Glisten

Glycogen Storage News

30th Anniversary Issue

June 2015

Volume 14 Issue 1

Contents

Chairman's Update

Michael Porter, Chairman

The time since our conference last year at Wyboston Lakes seems to have flown by so quickly. Last year's conference was our most successful in terms of attendance and the feedback received was very positive. I hope that the high attendance numbers continue at this year's conference particularly as we will be marking the 30 year anniversary for the charity.

I would like to place on record thanks to one of our trustees Wendy Bascal who has resigned due to external pressures on her time. Wendy has been a long time supporter of the charity committing lots of her personal time and experience in supporting GSD sufferers and helping AGSD-UK develop and grow.

The trustees last year announced that it was planning to submit a substantial bid to the Big Lottery Fund. The aim of the bid was to get funding that would support the charity in recruiting three full time Specialist Care Advisors. This would help alleviate pressure on our volunteer Type Co-ordinators and also allow us to expand the level of support that we can currently provide to GSD patients and their families/carers. The bid was formally submitted in January and we heard back around 8 weeks later that in this instance we had not been successful. No feedback or reason is given for the decision. The number and value of the submissions to the Big Lottery Fund far exceed the amount of funds available so we were always going to be competing with a large number of other equally worthy bids. It does not preclude us from making future submissions and the trustees will look closely at making a revised submission at some time over the next 12 months.

Notwithstanding the decision from the Big Lottery Fund we have managed, with the continuing support from Genzyme, to recruit a Specialist Care Advisor for the Pompe community. This post replaces the role that was undertaken by Joan Fletcher a NHS employee who worked out of Manchester Children's Hospital. The new role which has been filled by Jane Lewthwaite is directly employed by AGSD-UK and she has already made an excellent start in supporting the Pompe community. Please join me in welcoming Jane on board.

Work continues with getting our website updated and our IT infrastructure onto a more secure footing and as presented at the conference we will continue to target charitable trusts and foundations for additional funding. We still of course

still need you all to carry on with your excellent fundraising activities as this is crucial lifeblood to AGSD-UK. Allan Muir will provide further information on fundraising within this edition.

Finally there is a lot of coverage right now on the forthcoming EU referendum and what would be the likely impact of an in/out decision. As individuals we will all have our view and be able to vote accordingly. However the charity has an obligation to protect the interests of our members and therefore the trustees will carefully look at any potential impact on the UK GSD community – i.e. cross European research/ collaboration and voice any concerns accordingly.

Thank you all for your continuing support and I look forward to seeing those of you who are able to attend at this year's conference.

Development Update

Allan Muir, Charity Director

First of all, I must apologise for the lateness of this publication; in a way we are victims of our own success: The AGSD-UK is recognised globally and as such we receive many invitations to collaborate and requests from overseas patients for support. As a result, I and our small team of volunteers sometimes struggle to meet all of our objectives.

It has long been my dream to employ staff to support the needs of all GSD families and individuals. So I would like to join the Chairman in welcoming Jane Lewthwaite into the role of Specialist Care Advisor (Pompe). Jane has already met with many of our Pompe community and has joined professional networks of other support organisations to gain from their experience. I am sure that the experience we gain, through Jane's employment, will be crucial in giving other funders good reason to provide financial support to us, to enable us to employ additional team members; staff needed to support the whole UK GSD community.

Jane brings much knowledge (e.g. social services and benefits) that I and other volunteers can learn from so that we may offer improved guidance to people living with any GSD.

From her professional work with Age UK and Children in Need, Jane also has a great deal of relevant experience dealing with fundraising and grant applications so she will be a great help in that important area of our work.

And speaking of fundraising, we have received a small grant to enable us to employ a well-recommended fundraiser, initially for one day per week over three months. Vanessa Casey is well known to one of our members and will be working only on applications to Charitable Trusts and Foundations for grants to help us cover our management and office costs and also to find funding for the Specialist Care Advisors we would like to recruit.

Organising the conference venue for October this year has been fraught with difficulties lately. Our preferred option withdrew due to problems with their building schedule. We have finally chosen Tortworth Court Hotel, Gloucestershire; we have had to make a few compromises but the staff are very keen to provide a great experience for all delegates, whatever their needs.

The theme for this year's conference is "Back to the Future" to allow us to reflect a little on the past and spend some time scanning the horizon; so again, if you have any ideas that might reflect that theme, do let me know or bring them along with you. Fancy dress is not compulsory!

Moments at the conference will be a commemoration of all our achievements over the last 30 years but it will also be a grand occasion to speculate on advances we hope to witness in the next few decades. If you are planning to come along to Tortworth Court in October, please bring along your most treasured memories of the AGSD-UK and thoughts and hopes for the future. For example, if you have photographs or newspaper articles we would love to make a large display for everyone to enjoy.

We also plan to have space at the conference for everyone to write or draw their vison for the future of the AGSD-UK or for any aspect of living with a GSD. So please do give it some thought and bring along anything that you feel would add interest to the occasion.

I look forward to seeing you in October,

Allan

Staff Update

Jane Lewthwaite, Specialist Care Advisor (Pompe)

It is only a few weeks since I started in the new role of "Specialist Care Advisor (Pompe)" within AGSD-UK. The main purpose of this post is to provide non-medical support for people with Pompe, so it is quite a broad remit. I do not have Pompe so it will be doubly important to listen to members, appreciate their needs and understand their expectations for this new role.

It is crucial that my work supplements and enhances the huge amount of voluntary effort already made by members. Hopefully there will be chances to meet and communicate with as many people as possible, fairly soon. I am already setting up meetings with other Care Advisors and Support Workers to see if they have good ideas to share. I am also starting to gain an understanding of the mix of medical, social, private and public services available.

Perhaps the most important thing I can offer is time. Time to listen and time to spend on locating solutions for people with Pompe and their families. 'Advice' has many aspects and I really want to make sure that my work is adaptable for each person's needs and wishes.

Please get in touch with me if you have any comments about this new role. Suggestions and ideas are always welcome. I am especially keen to hear from people who have queries, unmet needs, or who would like to make use of my time. If possible I will visit you, we can arrange to meet either at your home or another suitable venue. We can talk by phone, email, Skype or Facebook, whichever suits you. Generally speaking, I work Monday - Thursday but this is very flexible.

Office : 0300 123 2792 (9-5, Mon to Thurs)
Mobile : 07484 055334
Facebook : jane-agsduk

Annual Conference – Change of Venue

Tortworth Court Hotel, Wotton-Under-Edge, South Gloucestershire
October 15th and 16th, 2016

This year's 30th anniversary conference was to be held at the Wokefield Park, however they have now cancelled our booking due to delays in their building work.

That's the bad news; the good news is that we have been given alternative accommodation at the very grand Tortworth Court, plus all delegates receive 10% discount on the Peels Spa* so I expect places to be booked up very quickly!

Given the late notice and options available, we did need to make a few compromises. The location is not ideal, being some distance from International airports, and the disabled facilities are not perfect, although we are working with the hotel to make the conference accessible for all who wish to attend. There are only five "accessible bedrooms" (but with step-in showers) so if you require one of those please please book early so that we can discuss your requirements well in advance. There is an accessible wet-room in the Peels Spa of which we will have sole-use.

*Peels Spa: 12m x 7.5m swimming pool, fully equipped gym, sauna, steam room, spa bath and beauty treatments.

www.tortworthcourthotel.co.uk

Annual Conference – Venue Directions

Directions

From the North:
Leave the M5 at Junction 14.
Follow the B4509towards Wotton.
On the B4509, go up the hill and take the firstright into Tortworth Road. After half a mile, the hotel is on theright hand side up a long drive. The hotel is signposted once you join the B4509.

From the South:
Leave M5 at junction 14, signposted to Dursley. At the T-junction turn right onto the B4509 and follow directions above. It is recommended that you drive to reception to check in. You will then be directed to the nearest and most convenient car park to your accommodation.

An area closest to the hotel accommodation will be reserved for blue-badge holders.

Airports

Bristol Airport 24 miles, Birmingham International Airport 83 miles.

Rail Links

Bristol Temple Meads 17 miles
Bristol Parkway 11 miles
The closest main line station to Tortworth Court is Bristol Parkway, approximately 30 minutes away. For timetable and information see www.nationalrail.co.uk.

Taxis

The taxi firm used by the hotel is **A K Taxis** who specialise in airport and train stations runs.
Contact: Tony Wheeler,
Phone: 01453 842673
Mobile: 07970058112
Email: timkwheeler@yahoo.co.uk

Bristol Parkway Taxis charge £30 – £40 so it may be advisable to try to share a 4-seater with others. They do have one taxi with an electric wheelchair lift, but this may need to be booked well in advance.
Telephone: 0117 254 1111
www.bristolparkwaytaxis.co.uk

Other taxis are also available, so please contact the office if you have special requirements.

Annual Conference – Programme

Back to the Future

The theme of this year's conference will allow us to reflect on the growth and work of the charity over 30 years and to look ahead to the next 30.

We are currently in the planning stage of the conference; so if you have any interesting ideas to make it a really memorable event, please do contact the office.

On that point, if you have any AGSD-UK memorabilia, photographs or recollections from past events (fundraising, conferences, meetings), please do share them with me, or failing that, bring them along to the conference in October.

Let's all work together to make 2016 a significant milestone in the history of the AGSD-UK, something to look back on in 30 years' time perhaps...

The Conference will have the usual structure of general topics and GSD-specific workshops. Details of these will be added to our website as they develop.

Conference Registration

Online registration is available at: www.agsd.org.uk

We would encourage you to arrive during the morning of October 15th, as this will give you time for networking, to get to know other members and to meet up again with old friends.

The Conference will commence on Saturday 15th October at midday with registration and lunch. During the afternoon there will be the usual eclectic mix of speakers and workshops plus a few surprises. The day will finish with a Gala Dinner, which will start with a drinks reception at at 7.00pm.

The event will continue on 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.

Annual Conference 2016

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 centre, and AGSD-UK does subsidise the cost of accommodation. Most rooms will accommodate up to 2 adults plus one child but families of 4 may use the larger suites and there are adjoining doors between some rooms. Book early to ensure that a room will be available.

Wheelchair Accessible Rooms

Please only ask for an accessible room if you really need it; there are only 5 rooms available at this venue. If you need any aids such as bath seats, raised toilet seats (with arms) or grab-handles, please contact us and we will make arrangements with the hotel. The Peels Spa has a wet-room for which we will have exclusive use, so you can shower in there if your own bathroom is not ideal.

Residential and Day Delegate Packages

Please indicate on the form whether you will be registering as a residential delegate or a day delegate.

Mini-bars – Fridges

Unfortunately, the hotel bedrooms do not have mini-bars/refrigerators, so please let the office know if you require a fridge for children's special feeds or medications.

www.tortworthcourthotel.co.uk

GSD conference 2015

GSD conference 2015 – GSD I

David Weinstein

David has been attending the AGSD UK conference for last 13 years, he is currently conducting a trial on dogs for enzyme replacement therapy for GSD. David got involved and interested in helping GSD sufferers after being invited to the US conference and had a conversation with a parent whom informed him the only way for a sufferer to survive was through a liver transplant. He could not believe this was so.

Latest developments from the US:

- Blood and saliva can be used to diagnose GSD
- Understanding complications and treating appropriately so no limitations to quality of life for patient.
- Complications from GSD 1 can be avoided by having good metabolic control thus possibly reducing brisk of kidney disease, hepatic adenomas, osteoporosis, anaemia, spleen damage ect.
- In America some of his GSD 1 patients have had successful pregnancies by having good metabolic control with monitoring. Some his patients have had 4 babies with both mother and baby being well.

GSD diet USA

Dr Weinstein in Florida USA gets his patients to have good metabolic control by following a high protein, low carbohydrate diet with plenty of vegetables. In this diet all vegetables are ok but need to limit intake of sweetcorn and sweet potato due to their sugar content. Artificial sweeteners are ok except sugar alcohols like malitol/sorbitol , Splenda and stevia are ok. No fruits except avocado and berries but in small quantities. No Milk is allowed as it contains lactose and naturally occurring sugar but not good for GSD 1 patients, to supplement lack of milk all patients are on VIT D supplements, and a multivitamin supplement due to no fruit allowed in diet. GSD diet in UK is not so restrictive as no official studies have been carried out here in UK.

Enzyme replacement therapy trials

Dr Weinstein mentioned that they may have plans to start a trial with enzyme replacement therapy on humans potentially in 2016. This would involve a safety trial with low doses to see the enzyme is accepted by the body, it will be monitored very closely to see of any long term side effects and issues that may or may not arise. The trial will be carried out on patients over 18years of age.

Helen Mundy

Dr Mundy spoke about Corn starch dosing is individual to the patient, as over treating a patient can lead to having empty calories which may affect weight gain.

Patient/ Physician experience living with GSD type 1a

Monica Dambska (has GSD type 1a and is also a doctor on placement with Dr David Weinstein in USA)

Monica was born in Poland in 1985, and showed typical GSD symptoms of failure to thrive, constant diarrhoea, and her mum found that for some reason giving Monica glucose was helping her. Monica eventually got diagnosed at 8 months old. In 1998 Monica has a severe hypoglycaemic attack, she was in need of support and contacted AGSD UK and AGSD US.

Her family came to 2002 Nottingham AGSD conference and met other families and were told about continuous overnight feeds and good metabolic control. Even so her control was not so great as from 2007–2001 she was developing adenomas, high lipids, high uric acid and lactid acid levels.

In 2010 Monica returned to the AGSD conference and met David Weinstein who advised her on how to achieve good metabolic control by following a strict sugar free, dairy free diet with supplements to provide vitamins and minerals fruit and dairy provide. Monica uses cornflour to keep her sugar levels in control as well as a strict diet, she has tried using glycosasde but it does not agree with her so she only uses glycosade once a week to get a full night sleep, other nights its cornflour.

Sample of her daily diet living GSD

7.30 AM		25grms corn starch
8–9 AM		¼ slice of bread, 5-10g carbs with salad, meat and a small amount of hard cheese
10.30 AM		corn starch
12–1 PM	lunch	10–15grms carbs with lots of salad, meat
13.30 PM		33grms corn starch
3–4 PM	snack	nuts or 5–10grms of carbs
4.30 PM		33grms corn starch
6–7 pm	dinner	small amount of carbs lots of salad and meat
9.30 PM		22grms corn starch
11.30 PM		last corn starch or glycosade
3.30 AM		corn starch

GSD conference 2015 – GSD III, VI, IX

By Caroline Calder

Wendy Bascal welcomed all to the Hepatic 2 workshop. Group of 20 present representing the Type III's, VI's and IX's (though Type VI numbers only reached 2!) Where possible the slides will be available on agsd.org.uk in the coming weeks.

Basics of III, VI, IX

Balancing science and application, Dr Urike Steuerwald from Hannover gave a great overview of basics of our disease, which in her words is not a disease more a 'genetic mistake'. This presentation took us back to the basics of where energy comes from, the sources and the uses. This lay the foundations to discuss sugars in more detail and how GSD patients react to them, including of course the challenges we all have in breaking down glycogen.

The presentation then covered what the body does in the absence of having glucose to run on, and how the body turns to protein. The breakdown in proteins causes a rise in ketones, which led very nicely to the presentation by Dr Helen Mundy and Tanya Gill later in the morning

Improving Exercise Tolerance in metabolic myopathies

Dr Nicolai Prisler, Copenhagen presented his research that he has found working with patients in Denmark. The presentation focused on Type IIIa. A common theme again was the increased use of proteins to enable exercise.

Ketone and GSD, Ketone Monitoring and GSK IV, IX outcome data

Taking the discussion on ketones further Dr Helen Mundy and Tanya Gill (from Evelina Children's Hospital) presented to us the science of ketones – what they are, where they are produced and a view of whether they are good or bad (to which the answer is, it depends!). Following clarity on the important on ketones as a reflection of how successful our bodies are at providing the right energy, ketone monitoring was discussed. The Evelina hospital wanted to provide ketone monitors to their patients to allow for home monitoring, which they felt would give more realistic results, reflected actual day to day levels. They tested two monitors and found the Nova monitor more successful especially at the lower end of the scale where GSD patients should be monitoring.

Expert Panel

Chaired by Dr Uma Ramaswami. Panel members: Dr Weinstein, Dr Steuerwald, Dr Munday, Diane Greene. All hepatic types attended this.

Questions were put to the team on gene therapy, the dosing of cornstarch, cornstarch vs glycoside, carriers and exercise.
The responses from the cross Atlantic teams for the vast vast majority were in agreement. The research on gene therapy is progressing with speed, with a potential for this to be available in Type 1a and 1b in the coming years. Research on Type III and Type VI is initiating. The advice on cornstarch and/or glycosade remains that is need to be individualised patient by patient. Exercise across all types is encouraged and beneficial. Carriers and research on the prevalence on symptoms continues and is gathering pace.

Dinner on the Saturday Night

Despite a wait for some we all enjoyed a great dinner, shared with great company and a pretty incredible magician!

Day 2

Dr Urike Steuerwald works with patients in Germany and the Faroe Islands. She has a significant amount of knowledge and real life examples. She shared with us all her knowledge and took our questions across the morning – the topics focused on were: metabolic control, nutrition and treatment, exercise and, emergency plans in GSD Types III, VI, IX.

Three of Dr Steuerwald's presentations are posted on our website with links given in the GSD III, VI and IX sections.

In combining metabolic control and nutrition it was explained that with a controlled diet we can prevent low sugars, protein breakdown and high cholesterol. With this the liver can be maintained at lower size and with low risk of damage to the liver. To achieve a healthy diet the following is recommended –

- Many small meals
- 30 grams of carb per meal
- No more than 5 grams of sugars per meal
- High protein diet (Type III 3-4 gram per kg weight, Type VI and IX 2-3 gram per kg weight)
- Bedtime snack
- Uncooked cornflour

She shared a very helpful slide outlining what 15 grams of carbs is across a variety of foods. We also had a practical test which had us check a variety of food packets for the nutritional information.

We took the discussion on protein further and looked at protein sources and discussed protein supplements, shakes, powders. The advice was to check with the manufacturer for any protein supplements whether the product contains steroids, which is not always stated. Another piece of advice in order to make the carbohydrate intake as effective as possible, was to cook pasta for 2 minutes less than the packet suggests, and even rinse under cold water after cooking. This will slow down the breakdown of starch pre-ingestion. Supplements, exercise, alcohol intake, sleeping in, illness and the impact/advice for GSD patients were also crammed into what was an incredibly informative morning.

The final piece of advice was to have a medicalert bracelet/necklace made. This should display: Name, Hypoglycaemia, IV Glucose 10%, No Glucagon

GSD conference 2015 – McArdle

By Andrew Wakelin, McArdle Coordinator

We had a very lively series of workshops for McArdle's. On the Saturday we were packed with professionals and on the Sunday we had more reflective time to ourselves.

Saturday

We opened with introductions including a guest from Malta, Anna Sultana, who we feel sure is the only McArdle person on the island.

Dr. Ros Quinlivan told us about new developments at the McArdle Service. There are plans to have "patient information days" three times a year for small groups of new patients; and a new type of management clinic to give more intensive support to those who are worst affected. In the past year four children had been diagnosed, whereas normally there might be one every few years. With currently five diagnosed children in the UK, the plan is to hold an annual clinic day for children under the auspices of Great Ormond Street Hospital. It will of course also be an opportunity for parents to meet.

Dr. Karen Madsen from the Copenhagen Neuromuscular Centre in Denmark, gave a presentation on the current trial of Triheptanoin which is on-going in Copenhagen and Paris. Triheptanoin is a type of oil which can be taken as a dietary supplement, although it is licenced as a drug. They are looking at whether this supplement can enhance exercise capacity or endurance or reduce McArdle symptoms. It has been beneficial in trials in fatty acid oxidation conditions. The oil is odourless and tasteless. The dose will provide 25%-35% of the daily calorie requirement and therefore the diet needs to be low in sugar and fat to compensate. There are about 25 patients involved and results are expected by the end of 2016.

Andrew presented the findings of the survey on the ketogenic diet, on behalf of Stacey Reason from Canada. He explained the benefit of nutritional ketosis for people with McArdle's. The approach involves very tight restriction of carbohydrate and much more fat in the diet. People are reporting success with around 10% carbohydrate, 25% protein and 65% fat. 80 people had responded and 49% had tried the ketogenic diet. 73% felt that they were in permanent 'second wind'. 85% reported improvement in daily symptoms and 76% reported an improvement in exercise tolerance. He also reported on the workshop organised in New York – see 'McArdle's News' in this issue.

Our last session on the Saturday was our 'Expert Panel' with Dr. Ros Quinlivan, Dr Jatin Pattni, Charlotte Ellerton and Dr. Renata Scalco - all from the McArdle Clinic in

London. Questions addressed included: sugar before exercise, statins and cholesterol and management techniques for stress.

Sunday

On Sunday, Andrew gave an update on patient numbers and support issues. We then had patient's own stories from those in the room, which helped us all to understand the breadth of issues that can be involved.

Dr. Renata Scalco then gave an update on the Sodium Valproate trial. This existing, low cost drug is used for other conditions. In animal models it has been shown to switch on the brain version of phosphorylase in skeletal muscle. That isoform of the enzyme normally only reactivates in a muscle that is recovering from an injury. Fifteen patients are included in the study, 8 in London and 7 in Copenhagen. Each participant will undergo a process of screening, induction, muscle biopsy, the drug trial period with exercise assessments, a final muscle biopsy and follow ups. All this will be over a period of approx. 8 months in total. At the time of the conference 5 patients had been recruited.

In our closing session before a farewell lunch, Andrew Wakelin gave a presentation about the "Walking with McArdle's" activities of the summer. There were three events. First was the annual walking course now in its fifth year and this time had three participants under age 18, including 11 year old Layton from the US who came with his mother Melissa and did really well. For the first time we had two people with GSD 7 (Tarui). In total there were fourteen people. Although the weather was bad, they had a great week. Next was the "Children and Parents" event, the first time that anything like that had been held anywhere in the world. And finally was the "Welsh 3000s" challenge when experienced McArdle walkers climbed 9 of the Welsh mountains over 3,000 feet. A recorded version of Andrew's presentation is available on the AGSD-UK YouTube Channel.

All told, a very successful two days of workshops. Thanks to all concerned – speakers and attendees.

GSD conference 2015 – Pompe

By Allan Muir, Pompe Coordinator

Celebrating a Decade of Patient Advocacy, Malcolm Johnson, Sanofi-Genzyme UK

Malcolm Johnson was one of the first Genzyme staff-members we were introduced to when I and a small Pompe team visited their Oxford offices in around 2004. Together we were making preparations for Myozyme to become a commercially available therapy in the UK. Myozyme was eventually approved in England in early 2006. So it was fitting that he should mark our decade of collaboration, and also the work of our Family Support Officer, Joan Fletcher. After working for the AGSD-UK for the last 9 years, Joan has now returned to her former role as Clinical Nurse Specialist within the Willink Unit of the Royal Manchester Children's Hospital.

After a fitting tribute from Malcolm, Joan was presented with a large bunch of flowers from Sanofi-Genzyme and a gift and card from the AGSD-UK.

Being your own Advocate. Ben Parker

Ben is a member of the Pompe Support Team (PST) and also a voluntary advocate with two disability charities in his local area. He helps people speak up for themselves and achieve things that they felt had too many barriers in the way for them to happen.

Ben said he has learned a lot through his experiences with volunteering, and of course living with Pompe on a daily basis, and he knows how hard it can be to get certain resources that would make his life easier. He also knows how difficult it can be to speak up assistance is needed. It is, he said, for some strange reason, always easier to help others than ourselves.

Over the last eight years of living with Pompe, Ben has learned a lot of useful information. In his words: "I have a new understanding of what it is to live with a chronic illness on a daily basis and I can empathise with people on a more level playing field. All of this can be invaluable to other people who suffer in the same way and every journey is better shared than kept to ourselves."

Ben feels he has to educate each and every one of the medical professionals he sees, meaning that he has to think about what he wants to say, what he needs to say and how he can make this process easier for both parties.

Ben always plans ahead of an appointment and advises people to:

» Write down questions beforehand, don't get into the room just to find you've forgotten everything!

- Think about the person you are seeing, what information you need to give them and what you need from them so that they can help you in the right way.
- Don't waste it trying to explain something that has no relevance to the appointment. Be precise and direct the conversation if needs.

Ben gave advice on a wide range of issues that affect the Pompe community, sadly too extensive to repeat here. He included GP visits and the many social services benefits that are available. Do contact Ben if you would like to discuss any of these points further.

Wrapping up, Ben gave a few pointers which he thinks are important to remember:

- Make professionals work for you in the right way and not just work around you.
- Keep pestering and bothering until they give you what you need or at least work together for a positive outcome.
- Use citizen's advice and local advocacy charities to help you achieve your aims.
- Don't be afraid to ask for help in all aspects of daily life.

Diet and Pompe. **Diane Greene, Royal Salford Hospital**

Diane is a Metabolic Dietitian at the Royal Salford and has developed some very effective dietary modifications for her Pompe patients. She began by describing some of the early evidence and recommendations (before ERT was available) which provided these general conclusions and recommendations:

- Progressive muscular damage may be slowed down through lifestyle changes based on a specific diet therapy and daily physical exercise.
- Dietary intake made up of proteins (25-30%), carbohydrates (30-35%), fat (35-40%), might be beneficial
- Proteins coming from meat, fish, egg, and dairy products are preferred (high biological protein); as they are rich in alanine, an amino acid playing a key role in glycose metabolism and, consequently, in the employment of glucose as a source of energy.
- Fats are recommended in the form of unsaturated fats: omega-3, which are mainly contained in oily fish and omega-6, which are mainly contained in olive oil, dried fruit and cereals. Try to reduce saturated fats within the diet.
- Carbohydrates to be reduced to 30- 35% spread out during the day. The "a little and often" rational consists in avoiding the build-up of glycogen on the one hand, and hypoglycaemia on the other.
- Complex carbohydrates such as wholemeal cereals, pasta, bread rice and legumes, and wholemeal pasta should form the basis of meals and snacks.
- Avoiding sugars and simple carbohydrates, but ensure the diet contains fruit (dried or fresh) and vegetables; aiming to increase the dietary fibre.
- Aim for a healthy Body Mass Index (BMI)

Diane showed preliminary assessments of patients who had undergone her dietary therapy, one who reduced her BMI and one who increased it; both had physical assessments that showed some improvement. However Diane issued the following health warnings:

- For most healthy people, a high-protein diet generally isn't harmful, particularly when followed for a short time.
- The risks of using a high-protein diet with carbohydrate restriction for the long term are still being studied. Some unmonitored high-protein diets restrict other nutrients that they can result in nutritional deficiencies which can cause problems such as bad breath, headache and constipation.
- Some high-protein diets include foods such as red meat and full-fat dairy products, which may increase your risk of heart disease.
- A high-protein diet may worsen kidney function in people with an existing kidney disease because your body may have trouble eliminating all the waste products of protein metabolism
- Normal recommended daily intake of protein for an adult is approx 1.0g Protein/kg
- ALWAYS DISCUSS INCREASING YOUR PROTEIN INTAKE WITH YOUR HEALTHCARE TEAM

Diane showed how her recommendation of 1.5 grams per kg of protein and daily energy requirement of 30 calories per kilogram could be achieved, breaking down foods into 6 gram protein "exchanges".

For example; for a 70kg person (11 Stone) they would require 70x1.5 = 105 grams of protein and 70x30=2100 calories each day to remain a healthy weight. 73 grams (70%) of the protein should come from High Biological Protein (meat, fish, eggs, dairy). Examples were given of foods and their protein content and other protein enhanced foods were mentioned

Finally, Diane gave the following advice:

- You need to consider all other micro and macro nutrients – vitamins, minerals
- Think about type and amount of fat in your diet
- Talk to your dietitian and healthcare team.

Pompe disease: Past, Present, and Future.

Dr Ralph Wigley, Enzyme Laboratory, GOSH

Dr Wigley started his talk with a historical look at Pompe, from its discovery in 1932; thereafter testing was done by clinical examination and looking at biopsy slides under the microscope. The Enzyme Lab was established at GOSH in 1959 and in 1966 tests began to develop assays to measure enzyme activity. After work by Drs A Reuser and A van der Ploeg in Rotterdam, and the development of Myozyme by Genzyme, a test was developed to measure enzyme activity in the blood. This test has been available at GOS since 2005.

At the present time the Enzyme Unit offers many tests for Pompe and other conditions, including both muscle and liver GSDs. Urine tests for a tetra-saccharide biomarker is available, to monitor the efficacy of ERT, and the CRIM status of infants can be determined to distinguish the severity of their disease.

The unit is now looking to the future where it hopes to be able to offer antibody testing very soon and it is also looking at a high-throughput multiplex assay
of dried bloodspots. They are undergoing a proof-in-principle study for mass spectrometry assay which, if successful may be applicable to new born screening.

In summary Ralf said:

- There is a long-standing history of Pompe diagnostics at GOSH.
- Currently they offer low-invasive diagnostic testing, with further testing to aid in clinical decision making and monitoring.
- Addition of antibody testing will make the GOS Enzyme Unit a 'one stop shop' for Pompe disease in terms of diagnosis and complete monitoring of patients on ERT.
- High throughput multiplex assay may provide us with ways of testing for more diseases from a single blood spot and could lead to a viable method for neonatal screening.

Respiritory Support and measures. **Dr Mark Roberts Neurologist, Greater Manchester Neurosciences Unit, Salford Royal NHS Foundation Trust.**
Dr Roberts gave a detailed presentation on the respiratory issues associated with Pompe, first describing the broad range of symptoms: *"Pompe disease presents at any age with remarkable phenotypic variation, including variable rates of progression, degree of organ involvement, and disease severity, at least in part correlating with residual enzyme level".* Muscle weakness and breathing problems are the most common symptoms in Adult Pompe and so Mark ran through a number of slides showing the natural history of the condition, common presentations leading to diagnosis and MRI imagery.

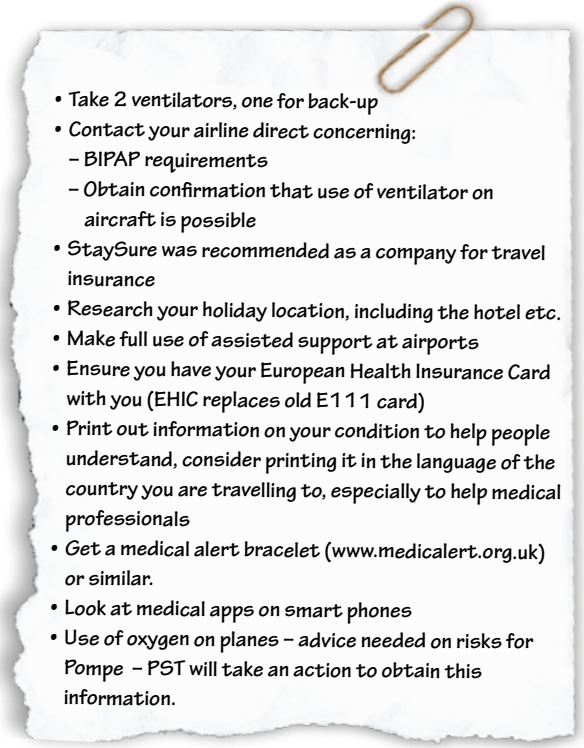
Mark discussed the need for adequate ventilation, the mechanism of respiratory failure, and pulmonary function tests used to monitor the severity or improvements under therapy.

He then showed how non-invasive mechanical ventilation has improved from the days of the negative-pressure ventilator (Iron Lung) to modern lightweight equipment and masks.

The Long-term Ventilation Service at Salford has a medical team of three consultants and a non-medical team of specialist physiotherapists and five other staff. Together they serve 1842 patients on home ventilation of which 38 are Pompe.

Travelling Abroad

A small group gathered during the conference to discuss points for further consideration when travelling abroad. These meeting notes, taken by Amanda Porter (PST), may be further developed into a leaflet in the future.



- Take 2 ventilators, one for back-up
- Contact your airline direct concerning:
  - BIPAP requirements
  - Obtain confirmation that use of ventilator on aircraft is possible
- StaySure was recommended as a company for travel insurance
- Research your holiday location, including the hotel etc.
- Make full use of assisted support at airports
- Ensure you have your European Health Insurance Card with you (EHIC replaces old E111 card)
- Print out information on your condition to help people understand, consider printing it in the language of the country you are travelling to, especially to help medical professionals
- Get a medical alert bracelet (www.medicalert.org.uk) or similar.
- Look at medical apps on smart phones
- Use of oxygen on planes – advice needed on risks for Pompe – PST will take an action to obtain this information.

Pompe Disease : A Treatable Multisystem Disorder: Research Update. **Dr Mark Roberts Neurologist, Greater Manchester Neurosciences Unit, Salford Royal NHS Foundation Trust.**

A last minute complaint from one company meant that companies involved with drug development were not able to present their programme updates to us in person, as they have done in the past. However, Dr Mark Roberts very kindly offered to present an independent update on the each of the studies covering:

» Genzyme NeoGAA
» BioMarin Gilt Technology
» Amicus co-administration of a Pharmacological Chaperone with ERT

» Audentes AAV-9 Gene Therapy
» Exosome Technology proposed by Mark Tarnapolsky

Mark described how each of the next generation therapies attempted to enhance targeting of their product to muscle cells and he gave an indication of the outcomes each would be measuring. Clinical trials for each are proposed in the near future and Mark concluded by saying that he believed that gene therapy could be available within five years.

It is exciting to see so much research and development being undertaken for such a rare condition and the AGSD-UK will certainly be monitoring the progress of these and drug programmes by other companies.

Exercise and Pompe:
Improving Exercise Tolerance. **Nicolai Priesler, Human Physiology Lab, Copenhagen**
Exercise & Hydrotherapy. **Ed Silk, Salford Royal NHS Foundation Trust**
Nicolai and Ed gave presented different aspects of Exercising with Pompe, but unfortunately my notes on both sessions are inexplicably scant. Nicolai's overall message was that lysosomal glycogen does not contribute greatly to the energy supply of muscle cells and so it should be fine for Pompe folk to exercise like everybody else. Obviously taking care not to over-exert which may lead to muscle injury. The problems with Pompe are more structural, rather than metabolic – so weakness is caused by damage to the cells, not an inability to process glycogen, as with other GSDs.

Ed Silk, on the other hand described a study that is now underway to examine the effect of exercising during infusions. The study protocol was described and we look forward with great interest to a detailed report at our 2016 conference in October. This will be given by both study centres, Salford and London (The National).

Ed talked briefly on Hydrotherapy saying that whilst it is excellent for many, in how it supports the body and allows a wide range of resistance exercises, however some people with diaphragm weakness cannot tolerate the water pressure.

Primary Healthcare and Pompe. **Mike Nichols, AGSD-UK Member**
Mike Nicholls led a workshop on the role of Primary Healthcare in the handling of Pompe patients. Whilst treatment was largely the responsibility of Trust-based consultants, it was also essential that GPs, when treating other ailments, did not take actions that were contra-indicated for Pompe. Noting the rapid changes in the NHS, it wasn't only the GPs that needed sufficient information to make the right decisions. Nurse prescribers, pharmacists, displaced GP specialists, specialist facilities (e.g. for scans) were taking over GP primary care responsibilities in many areas.
Mike's own experience was of one GP who was very interested in his condition, soon after diagnosis, and another who asserted that all responsibility lay with the consultant, to the point of being irresponsive to communications from the hospital. Only very recently had warnings about his breathlessness, limitations on the use of reinforced oxygen, and of anaesthesia, and balance problems, been emphasized by putting them on the front page of his medical records. This he believed was essential information to put on referrals for other conditions, and to catch the eye of others in the surgery standing in for one's own GP or with delegated responsibilities. Embarking on yet another description of Pompe and its impact can become quite a chore – in his surgery there were twelve GPs, eight nurses and two pharmacists with the authority to prescribe.

So searching out an interested GP, checking that Pompe and its impact are 'front page news' in medical records, and that appropriate references are included in referrals, are important. So maybe the further education of primary care staff through electronic training modules, already available for GSD V, McArdle Disease could be encouraged for Pompe.

A wide spectrum of experiences was expressed from those present, with perhaps the best experiences reported from those using smaller GP practices. The Pompe Support Team was asked to take the issues forward.

Editor's note
Since the completion of the PST's project to create a 101 Tips for Pompe Disease, the team are now focusing their attention on further publications, including materials for GPs.

Fundraising Campaign 2016

Bake or Burn
Cash for Calories!

Bake or Burn
Making Calories Count! Whether you prefer to bake-off or burn-off calories, you can join our calorific campaign to raise funds and awareness for people living with GSD.

BAKING - Self-raising awareness
If you prefer baking then why not hold a cake sale for your friends or work colleagues? Of course you don't have to make cakes; you can cook anything you like; how about selling hot soup and bread rolls after a brisk country walk? Or hold a "Come Dine with Me" evening for friends and neighbours?

BURNING – Shed lbs to Raise £s
If you're more of the active type; then try a sponsored walk, run, cycle, swim or any other activity to burn off a few calories. You can join an organised event or make up one of your own with family and friends.

Check out the AGSD-UK Facebook page to see what others are doing:
www.facebook.com/AGSDUK

Fundraising support:
fundraising@agsd.org.uk / 0300 123 2790

Sitting in a bath of baked beans

Jane Lewthwaite, Specialist Care Advisor, AGSD-UK

Prior to joining AGSD I worked in North Somerset Social services on a project for older people developing a community information service. Going way back to the beginning of my career I worked for Children in Need for about ten years. There I came across every possible style and form of fundraising from the funny, the dubious and the marginally legal, to the downright brilliant and inspired. People hired a double decker bus and wore fancy dress whilst stopping to collect at pubs. A group of runners had a massive sponsored jog every year usually starting in Cardiff and ending at the TV studios. Comic Relief and Sport Relief have really taken celebrity fundraising challenges to another level, with swims up the Amazon and down the Thames. They are very newsworthy and raise millions.

My job title was Regional Co-ordinator and I covered an area from Cornwall to East Sussex. The majority of my time was spent organising the grants that were made to groups and individuals. The budget for grant-making depended on the amount of income raised each year. During "Appeal time" work changed dramatically. We kept track of all the fundraisers who phoned or emailed in their details. We collected all the stories and passed them on to national and regional TV, Radio, Press Officers. Yes, including, sitting in a bath of baked beans.

You might know that Children in Need, Comic Relief and others have available an extensive, free fundraising pack. We are considering whether we could also provide something similar in AGSD-UK. Of course, it would have to be simpler and cheaper to produce. The good old sponsorship form is a trusty friend and might be a good starting point for us. If we design one would you use it? Would it help to have some pre-printed blank posters with our AGSD-UK logo on to help you promote your event?

Why do we need everyone to help?

AGSD-UK is very small and national charity so we slip through the net for many of the more obvious supporters. Large businesses and organisations, such as Sainsbury or Tesco, nowadays select a big, single, well known charity to support for a whole year. This is great for the big ones such as Diabetes UK, but means that there are fewer chances for the small ones like us. There are many local grant funders such as the Community Foundations, however, they operate only in their local area and do not consider national charities like us so again, we slip through the net.

Drive – it helps to have one or two key people willing to really push the event forward, these are the lynchpins. Can you persuade your most dynamic friend to help you?

Short term and time limited – We all have busy lives and cannot commit vast amounts of never-ending time. It can help to plan an event, devise a timetable of action and know how long people involved will need to make a commitment. This way people can see the end in sight.

Upsize a small idea – There are also some good longer term ideas. Could you obtain some large bottles or jars, label with AGSD-UK logo and ask friends and family to collect all their coppers for one year? If we could get 20 of our members to sign up 5 people each, we would have 100 people doing this. Guessing at £20 per jar we could make £2000.

Goal – Decide what event or idea you have and then think, realistically, about how many people could get on board with you and decide on a realistic financial goal to aim for.

Explain why your fundraising matters – try to include some explanation of why you are fundraising and what donations or sponsorship might buy.

Maximise money opportunities – if you have a cake sale, could you gather 10 nice items together such as a bottle of wine, toiletries, bisuits, and run a small raffle alongside it. It adds to the fun and adds to the income. Local shops will often give a small item.

Look for easy money – keep your eyes open for every possible, low input chance to make some money for AGSD-UK. Could you get our charity onto the Asda or Waitrose Plastic Token Scheme? They require a letter and prefer local charities, but they usually have a panel who decide which charities are to be included and we could help you write a letter explaining the work of AGSD-UK in your local area and making our case.

Local benefactors – Is there anyone in your area who regularly gives to charities that we could approach for support? There are often local business owners or organisations willing to make donations. Please contact Allan or myself to discuss the best approach and we could make a request to them for donations.

Groups – do you have any friends or family who belong to groups such as Choirs, bands, Chess Clubs or sports teams? Could you help them organise something fun for a large group to enjoy that also brings in donations such as a fancy dress parade, treasure hunt or mini-disco? Perhaps your local netball team request a bag packing day in their local supermarket?

Do what you enjoy – If you have a hobby, make it pay. It helps to work fundraising in to something you are already enjoying anyway. Gardeners; get sponsorship with friends to grow the biggest sunflower, hold a small open garden day, sell flowers, seedlings and seeds.

Always ask for GiftAid from tax payers – speak to Allan Muir about the details on it. We can ensure we claim 25% extra back on donations from people who pay UK tax, but they need to give a few details when they donate.

Make it easy to give – Virgin Money Giving is a web site where people can make donations to our cause and you can join through our "Bake or Burn" Campaign page: http://uk.virginmoneygiving.com/fund/BakeOrBurn

Just Giving pages are similar but they are not linked to our campaigns and their fees are much higher.

Fundraising

People often ask me if we have organised walks, runs cycles etc., we have tried in the past, but sadly really need full-time staff to organise them safely and efficiently. I may organise a small cycle ride (son of GSD Giant) in the South Downs again, but with such a small number of members, spread all over the UK, it is better for individuals or groups to find a local or National event to join and to use it to raise funds for the AGSD-UK.

If you are taking part in an event and would like to fundraise for the AGSD-UK, then please contact the office so that we can help you in several ways:

- Setting up a Virgin Money Giving (preferred) web-page
- T-Shirts supplied in your size(s)
- Collection tins
- Sponsor forms
- Charity leaflets
- Help with text for a personalised press release.

Below are a few organisations who are running or advertising events this year:

Big Fun Run

Run for the AGSD-UK

There are 5k runs all around the country that you can join in the name of the AGSD-UK. A full list of venues is listed here: www.bigfunrun.com/venues/

There's just over one month to go until the first of this year's events, which takes place at Brighton's Hove Park on Saturday 23 July!

Big Fun Run is an all-inclusive, family friendly series that you can enjoy all Summer long. The event is popular with participants of all ages, shapes and sizes

Once you have selected your event contact the AGSD-UK office and we'll make sure we have places available for you at your chosen location and for anyone else you'll be running with.

Parallel London

http://parallellondon.com/

Fundraise at the world's first push/run for everyone!
Parallel London 4 Sept 2016,
Queen Elizabeth Olympic Park

Exactly four years on from the London 2012 Paralympics, take your place on the start line at the iconic Queen Elizabeth Olympic Park for the world's first fully inclusive fun push/run.

This event celebrates our magnificent diversity as well as what unites us. All ages. All abilities. No cut-off times. No excuses! Run it, walk it, push it or be pushed – anything goes.

And that's not all. We're making the most of the spectacular venue with a free family festival. We'll be showcasing the very best accessible attractions and entertainment across technology, art and culture, food and drink, health and fitness and much more.

Join us in breaking down barriers and be part of sporting history.

Thunder Run

powvirtualrunning.co.uk/races/4589259225

Virtual Running allows you to run at any time and supply evidence to win a medal at any time throughout the year. Throughout July the organisers are supporting the AGSD-UK so that 20% of their profits will benefit the charity.

Stuart French helped us set up this arrangement and he has already clocked up 450 miles this year, very close to his target of 600 miles for the whole of 2016.

SiEntries

www.sientries.co.uk

SiEntries advertise and manage entries for a host of different organised activities around the UK and throughout the year.

- Adventure Races
- Challenge Events
- Cycle Races
- Cyclo-cross
- Cyclosportives
- Duathlons / Triathlons
- Fell Races
- Mountain Bike Events
- Mountain Marathons
- MTB Gravity Enduros
- Orienteering Events
- Road Races
- Running / Walking Events
- Swimming
- Trail Races
- Trailquest Events
- Ultra Running Events
- Walking Events

Wantage to Winchester Cycle Ride 2016

www.hyde900.org.uk/2016/05/17/w2w-2016/#more-3082

10am, Sunday 17th July

I felt that this would suit our Bake or Burn campaign very nicely. It is a cycle ride starting at Wantage, the birthplace of King Alfred the Great, and ending at his place of rest. Taking part in this we could really promote the line "Burning Cakes for GSD"

Charitable

https://charitablebookings.org/about

The AGSD-UK is registered with Charitable so that whenever you book a meal at one of their restaurants, we will benefit by £1 per person in your group. So keep their details handy and download their app onto your smartphone (if you have one).

CHARITABLE BOOKINGS

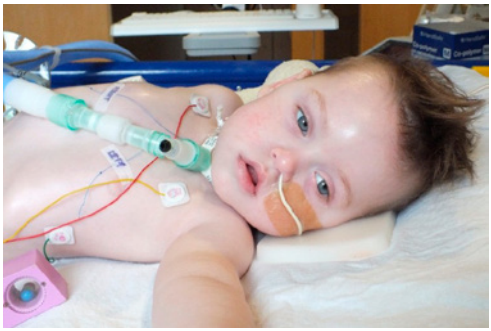
"Giving back has never tasted so good"

Bearded prisoners

Kara and Michael, parents of Leo received a donation from the prisoners at Altcourse prison, and they couldn't be more grateful! The donation was made along with this message:

"Please find enclosed a little something for Leo's appeal. The donation is from the prisoners at HMP Altcourse who have been moved by your story. The only way they could support you was by raising funds by growing beards and weird haircuts. For prisoners this was a big thing as image is very important inside. They hope and prey that little Leo becomes a strong person and achieves all he will dream for. Best wishes, Dave Mcalley and the lads at Altcourse."

Leo suffers from Infantile Pompe Disease and receives infusions of Myozyme. With this drug he is able to break down the glycogen in his body, and without the drug the build up would be fatal to him. He started weekly treatments February 2015 and was making steady progression until he was forced to start fortnightly treatments instead, and ever since then he has been steadily decreasing in health. His respiratory system is failing and the NHS isn't willing to give him weekly treatments again, which he so desperately needs. The family want to fund treatment every other week for one year so he can have, effectively, weekly treatment for a year. After that, they hope that the NHS will see sufficient improvement fund his treatment from then on.



If you too would like to support the family, or read updates on Leo's situation, they have an online fundraising page here: http://tinyurl.com/hwdyxux

Another family is also fundraising for their twins Orin and Olivia. You can read updates on their progress here: www.gofundme.com/helporinandolivia

Darlington and Stockton Times

Darlington Flower Club's Spring Fling raises funds for boy with rare disease

Darlington Flower Club, now nearing its 60th year, held its Spring Fling at Headlam Hall on Wednesday, March 23. Visitors enjoyed lunch, speakers, stalls and entertainment at the event, held annually in aid of good causes.

This year, the Spring Fling built on the prior fundraising efforts of members in supporting charities helping those with Glycogen Storage Disease (GSD). The charity was chosen in honour of Darlington boy, George Morrison. Six-year-old George is the grandson of Darlington Flower Club's chair and is one of just a handful of children in the UK to suffer from the rare condition.

Little George hit national headlines when his plight was revealed earlier this year. He is fed cornflour around the clock in a bid to prevent dangerous symptoms caused by GSD. The starch in cornflour creates a slow release of energy which stops the boy from suffering liver damage or falling into a coma – without it, he could die. His family are now fundraising to help charities in their work searching for a cure for the life-threatening condition.

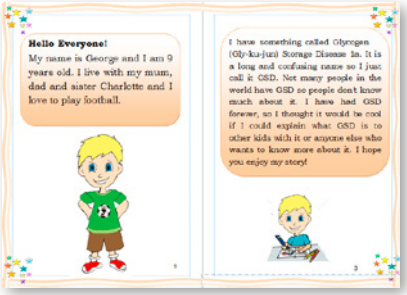
Members of Darlington Flower Club felt strongly about using their Spring Fling to support the family. The club's Doreen Howson said: "We all feel a personal connection to the family and wanted to support a charity helping someone living close to us. "George is such a lovely little boy and everybody wanted to help his family, who don't seem to be getting much support. "Hopefully one day, they will find a cure for children like George, it's heart-breaking to think of."

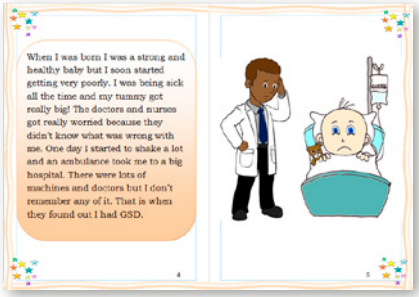
George's family are fundraising for the AGSD-UK. To donate, visit justgiving.com/curegsd.

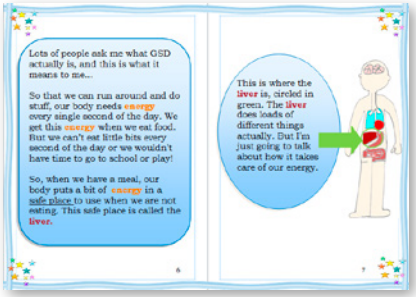
From left, Greta Metcalfe, Phyl Hodgson, Jane Currie, Doreen Howson and Christina Steel at the Flower Club annual Spring Fling at Headlam Hall.

GSD and Me!

Gabriella Clark was inspired to write a children's book about GSD as her close friend was diagnosed with GSD Ia. Gabriella is a fourth year medical student based in Truro, Cornwall, and she designed the book as part of a 'doctors as teachers' project. In the book a 9 year-old boy called George (GSD I a) describes his condition and how he manages his diet. Gabriella hopes to print a small number of these books to make them available to the AGSD-UK membership. Please let me know if you would be interested in buying a copy so that we can judge the potential demand.







Patient Story: Jae

Gene machine helps diagnose rare liver disease

A toddler with a rare disorder is the first patient to benefit from a new 'gene machine' system in Sheffield, which provides a faster diagnosis for rare and inherited diseases.

Jae Machin, aged two, was diagnosed with a Glycogen Storage Disease (GSD) after his blood was tested using the new next generation sequencers at Sheffield Children's Hospital.

GSD is a serious, but treatable metabolic disorder that can be caused by gene mutations that affect different tissues: the liver, muscles or the heart.

The new technology allows experts to screen up to 100 genes at once for mutations that may cause the disease, instead of one at a time. It means that tests can be returned in a matter of weeks instead of the previous system which could take up to a year.

Mum Jennifer Machin, aged 36, said: *"Having the diagnosis has already made a difference to Jae and to our family life and it is great that many other families will benefit from having access to this technology."*

Identifying Jae's disease

Jae's health issue was flagged up at Doncaster Royal Infirmary. Specialist Registrar Dr Charles Amobi, assessed Jae in a review clinic and noted his swollen abdomen and liver. Dr Amobi thought the youngster could have Glycogen Storage Disease, so referred Jae for further investigations to the metabolics team at Sheffield Children's Hospital.

Jae was screened and diagnosed with GDS type IXa. He is being treated by changes to his diet, including regular feeds of corn flour to stabilise his blood sugar levels. By his teens, it is hoped that Jae will have grown out of the condition. Mrs Machin, from Conisborough, South Yorkshire, said: *"Identifying the gene that caused the problem is making a difference, as we can make changes to Jae's diet and can now manage the condition effectively. It will also make it much easier when it comes to testing other members of the family."*

McArdle's News

Andrew Wakelin – McArdle (GSD V) Coordinator

Members of the Patient Liaison Panel for the McArdle's Service and some other McArdlites from the London area attended the Euromac Symposium in London on 1 April 2016. Although aimed at medical professionals, it was useful to have some patient involvement to make the subject more real for the professionals attending. I gave a presentation on the typical life experience of a person with McArdle's.

There are currently 266 of us diagnosed with McArdle's in the UK. These days most new people are recently diagnosed, but still some are appearing who have been diagnosed for many years and never been told of the support available.

Approx. 162 of us attend the McArdle Clinic in London, making it the largest 'centre of excellence' for McArdle's anywhere in the world.

An international group of us organised a workshop in New York on the patient-led initiative of the ketogenic diet over 15/16 August 2105. Dr Alf Slonim presented one session for us. Five patients who had followed the diet presented their experiences. A detailed report was produced and was promulgated via the "Ketosis in McArdle's" Facebook group.

An on-line survey of people using the ketogenic diet was carried out by Stacey Reason to help inform clinical research. There were 80 respondents, of which half had tried the ketogenic diet. Findings were presented at the workshop in New York and the AGSD-UK 2015 conference.

I attended a meeting of the Euromac Steering Committee in London in April and reported on my Dissemination role. We are now producing the 101 Tips book, the Reference for GPs (now called Medical Overview) and the Information Cards in a total of 9 languages. An "International English" edition of these publications has been produced and copies distributed to leading consultants in USA, Canada, Australia and New Zealand. In addition this edition has been useful for countries where most people have English as a second language – such as Denmark, The Netherlands, Sweden, etc.

The Euromac registry has been open for registrations for a while. UK McArdle patients registered now total 67, with Spain following with 37. We are hoping for a big increase by the year end.

Stacey Reason ran an on-line survey for McArdle people as part of her PhD studies. Over 400 people responded and 290 were suitably diagnosed and successfully completed the lengthy questionnaire. In addition to the results being written up in Stacey's PhD thesis, she is hoping that an article will be published in a medical journal.

A new leaflet is in preparation entitled "At work with McArdle's". It contains information aimed at employers, and aims to support employees with McArdle's by explaining the condition and how it may affect them in the workplace. If you have a need of such a leaflet, or are an employer with views on the employment of McArdle people, and would like to help with a pilot of the leaflet, please get in touch.

Last August an intrepid group took up a challenge event of climbing the "Welsh 3000s" – the 15 peaks in Snowdonia over 3,000 feet. Some other people attempt it within 24 hours, but we aimed to do it in four day-walks over a week. We suffered ridiculously inclement weather - often setting off into the rain and disappearing into low cloud! We had to abandon a number of the walks and finally achieved 9 of the summits, with a person-peak total of 64. So well done to all.

The Sodium Valproate trial has now recruited all the participants and the first ones have completed the trial process. Once the last participants have completed the results will be analysed and written up. We should have news at our Conference in October.

A trial of a supplement called Triheptanoin is currently underway in Denmark and Paris. This summer a small trial of the Ketogenic Diet is to take place in Germany. I think this must be the first time ever that three trials on McArdle's have been in progress at the same time.

This year we will run a walking course again, from 22-29 July. We are returning to the Pembrokeshire Coast National Park which we last visited in 2013. People are coming from the UK, USA and Australia.

On the three days before the walking course we are repeating last year's successful event for children with McArdle's and their parents. We are using

the same Pembrokeshire location, based at the National Trust's Stackpole Centre.

I shall be attending the AGSD US conference over 16/17 September, this time being held in Toronto. We are hoping to attract a strong McArdle's attendance.

"Having fun on the Children and Parents event, August 2015."

New Contracts for Homecare Services

The Department of Health have published the new Framework Agreements that the NHS have in place with homecare providers; these include Myozyme deliveries to homes and homecare nurses to reconstitute Myozyme and perform infusions. The agreement covers all Lysosomal Storage diseases and is published in the public domain under the Government's Transparency agenda. You'll note that there is one new company involved; Pharmaxo join Bupa and Healthcare at Home in providing this service. You may also notice that the guidelines for using special refrigerators has been relaxed for some Enzyme Replacement therapies, but sadly not for Myozyme.

Framework agreement for NHS National Framework Agreement for the Supply of Home Delivery Service - Lysosomal Storage Disorders
Offer reference number: CM/MSR/12/5381
Period of framework agreement: 1 October 2015 to 30 September 2017 with an option to extend for up to an additional 24 months.

www.contractsfinder.service.gov.uk/Notice/f7a4fb63-c8d9-45af-9f6b-259920dfa7f5

Bupa look to the skies

We understand from Bupa that their infusion nurses will be managed by Celestial Nurses from July 1st 2016. This is a new company for ERT nurses and injectable therapies. It is hoped that there will be more investment in team training in the future. Patients should by now have had a letter and should not see any changes to their care.

Living with 73 Years of Pompe's

By Mike Nicholls

Actually the above title, whilst technically true for me, is misleading. That's because for the first 45 years, after my birth in 1942, I did not know that anything was substantially wrong with me. I was not debilitated in any way that affected my work or social life. And for another 10 years or so I seldom dwelt on the condition, during that time diagnosed as muscular dystrophy, which became more irritating than disabling. So, compared to most sufferers, I feel I have been very lucky, since the day I was born. Which doesn't mean, when I (and family) look back, that deficiencies relating to Pompe's did not exist; they were submerged by my job and other interests.

I was the only child of parents who were impoverished for some years after the war; I became thin, pale and beset by a series of illnesses including measles, rubella, scarlet fever and yellow jaundice. The first thing a medical practitioner assessed (recently) as possibly Pompe-related was squint, in one eye, which developed whilst at primary school. For that I had a successful operation. Perhaps more clearly related was my inability to swim; my legs refused to propel me since they always hung down vertically. Dad was an outstanding distance swimmer, Mum not so bad either.

Then at senior school, though being very willing, some sporting activities were a problem. In races, I followed the field, about 10 yards behind in the 100 yards; so the long jump was also a problem. I could never do 'sit-ups'. At rugby, I was selected as prop in the house team, but had to stop after umpteen stones pushing at me from the front, and some 12 stones from (immediately) behind, buckled my back horribly. At cricket, I could belt the ball with all my strength from the middle of the bat only to see it drop into the hands of mid on or off. But that was strange, because when fielding, I could throw the ball, without a bounce, from the boundary to the wicket keeper. And I could hike for 20 miles in five hours with a heavy back pack, hitch-hike for weeks around Europe, climb the steep paths to the top of Snowdon, and cycle all day long. I was also an excellent roller skater, at a rink, down the hills, sometimes to and from school four miles away; I had no balance problems at all. And, whilst still at school, I worked in a shop for three years from 5.50 am for two hours every day, with no problems. At university, I carried on with the walking and extended my activities to weight training, though with some arm exercises being impossible, and to judo in which falling became a skill for later use!

At work, most of my first 14 years (to age 35) were spent directing and participating in research flights in various continents. We flew in the most hazardous weather situations we could find. Sorties in an unpressurised Canberra bomber involved wearing a pressure suit and breathing pure oxygen; I had no problems with the RAF medicals. A wonderful wife (Jill) and three healthy children were secured in this period; one daughter, in adulthood, became a top ladies marathon runner, the other an excellent swimmer, and my son followed Tour de France routes on his bicycle.

In 1977, I moved to the operational side of the Meteorological Office, my employer, at its Bracknell HQ, Berkshire, as one of six shift-working chief forecasters. Bracknell was the source of all weather forecasts for the public, the press, TV and radio, to merchant shipping, military and civil aviation, and to industry, covering the whole Northern hemisphere. The role involved continuous shift work, leaning over a bench to draw charts and write broadcast scripts. The work was tremendously exciting with the Office moving to the top of the league, globally, with its computer power, and in its use of data from weather satellites.

I was 'waddling' and leaning excessively backwards

On my move to management (a day job) in 1982, back pains, sciatica and a recurrent stiff neck became a nuisance, not however to the point of me taking any sick leave. I was told that my gait had become odd; I was 'waddling' and leaning excessively backwards. In 1987 my GP referred me to a consultant neurologist whose rounds included St Mary's and The NHNN (Queen Square) in London, as well as the local hospital at Windsor. Let's call him Dr X. During a week as an inpatient, mostly at the NHNN, tests, including a biopsy from my thigh, led to a diagnosis of a marked weakening of shoulder, hip and hamstring muscles due to limb girdle muscular dystrophy (MD). I was discharged on a Saturday morning after agreeing to assist Dr X in a lecture he was giving on muscular dystrophy. Later, he advised my GP that I had 'no convincing abnormality' and that the outlook for me was therefore 'excellent'. This is what was reported back to me at the time; how it quite squared up to the detail is, now, something of a mystery.

But there was, anyway, no impact on my work, even though, then as a Director with over 800 staff, and heavily involved in steering the Office from a pure civil service to a commercial organization, I was averaging 70 – 80 hours' work a week. I had annual check-ups at Dr X's Windsor clinic, though his personal interest had fallen away rapidly. I was normally seen by a junior doctor or registrar. In 1991, one of those, Shelagh Smith, a consultant neurophysiologist, made a referral back to the NHNN at Queen Square for repeat myometry. There had been some 'alarm bells'; I found, at a hydrotherapy session, that with my trunk immersed, I could not breathe at all and a frantic exit ensued; and once, crossing a stream by stepping stones, I could not bridge from one to another and nearly fell onto the rocks.

These tests revealed an abnormal drop, by about 2 litres, in my vital (lung) capacity when lying down, compared to the 1987 value. This triggered her suggestion, to a senior colleague, that my condition was Pompe's and not MD. His response was negative, since (he reported) the previous tests found no evidence of damaged cell structure in the muscle, no excessive deposit of glycogen, and because in his view such deterioration was not unusual in cases of truncal weakness. Years later, her diagnosis was shown to be the correct one.

I took an early retirement from the Met Office in 1995, aged 53, though carrying on full-time work as a private consultant. It involved a lot of travelling, suit- and briefcases in tow, which I could still manage. Overall, I had more spare time. But I soon found out that I could not do things I had given up some 10 – 15 years previously. Then, I had managed a large vegetable garden; now I was immediately out of breath and unable to bend forward to lift the spade; I could not play snooker, bending (over the table) was impossible; and I could not play darts, my aim was uncontrollable. Worst of all, I could not ride a bike – pedalling and balance were not a problem, but leaning on the handlebars caused too much shoulder pain.

In 1996, another of Dr X's registrars referred me back to the NHNN, concerned that I was not getting enough oxygen when sleeping; I had been having periods of severe breathlessness as soon as I lay down. I was admitted as an inpatient for five days. Various checks were made, though by 3pm on the Friday when I was due to be discharged nobody in Doctor X's team (nor any other) had been to see me, and few results made known to me. Literally at closing time, a registrar, contacted by an agitated nurse, appeared but with a main purpose to recruit me to assist at the Royal College of Physicians (MRCP II) training courses held at the National. I was pleased at this invitation. The routine would be, with other volunteers, to attend the NHNN a few times a year, and meet (separately) with about six groups of eight students (under the supervision of a registrar) who would be invited to diagnose my condition. Separately my MD was confirmed by letter, though qualified to 'limb girdle dystrophy with neurological problems'.

Many months later, after I attended a couple of MRCP sessions, I realized I had not received any appointment with Dr X or his team for my routine check-up, and no detail behind the hospital diagnosis. Indeed I was getting most of the information about my condition from the MRCP registrars, some of it quite worrying. After a phone call, a secretary arranged for an appointment with Dr X. A very curt appraisal ended with a reprimand 'for having made a self-referral which I had no right to do'; I would only be seen in future given a completely new, justifiable case from my GP. I left completely bewildered; how could my condition be so pronounced to warrant 'exhibition' status for senior medical professionals and students, and yet I find myself denied year-to-year check-ups and further

treatment. But I did continue with my commitments to the MRCP training, for another 2 ½ years, and received gracious letters of thanks from the Dean and Professors for doing so.

Whilst all this was going on, I was suffering an internal problem, for which an operation was deemed necessary, under general anaesthetic. I was also given reinforced oxygen. Afterwards, on lying down, hallucinations and severe breathing difficulties developed, and I was put under intensive observation at night. It was a few years' later that I was told how lucky I had been, two of the 'rules', applying to treatment of those with wasted diaphragms, having been (inadvertently) broken.

In 1998 my (new) GP, having declined to refer me back to Dr X, referred me to a consultant for rehabilitation medicine, Dr Collin, at Battle Hospital, Reading, to see if my gait difficulties could be treated. In separate tests, she found that my breathing capacity, when lying, had reduced further to about a third of what it should be, and made an urgent referral to Dr Hilton-Jones at the Radcliffe Infirmary, Oxford. He immediately took blood tests, and arranged for the hospital's senior anaesthetist, Dr Loh, to make direct measurements of the state of my diaphragm. Their diagnosis, confirmed by another biopsy, was that I was suffering from GSD Type 2, Pompe's. GSD 2 and limb girdle MD are similar in their presentation. The Oxford team set up a programme of regular monitoring, including sleep tests, and 6 monthly consultations at the Radcliffe; Dr Collin arranged for a six month period of weekly physiotherapy, which did bring back a state of overall fitness which enabled me to walk further and without discomfort. There is no doubt that this team's (including the Radcliffe's physio Jane and OT Jane) overall coordination and concern, quite literally, breathed new life into me. I owe them a lot.

This team (...) quite literally, breathed new life into me

I was put on night time ventilation at the end of 1999, after contracting influenza from a particularly nasty virus; my lungs could not cope with the amount of material that had accumulated in them. Some seven years' later Dr Hilton-Jones, staying in overall charge, referred me to Dr Lee's group at the NHNN for the ERT programme, and thus into the hands of another impressive and dedicated team, Drs Lachmann and Murphy and the very effective and supportive specialist nurses, Alison and Anna.

The most unsettled part of the treatment over the last 15 years has undoubtedly been the guidance on physical exercise. Early on I was encouraged, and did, and enjoyed, as much as I could, until it ached or hurt. Then came the day at one of the AGSD conferences, when I asked a presenter, one of the Dutch consultants, if exercises could be aimed at strengthening those muscle groups which were only partly affected. I couldn't have been more confounded by the answer, which was that I shouldn't be exercising at all since it would only cause further damage. This went unchallenged by any of the clinicians who were present. Thankfully the excellent work by Stephen Dando and his colleagues elsewhere has greatly helped to clarify the situation.

So now I am 73 and Pompes has advanced to the stage where my walking has become limited to 200 yards or so and I rely on a scooter most of the time. But other factors have played a significant part in the deterioration. For four years I gave priority to my mother's care when she had advanced dementia; it was physically and mentally exhausting. Then I was diagnosed as having osteoporosis in the hips and lower spine which meant avoiding falls at all costs; and more recently severe vertigo caused a total loss of balance over two months and denied me any chance to exercise. I only provide these details in order to make my final point: it is not Pompe's that has caused me the greatest difficulty, but always Pompe's plus something else (including even a cough and cold that non sufferers would find harmless). And the treatment for such combinations does, in my view, need more attention, particularly since GPs, with little or no knowledge of GSDs, also become involved; and, in my case, make very poor decisions.

But I must make one more point, regarding distraction therapy. It has played a massive role for me in offsetting the gloom associated with all of the above ailments. Jill, our children and grandchildren and some wonderful friends have played a big part in it. So has an absorbing hobby; in my case genealogical research and the writing and publication of biographies is so entrancing that I can forget, thankfully, everything else for months on end.

My glass is half full, of good medicine.

Glisten, March 2015

Duke Pompe Programme

Late-Onset Pompe Disease Patient Meeting
Duke University Medical Centre - United Pompe Foundation

A report by Allan Muir, Charity Director, AGSD-UK

The meeting was introduced and chaired throughout by Katie Berrier, a Certified Genetic Counsellor and a member of the Pompe multidisciplinary team at Duke. The topics covered showed the impressive extent of research studies being undertaken at Duke to improve the quality of live for Pompe patients, their care-givers and families. Below is a list of topics together with a few explanatory notes:

Physical Therapy and Assistive Devices. **Laura Case, and Karla Greene**
After a long round-up of assistive aids I asked Laura Case afterwards about robotic aids and she pointed me at the Kinova Robotics website, kinovarobotics.com, they are developing some interesting robotic arms to help people with limited upper body function.

Disability Issues, Caregiver Concerns and Resources. **Gina Miller**
Gina Millar raised some interesting points about "burnout" and coping strategies for care-givers. I think we will include some of her ideas in our AGSD-UK conference workshops this year. Making sure that you have sufficient time for yourself, every day, is essential for care-givers.

Words from our Sponsors. **Genzyme, Amicus and BioMarin**
Each of the sponsoring companies gave a brief talk about their study programmes and patient advocacy. Details of each of the studies are available on ClinicalTrials.gov (search for Pompe) and clinicaltrialsregister.eu. Each of the companies are seeking support from patient groups to help them better formulate Patient Reported Outcome Measures (PROMs), studies may include these functional measures and their results could, I imagine, be submitted to the regulatory authorities (FDA, EMA) along with clinical data.

Pompe Disease: An overview and Updates. **Priya Kishnani, MD**
Dr Priya Kishnani, Divisional Chief of Medical Genetics at Duke, gave an overview of the great strides that have been made since Myozyme (Lumizyme) became available 10 years ago. She also highlighted the large growth in patient numbers at this meeting since the first in 2015.

Since Newborn Screening (NBS) was introduced in Taiwan and now is being rolled out across the USA, As a result of recent NBS data Duke have amended the standard text to describe Pompe Disease in the literature; they now quote its incidence as 1 in

11,000 to 1 in 27,000 rather than earlier estimates of 1: 40,000. It's still an ultra-rare disorder, but many more patients are being missed than previously suspected.

As well as potential patient numbers increasing, the number of publications in the field is exploding, reflecting the huge amount of research being undertaken for such a rare condition. Highlights from the research stress that ERT provides good stabilisation of the disease in at least two thirds of patients – and there is no benefit in waiting to be treated. Work with patient organisations to get immediate treatment, she said. ERT is less effective in late-onset Pompe than infantile onset, it is most effective in the first 1 to 2 years of treatment; good prognostic factors include being female and young. She listed the main symptoms of Pompe along with a long list of symptoms less commonly associated with the condition including fatigue and gastrointestinal problems that are often under-recognised.

On ERT dosing Dr. Kishnani noted that the current standard of care may not be sufficient for all patients and that many are suffering a residual decline. An increase in ERT dose and/or frequency has been beneficial for many infant-onset children and several potential therapies (Enzyme replacement, chaperone, Substrate Reduction, Gene replacement) are currently being evaluated to hopefully give an improved response for late-onset Pompe.

Neurological and Small Fibre Neuropathy. Lisa Hobson-Webb, MD
Dr Hobson-Webb raised a few interesting points in her talk. For example, she described a small trial to examine alternatives to muscle biopsy to track the progress of ERT. For example a portable ultrasound device and electrical impedance monitor are being used to determine the amount of fat that has replaced muscle fibres.

There is little evidence to suggest that significant glycogen is stored in the brain, other than in blood vessel walls, although 30% of LO Pompe folk have dilated arteries. But the peripheral nerves are now being investigated to explain symptoms that several people have reported or may be present: Burning pain in arms hands and feet, temperature sensations, autonomic functions (heart-rate, blood pressure, gastrointestinal, absence of sweat, saliva, tears, sexual and urinary functions). A recent questionnaire of 44 Pompe patients found that half of them suffered from either: Restless Leg Syndrome, Dry eyes, Dizziness, Flushing, and Allodynia (unprovoked pain).

Lisa's take-home message was to "Keep Calm and Complain", suggesting that many of these symptoms should be taken more seriously, especially as there are many treatment options for them.

Update from AMDA Conference. **Paul McIntosh**
Paul, now a fourth year medical student has strong neuromuscular interests, especially as he has Pompe himself and yet ran a Marathon last year with no ill effects.
Paul's talk concentrated on a review of the Gene Therapy studies that were presented at the AMDA/IPA Pompe Patient and Scientific Conference in 2015. He discussed the different approaches being studied by Andrea Amalfitano, Barry Byrne, Dwight Koeberl ad Pim Pijnappel.

Respiratory Muscle Training and Lingual Weakness. **Harrison Jones and Kelly Crisp**
Harrison and Kelly ran a double act to explain the function of respiratory muscles and also the effect of weakness of the tongue and bulbar muscles that can lead to speech issues and sleep disordered breathing. Several tests and MRI investigations have shown that the tongue often has evidence of fatty infiltration is consistently involved in late-onset Pompe.

Kelly described respiratory muscle weakness as the primary obstacle to improving outcomes with ERT. She described some very "crazy results" of a study of respiratory muscle training (RMT: weight-lifting for breathing muscles) where subjects were encouraged to perform 12 weeks of training that involved a total of 9,000 repetitions (25 reps per set). They found that across the group an improvement of 20% in MIP (maximal inspiratory pressure) was achieved and 16% improvement in MEP (Maximal Expiratory Pressure). The "craziness" came from the continued improvement in MIP by all subjects after the training had ended; across the group MIP increased a further 12% but MEP declined by 1%. Overall, the group returned gains of 35% in MIP and 15% in MEP.

A further double-blinded placebo-controlled study is planned to measure outcomes through sleep studies and questionnaires of sleep symptoms and life/social activities. The placebo arm will use a sham-RMT devices to make subjects feel that they are exercising.

Treatment Advances, Albuterol/Clenbuterol Updates. **Dwight Koeberl, MD**
Dr Koeberl described in detail his groups approach to Gene Therapy; using an AAV8 vector to target enzyme production in the liver. One great benefit of this is the very low immune response to the GAA enzyme that is manufactured in the liver and delivered to muscles via the bloodstream. Clinical trials are hoped to commence in the near future.

The team are also encouraged by results from their trial of Albuterol (Salbutamol) as an adjunctive therapy to Myozyme. The hypothesis is that Albuterol should improve the receptors on skeletal muscle cells to ERT, so that take-up is improved and more glycogen can be cleared. Results in the clinic have shown an increase in measures such as the 6-Minute Walk Test (6MWT) and the team are designing double-blinded studies in LOPD that will follow patients for one year. The dose of Albuterol is crucial and

currently uses a 4mg extended-release capsule. Clenbuterol is more potent and longer lasting than Albuterol and, although it currently does not have FDA approval, a trial is planned for that as an adjunct also.

Finally, Dr Koeberl gave a strong warning about taking Propranolol (a beta-blocker for heart irregularities) with Myozyme. Testing of Pompe mice showed that they had worse survival and lost significant muscle mass with the drug, suggesting that it interferes in some way with ERT. Evidence suggests that the ERT binds to the muscle cell but is unable to enter the lysosome where it is needed. There are other, third-generation beta-blockers, that are more suitable and Dr Koeberl suggested that Carvedilol has been successfully studied.

Sleep Issues in Pompe Disease. Sujay Kansagra, MD
Dr Kansagra gave a lengthy talk about sleep and although his study objectives concern gaining a better understanding of "Pompe Disease and Sleep", his presentation was of interest to everyone who sleeps, except the few who dozed off during his talk, the first after lunch.

After describing sleep, Sleep Disordered Breathing, Obstructive Sleep Apnoea, and Hypoventilation, he gave a few approaches to improving sleep:
1) Practice good "sleep hygiene": Create an appropriate environment and bedtime routine. Prepare for sleep, by avoiding exercise, light (TV, Laptop, smartphone, tablet), caffeine, nicotine, alcohol, beta blockers, and some anti-depressants. But a hot shower can help to induce sleep. On caffeine he gave some interesting data and suggested that even a coffee in the morning can disrupt sleep patterns at night. Even decaffeinated coffee contains 5mg caffeine and Starbucks coffee could be lethal at 550mg per cup if you were to drink 20 cups (16oz)!
2) Manage sleep time: You can't cheat sleep so plan your day around sleep, its not a luxury.
3) Screen for Sleep Disorders: Sleep apnoea, restless leg syndrome, insomnia and he mentioned psychophysiological insomnia whereby a person focuses on their sleep and worries about not sleeping.

Glc4: A Urine Biomarker of Glycogen Accumulation. Sara Young, PhD
Dr Young described the process of reducing Glycogen to glucose by the GAA enzyme. Normally this would happen one glucose molecule at a time, but when GAA is deficient larger molecules are left and find their way to be filtered in the kidneys and passed in the urine. Measuring one of these compounds, the tetrasaccharide Gcl4, also known as Hex4, gives good

correlation with the breakdown of muscle glycogen to give an indication of the clinical severity of muscle disease. The technique can be used in diagnostics where it gives a similar response to CK, or Creatine Kinase, measurements. Using a dried urine strip it may be possible to pinpoint the best time to treat an asymptomatic patient. Alternatively, Glc4 may be used to monitor the effect of ERT over time.

Dietary Management in Pompe Disease. Surekha Pendyal, MSc
Surekha is a metabolic dietitian whose goals with Pompe patients are to decrease glycogen storage and increase protein synthesis. She mentioned the challenges people face such as fatigue, facial weakness, swallowing issues that can lead to under nourishment, and also lack of activity that can lead to weight gain and further muscle weakness.

Whilst individual diet must be designed in consultation with your medical team, she did offer her general recommendations for a balanced diet:
Protein: 25 to 30% to replace carbohydrates
Carbohydrates: 30 to 35% avoiding excessive intake of simple carbs (sugars)
Fat: 25 to 40% - avoiding trans-unsaturated fats

This is not an Atkins high protein diet but can be a challenge; foods should be chosen carefully to avoid high glycaemic index carbohydrates and trans fats. For example it may be wise to avoid too much meat, full-fat dairy products. Make sure you are well-hydrated and make sure you are taking your recommended daily allowance (RDA) of calcium and Vitamin D to avoid osteoporosis

Immune Modulation with Low Dose Methotrexate. Zoheb Kazi, MBBS
I arrived late into this lecture but I heard Zohab state that it is currently not possible to predict which patients will develop high antibody responses to ERT. Working with infants, effective protocols have been developed to treat CRIM- children with Rituximab and Methotrexate.

But for CRIM+ children and late-onset patients they are beginning to gain a better understanding and a study has produced encouraging results by treating them with a low dose of Methotrexate. Indeed 3 patients created no antibodies at all (did not seroconvert). Further studies are planned for this important work.

Whole Exome Sequencing in Pompe Disease. Mari Mori, MD
WES is a technique that can identify genes, and their mutations, by analysing only 1.4 of the whole genome. So it can be much quicker and cheaper to process. However, Dr Mori said that of the 350 pathogenic mutations known for Pompe disease, WES can miss 10%. For that reason, it would miss 10% diagnoses and so will not yet fully replace other diagnostic methods.

WES may also help to identify "Modifier Genes" that may give clues to the timing of disease onset or the association with high –titre antibody responses, for example.

The Utility of Muscle Biopsy and Pathology. Anne Buckley, MD
The need for muscle biopsies to aid diagnostics has reduced since blood tests became available, but there is still a role for them, particularly in adults, to aid research into the many unknown aspects of the condition. Dr Buckley presented many slides showing the deposition of lysosomal glycogen in muscle fibres and also unidentified debris in the spaces between muscle fibres. This debris may be impeding the flow of ERT from blood capillaries into the cells and so much further study is required, and muscle biopsies are therefore necessary.

The Duke team fully understand the discomfort of patients giving biopsies and so to show solidarity with the patient community, Dr Buckley has offered to undergo a muscle biopsy herself; they hope to present the slides at next year's Pompe meeting!

In summary, whilst the thoughts of the Pompe community are often concentrated on the future improved therapies, this meeting showed how the Duke team are working hard with their patients, and care-givers, to improve their lives in so many different ways. To see it all happening in one centre is very impressive and I would certainly recommend this meeting to others in future years.

See www.dukechildrens.org/services/medical_genetics/pompe for the latest news on Duke University's developments for Pompe disease.

Research Update

The US National Institutes of Health has a website that maintains a list of clinical trials being undertaken around the world. Anyone can visit **clinicaltrials.gov** for regular updates and use an appropriate search term to find studies that may be of interest, e.g. ("study number", "Glycogen", "GSD", "McArdle", "Pompe").

There is also a European Clinical Trials Register that provides information on EU studies: **www.clinicaltrialsregister.eu**. Similar search facilities are available as above.

Below is a small selection that may be of interest:

NCT02054832: Sleep and Quality of Life in Patients With Glycogen Storage Disease on Standard Versus Modified Uncooked Cornstarch
Principal Investigator: John J Mitchell, MD Montreal Children's Hospital of the MUHC
Patients above 2 years old and their parents (for children only) will be enrolled during their usual follow-up. Parents will be asked to complete a quality of sleep questionnaire (as it pertains to both child and parent) relating to the past month on their current dietary regimen (standard UCCS) and a quality of life questionnaire (as it pertains to the child only). Parents will then complete a sleep diary (for both child and parent) and both child and parent will wear an actigraph that will record movements during sleep over a 1 week period. Adult GSD patients will complete their own questionnaires.

Following this first assessment, they will be hospitalised over a 24 hour period as part of standard of care to start the modified UCCS, Glycosade, under supervision and with a continuous glucose monitoring (CGM) sensor. Following hospitalization, the family will return home. Glucose will be monitored with the aid of the CGM sensor for 5 to 7 days. The actigraphy and the sleep diary will be repeated after 2 weeks (for 1 week) while on Glycosade. One month after starting the modified UCCS, questionnaires on quality of sleep and quality of life will be repeated.

NCT02683512: Glycogen Storage Disease Type IV Database
Principal Investigator: Loren Pena, MD, PhD, Duke University, North Carolina
The long term goal of the proposed project is to develop a repository of information with the long term goal of expanding the understanding of the disease manifestation and natural history of GSD IV.

Specific Aims: To establish a repository of clinical, laboratory, and biochemical information on individuals diagnosed with glycogen brancher enzyme deficiency. In the long term, this information will permit a better understanding of the long term complications and clinical course in GBE deficiency.

This research project will enroll individuals from outside institutions. The goal is increase the robustness of data collection and analysis with a large number of participants.

NCT02385162: Biomarker for Glycogen Storage Diseases
Principal Investigator: Prof Arndt Rolfs, University of Rostock, Albrecht-Kossel-Institute for Neuroregeneration
Development of a new Mass-Spectroscopy-based biomarker for the early and sensitive diagnosis of Glycogen Storage Diseases from plasma and saliva. Testing for clinical robustness, specificity and long-term stability of the biomarker.

NCT02635269: Fat and Sugar Metabolism During Exercise in Patients With Metabolic Myopathy
Principal Investigator: Karen L Madsen, MD, Neuromuscular Research Unit
This study aims to characterize the pathophysiological mechanisms of 21 different metabolic myopathies (including GSDs 0, II, III, IV, V, IX, XV). The study will focus on exercise capacity and the metabolic derangement during exercise.

There have recently been a number of studies looking at dietary modifications for GSD III and particularly for GSD IIIa. Two of these are referenced below, but we must stress that our medical advisors recommend great caution; for example Dr. Ulrike Steuerwald told me:
My experience (underlined by similar findings by Dr, David Weinstein) proposes that a ketogenic diet should not be advised for children or adolescents. We lack knowledge about the long-term follow-up in adults. These diets should be tried only under supervision of a very experienced metabolic specialist and at best only in an adult.

www.ncbi.nlm.nih.gov/pubmed/19322675
Reversal of glycogen storage disease type IIIa-related cardiomyopathy with modification of diet.

www.ncbi.nlm.nih.gov/pmc/articles/PMC4302571
Glycogen storage disease type III: modified Atkins diet improves myopathy
Sebene Mayorandan, Uta Meyer, Hans Hartmann, and Anibh Martin

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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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