will include the important information for type 1a and 1b to use in an emergency. Our leaflets will be a more in depth information source for you to read and use but may also help others understand your GSD1.

Please look out for further information about these on our website and social media. **We will notify you when the Emergency Cards and Leaflets are being published.**

Report from the HTAi Annual Meeting

June 2021 (online)
Report by Luke Fraser

Health Technology Assessment International (HTAi) is a global scientific and professional society for those involved in HTA to support optimal policy and decision making.

The 2021 Annual Meeting was initially intended to be held in Manchester but was ultimately switched to an online event. Comprising three days of meetings with a further two days of workshops, there was a multitude of speakers and presentations to choose between.

I attended a presentation by Ray Milman of the University of Health, Israel, who discussed an upcoming publication on providing an evaluative framework for the development of new or early-stage healthcare technologies.

He discussed the process of collaboration between health organisations, policymakers and tech start-ups, and emphasised how productive collaboration between the three is key to developing the most effective new treatments.

The Framework itself aims to structure an analysis across a range of categories including a range of social and practical considerations alongside a traditional cost-benefit approach and should hopefully lead to more informed decision making in this area.

Meanwhile Mark Rasburn, a senior Public Involvement Advisor from NICE, gave a talk on establishing meaningful patient and public involvement in HTA. NICE has been keen to involve patient Stakeholder groups in the 'scoping' process for new HTAs from an early stage (AGSD-UK being amongst these.)

He described how it is important for HTA organisations to involve stakeholder groups in such a way that is not simply 'tokenistic but adds value'. He stressed that designing meaningful approaches to patient and public involvement must be about effecting change and practically removing barriers to involvement, rather than simply analysing them. With this is mind, NICE has undertaken a range of measures to further involve and support patient organisations both large and small and is continuing to review its practice.

Also representing NICE, Andrew Kenyon, Michael King and Charlotte Downing gave an analysis of the NICE Technology Appraisals process. This process was updated in 2018 with a view towards issuing more robust guidance as close to the point of a drug's marketing authorisation as possible. The effect has been to ensure sufficient evidence is available when required in order to give the best chance of making positive recommendations, alongside reducing assessment delays.

Meanwhile Karen Macpherson, from Healthcare Improvement Scotland gave a presentation outlining a review of patient and public perspectives on cell and gene therapies. She discussed how the review was undertaken in order to address the 'unique and complex challenges of bringing these therapies to patients.'

Thirty-five studies, mostly from North America and the UK were included. Outcomes revealed varying levels of knowledge and understanding among patients, with limited research relating to members of the public.

Levels of acceptance of cell and gene therapies among patients varied, but after information on therapies was supplied, acceptance levels were generally fairly high. Patients generally wished for more, particularly personalised, information, and the review also concluded that campaigning groups and political events can also exert an influence on public perception of such therapies.

Gene & Cell Therapy Research under Horizon Europe

Janka Mátrai, PhD
Scientific Project Advisor for the European Research Council Executive Agency

Europe's newest seven-years funding programme for research and innovation, called *Horizon Europe*, has just started in 2021. While it strongly builds upon the previous framework programs, it has some important novel features. Much more pronouncedly than before, it aims to strengthen the impact of research and innovation. In line with this aim, among its new instruments one can find the *European Innovation Council (EIC)*, which is an agency supporting innovations with potential breakthrough or disruptive nature that may be too risky for private investors.

By its nature, gene and cell therapy research is situated at the crossroad of many types of research, being it basic, frontier, disruptive, translational, clinical, applied or industrial.

This unique characteristic makes it ideally suited to many of the funding modalities, instruments and actions the EU programmes offer. *Over three thousand projects* have already been funded by the various instruments, and if we look at the single principal investigator driven frontier research grants of the *European Research Council (ERC),* over a hundred of groundbreaking gene and cell therapy research projects were funded so far.

Because of the vast amount of knowledge that has been built up in the field, in the labs as well as in the clinics, because of the truly paradigm shifting *success stories*, and because of its huge innovation and investment potential, the first *workshop* organised jointly by the EIC and ERC was held, this year, on gene and cell therapy research.

There were four thematic sessions covered: Cell therapy, including stem cell therapy; Disease modelling including 3D-bioprinted organs/ organoids and other regenerative medicine novel approaches; New vectors (viral and non-viral) for gene and cell therapy; and Novel gene therapy approaches including RNA-based technologies. The speakers were prominent representatives of the gene and cell therapy research and investment world. The event was concluded by a round table discussion on the transition from research to innovation in gene and cell therapy.

The take home message was well articulated, the European research teams have all the potential to bring their excellent science to the market for the benefit of many patient groups. However, a shift in attitude towards higher flexibility in risk taking, tolerance of failure, easier access to venture capital, easier communication and networking among the stakeholders, reduced bureaucracy and better organised facilities are necessary to make this next step.

These changes must happen to reduce the time spent on the transition from the bench to the bedside and to prevent stress building up in the research communities due to the present unnecessary hurdles, and must happen at all levels, starting with the individual, continuing with the leaders of our research institutes, universities and ending with the policy makers and our politicians. And we, the patients and their families, we have to continue to be around too, we need to make sure that initiatives do not settle into standstill, but turn into continued progress until all *orphan* and *rare diseases* are cured.

Fundrasing Round-up

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

- Thanks to the Bedford Park Lodge Masonic Hall who donated **£500**

Nigel Walker

I had always wanted to do a Marathon and decided in October of last year to enter the ballot for London being held in October. Not thinking I would get in as its very difficult to get a ballot place, I was happily surprised when I received the email to say I had been successful.

The training is going very well. As of the start of July, I have been heading out for three runs a week: 6-8 mile fast runs on Monday evenings, hill Sprint Sessions on Wednesday evenings and long distance runs at the weekend which I use to increase my mileage (currently up to 18). Most routes around Bourne are hilly so I am hoping this will help when the time comes to run the marathon in October. The running club which I am part of has been brilliant for me as other members have been recommending training plans and different routes to add some variety to the training.

I am proud to be running and fund raising for AGSD-UK as they have been a great source of information for me and my family.

https://tinyurl.com/bn35n2m6

£447.50 RAISED

Flo Osborne and friends raised £18,986 in their *Hike for Hugo*

£18,986 RAISED

It was a pleasure to raise money for AGSD-UK. All our friends, families and colleagues have been incredibly generous. We have raised £18,986 in total which is beyond all of our expectations.

The challenge itself was amazing – so many great moments. Walking through snow on Ben Nevis, watching the sun rise at the top of Scafell Pike and the moment we finished at the bottom of Snowdon in under 23 hours (the target was 24 hours).

Our little Hugo inspired us all the way through the training and up and down each mountain. Hopefully we showed some of the strength and resilience that he shows us every single day.

https://tinyurl.com/cdyphthy

Phil Sharman has raised £1357

I chose to run and fundraise for AGSD-UK in the London Landmarks Half Marathon. This wonderful organisation has helped my mum understand the rare genetic Pompe disease that she has suffered from for the last 4+ years.

This horrible disease makes everyday tasks extremely difficult. It is hard to believe that getting out of a chair or climbing some stairs are things that have become extremely challenging on a daily basis over the years for my Mum. I trained for quite a long time for this run with the event cancelled and dates changed a few times over the past year for obvious reasons. I achieved my goal of a sub 2 hour half marathon and completed it in 1 hour 56 minutes! This was the longest I have ever run and a personal achievement with the added bonus of a chance to raise some money for a great cause.

https://www.justgiving.com/fundraising/phil-sharman

Is it your birthday and you want to raise money for AGSD-UK? Did you know that you can fundraise though Facebook?

Sinead and Nikki have been doing just that!

- Sioned Wyn
  Total money raised: **£60**
- Nikki Christie
  Total money raised: **£269**

If you want to fundraise on Facebook but not sure how? Contact **info@agsd.org.uk** and we can show you what to do!

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

Remi's Challenge

Remi and his wife Mirian, who live in London, are parents to their one year old daughter Olani who was diagnosed with Pompe Disease.

Remi will be taking part in a challenge to fundraise for AGSD-UK. He will be cycling from Lands End to John O' Groats, starting on his Birthday, September 16th, 2021. Friends will join him on the trip to film and take photos so he can document his journey.

Remi is currently in the process of putting together his fundraising and project plan and will be creating a website for people to visit to find out more information about his challenge and why this cause is so important to him and his family. The website will advertise his route and will enable people to follow his cycle online.

Keep an eye out on our social media for further information and updates on ***Remi's Challenge*** as well as the fundraising link to donate should you wish to!! He would love to meet some families and adults in the AGSD-UK community along the way, so if anyone would like to arrange to meet him and his team, please email info@agsd.org.uk who can put you in touch with Remi's support team.

Just Keep Running Kieron!

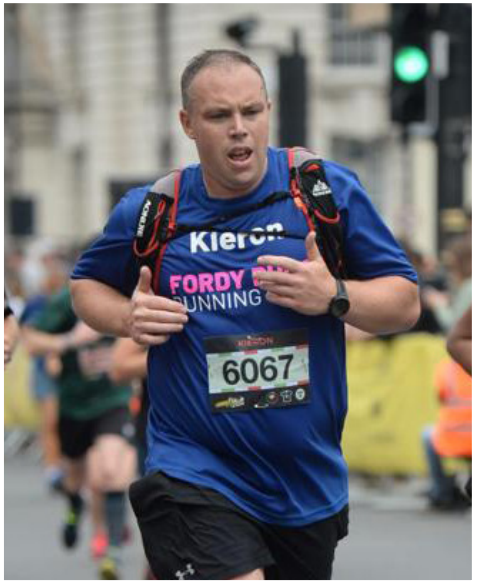
Kieron has been back running again, this time in the *London Landmarks Half Marathon 2021*!

His training for this run took a hit as he was isolating due to family members having Covid, but he ran a great race and finished with a personal best time of 2 hour 5 minutes!

Keiron has got a busy year ahead with lots of races planned such as the *Vitality Big Half* in London in August and the Chelmsford half in October. Also, in October he will be running in the Manchester Marathon!

These runs will set him up brilliantly to achieve his current goal to fundraise and run in the London Marathon 2022.

Good Luck Kieron!



Reggie's Gift UK

Liz & John-Paul Renouf in the memory of Grandson Reggie have raised £3312 so far.

Reggie's Gift has been founded in loving memory of Reggie Gordon Renouf, who sadly died in November 2019, at the age of two, from Infantile Onset Pompe Disease. It's mission is to support and enrich the lives of families who have a child with Infantile Onset Pompe Disease, by providing financial aid which is flexible and unconditional.

Reggie's parents, Tess and Chris who live in Australia, experienced the financial and emotional strain that comes with caring for a child with a life-threatening illness. They have had the opportunity to establish a program, in collaboration with the Australian Pompe Association, to help Australian families facing similar hardships in the future to access funds from Reggie's Gift.

Reggie was a beautiful, shy but determined, brave little boy who had to battle with his deteriorating abilities and functions as his illness took hold.

He adored watching cartoons, cuddling up with his four-legged friends and spending time with his family. He was very much loved by his dear mum and dad, Tess and Chris, and by all his family. Despite his progressive weakness and loss of physical ability, he enjoyed as happy a life as he possibly could during his short time with us.

During the course of his illness, Reggie received expert care from the Royal Children's Hospital in Melbourne and Very Special Kids, a children's hospice, in particular from the Metabolic and Palliative Care teams. He touched the hearts of all who knew him. The support Reggie and his family were given was invaluable & they wanted to do

something to help other families affected by Pompe.

Reggie's Grandparents, Liz and John-Paul who live in England, also felt compelled to help where they could, so they took on the challenge to walk 10,000 steps every day for 40 days during Lent to raise money to support UK families who have a child with Infantile Onset Pompe Disease. This money has helped to establish a Reggie's Gift UK Fund.

One grant has already been made from this fund to a family which has already helped them a great deal.

In April 2021 this family got the news that their 5-week-old daughter has Infantile Onset Pompe Disease. Their daughter had had a series of chest infections and was diagnosed after a cardiac scan highlighted myopathy. This was a very early diagnosis, and the baby was admitted to Manchester Children's hospital with Mum straight away. She was able to start enzyme replacement therapy within a few weeks and is currently doing very well.

She spent 7 weeks in hospital. A grant of £100 from Reggie's Gift UK was given to them within 48 hours, as they needed immediate help with transport costs so Dad could come to visit his baby and wife. They have now had other grants and can apply for more from Reggie's Gift UK if needed.

Thank you to those that have donated in remembrance

We thank those who have raised money in remembrance of lost loved ones. This money will go far in helping others and their families living with GSD.

Paul Beckett

Margaret Beckett has been raising money in memory of her son Paul who had GSD3; she has raised **£275.00**

Margaret raised money at a car boot sale in Torksey with her friend Bob to help families with children affected by Glycogen Storage Disease. They had a lot of fun in the process!

Aldwyn Cooper

Tributes have been paid to Aldwyn John Richard Cooper who died on 4th February 2021. Aldwyn is much loved and will always be remembered.
"Aldwyn was such a kind and caring person. He gave his time so freely to join meetings as a patient representative nationally and internationally. Only last August he joined a meeting of the *Medicines Health Regulatory Authority* trying to help the cause for new treatments for Pompe disease. He always had an interesting story to tell and was especially proud that his diagnosing neurologist Dr Jarman participated in the TV programme, *Diagnosis Detectives.*

Aldwyn also helped a homecare company create a video about having ERT infusions at work. He always wanted to help others gain better understanding. Aldwyn will be very sadly missed."

AGSD-UK Specialist Care Advisor
Jane Lewthwaite

AGSD-UK would like to say thank you to all that have donated in Aldwyn's memory.

To date £710 has been raised via the obituary website Beyond Life.

GSD Survey Research Study

By Sinead Smythe

Sinead Smythe is a health student who has recently conducted a research project on Ketotic Glycogen Storage Diseases. She has kindly written up a summary review of what she found. AGSD-UK connected with Sinead and supported the research by circulating to members by email and social media. We always want to support, not only families, but research too, and here is a parent doing their own piece of research. Sinead has a child that has GSD9 so this is a topic that is extremely important to her.

In my research paper I discussed the ketotic types of Glycogen Storage Disease in children between the ages of zero to eighteen. The reason for choosing this research was that it seemed an interesting topic and a rare condition that is not well known. I collected information through questionnaires from secondary and primary research which led me to be able to discover the strengths, limitations and improvements that could be made if this research task were to be carried out again.

My aim was to find out what care and advice is given to patients with this disease and if it differs depending on what country or county a person resides and how well known this condition is to doctors. I also noted any other medical conditions that these children have in addition to the effects of hypoglycaemia and ketones which impact the daily life of the child and their families.

After doing some secondary research and speaking to parents of children with this condition it seemed that different areas receive different information and treatment plans for the same GSD. A questionnaire was then developed to find out how this differs and what impact this has on the care provided to the child and families. Qualitative questions enabled parents of these children to have the opportunity to express their views on the care their child receives from both medical experts and the parents/carers. Quantitative questions were used to find how many children have the same type and if the same information is given in different areas.

Symptoms presented for participants with GSD3 were:

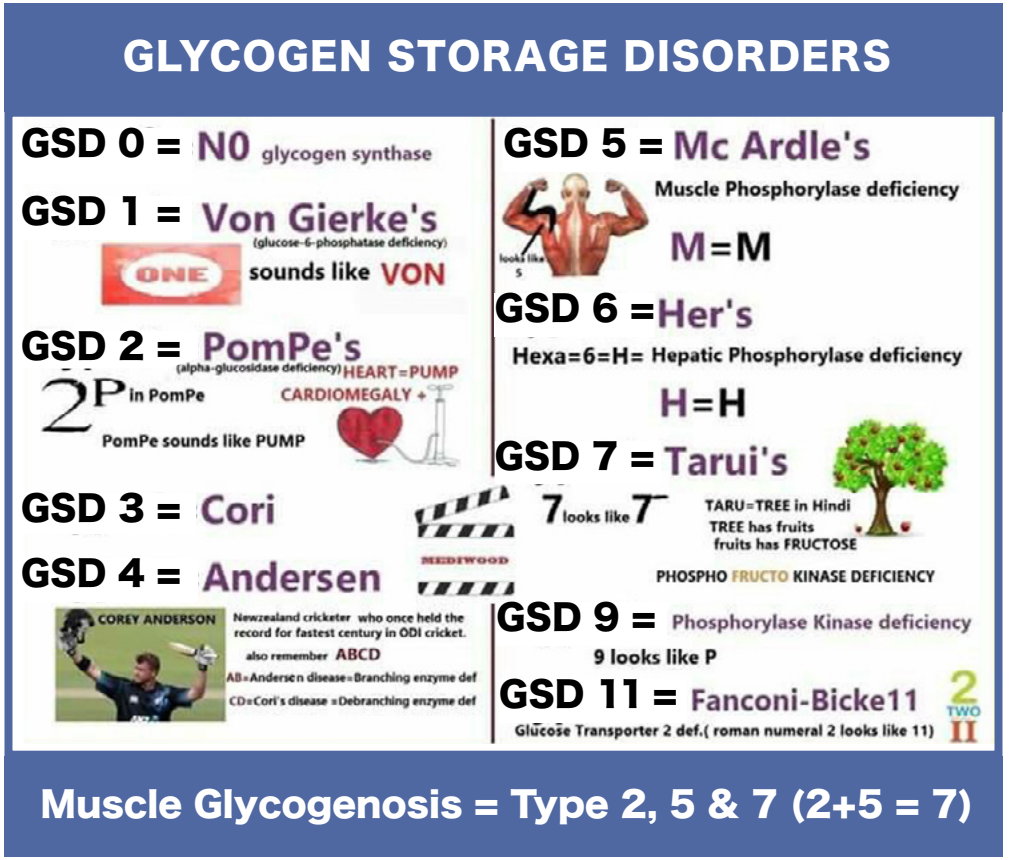
- Enlarged abdomen from the accumulation of glucose in the liver
- Floppy episodes and extremely sleepy in a morning
- Failure to thrive
- Yellowing of the skin and whites of the eyes

All were from the UK and were told the normal range of ketones was 0.0-0.6 except for one. Other health conditions found included one patient who has left ventricular cardiomyopathy, thought to be linked to GSD3 and one that has fattening of the heart valves.

Symptoms presented for participants with GSD0 were:

- Seizures
- Muscle weakness
- Slow growth
- Vomiting
- Development delay
- Constant hunger
- Sweating
- Unexplained crying
- Sweating

All were told ketones 0.0-0.6 ketones are normal range. Other health conditions found included one participant that had a cleft palate and strabismus.



Please note that research in regards to GSD0, appears to be only about the liver types, so the list of symptoms does not apply to the muscle type.

Mutations in GYS1 gene cause muscle GSD0, and mutations in the GYS2 gene cause liver GSD0. (MedlinePlus)

Symptoms presented for participants with GSD9 were:

- Ketotic hypoglycaemia
- Vomiting
- Pale complexion
- Lack of concentration
- Leg pains
- Short statue
- An enlarged abdomen
- Short statue
- Sweats
- Lethargy
- Frequent urination
- Unexplained behavioural changes, including distress and fear and hunger

All were told ketones 0.0-0.3 ketones are normal range. No other health conditions were found within this GSD group. It was reported that one patient was diagnosed accidentally. Please note GSD9d is a muscle type and so this information does not apply.

One participant has GSD11 (Fanconi Bickel Syndrome) which presented with:

- Slow growth
- Failure to thrive
- Developmental delay
- Renal tubular acidosis

There were participants from the United States, United Kingdom, Canada, Belarus and United Arab Emirates. Ketone levels within these countries ranged from 0.1-8.0 with treatment including sodium bicarbonate mineral water, additional protein, additional corn-starch, fluids, and hospital admissions. I found that there didn't seem to be much of a difference in the treatment plans in different countries, however, they are or should be specific to the child not the condition itself. One parent had to seek medical care by travelling to see a GSD specialist in a different country, as their local hospital treatment plan had caused complications and liver damage.

There is not enough research out there to do a full in-depth report and get enough information to get a full picture of the impact this has on daily life. Although around three hundred questionnaires were sent out, not enough were obtained to carry out the research paper in more depth. However, the questionnaires completed were well received and filled in by many in detail, giving a clear understanding of what was asked.

From the sixteen questionnaires only four participants said that their local hospital was aware of how to treat the condition. Many had to travel far to receive the correct care or have their local hospital contact their specialist to be informed on how to manage it. This included the use of IV dextrose to bring the ketones down and stabilise blood sugars.

If I were to conduct this research again, I would include a question on how long after developing symptoms it took to get a diagnosis. This would help to determine whether the lack of knowledge by doctors has an impact on the length of time before treatment is started and if this in turn impacts on the long-term outlook of health.

Thank you to all who took part in my research project

Sinead Smythe

COVID-19 UPDATE

It has been a strange and uncertain 18 months for us all, so it is important to reflect on how the charity took on the challenge to ensure we were there for all our members, providing fun things to do, supportive online groups and meetings and events to keep you all busy! We hope to continue to provide exciting events and opportunities to get involved in.

• Winter Season 2020/21

We started with our Winter Season, which was in the place of the 2020 conference. The season was held over 3 months and included presentations from medical practitioners as well as patient stories. We had some fantastic feedback from the Survey Monkey that was sent out to all members. It was great to see how many people enjoyed and found value from the Winter Season. After a great success we have decided to run another season in January 2022.

Keep your eyes peeled for more information towards the end of this year on the website and in your inboxes.

You can still access all of our Winter Season 2020/21 for free on our YouTube Channel *GSDScreen*.

• IOPD Tea Party from Muscle Warrior

On Sunday 28th March AGSD-UK members connected with Muscle Warrior Foundation for an online tea party! The activities were funded by *BBC Children in Need*.

It was organised by Jo Harford and included a talk by the founder and leader of the *Foundation Michael McGrath*. 15 children participated with their parents, all of which have a child with infantile onset Pompe disease in the family. The best part was getting a HUGE home delivered picnic hamper to share for the party.

If you have young family members affected by a muscle condition, check out their website here:

https://www.musclehelp.com/

Michael McGrath, founder of the Muscle Warrior Foundation

• Tune-in Tuesdays

Pompe Support Team held *Tune-In Tuesdays* for all-comers to join and have a chat. We reached out to newly diagnosed members and ensured they would feel welcome. *Tune-in Tuesdays* turned in to *Tune-In Thursdays* as it worked out better on Thursdays! We have stopped them for the summer because after lockdown eased people wanted to get out and about; thank goodness everyone could circulate more. See you all soon.

• Other Information

British Inherited Metabolic Disease Group (BIMDG) have not run any more of their useful webinars since the Spring. We always monitor this and will let everyone know if they do. Check out our website where we continue to update our news on Covid-19 and will keep adding important information.

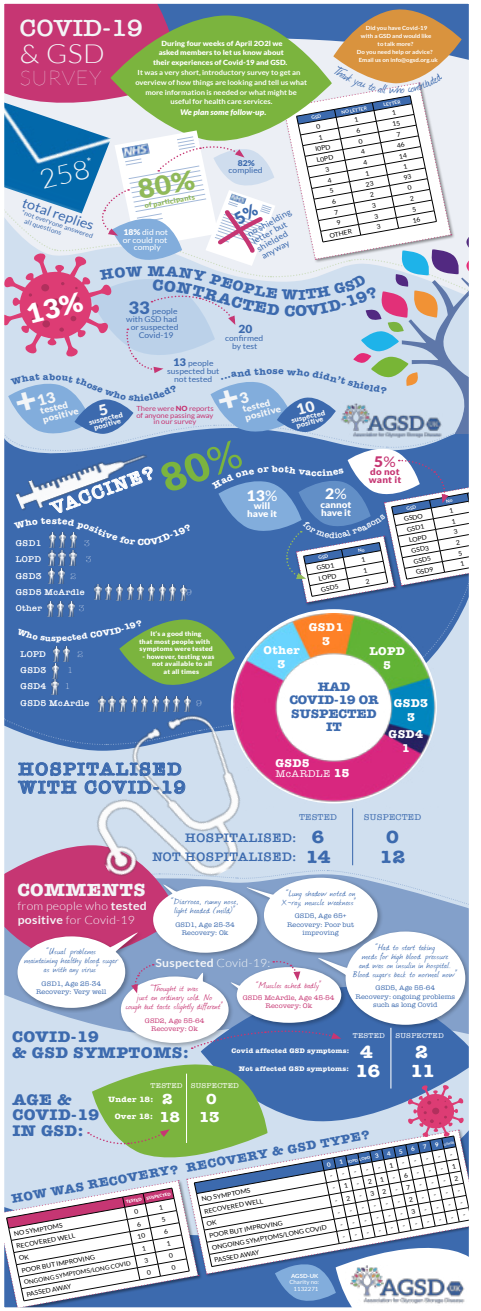
https://tinyurl.com/3uudjnan

In May we circulated the report from ARDent, *Action for Rare Disease Empowerment*. They completed a survey on the impact of the pandemic on rare disease communities concluding that there were delays in diagnosis and especially, delays and suspension of clinical trials.

https://tinyurl.com/7wy2m27j

We continued to be in regular contact with all the specialist metabolic centres, most are now running at full speed in spite of many staff being re-deployed during the worst of the crisis.

We completed our GSD and Covid-19 survey, thanks to all who participated.



My Assistance Dog

Jo and Azaria Moyse

After a couple of weeks in hospital fighting pneumonia a few years ago, we were visited by a PAT (Pets as Therapy) dog. This is where the idea of having a companion to help with both physical tasks and mental health started to grow. After some research we discovered that at that time none of the charities were taking new clients, so we just kept watching and waiting.

One day Azaria came into the room with the exciting news that *Dogs for Good* were again taking applications and we needed to fill in an expression of interest. We filled this in that same day. Names were then selected at random to be invited to an information day, and amazingly we got an invite.

In June 2019 we went along to Bristol to hear all about the amazing work of the charity and all the ways their dogs were trained to assist. We were given an application pack, which I completed over many hours. It was a very in-depth form, but I was determined that I would fill it in to give Azaria the best chance of getting an assistance dog. The process was broken down into many stages and we kept moving forwards, until finally we were offered a home visit! Unfortunately, the call came the day after we had accepted an offer on our house. With a move in the offing, the visit was postponed until after we had relocated 500m down the road to a wheelchair friendly bungalow.

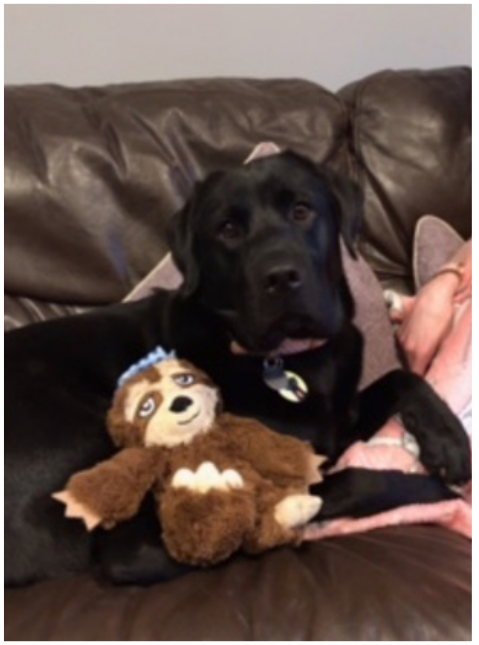
About 2 months after our move the home visit took place and we moved to the final stage of awaiting approval. But then the COVID pandemic hit so everything went on hold. Finally in October 2020 we heard that our application had been successful, and they would start looking for a match.

In February 2021, we heard that Bronte was considered a match for Azaria and a date was agreed for her to come to us to commence our training. The training took place in a compressed format due to the restrictions. Bronte moved in in March and we had visits from our trainer in the garden, went out for walks with her and did some training via teams. We were very grateful that the charity found ways of working in a COVID safe way to enable Azaria to finally get her assistance dog.

In May on a very very very wet Friday morning, just before the end of A level exams, Azaria and Bronte qualified as partners. Bronte is great at picking things up that Azaria can't reach down to get, opening and shutting doors and she helps with undressing, retrieving items and for me one of the best is that she has given Azaria confidence finally to go out on her own for the first time in many years.

There are still things that we are working on together, and our trainer will always be an email/phone call/text away with help and advice. Bronte has had a soothing effect on Azaria, and it is reassuring to know that when all her classmates head off to Uni in September, Azaria won't just be staying at home on her own. She will have a reason every day to go out and do things. It is lovely to see them working together, and the special bond they have formed.

When things are tough, that unquestioning affection, will, I'm sure, help all the more.



Bronte with Pompe toy mascot sloth ***Hope Travels***

My Work Placement with AGSD-UK

Lisa Massimo

Hi, I'm Lisa and I have GSD Type 1b. In my spare time I volunteer for AGSD-UK and I am responsible for their social media platforms. As part of my day job, I am training to be a genetic counsellor. For part of my course, I was fortunate enough to spend two weeks with AGSD-UK. I wanted to learn more about how individuals with Pompe were diagnosed and what medical management is involved. I also wanted to understand if individuals had any interaction with genetic services. There probably isn't room in the Glisten magazine to write all that I learnt, but I have summarised some of things that really stood out to me in these two weeks.

It was clear through talking to individuals that there is a huge variety in the amount of time it takes to get the correct diagnosis! While some people had to really push to be investigated, others were diagnosed much quicker. I found it interesting that most of the stories I heard involved a curious or thoughtful healthcare professional thinking 'outside the box' to find the right diagnosis for people.

It appeared that people diagnosed with Pompe were not always put in touch with the genetic service. I spoke with several healthcare professionals regarding this, and they explained there were a few reasons why. Firstly, that genetic results do not come back quick enough to help impact diagnosis, so are not always done. Secondly, genetics is not always introduced at the start of a diagnosis due to the huge influx of information that person will have received. They advocated people to think about genetics and seek a referral if they wanted to know more.

My two weeks also highlighted how important AGSD-UK has been to individuals with Pompe. The charity has been accessed for a wide range of support, from families who were struggling to find easy to read information about the condition, to helping apply for benefits and medical equipment. I was also made aware of all the background work that goes on to keep the charity running and the medical input the charity has to help support new treatment for people with Pompe.

I also realise how passionate I am about highlighting Glycogen Storage Disease to healthcare professionals, to help shorten the diagnostic journey and ensure people have access to the correct treatment. I would like to say a big thank you to everyone who spared some time to speak with me about their experience and thank you to Jane Lewthwaite for organising my busy timetable!

HEPATIC UPDATE

A bi-annual round up of recent research on the hepatic GSDs. Please do get in touch if you have any additions or comments.

Compiled by Ailsa Arthur

1 Effects of acute nutritional ketosis during exercise in adults with glycogen storage disease type IIIa are phenotype-specific: An investigator-initiated, randomised, crossover study.

IRENE J HOOGEVEEN ET AL. J INHERIT METAB DIS JAN 2021

This study carried out on six adults shows that the effects of acute nutritional ketosis (ANK) are phenotype specific. The authors suggest ANK presents a promising therapy in GSD IIIa patients with a severe myopathic phenotype.

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

TERRY G J DERKS ET AL. J INHERIT METAB DIS MAY 2021

An observational study has been commenced to assess the outcome of a medical diet to limit the accumulation of glycogen in light of the success of dietary treatment for the other forms of hepatic GSDs.

3 A retrospective in-depth analysis of continuous glucose monitoring datasets for patients with hepatic glycogen storage disease: Recommended outcome parameters for glucose management.

FABIEN PEEKS ET AL. J INHERIT METAB DIS APR 2021

This is an in-depth retrospective real time analyses of continuous glucose monitor (CGM) parameters in 15 individual GSD patients. Night time measurements reduced confounding by physical activity and diet. The conclusion being that in-depth CGM analysis can be a powerful tool to assess glucose management and optimise treatment in individual hepatic GSD patients.

4 The natural history of glycogen storage disease type 1b in England: A multisite survey.

REBECCA HALLIGAN ET AL. JIMD REP JAN 2021

A retrospective observational study on a cohort of (35) patients with GSD Ib across England (age range 1-39) The phenotype in England involves a short fasting tolerance that extends into adulthood and a high prevalence of gastrointestinal symptoms. Growth is difficult to manage and neutropaenia and recurrent infections persist. Liver transplantation was performed in 9 patients which normalised fasting tolerance but did not correct neutropaenia.

5 Successful pregnancy in a woman with glycogen storage disease type 6.

SARAH CATHERINE GRUNERT ET AL. MOL GENET METAB REP MAY 2021

A report on a successful pregnancy in a GSD VI patient that resulted in a healthy offspring.

6 Hepatic glycogen storage diseases: pathogenesis, clinical symptoms and therapeutic management

EDYTA SZYMARISKA ET A. ARCH MED SCI 2021

The article addresses the problem of diagnostic procedures for patients with a suspected disease.

7 Dietary lipids in glycogen storage disease type IIIa: A systematic literature study, case studies and future recommendations

ALESSANDRO ROSSI ET AL. J INHERIT METAB DIS. JULY 2020

A potential role of dietary lipids in the management of hepatic glycogen storage diseases has been proposed but no consensus on management guidelines exists. An international study was set up to identify published and unpublished cases describing hepatic GSD patients with a dietary lipid manipulation. 28 cases with GSD III were found. High fat diet may be beneficial in paediatric GSD IIIa patients with cardiac hypertrophy, but careful long-term monitoring for potential complications is warranted, such as growth restriction, liver inflammation and hepatocellular carcinoma development.

8 Neurological Characteristics of Paediatric Glycogen Storage Disease

JULIO HENRIQUE MUZETTI ET AL. FRONT ENDOCRINOL MAY 2021

The aim of the study was to investigate the metabolic, genetic, and neurological profiles of children with GSD and to test the hypothesis whether GSD type 1 would have greater impact neurological impact than type IX. 5 with 1a, 1 with 1b, 4 with IXa and 1 with IXb were looked at. Biochemical data and magnetic resonance imaging were of the brain were evaluated. Pathogenic mutations were identified using multigene panel analyses. The study reported six GSD variants, previously unknown and neurological consequences of GSD1. The principal neurological impact of GSD appears to be related to inadequate control, especially hypoglycaemia.

POMPE RESEARCH UPDATE

Amanda Porter

1 First Patient Dosed in FORTIS Trial of AT845 for LOPD

- The first patient has been dosed in the FORTIS Phase 1/2 trial of AT845, a single-dose gene therapy for adults with LOPD.
- AT845 is designed to deliver a healthy copy of the gene that encodes the enzyme acid alpha-glucosidase (GAA) directly to muscle cell.
- FORTIS (NCT04174105), which is expected to enroll 8 participants, will be conducted at eight sites across the U.S., Germany, and the U.K. The first patient was dosed at one of the three U.S. sites now recruiting.
- The trial is being conducted by Astellas Gene Therapies, the result of the acquisition of Audentes Therapeutics by Astellas Pharma.
- It will evaluate the safety and efficacy of AT845, given as a single dose via intravenous delivery, to LOPD aged 18 to 80, followed by a 48-week observation period and a five-year follow-up.
- The trial's primary outcomes are the safety and tolerability of AT845, as well as the change in GAA protein expression levels and activity from the study's start to 12 weeks post-treatment.

2 FDA Filing of AT-GAA for Late-onset Pompe Expected my Mid-Year

- Amicus Therapeutics anticipates completing a rolling application by mid-year to seek approval of its investigational therapy AT-GAA for LOPD in the U.S.
- AT-GAA is a two-component therapy comprising Cipaglucosidase alfa, a lab-made version of GAA designed to enter cells more effectively, in combination with miglustat to stabilise the enzyme's structure. Cipaglucosidase alfa is administered directly into the bloodstream, while miglustat (sold as Zavesca as a treatment for Gaucher disease) is taken as oral capsules.
- The application is based on AT-GAA safety and effectiveness date from the Phase 1/2 ATB200-02 trial, the Phase 3 PROPEL study and its open-label extension study.
- Top-line data showed the AT-GAA led to significant improvements in physical and lung function, particularly among patients who were previously on alglucosidase alfa, compared with those who continued on alglucosidase alfa.
- AT-GAA-treated patients also showed improvements in measurements of muscle and motor function, reductions in biomarkers of muscle damage and increase glycogen clearance, relative to those given the standard treatment.

3 Duke Department of Neurology Research

- A Duke University study finds that an ultrasound of the tongue may be used as an effective diagnostic tool for LOPD and distinguish it from other diseases.
- The research assessed at tongue structure and function in 70 subjects. Ultrasound was used to assess tongue overall appearance, echo intensity and thickness.
- Differences in tongue strength, qualitative appearance, echo intensity and thickness between LOPD subjects and neuropathic controls were statistically significant.
- Greater tongue involvement was observed in LOPD subjects compared to those with other acquired/hereditary myopathies, based on tongue strength and sonographic muscle thickness.
- These findings provide additional evidence for tongue involvement in LOPD characterised by weakness and sonographic abnormalities suggestive of fibrofatty replacement and atrophy.

4 Different Types of Mutations Appear to Predict Pompe Subtypes

- The type of mutation carried by a Pompe disease patient appears to predict whether the disease is infantile or Late-onset, according to a study published in the Journal of Human Genetics
- While GAA activity under 10% of normal is generally found in Infantile-onset, the Late-onset form associates with a wide range of enzyme activity, which do not correlate well with disease severity.

- All infantile-onset patients had clinically significant cardiac hypertrophy (enlargement) and had GAA activity under 20% of normal. AS for the LOPD group, no patient had significant heart muscle disease and most had elevated levels of muscle or liver damage markers.
- Mutations known as "splice site variants" - which can disrupt protein formation by resulting in the loss of protein-coding sequences or the inclusion of gene subsections normally removed - associated with LOPD. These splice variants often occurred in only one of the two GAA gene copies.
- Several of the splice site variants in the study appeared to associate with milder cases, implying that disease severity might be predicted from a person's individual genetic features 0 their "genotype".
- The children with IOPD tends to have stations in both GAA copies. Rather than splice site variants, they had mutations that result in the loss of genetic material, a change in amino acids, a premature stop in the coding sequence the less to a shorter, unfinished protein, or an alteration in the way that sequence is read.
- Although this study's findings imply a greater role of genetic testing and reinforce the value of newborn screening, the remaining uncertainty involved in making predictions based on patient genotype means the enzyme activity levels continue to have utility for supporting the diagnosis when the genetic variants are ambiguous.

5 Muscle Damage Not Evident in LOPD Patients after Resistance Exercise

- Strength training exercises did not aggravate muscle damage in people with LOPD, although patients with significant changes including certain markers of muscle damage should be monitored while determining their best training regime.
- Physical exercise can be broadly categorised as aerobic or resistance exercise, the latter better known as weight training. Although resistance exercise is an established way of developing strength of healthy individuals, there are concerns regarding its safety in LOPD.
- Unaccustomed exercise typically causes muscle some damage - known as exercise-induced muscle damage (EIMD) - that is marked by heightened levels of creation kinase, lactate dehydrogenase and myoglobin, as well as muscle soreness and temporary strength loss. Studies indicate the EIMD is less severe when muscle contractions are performed concentrically, or in ways that cause muscle tension as muscles shorten (as with a bicep curl).
- Researchers at the University of Pecs, in Hungary, investigated how EIMD markers change in response to resistance exercises - repeat sets of concentric knee flexions with resistance - among LOPD patients.
- Starting levels of all tree blood markers - creatine kinase, lactate dehydrogenase and myoglobin - were higher in patients than controls.
- Levels of these markers increased significantly among the healthy controls at 24 hours post-exercise, before lowering. In contrast, only myoglobin levels rose among those with LOPD, remaining elevated at 48 hours after the exercise session.
- Overall, it appeared that resistance exercise increased patients' strength over 24 hours, and that with the exception of lactate dehydrogenase, changes in EIMD markers were less severe among patients than controls.

LATEST NEWS

We try to keep you updated with all Pompe related news, usually by direct email.

Importantly, keep checking our website news area too!

Whole Genome Sequencing for newborn screening

Step- Free Access in London - BBC Politics Show

So far, in 2021, Early Access to Medicine Status was given to two treatments;

Avalglucosidase Alfa and also to Cipaglucosidase alfa with miglustat.

Links to Government website news for more information are below.

https://tinyurl.com/pw878rcf

https://tinyurl.com/5ejzj4k6

There is certain to be more news to come; we hope to hear about the first UK patient on a gene therapy trial.

We are also closely monitoring NICE, the National Institute for Health and Care Excellence. Specifically, they appraise highly specialised technologies for wider NHS use.

FDA approves Nexviazyme® (avalglucosidase alfa-ngpt), an important new treatment option for late-onset Pompe disease.

https://tinyurl.com/48d5ztaf

THANKYOU!

AGSD-UK Pompe Survey 2021

THANKS for all the replies!

We are collating everything together and a report will be prepared.

We will let you know the headlines and the report will be available on request.

There have been a lot of surveys in 2021. It helps prepare for important work representing our community. We appreciate your patience.

Our latest publication for Pompe disease

The booklet was sent out to members for International Pompe Day 2021. We had great feedback, including one person who ordered FIVE copies!

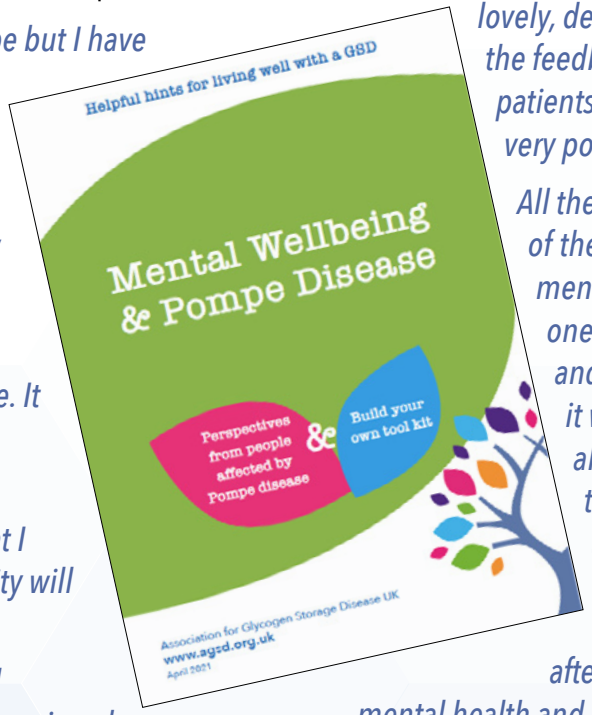
" I don't have Pompe but I have a bone-wasting condition, I saw this and found it very helpful indeed, thank you."

"I really love the practical approach you have taken here. It is so important."

"Well done on such a great resource that I know the community will value."

"This is a wonderful resource and I really enjoyed learning from it."

"I loved the Kit Bag ideas and am inspired by the community's contributions! I'm looking forward to sharing this more widely."



We received this response from a mental health professional:

"It seems to have something for everyone, and I particularly liked the toolkit and Annabel's story. It is lovely, detailed and the feedback from patients has all been very positive.

All the models of therapy mentioned are ones I advocate, and I am sure it will benefit all patients to hear about the importance of looking after your mental health and wellbeing and the variety of psychological help that could be made available to them as well. No health without mental health is a phrase I like, and this leaflet captures that well."

Research Opportunity

Nicola Condon

Nicola Condon Senior Physiotherapist Queen Elizabeth Hospital Birmingham & Birmingham University MRes Clinical Health Research student

I am a Masters Student studying Clinical Health Research at the University of Birmingham and also a Physiotherapist working at the Queen Elizabeth Hospital Birmingham. For my thesis I will be conducting a research study exploring barriers and facilitators to physical activity for people living with Pompe disease later this year…… and I will be needing your help!

Are you diagnosed with Late onset Pompe Disease (LOPD), over 18 years of age, living in the UK? Would you be willing to join me for a chat either online or by phone?

If so, please would you consider taking part in an interview discussing your views on Physical activity and exercise, your experiences, and any challenges you encounter incorporating physical activity and/or exercise into your lives.

The interview is likely to take no longer than an hour and can be completed over the phone or on-line video call, whichever is easiest for you.

I am keen to hear from all perspectives

- Whatever your age, physical abilities or limitations
- Whether you see yourself as generally physically active or not
- Any experience with exercise – good or bad / past and present

Research Title: Physical activity barriers and facilitators in Pompe Disease: perspectives and lived experiences

Due to commence recruitment - Nov 2021 – Feb 2022

What is the purpose of the research?

We should all be engaging in healthy lifestyles, however, data on physical activity in adults demonstrates significant inequalities for people with disabilities. They are twice as likely to be inactive when compared to non-disabled people and only 25% of adults in the UK with a disability reach recommended amounts of Physical Activity. The World Health Organisation (WHO) defines physical activity as any bodily movement produced by the muscles that requires us to expend energy. This includes exercise as well as other activities which involve bodily movement such as movement as part of working, commuting, household chores and recreational activities.

The purpose of this research study is to explore attitudes towards engagement in physical activity and exercise from the perspectives of people living with the rare condition, Pompe disease.

I hope to develop a more in-depth understanding about perceived barriers and facilitators to engagement which could inform future service developments and new research.

Contact and Further information:

If you want any further information about this study, please contact Nicola Condon via e-mail :

NJC645@studentbham.ac.uk

Andrew Wakelin
McArdle's Coordinator

The McArdle Experience 2022

Although the Walking Course and the Children & Parents event have both had to be cancelled in 2020 and 2021 due to the COVID-19 pandemic, we are planning to return to events in 2022. The walking course will be rebranded as ***The McArdle's Experience*** to better reflect the nature of the week-long residential course. Walking is really only a part of the event and it's much more to do with meeting people, sharing experiences, learning all the tips and tricks, and usually having quite a laugh!

We shall again be returning to the Stackpole Centre in Pembrokeshire, Southwest Wales for both the adults and children's events. There it will be some overlap between the events and the dates are:

The McArdle's Experience:
Saturday 30 July to Saturday 6 August.

Children & Parents Event:
Friday 29 July to Tuesday 2 August.

Drop Andrew an email - type5@agsd.org.uk if you wish to provisionally reserve a place, or just watch out for opening of bookings early next year. There will be an announcement on the website and in the Facebook groups.

McArdle's walkers enjoying the Pembrokeshire Coast National Park

The National Trust

In the last issue of Glisten we had a hint about some exposure which was expected in the National Trust magazine, going to almost 6,000,000 members. Well, it appeared in the summer issue and was a story in their series ***My Membership and Me*** in which I talked about my many connections with the National Trust and in particular about ***Walk over Wales*** and running the walking courses – which have heavily used National Trust accommodation bases and countryside. There is a news story about it on our website, from which you can read/download a copy of the article.

https://tinyurl.com/2p2hpyp2

Read the story via our website

The Winter Season is coming again

Following the cancellation of the AGSD-UK annual conference last year due to the COVID-19 pandemic, we ran a winter season of videos and online events over the turn of the year. For McArdle's we produced 19 videos which have now had about 1800 views. Most of the videos have long-term relevance and value, so if you have not seen them yet do give them a try on the AGSD-UK YouTube channel ***GSD Screen***.

https://www.youtube.com/user/GSDScreen

The trustees have decided that the pandemic makes it necessary to make similar arrangements this year, so watch out for the ***Winter Season*** during January, which will include McArdle's videos and events. NB: if you have any special requests of subjects for videos or online events, please drop Andrew an email in plenty of time.

type5@agsd.org.uk

Changes at the McArdle Clinic

There have been some changes at the national McArdle's Clinic at Queen Square. Prof Ros Quinlivan remains the clinical lead but some of her other duties have changed significantly. One impact is that the annual children's clinic at GOSH will be led by another doctor but Prof Quinlivan will still attend. Also, Sherryl Chatfield has moved on and our Neuromuscular Physiotherapist is now Kate Russell.

The COVID-19 pandemic continues to mean that some appointments will be on-line rather than face-to-face. Make sure you know which type yours is before travelling.

Paper on Scottish cohort with McArdle's

Dr Maria Farrugia and a team of co-authors have published a paper on a cohort of 14 patients with McArdle's who attend the West of Scotland adult muscle clinic in Glasgow. The paper reports on the phenotypic and genotypic features of these patients, that is their clinical presentation and their genetic mutations.

Hopefully this will further raise awareness of McArdle's in Scotland. AGSD-UK supports the clinic with books and leaflets for patients.

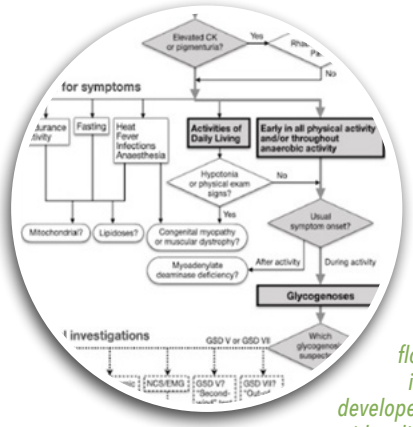
There are many interesting or useful aspects to emerge. One such is that in this cohort 58% did not report any *second-wind* like feature before diagnosis and explanation of the phenomenon. This is really important. As *second-wind* is universal when tested under controlled conditions, some clinicians around the world will dismiss patients saying they can't have McArdle disease if they don't experience *second-wind*. They must understand that self-reporting is very different to testing in clinic.

We are attempting to obtain Open Access to this paper for our members, and will keep you informed. The Abstract can be read here: https://tinyurl.com/yk2wd4jn

Progress on Clinical Practice Guidelines

Dr Stacey Reason, President of *IamGSD*, has been steering the development of Clinical Practice Guidelines for McArdle's (GSD5) and Tarui disease (GSD7), with a team of 15 co-authors from around the world. Almost all the leading clinicians and researchers in these conditions have been involved. It is now in its final stages and will be published in *Neuromuscular Disorders*, the journal of the *World Muscle Society*, during the autumn. It will be about 12 pages with a supplementary document available online.

IamGSD has arranged for the paper to have Open Access, thanks to the financial support of *Reneo Pharmaceuticals*, a Californian biotechnology company focusing on the development of therapies for patients suffering from rare genetic mitochondrial diseases.



A flowchart is being developed as an aid to diagnosis

IAMGSD's Global Walking Week

IamGSD's annual walking week is an event in which people with McArdle's around the world walk where they are and contribute their mileage, any tips and how they felt. It took place again this year over 22-28 May but of course was impacted by the COVID-19 pandemic. Hopefully next year will see very many more people participating.

Thank you for continued support from the Industry, Foundations and Trusts that help AGSD-UK.

These grants allow us to work on projects and ensure our members at AGSD-UK have access to a high level of support and enable us to run events and contribute to research in GSD.

Thank you for the continued support from:

Amicus

Astellas Audentes

Avro Bio

Edward Gostling Foundation

FMR Global Health

FYMCA

Sangamo

Sanofi Genzyme

Spark Therapeutics

Ultragenyx

Vitaflo

AGSD UK
Association for Glycogen Storage Disease

Take aways from this Glisten...

Up coming event!

Keep an eye out on our social media for more information on Remi's Challenge where he is cycling from Lands End to John O' Groats!

- Please help us with fundraising and donations
- Keep watching our social media and website for the latest information and news!
- Phone us for advice on completing benefit forms before you apply. We're here to help
- Help us with research and get involved in a Pompe project with Nicola Condon

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