Glisten

Glycogen Storage News

Summer 2017

AGSD UK
Association for Glycogen Storage Disease

Rare as Rocking horses

by Gemma Seyfang

Rare as rocking horses that's what we are,
Join me as I explain a snippet of life so far.
The difficulty I feel to rise from certain chairs,
And the way I struggle to climb up the stairs.
Often passers-by will presume that I'm drunk,
'Cos when I walk my hips sway to the front.
It's very easy for me to trip, stumble and fall,
And if you're with me you may feel uncomfortable.
I've been told there are only 140 people in the UK,
Being treated for this disease by the NHS today.
The treatment that I'm receiving is aiming to keep,
My muscles in a stable condition so I can stay on my feet.
My every day struggles include tying up laces,
Picking up something off the floor and reaching high places.
Exercise is difficult even to walk for just a while,
Will make my muscles hurt and ache even though it was just a mile.
Climbing in and out the car or rising off the loo,
Don't get me started on shaving my legs, it's difficult, it's true.
My Lung muscles are also weak, especially at night,
It leaves me feeling unrefreshed when I wake with the morning light.
My family and close friends are always there for me,
I cannot thank them enough and I love them, I hope this they can see.
This is my life, this is who I am,
I will remain as independent as I can.
This disease is part of me and so I must embrace it,
Become more knowledgeable, do all I can, so I can better face it.
Pompe Disease will continue to make my body suffer,
But "Together We Are Stronger" is the motto we all utter.
I am like a diamond that's precious and RARE,
I'm not like those stones that can be found everywhere.

Glisten

AGSD UK
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Chairman's Update 2017

Since our 30th anniversary conference at Tortworth Court the trustee group has been extremely active and engaged. Unfortunately due to family circumstances we have had one trustee resignation – Shaun Griffin and I would like to place on record our thanks to Shaun for his support and guidance, particularly utilising his skills as a chartered accountant in helping us with our financial governance. The addition of three new trustees however has been a real boon to the trustee board, though we are still looking to add further trustees to support both strengthening our all round skill set and also meeting our diversity aims.

In addition to business as usual the main focus areas for the trustee board since the last conference have been: rebranding and web site development, the creation and implementation of a wider suite of policies and procedures, the implementation of a secure IT infrastructure and environment and in order to support the growing workload the creation of a number of trustee sub committees. Aside from the rebranding and web site redevelopment most of results of this work will be invisible to the members but is essential in ensuring that the charity remains legally and regulatory compliant and also positions itself to be able to apply for and be successful in securing larger charitable grants.

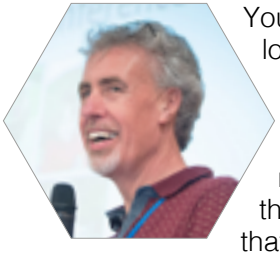
Working with an experienced web development company and a small focus group from within the charity a fresh new brand that seeks to reflect the values and aspirations of AGSD-UK has been developed and we hope you all come to identify with it.

The feedback received to date regarding the work being undertaken by our Pompe Specialist Care Advisor – Jane Lewthwaite has borne out the view of the charity that there was a large unfulfilled need in the Pompe community. That need is now being addressed and is delivering significant tangible benefits. Our aim is to secure funding to recruit additional SCA's to support the other GSD types.

Income generation continues to be a challenge as it is for most small charities. Our newly formed fundraising sub-committee, ably supported by an experienced associate, has identified a number of workstreams that we feel have real potential. We recognise however that we could not survive without the fantastic fundraising activities that you all undertake on behalf of the charity so please keep up the invaluable and much appreciated work. I would also like to thank our industry partners for their ongoing support.

Mike Porter, Chairman

Charity Director's Update



You will no doubt have noticed the fresh new AGSD–UK logo embellishing the cover of Glisten, and used elsewhere in our more recent publications; the logo is already turning heads on our running vests, cycling jerseys and T-shirts, and we hope to launch a bright new website to match at our October conference; that will complete the rebranding exercise. I do believe that our colourful new image reflects not just the diversity within the organisation but also a feeling, within the AGSD–UK, that we are extending our reach and working hard to provide improved levels of support to our members and other stakeholders.

There is so much activity at the moment, including:

- Vital work to ensure that we are compliant with all the latest charity rules and regulations
- Staff and volunteers working together to prepare new publications and online content
- Fundraisers engaging in a wonderful variety of activities to raise both awareness and sponsorship
- Improved support to individuals and families by our Specialist Care Advisor
- Efforts to raise funds to employ additional support staff and care advisors
- Preparation of an eLearning module for GPs and other health workers
- Preparations for our October conference

It is a huge frustration to me that the increased workload has meant that we have failed to publish more issues of Glysten over the last twelve months, but please be assured that we are very active on behalf of our members and we are hopeful that frequency of this vital publication will be restored very soon.

Conferences are always wonderful opportunities to exchange knowledge and meet old and new friends; the second International GSD Conference held in Groningen, The Netherlands, was no exception. It had both scientific and patient-focussed presentations and it was great to see so many of our members contributing to the sessions.

The eLearning module I mentioned above is a collaboration with the Royal College of General Practitioners. The intention is to provide a very short introduction to GSDs that will encourage GPs to refer their patients to appropriate specialists (metabolic, neurology, pulmonology etc.) to improve the chances of early diagnosis; late diagnosis is common to most GSDs and is life-threatening in some cases. We are hopeful that the module will be online in time for our conference.

So, our conference promises to showcase the great strides we are making as a charity, as well as highlighting some of the research into therapeutic and improved care for GSDs. I hope that you can join us in Nottingham this year.

Allan Muir, Charity Director

Gosh Family Day

Jane Lewthwaite, SCA

On Saturday 20th May families with children with hepatic GSD and Pompe attended a fun filled family day at Great Ormond Street Hospital, London. The event was the first one held and was kindly sponsored by the AGSD–UK.

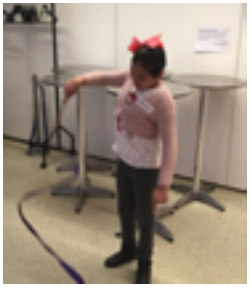
The day started off with 'Aunty Julie' the entertainer who kept the children amused with tricks, jokes and music. This was then followed by art and craft activities and games for the older children, coffee, tea and juice as well as biscuits and fruit were available throughout the morning.

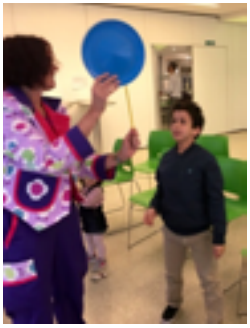
Parents were able to sit and chat with each other and many of the young people interacted and talked about their condition with each other and certainly plenty of friendships were made. Newly diagnosed families were able to meet other children and their families, which they found helpful to know there are other families out there. It was lovely to see a group of dad's chatting over coffee discussing common issues regarding their children resulting in questions from them to the professionals.

Mel and Mathilda (Metabolic Nurses) were on hand to not only make introductions but to answer and questions raised and Dr James Davison provided an overview of the hepatic GSDs and Pompe, including current research and treatments, his talk was very adaptable as some of the older children sat and listened so he made sure it pitched his talk at all levels.

Fun was had by all; feedback from the families was very positive asking for another one to be done next year. One mum said *'it was nice to see other families and know that others are going through the same issues'.*

"Its nice for the children to meet other children with the same condition"





Safeguarding

Jane Lewthwaite, SCA

What is Safeguarding?

Last year AGSD–UK celebrated its 30th year of existence. As a small charity we have achieved a huge amount in those 30 years and now we are growing further and hopefully, stronger too. We want to expand and employ more staff, including Care Advisors, which brings with it additional responsibilities.

Our Trustees have had help in recent months from Jane Guy OBE in reviewing and updating new governance for AGSD–UK. Jane has vast experience in helping organisations ensure they are compliant, not only with the Charity Commission requirements, but also with the Law.

Safeguarding includes protecting people's health, wellbeing and human rights, and enabling them to live free from harm, abuse and neglect. Children and vulnerable people need extra protection. We always aim to ensure that anyone who has concerns about vulnerable people can get them checked out, and know they will get a response.

Jane Guy has updated our Safeguarding Policy. This means that we will ensure everything within AGSD–UK is as safe as it possibly can be for everyone who participates in it. So, volunteers might need to have a DBS check depending on what work they are doing. Designated people, in our case the Chair of Trustees and the Specialist Care Advisor will have updated training in Safeguarding and Child Protection. They have both already completed NSPCC training.

Jane Lewthwaite will be in touch with volunteers individually to discuss whether the implementation of the Safeguarding Policy affects them, but if you have any questions or suggestions please get in touch on 0300 123 2792.

The full Safeguarding Policy is posted on our website on the Governance page.

Personal Health Budget

Larissa Lowe, GSD 4 Coordinator

As a person classed as "disabled" pursuant to the Equalities Act, I habitually screen for new developments in equality legislation and in our health care system (through statute, regulations and/or case law or local health initiatives).

I have been following the progress of the 'Personal Health Budget' (PHB) a non-means-tested sum for which people who are eligible for NHS Continuing Health Care and children in receipt of continuing care may apply, in order to fund their identified health and wellbeing needs.

Personal Health Budgets have been around since 1 April 2014 but it won't come as a great surprise that not all hospital trusts are as familiar with PHBs as they ought to be. Implementation is further affected due to lack finances available to the NHS in general and specifically due to a delay in the development of the systems and processes required to implement PHBs.

A PHB is a sum of money (calculated and agreed between the patient and their local NHS Trust) available to the patient to support their health and wellbeing needs in order to give patients with long term conditions and disabilities greater choice, flexibility and control over their health care and the support they receive.

The benefits of PHBs reported to date are that the patient (or their representative):

- **know the budget available for their healthcare and support.**
- **have a greater choice in how to spend their budget in a manner that promotes their physical and mental health**
- **be involved in the design of their care plan.**
- **can spend the money in ways and at times that benefits their treatment.**

Patients suffering with a rare disease have had to become "experts" on their disease, often having to educate their physicians. These would be the same physicians making decisions on immediate and long term care requirements, with patients often having to sit through unnecessary treatment to prove said treatment is not conducive to their health (and at best is a waste of time). With the patients' input in their care plan and in deciding how to spend their PHB on treatment and care that is appropriate, where it is needed and when it is needed is a step in the right direction.

If you are eligible for NHS Continuing Health Care or any of your children are in receipt of Continuing Care, do remind your local NHS trust of the existence of PHBs.

If you have any questions or for any other legal advice, do contact me at: larissa.lowe@michelmores.com

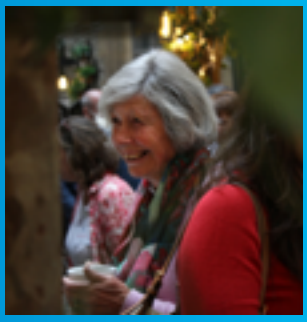
Thank you for your participation!











AGSD–UK Annual Conference

Date
Weekend of October 28th and 29th 2017

Venue
East Midlands Conference Centre
Beeston Lane
University of Nottingham
NG7 2RJ

Our conference this year will be hosted at the impressive conference facilities in Nottingham. Again, the event will run from Lunchtime on Saturday through to Lunch on Sunday. A gala dinner will be provided on the Saturday evening.

We would encourage you to arrive during the morning of October 28th, as this will give you time for networking, to get to know other members and to meet up again with old friends. Bedrooms at the Orchard Hotel won't be available immediately, but Room-keys will be handed out at registration or during the afternoon break.

The Conference will commence on Saturday 28th October at midday with registration and lunch. During the afternoon, there will be the usual eclectic mix of speakers and workshops. The day will finish with a Gala Dinner, starting with a drinks reception at at 7.00pm.

The event will continue Sunday morning, starting with the AGSD–UK Annual General Meeting (AGM) and finish at 1pm with a buffet lunch provided. Again, if you wish to stay for the afternoon, there will be opportunities for networking.

Raffle
Volunteers will again be running a raffle during the conference and we already have one very special prize. People were very generous with prize donations last year and, we will be very grateful for whatever you can donate this year.

Conference Registration
Online registration available at:
www.agsd.org.uk
If you do not have Internet access, then a pull-out form is included within the pages of this newsletter.

Speakers
Invited Speakers are not required to officially register, and all meals will be provided, but it would be helpful to know your requirements in terms of dietary requirements and conference dinner. Please use the Health Professionals' registration form to provide this information.

Accommodation
We have reserved a large number of bedrooms at the adjacent Orchard Hotel, AGSD–UK subsidises the cost of accommodation. Most rooms will accommodate up to 2 adults plus one child but families of 4 may need to book two rooms.

Accessible Rooms
Please only ask for an accessible room if you really need it; there are limited rooms available at this venue. If you need any aids such as raised toilet seats (with arms) or grab-handles, please contact us and we will make arrangements with the hotel.

Residential and Day Delegate Packages
Please indicate on the form whether you will be registering as a residential delegate or a day delegate.

Travelling to the conference centre and the hotel:

Rail
The nearest stations are Beeston Station and Nottingham. There are over 30 direct trains that travel between St Pancras and Nottingham daily.

Road
For travel by car, use the following navigation coordinates (52.939349, -1.202139) and postcode (NG7 2QL) to find the hotel. Guests are entitled to complimentary car parking.

Air
East Midlands Airport is 14 miles from De Vere Orchard Hotel via the M1 North. There is a regular Skylink Nottingham bus from the airport directed to Broad Marsh Bus Station—minutes from the hotel.

We would like to thank our generous sponsors for their support:

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30th Anniversary Conference 2016

Feedback Summary

The AGSD–UK annual conference was held at Tortworth Court in Gloucestershire. A last minute glitch at the original venue meant we switched to this fine, four star hotel and conference facility. This change meant a lot of extra work but also resulted in us spending our time at an extremely pleasant hotel, much higher standard than we can usually afford.

Feedback forms were placed in all delegate packs and we had a 25% return rate. Through feedback we hope to;

- Continually listen to our members and stakeholders
- Improve their conference experiences year on year
- Consider the changes we might make, subject to cost

97% rated organization and value for money as excellent

Asked what was least useful, over 50% of responders said 'nothing'.

50% of the sessions were rated as 'most useful' by at least one person

–

"I gained knowledge for the future, and also gave and received support from other people"

"Usually I feel as though I am the 'different one', here I feel 'normal'"

"I really enjoyed meeting people, it made a relaxing getaway from being a 24/7 carer"

Organisation and Value for Money
Universal satisfaction:
- 97% said both were EXCELLENT
- 3% said FAIR.

Most Useful Session
- 4 people said 'all'.
- There were 31 sessions in total and 16 were rated by at least one person as their most useful. So over 50% of the possible sessions were top favourite for at least one person.
- Dr Roberts' Pompe & ERT session scored highest with 9 rating it as the most useful.
- Research updates from Dr Weinstein and in McArdles also rated highly.

Least Useful Session
- 50% of people said 'no session was least useful'.
- Only 11 ratings were given for six different sessions.
- Sessions on 'Gene Therapy' and 'Knowledge, Value and Patients' both scored most with 3 ratings each.

"Friendship, information and more confidence in humanity"

What did you gain from attending?
Comments grouped and in order of frequency;
- Additional knowledge/insight
- New information, new research
- Networking, friendship, new people, companionship, support
- A sense of belonging
- New insights and advice on coping
- Hope, encouragement, positivity
- Giving support to others, sharing

"Brilliant conference, proud and positive"

What could be improved?
Comments grouped and in order of frequency;
- More informal patient discussions, more round the table time
- More short specific sessions, less general sessions
- Practical suggestions ; allow 5 minutes between sessions for people to move about, microphone and acoustics, more space
- Fewer slides and fewer very detailed slides
- Less technical jargon

"More of the same as this year, it has been fabulous"

What would you like at future conferences?
Comments grouped and in order of frequency;
- More time
- More informal or practical sessions
- Sessions for carers
- Keep doing what you are doing, more of the same
- Entertainment, fun element
- Dietician

Lastly, a note on resources. We are a small charity, the Charity Director plans and organizes the full conference single-handed with Type Co-ordinators contributing to content arrangements. He was ably helped over the weekend Barbara, Jane and PST members; Theo, Angela, Ben, Sam and Amanda. Thanks to Jamie and Dennie for photography. Thanks to Diane Evans, our venue finder. Thanks too to our Trustees who contribute hugely. Thanks to Amanda for organizing the raffle and to all who donated prizes or bought tickets, raising £650.

Mental Health and Wellbeing

Extracts from Annabel's talk during the Pompe Workshop at the AGSD–UK Conference.

I'm now 34 years old and was diagnosed with Pompe about 5 years ago. I'd like to talk to you both from a personal and a professional point of view. I'll share a little about my diagnosis of Pompe, the effect that it had on my mental wellbeing and how I coped. I'd also like to explain a little about some of the common causes and maintenance issues surrounding one of the most common mental health conditions, depression.

My background

Professionally, I started my training as a Cognitive Behavioural Psychotherapist, or CBT, about 10 years ago, and I specialise in the more common mental health problems such as Depression and Anxiety" Disorders. CBT is rapidly being offered to more and more people across the country suffering with all sorts of psychological problems. This is through a government initiative, started a few years ago called IAPT or improving access to Psychological Therapies. IAPT is an NHS programme rolling out psychological services across England offering interventions approved by the National institute of Health and Clinical Excellence (NICE) for treatingpeople with depression and anxiety disorders.

What this should mean, is that if you ask your GP for Psychological help, you should be offered an assessment, and in some cases, that can take only a few days. CBT is being offered as an alternative to medication or used alongside medication depending on the need. It's a fantastic service of which I've very proud of being part of.

CBT isn't for everyone and it isn't a miracle cure. It takes tremendous skill, strength and courage of behalf of a patient, to actively overcome their difficulties. But time and time again I'm humbled by the effectiveness of CBT, the determination of people to help themselves and the improvement in people's day to day lives. If CBT, or other therapies within primary care aren't suitable, then patients can be referred to secondary or specialist services for further care.

Annabel Rajgor

Depression

Depression is a genuine health condition, it's not all in the mind. It often needs treatment from a professional and often it doesn't just always go away on its own. It is a common mental health condition. Globally, an estimated 350 million people of all ages suffer from depression. It is the leading cause of disability worldwide, and is a major contributor to the overall global burden of other diseases.

For a diagnosis of Depression, core symptoms, must occur consistently over a minimum of two weeks. These symptoms include reduced pleasure and interest in everyday life, persistent low mood, reduced energy and reduced activity. Problems with sleep, reduced appetite, increase irritability, reduced self-esteem, feelings of worthlessness, powerlessness and hopelessness.

Triggers for depression can often involve:

- Significant losses in a person's life
  Bereavement, good health, relationship ending
- Major life transitions
  Having a baby, changing jobs, retirement
- Relationship disputes
  Partners, employers
- Role changes
  Becoming a parent or a care-giver
- Prolonged periods of stress and anxiety.
  Bullying at work

I'll try to explain these triggering factors through my own experience.

The diagnosis of Pompe for me, like many of us here, took a while. I would often see my GP for pain, but not for muscle weakness. At the time and growing up, I didn't realise my body was weaker than other people's; I just thought I was unfit. When the testing got a little more serious, you'd often here me speak about feeling like a piece of meat. What I meant was, I felt I wasn't being treated holistically like the complex human being I was. Health professionals, lovely and highly skilled, as they were, were only interested in their specific area of testing; no one really asked me how I felt. Until I met my specialist team in Cambridge who then directed me to AGSD–UK.

But for all those years the effect of ongoing pain, weakness and uncertainty were taking their toll on me. I was working in London, and I couldn't keep up with friends going out and partying, I'd get frustrated, I couldn't get up and down the stairs on the tubes. I felt exhausted, every day struggling to develop my career and be competitive.

So here we have prolonged pain, ongoing stress of appointments and time away from work, loss of quality of life through weakness, anxiety and uncertainty. Some of those key triggers for depression.

When I was diagnosed with Pompe on some level I felt relieved after many years of not knowing what was wrong with me, but there was also a tremendous sadness and despair, at the sense of loss I felt. I would never run a marathon for example, I mean, not that that was ever one of my goals in life, and it might sound trite, but to have those sorts of choices taken away was brutal. I felt weird, weak and vulnerable. It was also a role transition. I now saw myself as a disabled person, and all the stigma and discrimination people with disabilities face today was now going to be directed at me. I had to adapt to dealing with the fact that in my late 20's I needed a walking stick when out on my own. I needed hand rails, flat unsexy supportive shoes, lifts, raised toilets, radar keys, grab rails, an automatic car, etc. etc.

I grieved, I got angry, I cried, I ignored it, I got depressed, I faced it, and eventually I worked through it.

These things helped:
1. Family, friends and my husband were the most important
2. Homecare team and specialist doctor
3. AGSD–UK

But ultimately it was down to me.

I never like to be told I can't do something. If Pompe said I couldn't go travelling. B***er that, I was going travelling! I developed my career, I got married and had a beautiful baby daughter. If I couldn't ride a bike, I'd buy an electric one. I wouldn't be beaten.

I realised I had to make adjustments to have equal access to things other people took for granted. Travelling: we planned mobility issues, which basically involved my husband carrying everything! Career: I moved back home and reduced my time at work. In this case having a supportive boss was essential. Having a baby: well, that's an ongoing "make it up as we go along" strategy!

Being a therapist also meant that I had some of the skills and insight to help me work through some of my low mood and anxieties. When I felt anxious about what people would think of me with a walking stick, I went out to find out. I tested my assumptions. And in fact, I found most people were fantastic, they moved out my way because now they had a visual cue that I struggled a bit more. I was less bitter about the world.

I was also able to argue some of the negative and unhelpful thoughts which popped through my head frequently. I could see that having a disability, and struggling emotionally with the diagnosis and management of it was not a weakness; it was normal, in fact it was a sign of my strength.

Counselling

I also sought help from a professional counsellor. In a safe place I could fully explore without judgment and shame all the things I was feeling. I felt that the Pompe was taking over my life. My mobility everywhere, my diet, my career, my relationships, my future. But I learnt that I was a person who now lived with a long term condition, I wasn't that condition.

Depression is not only a consequence of living with a long-term condition, it can severely disrupt a person from properly looking after themselves. Depression means that its harder to do things, even like getting out of bed is a milestone for some people, So finding the motivation to do some light muscle strengthening exercise is a long way off. People with depression often don't eat well, reduced energy and activity can mean eating more convenience food or snacking or high carb or sugary foods, people may go out less due to

anxieties about mobility issue or social stigma. Worry and anxiety can often lead to procrastination and poor problem solving, not going to routine appointments or getting things checked out. Conditions can get worse. It's a downward spiral.

There are new challenges I face now. That grieving process isn't linear, its messy. I still have times when I get angry and anxious. I feel anxious that I'll lose my car and my blue badge one day soon, I feel sad that I won't be able to do things with my daughter that other mums can do. Thankfully for her, she'll grow up thinking disability is the most normal thing in the world, and hopefully the new generation won't have so many hang ups about it. I feel sad when I struggle to do something or something is taken away, but I remind myself that Pompe and my experience of depression has given me something. For me, it's given me a deeper understanding of what it is to be human. "To err is to human". We're not perfect, we're not all the same. I totally get that and I'm more comfortable in my own skin. Depression is something you can hide, but disability isn't so easy to cover up. I've been forced to confront some of my demons about my Pompe, and how it affects my life, and in that I've developed a better understanding of myself and what it means to be human.

If you are concerned about your own, or someone you care about's mental wellbeing:
Talk about it.

- Seek help from friends, family, GP.
- Research online about CBT and mental health.
- Look after your basic wellbeing - eat well, sleep well, exercise, socialise, get a hobby, do something for other people, these are all good for our mental health. Mental health first aid!
- Go and see your GP, if they're not very psychologically minded, find another GP.

What health professionals can do:

- Collect some leaflets for your local NHS IAPT services to hand out, or just ask people how they're coping with it all. Allow an extra 5 minutes if you can.
- Get involved in mental health awareness week. See what your organisation is doing to support mental health.

Where to find help:

- AGSD–UK website and Glisten
- IAPT website (see below)
- Self-referrals in some areas
- Living life to the full
- Mobile apps
- Private CBT - BABCP website
- Books
  "I Had a Black Dog" by Matthew Johnstone
  "Overcoming Depression" by Paul Gilbert.
  Audio CD version also available.

Websites:

- NHS Improving Access to Psychological Therapies (IAPT) programme
  www.iapt.nhs.uk
- British Association for Behavioural and Cognitive Psychotherapies
  www.babcp.com
- Living Life to The Full
  www.llttf.com
- World Health Organisation
  www.who.int

Conference Report

A flavour from Groningen International GSD Conference

Jane Lewthwaite

The first International GSD Conference was organised in Heidelberg by the German GSD patient support group. This year it was organised by the Netherlands and provided a scientific programme together with parallel sessions for patients and families. The programme and abstracts of presentations are still available on the conference website: www.igsd2017.com.

The conference was attended by several AGSD–UK representatives and individuals; below are the impressions of our Specialist Care Advisor, Jane Lewthwaite, who represented AGSD–UK in Groningen at this important event.

Patients, Families and Professionals.

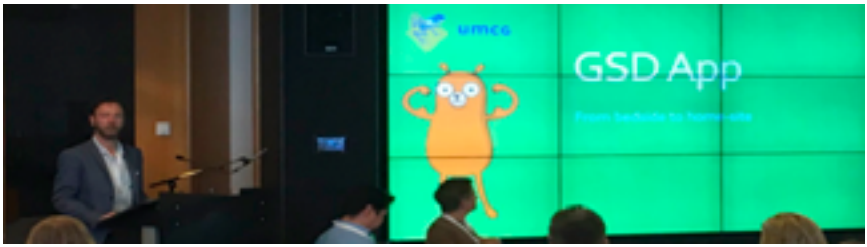
IGSD2017 was a chance for all three to meet, exchange ideas, views and share experiences with a view to providing better care and more agreed, common approaches. AGSD–UK's Jonathon Mosedale ran a networking session on 'Charting Experiences of Myopathy in GSD III'. Andrew Wakelin talked on McArdles - Inspirational Patients and also on 'Latest Developments in Walking Courses for children and parents with McArdles'. He also introduced the International Association of Muscle GSDs (IAMGSD) to the conference. As an example of who attends, one person was there find out more about GSDs because his niece in India is going through testing. Another person attended, who had McArdles, it was the first time he had ever met anyone else with it though he was diagnosed a long time ago.

Exercise

Dr Ulrike Steuerwald spoke on her research in 'Sports for Liver GSDs'. Clear, concise and motivating, she said *exercise is vital*. The body acts like a tool to metabolise food intake and if it is not kept in good use it becomes blunt and ineffective when asked to be active. The more people take appropriate exercise the less dependent they will be on burning carbs and glycogen because muscles are well exercised. People who exercise benefit from better mood, quicker learning and prevention of lifestyle problems [reducing the risk of Alzheimers disease and so on]. She also found that it improved lipid profiles, blood pressure, bone density and insulin sensitivity. She suggests 30 mins walking 3 times per week. Dr N Preisler spoke about Exercise Tolerance in GSDs, ' The less you exercise, the harder it is – a cycle of disuse'. Don't exercise if you are still tired from the last sessions, monitor CK levels after exercise and be realistic. Aim for endurance rather than short, sharp bursts. Start low – go slow.

The GSD App – by Sebastien te Boekhorst

This App might become very useful in the future, so watch this space and we will include something in a later newsletter. The App aims to collect data from wearable technology, diet, glucose and ketone concentrations to inform medical teams and enable quicker treatment. It also stores emergency and diet information.



James Lind Alliance
The James Lind Alliance explained that they aim to tackle treatment uncertainties together with patients and medics, and set priorities for research through gathering information. A later session was the inaugural meeting for the 'Worldwide Hepatic GSD Partnership'. They might be seeking people to contribute their views so we will alert AGSD–UK members by email.

Peter Berghardt – from Heidelberg.
'Negotiation is for politicians, not for parents of a child with a GSD' he said 'following a diet or treatment regime for a GSD is non-negotiable. So, it should be presented as clearly as not running across a road in traffic'. He also said the most common question he gets asked is how to reply when a child asks,' Why do I have a GSD?'. He suggested, *'We are all humans and humans are different, we have different faces and different hair. Some people have an illness and some people do not. Some people have it for life and some people do not.'* Finally, he made an important point trying to help parents with their feelings of guilt in having passed on a disease to their child. He said it is understandable but parents must overcome those feelings because they could not change genetics and most child learning is observational, and *"parental guilt leads to childhood shame'.*

Learning habits in managing your condition – Slow thinking is hard work and leads to exhaustion (Watch a child learning to fasten a button or tie a shoelace – they are slow thinking). So, as much as possible build routines out of crucial self-care tasks and ensure they are passed over to your Fast Processing, they will be less exhausting.

Gene Therapy
Dr David Weinstein presented on GSD 1a, he has had successful trials with mice and dogs and expects to start full trials with adults in 2020. The main challenge for humans is pre-existing anti-bodies which might cause a reaction to the carrier (a virus) that is used for the replacement gene. People who have pre-existing anti-bodies to the virus will not be able to join that trial. For gene therapy to be effective it only needs to increase enzyme activity by 5–10%.

Newborn screening (NBS) for Type II Pompe.
Dr Priya Kishnani found that in a small group of infants where Pompe was diagnosed through symptoms alone, ventilator free survival at two years old was 67%. However, for the 5 infants detected through newborn screening, ventilator free survival was 100% at aged two. In three different US states NBS detected Pompe at 1:9000 to 1:24000 compared to the previous view that it is about 1:40,000. There is a still a big question about identifying adult-onset disease through NBS, she suggested those infants should be monitored for motor functions, hip extensions, gait, CK/ALT/AST increase in blood and Glc4 in urine. The ERT could be started when these factors show disease progression.

Cornstarch Kids Camps USA
Susan Williams, a nurse from Texas, developed disease specific camps for kids in the 1980s. When two of her grandchildren were diagnosed with Type IXb she helped to set up this camp. She explained the whole process; medical needs, equipment needs, location and venue needs. Families and children had a massively positive reaction to the camps. They made friends, did not feel 'unusual', shared tips and learned about their condition all of which lead to better health and wellbeing. Very inspiring.

IGSD2020
It's nearly 4 years since the last international GSD conference in Heidelberg, and dates for the future were discussed; Brazil offered to organise the event in 2020, so perhaps fewer people will be able to attend from Europe. AGSD–UK offered to host the conference in 2023 and 2026 was suggested for the USA. These dates and venues require agreement between the GSD patient and scientific communities.

Conference Report world 2017

WORLD Symposium 2017
Allan Muir

Each year this major international conference is held in the US to follow and support research into Lysosomal Storage Disorders, of which Pompe disease is one. I have been very lucky to find a sponsor to support my expenses at these meetings for the last 3 years. Most of the delegates attending these events are medical professionals who have interests across all GSDs and so they are very useful occasions to network closely with researchers, clinicians, specialists and company representatives.

The symposium in February 2017 was no exception and I was fortunate to spend time with many professionals working towards improved treatments for GSDs around the world. Whilst in San Diego I was also able to meet with Viking Therapeutics who are currently developing a treatment for GSD 1a.

Despite being on the East Coast of the USA, I couldn't escape the familiar faces of AGSD–UK members as they appeared on three different displays at the symposium, a few of whom are shown here.

Legacies

Herbert James Leadley

(d. March 2017)

We are most grateful to the family of Herbert Leadley who have made a donation of £2,000 to AGSD–UK through a Deed of Variation dated 20th March 1917.

Henri Termeer

Former Genzyme CEO Henri Termeer passed away unexpectedly at his home on the evening of Friday 12th May 2017.

Henri led Genzyme for 28 years, joining the company relatively soon after its founding and overseeing its expansion and growth until stepping down following the Sanofi acquisition in 2011. His contributions to the Biotech industry were impressive, not least through his understanding of the enormous role that patients have in ultra-orphan conditions.

John Ernest Clarke

(4 Jan 1946 – 24 Apr 2017)

AGSD–UK has received donations of over £2,000 In loving memory of John Ernest Clarke FRICS, who sadly passed away on 24th April 2017, aged 71. He is survived by his mother Joyce, wife Olga, 3 children and 7 grandchildren.

John lived with the effects of glycogen storage disease for the latter part of his life, but continued to work in the field that he loved. None of this could have been achieved without the support of his wife and family, as well as numerous friends and colleagues. He will be sorely missed.

Donations were made through FuneralZone, a website prepared for funeral directors. John's page can be viewed here: www.funeralzone.co.uk/obituaries/30136

Fundraising

Our members and supporters have been incredibly active lately, joining events we advertise and running their own fundraising activities. We are heavily reliant on charitable donations and so this is all wonderful to see. Below is a small selection of activities and I apologise in advance if I haven't given yours a mention.

Case Studies for fundraising

A small fundraising subcommittee recently held a very productive meeting to focus minds on the current and future needs of the charity. Whilst examining various means of financing our core work and projects, we developed a comprehensive list of essential items to assist fundraisers and funding applications. At the top of the list is Case Studies; we need powerful stories, from the people we represent, that show the difficulties faced by people living with a GSD, and also the benefit they have received from AGSD–UK or other organisations. We hope to build a comprehensive library of articles, photographs and other media, to build compelling and touching stories to help us stand out above our countless competitors.

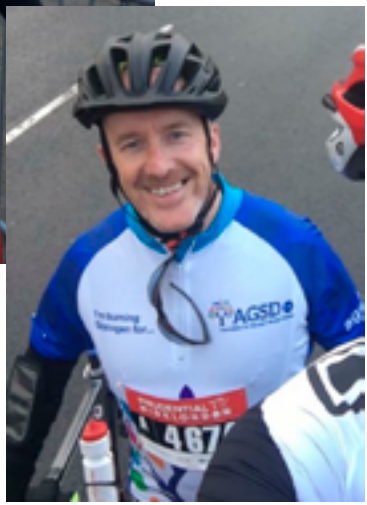
Please take a moment to consider if you would like your story to be told, if you would like to contribute an article, please send it to me, Allan Muir (contact details on the back cover).

Ride London-Surrey 100

Many congratulations to each of our five TEAM GSD cyclists who successfully completed the 100 mile circuit through London and the Surry Hills on 30th July. Between them they raised over £3000 in sponsorship for AGSD–UK.

AGSD–UK has five places reserved for the 2018 event, but we ask anyone who would like to join our team to enter the public ballot first; places are much cheaper for individuals and we can increase our team size that way:

www.prudentialridelondon.co.uk/events/100/entries/



Left page: Emily Thompson and Chris Davies
This page top: Luca Venditto and Mark Bowles
This page bottom: Gary O'Donnell

London Vitality 10,000

This is an annual running event taking place at the end of May each year. AGSD–UK has 10 places of which seven were taken by our supporters, but only five ran in the event because Noor Abdul Ghani and Allan Muir both had muscle injuries on the day. But many thanks to Team GSD who raised over £1050 for AGSD–UK and enjoyed a perfect day for running in London.

Team GSD

Aseel Bdalwi David Burton

Jenny Liquorish Sharon Gilroy Darren Josiah

Jenny Liquorish

So – what can I say?! The Vitality London 10k was one of the most organised, enjoyable runs I've ever done. I hadn't run for a while so I was obviously apprehensive & the normal nerves had definitely kicked in when I woke up on bank holiday Monday, but I needn't have worried. The journey to Victoria was simple & then it was just a few minutes' walk to the start line in glorious weather with two of my lovely running buddies.

The run is suitable for all levels of fitness - the elite runners were finishing when I was less than half way (I won't lie, it's a bit demoralising seeing runners on their way to the finish when you have only just started) but equally there were those who paced themselves & walked most of the route. It's a flat run with amazing landmarks to see.

If you are thinking of trying your first 10k, I would definitely recommend this one. So, of course I was flying the AGSD–UK flag & raising money in the process (top tip - take your sponsorship form to a pub & wave it around when everyone has had a few pints - they are very generous!) - my vest had my name printed across it & I had email support from the other runners. And to top it all, I crossed the finish line with another AGSD–UK runner (David Burton) who I hadn't met before.

All in all, a lovely run in unusually sunny London raising money for such a worthwhile cause at the same time

Darren Josiah

I really enjoyed this run and my time was 43 minutes. The best thing about this event was being able to explore London at pace and be surrounded by so many people with the same goal. Very inspiring. The musicians that were dashed along the route gave the event some life and helped to distract from the pain of running that distance. The day was fantastic and we even managed to explore London and see the sights while we were there.

South Downs Half Marathon

17th June 2017
Many thanks to Adam Whitby who raised over £1330 for AGSD–UK. Adam was running in support of his nephew, Archie, who has Pompe disease. He says on his fundraising page:

Because Pompe is so rare, there is very little known of the disease and research to find new treatments and improve outcomes is vital. This is why, when I decided to run the South Downs Half Marathon, the choice of charity I wanted to raise money for was a no-brainer. I hope you will give generously to this very worthy cause and help the lives of Archie and others who suffer with GSD.

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!

Check out the venues at www.bigfunrun.com/venues/
Contact the AGSD–UK office if you would like a place in one of these runs.

London Marathon

Wrexham woman with rare muscular disorder makes history by completing London Marathon

The Wrexham Leader, Published 24 April 2017 I Article by: Geraint Jones and David Humphreys

A woman with a rare muscular disorder was all smiles after completing the gruelling challenge of the London Marathon. Deborah Corcoran, 49, was not going to let her battle with McArdle disease stand in her way of completing yesterday's marathon and she overcame the debilitating condition to conquer the 26.2 mile course. She is believed to be the first woman with McArdle disease ever to take part in the London Marathon.

McArdle disease, also known as glycogen storage disease type five, is a rare metabolic disorder which causes muscle pain in everyday activities and exercise.

As the elite and professional runners whizzed by, Mrs Corcoran, of Coed y Glyn, Wrexham, managed to walk the course in a time of seven hours, nine minutes. She described the elation of completing the route as "hugely satisfying". "It was tough but one of the best feelings," she said. "It was hugely satisfying. The last mile came and went and I'm so pleased to have done it for everyone. It was a real mental challenge to keep going."

Deborah hopes to raise £3,000 for the Association of Glycogen Storage Disease UK, which helped her after suffering a heart attack five years ago. She explained the difficulties of living with her condition. "It's a very rare disorder and it's very difficult because people aren't aware of it, so GPs and doctors in the hospitals don't know what to do with people when they have symptoms, so people aren't getting diagnosed because there's not enough awareness out there," Deborah said. The condition has been linked to heart problems.

"When I was a child I was quite lazy at school, I mean I got lapped at the 800 metres and carried home by one of teachers in the cross country race and I never got picked for any teams," she said. "Then, when I was 33 I got ill and was diagnosed with this muscle disorder. "You get tired very very quickly and unless you're aerobically fit, it's very hard to become aerobically fit with it, so it wasn't until I had my heart attack at 44 that I realised that I needed to do something. "Since then I've built up my strength, I walk a lot and just thought I'd do the marathon as a challenge not just for me, but also to raise funds because they supported me really well when I had my heart attack."

Some people with McArdle disease are wheelchair bound and some have a very short life span. Deborah said the condition affected her upper body strength, and she has to spend 10 minutes getting into a "second wind" when exercising to avoid muscle contractions or kidney failure. "There's a lot of people who struggle to put one foot in front of the other, but because I was reasonably young at 44 to be able to increase my fitness and take it steady to get to a position where I'm comfortable, I'm lucky. But a lot of people don't get to that position," she said. "I do have good days and bad days, but it varies in different ways and I manage to keep quite active and I think that helps a lot." Deborah has gradually built up her training, which eventually included a walk from Wrexham to Chester and back.

She said she had received an "amazing" response through donations, making £550 at a coffee morning in Wrexham and receiving a £250 pledge from an elderly man with the condition. "So many people have been so generous, even people I have never met, never spoken to," she said. Paying tribute to the supporters who lined the route, Deborah said: "I took supplies with me but I didn't need them because the people on the street were so supportive and helpful sending over sweets and water, it was great," she said.

Lyke Wake Walk

June 2017

On June 10th, I and eight classmates from my schooldays made an attempt on the Lyke Wake Walk, a 40-mile hike across the North York Moors. Four of us aimed to complete the challenge in under 24 hours to qualify for a cute little pin badge like this:

The Lyke Wake Walk is a forty-mile crossing of the North York Moors from Osmotherley to Ravenscar following the line of the watershed across the moors at its widest point. The walk commemorates the practice – long since abandoned – of carrying the bodies of the dead from various parts of the moor to their final resting places.

We arrived at Rosedale Abbey on the Friday afternoon to enjoy the beautiful vistas of Rosedale and enjoyed a small reunion in the warm evening sun, before an early night. We awoke at 3am and drove off to the start near Osmotherley. The glorious red sky at 4:30am should have prepared us for what was to come, but we were in good spirits and marched on with enthusiasm onto the North Yorks Moors. The rain started by 5am and by 7:30 it was driving horizontally we were drenched and cold. After changing clothes, we carried on to the half-way checkpoint where we agreed to postpone the second 20 miles to the following day. The decision was not easy, but one of the group was almost hypothermic, another struggled with sciatica and all the enjoyment of the walk had been extinguished.

The next day was a beautiful day for a walk! We completed the second 20 miles, much of it alongside the second group of walkers, so finished as a complete group. We celebrated the whole event, reunion and walk, with a meal and fine ales in Rosedale's White Horse Farm Inn.

So, I didn't earn my little pin badge, that's still on my bucket list, but we raised nearly £3000 for AGSD–UK and the reunion rekindled friendships from over 40 years ago.

Allan Muir

Buriton Art Trail

AGSD–UK was invited to provide cakes and refreshments at an annual Art Trail event held in the village of Buriton, Hampshire. Barbara Muir organised the baking and drinks on our behalf and raised an impressive £360 for the charity.

Ride Run or Walk in Richmond Park

Sunday August 13th 2017

AGSD–UK member, Matthew Arthur, is organising a day in Richmond Park to support glycogen storage disease research. Matthew is following in the footsteps of his father, Tim Arthur, who organised the AGSD–UK Vietnam Bike Ride in 2003. Matthew has GSD III and is passionate about promoting further research for his and other conditions.

Whether you are an endurance athlete or struggle with GSD yourself, this event is designed so that you can meet your own personal challenge within a safe and supportive format, even electric bikes are welcome.

Check out Matthew's fundraising website for more details: www.gsdfundraising.com

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

Blister Sisters Challenge

Fundraisers:
Kastur Pindolia, Pushpa Meghani, Sangita Kerai
http://uk.virginmoneygiving.com/team/Blistersisterschallenge

Message from Kas:
I am walking 100km with two good friends of mine Pushpa & Sangita. This a personal challenge to test our physical and more so mental capacity to keep walking day and night continuously.

AGSD–UK is dear to me as both my children suffer from this condition and the association has always been there to support and advise when we really needed them. They support families, organise conferences and provide financial support where possible for families to get much needed mobility aids or help fund any treatment not available on the NHS and grants for research into new treatments.

Giro d’Italia Gran Fondo, Northern Ireland

Bill Corr

On 4th June, Bill Corr cycled the 70km Gran Fondo NI, a legacy event from the 2014 Giro d’Italia Big Start.

As well as exercising his McArdle muscles, Bill raised a very impressive £350 for AGSD-UK.

NHS Specialised Services for Metabolic Conditions

The article below highlights the difficulties experienced by one family living with a GSD. It is authored by Jason McMillan in his personal capacity as a parent living with a child with a GSD and is aimed at stimulating discussion and providing feedback to the NHS.

Over the years we have heard a number of stories of late diagnosis, misdiagnosis and mismanagement of glycogen storage diseases. Awareness of GSD's and the appropriate care individuals is still far from perfect especially as each patient may require very personalised treatment. The UK is very fortunate to have specialist treatment centres throughout the UK with specialist teams of dedicated medical consultants, dieticians, physiotherapists, clinical nurse specialists and diagnostic laboratories. Medical teams are working towards an improved understanding of GSDs and a consensus on the management of the disease is constantly under discussion at medical conferences and through emerging rare disease networks. As a result of these advances many individuals are receiving much improved care and management of their GSD than was the case just a decade ago, though clearly there is still room for improvement.

Opinions in the article below are the authors own and do not reflect the official view of the Association for Glycogen Storage Disease (UK) Ltd.

Family Experience of NHS Care for GSD 1b

Have the Wheels Fallen Off NHS Care and Diagnosis of GSD?

Jason McMillan, father of Catherine

We have not come to this conclusion lightly or purely through our own poor experience of care of our daughter with GSD1b (The deficiency impairs the ability of the liver to produce free glucose from glycogen it causes severe hypoglycaemia and include lactic acidosis, Hyperlipidaemia, Hepatomegaly and other liver problems plus in 1b Infection risk due to Neutropenia), it is through seeing the treatment offered in France, Spain, and the USA. It is also from attending a few GSD Conferences in other countries and also spoken to doctors who themselves live with GSD or have children with the condition, all of whom seem bewildered by the dated approach taken by doctors within the NHS.

We have seen, first-hand, patients doing much better elsewhere than those in the UK and I believe NHS doctors have a lot to learn from those with significantly more experience, particularly the doctors in the USA.

The major issues with our experience are;

- The inability of the NHS to diagnose her, despite clear symptoms
- Their blasé attitude once the problem was clear
- The frustrating interactions and treatment provided once Catherine was correctly diagnosed
- The stubbornness shown by the NHS doctors to work together or with experts in the field
- The delay in Catherine's diagnosis which has left us with concerns for her long-term health and future care

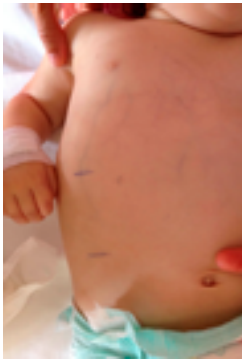
All of this has led to our seeking treatment from outside of the UK.

I am Catherine's father, Jason McMillan (British), her mother, Maria Jesus who is from Spain. Catherine was born in the UK 5 years ago.

We are attentive parents who have never missed any check-ups and were quick to seek help if she was ill or if we had any reason for concern. My wife Maria had noticed the differences between Spanish and UK healthcare and during our visits to Spain would always arrange check-ups for the children. When Catherine was 6 months-old a doctor in Spain noticed, her liver was quite large and suggested we should have this checked by our doctor in the UK. However, the UK doctors and midwives said there was no cause for concern and did nothing further was done.
By the time, she was 1-year-old we began to be very anxious as saw her weight and height plateau and her stomach seemed to protrude a lot more. Doctors ignored these things again.

This was further compounded when an owner of our local nursery pointed out that Catherine seemed disproportionately small with a large tummy, and recommended we have this checked by a doctor. Again, these concerns were dismissed by our Doctor.

Symptoms of GSD such as large nose bleeds were also dismissed at check-ups even after calling 111 for help on occasions. The discoverer of GSD1 disease (Von Gierke) describes frequent nosebleeds before the deaths, of children who had enlarged livers. The symptoms were ignored once more by the doctors.



Catherines liver size 28 Aug 2014

In August 2014, we went for a Holiday to France. On the night of the 21 August, Catharine was sick with high fever and we decided to get her checked out at the local Hospital, the CHU Hopitaux de Rouen. They quickly assessed that she had a serious health problem based on her protruding stomach, thin arms and legs and due to her overall size. She was assessed by several doctors who were all very concerned with her condition. The same symptoms had been disregarded and ignored by the UK doctors

They carried out a huge range of tests and considered several possible diagnoses, which was traumatic and distressing. Within 48 hours of being admitted Catherine had a full-body CT scan, lumbar punch, bone marrow biopsy, MRI, X-Ray, Hepatic Ultrasound and every other test they could do. We had been to the UK hospital and doctors with the same symptoms and they had done nothing. We were in the French hospital for two weeks at which point they thought she may have HHV6 and strongly advised our doctor in the UK carry out a full blood test and check of her liver to ensure a correct diagnosis.

The French doctors seemed quite bemused by the treatment provided in the UK and the fact our doctor had not noticed how drastically oversized Catherine's liver was (we were told 11cm larger than average). (please see attached photos titled 'Catherines liver size 28 Aug 2014 at CHU Hopitaux de Rouen' of Catherine marking with the liver should be and where the liver was).

Upon our return to the UK, we provided all the documentation and advice from France, but the doctors seemed content to leave her diagnosis at HHV6 rather than carry out the recommended tests. She was hospitalised a few times for a few days after we insisted they should monitor her as suggested by the French doctors, but we continually seemed to come up against the paediatrician's egos and unwillingness to answer any questions. The doctors were uncooperative and unprofessional and refused to work together to care for our daughter. We were even told that 'it doesn't matter that her liver is big it's working and that was fine', and her regular nosebleeds were because of her picking her nose. Any simple search on Internet of enlarged livers and nosebleeds would've highlighted a liver problem which they should have followed up on.
On more than one occasion the blood test results were lost and had to be taken again, further delaying her appropriate diagnosis and treatment. Even when some of the results appear to show she might have Neutropenia, high cholesterol, and very high triglycerides these where all ignored again, even when Neutropenia was a clear red flag to symptoms for GSD 1b. We were continuously dismissed and ignored as troublesome parents.

In December 2014, we decided to go to the doctor in Spain out of sheer frustration, we were referred to Dr. Carlos Ochoa who provided outstanding care and attention to our case. Without his expertise and help, we feel we may still be without a diagnosis and our child could be dead. In his report, Dr. Ochoa wrote that this could be GSD as: *"Glycogenosis (these patients usually present symptomatic hypoglycemia that our patient has not had; the diagnosis requires genetic study and/or liver biopsy)"*. We were told at our Local Hospital, that what Dr. Ochoa prognosis' we're not possible.

Eventually through continuing persistence and Dr. Ochoa's report we were finally able to get referred to a London hospital's liver ward for a biopsy. This was an extremely emotional experience. As we would later found out that Catherine could end up in this ward waiting for a transplant herself if we did not find some better professionals to care for her.

We were then transferred to one of the main children's hospital in London dealing with metabolic disorders where we finally got the diagnoses that it was Glycogen storage disease. At this stage, she was nearly 3 years old.


Catherine in March 2015

GSD is a very hard disease to live with and at that time, on the UK care plan, Catherine required a feeding tube down her nose into her stomach, this made us live in constant fear of a tube leaking and her going into a coma or of her becoming tangled up in her tube during her sleep. We put a bed for her next to ours and would wake up several times in the night to check on her.

On the UK Care plan, there were no restrictions on eating fruit or having sugars or lactates. This was even with the knowledge that high sugars and fructose can damage the liver. We were not given or allowed to have a glucose checker. We seem to be in the dark and constantly concerned with what her sugar levels were. There was very little concern for accurate measurements of foods or of cornflour. There seems no concern about long-term issues with Crohn's disease and no use of probiotics.

We began researching her condition ourselves and found that doctors in the US and elsewhere used a significantly different treatment plan to the one Catherine was given. Our questions about this other treatment method were rejected and we were told this treatment was backward and provided less care and support. But Catherine was frequently sick on her UK care plan. So, we start to have our doubts that her treatment was appropriate.

In early October, we attended the AGSD Conference in the UK, where we met the world's leading authority in GSD Care. We came to our own conclusion about the different care plans and it seemed to us that the UK method was easier for the hospital to manage and reduce questions or queries and did not put patient care first. With the UK care plan, she was more likely to develop liver and kidney problems and very likely to develop digestion problems.

We have spoken to different doctors who themselves live with the disease and who have switched to the US method of treatment and had and have seen vast improvements in their health. But the UK doctors seemed to be refusing to acknowledge any other treatment options other than their own.

One night, we had major problems with Catherine's feeding tube which started to leak. We knew this was a dangerous situation for our daughter and could potentially result in her going into a coma or even dying.
This was the final straw for us and we decided then to seek out an alternative treatment method in Spain. We found a doctor who has GSD herself and as training from the US who had been on both treatment plans herself. We finally felt confident we could get the right treatment for our daughter.

Our lives have changed dramatically, and within a week of being on the new care plan, Catherine no longer needed the feeding tube. On the UK care plan, she was sick after only

having a small amount of cornflower and we were told by UK doctors that children with GSD would vomit quite regular because of the cornflour. But on the US plan, Catherine was taking a considerable amount of cornflour throughout the day without being sick.

What seems clear, is that on the UK care plan, with the overnight feed and the inaccurate cornflour measurements were sending Catherine into a rollercoaster ride with her glucose, which was making her nauseous and sick. It would seem this rollercoaster ride on the continuous feed they could also be the long-term consequence of liver and kidney damage.

Equipment required for a few days with UK care

All required for US care plan

On the US care plan, she can sleep in her room with her sister, she can play freely without fear of getting tangled in tubes and is now more alive than ever. Her hair has started to grow and her liver is now just 1cm under her ribs and her blood labs have vastly improved something that we were told in the UK could not be achieved. Now that she isn't dependent on feeding tubes, her quality of life has vastly improved.

She is no longer at risk of infection from feeding tubes. Under this plan, we are encouraged to monitor her glucose levels, which was strongly discouraged under the UK plan. There is so much more freedom in our lives to travel and move around (please see attached photo's titled 'Equipment required for a few days with UK care' and photo 'All required for US care plan')

The NHS doctors seemed to have completely the wrong outlook on her condition, they appeared to take for granted the idea that she would have complications later in her life and would deal with them then, rather than taking preventive care. On the other hand, the doctors all around the world advised that with the correct treatment complications would be significantly reduced.

The major issue with UK care plan seems to be the egos and the unwillingness to take outside advice or think outside the box. Can the doctors change? It seems they're not going to change of their own accord.

If you have doubts about your care plan, it is time to put politeness to one side and time to act. Write letters to your doctors and your consultants dealing with your GSD. Asking why you're not been given the full choice of possible different care plans and ask why they do not wish to talk to other doctors. Why are they not speaking to doctors who have GSD themselves?

The difference in treatment and quality of life is day and night, and it is important to ensure other children get the treatment they need and do not have the same experience that we have had.

My email address is jasonjmc@gmail.com. Feel free to contact me if you have any questions.

Catherine now

McArdle news

Walking with McArdle's courses

The name is a bit of a misnomer as these weeks are about so much more than walking. Many people attending have never met anyone else with McArdle's, so sharing experiences, comparing notes and interchanging ideas are high on the list of benefits. The courses are really about the techniques of coping with McArdle's on a day-to-day basis, with the walks being the main vehicle for doing that. We are just about to add a new short video to our YouTube Channel with testimonials about people's experiences on the courses.

North Wales

2017 will be our seventh year of running these courses, so about 60 or more people have attended. They have come from 15 countries and have ranged in age from 11 to 69. This year we are back in North Wales based at Capel Curig in the very comfortable Bryn Engan converted farmhouse, with single en-suite rooms.

Children & Parents

For the third year running we are running the very successful "Children & Parents" event for 3 days prior to the course, at the same venue. Full to bursting this year, it is amazing to see how much the young people can take on board at such an early age.

New opportunity in California

This year a new course is being run in California, USA, for people with McArdle's (GSD5) and Tarui's (GSD7). It is just 5 days and would fit well with a more extended holiday over there. The dates are 13 to 17 October (still great weather in California!). If you are interested please contact the organiser for a flyer. jeremy.michelson@iamgsd.org.

Launch of IamGSD

A new patient-led organisation was launched at the IGSD2017 Conference in The Netherlands in June. The International Association for Muscle Glycogen Storage Disease (IamGSD) will initially be very focussed on McArdle's and Tarui's, but will gradually expand to cover all the muscle GSDs except Pompe (GSD2) which already has its own International Pompe Association.

With eleven trustees from around the world, the new Association aims to enhance the quality of life of all those affected by muscle GSDs.

The IamGSD objectives are to:

1. **Raise Awareness amongst the medical profession, schools, sports clubs, and other relevant groups.**
2. **Provide support for patients, organisations and medical professionals.**
3. **Advocate the patient viewpoint and needs to governments, patient organisations and medical professionals.**
4. **Disseminate standards and best practice.**
5. **Contribute to the planning of research projects and to support and assist those projects.**
6. **Facilitate communications between organisations, especially internationally**
7. **Work towards reducing the average age of diagnosis to age 10**

The web site is being developed and should launch in the next few months. It is planned to eventually have information in about 6 languages.

McArdle's research coming to fruition

A number of significant research projects are about to report results. In the next few months we should hear about the Sodium Valproate trial (London and Copenhagen), the Triheptanoin trial (Copenhagen and Paris) and the initial ketogenic diet trial (Germany). There will be a report on the Sodium Valproate trial at our AGSD–UK Conference over 28/29 October.

The two exercise related trials at Brunel University are still active, one looking to do a second phase and the other still in the initial planning stages. If you have already volunteered for the latter one, you will be contacted once the team are able to recruit participants.

This is by far the busiest time ever for McArdle's research, so a very exciting period with the prospect of good things coming up.

Ketogenic diet

The joint position statement on the ketogenic diet between AGSD–UK and the McArdle Clinic (circulated last October) has started to have an impact. If you attend the clinic and have decided to try this developmental diet, do please tell the clinic by letter or email. We hope that in time this will facilitate the collation of data and feed in to research projects which are now being planned. Of course, in the meantime keep sharing your experiences on the “Ketosis in McArdle’s” Facebook group.

McArdle's numbers up again

We have just reached 303 people currently diagnosed with McArdle’s in the UK. And we are succeeding in getting more people diagnosed in childhood, with three more children diagnosed in the last 12 months. This is great news for those young people, as they can get the correct advice from an early age and thus avoid the worst of the problems later. Over 190 of those UK patients manage to attend the McArdle’s Clinic in London.

Pompe Support Team

After the great success of the PST publication "101 Tips for an easier life with Pompe Disease", They are now very busy working on a number of smaller but very important publications. First will be a Medical Alert Card that will be printed very soon and will be available to the UK Pompe community. The second will be a booklet providing a comprehensive "Medical Overview of Pompe Disease" that will be given to GPs and other non-specialist healthcare professionals to help them understand the condition and provide improved care for their clients. This is currently at its first draft and is still to be reviewed by our medical advisors.

Research update

Research Updates

There were many papers presented at the recent International IGSD Conference in Groningen. Abstracts of the research presented are available in a downloadable book from the conference website which covers many aspects of many Glycogen Stoarage Disease: http://igsd2017.com/program/

Below are examples of some of the research discussed, but please refer to the website above for details of all the discussions and research presented:

Development of renal cysts in patients with glycogen storage disease type I
Philippe Labrune, APHP, Hôpital Antoine Béclère, Centre de Référence Maladies Héréditaires du Métabolisme Hépatique,
Clamart and Paris Sud University, France.

Dietary management of ketotic GSDs
Dr. Bhattacharya, K., Sydney Childrens Hospital network, Westmead, Australia

Renoprotective effects of fenofibrate in glycogen storage disease type i in mice
Laure Monteillet – Lyon

New generation biopharmaceuticals for the treatment of glycogen storage disease TYPE III (GSDIII)
Doriana Triggiani – Rome

Inhibition of glycogen synthase ii with rnai prevents liver abnormalities in mouse models of GSDs
Chengjung Lai – Cambridge

In vivo gene therapy for monogenic diseases
Federico Mingozzi, Paris
INSERM U951, Genethon, and University Pierre and Marie Curie-Paris 6, France

Gene therapy for glycogen storage disease type Ib
Janice Y Chou, Bethesda and Joon Hyun Kwon
Section on Cellular Differentiation, Eunice Kennedy Shriver National Institute of Child Health and Human Development, Bethesda, MD 20892

In Vivo Genome Editing to Achieve Integration of a Glucose-6-Phosphatase Trans-gene in Glycogen Storage Disease Type Ia
Hye Ri Kiang, Elizabeth Brooks, Songtao Li, and Dwight Koeberl
Division of Medical Genetics, Department of Pediatrics; Duke University School of Medicine, Durham

Valerion Initiates VAL-1221 Dosing in Patients with Pompe Disease

Potential New Treatment Option Designed to Improve Glycogen Clearance for Better Patient Outcomes

Top-line Clinical Data Expected in the Fourth Quarter of 2017

CONCORD, Mass., July 11, 2017 /PRNewswire/ --
Valerion Therapeutics, a clinical-stage biotechnology company that specializes in the development of therapies for orphan genetic diseases, today announced that it has initiated dosing in a Phase 1/2 clinical trial evaluating VAL-1221 in patients with late-onset Pompe disease. VAL-1221 is a novel fusion protein that combines Valerion's antibody-mediated delivery technology with recombinant human acid alpha-glucosidase (rhGAA) to uniquely target both lysosomal and extra-lysosomal glycogen in the cytoplasm through enhanced intracellular delivery to affected tissues. By leveraging two uptake mechanisms, rather than one, VAL-1221 offers the potential for improved glycogen clearance and better patient outcomes. The Company expects to report top-line data from the Phase 1/2 study in the fourth quarter of 2017.

"After a brief period of modest improvement with currently approved treatments, nearly all patients with late-onset Pompe disease resume their downward trajectory in skeletal muscle and respiratory function in association with accumulation of glycogen in the cytoplasm," said Deborah Ramsdell, CEO of Valerion. "While current Pompe disease therapies are directed only to lysosomal glycogen, VAL-1221 has demonstrated in preclinical studies the potential to uniquely target and clear both lysosomal and extra-lysosomal glycogen in the cytoplasm. We look forward to reporting top line data from this study in the fourth quarter of this year."

"The Association for Glycogen Storage Disease (UK) is excited by the potential benefits promised by this novel therapy, to improve the lives of individuals and families living with Pompe Disease," said Allan Muir, Development Director, The Association for Glycogen Storage Disease (UK).

"Pompe is a very debilitating disease often leading to a loss of limb mobility and breathing function so serious as to require the use of wheelchairs and full time respiratory support, in addition to terrible and constant pain. Pompe dramatically limits quality of life in most patients, and in its most severe forms, can even be fatal," said Maryze Schoneveld van der Linde, Pompe patient. "Significant unmet need exists for a variety of new therapeutic options that offer the potential to treat all Pompe patients."

Amicus announce positive Pompe phase 1/2 functional data.

On May 15 2017, Amicus announced positive preliminary functional data from the ongoing clinical study (ATB200-02) to investigate ATB200/AT2221 in patients with Pompe disease; this is a global study of 19 individuals, including ambulatory and non-ambulatory, ERT-naïve and individuals switching from Myozyme. The data is from those study participants who completed six months of treatment with the investigational ERT together with the pharmacological chaperone. The positive preliminary data announced include: safety, tolerability and pharmacokinetics (PK); pharmacodynamics (PD) on muscle damage and glycogen biomarkers; motor function, pulmonary function and muscle strength.

For further information, you can visit the Amicus corporate website to see the full press release. A conference call and webcast was held on May 15. The slide presentation accompanying the webcast is still available at http://ir.amicusrx.com/events.cfm.

Extract from Viking Therapeutics press release

SAN DIEGO, March 21, 2017. Viking Therapeutics, Inc. a clinical-stage biopharmaceutical company focused on the development of novel therapies for metabolic and endocrine disorders, today provided an update on its clinical pipeline and other corporate developments.

New program initiated to evaluate VK2809 for the treatment of glycogen storage disease type Ia (GSD Ia)

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. Initial results from an in vivo proof-of-concept study showed that treatment with VK2809 produced rapid and substantial reductions in liver triglyceride content, liver weight and liver weight as a percentage of body weight compared with vehicle-treated controls. Mean liver triglyceride content was reduced by more than 60% in VK2809-treated animals relative to vehicle-treated control animals, while average liver weight was reduced by more than 30% vs. controls. Complete results from this study, which is being conducted under a sponsored research agreement between Duke University and Viking, will be presented at a future scientific meeting.

Funding secured to advance GSD Ia program

In February 2017, Viking entered into an agreement with a dedicated healthcare investment fund to support the initial clinical development of the company's GSD Ia program. With this funding, the company plans to file an Investigational New Drug (IND) application for VK2809 for the treatment of patients with GSD Ia and initiate a human proof-of-concept study in the second half of 2017.

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

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