March 2021

AGSD UK
Association for Glycogen Storage Disease

GLISTEN

GLYCOGEN STORAGE DISEASE NEWS

Support Hugo's Family in their AGSD-UK fundraising challenge!

Association for Glycogen Storage Disease UK
PO Box 1232
Bristol
BS48 9DD
Phone: 0300 123 2790
Email: Info@agsd.org.uk
Charity Number :1132271
March 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:
Hugo Osborne with family & friends

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

The last Glisten hit the streets in August 2020. Since then our conference moved on line and mutated (is that a bad word these days?) into the *'Winter Season'* of online talks and presentations. We recruited Producer, Vicki Lucass, who has made it all happen. We have had great feedback on the season. So, it shows how well we can adapt when needed.

Patient representatives:

On pages 26 and 28 you will see reports from Luke and Gemma who attended online sessions. These are extremely informative for patients. Are you interested? If yes, email info@agsd.org.uk. We will ensure we circulate all opportunities to you. Many events are free and we also sometimes fund reps to attend, in return for a little written round-up.

We have only had very few reports of cases of Covid-19 amid the GSD community via ad-hoc information gathering.

We are planning to send out, very soon, a quick questionnaire asking for more information; please complete it for us and we will report on it to you. So far, we know that everyone has recovered well, even those in the extremely high-risk groups. Vaccinations are happening fast, and, as I write, most of our membership with a GSD is receiving a vaccine, being in the higher risk groups. All good news.

An important word to our parents:

We all know, appreciate and recognise the ongoing care needed for children with a GSD. The extra vigilance, hospital appointments, feeding and treatment regimes are crucial but can be extremely tiring. Although I am an adult living with a GSD I know well, hearing from others, just how all-consuming this can be. Parents could have a look at page 33, where you will see that *Over The Wall* camps have also moved online to the *Camp in the Cloud.*

This makes us all the more appreciative of what Hugo's parents are doing with their fundraising walk for AGSD-UK. Hugo was diagnosed with GSD3 in early 2020.

More good news! We have a new Industry funder in Ultragenyx Ltd who have granted us £5000 towards development of new information resources.

Not such good news; we have not been able to apply for the Covid-19 related charitable funding which either targeted regional charities or those who had already been funded by the Lottery and others. So, donations and fundraising are desperately needed. Could you help?

See pages 12 & 13 to see how others have helped us, for which, THANK YOU.

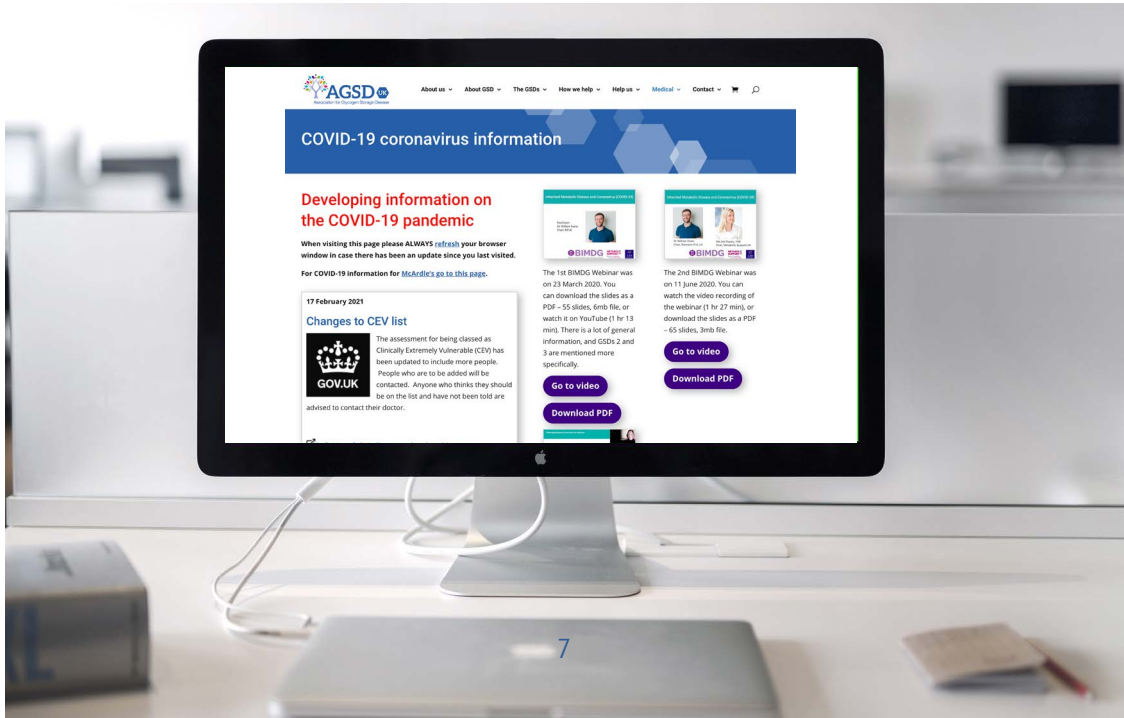
Nick Jones
Acting Chair of Trustees

Hugo Osborne

New AGSD-UK leaflet

Our GSD COVID-19 year?

- See our website for regularly updated Covid-19 GSD-related news
- The first news item was added to it on March 18th 2020 and since then added 40 more important items and links
- We issued 10 urgent updates and alerts to the whole membership, the first was on March 6th 2020
- We advertised all the Metabolic Covid-19 webinars and there are three videos available on our website
- 87 Patient welfare checks or phone calls were made by the Care Advisor
- We added regular social media posts, several times per week
- Trustees had a series of fortnightly Zoom meetings from March to June 2020 to provide direction and support
- Trustees awarded nine small grants for those in very urgent need
- We offered 100 free places to join online Mindfulness and mental health training
- Our Mailchimp messages have excellent readership clicks at 50-60%
- We applied to Sanofi and received two grants towards a benefits support advisor



Her Indoors...

A lyrical round-up of Esther Prosser's Covid-19 year

It's been alright I suppose,
I can't really complain,
For me, this is not the first time I've
been stuck indoors,
For me things are mostly the same

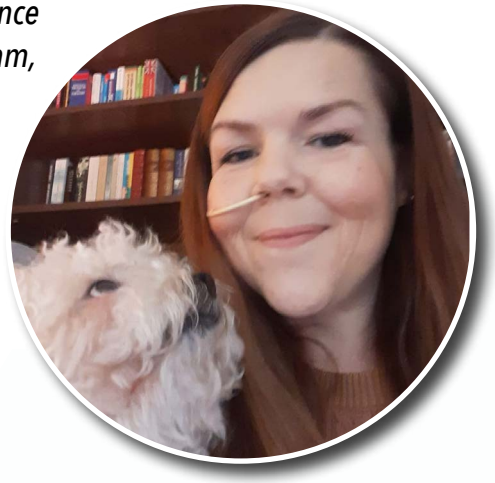
Except, shopping with friends
And long lunches in town.
Have been replaced
With staring out the window
In my dressing gown

Oh and of course
My beauty regime
Definitely has had a slip
as I watch my hair grow
Over my top lip

And no cuddles,
No kisses,
Not even a snog
Unless you count the ones
From Bella …my dog

I do like to binge on TV and films
And eating dinners in bed
And finally got round to fixing my
SHE-SHED.

Found a passion for the garden
giving orders from a distance
While my lone worker, Mam,
shouted back what!?
Pardon!?



Another little
positive
Is accessibility,
Because now my
favourite restaurants
Do home delivery.

It's been good catching up with friends
From home and away
Making future plans to visit,
Invitations to come and stay.

Started doing gentle exercise
And into heathy eating
So far so good
I'll tell you about it at our next meeting

Until then
I hope you are well and feeling fine?
That my little rhyme made you laugh
but
Mostly helped just pass the time.

Esther Prosser

COVID-19 HERO

What colour is a flamingo?

Gemma Seyfang of the Pompe Support Team created fun quizzes for all–comers (not just Pompe) during lockdown that have been a BIG HIT. Not only that, but she ran two prize quizzes in January for children as part of the Winter season (including the question above!)

An unexpected hero in this lockdown time
Arose called Gemma who is so sublime
To make us smile with her wicked quiz
She really was a technical whiz
Gemma is now our quiz mistress.
She raised our spirits and eased distress.
When lockdown meant we were all alone
Gemma zoomed into our living room.
To give us a laugh and stop us feeling glum,
She made the quizzes hard, easy and fun.
So, for an hour we forgot our woes,
As she kept us on our mental toes.
So thank you Gemma from all of us
For keeping us entertained without a fuss
You will never know how glad we are,
That you are there, our zooming star.
John

THANK YOU GEMMA!

Jimjams, THANK YOU so much for starting and continuing to organise and present the zoom quizzes throughout lockdown and beyond. For us, it has been a connection we have thoroughly enjoyed - seeing you and other people with GSDs, laughing and having fun has been much needed. You're a star! **Rae**

Thanks Gemma for adding some much-needed fun to last year's lockdown! **Kempton**

I have loved the quizzes that Gemma has set - quirky, varied & very entertaining! They each must take her hours of preparation beforehand and are always very well delivered. Gemma doesn't bully us like Jeremy Paxman, either!!! **Bridget**

Just when it feels bleak and difficult, just when there seems to be little inspiration, Gemma creates FUN. Not just the quiz, (which is brilliant) but kindness, caring, always bringing positivity and her lovely laugh too! You've been a massive part of making everything brighter, thank you. **Jane**

Gemma, thank you for all the effort you put into your amazing quizzes. I've really enjoyed meeting new members of the GSD community through them (including you) and I've definitely learnt a few new facts! **Lisa**

Gemma has been an absolute star arranging quizzes for us. They're great fun, a good distraction from the covid 19 stuff and a good way to catch up with other GSD folk. I have loved taking part and really appreciate all Gemma's hard work. **Sue**

Fundraising Round-up

Hugo's family fundraising for AGSD-UK

Flo, Jenny, Mads, Lauren, Nic, Dale, Nathan & Andy will all be ***#HikingForHugo*** and climbing the three highest peaks of Scotland, England and Wales within 24 hours.

Hugo's Mum, Dr Florence Kinnafick said; *'Our fundraising challenge is getting close! We are all doing well but as you can imagine the COVID restrictions are hampering any long walks and training as we can only go from the house and stay fairly local. A few of us did fit in a good walk (24km) before lockdown in December, as you can see in the photos.'*

The challenge is set for April 17th weekend. However, just in case things are still looking difficult because of COVID, they have set a second weekend aside on July 10th to complete it then.

So far they have raised £4,400

Our thanks extend to everyone able to donate and fundraise for us, especially during difficult lockdowns.

A special mention goes for Jenny Cox, she is in the centre of this photo. She is taking on this challenge and raising money for AGSD-UK as a challenge to herself to mark 10 years since her dear father, Pete Chamberlain, passed away.

Although he did not have a GSD, she has chosen to complete this challenge for us as he liked his fitness training and would have loved her to do it!

https://uk.virginmoneygiving.com/HikingForHugo

Thank you also to...

- Eileen Rice kindly gave a donation after having help from Julie Jackson with a complex issue which was resolved **£100**
- Father Christmas?! Thanks to Ashley Eustace for his **£100** donation on 25th December, all the way from Australia
- Lynne Sharman continues to sell home-made face masks. She was helped by her son who sold masks at his rubgy club and her daughter who made beauty bags. Total raised **£180**
- Thanks for the Bedford Park Lodge Masonic Hall who donated **£500**
- Update from Gemma and Kieron on their mammoth fundraiser last year; the whopping total from the sponsored run, curry night, bingo night, and online auctions was well over **£6000**
- Frank and Irene Green, Christmas lightshow at their home. The total raised beat last year **£770**
- Congratulations to Grant Fraser for his 1st prize win in the Rhodar competition; dontation of **£250** to AGSD-UK

Rhodar Ltd, a UK leader in asbestos removal, demolition and remediation services ran an innovative Christmas promotion in December to help raise spirits at a challenging time....whilst also supporting charities.

RHODAR

The online competition game titled **'Hoppy Xmas'** allowed players to navigate Santa's Elf through a winter wonderland and register a score of over 1,000 points. All players achieving this were entered into a prize draw to win a donation for their chosen charity.

The three winners of the prize draw were announced in early January...along with the charities they had chosen to receive the donations:

1st prize winner - Grant Fraser

Grant chose to split his £500 donation between two charities: AGSD UK & St Columba's Hospice

2nd prize winner - Michael Cosham

Michael chose to send his £250 prize donation to Dementia UK

3rd prize winner - Andrew Brown

Andrew will be donating his £200 prize to MQ Mental Health

Rhodar was delighted that the game created such a positive impact during what was a challenging run up to Christmas...and that the above four charities were also able to benefit from this initiative.

Nigel Walker

Nigel has a won a place in the London Marathon in October 2021.

He said "I would like to fundraise for AGSD UK as my chosen charity. AGSD UK is something that is close to my heart". He has many months of hard training ahead. Good luck!

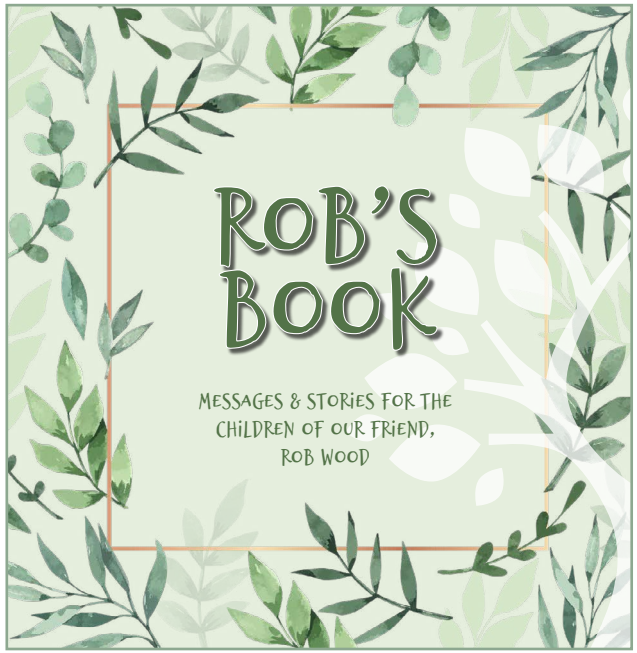
ROB WOOD

We must mention our friend Rob Wood.

He sadly passed away in October 2020.

His wife, Jennifer, kindly asked for donations to AGSD-UK and we gratefully received £1424.00 in Rob's memory.

Thanks to all Rob's friends and professionals who contributed to a book of memories we created for his children.



FINANCE UPDATE

The AGM was held in January 2021, a little later than usual because it is normally held at the Conference in October.

Even though it had to take place online we had a good number of people who joined us.

Below is a summary of the last five years' accounts. The 2019/20 figures are yet to be confirmed and may be subject to change.

Income	2015/16	2016/17	2017/18	2018/19	2019/20
Restricted	90,458	227,966	229,071	103,566	137,759
Unrestricted	41,859	72,787	70,354	64,074	68,043
Total	137,179	300,753	299,425	167,640	205,802
Expenditure	116,609	265,960	232,536	228,035	217,134
Surplus/Deficit	20,570	34,793	66,689	60,395	11,332
Total funds					
Restricted	85,563	128,147	179,059	99,890	121,307
Unrestricted	12,766	4,975	20,952	39,726	6,977
Total	98,329	133,122	200,011	139,616	128,284

AGSD UK
Association for Glycogen Storage Disease

Welcome to our Winter Season 2020/21

The Winter Season is our offering in lieu of the cancelled October conference. Three months, December 20 - February 21, of talks, presentations, events and fun to join in with, for all patients. There are three streams: Hepatic, McArdle and Pompe. All patient and professional presentations will be available after the end of the Season. A huge and valuable resource for everyone.

The AGSD-UK ***Winter Season 2020/21*** has been a huge success and we are so grateful to all that contributed and to all that have watched so far.

We couldn't hold our annual conference in 2020, so instead we had an online offering of videos that covered two playlists. Our Winter Season 20/21 playlists included presentations from some of the top doctors in the field on GSD such as Dr Priya Kishnani (GSD3), Dr Jon Rajan (GSD2) and Dr Jared Schreiner (GSD5) as well as many other top medical professionals. Our other playlist, ***Winter Season Patient Stories***, includes some insights and experiences in the lives of some of our GSD community.

It has been a pleasure to work with so many incredible people to create something that can be enjoyed by so many affected by GSD. The response has been exceptional from the medical professionals and members and has enabled us to provide you all with lots of different presentations covering a wide range of topics. The feedback from members has been overwhelmingly positive and we hope to offer similar events in future. After the Winter Season ends, we plan to keep the videos on our YouTube page for you to visit and watch at your leisure. All details of our Winter Season can be found on the AGSD-UK website.

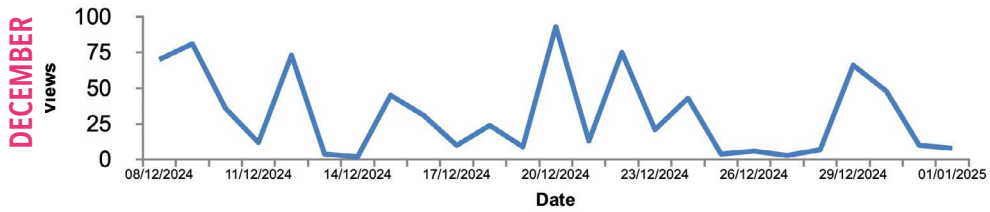
Vicki Lucass, Season Producer

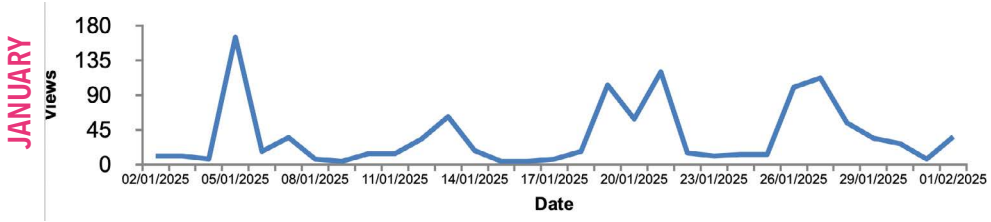
Our viewing figures

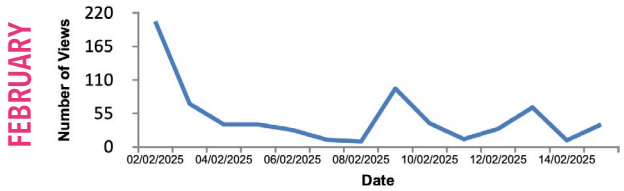
Since we launched our Winter Season, our YouTube page ***GSDScreen***, has had **1928** views, which is a significant increase for the channel, as well as **39** new subscribers! We have had views from all over the world including the UK, USA, Canada, Ireland and Malta. Please see below some monthly graphs, to give you a visual on how successful our viewing figures have been over the Winter Season. There have been people visiting our channel to watch presentations every day since the launch on December 7th.

Breaking down the Winter Season Playlist

Over the last few months we have had many contributions and have so far got







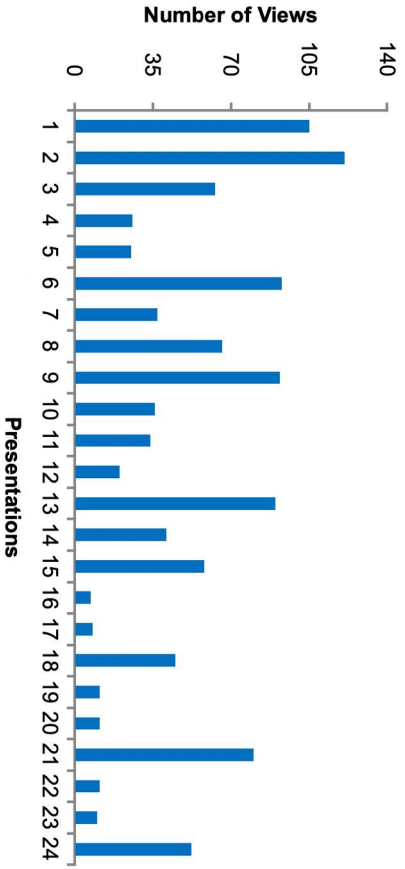
lots of views on our YouTube. We had 9 contributors for GSD2 Pompe, 5 for GSD5 McArdle's and 6 for our Hepatic stream. In such uncertain times with the global pandemic and how much extra pressure this has put on to our medical professionals, we are incredibly grateful to all who have taken the time to record these presentations for us. They have been a great success so we would like to send a big thank you to;

- Sarah Steeds & Helen Gallagher (GSD2 Pompe)
- Andrew Wakelin (GSD5 McArdle's)
- Dr Jordi Diaz-Manera (GSD2 Pompe)
- Dr Terry Derks (Hepatic)
- Kathryn Pierce, M.A. (Hepatic)
- Stacey Reason (GSD5 McArdle's)
- Dr Gisela Wilcox (GSD2 Pompe)
- Dr Kris Saha (GSD2 Pompe)
- Ian MacDougall (GSD5 McArdle's)
- Dr Priya Kishnani (Hepatic)
- Dr Stephanie Austin (GSD2 Pompe)
- Rebecca Gibson (Hepatic)
- Andy Williams (GSD5 McArdle's)
- Chef Neil (Hepatic)
- Dr Jon Rajan (GSD2 Pompe)
- Carrie Connors (GSD5 McArdle's)
- Dr Yin-Hsiu Chien (Baebies) (GSD2 Pompe)
- Dr Ulrike Steuerwald (Hepatic)
- Dr Mark Roberts (GSD2 Pompe)
- Bryn Edwards (GSD2 Pompe)

Winter Season Playlist

Here are just a few of our presentations from the medical professionals that we have featured on YouTube. Visit our channel to watch these as well as many more!!

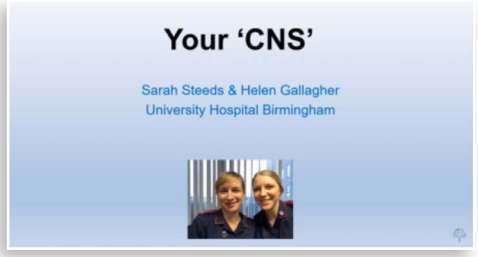
Clinical Nurse Specialists Sarah Steeds and Helen Gallagher kicked off our Winter Season with their presentation on how their role can support people living with GSD, specifically GSD2 Pompe.



This graph shows how many times the medical presentations have been viewed over the Winter Season. Graph data as of 14/2/21 (2 more weeks of presentations left of the Winter Season)

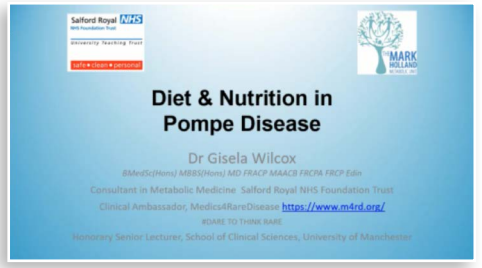
Within this talk they cover topics such as 'what makes a CNS?' How, as part of a multidisciplinary team, they can continue to provide a high level of support whenever it is needed, and how important it is to maintain these high standards during this pandemic.

We had some great comments from some viewers who watched the presentation. Anne found that it was '*very well explained*' and Linda commented '*thank you, very useful info*'.



Stacey Reason, President of IamGSD provided us with a great presentation which has been enjoyed by many so far on our YouTube channel. In her talk she introduces IamGSD, its overall objectives and the progress that is being made towards achieving this by driving activity in its 5 campaigns. Watch on YouTube to find out more!

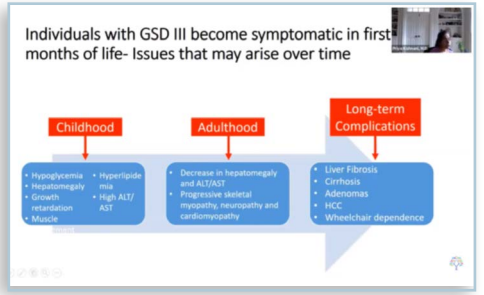
Dr Gisela Wilcox discusses Diet and Nutrition in Pompe Disease. Within her presentation she covers topics such as body composition, the impact of Pompe on nutritional status and the role of nutritional interventions.

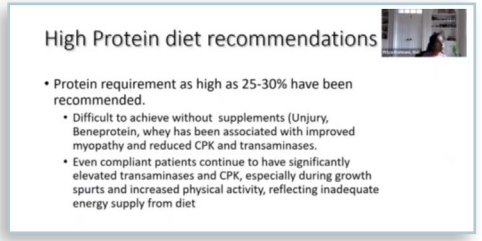


Dr Wilcox also talks about Covid-19 and how to stay well in this current situation.

Amanda commented '*I really enjoyed the presentation on diet for Pompe by Dr Gisela Wilcox - it was really interesting to hear the science behind diet. Thank you*'

Dr Priya Kishnani is Chief in the Division of Medical Genetics at Duke University Medical Centre, which focuses on developing new therapies for rare genetic disorders. She is very established in this field and has a keen interest in glycogen storage disease.





In her presentation she takes us through an in-depth update on Cori disease. She discusses what GSD3 is caused by, the sub types of GSD3 and the recent research conducted in this area.

Over the Winter Series our McArdle's Coordinator Andrew Wakelin has put a lot of time and effort into creating great content for you all to enjoy. In this presentation, Andrew reports on activities and development during 2020.

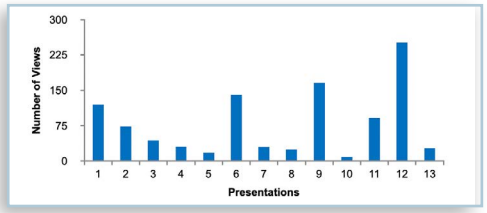
The year was greatly disrupted by the Covid-19 pandemic, but nevertheless there was progress in many areas and considerable promise for the future.

Breaking down the Patient Story Playlist

We had 4 contributions for GSD2 Pompe, 7 for GSD5 McArdle's and 5 for our Hepatic stream. Many of our lovely members that have GSD have kindly recorded short talks for you all to

enjoy. They have been a very popular addition to our YouTube channel and it's been great to gain insight into how people have been coping with the pandemic, especially over the last year. They have been fun, creative but also informative with many of our patient stories giving others who suffer with GSD tips and advice. We would like to thank:

- Esther Prosser (GSD3a)
- Sioned Hosseinian (GSD5 McArdle's)
- Matt Arthur (GSD3a)
- Lisa Massimo (GSD1b)
- Gemma Seyfang (GSD2 Pompe)
- Luke Fraser (GSD2 Pompe)
- John Foxwell (GSD2 Pompe)
- Georgina Chalkley (GSD3a)
- Andrew Wakelin (GSD5 McArdle's)
- Flo Osborne (GSD3b)
- Michael Wilson (GSD5 McArdle's)
- Bronte Thomas-Bush (GSD5 McArdle's)
- Lucy Smith (GSD5 McArdle's)
- Sonia Worthy (GSD5 McArdle's)
- Andy Williams (GSD5 McArdle's)



This graph shows how many times the medical presentations have been viewed over the Winter Season. Graph data as of 14/2/21 (2 more weeks of presentations left of the Winter Season)

Patient Story Playlist

Here are just a few of our patient stories that we have featured on YouTube. Visit our channel to watch these as well as many more!!

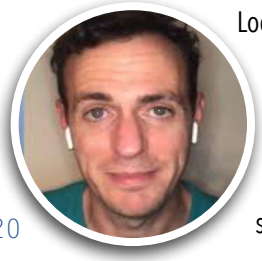
John Foxwell is one of our GSD2 Pompe patient stories and is also part of the Pompe support team. In his short talk, John tells us about his experience with GSD. He talks about the BiPap machine and how it has benefitted him.

John did a second talk for us about the continence issues that people with GSD may face when going about their daily lives and how helpful having a toilet card can be in these situations.

Esther Prosser, who has GSD3a, takes us on a hilarious journey through her experience of 2020. She gives us an insight in to how she has coped throughout the year.

Jane commented *'thanks Esther, that's my biggest laugh this year!'* and Matt also commented *'This is great!'*

This is well worth a watch!



Locked down during the Covid-19 pandemic in a multi-storey block in New York City, Michael Wilson turned to the stairs for his exercise.

Using a technique of two flights down and two flights up repeated, over months he built his aerobic capacity to the point where he can climb all 16 flights of the block in 3 minutes 22 seconds, non-stop. It has given him a new appreciation of what can be achieved with McArdle's.

Daniela commented '*Thanks, Michael, for sharing your experience! I will give it a try*'.

As part of the Winter Season, we ran some events for the AGSD-UK community to get involved with. One of our lovely members Gemma Seyfang, who has GSD2 Pompe, organised and held a Children's quiz. There were two age group categories, children up to the age of 10, and children aged 11 to 16. The questions were fun and everyone had a great time!! We hope to host more of these in the future. Gemma also started a social call '***Tune in Tuesday***' for anyone feeling stuck at home who wanted some company or just fancied a chat.

As part of something a little different, on the website we featured Luke Fraser who has GSD2 Pompe. Luke has a podcast of his monthly radio show, ***The Tonic,*** featuring a global mix of electronic and instrumental classical music from the 20th Century up until now. You can find the link to listen to his podcast on the Winter Season page.

How can I watch?

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

Our YouTube Channel

www.youtube.com/user/GSDScreen

Through Google

1. Go to www.google.co.uk
2. Search GSDScreen YouTube
3. Click GSDScreen – YouTube
4. Go to playlists
5. Here you will find Winter Season 2020/21 and Winter Season Patient Stories playlists
6. Click on the one you want to watch and you will find our ***Winter Season*** presentations from medical professionals and patients

Through our website

1. Go to https://agsd.org.uk/
2. Hover the mouse over 'how we help'
3. Under events you will see 'Winter Season' Click on this
4. Here you will find all of the information on our Winter Season

Look for the two YouTube logos with links to the two playlists. If you click on ***'Presentations'*** this will take you directly to the Winter Season 2020/21 and if you click on ***'Patient Stories'*** this will take you to the Winter Season Patient Stories.

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

Please Donate!

At AGSD-UK we help individuals and families affected by Glycogen Storage Disease by putting people in contact, providing information and support, publishing a magazine and holding conferences, workshops, courses and family events.

Throughout each year we seek to raise funds for specific projects to help deliver our work aimed at improving the lives of the UK GSD community.

Your donations go towards providing both practical and emotional support across all GSD's and their variants. AGSD-UK would like to see all people affected with GSDs, given the opportunity to lead full and productive lives.

If you have enjoyed our *Winter Season* and the events and presentations that we have featured on our YouTube, please donate to help us continue our work in to 2021 and beyond!

AGSD-UK could not exist without support and commitment from a broad range of sources. We appreciate the contribution and efforts of all of our fundraisers, donors and sponsors, many of whom support us each year.

Thank you to all of our contributors, to all members that have supported and watched the *Winter Season* presentations and to our sponsors Spark, Sanofi, Audentes, Vitaflo and Amicus

Your options for a one-off donation:

- **Virgin Money Giving Page:**
- **tinyurl.com/y36n5v4g**
- **PayPal Giving Fund: tinyurl.com/pp-agsduk**
- **By cheque or CAF voucher sent in the post:**

Please make payable to AGSD-UK and post it to:
AGSD-UK, PO Box 1232, Bristol, BS48 9DD

Reports

NHS RARE DISEASE FRAMEWORK

In early January 2021 the Department of Health and Social Care published the new UK Rare Diseases Framework.

The Framework is aimed at raising awareness of rare diseases, speeding up diagnosis and improving care and treatment.

All UK regions are holding separate meetings during 2021 to discuss implantation of the framework and we will update you on those soon.

It sets out four key priorities for rare diseases in the UK over the next five years, covering:

- Helping patients to get a final diagnosis faster
- Increasing awareness of rare diseases among healthcare professionals
- Better coordination of care
- Improving access to specialist care, treatments and drugs

Overview

The National Conversation on Rare Diseases Survey formed the basis of the Framework. This was followed up by an Editorial Board of policy officials, representatives from clinical practice and patient organisations, to formally identify and refine the priorities and underpinning themes for the new Framework.

In order to achieve the four priorities outlined in the Framework, five underpinning themes have been identified in which work will be undertaken to support the priorities of the Framework and improve the lives of those living with rare diseases:

Patient Voice: Any commitments will be developed in consultation with patient representatives, giving particular consideration to ensuring representation from those whose voices can often go unheard.

National & International Collaboration: DHSC is committed to continuing collaboration with the rare disease community across the world, including patients, doctors and industry, to share knowledge and ideas to improve outcomes.

Pioneering Research: DHSC will continue to support and invest in innovative research for rare diseases and ensure that the outcomes are translated into frontline clinical care.

Digital, Data & Technology: DHSC will utilise the benefits that technology can bring to rare disease patients, and consider how digital tools could be appropriately used to improve efficiency and patient experience and support research.

Wider Policy Alignment: DHSC will work to ensure that the needs of rare disease patients are recognised in wider policy development, whether that be mental health, social care, specialist educational support, or long-term workforce planning.

Priorities

Priority 1: **Helping patients to get a final diagnosis faster;**

The vision of the Framework is for rare disease patients across the UK to get a final diagnosis faster, and for research into previously unrecognised conditions to identify new rare diseases and provide new diagnoses.

Priority 2: **Increasing awareness of rare diseases among healthcare professionals;**

The vision of the Framework is for HCPs to have an increased awareness of rare diseases and use of genomic testing and digital tools to support quicker diagnosis and better patient care.

Priority 3: **Better coordination of care;**

The vision of the Framework is for rare disease patients to experience better coordination of care throughout the patient journey.

Priority 4: **Improving access to specialist care, treatments and drugs;**

The vision of the Framework is for rare disease patients to have improved access to specialist care, treatments and drugs.

Westminster Health Forum Policy Conference

Luke Fraser

Luke attended this conference in January 2021.

Titled *'Next steps for rare diseases and specialised commissioning'*, this was an informative half-day online conference covering the current landscape for rare diseases in the UK and focusing on policy priorities, utilising genomics, patient engagement and co-ordinating care.

The event featured a wide range of speakers including parliamentarians, senior NHS representatives, rare disease advocacy group leaders and industry professionals.

Common threads of discussion throughout the morning included the following:

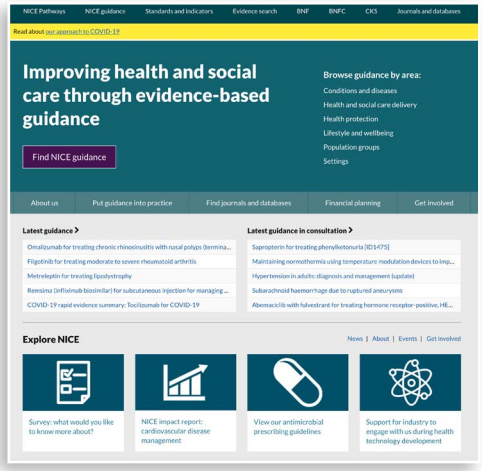
- Assessing the current state of play with regards to the Rare Disease Framework - a government-led strategy outlining how the UK will address the challenges faced by those living with rare diseases over the next 5 years. General consensus was that the creation of the framework is a positive step, but also that it needs to be properly implemented going forwards.
- An awareness of the need to better involve patients in both clinical research, the devising of strategy with regards to rare diseases and an acknowledgement that patients are experts in their own conditions.
- How to progress and integrate the innovations of genomic medicine from data gathering and testing to direct patient care.
- Reducing the 'diagnostic odyssey' for patients with rare diseases and providing better patients access to specialist care.
- How to continue to raise the profile of rare diseases in the UK, bearing in mind that whilst individual conditions may be rare, when taken together they amount to a significant area of focus for policy providers.
- On the impact of Covid-19, from disruption and suspension of healthcare services to the accelerated trend towards remote healthcare appointments, and a discussion of the pros and cons of such an online shift – the rough consensus is that such a model, whilst working for some so far, is not working for others. Diagnostic subtleties may be lost via the online medium, and there are challenges posed by digital poverty and the difficulty some patients may have accessing services online.
- There was also brief discussion on the mental health impacts of the Covid-19 pandemic on those living with rare diseases, many of whom have been advised to shield for significant periods of time.

NICE Technology Appraisals Induction & Training

Gemma Seyfang,
Pompe Support Team

When a new treatment is created NICE (National Institute for Health and Care Excellence) will put it through a process called a technology appraisal, where the health impact, the cost effectiveness, the implications for the quality and length of life, social value and pricing deals are all taken into consideration. If a treatment is approved NICE will issue guidelines on how the treatment should be delivered and managed.

Here is Gemma's report on how patients can become part of and influence the process.



The whole assessment process takes up to one year to get an answer when a new treatment is going through.

The Scoping Stage

Firstly, the *scoping process* is what happens when the questions that are going to be raised during the appraisal and assessment process are checked. This identifies which patient organisations might be involved, which stakeholders and the important outcomes for the treatment, improving quality of life and equality issues. At the evidence-submission stage, family, patient and carer evidence is useful. However, do not include industry evidence, pharmaceutical company evidence or emotive language.

The Patient Expert

NICE is looking for a mixture of broad experience, personal experience and written individual statements. Patients attend as individuals and there are only a certain number of places for patient speakers.

Include written statements about unmet needs and also from people on the trial who have something to say.

Technical Engagement Stage

Industry submit this scientific evidence and a call is held with a technical team, like a pre-committee meeting.

Committee Stage One

This looks at value for money and is based on all evidence given so far.

Approval of a new treatment does not bring any extra money to the NHS to pay for it. So, a new treatment/technology can just add to the budgets being more stretched. You can use international surveys. Committee members attend the meetings, including two lay members alongside NHS commissioning experts.

Public can watch meetings in a public gallery (online).

Committee Stage Two

This looks at final draft guidance. Stakeholders can correct factual errors in the final statement.

Tips

1. Always ask if you have any questions, the PIP team is there to help.
2. Always check deadlines.
3. Look out for a committee meeting so you can join one as an observer prior to your own one coming up.
4. Look at the NICE website for more information: www.nice.org.uk

In Conclusion

I found it useful because I have gained more information and knowledge about the stages and how to contribute. It was good to know that we can watch from the public gallery at a virtual meeting to get some insight.

Oddly, Covid has helped because there are more meetings going online which makes them more accessible for people like me. I have bad eyesight and am nervous of new places so prefer when it is done by Zoom and by webinars; that is really good.

FOR CHILDREN...

Education and Health Care Planning (EHCP)

This is a legal document for a child that is drawn up by a local authority in consultation with the family and the child.

It is drafted after a special educational needs assessment has been completed.

This is likely to be very important for many children living with GSD. Most children have some form of extra help at school.

The Special Educational Needs Co-ordinator in the nursery or school will give you advice and help.

If you are unsure, ask your GSD nurse or phone 0300 123 2790 and AGSD-UK will be able to point you in the right direction.

Sometimes, parents feel it is a hard effort to get the EHCP, so be firm, if you think it is needed for your child.

Professional supporting letters are important; again, contact info@agsd.org.uk and we can help provide or source the right letters.

This handy guide gives good tips on how to compile an EHCP.

SECTION A:

The views, interests and aspirations of the child and their parents

- Outline aspirations and goals for the future
- Include play, health, schooling, independence, friendships, further education and future plans in this section
- Detail how to communicate with the child and his or her parents, and how they will be involved in the decision making
- Include a brief summary of the child's history

SECTION B:

Special educational needs

- Map out all special educational needs

SECTION C:

Health needs which relate to their SEN

- Outline health needs which are related specifically to their SEN
- The Clinical Commissioning Group (CCG) may wish to include other healthcare needs

SECTION D:

Social care needs

- In this section list social care needs identified through the EHC needs assessment which
  - → relate to the child's SEN, or
  - → require provision under section 2 of the Chronically Sick and Disabled Persons Act 1970

SECTION E:

The outcomes sought for the child

- Include a range of projected outcomes with timelines and include provision for monitoring progress

SECTION F:

The special educational provision required

- Provision must be detailed and specific and should be quantified, in terms of the type, hours and frequency of support and level of expertise, and should include where the support is secured through a personal budget
- Flexibility should be allowed in the use of the personal budget, should it be needed, to meet the changing needs of the child
- The plan should specify:
  - → any appropriate facilities and equipment, staffing arrangements and curriculum
  - → any appropriate modifications or exclusions from the application of the National Curriculum
  - → the outcomes to which it relates
  - → speech & language therapy
  - → physiotherapy
  - → occupational therapy
  - → CAMHS services (child and adolescent mental health services)

SECTION G:

Any health provision required

- This section lists the type of health support required and who will provide it
- It may include specialist support and therapies, such as medical treatments and the delivery of medication, occupational therapy and physiotherapy, nursing support, specialist equipment, wheelchairs and continence supplies

SECTION H:

Social care provision

- Include all the services needed for a disabled child, under section 2 of the Chronically Sick and Disabled Persons Act 1970 including
  - → practical assistance in the home
  - → provision or assistance in obtaining recreational and educational facilities at home and outside the home
  - → assistance in travelling to/from recreational and educational facilities
  - → adaptations to the home
  - → provision of meals
  - → provision or assistance in obtaining any special equipment
  - → non-residential short breaks

SECTION I:

Placement

- The name and type of school to be attended by the child
- These details must be included only in the final plan, not the draft plan sent to the child's parents

SECTION J:

Personal budget (including arrangements for direct payments)

- Include detailed information on any personal budget that will be used to secure provision in the EHC plan

- It should set out the arrangements in relation to direct payments as required by education, health and social care regulations
- The special educational needs and outcomes that are to be met by any direct payments must be specified. Any amount of money specified in this section must be enough to secure the provision specified. It is, therefore, essential that type and amount of provision is adequately specified, e.g., as well as amount of time per week, the qualifications and experience and therefore grade of a specialist teacher

SECTION K:

Advice and information

- The advice and information gathered during the EHC needs assessment must be set out in a list in the appendices
- The list should include brief details of who gave the advice and when, e.g., Joanne Donald, NHS physiotherapist, 19 May 2016
- Copies of all the advice and information gathered during the statutory assessment process should be attached to the EHC plan as appendices

For independent EHCP advice contact IPSEA. This is the Independent Provider of Special Education Advice (known as IPSEA), a registered charity operating in England.

IPSEA offers free and independent legally based information, advice and support to help get the right education for children and young people with all kinds of special educational needs and disabilities (SEND).

We also provide training on the SEND legal framework to parents and carers, professionals and other organisations.

If you are having problems, try phoning 0300 123 2790, we might be able to put you in touch with another parent with a child with GSD who has been through the same thing.

SHORT BREAKS

What are 'Short Breaks' for disabled children?

A 'short break' is a chance for an activity or support that offers disabled children an opportunity to have fun, try new activities, socialise, make friends and promote independence. The short break allows families to do positive things together, or supports the child in a safe and secure environment. As a result the parent/carers get a break from their caring responsibilities.

All local education authorities must provide short breaks; they must advertise how to apply for them and help parents and carers to apply. Families should be able to access services using a direct payment (money in your pocket in the form of a grant).

Sources of help & advice on short breaks

Contact a family - 0808 808 3555

Over The Wall Camps for children with medical conditions

During 2017 we were lucky enough to gain funding to send a group of GSD families on an Over The Wall camp.

Since Covid-19 hit they now offer an online version called Camp in the Clouds. This is going to continue for at least a few years. Would your child like to join in? Visit the website for more information.

They have a series of different camps, including some for siblings, running all year, online.

www.otw.org.uk

WHAT IS SPOON THEORY?

Amanda Porter

Spoon Theory may help explain how your GSD affects you.

If you have a GSD, perhaps even late-onset Pompe disease as I do, you might be dealing with muscle weakness, aches and cramps and fatigue. Many people may not understand how overwhelming this can be, and it may not always be easy to explain. However, an analogy called the ***spoon theory*** may help family and friends understand what you go through regularly.

The spoon theory was conceived by a lupus patient, Christine Miserandino. She used it to explain to an inquiring friend what it was like to live with a chronic disease.

What is Spoon Theory?

Energy, for many of us with a chronic illness, is limited and depends on many factors including stress levels, how we're sleeping and pain.

According to the theory, you start each day with 12 theoretical spoons. You have to give up one spoon for each task you perform: brushing your teeth, putting on clothes, visiting the doctor, making dinner etc. When you finish using all the spoons, that's it. You are done for the day!

Healthy people have all the energy necessary to do whatever they need to do in a given day. They, unlike people living with GSD, have a seemingly infinite spoon supply.

The spoon theory illustrates how those with a chronic disease, as well as their caregivers, have a finite amount of energy that must be carefully rationed.

Performing one errand or task can limit what you can do for the rest of the day. In the words of a Pompe patient: *'The main thing for me is that it helps explain to other people about why I have to plan ahead and think carefully about what I'm doing as I don't have the same amount of energy as someone else might have'.*

How do I put the Spoon Theory into use?

Understanding that you have only so much energy renders daily prioritising and planning crucial. Show yourself compassion if you don't complete everything you set out to do. When you've spent all your energy, you are done for the day.

The guilt associated with having a chronic illness is a heavy burden. One of the things the Spoon Theory can help with is that separation between what we'd like to do and what our illness dictates.

Practicing self-care is vital. If part of that means ***using a spoon*** for, say, practising with a walking aid in lieu of another get-together with friends, then so be it. You know your body and needs best. When you've exhausted your spoon set, ask for help from others.

Partnership with Vitaflo Ltd: Living with Hepatic GSDs

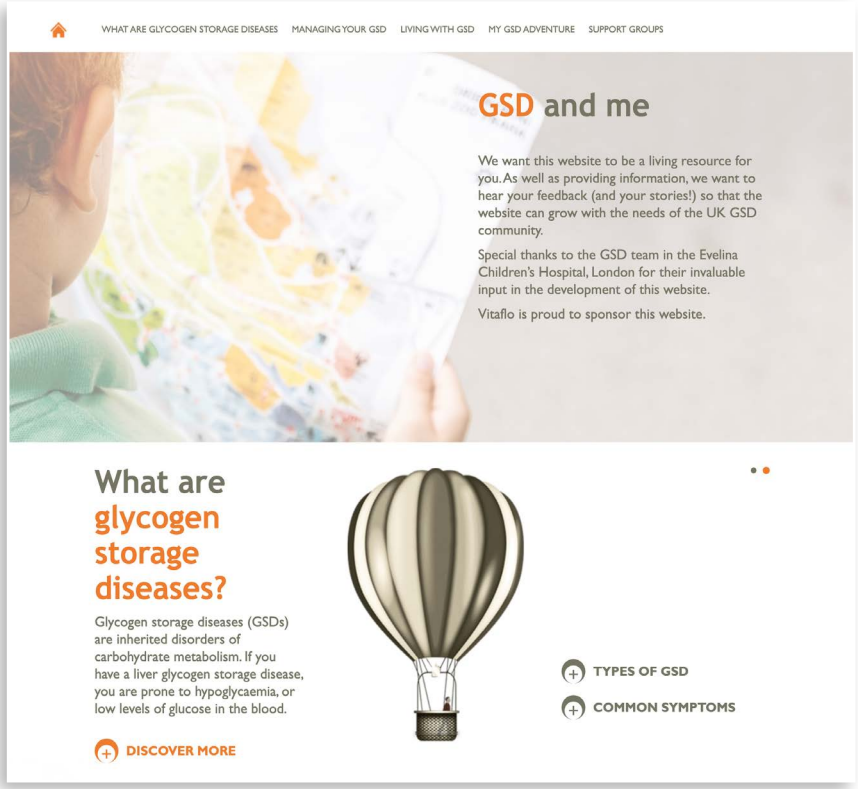
During 2020 we partnered with Vitaflo on a patient survey for our Hepatic GSD members. Ailsa worked with Tiina Heinenon-Kirk to create a questionnaire which was widely circulated. It was a win-win because the total of 55 responses meant that AGSD-UK gained £1100 from Vitaflo as a donation.

Very useful information was gathered and on 1st Feb a focus group of eight patients and carers gathered together to delve deeper in to the themes that were raised.

All this will not only inform AGSD-UK about our members' needs but will help Vitaflo enhance their patient information website. The current version can be found here:

www.gsdandme.com

Jason McMillan, GSD1 Co-ordinator said, *'Good to be part of it and to know that Vitalfo are listening to patients and parents about what is needed and what can help them going forward.'*



Anonymous Industry partner

Also in 2020 we worked alongside another USA based company and gathered together a group of GSD1a patients who joined a focus group discussing how they live with their condition. They were joined by patient advocates, staff and scientists.

Partnership working with Ultragenyx Ltd

In early 2020 we liaised with Ultragenyx in their two endpoint surveys for GSD1a and GSD3. Links to the survey were circulated widely and many members responded with valuable information. Amongst other things, information gathered goes toward helping design trials for new treatments.

An international focus group was convened with Ultragenyx in early 2021, including three UK GSD1a patients.

One participant, CH, said;

'The online focus group lasted around three hours in which we exchanged our thoughts living with GSD1a. During the board I, as well as a few other people from around the world, exchanged our personal experience and stories to Ultragenyx, it was nice to talk and share everything hoping that it will aid in the development of treating GSD1a'

GSD1b prevalence

We responded to a USA enquiry about how many patients there are in the UK with GSD1b. As you can see, there is more activity around GSD1a and b over the last year.

This is why it is so important to have up to date information.

Please, if you have GSD1, or any other GSDs, ensure you are registered with updated details at **www.agsd.org.uk/help-us-help/register-or-join/**

Hepatic Research & Articles

We have done a round-up of some recent research and articles about hepatic GSDs, that were published 2019-2020. If you want to comment or add to them, please get in touch. We would love to hear from you.

1 Glycogen storage disease type VI: clinical course and molecular background.

TIM RJ AEPPLI, JOHANNES HABERLE. EUR J PAED. 2020

This study carried out in Zurich, Switzerland, used an observational retrospective case study to look at the long-term outcomes in six patients with GSD VI or Her's Disease. Small, frequent meals were introduced as well as cornstarch. Following this diet led to an absence of hypoglycaemia. Outcome was excellent in all patients, yet half showed persistent hypertriglyceridaemia.

The use of molecular genetics identified four novel mutations, which adds important information to the understanding of clinical variability in this disease.

2 Diagnosis and management of glycogen storage diseases type VI and IX: a clinical practice resource of The American College of Medical Genetics and Genomics (ACMG).

PRIYA S KISHNANI GENET MED. 2019 APRIL

Guidelines have been produced in America to facilitate the diagnosis and management by health care providers of patients with GSD VI and IX. It is also hoped that the information will help identify gaps in knowledge and suggest future studies. It should be borne in mind that the techniques and tests may vary on an inter-national basis.

3 Neutropenia in glycogen storage disease 1b: outcomes for patients treated with granulocyte colony-stimulating factor.

DAVID C DALE CURR OPIN HEMATOL 2019

Granulocyte colony-stimulating factor (G-CSF) is commonly used to treat neutropaenia in GSD 1b. A co-operative group was formed to look at the long term outcomes of this treatment. In summary G-CSF ameliorates inflammatory bowel symptoms in conjunction with other therapies but the authors recommend that doses should be limited to minimise increased spleen size and abdominal pain.

4 Glycogen storage disease type 1b: role of glucose-6-phosphate transporter in cell metabolism and function

SANG WAN SIM FEBS LETT. 2020 JAN

A collaboration between the Universities of Korea and Connecticut described the broad impact of altered cell metabolism due to a lack of G6PT activity on cell function and considers the prospect for developing novel approaches for treating GSD 1b.

5 Molecular diagnosis of glycogen storage disease 1: a review

ZAHRA BEYZAEL, BITA GERAMIZADEH. EXCLI J 2019 JAN

The authors discuss molecular tests as a platform for the diagnosis of GSD 1, with an aim to overcome the difficulties in diagnosing diseases with broad clinical and genetic heterogeneity.

6 Gene therapy for glycogen storage review

PRIYA S KISHNANI. HUMAN MOL GENET, 2019 OCT

Here is described how the lack of therapies for the GSDs has driven the need for new treatments. The early phase clinical trials for GSD1a and GSD II are described along with proof-of-concept studies for GSD III, IV and V. The future of gene therapy for the GSDs is seen to be promising.

7 Whole-Body Magnetic Body Imagibg in Glycogen Storage Disease Type III

DAVID TOBALY, ROBERT-YVES CARLIER. MUSCLE NERVE 2019 JULY

Fifteen patients, between the ages of 16 and 59 underwent MRI scans. The discussion suggested that whole body muscle imaging provides clinically relevant information regarding muscle involvement in GSD III.

8 Spectrum of amyloglucosidase mutations in Asian Indian patients with glycogen storage disease type III

SHAMA PARVEEN, MADHILIKA KABRA. AM J MED GENET A 2020 MAY

To date, 258 mutations have been found in the amyloglucosidase (AGL) gene, worldwide. Based in Delhi, 24 patients from 21 families with a provisional diagnosis of GSD III were investigated. Eighteen different variants were identified, out of which 78% were novel.

9 Dietary lipids in glycogen storage type III: a systematic literature study, case studies and future recommendations

ALESSANDRO ROSSI, IRENE J HOOGEVEEN. J INHERIT METAB DIS 2020 JUL

Twenty-eight cases of GSD III involving dietary lipid manipulation were identified internationally. The authors suggest a high fat diet may be beneficial in paediatric GSD IIIa patients with cardiac hypertrophy, but careful monitoring for long-term complications is warranted.

10 Liver fibrosis during clinical ascertainment of glycogen storage disease type III: a need for improved and systematic monitoring

CARINE A HALABY, SARAH P YOUNG

The authors present a GSD III liver natural history study and conclude that liver fibrosis can occur at an early age, which may explain the decrease in aminotransferases and Glc4 with age. They suggest this shows the need for systemic follow up and biological and radiological tools to monitor the silent course of the liver disease process.

Message from Ben Parker of the Pompe Support Team

Annual April Stoke Social for Pompe

This year the Stoke Social gathering won't be going ahead, but I hope to arrange some form of meet up, maybe a pub meal outside, but it's all still uncertain and will depend on where we are with the pandemic in the next few months. I hope everyone is keeping safe and well x.

Ben

Hooked on mud

Like many people, Ben has been enjoying new hobbies during lockdown. He said;
'Clay has a created a new chapter in my life which I think will last for the rest of it. I love all aspects of clay, imagining a piece, creating, firing and glazing. It has me hooked on mud!!'

New treatments for Pompe?

For many years Pompe patients have participated in a series of trials for different treatments and therapies.

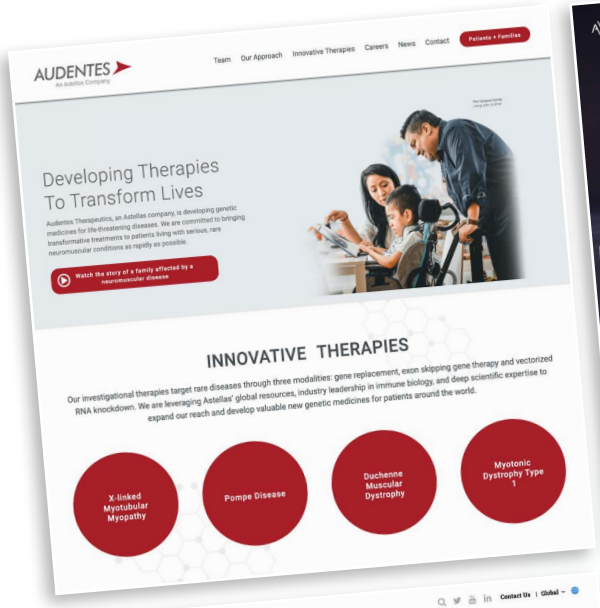
As reported on our website last September, AGSD-UK submitted important information for the National Institute for Care Excellence (NICE) scoping meeting for Sanofi's 'NEO-GAA' ERT and two patient reps attended. We await more news.

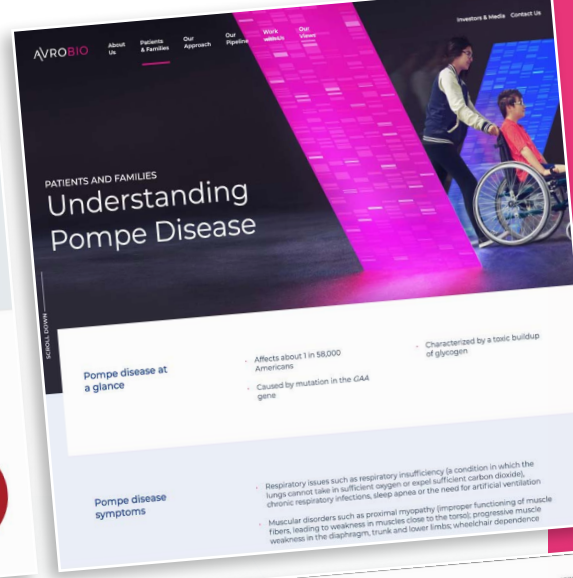
Amicus's ERT offering is an enzyme with a chaperone treatment which has been under trial in the UK. It was awarded Promising Innovative Medicine status by the MHRA. This means it could become available under certain circumstances. Again, we await news.

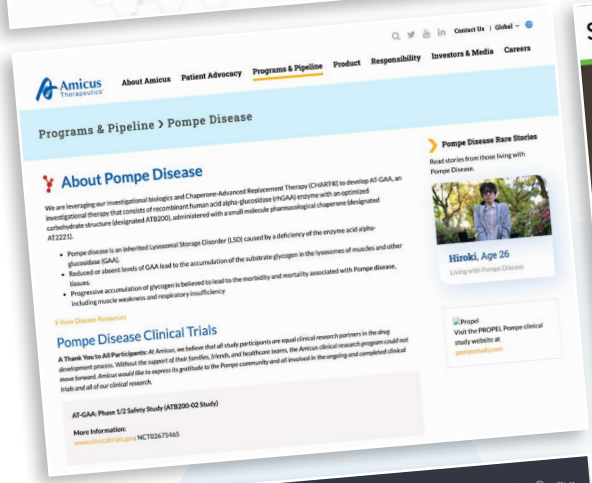
In February 2021 Sparks reported that they had dosed the first patient with a trial gene therapy in the USA. Salford Hospital will be the site for a similar trial in the UK.

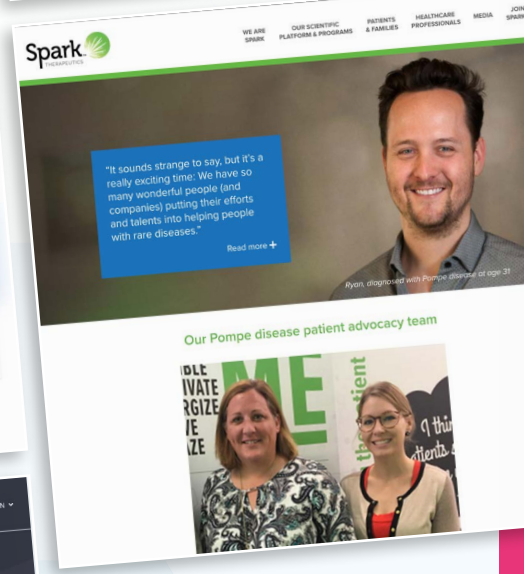
Newcastle Centre for Life is expected to commence the Fortis gene therapy trial soon for Audentes Therapeutics. This is a proof of concept trial with just eight patients globally at first.

View information about clinical trials at www.clinicaltrials.gov website.









Pompe Research Update

Amanda Porter

1 Is exercise alone, or in combination with a high-protein diet, best?

JUNE 2020 ORPHANET JOURNAL OF RARE DISEASES SECHI ET AL.

Synopsis: A high-protein diet associated with moderate-intensity aerobic exercise improved exercise tolerance, muscle enzymes, pulmonary functions and quality of life of late-onset Pompe disease (LOPD) patients chronically treated with ERT.

Background: Enzyme replacement therapy became available for Pompe disease in 2006 and showed to be effective on motor and respiratory functions of LOPD patients. However, ERT does not completely counteract disease progression. Prior to the approval of ERT, many clinicians prescribed diet and exercise to patients with LOPD. After the advent of ERT these life-style interventions were mostly disregarded in clinical practice.

A study evaluating, in LOPD patients who are long term treated with ERT, the effects of exercise alone or combined with a high-protein diet. Measures on indices of exercise tolerance, motor and respiratory functions and quality of life were performed.

Methods: 13 patients were recruited onto the study. Exercise training was conducted at home and consisted of 4 * 1 hour session per week. Each session included 1) warm up exercise. 2) Stretching and balance exercises (10-15mins). 3) Strength training at very moderate loads, using stretch bands. 4) Moderate-intensity aerobic exercise on a cycle ergometer. The diet was individually personalised and based on 25-30% protein, 30-35% carbohydrate, and 35-40% fat.

Results:

- Peak aerobic power (peak pulmonary O2 uptake) increased after exercise, and more markedly after exercise & diet.
- Serum levels of lactate dehydrogenase (LDH) significantly decreased after exercise & diet; both creatine kinase (CK) and LDH levels were significantly reduced after exercise & diet compared to exercise.
- CK is an enzyme that leaks out of damaged muscle. When elevated CK levels are found in a blood sample, it usually means muscle is being destroyed by some abnormal process, such as a muscular dystrophy or inflammation.
- High levels of LDH indicate some form of tissue damage.
- Pulmonary function showed no changes after exercise, whereas a significant improvement of forced expiratory volume in 1 sec (FEV1) was observed after exercise & diet.
- There was a slight improvement in the "mental component" scale after exercise, and a significant improvement in "general health" and "vitality" scales after exercise & diet.
- BMI significantly decreased with exercise & diet although there were no differences in body composition.

Conclusion: Exercise tolerance (as evaluated by peak aerobic power) showed a tendency to decrease in LOPD patients on long-term ERT. Exercise training, particularly if combined with a high-protein diet, could reverse this decrease and result in an improvement, which was accompanied by improved quality of life. The association of the two lifestyle interventions resulted, also, in a reduction of muscle enzyme levels and improved pulmonary function.

2 Chest MRI to diagnose early diaphragmatic weakness in Pompe disease

DR VAN DER PLOEG ET AL

JOURNAL OF RARE DISEASES JAN 2021

Background: Respiratory insufficiency due to muscle weakness is often observed in patients with muscle disease. Respiratory dysfunction in people with Pompe is caused mainly by weakness of the diaphragm, and is demonstrated by a decreased forced vital capacity (FVC) particularly in supine position (lying down). In adults, treatment with enzyme replacement therapy (ERT) resulted in an improved walking distance, muscle strength and a stabilising of respiratory function. However, the effect of ERT is much smaller on respiratory function than on skeletal muscle function.

The international guideline for starting ERT, indicates that ERT should start when patients have skeletal muscle weakness and/or respiratory muscle weakness (defined as FVC of below 80% predicted). However, it is possible that damage to the diaphragm had already become irreversible before ERT was started. Early recognition of diaphragmatic weakness is important to allow a timely start of treatment.

Routine pulmonary function tests, such as vital capacity, FVC and mean inspiratory and expiratory pressures (MIP and MEP) do not differentiate between the function of the diaphragm and the intercostal muscles. Improved insight into the contribution of the different respiratory muscles to inspiration can be provided using spirometry-controller MRI, evaluating the entire diaphragm and thoracic wall during respiratory movements.

Method: A study of 35 people with Pompe disease was conducted to identify early signs of diaphragmatic weakness in Pompe patients. The study included a large group of children and adults with Pompe disease covering the spectrum of disease severity, ranging from those with normal spirometry results to those who need nocturnal ventilation due to respiratory dysfunction.

The study used advanced image-analysis techniques to evaluate the motion and shape of the diaphragm in detail.

Results: 35 of the 53 participants had Pompe disease; the remaining 18 participants were used as the healthy control group. 13 patients had spirometry results in the normal range; none of these patients had significant dyspnea or signs of nocturnal hypoventilation. 22 patients had decreased spirometry results, one of whom used nocturnal non-invasive ventilation due to hypercapnia.

The mean cranial-caudal ration was only 1:32 in patients with decreased spirometry results, 1:60 in patients with normal spirometry results and 1:72 in health controls. Anterior-posterior ratios showed no significant differences. The mean height ratios of the diaphragm curvature were 1:41 in patients with decreased spirometry results, 1:08 in patients with normal spirometry results and 0:82 in healthy controls, indicating an increased curvature of the diaphragm during inspiration in Pompe patients.

Patients with normal spirometry results were younger, had a shorter disease duration since symptom onset, and fewer of them had been treated with ERT than patients with decreased spirometry results. Healthy controls had better FVC, MP and MEP outcomes than Pompe patients. None of the patients had a significant kyphoscoliosis (abnormal curve of the spine) needing surgery or referral to an orthopaedic surgeon.

Conclusion: Even in early-stage Pompe disease, when spirometry results are still within normal range, the motion of the diaphragm is already reduced and the shape is more curved during inspiration. MRI can be used to detect early signs if diaphragmatic weakness in patients with Pompe disease, which might help select patients for early intervention to prevent possible irreversible damage to the diaphragm.

3 Multi-discipline teams for the treatment of Pompe disease

BRIAN MURPHY, PH.D. HEALTH INSIGHTS JAN 2021

Late-onset Pompe disease (LOPD) can affect a number of different organs and systems in the body. If you have the disorder, you will need to see a multidisciplinary treatment team to ensure you receive the best possible care. Below lists the main areas affected and symptoms, and the specialist that can help diagnose and provide treatment.

Muscle weakness: The main symptom of patients with LOPD is muscle weakness, which primarily affect muscles closer to the body (proximal) rather than muscles of the limbs (distal). To help monitor and diagnose any muscle weaknesses you will need a neurologist or neuromuscular specialist.

Everyday life complications: A number of different therapists can help you with various aspects of your daily life. A physical therapist can help you with stretches and exercises to try and maintain muscle strength and flexibility. An occupational therapist can work with you on strategies to perform activities of daily life. You may be able to improve the strength of your breathing with the help of a respiratory therapist. A speech therapist can help you with any difficulties speaking due to weak muscles of the mouth, throat, and diaphragm.

Nutritional requirements: There is some evidence that a high-protein, low-carbohydrate diet may help reduce some symptoms of LOPD. A dietician can help you plan out meals to ensure you are following recommendations and getting adequate nutrition.

Breathing difficulties: The diaphragm and other muscles related to breathing become weak and make it difficult to breathe. Some patients experience sleep-disordered breathing such as apnea. Some may also require breathing assistance through a ventilator. A pulmonologist can help diagnose, monitor, and treat breathing problems.

Mobility impairments: When muscles of the body become weak, you may lose some of your mobility and your ability to maintain your posture. An orthopedist can help diagnose and treat disorders of the joints and bones that are common in LOPD as the disease progresses such as scoliosis (a sideways curvature of the spine), contractors (a shortening of the muscles and tendons), and osteoporosis (a decrease in the density of the bones).

Mental health issues: A chronic, progressive illness such as LOPD can cause you and your family a great deal of stress and anxiety. You may also feel depressed and overwhelmed from time to time. Psychologists can help you learn strategies to deal with these emotions. In more serious cases, psychiatrists are able to prescribe medications.

Feeding issues: As LOPD progresses, you may have difficulty with swallowing. When swallowing becomes too hard, you may need a feeding tube. A gastroenterologist can place this to ensure that you are receiving sufficient

nutrition and are not at risk of accidentally breathing in food or liquid.

Heart problems: While children with infantile-onset Pompe disease often experience severe disease of the heart muscle, this problem appears to be rare in LOPD patients. Heart arrhythmias (irregular heartbeats) may still be a concern, though, and you should consult with a cardiologist for monitoring and possible treatments.

Genetic issues: Genetic counselling can help you better understand LOPD, including its causes and implications. The counsellor can also explain the odds of passing on the disease if you are planning to have children.

4 Pompe Disease and Sleep

SHAH ET AL JOURNAL OF THORACIC DISEASE OCTOBER 2020

Sleep-disordered breathing (SDB) and respiratory failure are complications of Late-onset Pompe disease (LOPD). In untreated LOPD, the most common respiratory failure can have severe consequences.

Diaphragm weakness is the major cause of SDB and respiratory failure. Fatigue, excessive daytime sleepiness and sleep disturbances are also common, and as such health-related quality of life worsens as sleep disturbance increases.

The main treatment for SDB is non-invasive ventilation (NIV), which aims to ensure adequate ventilation, particularly during sleep, and prevent acute hypercapnia failure (too much carbon dioxide, and near normal or not enough oxygen in your blood) and obstructive sleeps apnea. Patients who need NIV but are not receiving treatment are at risk of acute deterioration due to lower respiratory tract infections; effective secretion clearance. Vaccination against common pathogens (viruses, bacteria etc) is also an important facet of care.

Whilst disease-modifying enzyme replacement therapy delays progression of locomotor dysfunction and prolongs life, its effect on respiratory failure and SDB remains unclear. Non-invasive ventilation is proven to significantly improves respiration, oxygenation and sleep quality from the first night of treatment.

5 Establishing Pompe Disease Newborn Screening

JOAN KUTZER, JOURNAL OF NEONATAL SCREENING JULY 2020

We frequently hear that '*it takes a village*' to get things done. In the case of Pompe disease newborn screening (NBS), '*it took the world*'.

Since 1999, an unprecedented and selfless collaboration of countless international experts have been working together to establish a newborn screening programme for Pompe disease.

Two methods for NBS for Pompe disease by measuring acid a-glucosidase in dried blood spots on filter paper were developed in an international collaborative research effort led by Genzyme. Both methods were used successfully in NBS pilot programmes to demonstrate the feasibility of NBS for Pompe disease.

Since 2009, all babies born in Taiwan have been screened for Pompe disease. Pompe disease was added to the Recommended Uniform (Newborn) Screening Panel in the United States in 2015.

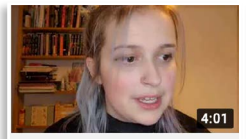
WINTER SEASON McARDLE'S VIDEOS

Andrew Wakelin, McArdle's Coordinator

You are not too late! Although the Season has now finished, the videos will remain available for the long-term. We managed to produce a total of 21 videos on McArdle's (some also relating to other muscle GSDs) for the AGSD-UK Winter Season, which was introduced to replace the annual conference due to Covid-19.

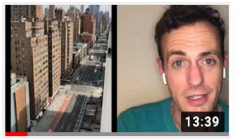
If you have not watched them already please do catch up with them, they vary from the personal and funny to the helpful and educational. They are each well worth a few minutes of your time. There are around 4 hours of watching in total.

As the Season finished at the end of February the McArdle's videos had clocked up over 1500 views. From past experience, this number is likely to double in the coming year. Lucy Smith, age 11, got the highest number of views. Very many thanks to her and ALL those who assisted in the productions, not just in the UK but from overseas as well. Their names are up there in lights (back lit!). Here are a few screen grabs to whet your appetite.

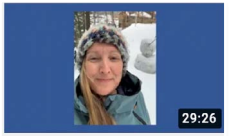


GSD5 McArdle's – Bronte Thomas-Bush – Challenges...
96 views • 4 weeks ago

Muscle GSDs – Celebrating 10 years of the walking...
38 views • 1 month ago



GSD5 McArdle's – Michael Wilson – Building aerobic...
171 views • 1 month ago



Muscle GSDs – IamGSD and International Developments
96 views • 1 month ago

GSD5 McArdle's – Lucy Smith – From McArdle's diagnosis...
263 views • 3 weeks ago

GSD5 McArdle's – Andy Williams – A personalised...
24 views • 1 week ago

Muscle GSDs - Carrie Connors - Lessons from an...
55 views • 2 weeks ago

GSD5 McArdle's – Stacey Reason – My shoelace mus...
23 views • 1 day ago

GSD5 McArdle's - Sioned Hosseinian - My Year with...
75 views • 2 months ago

Muscle GSDs – Andrew Wakelin – Lucky Dip of...
13 views • 1 day ago

IMPACT OF THE COVID-19 PANDEMIC

Andrew Wakelin, McArdle's Coordinator

The big impact has of course been the cancellation of the walking course which was to be held in Snowdonia in 2020, along with the Children & Parent's event. However, we do have a booking in place for the Stackpole Centre in Pembrokeshire for 2021. At the time of going to press it is looking as though all restrictions will be relaxed in time for us to run both events. The course will be 21-28 July and the Children & Parent's event will be 24-28 July.

The introduction of the on-line *Winter Season* in place of the annual conference meant that we finally got to grips with making videos on a number of aspects of McArdle's. This had been considered for some time. The push provided by the pandemic was at least one benefit coming out of the whole episode. We are going to try to continue this initiative in some way, possibly under the IamGSD banner.

Overseas events which I would have attended were also affected, notably the annual conferences in the US and Germany, and the walking course in Germany.

Check out the IamGSD web site for details of an anonymous Covid-19 survey which will help the medical world to understand the impact of the such pandemics on people with muscle GSDs.

The McArdle's clinic has been severely impacted in two ways. Firstly by reallocation of some staff, but also because of the need to switch from face-to-face consultations to online or telephone. There was a brief return to normality during the summer last year, but they very soon had to revert to remote consultations. We are hoping that things will be normalised by summer or autumn this year.

Back in Pembrokeshire for our 2021 walking course

The social aspect is as important as the walking.

More on-line McArdle's coffee mornings?

During the Winter Season we trialed two on-line coffee mornings as a way of people getting together during the pandemic. We had about 16 to 20 people attend each event. Now we are wondering whether this would be a good thing to offer on a regular basis even after the pandemic is long forgotten.

As a rare disease, we are all geographically rather remote from each other. The online get-together is a way of people meeting and sharing experiences, or just having a bit of a laugh. The two which we trialed were on Sunday mornings at 11am for about an hour. It would be interesting to know if people feel this is the best time. Would you like more of these events? On a regular basis? On different subjects?

If you have any comments please do drop an email to Andrew Wakelin on type5@agsd.org.uk.

And in other news...

UK numbers continue upwards

We have had 9 more people get in touch since the last issue of *Glisten*. A few had been recently diagnosed in their teens, but some others were diagnosed many years ago, even up to 45 years, and never told of our existence. Our UK and Ireland numbers are now up to 348 people diagnosed with McArdle's. The breakdown is England 242, Scotland 36, Wales 38, Northern Ireland 14, Isle of Man 2, Ireland 13, ex-pat 3.

Interestingly the diagnosed rate in Wales is about 1 in 84,000, whereas in England it is about 1 in 230,000. It makes me wonder whether the very common R50X mutation started in Wales, I don't suppose we will ever know.

Changes to the genetics service

For about 20 years we have been well served by genetics services in Sheffield and Birmingham and have built relationships with them. However, NHS England has reorganised genomics services into seven main hubs. We now need to build relationships with these laboratories and try to ensure that they pass on our details to the doctors who referred the people who get a positive test result for McArdle's.

Please complete the survey study

The Copenhagen Neuromuscular centre is conducting a big international survey study to investigate the personal experience of people with McArdle's. It investigates the ketogenic diet, physical activity, fatigue, quality of life and sleep patterns. The survey is open internationally and is anonymous. It is important that as many people as possible finish the survey (closing 31 March) for the results to be reliable. See the IamGSD website / Get Involved / McArdle Disease Survey.

Clinical Practice Guidelines – coming soon

A large international group of McArdle's clinicians and researchers is developing Clinical Practice Guidelines under the leadership of Dr Stacey Reason, President of IamGSD. It has been a project of more than a year already, but it is hoped to have it finalised and published in a medical journal during this summer. This represents a very significant advance in the potential for high quality care for people with McArdle's worldwide.

A new condition from the PYGM gene

As an autosomal recessive condition, people with McArdle's have a causative mutation on both copies of the PYGM gene, which they inherit from their parents. Recently a paper has been published on the discovery of a family you have a dominant mutation on the PYGM gene. This means that to be affected they only need this causative mutation on one copy of the gene, so the condition is passed on very much more easily than normal. However, the big surprise is that the particular mutation does not cause McArdle's, but a rather different condition which only develops later in life, compared to McArdle's which is almost always evident from around age 5 to 10. The new condition has not yet been given a name.

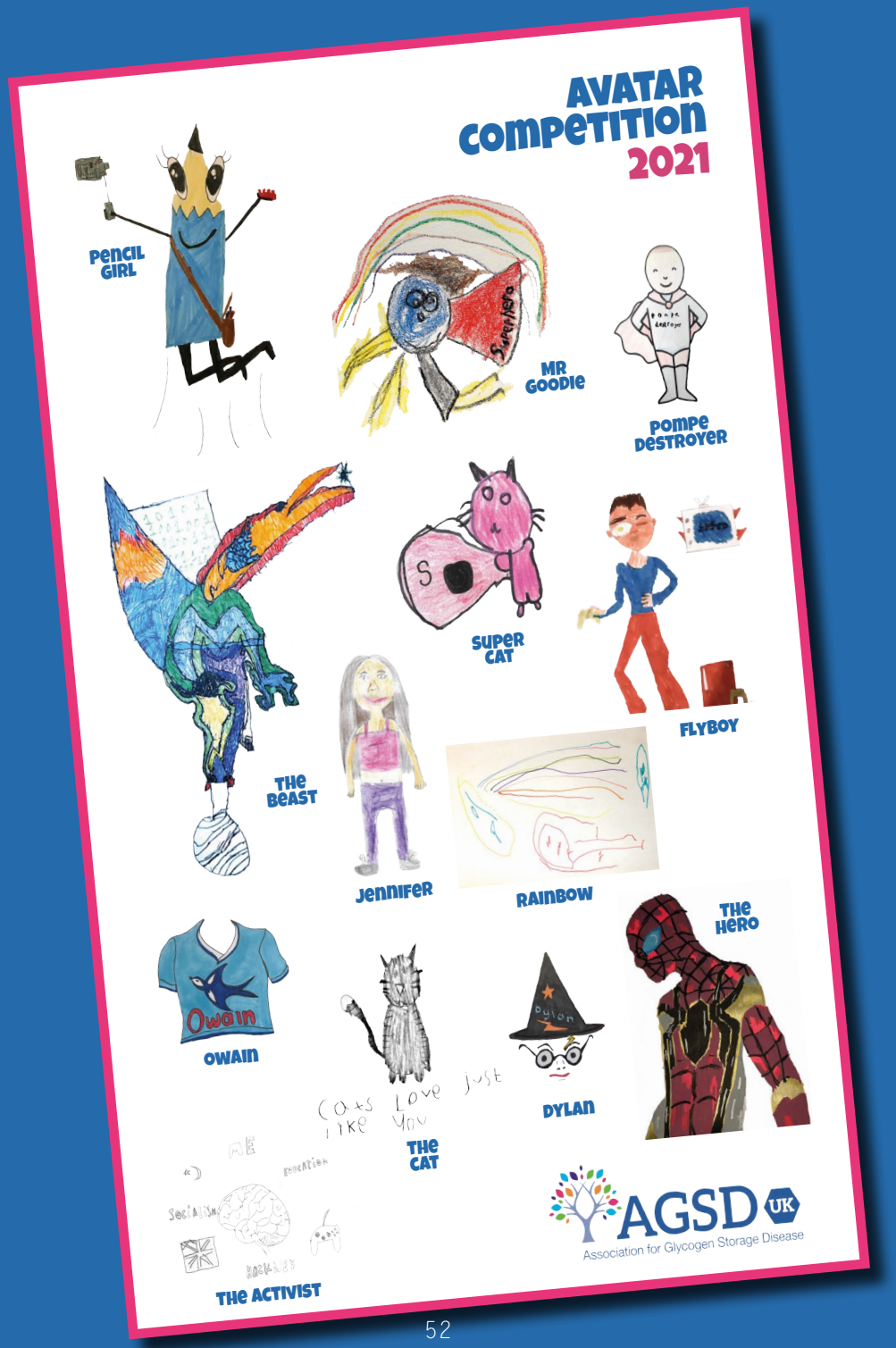
ALJAZEERA

National Trust

Press exposure for McArdle's

Watch out for some press exposure for McArdle's, now and in the near future.

- Published on Rare Disease Day (28 February) is an article on rare diseases, including GSDs, represented by McArdle's. It is online on a website called AlJazeera.com, in their Features section.
- If you are a member of the National Trust (and almost 6 million people are!) keep an eye out for the Summer edition of their magazine. We can't talk about it yet, but there is likely to be a one-page feature with mentions of McArdle's.



CHILDREN'S AVATAR COMPETITION

Results:

Winner! **POPPY**

Second! **MAXIM**

Third! **ABDERRAHMAN**

Well done to everyone who entered the competition. The judges were impressed with your artwork!

We've produced a fundraising teatowel (see opposite page) which can be ordered for £7.00 including postage & packing.

If you'd like to support us in this fundraiser, please contact the team at

info@agsd.org.uk

Thanks to our industry sponsors for their support and grants in the past year.

Sanofi Genzyme Ltd
Sparks Therapeutics (Roche)
Audentes (Astellas)
Amicus Therapeutics
Vitaflo UK
Ultragenyx

AGSD UK
Association for Glycogen Storage Disease

TAKE AWAY FROM THIS GLISTEN...

AMAZING!

Hugo's family fundraising for AGSD-UK

Flo, Jenny, Mads, Lauren, Nic, Dale, Nathan & Andy will all be #HikingForHugo and climbing the three highest peaks of Scotland, England and Wales within 24 hours.

https://uk.virginmoneygiving.com/HikingForHugo

- PLEASE help us with fundraising and donations
- 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.

OFFICE CONTACT DETAILS:

AGSD-UK, PO Box 1232,
Bristol, BS48 9DD
Phone 0300 123 2790

Email info@agsd.org.uk

Charity number 1132271

Emma Grove Design 07990 520 807

AGSD UK
Association for Glycogen Storage Disease