Glisten

Glycogen Storage News

AGSD UK
Association for Glycogen Storage Disease

SPRING 2018

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CHAIRMAN'S UPDATE

Welcome to the latest edition of Glisten. Since our last edition has been another very busy time for the Trustee Directors, staff and volunteers. As always the financial environment for all UK charities, particularly for those smaller rare disease such as ourselves, remains challenging. Despite this we have continued to deliver sustainable year-on-year financial improvement helping us deliver services to our beneficiaries.

Since the conference last year the trustees have held one trustee board meeting which was in January 2018 with the next one scheduled for early June. In addition a number of trustee board sub committees met throughout the year. We have had one resignation from the trustee board, Phil Prosser, due to family and personal pressures and I would like to place on record our thanks to Phil. However I am delighted to report that we have appointed three new Trustee Directors; Jane Guy OBE, Heneage Legge-Bourke and Ailsa Arthur, with another one, Rob Seabourne about to come on board. One of our major objectives was to further strengthen the trustee board and I believe that we now have a group of committed trustees with a broad mix of backgrounds and appropriate skill sets.

As you will see from the content of this edition there has been a significant amount of activity going on and I am never ceased to be amazed at the positivity, creativity and energy of our members, volunteers and staff.

On the flip side it is unfortunate that the spotlight has fallen on the charitable sector recently for the wrong reasons – aggressive fundraising tactics, data breaches and the safeguarding issues come to mind. All of these serious issues remind us of the need to ensure that we have appropriate policies and governance in place to protect individuals. Having the appropriate levels of governance in place is always a larger burden on smaller charities but we are committed to ensuring that our practices and procedures are commensurate with the values of AGSD-UK, are robust enough to allow early identification and treatment of issues and provide appropriate assurance to our funders and donors.

I am never ceased to be amazed at the positivity, creativity and energy of our members, volunteers and staff

As a small charity supporting a number of ultra rare conditions we will always be a long way down the queue in terms of people's awareness of who we are and what we do. However we have a track record of punching well above our weight and we will continue to strive to beat the drum for the glycogen storage disease community in the UK.

CHARITY DIRECTOR'S MESSAGE

Trying to explain Glycogen Storage Disease in simple terms to the non-medical lay-person is something that I often struggle with, mainly because I try to include all forms of the condition; liver and muscle. So, things have just got a little harder for me; I and the trustees have decided that AGSD-UK should support all conditions that are medically described as glycogen storage diseases. So, Danon disease (GSD 2b) and Lafora disease will soon be given sections on the new website, once it is launched.

Through internet searches and talking with biotechnology companies, I recently learned three new words: Polyglucosan, polyglucan and polyglusan. It seems, however, that they all mean the same thing: Polyglucosan bodies are accumulations of abnormally formed glycogen and are present in four different GSDs: Late-onset GSD 4 (Adult Polyglucosan Body Disease), GSD 7 (Tarui Disease), GSD 15, and Lafora disease. It seems that GSD terminology is also occasionally abnormally formed!

RCGP eLearning Module for GSD is Launched

I am very pleased to report that a Glycogen Storage Disease course is now available on the Royal College of General Practitioner's website. Developed in collaboration with AGSD-UK and Funded through a Sanofi-Genzyme Patient Advocacy Leadership (PAL) Award, the course was designed to educate primary health professionals. It is available to anyone with an interest in the conditions.

The course comprises two modules, Hepatic GSD and Muscle GSD. There wasn't space to include a huge amount of detail on all GSDs, but hopefully the course will raise awareness amongst primary care healthcare professionals. We will be monitoring its use and I'll be able to provide feedback in due course.

The muscle and hepatic GSD modules can be accessed here: http://elearning.rcgp.org.uk/course/view.php?id=269

You can sign up to the platform by completing the registration form here: www.rcgp.org.uk/my-rcgp/basic-registration.aspx

Trying to keep up with medical terms is one problem, but more recently my problem is trying to keep up with the wide range of activities within the GSD community; the formation of small active groups to support our GSD Coordinators is something I have often promoted, and so I am very pleased to welcome CATS to support our new Cori-disease coordinator, Sylvia Wilson, and also the prospect of volunteers supporting Andrew Wakelin at McArdle Clinics. On the fundraising front, I'm very envious of fundraiser Lucy Wright in her John O'Groats to Lands' End cycle challenge; but not so envious of the Faffing Frosties who'll be swimming the English Channel in June, but best of luck to you all.

My sincere thanks go to the GSD community who support AGSD-UK ambitions in so many different ways; you can read about some of them in this issue. If that inspires you, please let me know how we can support your ideas for the year ahead.

SPECIALIST CARE ADVISOR

Jane Lewthwaite

The Care Advisor role has now existed for two years and in that time is has developed in response to requests from people affected by Pompe disease, and so it is largely led by the people who use it. The Care Advisor reports monthly to the Pompe Support Team (PST) and also provides ideas and suggestions.

- ***INDIVIDUAL HELP*** accessing grants, benefits, meetings and the right answer to a problem. In the last year over £5000 was obtained for individuals through grants.
- ***TIME TO TALK*** or chew over a difficult issue and reflect on solutions. The Care Advisor meets and talks to many people with Pompe and tries to share and pass on knowledge, tips and ideas from one to another.
- ***INFORMATION AND PUBLICATIONS***. Two mailshots have been sent recently. One including the new Risk Alert Card and the second for the new Medical Overview leaflet for Pompe disease.
- ***LINKING TO SPECIALIST CENTRES*** to iron out communication difficulties and help create better services.
- ***OUTREACH and SUPPORT*** during and after diagnosis. New contacts come via Facebook, email or referrals from the NHS specialist centres. During this anxious time the Specialist Care Advisor can listen, counsel and send out materials, answer questions and put people in touch with someone who has Pompe local to them.
- ***IMPROVING DIAGNOSIS AND RAISING AWARENESS***. The Care Advisor helps to promote knowledge and understanding of Pompe disease and ensure the Pompe voice is heard. Recently, members with Pompe have given eight talks or attended meetings as Patient Advocates, with more talks planned for the future.

Benefit Claims

Whether you can work or not, living with a disability brings many extra costs with it, especially travelling or home adaptations.

In the last year, 28 members were supported in making applications for disability benefits and a few more were helped to obtain full benefits checks or local dedicated advice. These awards have succeeded in obtaining benefits to an annual value of just over £100,000.

OUR WORK WITH CHILDREN

GSD Family Days

In the last year, we have organised GSD Family days at Great Ormond Street, Taunton and Birmingham. We provide craft and fun for the children and include local staff from the medical centres who have time to talk with everyone. Children get a lot from these events, especially because they can meet others with the same rare condition.

Teenagers Project for all GSDs

In 2018 we will run an event for young people which will be designed by the young people themselves. We have obtained funding for a small project. For example, we could aim to create a video presentation which to be given to schools or medical staff explaining GSDs.

Should we organise a meeting or run it online? Contact Jane Lewthwaite with your ideas and if you would like to be involved.

Books and Resources

The last year has seen new resources. Great Ormond Street Hospital together with Genzyme and AGSD-UK created the new Infantile Onset Pompe Disease School Resource pack with sections for the school and sections for families.

AGSD-UK also created a book designed for under 7's called Pompedoo.

Involving young people

Please let me know how you think young people could be included?

Recently one of our young members aged 12 who has Pompe disease went on a protest against cuts to Care Budgets. Local Councillors talked to him, invited him in to their meeting at which he spoke, off the cuff, about what care means to him and his family. He was so persuasive and eloquent that he has been invited back.

Trustees and young people

Trustees are ready to welcome young visitors to all or part of a Trustee meeting so they can see how the charity runs. Would you like to have your say at a Trustee meeting?

Free Kids Camp

In September 2018, we have free places at an "Over The Wall" kids camp. See the following pages for more details. Four families have already booked but there are still places available.

Over The Wall has partnered with AGSD-UK to provide:

A FREE AND FUN ACTIVITY CAMP FOR FAMILIES LIVING WITH GLYCOGEN STORAGE DISEASE

Specialist Camps for UK Charities

What & Who?

A **free**, 2 night residential camp for families who have a child or teenager who are part of the AGSD-UK community.

When & Where?

Fri 28 - Sun 30 September 2018 at Whitemoor Lakes, Lichfield, Staffordshire. WS13 8QT

info@otw.org.uk
02392 477 110

Sponsored by Shire

SYLVIA WILSON

Autobiography of our new GSD III Coordinator

I had hoped that retirement from a lifetime of teaching would prove relaxing and rewarding but being of 'the sandwich generation' that hasn't been the case. I think the 'relaxing' element disappeared six years ago when a family member was diagnosed with GSD III, Cori disease, and I first became involved with supporting AGSD-UK but the 'rewarding' element has well and truly been met.

I live in Suffolk with my husband and a rescue dog. If it counts as 'relaxation' I am actively involved with two male choirs, the St Edmundsbury Male Voice Choir and the Edmund Octet, both of whom have helped to raise money for AGSD- UK. (In case you're wondering about the male bit.... I'm the page turner!)

I have been so inspired by the commitment of Esther James and her AGSD-UK colleagues and I hope I can make a useful contribution to the team.

RARE DISEASE DAY

Each leap-year, Rare Disease Day is held on the rarest day, 29th February; on other years, it is celebrated on February 28th throughout the world.

AGSD-UK had three willing participants in a major promotional and awareness raising event at the Houses of Parliament in London this year.

Sanofi-Genzyme organised a "One Voice Choir" for the occasion, singing in Westminster Underground Station. Choristers included Jayne Spink, CEO of Genetic Alliance UK, parliamentarians and advocates for different rare conditions.

Bronte Thomas, Ailsa Arthur and Alex Fraser sang their hearts out for AGSD-UK, and gave press interviews during rehearsals.

Bronte Thomas said "I attended the event to raise awareness and create publicity for McArdle's disease, the rare disease that I have been diagnosed with. It was an enjoyable day, where I met many interesting people and learnt so much about other's rare diseases. My rare disease means that I am different and it was encouraging to meet others who are different like me."

PGD

Preimplantation Genetic Diagnosis

The Human Fertilisation and Embryology Authority (HFEA) has received an application to licence preimplantation genetic diagnosis (PGD) for glycogen storage disorder type 3. This means that a couple in the UK with a family history of the condition have applied to use PGD to conceive a child who would be free from the condition. You can find out more about the reproductive technique, preimplantation genetic diagnosis, here: www.geneticalliance.org.uk/information/services-and-testing

The HFEA call for information on this condition can be viewed here: www.hfea.gov.uk/pgd-conditions Type "Glycogen" into the Condition search box

This table shows all PGD conditions currently approved and awaiting consideration by the HFEA.

Glycogen Storage Disease Type Ia	Von Gierke disease	approved
Glycogen Storage Disease II	Pompe Disease (early onset)	approved
Glycogen Storage Disorder Type III	Cori disease	awaiting approval
Glycogen Storage Disease Type IV	GSD 4	approved

POLICY DEVELOPMENT UPDATE

Jane Guy OBE, Trustee

Trustees have spent considerable time researching and agreeing new policies and our staff and volunteers have worked hard to implement them. We are not quite there yet but work on the most important ones, where we must comply with government legislation and protect our membership, is complete. These are:

- Acceptable use of ICT
- Conflicts of interest (mainly to ensure trustees are transparent with any related loyalties)
- Contracts of employment (for our staff)
- Financial procedures (to ensure financial stability and probity)
- Handling complaints
- Risk assessment (to minimize risks to people and AGSD-UK)
- Safeguarding (particularly important for us as we support very vulnerable people)
- Equality and Diversity
- Lone Working
- Whistle Blowing

We are also preparing for the new legislation around storing personal data and will soon be surveying all our stakeholders and advising them on what we store, how it is stored and shared/not shared.

We are now working to implement the final group of policies which are:

- Dignity at Work
- Expenses
- Grievance
- Supervision and Support (for staff and volunteers)
- Volunteer Performance
- TOIL (Time off in lieu, i.e. in return for working longer hours than staff should)

These should all be implemented before the summer of 2018

AGSD UK Charity Governance

We have decided it would be a good idea to include something about charity governance in each issue of Glisten. It is not always the most interesting topic, however, it is very important and we want to ensure everyone is up to date on our progress and improvements.

Safeguarding

In Glisten Summer 2017 we reminded everyone about our Safeguarding policy. Dan Machin is the Designated Safeguarding Lead and Jane Lewthwaite is the day-to-day safeguarding officer. This means that both have to undertake training in protection of children and vulnerable adults. For example, our Safeguarding policy means that we check carefully on the standards, training and protocols of the Crèche providers at Conference.

We have renewed our method for Disclosure and Barring Service (DBS) checks. If you volunteer or work for AGSD-UK, you will have been asked about the type of work you do and whether this means you need a basic or enhanced DBS check. If you have any questions and would like to discuss this further, please contact Jane Lewthwaite.

Equality and Diversity

It is a legal requirement to monitor our work to ensure AGSD UK maintains the highest standards for equality and diversity. However, for AGSD UK Trustees and staff it is also an important part of our mission and a core part of the charities ethos that we are inclusive and non-discriminatory. All staff and volunteers have had a copy of our Equalities and Diversity Policy. We are planning how we can include as many people as possible in important Equalities and Diversity training.

Equality and Diversity statement

AGSD-UK is committed to providing a supportive and inclusive culture for all those who need our services, our volunteers, our staff and other stakeholders. We recognise the positive value of diversity, promoting equality and fairness, and challenging discrimination.

We will not discriminate or tolerate discriminatory behaviour on the grounds of race, colour, sex, gender identity (trans-gender), disability, nationality, national or ethnic origin, religion or belief, marital/partnership or family status, caring responsibilities, sexual orientation, age, social class, educational background, employment status, working pattern, trade union membership or any other factor.

GDPR – General Data Protection Regulation

In the last few months you have probably been contacted by organisations and service providers to update your permission to store and use your personal information.

AGSD-UK must also comply with these new rules and all people registered with us will soon get a letter or email, please help us by responding.

CALENDAR ART COMPETITION

AGSD-UK member Gemma Seyfang is designing a 2019 calendar for AGSD-UK as a fundraiser and to raise awareness of Glycogen Storage Disease. We expect the calendars to be on sale from September 2018, but we need our members and supporters to provide the images and words.

Message from Gemma:
Rare as gold dust, that's what we are Join the challenge to be a star!

Please send in your photographs and stories, quotes and poems to reflect one or more of:
Achievement
Goal
Success
Determination

Photographs can be of any subject and should have a brief description explaining why you have chosen your particular composition.

PRIZES

The best 13 photographs and the best 13 quotes/stories/poems will be chosen to appear in the 2019 calendar (12 months + cover).

The overall winning entries in the over-18 category and the under-18 category, will each receive a free calendar.

ADDRESS FOR ENTRIES

Please reply send entries by email to info@agsd.org.uk,
or by post to the office address

Closing date for entries is JULY 31 2018

Winning entries will be notified before September 1 2018.

Please include with your entry:

- Name, Age (if under 18), Address and Phone Number.
- Under 18s need confirmation of parental approval to be included.

Competition is open to anyone – but you must be registered with AGSD-UK. Entrants can register through our website here: www.agsd.org.uk/tabid/1010/Default.aspx

Independent judges will be selected from AGSD-UK supporters with no connection to any particular GSD or individual entrant.

AGSD-UK CONFERENCE 2018

Weekend of
20 and 21 October 2018

Wyboston Lakes
Executive Centre
Great North Road, Wyboston,
Bedfordshire, MK44 3AL

The AGSD-UK annual conference will return to the Executive Centre at the Wyboston Lakes Resort. The centre is currently being refurbished and so we can expect a larger conference area than our last visit, and a bamboo conservatory – no pandas please!

We expect to have welcome refreshments at 11am on Saturday for those arriving early. The conference itself will start with lunch at 12 noon on Saturday and close with lunch at 1pm Sunday.

There accessible rooms in the Executive Centre accommodation block, but they have small bathrooms without showers. Those in the Training Centre, nearby, have better access and better bathrooms. The Training centre is a short drive away, or a long walk.

Bedrooms can accommodate two adults and one child, so we will need to make special arrangements when more than one child is attending.

Further information about making reservations, workshop programmes and travel directions will be updated on our website as the arrangements develop. Go to www.agsd.org.uk and follow links to the conference page.

If you don't have access to the internet, please contact the Droxford Office for further information and booking forms.

CONFERENCE 2017 REPORTS

AGSD UK Conference

October 28th–29th 2017
East Midlands Conference Centre, Birmingham.

Once again we enjoyed a highly enjoyable and informative conference in a most impressive conference centre. Feedback was overwhelmingly positive, but we do need to work harder to provide meals suitable for all delegates.

Below are a collection of notes and reports from the hepatic and Pompe workshops.

We would like to extend our great thanks to the speakers and contributors who gave up their valuable time to be with us during the conference.

Hepatic Workshop Report

Dan Machin, GSD IX Coordinator

This report covers GSD I, III, IV, IX and O.

Clinical Research on hepatic GSD

Irene Hoogeveen from Groningen in the Netherlands

Summary

Irene outlined some of the research and projects that are being done by the UMC Groningen Centre in The Netherlands. These include the GSD Communication platform, their Ketones Trial and The Liver GSD Priority Setting Partnership.

Points that may be of interest

- In the Netherlands they have developed The GSD Communication Platform that combines two parts: The Dutch language GSD App (for patients) and the GSD Clinical Dashboard (for health-care professionals, in English) The App records prescribed and registered diets, data from both manual readings and automatic retrieval from continuous glucose monitor devices as well as results from activity wearables.
- The dashboard is more of an analysis tool that allows more professional evaluation of the recorded information. This allows professionals to analyse the data and draw conclusions and therefore advice based upon it.
- It was devised to try and improve the accuracy of measurements that are more accurate at home compared with those that might be seen in hospital.
- The Ketones trial provided evidence that supported the idea that GSD type 3 patients that had liver disease in childhood, developed into muscle problems in adulthood. A lot of preliminary research was done in the Faroe Islands in 2015 where they use a very high protein diet. They got patients to do a series of visits to do various physical tests to compare against blood data.
- In Feb 2017 they got permission to start sampling patients. They are working with 6 subjects. One test was to use a cycling type machine while in a MRI machine. It analysed muscle metabolism. These tests will try

to compare the difference between the use of a high protein diet versus carbohydrate as the main fuel source. Full results were not available at the time of the conference.

- Liver GSD Priority Setting Partnership. This is a means to encourage the GSD community to submit research questions to the group. These would be reviewed and they hope to create a top ten list of research priorities. Anyone can submit questions to the group.

http://igsdpsp.com/

Ciara Harkins

Personal Experiences of dealing with GSD

Ciara Harkins

Summary

Ciara provided an entertaining and uplifting presentation about her experiences through childhood and in particular in the last year after significant changes to her lifestyle that have led to improvements in her growth and general health.

Points that may be of interest

- Ciara is 17 years old, has Type 1 and lives in Newcastle.
- She noted how valuable the conference was last year, where she spoke to Dr Weinstein who gave her advice on how to improve her treatment and diet. Improvements she has seen included reduction in liver size, gain in height (6cm in a year) and improvement in hair strength and quality.
- The talk was great and the audience were impressed with her courage and the delivery of her personal and incredibly optimistic tale.

Hepatic GSD Research Update

Prof David Weinstein, Connecticut Children's Medical Centre

Summary

Dr Weinstein spoke about both his reflections on returning to the AGSD UK Conference in both 2002 and again now in 2017. He later outlined some recent research in the field of GSD and the prospective projects that may be coming soon.

Points that may be of interest

- He explained how gene therapy works. The method that he explained uses the benign virus AAV8 that transports an artificially created gene to the liver. The AAV8 virus is used because it is a practically harmless virus that only targets the liver and therefore perfect for specific transportation. This gene then can unlock the abnormal gene functionality and start the ability to break down Glycogen.

- He explained how this process was broken down over the recent years:

Step 1: Tests on animals were started (GSD 1a mice 1999-2005 and in Dogs 2005-2016) both have been successful and protected them against complications later in in life. He showed a compelling video of a type 1a mouse who fasted to 2hr 20mins before going into a hypoglycaemic coma. After treatment with gene therapy it fasted for at least 48hrs without problems. The test was only stopped as the mouse was starting to lose weight unnecessarily.

Dimension was a company that asked people to submit suggestions for gene therapy projects. After considering all applications they chose GSD as the one they would work on. This unlocked funding for human trials.

Step 2: Creation of a genetic protocol for human trials. A board was created of international experts that had to be comfortable with the proposed protocol. All 8 experts agreed that the project should be granted permission.

Step 3: The protocol was submitted to a NIH committee in America who had to agree with the project. This was approved in June 2017.

Step 4: An application was submitted to the FDA. If this is approved they should be able to start trials in Feb 2018.

The potential of the trial's outcome and financial viability has meant that Dimension has now been bought out by another company for a much inflated price which shows the interest in the project.

Prof David Weinstein

- New research – Glyde trial – This will attempt to show how Glycosade compares to CS in the day time as well as night time.
- Viking Therapeutics – They have created a drug that lowers triglycerides by up to 79%. This is a problem with many GSDs and can be transferred to the general population.
- There has been research in GSD 1b including the use of gene therapy in mice. These have been successful however, they only fixed the liver problems but the mice still had neutropenia in the bone marrow. Scientists believe that they can take stem cells from these mice and later in turn use these in gene therapy trials to tackle the bone marrow too.
- GSD III – There are plans to test new drinks to prevent muscle damage.
- GSD IV – Dover Lifesciences have developed a drug that blocks glycogen formation.
- GSD VI an IX There have been mouse models created for VI and IX b and there are plans for IXa in 2018.
- Teams at Viking Therapeutics are also trying to create treatments that prevent complications and liver scarring.

Sick Day Management and Kids Camps

Iris Ferrecchia, Connecticut Children's Medical Centre

Summary

This was a general advice session for suffers and families. She discussed the influence and effect of hypoglycaemia. She outlined the way it felt for different people including the pain or discomfort felt in the feet. She also compared different foods as a means to tackle hypos.

Points that may be of interest

- Iris explained how some foods are better for recovery from hypos. Carbohydrates are the fastest but you need to recover to 4.1mmol/L with a quick glucose product such as a glucose gel and then treat with slower release food to maintain the levels in the normal range.
- She also discussed the importance of having medical bracelets or chains to inform a member of the pupil or medical professionals in the case of emergency.
- Iris finished by showing a video about GSD camps that are run in the US to get families together to share stories and build relationships and friendships within the community.
- At the end of the talk there was a discussion about the most accurate Blood Glucose monitors. Freestyle Lite or Freestyle Freedom are generally thought to be reliable and accurate.

Question and answer session

Question and Answer session

Summary

The professionals in the room were invited up to the front to answer questions fielded by the audience. Here are some of the questions, answers or general topics that were raised.

Points that may be of interest

- The panel stressed the importance of blood glucose monitoring.
- Someone asked Dr Weinstein who would be included in the human trials for gene therapy starting in 2018, which was discussed earlier in the day. Dr Weinstein explained that there would be 9 subjects 18yrs and above from around the world: 3 at Connecticut, and the remaining subjects would be somewhere in Canada, UK or Europe. There were no confirmed locations as yet. The tests will increase dosage gradually after each run was analysed and approval gained

for the next higher dosage. Importantly, subjects would be only chosen if they had their treatment in good control and no noticeable internal damage to their liver or kidneys.

- It was raised that there is still great discrepancies between medical establishments in this country and around the world, especially with regards to the use of sugars in the diet. Dr W reassured people that there are many good doctors and you should discuss ideas with your doctor.
- The panel explained the importance of treating with protein with the ketotic types 0, 3, 6,9 to promote and maintain growth. They also explained that ketotic GSDs can eat fruits and some sugars but not excessive amounts. Whereas it is not advised for GSD 1 unless in incredibly small amounts, but should be avoided whenever possible.
- It was discussed that CS or Glycosade can take between 20- 60mins to work so it is not always good for use in sports. They recommended lower impact exercise for ketotoc types rather than sprinting or weightlifting.
- When swimming you can take CS or Glycosade 20 mins before sport and possibly to periodically sip SOS or some similar faster release drink if swimming for a longer period of time.
- Protein is very important to ketotic types and should not affect the kidneys as long as the creatinine levels are normal. They recommend BUN (Blood Urea Nitrogen) forms of measurement.
- They recommended patients to find out when their ketones are high. They should not be above 0.3mmol/L and it was still important to maintain Blood Glucose in the normal range.
- It was recommended that patients have a higher level of protein intake when trying to catch up growth or during puberty.
- It is important to get all the lab results available, and seek to get everything balanced.
- The room was reminded that alcohol is very dangerous to GSD 3, 6, 9; when drunk, such people cannot process energy properly and can behave more like GSD 1 which is very dangerous.
- It was discussed how many patients will not feel affected by low BG down to low 2mmol as they are used to it and will adapt the way it feels. However it is only disguising the underlying damage being done by the other metabolic issues at play.

- The Freestyle Libre Continuous monitor was recommended as it was a simple and easy way of plotting highs and lows throughout the day. These are available as a starter kit of a

monitor and 2 pads at around £160. The patient put the sensor pad on their arm for 2 weeks and measure the BG every 5minutes. The monitor simply needs to be held close to the sensor and downloads the information, turning it into a series of tables or graphs to show highs and lows over the 2 week period.

- It was recommended that Type 9 patients should take up to 4g of protein per kg body weight per day. However, this depends on other lab results being in the normal range. This protein can be either dietary or supplementary such as whey protein, Renapro etc.

Paediatric GSD

Efstathia Chronopoulou, Bristol Royal Children's Hospital

Summary

Efstathia outlined a number of case studies that she had worked with over the last year or so and explained their successes and points of interest to the group.

Points that may be of interest

- Efstathia works at Bristol Children's Hospital and worked with a number of patients including one type III with whom she was not happy with her careplan. Last year she attended the AGSD UK conference in order to research further and has since adapted the way she was treated. The young girl is now on about 4g/kg body weight/day (including synthetic powder) and is now doing well with good lab results.
- Dr Chronopoulou also outlined details for a girl with GSD Ia and a girl with GSD IIIb.

She then explained the way that diagnosis and treatment differs to historical methods. Also how they use monitoring at Bristol CH. She discussed:

- Continuous Glucose Monitoring every 6 months up to the age of 5 years and over 5 years old they use continuous monitoring annually (or more frequently if clinical indicated)
- They complete a 24 hour pre-prandial profile annually
- UCCS load test annually. Regular weight, height and BMI
- They complete a Nutritional Profile every 3-6 months
- They believe that transplants do not help renal disease or neutropenia
- Upcoming/potential strategies they promote
- Medium chain triglycerides and ketogenic diet for GSD III
- Gene therapy GSD Ia, GSD III, GSD IV

At the end there was a discussion about the pros and cons of overnight feeding pumps. Dr Weinstein avoids overnight feeds as it can cause peaks

and troughs once they come off the feed due to the elevated insulin during the night.

Some people raised the issue of damage to teeth due to overnight CS feeds or other products. A study in the States found no significant link to tooth cavities. However, a Fluoride coating is available to help prevent damage.

Multivitamins

Kathy Ross, a dietician at Connecticut Children's Hospital, said that mixing fruitivits or other vitamin supplements with CS feed can be a problem. The CS can inhibit the uptake of the vitamins, especially iron and calcium.

Liver transplants

Dr Weinstein said that liver transplants should not be necessary if patients are under optimal metabolic control.

Promises and challenges in the development of a gene therapy approach for GSDIII

Dr Guiseppe Ronzetti, Genethon

Summary

Dr Ronzetti explained how his project was developing gene therapy techniques for GSD type III. He outlined the history of the project, through their routes for funding to early experimental results.

Points that may be of Interest

- He worked for a company called Genethon but he project was part funded by AFM Telethon – a fund raising organisation that focuses on muscle disease.
- Their research shows there is link between age and reduced muscle Function. This led to them investigating gene therapy as a treatment.
- They use non-infectious AAV Vectors to transfer the genetic material into the liver.
- They based a lot of their research on the links with similar gene therapy studies on Haemophilia b. In 2002 research was able to treat Haemophilia b in dogs. In 2011 they had permission to treat in humans. All the results were successful and there are plans to continue research in Haemophilia a (the more common variation of haemophilia).

Type GSDIII

- They started with a mouse model where they removed the gene from the type III mice which resulted in a raised accumulation of glycogen in the muscle and liver. They also had twice the amount of Hepatomegaly (liver enlargement).
- When trying to insert the specific gene created, they found that it was too big to fit in one vector so they split it and sent in on 2 separate vectors. Mice treated with the therapy returned to expected results for normal wild mice with regards to hypoglycaemia.

- In the future they want to experiment with the effectiveness of this split gene method to improve efficiency.
- They do not have permission to work on humans yet but plan to submit projects that could have results in the next 5 years.

Dr Steuerwald provided a very informative session that explained how a GSD person uses energy compared to normal metabolism

Hepatic GSD and Sport

Ulrike Steuerwald, Hanover, Germany

Summary

Dr Steuerwald provided a very informative session that explained how a GSD person uses energy compared to normal metabolism. She also made some recommendations for GSD patients (hepatic types in particular) with regards to nutrition and safe sport or exercise activities. She is based in Hanover but does a lot of work in Torshavn, Faroe Islands, as they have a large GSD population.

Points that may be of interest

- The Faroe Islands have a population of 50,000 and 16 cases of type 3. This is way above other averages in other countries, partly due to the limited gene pool in the community.
- Dr Steuerwald outlined the importance of understanding how muscles get energy and where we get it from (protein, fat and carbs).

  Sugars = fast fuel and nutrition for the brain.

  Fat = fuel for muscles and transport for vitamins

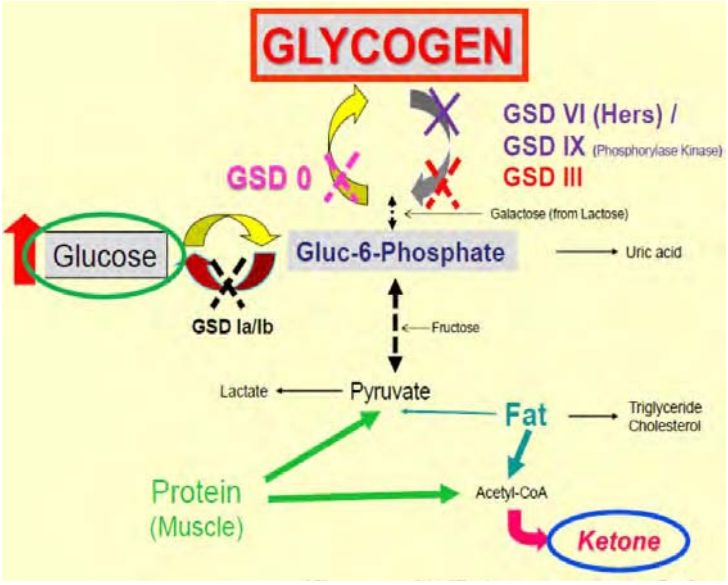
  Protein = growth , muscles, second-line energy source

- Brain – more than 60% of sugar in your blood goes to brain function
- The second line of fuel for the brain is fat. This is broken down when there is an absence of glucose in the blood and glycogen is unavailable. However, this causes ketones to be produced, especially in type 3,6,9,0. The main issue in type 1 is the levels of lactate, not ketones. But ketones are not ideal and they can give you a headache or cause vomiting when in high levels.
- Muscles can use sugar, fat, ketones, lactate for energy
- ATP (Adenine Tri-phosphate)– is the best energy for immediate use eg 2 secs sprint or weight lifting. ATP is stored in each cell but runs out very quickly. Next the body uses whatever glucose is in the blood. Next it will use the glycogen stored in muscles or liver.

- Fat is only used during low level exercise over longer periods of time.

Benefits of exercise

- Exercise is great for your brain. It can reduce depression, improve memory, concentration, lessen ADHD and quicker learning.
- It has been proven to be one of the best ways to prevent the onset of Alzheimer's disease!
- Exercise releases chemicals in the brain, serotonin, norepinephrine, endorphins, dopamine – these cause a 'feel good' sensation.
- Exercise strengthens the heart and lowers blood pressure.
- Exercise makes muscles grow,
- Helps creation of new blood vessels
- Improves bone density.
- Decrease risk of breast and colon cancer.
- The more you exercise the more efficient your body will become at burning fat. You will be less dependent on carbs and glycogen and lose weight more easily.
- Exercise can improve lipid profile. A lipid profile is a blood test that measures the amount of cholesterol and fats called triglycerides in the blood.
- Exercise increases insulin sensitivity and regulates appetite.
- Dr Steuerwald explained that she has patients that exercise well (walk at least 30mins a day) and are not dependent on CS.
- Recommendation - 3x per week 10 mins aerobic activity at medium to sub maximum effort level.
- Dr Steuerwald explained the chemical pathways involved in the different types of Hepatic GSDs. All kinds of carbo-hydrates are broken down into glucose.



- Type 3,6,9 cannot break the glycogen back into Glucose, so they have to go to pyruvate for an energy source which can be produced by breaking down protein or fat. When you break down fat you produce ketones as a bi-product.
- Problem sports are those that require a quick release of energy.
- Preferred activities are endurance sports as they promote a longer, gradual release.
- How do you know when you have done too much? – You see an elevated Creatinine kinase level or better to test for myoglobin. This can be tested at home with testing strips. Myoglobin is muscle protein and is passed out in urine.

We watched a video about a young man called Jay Gordon who successfully manages his type 1a and carefully exercises too. The 'Fight for a Cure' YouTube video can be found on this link:

https://youtu.be/MExb7gA3Zeo

GSD III tolerance study

Dr Phillip Hennis, Nottingham Trent University

Summary

Dr Hennis outlined the research that the team at Nottingham have been completed looking at GSD 3 patients and their performance in exercise trials. The projects looked at the apparent strength and maximum output compared to other measurements and variables.

Points that might be of interest

- Patients were given 2 main tests: a stationary cycling test and later leg extension tests to measure a physical functioning measurement.
- The GSD 3 patients maximum work rate was not limited by their heart rate as they stopped before their heart rate maximum was reached (Only 77% of the heart rate maximum) but researchers believe this was partly due to relative strength of their muscles and the muscle quality they have compared to comparable individuals without GSD 3.
- They also found that GSD 3 people are unusually sedentary with subjects reporting to be seated for longer than 10 hours a day. They did not do as much low level activity during the day for example general standing or 'pottering around'.
- The sample group were split into 2 groups: high and low physical functioning groups. They found that the higher physical functioning subjects (2 subjects with an average age of 27) had far better results that measured close to 'normal' people. However the lower physical functioning group (3 subjects average age 46) did not fare so well. What Dr Hennis could not make clear was why the lower physical functioning group performed worse.
- It was stressed that this study was only descriptive of 5 subjects in the UK who had not been treated with a restricted diet or high protein/low carb diet which is commonly accepted in other countries around the world.
- Similar studies have been done in the US and Netherlands but have not been published and were not discussed in detail at the conference.

CARERS DROP-IN SESSION

Summary of Carers' Drop-in Session

Jane Lewthwaite, AGSD-UK Specialist Care Advisor

Feelings about being a carer

- Patronising information and suggestions
- Crisis
- Feeling out of control
- Cannot fix it
- Grieving for a life we thought we would have
- Overlooked
- In the background
- Loving
- Caring
- Lack of time
- Caring for someone who doesn't want it
- Powerlessness
- Guilt for not doing more
- Guilt for doing something for myself

Suggestions from my experiences

- Counselling can help
- Don't plan too far ahead
- Make gradual changes
- Ensure GP knows you are a carer
- Learn about the condition
- Acceptance
- Stay well informed
- Meeting others
- Know the person is still a person
- Join a carers group
- Get a Lasting Power of Attorney, especially if you are not married

ASGD-UK now has a private Carers Facebook group. Contact Jane Lewthwaite (Facebook: "Jane Agsduk").

Websites*

- carersuk.org
- gov.uk/carers-uk
- carers.org
- ageuk.org.uk › Home & care
- www.carerssupport.org.uk
- musculardystrophyuk.org/get-the-right-care-and-support/entitlements/carers-support/
- carersclub.org

Local

- Search for Carers Support Meetings
- Social Services Help Desk – phone to request someone to review your needs [a carers assessment] and ask then to tell you what is available
- Register as a Carer with your GP

Benefits Advice

- Phone your Citizens Advice Bureau, local Carer's Organisation or local Disability Organisation and ask for an appointment for a full benefits check.
- Contact AGSD UK for supporting letters and advice on completing the forms.

Emotional Support

All counties have an NHS counselling service, available by referring yourself via a phone call or through your GP. They are usually called Positive Steps, First Steps or similar. Some offer groups for Carers, for Anxiety, for Managing Mood. Also offer 1-1, email and other. Some Carers organisations offer phone support.

Guides

Until we have a guide, use one for another condition, much information is similar.

Legal

- Lasting Power of Attorney
- Making a Will and Advance Directive

[All of these are relevant to everyone, not just carers].

Emergency Planning

Gather a folder; phone numbers, helpers, other carers, Whatsapp group of supporters.

Get a Community Care Alarm, Risk Alert Card, talk to others. By doing this, you could reduce your worries about what will happen in a crisis.

*** If you do not use a computer please phone Jane on 07484 055334 she can print and post information to you.**

There are only four kinds of people in the world.

Those who have been caregivers.
Those who are currently caregivers.
Those who will be caregivers,
and those who will need a caregiver.

Rosalyn Carter

AGSD-UK Conference 2017 – Lunchtime

NUTRITION ADVICE FOR POMPE DISEASE

Nutrition advice for Pompe Disease

Diane Green, Dietitian, Salford Royal Hospital

Diane Green led two workshops whilst others attended a physiotherapy or carer's workshop. Below are points captured by workshop participants.

Luke Fraser's notes:

- 70% of protein consumed should be from High Biological Sources [meat, dairy]
- The other 30% taken should be from carbohydrate-based foods such as fruit and vegetables.
- 30-35% of calories should be from protein [current recommendation].
- Protein boosted foods such as high protein yoghurt [increase the protein without increasing the fat].
- Potential benefits to consuming protein in the evening.
- Patient concern over protein-enriched foods. Try skimmed milk powder [high protein and low in fat]. Nuts [these are higher in fat but it is a healthier fat].
- There is no evidence that higher fat content is beneficial to Pompe patients.
- Reduce carbohydrates, but consistent intake still important for brain and body function.
- Bone strength: Lack of weight bearing exercise because of reduced mobility can affect bone strength. So, maintain calcium and vitamin D levels.
- Avoid the ketogenic diet – still need carbs. Liver can still convert glycogen to glucose.
- Canadian study – suggest 20% of calories from carbohydrates spread throughout the day.
- Low GI foods (avoid simple sugars) – slow burn calories [e.g. nuts and wholegrains].
- Need to consider general health effects of diet - apart from Pompe [eg avoiding high salt, saturated fats, raising LDL cholesterol].
- Use a slow-cooker to avoid having to cook in the evening
- Avoid sugary drinks in the evening.
- Tea/Coffee – up to 5 cups per day, but try skimmed milk

Barbara Muir's additional notes:

- Double check your kidney function with your medical team
- There is 6g of protein in 1 oz. meat
- Maintain protein but move towards lower fat products. E.g. a normal yogurt has 6gms of protein but others have up to 20gms.
- Take advice from your metabolic dietitian
- Skimmed milk powder is low in carbs and fat, but high in protein
- Nuts have 26g of protein per 100g. Useful to have as snacks.
- Small blocks of cheese for snacks, avoid carbs.

PHYSIOTHERAPY WORKSHOP

Note regarding fats

- Patients in ITU coming off a ventilator often require a higher percentage of fats.
- No evidence that healthy fats have a detrimental effect.

On Avoiding Carbohydrates

- People with Pompe need less carbs; not no carbs.
- Have smaller amounts of carbs throughout the day
- Be aware of the risks as you age e.g. diabetes or heart disease.
- Avoid processed foods and meat, recommends fresh unprocessed foods.
- Consume red meat in moderation.

Bryn Edwards and Jonny Smith, Neuromuscular Centre, Winsford, Cheshire.

Contact details

bryn.edwards@nmcentre.com

jonny.smith@nmcentre.com

www.nmcentre.com

General enquiries: 01606 860 911

Notes by Jane Lewthwaite, AGSD-UK Specialist Care Advisor

People with Pompe experience weakness in the core and respiratory muscles as well as the large muscles such as hamstrings and gluteus.

Remember that people with Pompe are still at risk of the health issues faced by everyone through lack of exercise and poor diet, such as Type II diabetes and heart disease.

While you have the muscle function, try to use it. You cannot change the diagnosis of Pompe disease, but you can take control of exercise.

Muscles have access to glucose and they will use it. Through regular exercise you can improve fitness. Improvements can be made. Including improvements to gas exchange in the lungs.

- Anti-gravity exercises are best.
- Extension exercises are good.
- Do something every day if possible.

Fitness Applications for phones, tablets and computers can help to monitor and motivate both diet and exercise. For example, MyFitnessPal.

Exercise Advice

Regular aerobic exercise

- NHS recommends 150 minutes per week.
- Start low and avoid over working. E.g. start for 5 minutes or less.
- Just get your heart rate and respiratory rate up a little bit until you can do 20 minutes per day.
- On a good day, you might risk overdoing it
- At the Neuromuscular Centre (NMC) they rarely see people with injuries but they do see people feeling sore and fatigued.

Safety

- Avoid perilous surfaces and machines such as treadmills – these can be dangerous.
- Use a static cycle or small pedal system.

Aerobic vs Resistance

- Try 3 sets of 10 repetitions of a simple exercise.
- If you are struggling to do that then cut down and start lower.
- If you cope well, then move up to 4 sets and then 5 and then 6.
- Check muscle pain post-exercise and if you feel it then you did too much.
- Fatigue after exercising also shows that might have done too much.
- Ensure you rest well afterwards.

Core Strength

- Try a wobble cushion, cheap and easy to use, stay up straight on it and practice several times per day to help balance. By sitting on an unstable platform.
- Hydrotherapy is recommended for those who enjoy water and do not experience breathing problems in water. The buoyancy offsets the effect of body weight. However, this can lead people to doing too much in water, because they feel light.
- Pilates style exercises have worked well for people.
- Anyone can attend the NMC at Winsford for a few sessions to practice. Get a referral from your GP and if they won't help, then phone Winsford directly and ask for the help in getting a referral (your specialist centre might be willing).

Bryn and Jonny will also offer to speak to your local physiotherapist about special needs for Pompe disease.

"OUR GOOD STUFF"

Amicus Therapeutics provided "Our Good Stuff" kits to encourage people to focus on the positive events in their lives. Below are a few examples of the notes people left in the jar during the conference.

Enjoyed being with people the same as me without feeling judged by my disabilities

Wonderful to meet old and new friends in Nottingham

Super place and super people, thank you

Spending time with other people with Pompe and their families

It was good to talk with Jane she is a good listener

I experienced the love from the Pompe friends. Feeling normal with them (sharing and caring)

So happy to be where Robin Hood lived

New Trustees on board

Loved the hotel and facilities at this conference

Meeting enthusiastic people and sharing ideas

Meeting other Pompe friends. Gaining helpful knowledge

AGSD community is wonderfully mutually supportive... a real boost in the face of adversity

Listening to Joan's inspirational talk and being inspired to do something for my 70th next year

Great to share

WORLD Symposium 2018, San Diego

Visit Report, Allan Muir

Overview

The WORD Symposium is held every year in either San Diego, California or Orlando Florida. It attracts all those with an interest in Lysosomal Storage Disease research, including medical professionals, pharmaceutical and biotechnology companies and patient groups. Many global organisations use the occasion to coordinate their own meetings and so it is an excellent event for networking with the international LSD community. Attendance was reported to have risen to over 1700 delegates this year.

Pompe disease is the only GSD that is also an LSD, however many of the conference delegates work in rare diseases or in metabolic diseases; so, although other GSDs get little attention in the presentations, they are well understood by the target audience.

The conference programme started on the Monday with the Council of Patient Advocates, a meeting where patient groups discuss LSD research with the aim of involving patients in the application of research grants, particularly from the National Institutes of Health (NIH).

The following three days provided research presentations including natural history studies, novel therapies, new-born screening and sponsored satellite symposia. Several hundred posters were also displayed including one that I was a co-author, showcasing the work of the UK LSD Collaborative.

Whilst at WORLD I was also able to meet with medical teams from both the UK and abroad, and managed to discuss several important issues:

- Speakers and topics for the AGSD-UK Annual conference.
- A proposal to price current and future ERT per patient rather than by drug volume.
- A proposed meeting to explore the creation of management guidelines for hepatic GSD.
- The development of the GSD laboratory service at GOS. We are invited to arrange a visit in the near future.
- I also managed to recruit a UK metabolic consultant and his wife onto our cycling team for the Prudential RideLondon–Surry 100 event in July!

Council of Patient Advocates

This year's COPA meeting was very well attended with over 100 people in the room representing a mix of patient support groups and patient advocacy and other staff from various companies.

The meeting this year was designed to allocate themes to each table of around 10 people who were tasked to a) define an unmet need, b) hypothesise a solution and then c) create a short pitch for research funding. The themes were:

- Natural History
- Quality of Life
- Pain
- New-born Screening
- Towards Treatment
- Joints and Musculoskeletal
- Cognitive Endpoints
- Role of Genetics
- Societal and Emotional Issues
- Clinical Management Guidelines

As you can imagine, there was a great deal of information and discussion within the allocated three hours; what was most interesting to me was how just about every table highlighted the need for natural history studies and registries for each rare disease. This is certainly something we should be considering for all GSDs, both to assist research funding applications and to assist future therapies seeking market approval from regulators.

Presentations

There were a number of presentations dedicated to Pompe disease, although many more had some relevance to it and other GSDs. For example, a presentation by Arcturus Therapeutics made mention of their messenger RNA program (LUNAR-RARE1) that is developing a therapy for GSD 3, if successful, that will be commercialised by Ultragenyx.

NIH introduced their new projects devoted to rare diseases under NCATS (National Center for Advancing Transtlational Research). GARD is the Genetic and Rare Disease Information Centre that is similar to the EU Orphanet. Also RaDaR (Rare Diseases Registry Program) which aims to define best practices for patient registries. NIH are developing a toolkit for patient-focused Therapy Development. All very useful resources and illustrating NCATS goal to engage patients as essential partners in R&D.

Satellite Symposia

Responding to the Challenge of Pompe Disease

Supported by an independent educational grant from Sanofi Genzyme. The main speaker in this session was Priya Kishnani who spoke of emerging new phenotypes of Pompe disease as a result of new-born screening in the USA. For example, the atypical Infantile-Onset children who are diagnosed under the age of one year and have no cardiomyopathy.

Novel Treatment Approaches and Study Designs for Rare Diseases

Supported by Ultragenyx. Clinical trial designs were discussed for ultra-rare diseases with very small heterogeneous populations. Individual Clinical Responses (ICRs) were gathered and combined with Patient Reported Outcomes (PROs).

Creating Hope for the Unreachable, Unprofitable and Unthinkable Patient

Sponsored by Care Beyond Diagnosis Foundation, in conjunction with the MPS Society UK and the European Gaucher Alliance. Supported via unrestricted educational grants from Chiesi Farmaceutici, Shire and FYMCA Medical Ltd. Care Beyond Diagnosis is a new organisation formed by Chris Hendriksz and his family. Chris was, until recently, the clinical lead at the adult LSD specialist centre in Salford. Before that he worked at a paediatric centre in Birmingham. They work to improve the care of people with rare diseases in areas of the globe where high standards of medical care do not exist.

Early treatment of lysosomal disorders: a closer look at Fabry and Pompe disease

Supported by Amicus Therapeutics, Inc. Mark Patterson gave an interesting talk about the need for early diagnosis across all diseases, and Priya Kishnani gave examples from the New-born Screening programme in the USA.

Sanofi Genzyme's 6th Annual Forum for patient advocates

This was a small meeting designed to introduce the global patient advocacy team and the support they can provide, and to inform Patient groups of grants available and the process for making grant applications.

An extremely successful art exhibition, "Beyond the Diagnosis" by Patricia Weltin was highlighted as a successful way of raising awareness of rare diseases.

Industry Meetings

Before arriving in San Diego, I had pre-arranged meetings with more than ten organisations, including:

Developing Next-Generation Enzyme Replacement Therapy

- Amicus Therapeutics
- Sanofi-Genzyme
- Greenovation
- Valerion – ERT for Pompe and interests in late-onset GSD 4, 7, 15 and Lafora disease (Polyglucosan Body Diseases)
- Regeneron

Developing Gene Therapies

- Audentes Therapeutics
- AvroBio
- Duke University with Actus Therapeutics
- University of Florida, Dr. Barry Byrne
- Spark Therapeutics
- Ultragenyx Therapeutics – GSD 1a and interest in GSD III

It was fascinating to hear so much interest in our group of ultra-rare conditions and I look forward to hearing more from these organisations as their research and development progresses.

FUNDRAISING

Below is a small selection of past and present activities and I apologise in advance if I haven't given yours a mention.

Pompathlon

Member Joan Wright (Pompe) talked at the Conference last year; she undertook a 60 lengths swim, 60km bike ride and 6-mile walk over 3 days to raise awareness of Pompe. Her daughter, Lucy, is planning to cycle 1200 miles on mountain bikes from John O'Groats to Land's End next July/August with two friends; Lucy is raising money for AGSD-UK.

Swimmers to take on English Channel to raise money for charity.

https://uk.virginmoneygiving.com/Team/FaffingFrosties

In the first week of June, four intrepid swimmers will be aiming to swim in relay between Dover and Calais.

Each team member will swim for an hour in succession until they reach the shores of France. This will be a tough swim, because at the start of the season the temperature of the English Channel will only be between 12° and 14° and it could take 18+ hours to complete, but hopefully less.

The team members are: Melissa Compton, Keith Clarke, Emma Heggenbath, Janet Mills and in reserve Karin Daelemans.

The team have been training throughout the winter to make sure they are ready to take on this demanding challenge. The swimmers are raising money for two very worthwhile causes and would be most grateful if you could sponsor the team and help them on their way. The two charities are AGSD-UK and the Shrewsbury and Telford Hospital Charity.

Quotes from online fundraising pages:

Michele Watson

M&Ms Annual Fundraising Barbie 2018

https://uk.virginmoneygiving.com/MandMsAnnualFundraisingBBQ2018

"Mark's journey towards a confirmed diagnosis for a rare muscle disease continues. We started the journey in late 2015 and although we're not reached the final destination, we do know that this charity provides wonderful support for people just like Mark and myself who are unfortunate enough to have one of the rare 'AGSD' diseases."

Ceri Reed

Fundraising for AGSD UK

https://uk.virginmoneygiving.com/CeriReed

"George was diagnosed with GSD type IX in 2013 at age 10. The late diagnosis affected him neurologically. Today he often misses school as a result of GSD and its complications but is improving! He manages his diet daily and is working hard with his school work with grades being a credit to his determination. He is a beautiful person inside and out and we couldn't be more proud of him. George wants to raise awareness of GSD so that other children have an earlier diagnosis."

Alexandra Carey

Brighton Marathon

https://uk.virginmoneygiving.com/AlexandraCarey

"AGSD-UK does a lot of work to help those affected by this disease from contributing to the achievements of treatments and cures to protecting the interests of those who suffer from it. So, whilst I'm training, working up a sweat and using my glycogen levels, I will be raising money to help those who are facing their own marathons on a daily basis!"

John Critchfield

"I am doing a 17.5 mile kayak race to raise funds for AGSD-UK because I know how bad this disease is and I want to help."

"Hurray. We finished. We came about mid field which is great in just over three hours and fifteen minutes. Thank you again for your sponsorship as I heard I met my target and the money is going to a great cause. Thank you, thank you, thank you."

Steve's The Murray family's 'give something back' event

"Our eldest son, James and youngest daughter Elsie-Mai have both been diagnosed with GSDV in recent months, we are currently awaiting genetics results for younger son Alfie and tests to be carried out on eldest daughter Charlotte."

"Without Andrew of AGSD, we would have been lost - it was Andrew who pointed us in the direction of the clinic at GOSH and whom has been a constant support with advice and assistance."

Gemma Seyfang's POMPE Christmas Jingle

"We face many challenges, but one of the bittersweet things to have come out of getting diagnosed with Pompe Disease is that we have a FANTASTIC support network in each other. We communicate with others across the world with Pompe Disease. AGSD-UK helps to support us in many ways and we would LOVE it if you can help to support this small charity."

Vik's Guinness World Record Attempt: Most Punches in 60 secs with a single arm

Vik Hothi

"In late November 2017, I was stuck down with a condition that almost claimed my life and threatened to take away my life's passion, martial arts and movement."

"To give myself a goal and to reframe this situation, turning something that was difficult for me to something that would help others, I have taken on the challenge of breaking this record in an attempt to raise money and awareness of Mcardle Disease and inspire people to try and find a way around bad physical, mental and spiritual situations."

PROJECT JOGLE 2018

Lucy Wright

Just an ordinary girl attempting an extraordinary challenge.

With only 13 weeks to go until I embark of the biggest challenge of my life, I am now starting to believe I might actually have a fighting chance of completing it!

On the 23rd July, myself and 2 friends will be cycling 1200 Miles in 13 days from John O'Groats to Lands End. We will be riding on mountain bikes using the UK's National cycle trails and will be staying in hotels and hostels along the way. We will also be supported by our very own support vehicle driven by a fantastic lady who has offered her spare time to help us out.

The main aim is to raise money for two fantastic charities, which are very close to our hearts. I will be raising money for the Association of Glycogen Storage Disease as my Mum, Joan, suffers from Pompe. Those who know Joan will also know how passionate she is about promoting the benefits of exercise and keeping active despite her condition. I hope to do her proud!

The training has been relentless over the winter months but I've stuck with it and I'm finally starting to feel the benefits! The support we've been given has been overwhelming and, ultimately, I know this is what will get us over the finish line!

If you would like to follow our journey, please like our Facebook page 'Project JOGLE 2018'. Any sponsorship is much appreciated and can be made via my justgiving page: www.justgiving.com/lucyjoanne3rd

However, my biggest wish would be to see as many AGSD sufferers and their families as possible on route to cheer us on, give us a much needed boost of morale and remind me while I am doing it!

Below is the route planned for Project JOGLE. If they pass close by your home town, you may like to line the route or greet the girls by waiving them off after breakfast, meet up for lunch or welcome them at the end of the day. Please contact the Droxford office if you'd like to send a message to Lucy.

Day	Miles	Date	Start	Passing through	Finish
1	89	Sat 23 June	JOHN O'GROATS	Thurso, Tongue	Crask Inn
2	65	Sun 24 June	Crask Inn	Ardgay	Inverness
3	100	Mon 25 June	Inverness	Carrbridge, Newtonmore	Pitlochry
4	97	Tues 26 June	Pitlochry	Kenmoreq, Callander	Balloch
5	97	Wed 27 June	Balloch	Glasgow, Abington	Lockerbie
6	80	Thurs 28 June	Lockerbie	Gretna Green, Penrith	Appleby
7	92	Fri 29 June	Appleby	Orton, Borwick	Whalley
8	81	Sat 30 June	Whalley	Haslingden, Manchester	Nantwich
9	97	Sun 1 July	Nantwich	Market Drayton, Bridgenorth	Worcester
10	108	Mon 2 July	Worcester	Tewkesbury, Thornbury	Wells
11	95	Tues 3 July	Wells	Glastonbury, Wellington	Bratton Fleming
12	96	Wed 4 July	Bratton Fleming	Bideford, Bude	Bodmin
13	91	Thurs 5 July	Bodmin	Padstow, Truro	LANDS END

BIG FUN RUN

Big Fun Run is a series of 5k untimed runs staged within scenic settings throughout the UK from July to October. There are 18 locations to choose from, all you need to do is find the one closest to you! The Big Fun Run series is strictly for FUN where it's all about relaxed exercise. No times, no pressure, no sweat – just some easy moves and lots of laughs. The runs are suitable for all the family where you can accomplish your personal goals; whether you are raising money for a worthwhile cause close to your heart, remembering a loved one or simply keeping active. This isn't about Olympic level athletes charging about in record times, it's about mums with prams, dads with toddlers, groups running together, fancy dress and a fantastic fun mix. Are you getting a picture of something a little bit different? There really is something for everyone at a Big Fun Run! Plus, under 5s can take part for FREE!

Entries now open for more 2018 Big Fun Runs!

The next 'wave' of 2018 Big Fun Run Event locations! In past years, they have launched all the Event Locations at the same time, however, for 2018 they are launching locations on an individual basis to give us more time to recruit runners.

For example, charity entries for the following 2018 Big Fun Run locations are now open:

- Big Fun Run Gateshead
  Saturday 20th October 2018
- Big Fun Run Middlesbrough
  Sunday 21st October 2018

Look out for other venues at www.bigfunrun.com/venues/

You will see that AGSD-UK is an affiliated charity for these events, but pleasecontact the AGSD-UK office if you would like a place in one of these runs.

Entries now open for more 2018 Dog Jogs!

The organisers have also opened the next 'wave' of 2018 Dog Jog Event locations!

Charity entries for the following 2018 Dog Jog Locations are now open:

- Dog Jog Gateshead
  Saturday 20th October 2018
- Dog Jog Middlesbrough
  Sunday 21st October 2018

Look out for other venues at www.dogjog.co.uk

You will see that AGSD-UK is an affiliated charity for these events, but please contact the AGSD-UK office if you would like a place in one of these runs.

RIDE LONDON-SURREY 100

Ride London-Surrey 100

AGSD-UK has sixteen places reserved for the 2018 event,
we have filled eleven and so we will have our greatest peleton yet;
look out for our colourful jersies during the TV coverage.
The remaining places will carry over to the 2019 event if not filled.

Our team of riders this year includes:

Suresh Vijay	http://uk.virginmoneygiving.com/SureshVijay
Kate Penny Thomas	http://uk.virginmoneygiving.com/kate-penny-thomas-mrx-PRLS-2018-1470
Sam Arthur	http://uk.virginmoneygiving.com/SamArthur
Vicki Lucass	http://uk.virginmoneygiving.com/vicki-lucass-ytr-PRLS-2018-11295
Allan Muir	https://uk.virginmoneygiving.com/AllanMuir
Laurence Plant	
Martin Lucass	
Robert McKenna	
Tommy Munday	
John Curnow	
Michael Galllucci	

2017 participants Emily Thompson and Chris Davies

A DAY AT THE RACES

Members Alan and Coren Moore, whose son Mylo has GSD 3, managed to persuade the Camping and Caravan Club to select AGSD-UK to be one of two charities to benefit from the August Bank Holiday “National Feast of Lanterns” (NFOL).

Allan and Barbara Muir together with Sylvia and Ed Wilson (GSD 3 granddaughter) manned a charity gazebo to raise additional donations from two activities:

- Smoothie Bike
- Glycogen Challenge – Burn 10grams of Glycogen by cycling one mile on an exercise bike – fasted time won a small prize.

Allan Muir was invited to address the assembled campers at the event’s opening ceremony.

Allan was photographed with Mylo and his family receiving a cheque to the value of around £691 from collections over the weekend. In total, the weekend raised £1293 for AGSD-UK.

From Left: Coren, Enya and Mylo Moore, Allan Muir, NFOL Chairman Allen Russell and Alan Moore.

GIVE AS YOU LIVE

Collecting donations for AGSD-UK while shopping online

Give as You Live

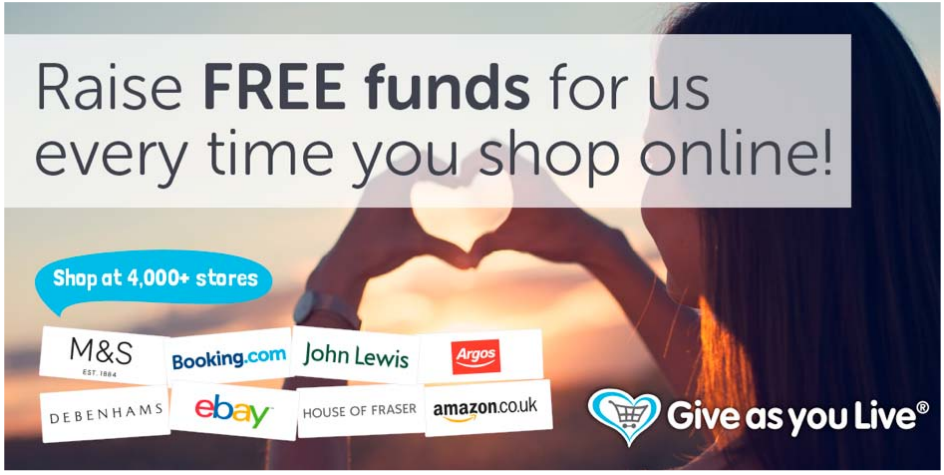
Did you know you can raise free funds for us every time you shop online? Simply shop via “Give as you Live” and each purchase you make will raise money for AGSD-UK!

www.giveasyoulive.com/join/agsd-uk

Already signed up?

Check that the Give as you Live extension is enabled in your browser; its bar should appear at the top of shopping pages. If its missing, you may need to reload it by visiting:

www.giveasyoulive.com



AMAZON SMILE

AmazonSmile is an alternative way for you to collect donations for AGSD-UK every time you shop, at no cost to you.

To shop at AmazonSmile simply go to smile.amazon.co.uk from the web browser on your computer or mobile device.

You use the same account on Amazon.co.uk and AmazonSmile. Your shopping cart, Wish List, wedding or baby registry, and other account settings are also the same. On your first visit to AmazonSmile, you need to select AGSD-UK to receive donations from eligible purchases before you begin shopping. Amazon will remember your selection, and then every eligible purchase you make will result in a donation.

LEGACIES

Leaving a legacy

A legacy is a wonderful way to ensure that we can continue our vital work. It can also be a valuable way of reducing inheritance-tax liability on your estate, because a legacy to a registered charity such as AGSD-UK is tax-free.

Whatever their size, bequests can make a big difference. Thanks to the kind support of our legacy givers, we can plan our future work and provide support to persons affected by Glycogen Storage Disease and their families in the UK.

For more information please contact our Droxford office.

CORI ACTION TEAM SUPPORT – CATS

My name is Lauren and I've been living with GSD for 17 years; I was born on February 29th 2000, which coincidentally is also Rare Diseases Day! I was diagnosed at 11 months at Birmingham Children's Hospital. There have been a fair share of ups and downs but through the help of my parents, the medical team at Birmingham Children's Hospital, and now at the University Hospital Birmingham, I have managed to live a life I am proud of.

Since becoming an adult I've wanted to help more within AGSD-UK, I now have a role within the charity as part of the recently formed Cori Action Team Support (CATS) along with Ailsa Arthur, Nikki Christie, Sylvia Wilson, and my Dad, Gary Thompson.

We are producing a CATS leaflet which will be sent to everyone with GSD 3. We aim to provide some support and to co-ordinate information for people affected by Cori disease, including parents and carers.

Nikki has been working on "101 Top Tips for GSD 3" and it will hopefully be published soon. Ailsa, helped by Jane Lewthwaite, has been trying to check all the contacts we have with members with GSD 3. She has been emailing and trying to refresh everyone's contact details. Please help by replying or just go to AGSD-UK website and renew your registration. Registration is free, but of course, if you could pay membership of £15 per year - it helps enormously.

Sylvia Wilson has taken over as Type Co-ordinator for GSD 3 from Esther Prosser, but Esther will still be involved because we need her knowledge and expertise.

As part of my eighteenth birthday celebrations we spent Christmas as Walt Disney World as you can see from the photo below with Mum and Dad. So, I hope people can see that I live a full life and stay positive with Cori disease.

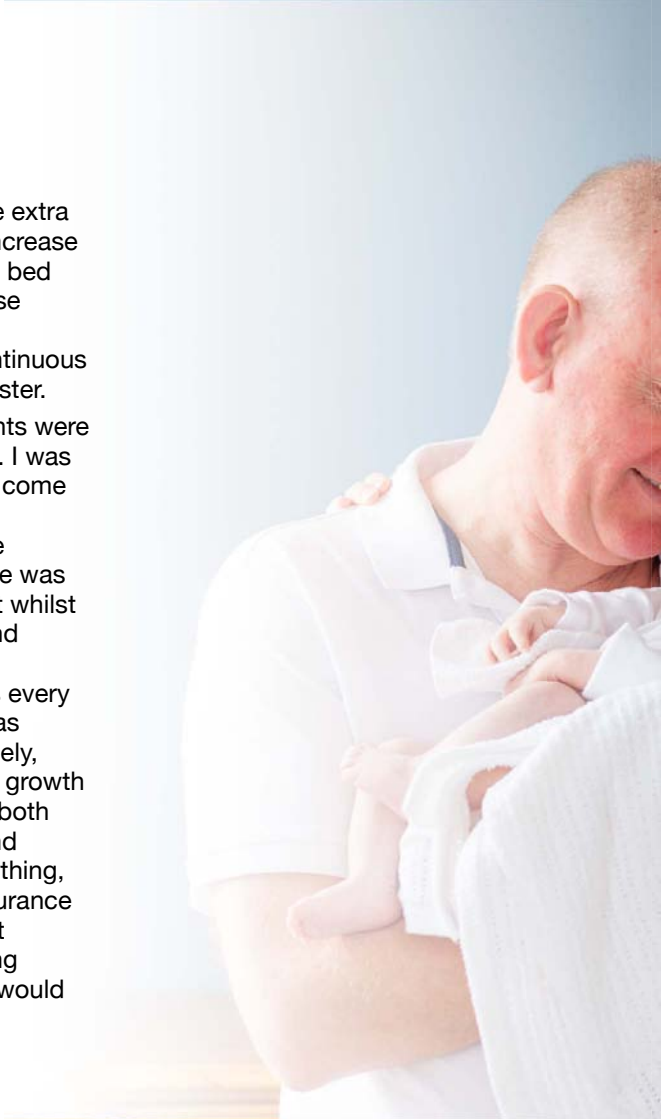
Lauren Thompson

Gary, Andrea, Chewbacca and Lauren Thompson

GLISTEN WITH MOTHER

One of the things that has always been a small question mark in my mind was how would GSD VI affect my chances of falling pregnant and having a healthy baby. After deciding alongside my husband that we wanted to try for a baby I shared my concerns with my doctors at UCLH. I equally asked on the GSD Facebook forums but there seemed to be no one with Type VI who had given birth whom I could contact. UCLH advised that GSD should have no impact on falling pregnant and hopefully little impact on the baby however there were some ways we could best prepare and manage during pregnancy. I underwent continuous glucose monitoring to get a stronger sense of how I was managing my sugar levels throughout a 5-day period. This came back positive with any worrying lows only after periods of exercise alongside fewer carbs. With this knowledge behind us we were confident that the risks were low. I conceived in October 2017. To give the baby and I the best chance of progressing healthily alongside extra check-ups UCHL advised to increase the amount of cornflour before bed and start monitoring my glucose levels and ketones daily, I also undertook a second 5-day continuous monitoring in my second trimester.

My antenatal appointments were managed at Kingston Hospital. I was the first case of GSD they had come across and so intriguing to the midwives and consultants! The consultant I was assigned there was fantastic and happy to support whilst letting the UCLH specialists and myself take the lead. Kingston Hospital proposed extra scans every 4 weeks to ensure the baby was growing as expected. Fortunately, both the glucose, ketones and growth scans showed every sign that both the baby and I were healthy and developing as expected. If anything, it was lovely to have the reassurance through all the extra scans that things were on track, something that a healthy low risk mother would not have.



THE STORY OF BABY EDWARD!

The next hurdle was to be labour. Labour is notorious for being a long draining event - hence the name! Equally it often causes nausea either causing vomiting or preventing you from eating. The concern was that I would not have enough energy to continue through a lengthy labour. The plan was that I should monitor my blood glucose levels once an hour during labour and should my glucose drop below an agreed level I would be given an IV drip. This was proposed by UCLH and supported by Kingston Hospital.

At 40 weeks they suspected I had Pre-Eclampsia (PE), this was not thought to be related to my GSD in any way. The advice for a baby at term where PE is suspected is to be induced immediately as if this should develop to Eclampsia it can be life threatening for mother and baby. Often induction comes with a risk of a longer labour, as such I was concerned but the risk of not taking this route were too high for the baby and myself. Indeed, my labour was on the longer side at 60 hours, but I could eat and drink until quite far in. We had in our hospital bag an array of snacks and sugary drinks. With this my blood sugar levels never dropped below the agreed level and an IV drip was not required.

On July 15th at 8:31 we welcomed Baby Edward into the world. He is a thriving baby and shows no signs of being impacted by GSD. He is now 8 months old and having learnt to crawl and climb is getting into all sorts of mischief! I owe a huge degree of thanks to the partnership UCLH and Kingston Hospital worked under. Equally for the additional monitoring, advice and support which I believe helped lead to a very healthy happy baby boy.

Caroline Calder, GSD VI Coordinator

UK MCARDLE'S NEWS

Newly diagnosed

Two more UK children have been diagnosed in the first quarter of 2018. This is so unusual for McArdle's. It represents a reduction of about 20 years in the typical age of diagnosis. Of course, they grow up and get to be over 18, so numbers go down as well as up. We currently have 11 children under 18, with two more currently undergoing tests and looking likely to be confirmed.

We currently have 309 people diagnosed with McArdle's in the UK, and approximately 210 of those attend the McArdle Clinic in London. The diagnosed incidence is still only 1 in about 250,000 people in the UK. However, the best estimates we have suggest that there are probably at least 1 in 100,000, so we have some way to go to diagnose all who could be helped. In addition, a recent paper on genetics suggested that they incidence may be a great deal higher.

New arrangements at McArdle Clinic

Last October Andrew Wakelin stopped attending every McArdle Clinic, where he has provided support to patients for about 14 years. Discussions have been on-going about a new arrangement for support to patients at the clinic. The clinic's program has been adjusted to create a one-hour slot at lunchtime for a patient support group session. This limited commitment should make it possible for other volunteers to help with those sessions. There are logistical and contractual issues to overcome before we can recruit volunteers (probably from London and the South East) to each cover about 3 or 4 clinics per year. The new arrangement was meant to start in April, but we are not quite there yet. Hopefully it won't be long.

UK map of McArdle's people

National Hospital for Neurology and Neurosurgery

IAMGSD NEWS

New web site launched

The international body for Muscle GSDs, IamGSD, launched its new web site in early March. This now takes over much of the information developed over the last approx. 15 years by AGSD-UK. This will help to bring it to a larger audience and it is hoped to eventually run the site in five languages. It is bringing together many aspects that have previously been somewhat separate or isolated, so it will become a powerhouse of information on muscle GSDs.

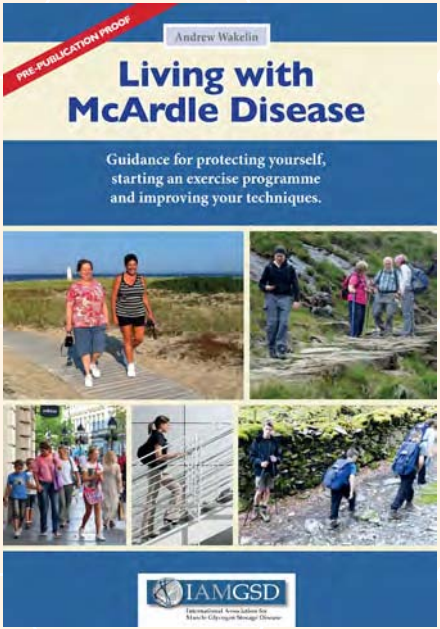
Our AGSD-UK web site's new incarnation (coming soon) will remain the place to go for UK-specific content.

The site is endeavoring to cover all muscle GSDs where exercise intolerance is the main symptom. With some muscle GSDs having as few as four diagnosed cases worldwide, the variation in knowledge about these conditions varies considerably. This is reflected in the variable amount of information that IamGSD can offer on the different muscle GSDs.

It is thought that there are now about 4,000 people diagnosed with McArdle's worldwide, more than the combined total of all the other related muscle GSDs.

Living with McArdle Disease booklet

IamGSD has produced a pilot edition of a new booklet called "Living with McArdle disease". 200 copies have been distributed at walking courses and conferences. A review has suggested that's the booklet could beneficially be expanded by another 10 pages, and focused even more on those who are worst affected. The title is to be changed to "Improving life with McArdle's". It is hoped that the full edition will be published around the middle of the year.



Nutritional Ketosis – Situation Report

IamGSD has prepared a 'Situation Report' on 'Nutritional Ketosis in McArdle Disease'. Around 200 people are now adhering to the diet in various guises. The intent of the report is to provide clinicians and researchers with insight into the knowledge gained from patients' own testing and development of nutritional ketosis.

As patients, we are 'experts' in managing McArdle disease. The experiences we have all shared can hopefully serve to inform formal research and speed the path to an eventual determination of whether this is a suitable nutritional strategy for individuals with McArdle disease.

The report has been sent to over 50 clinicians and researchers worldwide and received a very positive response. A PDF may be downloaded from the IamGSD web site, see 'Research' then 'IamGSD papers'.

"Keto Summit" to be hosted by UCLH and IamGSD

IamGSD and UCLH are to jointly host discussion of nutritional ketosis for the management of McArdle disease and related muscle GSDs. It is palnned for Friday 16 and Saturday 17 November 2018. Invites are going to leading researchers, clinicians, dieticians, patient advocates and leaders in other medical uses of nutritional ketosis.

Individuals with McArdle disease are finding that nutritional ketosis provides a consistent flow of substrates (free fatty acids and ketones) for working muscles throughout the course of DAILY life. While consumption of sucrose [37g] 5 min pre-activity helps to mitigate symptoms prior to second wind, it should not be used frequently and is therefore not a practical solution for activities of daily living (ADL).

Ketones seem to provide a steady supply of energy for ADLs, which has been identified as an important patient-centered outcome measure.

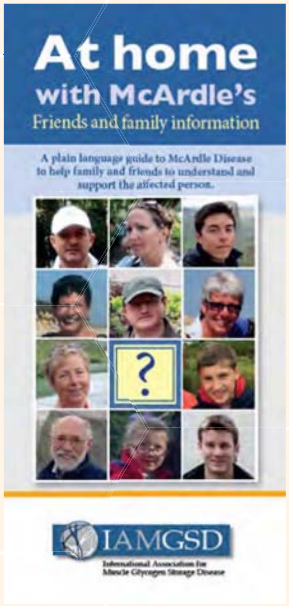
The objective is to discuss considerations and clinical outcomes (patient-reported outcomes; clinician-reported outcomes; and performance outcomes) related to the use of nutritional ketosis in the management of GSD5 and related muscle GSDs.

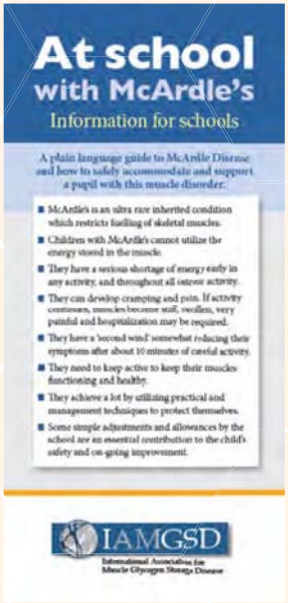
NHS
University College London Hospitals
NHS Foundation Trust

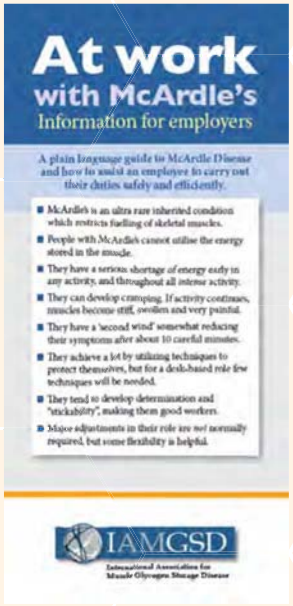
New leaflets

IamGSD has produced three leaflets. The first is based on the AGSD-UK's own leaflet "At work with McArdle's", with has been very useful for explaining things to an employer. The second of the leaflets is "At school with McArdle's" aimed at teachers and other school staff. Lastly there is "At home with McArdle's" – information to help family and friends understand the affected person's situation.

In the UK, the schools leaflet has been distributed to the parents of diagnosed children and the other two leaflets will be sent to everyone in the next few months. If anyone needs them sooner, please just let me know on type5@agsd.org.uk and I can send you copies.







WALKING COURSES

Up a mountain in a hailstorm!

Despite the name “Walking Course”, these events are about so much more – meeting others with McArdle’s, swapping stories, sharing experiences, learning day-to-day techniques and gaining a sense of camaraderie. Oh, and of course having a great deal of fun.

Last summer we had a first for the walking courses. At one point on the summit ridge of Talyfan we found ourselves in a hailstorm and a strong, cold headwind. We soon retreated to safety and pleasant bit of evening sun as we strolled back to the cars. Not that it was all quite so dramatic as those few minutes. Sioned Hosseinian has made a video of still photos of memories from her week on the course last summer, accompanied by her playing her harp. The video is available to view on the McArdle’s YouTube channel.

“Walking with McArdle’s” course 2018

This will be held at a new location in the Brecon Beacons National Park in South Wales, near Hay on Wye. The dates are 21 to 28 August and the cost £300 including accommodation, meals and activities. As in recent years the emphasis is on level and downhill walking, improving techniques and fitness during the week. Towards the end, and subject to weather, on one day we expect to split into two groups, high and low, so that those who wish can have a chance of reaching a mountain peak – possibly Pen y Fan, the highest in southern Britain. If you are interested, contact Andrew Wakelin for more details and bookings – type5@agsd.org.uk.

“The best four days of my life”

So said a youngster attending the “Children & Parents event” in North Wales last summer. This was the third year running that we had run the event and a huge success it was again, with children attending from England, Scotland, France and the USA. A major highlight was to see two girls who had been in wheelchairs in the past, who had learnt the techniques and improved aerobic fitness to the point of being able walk with us for hours, and even play hopscotch together.

Children & Parents event 2018

For 2018 the event will be at the same venue as the walking course, over 17 to 20 August. Contact Andrew Wakelin for more details – type5@agsd.org.uk.

California report

The first walking course organised by IamGSD took place in October 2017 in California. It happened despite our base being burnt to the ground in one of a series of forest fires, just a week before we were due to arrive. Some heroic efforts by Jeremy and Terri, trustees of IamGSD, saw us hurriedly relocate to a new base 100 miles south. Jeremy and Andrew got down there a few days ahead and checked out three routes in a day! Twenty people in all attended with ages from 15 to 78. Twelve of us had McArdle's (GSD5) or Tarui's (GSD7), plus we had parents of the younger members. It was the first time that so many carrier parents (or "hangers on" as Andrew called them!) had got together and it made for some interesting "parents only" discussion sessions. There is a video interview with Jeremy Michelson who organised the course on our YouTube Channel.

For 2018, IamGSD is planning to trial a much shorter course, just 3 days, concentrating on learning the techniques in theory and practice. It will be in immediate succession to the AGSD US annual conference on 21 and 22 September, staying at the same hotel location in Davenport, Iowa. It is a chance to visit both events in one go. Watch out for details on the IamGSD web site.

Walking in the redwoods

Trying walking poles

Our base for 2018

POMPE NEWS

Adjournment Debate on Pompe Disease

House of Commons,
31st October 2017

Speakers:

Nick Thomas-Symonds
Shadow Solicitor General, Shadow Minister (Home Office) (Security)

Steve Brine
Parliamentary Under-Secretary of State for Health

Jim Shannon Shadow
Shadow DUP Spokesperson (Human Rights and Health)

One of our Pompe Support Team members, John Foxwell, raised the question of access to treatment for Pompe disease with Nick Thomas-Symonds, MP for the Welsh county borough of Torfaen. Wales currently approves Myozyme only for children with Pompe disease; Adults must have an Individual Funding Request (IFR) submitted by their medical consultant to the health authorities, and show that they are an exceptional case of the condition.

The adjournment debate, held after the main business of the House of Commons business, contained considerable understanding of issues around Pompe disease and its transcript is recommended reading for the Pompe community in England and the devolved nations.

The full debate can be read on Hansard Online by following the link below and entering “Pompe Disease” into the search field:

https://hansard.parliament.uk

INTERNATIONAL PO

The International Pompe Association (IPA) is a federation of nearly 50 Pompe disease Patient Organisations(PO) around the world. Allan Muir sits on the IPA board and currently holds the office of Vice-Chair. Below is a list of features that describe the IPA.

Mission

The mission of IPA is to campaign for early diagnosis and effective, affordable and safe therapies.

Strive to provide information and support to all patients, their families and others with interests in Pompe disease.

The IPA boardmembers are spread around the world and so most interaction is electronic or by teleconference. We try to meet face-to-face at least once per year. Our activities include:

- Meetings with our members (Including our Annual General Meeting)
- Represent patient community with industries and scientists
- Answer questions of individual members / patients
- Coordinate and support the activities of national groups
- Stimulate the formation of new national groups
- Collect and communicate information through website and national groups.

Pompe connections

A series of information leaflets are produced and published on the IPA website in many languages:

http://www.worldpompe.org/index.php/publications

01 The Signs and Symptoms of Pompe Disease
02 Getting the Right Care for Pompe Disease
03 Adapting to Living with Pompe Disease
04 Breathing Problems in Pompe Disease
05 Nutrition and Dietary Therapy
06 Exercise and Physical Therapy
07 Medical Progress in Pompe Disease
08 The Emotional Impact of Pompe Disease
09 Common Health Concerns
10 Having Children When You Have Pompe Disease
11 Resources for Learning More
12 Acknowledgements
A1 History of Dr. Pompe

Treatment edition

01 About enzyme replacement therapy and Myozyme
02 Finding a treating physician
03 Indication, effectiveness and safety of Myozyme
04 Preparation, administration and monitoring of Myozyme
05 Treatment precautions for Myozyme
06 Practical considerations and supportive management for treatment with Myozyme
07 The cost of Myozyme
08 Pompe patient information collection
09 Outcomes of Myozyme treatment

...INTERNATIONAL POMPE ASSOCIATION

EmPower Programme

The goal of this educational programme was to find young people with Pompe or related to someone with Pompe, to learn to advocate for the Pompe community and maybe join the IPA Board one day in the future.

Erasmus survey

The Pompe Survey is a Patient-reported questionnaire that was initiated in collaboration with Erasmus MC, Rotterdam several years ago. It recorded the natural history of the disease before Enzyme Replacement Therapy was available and now monitors the response of Pompe individuals to treatment.

European POmpe Consortium (EPOC)

EPOC is a consortium of medical specialists throughout Europe (and Switzerland) who have a strong interest in Pompe disease. Membership has grown considerably over the last few years and one IPA member is invited to attends each meeting and has voting rights.

The scope of the consortium is to:

- Improve prospects for patients by combining efforts on understanding the disease process, existing therapies and development of innovative treatment strategies
- Provide guidance on treatment, care and outcome measures
- Harmonize the views on access and reimbursement of therapies in Europe to create equal chances for patients with Pompe disease across Europe

Here is a link to an important paper EPOC prepared last year:

van der Ploeg AT, Kruijshaar ME, Toscano A, et al.

"European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience".

Eur J Neurol. 2017 Jun;24(6):768-e31.

https://www.ncbi.nlm.nih.gov/pubmed/28477382

HOW PHYSIOTHERAPY HAS HELPED ME

Andrea Duckworth, October 2017

I was referred to the Neuromuscular Centre (NMC) in Cheshire, by the Consultant Neurologist following my diagnosis in June 2002. I had an assessment at the NMC in September of that year and have been going for regular treatment ever since.

My condition has changed significantly over that time and my physiotherapy has been adapted accordingly. I now do active exercise as well; under the supervision of the physiotherapy team.

Physiotherapy has helped me in so many ways…

1. Passive stretching by the physio helps stretch out muscles that become tight due to the nature of the condition. E.g. leg, hip and quad stretches. This is particularly important if you are sitting for long periods of the day in a scooter or wheelchair.
2. Trigger point massage by the physio helps relieve pain. This can be a bit uncomfortable but it really helps disperse knots. With pompe disease as with many other diseases, posture can be weak, so neck muscles can become very weak then other muscles overcompensate and everything just tightens and stiffens up.
3. Warm up - The use of heatpads help warm up the area before the physio starts working on them. The warmth is wonderful.
4. Neck stretches - the physio takes your head in their hands and lifts your head up to stretch it out. Then lifts and turns your head to the left and holds it for a bit and then does the same on the other side. I always feel so much better after these stretches.
5. At home. I have been taught to do neck stretches at home, which I try and do on a daily basis to relieve pain and stretch out the neck. This does help.
6. Falls – Due to the nature of Pompe disase I fall regularly. The NMC are able to help with the resultant pain and bruising through massage and physiotherapy. It really does aid recovery!

My top tips..

1. Regular physio really does help keep you as mobile as possible. I always feel I move that bit more easily after being stretched out.
2. It's great to be able to target specific areas. eg: the neck.
3. As your condition changes physiotherapy becomes even more important.
4. The physiotherapist should get as much information about your condition before treating you. They should also ask what particular areas you would like to work on during a session and any problems that you have.

5. Physiotherapy should not hurt! If you are uncomfortable doing anything just ask to stop. We all have different pain thresholds!
6. You can help yourself by asking your physio for exercises that you could do safely at home. You might like to do this with a personal assisstant, friend or family member.
7. Don't be afraid to have physiotherapy. It is a positive step and you will see the benefits.
8. You might like to consider active exercise too. Physiotherapists can advise. I have found active exercise at the NMC has given me a new lease of life and helped with my mobility and general well being.
9. Consider the use of heatpacks and heatpads at home to help manage pain.
10. Don't delay, speak to your consultant for advice on finding your local neuromuscular physio.

POMPEDOO!

Jane Lewthwaite has worked closely with Hania Myers who wrote the book "Smasheroo" for children with Spinal Muscular Atrophy (SMA) to produce a charming children's book "Pompedoo". Jane worked with illustrator Mary Hall to make the book appropriate for our very special Pompe children.

RESEARCH UPDATES

Below are a few research studies that have come to my attention recently:

FDA Clears IND Application for New Gene Therapy

APRIL 23, 2018

Mathew Shanley, Rare Disease Report

The U.S. Food and Drug Administration (FDA) has cleared the Investigational New Drug (IND) application for DTX401 for the treatment of glycogen storage disease type Ia (GSDIa). DTX401 is being developed as an adeno-associated virus (AAV) vector based type 8 gene therapy, and is intended to deliver stable expression and activity of the enzyme glucose-6-phosphatase-α (G6Pase-α) following a single intravenous infusion.

"GSDIa is a devastating disease that requires patients to adhere to a strict and burdensome cornstarch feeding protocol to maintain normal blood glucose levels and prevent hypoglycaemia. Failure of dietary therapy can lead to episodes of severe hypoglycaemia resulting in seizures and death," said Emil D. Kakkis, M.D., Ph.D., Chief Executive Officer and President of Ultragenyx in a press release. "We look forward to initiating our clinical program for DTX401, our gene therapy designed to replace the deficient enzyme in the liver to improve glucose control and prevent the devastating short and long-term consequences of this disease."

An open-label, multicentre Phase 1/2 study of the gene therapy is expected to begin in the first half of 2018, and will evaluate the safety, tolerability and therapeutic response of DTX401 in adults with GSDIa. Key efficacy assessments include time to hypoglycaemia, impact on biomarkers such as lipids, uric acid, and measurement of glycogen in liver.

The U.S. FDA has cleared the Investigational New Drug application for DTX401 for the treatment of glycogen storage disease type Ia

There are 3 potential dosing cohorts in the study, and 3 patients will be enrolled in each cohort. Data from the first cohort expected in the second half of 2018.

In previous preclinical studies, DTX401 has been shown to improve G6Pase-α activity and reduce hepatic glycogen levels, a well-described biomarker of disease progression. As of this writing, there are no approved pharmacological therapies intended for GSD Ia, and approximately 6,000 patients are affected by it worldwide.

...RESEARCH UPDATES

Viking Therapeutics Reports Fourth Quarter and Year-End 2017 Financial Results and Provides Corporate Update

MARCH 07, 2018

Proof-of-concept study of VK2809 in GSD Ia expected to begin in first half of 2018.

GSD Ia is an orphan genetic disease that results in an excess accumulation of glycogen and lipids in the liver, potentially leading to hepatic steatosis, hepatic adenomas, and hepatocellular carcinoma. In 2017, the company announced positive findings from an in vivo study evaluating VK2809 as a potential treatment for GSD Ia. In this study, treatment with VK2809 led to significant reductions in liver triglycerides, liver weight, and other metabolic markers in the glucose-6-phosphatase (G6PC) knockout mouse model, which is intended to replicate the impairment of this enzyme's function in patients with GSD Ia. The results demonstrated VK2809's potent, rapid-acting effects in liver tissue and support advancement of the candidate into the clinic. The company plans to begin dosing patients in a Phase 1 clinical trial in the second quarter of 2018.

Long-term neurologic and cardiac correction by intrathecal gene therapy in Pompe disease

Published on 06 Sep 2017 - 06:00

Abstract

Enzyme replacement therapy (ERT) has recently prolonged the lifespan of these patients, revealing a new natural history. The neurologic phenotype and the persistence of selective muscular weakness in some patients could be attributed to the central nervous system (CNS) storage uncorrected by ERT. GAA-KO mice were treated with a single intrathecal administration of adeno-associated recombinant vector (AAV) mediated gene transfer of human GAA at 1 month and their neurologic, neuromuscular, and cardiac function was assessed for 1 year. We demonstrate a significant functional neurologic correction in treated animals from 4 months onward, a neuromuscular improvement from 9

months onward, and a correction of the hypertrophic cardiomyopathy at 12 months. The regions most affected by the disease i.e. the brainstem, spinal cord, and the left cardiac ventricular wall all show enzymatic, biochemical and histological correction. Muscle glycogen storage is not affected by the treatment, thus suggesting that the restoration of muscle functionality is directly related to the CNS correction. This unprecedented global and long-term CNS and cardiac cure offer new perspectives for the management of patients.

GSD XIV (GSD 14)

https://clinicaltrials.gov/ct2/show/NCT03404856

Study of ORL-1G in Patients With Glycogen Storage Disease Type 14

Orpha Labs, Ankara, Turkey, 06100

Primary Outcome Measures :

- Improvement in liver function.
- Statistically significant decrease in plasma liver enzyme levels

Secondary Outcome Measures :

- Improvement in serum transferrin glycosylation pattern.
- Decreased serum level of hypoglycosylated transferrin.

Structural Insights of Glycogen Metabolism in Exercise and Health

www.advancedsciencenews.com/structural-insights-glycogen-metabolism-exercise-health/

By Anar Murphy, Posted on January 23, 2018, Advanced Science News

An integrative approach of modelling and hydrogen-deuterium exchange helps to describe the enzyme that regulates glycogen levels in muscles and liver.

“In the human body, the cells of the liver and skeletal muscle are largely responsible for synthesis and storage of glycogen. Phosphorylase kinase defects and deficiencies can lead to a glycogen storage disease with mild to severe metabolic myopathies, exercise intolerance, enlarged liver or heart, low blood sugar, and many others.”

OBITUARIES

Charlie Golder in 2016: he had a passion for sport and music and spoke many languages

Charlie Golder
1990–2018

Although the family home is now in France, many of you will have met Charlie Golder and his mother Lucy at several of our past conferences. Lucy recently wrote to me saying:

"As you know Charlie's wishes were that he became a tree. He wanted people to visit his tree and for the closest members of the family join him at the end of their lives in his garden.

Rather than asking for flowers, we have asked that those who wish, to contribute to the Charlie's Garden fund and there is to be a percentage of that which I would like the International Pompe Association to have, and a percentage towards cancer research."

Charlie is now in the guardian newspaper, what an amazing person he was, I am so proud to be his mum."

CHARLIE GOLDER OBITUARY

www.theguardian.com/media/2018/jan/31/charlie-golder-obituary

Charlie Golder, who has died aged 27 of cancer, loved writing and football, and last year combined these passions when he started his own agency, Golder's News and Sports, to provide what he called "off-the-wall" sports news.

Charlie studied demand and identified patterns, especially in foreign sports news that was not being reported in the UK and which he thought might interest his readers. The more outlandish, quirky, funny or tongue-in-cheek the story the better. Charlie had a real knack for knowing what readers wanted, not least because he was a sports enthusiast himself, a huge Chelsea fan with a tremendous knowledge of football, says his brother Joe.

Charlie was born in Chichester, West Sussex, son of Andrew Golder, a singer and narrator, and Lucy Strachan, a librarian. While he was still a child, the family moved to Thiviers, a small town in south-western France, where Charlie went to school. As well as sport, he loved wildlife and music. He learned the oboe at the age of seven, and later played acoustic, electric and bass guitars.

He was 15 when he was diagnosed with the rare and potentially fatal Pompe disease, which wastes the muscles and the heart. This required fortnightly intravenous treatment for the rest of his life, but that did not stop Charlie from obtaining a first degree and then a master's in linguistics from Nottingham Trent University.

He then moved to Vienna where he became fluent in German, obtained a teaching diploma, and worked at the University of Vienna's Language Centre, before realising his dream of living in Japan. He had taught himself Mandarin Chinese and Japanese as a child, and had a lifelong fascination with Japanese culture.

In 2014, Charlie was given the all-clear after treatment for a tumour behind his nose, and immediately started a full-time job with the Central European News (CEN) agency, rising rapidly to be chief subeditor. He met his soulmate Benedetta Tamburini at a party, and enjoyed life. With CEN's approval, in 2017 he hived off its sports coverage into Golder's News and Sports.

In December 2016 Charlie was told the cancer had returned, and was metastatic and very aggressive. He was in constant pain but continued to keep an eye on articles from his hospital bed.

I met Charlie's family 15 years ago. Like everyone he came across, I was immensely impressed by his positive attitude, charm, sense of humour and, later, his will to live against odds that would have defeated many people long before. He never complained.

He is survived by his parents and his siblings, Joe, Luke, Georgi and Anaïs. Joe will take over the sports news agency.

Barbara Casassus, wed 31 Jan 2018

CHRISTINE LAVERY

Announcement by MPS Society

It is with the heaviest of hearts that we announce the death of Mrs Christine Lavery MBE, Chief Executive of the Society for Mucopolysaccharide Diseases (MPS Society)

Christine sadly passed away on Tuesday 19th December 2017 in hospital surrounded by her family, following a brief illness.

Christine has tirelessly championed the MPS Society from its very conception in the early 1980s up until her untimely death. She was a formidable lady who cared passionately for every MPS Society member, past and present. Her efforts saw her work with patients, families and professionals all over the world as she dedicated her life to improving the knowledge, advocacy support and clinical outlook for patients with MPS. We are all deeply saddened at the tragic loss of her life.

The MPS Society will endeavour to make Christine proud and continue her legacy of providing professional support to individuals, families and professionals affected by MPS and related diseases throughout the UK.

Signed: The Chairman of the Trustees on behalf of the MPS Society

The MPS Society were founding members of the LSD Collaborative and Christine's experience and wisdom were vital ingredients to the success of that group. She and the MPS Society have been a great influence on the work and ambitions of the AGSD-UK. She will be greatly missed by all the charities to whom she willingly offered her advice.

Allan Muir

CONTACTS

The AGSD-UK Ltd is managed by a Board of Trustees elected by its members at the AGM held each year during our Annual Conference.

Board Members (trustees)

Michael Porter (Chairman)
> chairman@agsd.org.uk

Jayesh Pindolia
> jayesh.pindolia@agsd.org.uk

Stuart Alderson

Nick Jones

Ailsa Arthur

Jane Guy OBE

AGSD-UK Office

Allan Muir (Charity Director)
> allan.muir@agsd.org.uk

Old Hambledon Racecourse, Droxford, Southampton, Hampshire, SO32 3QY

Monday to Thursday: 0300 123 2790

Out of hours: 0300 123 2799

International: +44 1489 877 991

Volunteer Coordinators

GSD 0 – Abbie Maguire
> type0@agsd.org.uk

GSD I (1) – Jason McMillan
> type1@agsd.org.uk

GSD III (3) – Sylvia Wilson
> type3@agsd.org.uk

GSD IX (9) – Dan Machin
> type9@agsd.org.uk

Pompe – Allan Muir
> allan@pompe.org.uk

McArdle – Andrew Wakelin
> type5@agsd.org.uk
Phone: 0300 123 2795

Fundraising enquiries

> fundraising@agsd.org.uk
Phone: 0300 123 2790

Specialist Care Advisor – Pompe Disease

Jane Lewthwaite
> jane.lewthwaite@agsd.org.uk

Phone: 0300 123 2792

Mobile: 07484 055334

Facebook: jane-agsduk

Support Teams

Pompe Support Team
> pst@agsd.org.uk

Cori Action Team Support
> cats@agsd.org.uk

If you have anything interesting for the newsletter we'd be very pleased to hear from you.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WWW.STUDIOHOI.COM