Glisten

Glycogen Storage News

AGSD

August 2014

Volume 12 Issue 2

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GSD Giant

Going for Gold

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Developement Update

I want to take this opportunity to give my wholehearted thanks to Andrew Wakelin for steering this little support-vessel along a new course over the last four or five years, and for having the courage to push-through our three-year development plan and to make significant changes to the charity's governance. Many of his changes may have gone unnoticed by most people but they have significantly reduced our exposure to risk and improved our potential to grow as an organisation. I also thank Andrew for his continued and unerring support throughout my period in post as Development Director. I wish Andrew good health and success with his continued efforts on behalf of the McArdle community.

And so I welcome Mike Porter as our new chair; coming from a business management background I know that he perfectly understands the issues still faced by the Association and I'm sure that he is the right man with a vision to help us morph into a magnificent battle-cruiser, fighting for the GSD community in all directions over the coming years. I leave it to Mike to fully introduce himself in the following pages.

It is also my duty to report that three years since establishing the AGSD-UK office in Droxford the Board of Trustees were unable to find the necessary resources to renew the contract of our Administrator Wendy Griffiths.

Many of you have expressed your sadness at Wendy's departure and I have passed those on to her; I am certainly missing the office support as my own workload has increased as a direct consequence. With our new Strategic Plan in place and renewed effort to improve the charity's financial position we are ever-hopeful that an office manager can be afforded, but that may take some time.

It has always confused me as to why the AGSD-UK finds it particularly difficult to develop an income stream to cover what are quite modest running costs of such a small office.

I network closely with several other charities of similar size that don't seem to struggle in the same way. But something that may be indicative of an underlying problem is the confusion amongst many of our supporters (families, medical professionals, industry) about which organisation supports which GSD condition in the UK. Here are a few examples:

- NHS nurses have pointed out to me that talking about Glycogen Storage Disease may not be fully appreciated by many parents caring for Pompe children; they don't necessarily relate Pompe to GSD.
- A senior research professional in a large charity told me that another

charity existed in the UK that supports McArdle disease – it's run by a chap called Andrew Wakelin (our former chairman) apparently! In fact she was referring to the AGSD-UK without realising because she didn't immediately associate McArdle with GSD.

- I am often referred to as the leader of the "Pompe Association" by NHS commissioners and other health professionals. At the present time there is no such Association

So I and the trustees are currently exploring how we might exploit these misunderstandings in our favour, by creating separate identities within the Association in order to improve awareness of our conditions, improve public understanding of the charity, improve our fundraising performance and help the GSD community appreciate the benefits of our association.

One solution might be to form three distinct identities under the AGSD-UK umbrella:

1. Liver GSDs: Conditions managed by close attention to dietary restrictions – GSD 0, 1, 3, 6, 9, as well as the much rarer types 4 and 11 (Fanconi-Bickel)
2. McArdle and related GSDs: GSD 5, 7 and Muscle-affected GSD 4, 9, 10, and 13 where low intensity aerobic exercise is found to alleviate the primary symptoms.
3. Pompe GSD: Infantile, Juvenile and Adult forms where muscle weakness and respiratory weakness are primary symptoms and the only available treatment is an Enzyme Replacement Therapy (ERT).

I'd like to explore this idea further at our conference in October where we can hold separate workshops for the three main sub-groups to consider views on current perceptions of the AGSD-UK and what changes may benefit the organisation.

So put your thinking caps on now and bring your bright ideas to our Cheshire conference on 18th October.

Allan Muir

Development Director

Five years? Where did they go?

Andrew Wakelin

It doesn't seem like five years... but that is how long it is since I steered the Executive Committee of the then unincorporated AGSD-UK to become protected by incorporation. All I was after was a sound basis for developing the activities and publications for my GSD Type - McArdle's - for which I was the Coordinator.

However, immediately after delivering the "package" at the AGM in 2009 I mysteriously found myself elected as a Trustee and appointed as Chairman of the Board.

We recently worked out that about 4,500 people in the UK are fairly directly affected by Glycogen Storage Diseases. Not just those with a diagnosis, but siblings, parents, grandparents, etc. Over the last five years the association has moved on considerably, underpinned by legal protections, insurances, enhanced ability to enter into contracts, staff, awards of grants, growing sponsorship. We have more people than ever helping, having devolved the role of the Type Co-ordinators from that of the Trustees. We now have Coordinators for more of the Types than ever before, and our Type Coordinator meetings provide peer support and encouragement to them all. We have delivered improved services, information and support to those affected by GSDs. And in many areas we have played important parts on the international scene.

But, my goodness, is it hard work. With relatively few people affected we don't have a big pool of people from which to draw volunteers and fund raisers. We desperately need more people with professional skills, with in-depth knowledge of the various GSD Types or with a passion for fund raising. Being a group of ultra rare diseases is bad enough, but then add to the mix the fact that few people recognise the word "glyocgen" let alone understand the various conditions. We must all pull together to tackle the challenges.

I am standing down as Chairman and Trustee to look after my health, and to concentrate my remaining energies on my own GSD Type as McArdle Coordinator. I am very pleased to be able to hand over to Mike Porter, recently elected as Chairman by the Board of Trustees. Although he has been on the board for only a relatively short time, I have every confidence that Mike will steer the Association successfully in the coming months and years.

Please give him your wholehearted support.

Meet our new Chair

Mike Porter, Chair AGSD-UK

Saturday 31st May 2014 was in my opinion a sad day for AGSD-UK. Our longstanding chairman Andrew Wakelin tendered his resignation as both Chairman and Trustee of the charity on health grounds. Under his tenure the charity developed significantly and his absence and stewardship will be sadly missed. However Sir Alex Ferguson's departure from Manchester United proves that no one goes on forever and an organisation has to adapt and further evolve under new leadership. I would like to personally thank Andrew for his unstinting devotion to the cause and development of AGSD-UK.

Continuing the football analogy I sincerely hope that under my leadership AGSD-UK does not become the Manchester United of last season and that I get more time in the role than David Moyes did! My hope and aspiration for the charity is to build on the splendid work that Andrew and his fellow trustees have delivered to-date. I will be looking to move the organisation further along its path as set out in its strategic plan which is underpinned by the primary objective of providing those services that everyone affected by a GSD needs.

As I see it three of the key challenges to achieving our strategic objectives are: to get a much higher level of engagement with the UK GSD community, to significantly improve our financial position and to further strengthen the trustee board.

As a board of trustees we exist only to serve the needs of the charity – your needs. Without your engagement we will be in danger of operating in a void, we need you to interact with us in a way that helps us identify and meet your needs. Linked to that is that the services we provide have a cost. To deliver, improve and enhance those services we need your help to increase our fundraising efforts because as a charity we are entirely reliant on donations. Our fundraising partners Naked Fundraising have commenced the groundwork required to raise the profile of AGSD-UK with charitable foundations and trusts thereby increasing our chances of obtaining much needed grants but local fundraising needs to increase to complement other potential sources of income streams. Widening the skill set of the current trustee board particularly in the areas of finance and marketing and communications will I firmly believe help us deliver our strategic objectives.

I will be looking to keep you all regularly updated on our progress in future editions of Glisten and look forward to serving AGSD-UK as its chairman.

Strategy

AGSD-UK Strategy update (by Brian Topping, Non-executive Trustee)

The charity's strategy went live on 1 April 2014 as planned but much has happened in the intervening time that's not allowed full visibility of this. However a summary will be placed on the website very soon alongside regular updates on how progress is being made and how you can help.

The Strategy will run for three years from 1 April 2014 to 31 March 2017 but what will happen during the term of the Strategy.

But why do we need a strategy?

The charity is small and reflects the rare condition of GSD and its various types. We have limited resources and not enough to cover all that we need to do if we are to survive and develop. We need to improve and build on our financial position and that's why Naked Fundraising is helpfing us with this task. We also need to ensure that we get the right level of stakeholder involvement and that doesn't only mean financial support. It means looking for ways that we can get the right skills and experience to share in our drive to improve the position of the charity.

Our communication needs to be looked at and overall we must ensure that all our members and people affected by GSD are properly supported. So those are the reasons why we need a strategy.

How will we make sure all this will happen?

With this in mind, we have set out three key objectives as follows:

1. To ensure sufficient funding is available for the charity to maintain financial stability; to be confident and transparent in our strategic planning; and to improve the on-going financial position of the charity. (Overall responsibility Allan Muir)
2. To identify, engage and mobilise appropriate resources to ensure achievement of AGSD objectives. (Overall responsibility Mike Porter)
3. To establish an easy to understand and inclusive information and support structure, which fully meets the needs of everyone affected by GSD at any point in their journey. (Overall responsibility Wendy Bascal and Amanda Porter)

There is a very detailed Delivery Plan that has stepped processes to make sure we can track how we are progressing the successful delivery of all of this work and to pick up early where difficulties are being experienced.

This is a new approach for the charity and it's already clear that there is the need for a settling in period but with some staffing and personnel changes on the Board, this will help provide a stable platform from which we can progress successfully.

However this is not the Board's plan but your plan. It's been said on many an occasion that the success relies on everyone in the AGSD community pulling together and making this happen. This was just to let you know that change is happening but it's akin to turning an oil tanker. It may take time but it will happen with your help and involvement.

We need you

Fundraising is an integral part of AGSD-UK, without voluntary donations and people doing fantastic things to fundraise for us, we would not be able to do the vital work that we do. Which is why **we need you!**

For any hard core runners that missed the London Marathon ballot this year you are in luck! AGSD-UK are lucky enough to have been given one 2015 place, all you need to do is pledge to raise more than £1500 through Virgin Money Giving and the place is yours, please contact us asap to let us know how much you think you can raise.

Summer is finally in full swing and us fundraisers couldn't be happier. Brilliant weather always puts us in the mood for events and what better way to combine a favourite pastime (the pub) with fundraising than by taking part in or even better, running a **pub quiz!**

If you are one of those lucky people to have a great local then why not ask them if they are able to host a quiz in aid of AGSD-UK. All the materials are ready to go for you to set up the night, you just need to gather friends and family for a fun night out raising money for a great cause.

You could also take advantage of any sunny spells by holding a **fundraising BBQ** for AGSD-UK. Why not invite friends and family over for a BBQ and ask people to donate £5 for a burger and a beer?

We also need your help in asking **local companies** if they can support AGSD. More often than not, companies have a corporate giving scheme where they will select charities to support with employee fundraising. Please put AGSD-UK forward as a charity to benefit!

Well done to Kezia Moyse, who after recently raising an incredible £1,500 with a sponsored walk, has now got her school involved with raising funds for AGSD-UK.

They are holding a raffle, a 'guess the amount of sweets' jar and the whole school are taking part in a sponsored walk. A huge thank you to Kezia for this fantastic effort!

If you and your children would like to fundraise with your school, please contact us for more information.

Memory Walk for Pompe Disease

Kezia Moyse raised an amazing £1500 (three times her original target) for the AGSD-UK by organising a sponsored memory walk around her family's old home town of Plymouth. The walk celebrated International Pompe Day and also marked the 10th anniversary since her sister Azaria started her life-saving Enzyme Replacement Therapy.

Kezia's memory walk featured prominently in several local newspapers, including the Plymouth Herald with the headline:

Hair Show

Beck Walton's students of Hair Design at Tresham College in Corby put on a show with all ticket proceeds donated to the AGSD-UK.

If you have any other ideas about how you can support AGSD-UK or for further information on fundraising, please contact us on info@nakedfundraising.co.uk or give us a call on 01489 877 319.

Singing for Nora

Sylvia and Edwin Willson have been raising impressive sums on our behalf through the Gipping Valley Rotary Group and St Edmundsbury Male Voice Choir.

A sick three-year-old girl has had a concert staged in her honour after a Bury St Edmunds choir felt compelled 'to do something' to help.

The Edmund Octet's recent performance at The Unitarian Meeting House was held on behalf of member Edwin Wilson's granddaughter, Nora, who has Glycogen Storage Disease (GSD). It raised £1,000 for the AGSD-UK charity. Mr Wilson, of Elmswell, who sings in the Octet's top tenor section, said his son, Huw, and wife, Marita, were told 'they had more chance of winning the national lottery than having a child with GSD'.

He said: "Nora's condition was first spotted by her GP during her routine 12 month check up, although it took a further eight months of tests, including muscle and liver biopsies, before it was confirmed she had Type 3 GSD.

"She has an enlarged liver and this prevents her lungs functioning properly because they cannot expand fully."

Marcel Rousseau, Octet founder, chairman and top tenor, said:

"We are humbly pleased to have had tremendous support from our regular supporters and the general public who enabled us to raise the magnificent sum to give to the Trust which is seeking a cure for this insidious condition.

GSD Double Century Challenge

Cyclists on the 208 mile cycle around the South Downs, reported elsewhere in this issue, raised an impressive £5000 for the AGSD-UK.

Anyone fancy riding 300 miles next year??

Conference Programme Outline

We are still awaiting confirmation from several speakers and so the programme outline below should be regarded as provisional. However the purpose of the conference is to discuss issues that affect individuals affected by Liver GSD, McArdle (and associated conditions) and Pompe, so we do intend to leave space in the programme for you to raise your own issues and questions on both the Saturday afternoon and Sunday sessions.

Liver GSD Workshop

There will be opportunities for separate Liver GSD workshops depending on the number of delegates registering. For example we often split GSD 1 and GSD 3 but could also organise small meetings for GSD 6 and GSD 9 if the need arises.

Personal Experiences of GSD 1
Martin Grinnel

Research Developments
Dr David Weinstein, University of Florida

Liver GSD Carrier Study
Tayoot Chengsupanimit, University of Florida

Panel of experts Q&A
Panel of medical professionals

Dietary Workshop for children and Adults Marjory Dixon, GOSH

Liver GSD Publications
All present

Fundraising initiatives for Liver GSD
All present

Workshop for McArdle & related GSDs

Discussions will be led by Andrew Wakelin, our McArdle Representative, but will be relevant in some cases to other muscle GSDs: 4, 7, 9, 10 and 11.

Traditional format of a small discussion group with varied agenda including:

Publications
Walking with McArdle courses
Euromac European registry for McArdle and related neuromuscular conditions
Fundraising initiatives for McArdle and related GSDs.

Staff members from the **McArdle Clinic** at the National Hospital, London, will be present to join in the discussions.

Pompe Infants Workshop

Saturday afternoon will host a 3-hour workshop led by Joan Fletcher CNS for families caring for Pompe infants. We'll have a number of specialists on hand to answer questions including Clinical Nurse Specialists from the LSD centres. Topics will include:-

ERT Outcomes
Dr Alex Broomfield (Manchester Children's Hospital)

Clinical psychology Physiotherapy
Michelle Wood (GOSH)

Speech and Language
Sonia Lozano (GOSH)

Newborn Screening
Prof Simon Heales (GOSH)

Treatment Guidelines
Dr Simon Jones (Manchester Children's Hospital)

If time permits
Pompe publications Fundraising initiatives

Pompe Adult Workshop

We will have a varied programme over Saturday and Sunday. Not too intense and hopefully some fun in the interactive workshops and relaxation as we all experiment with Mindfulness.

Saturday Workshop

Respiratory Issues and Back Pain
Mark Roberts, Royal Salford Hospital

Mindfulness
Sam Murduck (PST) & Annabel Rajgor

Exercise and Diet
Steve Dando (National, London) and Nicola Condon(UHB)

Programme Update
Amicus Therapeutics

Sunday Workshop

Pompe Publications – 101 Top Tips
Angela Biggs (PST)

Research Developments
Powell Gene Therapy Centre, University of Florida

International Pompe Day
Ben Parker (PST)

Fundraising initiatives for Pompe
Allan Muir (AGSD-UK)

Pompe Advocacy Requirements
Joan Fletcher & Allan Muir (AGSD-UK)

Pathways to Diagnosis
Genzyme UK

AGSD-UK AGM

The Annual General Meeting will be held first thing on Sunday morning and will be an opportunity to meet the Trustees and officers of the Association.

AGSD-UK Annual Conference 2014

Wychwood Park, Weston, Crewe, Cheshire, CW2 5GP
October 18th and 19th 2014

Registrations should be received before 15th September to qualify for subsidised rates - Please complete registration form inserted to rear of book.

We would encourage you to arrive during the morning, 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 start on Saturday 18th October at midday with registration and lunch. during the afternoon there will be the usual eclectic mix of speakers and workshops. The day will finish with the Conference Dinner, which will take place at 7.30pm.

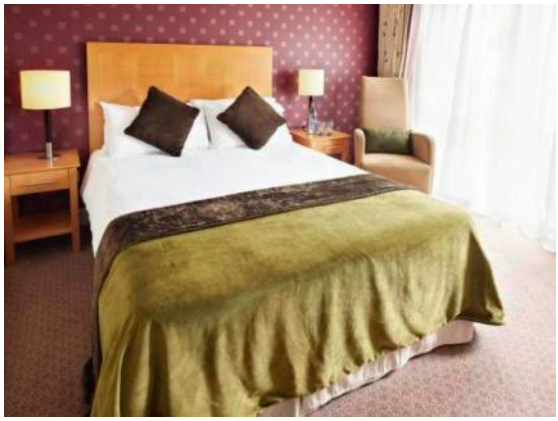
The Conference will continue on Sunday morning, and finish at 3.00pm (TBC) with a buffet lunch provided at 1pm. Again, if you wish to stay for the afternoon, there will be opportunities for networking.

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 number of rooms at the centre, and AGSD-UK will subsidise the cost of accommodation for members of the association.



Residential and Day Delegate Packages

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

Getting to the Conference Centre

By Road from the M6 Junction 16, take the A500 toward Nantwich. At the roundabout take the A531 towards Keele and Nantwich. At the next roundabout turn left on to the A531 towards Keele. Wychwood Park is on the right hand side and the entrance is at the right exit on the next roundabout. By Road from Chester take the A51 to Nantwich. Follow this road until you reach the Burford Crossroads (Junction with A534) where you turn left, signposted to Stone A500. Stay on this road through Shavington and Hough. Go over a railway bridge and at the next roundabout take 3rd exit on to the A531 towards Keele. Wychwood Park is on the right hand side and the entrance is at the right exit on the next roundabout.

By Rail

The nearest train station is Crewe, approximately 5 miles away from Wychwood park. There are direct trains from Manchester Airport to Crewe Railway Station which take 33 minutes

By Air

Wychwood Park is 35 miles south of Manchester Airport via the M56 and M6. If you are travelling by air please contact the AGSD-UK office with your flight details; we may be able to assist with your connections between the Holiday Inn and the airport.

If you require a taxi please call Cars R Us: 01270 505999

Residential Packages and Day-delegate rates

Patient and Family Residential Package

For **one** person staying overnight: Includes conference fees, Sunday breakfast, lunches on Saturday and Sunday, all refreshments and conference dinner.	£60.00
Additional adults (sixteen or over) sharing this room: Includes conference fees, Sunday breakfast, lunch on Saturday and Sunday, all refreshments and conference dinner.	£30.00 per person
Children under sixteen	FREE

Bursaries are available to help patients and families with these costs. Please apply to the charity office if you require financial support to attend this conference.

Patient and Family Day Delegate Costs

For **one** person attending each day (not staying overnight) Includes conference fees, lunch and refreshments.	£15.00 per day
Conference Dinner:	
Adults	£15.00 per person
Children under sixteen	£5.00 per child

Health Professional Residential Package

For **one** person staying overnight: Includes conference fees, Sunday breakfast, lunch on Saturday and Sunday, all refreshments and conference dinner.	£100.00
Additional persons sharing this room: Includes conference fees, Sunday breakfast, lunch on Saturday and Sunday, all refreshments and conference dinner.	£50.00 per person

Health-Professional Day Delegate Costs

For **one** person attending each day (not staying overnight) Includes conference fees, lunch on Saturday and Sunday, and refreshments.	£30.00 per day
Conference Dinner on Saturday evening	£30.00 per person

News in brief

Transformative gift to create the world's first centre for research into rare diseases in children

Plans to open the world's first centre dedicated to paediatric research into rare diseases at Great Ormond Street Hospital (GOSH) are to become a reality thanks to a £60 million gift from Her Highness Sheikha Fatima bint Mubarak.

Her Excellency Professor Maha Barakat, representing Her Highness Sheikha Fatima bint Mubarak with Jade, a 15-year old cardiac patient at Great Ormond Street Hospital

The gift, given by the wife of the late Sheikh Zayed bin Sultan Al Nahyan, founder of the United Arab Emirates, is in recognition of GOSH's unique position to advance treatments and cures in this area. It builds on the legacy of her late husband who had an active interest in global issues affecting the wellbeing of children. It is also given in gratitude to the hospital for the pioneering treatments it has provided to children from around the world, including those from the UAE.

Phase 1 of our GSD National Audit completed

In late March AGSD-UK employed a social researcher to reach out to all acute hospitals in England to inform them about the AGSD-UK and to encourage their GSD patients to register with the AGSD-UK. That work is now complete and we are currently compiling a report and executive summary for dissemination to all our stakeholders.

The most important piece of that research was the small card that was sent to patients and carers asking them to register with the Association. As a charity that has been running for nearly 30 years is no surprise that our records need updating; we have many members for whom we no longer have valid contact details.

It was hoped that both new patients and our existing members would use the card to help us update our records and give a better understanding of the UK demographics relating to GSD, but sadly we have received only one third of those we expected.

So if you haven't updated your details with us lately please use the card included with this issue of Glisten, or alternatively fill in the on-line form that you can find from the home page of our website: www.agsd.org.uk.

Double-Century Challenge promotes GSD Giant charity Sportive

By Allan Muir

On 8th June this year 13 cyclists gathered nervously at dawn to embark on a 208-mile cycle ride around the South Downs National Park in support of the AGSD-UK. For some their longest training ride had not exceeded 100 miles, let alone a double-century, so the anxiety was high: What would they feel like after climbing the 2-mile steep climb to Beachy Head? Would their glycogen reserves be sufficient for the undulations of the South Downs on their return from Eastbourne to Petersfield?

The ride set out from my house in Buriton, near Petersfield, Hampshire, at 5:30am and travelled west towards Winchester before swinging round to the east and following the coast roads all the way to Eastbourne for lunch. After 125 miles, a bowl of pasta and a sports massage, they were ready for the return trip of 85 miles along the northern edge of the South Downs back to Petersfield.


SPORT

Rob's Charity Challenge

Crucial cash helps battle to cope with rare disease

Chapeau to Tommy! Pride but there's no way I could

in a mission accomplished make it into daily routine

SPORT

RIDER TRIBUTES

RIDE IN FIGURES

207.9

2,419

12:41.39

134

77

7,148

16.3

Take on your own challenge

Why massage?

SNACK ATTACK

The fine weather and varied conditions throughout the day, kept the riders alert and motivated to complete their mission. Passing by mist-covered Petersfield lake in the early morning, hard climbing under the hot mid-day sun, and finally riding in close formation in the dark with bright front and rear lights all made the event a magical experience.

The fact that all the riders were still pushing hard in the final approaches to Petersfield at 10:30pm showed just how effective our training had been and how important the nutrition and hydration had been on the day. Scheduled stops were made every couple of hours to fill mouths and pockets and a mechanic was on hand to sort out any issues with our trusty steeds.

The ride showed that 208 miles is more of a mental than a physical challenge and that the charity's Sportive courses of 105, 78 or 48 miles should be accessible to all cyclists, even those new to the sport. It also showed what wonderful event organisers are GSD Cycling Events!

Sportives will usually have feeding stations every 25 miles or so, topping up muscle and liver glycogen after around 2 hours of effort. It is no coincidence that kids living with hepatic Glycogen Storage Disease (Liver-GSD) need to eat every 2 hours whilst at rest and during exercise because they can't fully utilise glycogen stored in the liver.

You can read more about the Double Century Challenge on my GSD Giant training blog (www.gsdgiant.org.uk/

A Change of Lifestyle

By Trushal Pindolia

In January my family and I went to Florida for a winter break whilst we were out there we went to see DR. Weinstein who comes over to the GSD conference every year. I told him about the fact that my triglycerides and cholesterol were very high and asked him if he knew of a way to rectify/normalise them. He said that it was simple; a change of diet was needed. This meant that I had to cut out dairy products (lactose), fruit sugars (fructose) and cut down on carbohydrates. He told me that it would, along with a fair amount of exercise, shrink the size of my liver and slowly bring my triglycerides and other markers in my blood back into normal range.

Over the course of this year my family has changed our diets; I increased my exercise by starting to train for the GSD Giant and playing more hockey. Upon receiving my blood results, only two months after having started the diet, the results showed that both my triglycerides and my cholesterol were reduced significantly from my last blood test. This meant that my bloods were back into the normal range for someone with GSD.

My bloods weren't the only thing the diet helped change; it helped me lose weight more easily than before. I haven't had any problems with the diet so far as it is easy to source lactose free dairy products. Also I take vitamin supplements to help my body cope with not having any fruit.

Here in the UK there are no dietary restrictions at present, however this diet has shown me that for me the American dietary restrictions have worked. I personally would recommend this diet to any GSD type 1a patient who is looking to bring their bloods back into control or to lose weight.

Before starting any new diet it is a good idea to check with both your dietician and consultant first.

Highs and Lows of GSD VI

Hi my name is Caroline and I would to share with you an account of my condition, Type VI Glycogen storage disorder. I very much appreciate that everyone will have different experiences and symptoms may vary, but here is my personal account, followed by comments from my parents.

My childhood memories connected to my GSD include a giant rocking horse, a school trip, a slide and sandwiches. Anyone who was privileged to be treated by Dr Phillip Lee might recall the giant rocking horse in the waiting area outside his room at Great Ormond Street. I think I used to dread the visit but I also looked forward to it, thanks to that Rocking Horse and the promise of an afternoon treat! The school trip occurred shortly after it was suggested I take cornflour as a way of maintaining my energy through the following day. I recall the fuss made over me as the teachers of our Year 6 trip mixed up this strange concoction.

Every year I used to attend an event for trainee doctors – a sort of guess the disease, there was a lovely slide and really nice sandwiches that made the event enjoyable –despite the prodding and poking of the trainees.

Luckily I don't have so many memories of the fear, concern, and sickness side of my condition. I can only put this down to the incredible doctors and my endless love and support from my parents and family. That said I know that I wasn't easy as a child. Sickly and below par on my development, it caused my parents significant cause for concern.

The low point for me of the condition was the research I agreed to be part of under Professor Leonard.

I understood at age 9 accepting to take part would entail 2 days off school and a trip to London, it involved blood tests but after it was confirmed I could choose which arm, and thus would be free to draw, had little other concern!

The 2 days were not the fun I expected (!) with the blood tests every 20mins and to this day I am uncomfortable around needles. Ironically I have since grown up and my career involves work for a pharmaceutical company, where I spent a significant amount of my time focused on injectable medicines.
The other coincidence is that the firm

used to own and make Lucozade – a drink got me through many activities and indeed through the night before the cornflour concept.

Aside from work I am lucky to be living with my partner, Ian, in Teddington a lovely London suburb in SW London. I live a healthy life with check ups every one to two years. There are days I struggle with my energy levels; mainly these follow periods of exercise. Throughout my childhood and early adult years I was above the limits on my weight, in part from my liver but largely due to a bad diet. In 2007 following mystudent/gap year life, I had run out of excuses for a bad diet. I changed this and started exercising. The first plod round the park turned into a half marathon that I ran for GOSH in 2010. I still struggle during the day if I exercise first thing, but then equally so do others. The thirst for fitness combined with my (at the time) new partners love of cycling brought us to the GSD Giant cycle event. I suggested this would be a great way to get to know each other and get fit, all with connections to my GSD. It wasn't easy, the signs with 'shut up legs' being the most appropriate message of the ride, but I made the finished line (relationship in tact!).

My early memories of GSD were for the large part snapshots of visits and trips, but I think the longer-term impact, other than the medical is in my approach to life. I have a can do approach, I am up for trying anything once and lucky to have the support which enables me to see through some of my more crazy ideas. I think this comes from the amount of talk of what I might not be able to do when I grow up due to the condition. I am not denying there have been some difficulties, particularly for my parents, but as an adult I look back and don't wish it away – believing it has had a positive impact on my career, my relationships and my approach to life. I would like to think that in sharing this there is some increased light at the end of the tunnel for parents and children newly diagnosed.

The view of my parents.

Caroline was our first child so we were not really aware that anything was different about her until her sister was born 15 months later. Despite routine visits to the doctor and health clinics no one had picked up on the fact that something was not quite right. She was a "sicky" baby, she was chubby and she did not walk until she was 17 months but she talked early (and has not stopped since), she understood everything, these facts could apply to hundreds of children who just develop at different rates. However once we saw how differently her sister developed we insisted that her condition was investigated. This involved many hospital visits, both local and to Great Ormond Street that were stressful for us but once her condition was diagnosed we felt much happier.

As Caroline got older she was very short for her age and had a very large stomach so dressing her at times was difficult but dungarees, elasticated waists and loose dresses made it easier. She was a very fussy eater but her condition meant that we really had to let her eat what she would, but again there are lots of fussy eaters out there (and she did grow out of it when she was 22 and working in a ski chalet!)

From the start she was a very determined little girl who learned to put a brave face on life and she accepted the hospital visits, tests and comments about her shape but underneath we knew she hated them. She grew out of her "sicky" phase when she was about 4 but up to that point we just became used to carrying a bucket with us wherever we went.

Starting school was a big hurdle but we found a small village school where she thrived. As she got older so she began to grow and her enlarged liver got less prominent. Despite the fact she had less energy than her school friends she was determined to carry on and she was often the "last man standing". She has made it through school, university, gap years (she managed to have two!), numerous trips abroad, glandular fever, cellulitis and now has a successful job. We believe that her condition has made a stronger character and given her an enthusiasm for life.

Cornflour made a big difference to Caroline's life (despite her hatred of it), as did Lucozade, and her doctors at Great Ormond Street. Our advice for parents of children diagnosed with type VI GSD is don't worry if they won't eat wholesome foods, at one point all Caroline would eat for breakfast was Wotsits - when I mentioned this to her consultant he said - "that's fine let her eat Wotsits"!

After that it would be to treat them normally as much as possible, don't mollycoddle them and make sure they always have a backup supply of energy. They will grow up and live a nearly normal life – just look at Caroline!!

Euromac workshop in Madrid

By Andrew Wakelin, GSD V Coordinator

On 11 and 12 July McArdle-ites from seven countries met at the Euromac sponsored Exercise Testing Workshop at the Universidad de Europea de Madrid, Spain. We travelled from USA, France, Germany, Netherlands, UK and of course Spain. Also attending were health professionals, exercise physiologists and researchers from across Europe and further afield. Patients from the UK were David Thompson and Yasar Ayab. Dr Ros Quinlivan, Dr Richard Godfrey, Dr Renata Scalco, psychologist Jatin Pattni and physiotherapist Sherryl Chatfield attended from the McArdle service.

The objective was to educate more professionals about McArdle's, raise standards and explore protocols for standardised exercise assessment ready for future multi-centre studies.

As well as presentations on McArdle's, exercise physiology and the Euromac Registry, there were practical workshop sessions on exercise assessment, and the Spanish team presented their work on strength training. Jatin gave a presentation on the psychological impact of McArdle's.

The McArdle-ites were the subjects in demonstrations, including ways to assess second wind, the 12 minute walking test and establishing a person's VO2 Peak (essentially their level of aerobic fitness). Then there was a chance for some of the patients to do some strength training - under highly supervised conditions. All very interesting and a good networking opportunity with patients and doctors from all over Europe and a few from beyond.

We learnt of Dr Andrea Martinuzzi's great success in running the first walking course in Italy, modelled initially on or own courses. Being run by medics they had a slightly different approach, but also ended the week with some walks. The course members climbed an ascent of 400 metres and were amazed and delighted with their progress and achievement.

David Thompson (UK) tries the strength training (3 lifts in very quick succession, within the limits of immediate ATP).


David Thompson (UK) and Niekie Kleintjens (Netherlands) getting another muscle group into second wind.

Coping with a genetic disorder: 'Why I won't let disease beat me'

Article published in the Jewish News: May 8th 2014

Marilyn Silver talks to Alex Galbinski about how she took control of her life to cope with a rare genetic disorder.

Looking back over her life, Marilyn Silver can't quite believe how much she pushed herself to do despite her illness

Marilyn suffers from an extremely rare genetic disorder called Glycogen Storage Disease Type VII (there are 10 types) – only a handful of people in the UK have been diagnosed with it. It is more common in Ashkenazi Jews, but the exact incidence is not known. Now 63, Marilyn (pictured) was around 11 when she realised things weren't quite right. Her legs always ached and she'd have to stop often to rest whenever she walked.

GSD VII sufferer Marilyn with her husband Laurence

"As a child, I was very slow. I found I couldn't keep up with my friends when walking, especially uphill," she says. "I remember when I was 18 going on a Matzah ramble with a boyfriend and he literally dragged me up a hill to where I could sit. All this was very embarrassing.

"I was also no good at sport. I never ran. I just struggled on – that's just how it was."

It was not until Marilyn was 32 that she was diagnosed with GSD VII, which is also known as Phosphofructokinase deficiency or Tarui's disease, and for which there is no cure.

One night while suffering from flu, Marilyn was so weak she fell halfway out of bed and could barely move. Scared, her husband Laurence rang for an ambulance but in hospital the

doctors initially had no idea what was wrong, or how to treat her. They gave her a massive dose of steroids, which enabled her to sit up, but she had no strength in her muscles. She had many tests, but was discharged without a formal diagnosis after two weeks, although for the first time she heard reference, in relation to her symptoms, to her being an Ashkenazi.

A few weeks later, Marilyn, who lives in Watford, was referred to the National Hospital for Neurology and Neurosurgery in Queen's Square, London. There, she had numerous blood tests and a muscle biopsy and was finally diagnosed. At first, her life did not change, as she had to carry on running a busy home, looking after her two boys – then aged two and four – and later going back to her secretarial career.

"However," she remembers, "I was always tired and in pain. At the end of the day, I would have to sit down for half an hour before I could even walk up the stairs.

"As there was no cure, I just carried on, not realising what damage I was doing to myself. I did much too much, but I didn't want my family to suffer."

The Hemel Hempstead synagogue member was advised that overdoing things could cause the body to react as if in shock. She recalls one particularly bad incident on her first day at a temporary job: "I walked up two flights of stairs and just about fell into a chair. I had to ring Laurence to come to get me and I literally crawled back down the stairs. I remember sitting on the couch shivering for the rest of the day."

Marilyn's specialist, Dr Ros Quinlivan of the Queen's Square hospital, explains: "GSD Type VII is a very rare disorder – there are probably not much more than a handful of patients diagnosed in the UK. The condition is caused by the absence of an enzyme called phosphofructokinase, which is essential for skeletal muscle to burn glucose during exercise.

"The consequence is muscle fatigue, pain, cramping and sometimes severe muscle damage, which leads to a Coca-Cola discolouration of the urine (myoglobinuria). Episodes of acute muscle damage can be very serious and require intensive care with kidney dialysis."

The condition is caused by the inheritance of two abnormal copies of the PFKM gene, one from each parent. "In the Ashkenazi Jewish population, the number of individuals carrying this abnormal gene copy is much higher than the general population," adds Quinlivan, although she says it is not clear why – presumably because of intermarriage. "Carriers do not have symptoms and discover that they carry the abnormal mutation only when they have an affected child".

Marilyn, who had a kidney transplant in 2009 after her kidneys were found to be failing, started to take things easier after her referral to the Queen's Square clinic. Following her diagnosis, her blood relatives were tested for the disorder, but thankfully no one was found to have it.

The grandmother-of-two describes herself as absolutely "frustrated" at her condition,

which caused her to need a walking stick when she was only 50. "I have to think carefully before I do anything whether I am going to be in pain afterwards," she says. "Even the simplest tasks like washing and drying my hair can cause me discomfort if I put too much pressure on my arms."

She adds: "I used to say to my GP: 'My body is my prison'. Now I don't attempt to walk far or uphill and I refuse some invitations. The whole thing is very debilitating, especially when you look alright."

Marilyn is now a co-ordinator for the GSD VII branch of self-help group for AGSD-UK – the Association for Glycogen Storage Diseases – and says people with similar symptoms should discuss them with their GPs.

Her current GP agrees, adding: "GSD Type VII is very rare, and the symptoms can be very non-specific. However, if you recognise the symptoms Marilyn had – particularly if you are from an Ashkenazi background – and where no other diagnosis has been reached despite baseline blood tests being done, it is worthwhile raising the potential diagnosis with your doctor. This is because most GPs are unlikely to consider the diagnosis or be aware of the specialist clinic at Queen's Square."

For her part, Marilyn wants to raise awareness of the disorder. "With this condition, you've got to go at an even keel. The minute you put your foot on the accelerator, that's when you hit trouble. If anyone else is suffering from it, they shouldn't suffer alone."

Sodium Valproate drug trial

The first ever drug trial in McArdle Disease is now getting underway, supported by the Muscular Dystrophy Campaign and AGSD-UK. This study is very promising and it could be a big breakthrough in treatment. It is a Phase II pilot trial involving 8 people in the UK and 7 people in Denmark. Another two people are still needed to be recruited here. There are several exclusion criteria, but if you are interested please email Dr Renata Scalco on r.scalco@ucl.ac.uk to see if you qualify.

Walking with McArdle

Andrew Wakelin, GSD V Coordinator

Our photo was taken on the first of two "Walking with McArdle's" courses in Wales this year, after crossing the railway viaduct over the Mawddach Estuary. We walked down from the foothills of Cadair Idris to the seaside town of Barmouth. Like most of the walks, this gave the opportunity for people to join or leave at various points - thus providing the right level of challenge for a wide range of abilities. That day people undertook sections from 1.5 miles to 8 miles, with terrain from easy level walking on a disused railway to rough paths through woodland and even an invisible path steeply down hill over fields.

We had an excellent week with great weather - just one day we headed west instead if the planned east to almost completely avoid the rain. It was incredibly hot most days. Our programme included some of us summiting a 2,000 feet peak called Tal-y-fan and finished with a challenging four-mile descent of Snowdon - the highest peak in Wales.

Regarding the photo - we walked this route in the opposite direction on "Walk over Wales" in 2010 - there is a photo at the same spot in Stacey's book. Photo thanks to West End Cycles at Llandudno very kindly gave us an electric bike demo as last year and let us ride five models. That day we did a new walk on the coast with an approach along the beach (testing out the resistances of different hardness of sand) to a pub for lunch! Then in the afternoon we got some snooty attitudes from golfers as we tried to negotiate public

rights of way through a golf course. They appeared to have removed most of the signs!

This photo was taken from where we stayed in a very comfortable converted farmhouse, part of Plas-y-Brenin, the national mountain sports centre - looking up Dyfryn Mymbyr (Mymbyr Valley) to the Snowdon group about 7 miles away. Snowdon, at 1085 m (3,560 ft), is the left hand peak of the group of three. We got the rack and pinion train up, did the last 10 m (30 ft) to the summit. Then we walked down (at times scrambled) the sometimes exposed and difficult south ridge and the easy Watkin Path to Nant Gwynant - involving a few short bits of ascent totalling 40 m (130 ft) and a total descent of 1068 m (3,503 ft). The descent took us almost seven hours.

Charlotte from Shoots & Leaves Films was with us for the week, filming for a video on the benefits of the walking courses.

Some great personal achievements by all concerned, some good lessons learnt and a lot of sharing of McArdle's experiences... and a fair bit of laughter. The second course of the year is starting as we go to press.

PST! Update from the Pompe Support team

We would like to introduce ourselves as the new UK Pompe Support Team established at the 2013 AGSD-UK conference. Our aim is to keep you updated on current Pompe news and to advise the AGSD-UK on resources needed within the Pompe community.

What's the PST (Pompe Support Team)?

The PST was set up by a group of patients and carers following the 2013 AGSD-UK conference. During discussions at the conference it became clear that one of the most important factors for patients was contact with others who understood the challenges of living with Pompe. We therefore agreed to set up the PST in order to reach out to others with Pompe to offer support and advice to both newly diagnosed and existing patients. We also felt we could have a role in working on Pompe projects within AGSD-UK that would be of interest and of benefit to patients and their families.

The members of the PST are scattered around the country but we communicate via email and Skype and have met face to face. We cover a wide age range and have different backgrounds, interests and experiences to draw on. Our main goals for the PST are as follows:

1. Offer support to Pompe patients (respecting patient and family confidentiality).
2. Work on projects related to Pompe Disease.
3. Act as a Patient Advisory Board for commercial and non-commercial research.
4. Keep the UK Pompe Community updated on current Pompe News.
5. Advise the AGSD-UK on resources needed in the Pompe Community.
6. Contribute to the AGSD-UK annual conference and Pompe Workshop.
7. Promote International Pompe Day.

Many people report that speaking to others with Pompe can reduce the sense of isolation that can come with living with a rare disease. We know it can be hard to make that first step to contact someone but please feel free to get in touch a member of the PST if you have any questions or need support. We may not have all the answers but we will do our best to help. We also welcome any ideas of projects you would like us to work on.

We are currently engaged with:

- Acting as a review panel for a Genzyme leaflet for newly diagnosed Pompe folk.
- Developing new leaflets for Pompe disease

- Collecting hints and tips for a pocket-size book

We would love to hear from you if you have any questions, ideas or suggestions as to areas that you would like AGSD to develop in relation to Pompe Disease.

International Pompe Day

The International Pompe Association (IPA) is held on April 15 each year, its goal is to foster international awareness of Pompe Disease and the slogan for our day is **"Together We Are Strong".**

Fundraising

As you know having the correct resources and funding so the AGSD-UK can carry on supporting its members is vitally important. Why not think about doing your own fundraising event to support the organisation. You could link your event to International Pompe Day to also raise awareness of the disease and highlight the day itself. The AGSD has it's own team of fundraising advisors, Naked Fundraising, who would be happy to help you further:
Email: fundraising@agsd.org.uk
Telephone: 01489 877 319

Pompe hints & tips

Following last year's conference we thought it might be useful to produce a helpful "Hint and Tips" booklet for both newly diagnosed people as well as existing patients. The idea is for it to be a booklet for patients from patients; the McArdle team has done something similar and it has been well received by their group.

To produce a quality, beneficial product we need your help. We would be extremely grateful if you would send us some examples of everyday things you do to make your life a little easier?
Some examples are:

- Using a perching stool for ironing, washing the dishes, in the shower etc.
- Using a long handled grab stick to pick things up from the floor
- Re-arranging kitchen cupboards so the items you use most are within easy reach and don't require you to bend down
- Using a toweling dressing gown to dry yourself
- Buying grated cheese
- Using an electric whisk rather than hand whisk
- Invest in a mobility scooter to save energy when out and about for periods of time
- Use a frame / handles to get on and off the toilet etc., etc., etc.

We're interested in all types of ideas from those that don't cost anything to those that will require some investment. We would be grateful if you could send your ideas to us and we'll further discuss them at the Pompe Workshop in October. You can send ideas to any member of the team, but Amanda will be collecting all your ideas for the publication.

We look forward to hearing from you and are keen to work closely with our Pompe community. We would welcome any new members to our Pompe Support Team, so please contact us if you would like to get involved.

Amanda, Ben, Angela, Theo, Sam and Emily - PST!

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Or contact the group via the AGSD-UK office (inside back cover)

Together We Are Strong

International Pompe Day

Low-intensity exercise programme for Pompe Patients

Experience of supervised low-intensity exercise programme for Pompe Patients undergoing Enzyme Replacement Therapy (ERT) infusions at Queen Elizabeth Hospital Birmingham.

Nicola Condon, Senior Physiotherapist, QEH Birmingham

Pompe Disease (Glycogen Storage Disease Type II, or Acid Maltase Deficiency) is characterised by a deficiency in an enzyme 'acid a-glucosidase' which breaks down glycogen, a form of sugar that is stored in muscle cells. As a result, too much glycogen builds up in the cells which weaken muscles throughout the body that support balance and mobility.

In late-onset Pompe disease, muscle weakness is seen in the proximal muscles (nearest to the trunk) of the lower part of the body (legs, hips, pelvis, and spine), and of the upper part of the body (neck, shoulders, and upper arms) and the muscles used for breathing.

Exercise can have many benefits for people with Pompe Disease, improving flexibility, mobility and help increase energy levels. Recent evidence suggests that patients with late-onset Pompe disease receiving Enzyme Replacement Therapy (ERT) who engage in exercise training can benefit from increased muscular strength and help maintain their functional level for longer. However, the wrong type of exercise or pushing

too hard can cause damage to the muscles and increase fatigue. Exercise should be individually tailored to a person's needs, abilities, and disabilities so guidance from a specialist Physiotherapist is essential.

The aim of ERT is to increase the level of enzyme within the body to clear these substances that would otherwise build up in the body.

Exercise during an ERT infusion has been thought to potentially increase the effectiveness of ERT through increased blood flow to the exercising muscles. However, as yet has not been proven and there has not been any adverse reactions reported.

The Inherited Metabolic Disease (IMD) Service at the Queen Elizabeth Hospital Birmingham provides a clinical service for the diagnosis, assessment and treatment of adult patients with IMD's and consists of a multi-disciplinary team led by Consultant's in IMD.

Many patients reported access to Physiotherapy locally as being patchy, sporadic and often Physiotherapists lacked knowledge of their condition. Some reported it would be beneficial to have seen a specialist Physiotherapist early to help to provide confidence with starting exercise programmes for the first time.

In 2012-2013 the protocol for commencing ERT dictated that patients should receive treatment in hospital fortnightly for the first six months. The typical infusion period is over four hours and supervised by IMD Specialist Nurse, monitoring for any signs of infusion related adverse effects/reactions. Normally patients are in a day-case area and did very little over the four hour infusion.

We were keen to use this time as an opportunity to introduce exercise and allow one to one time with the Specialist Physiotherapist. Over this period there were two patients commencing ERT and newly diagnosed with Pompe Disease. These patients were asked if they were keen to participate in an exercise programme during the time of their infusion. None were engaged in exercise other than their normal activities of daily living prior to these sessions.

One hour into their ERT infusion patients were walked to the Physiotherapy department with the IMD Specialist Nurse for their 45minute individualised exercise programme that was set and supervised by the Specialist Physiotherapist. Both IMD Specialist Nurse and Specialist Physiotherapist were present throughout the session and compliant with the recommended resuscitation training.

As part of the baseline assessments and on-going management within the clinics, assessments were made of respiratory status, muscle strength (myometry), cardiology, endurance walking tests and functional ability.

Exercise sessions consisted of a variety of exercise types set at a level and intensity that patients found to be somewhat hard but not causing excessive fatigue. They were encouraged to monitor their breathing and heart rate in relation to their exertion. Exercises were set to target areas of specific

weakness or compensations following the Physiotherapy assessment, for example;

- Submaximal aerobic exercises; in the form of recline exercise bike, seated cross trainer, Wii
- Postural and core stability exercise – involving correction and guidance from physiotherapist focusing on muscle activation around the pelvis, trunk and shoulder girdles.
- Equipment was used to progress these exercises using gym ball, small weights and resistance bands.
- Functional exercises – These were set to what each patient found the most challenging and the task was broken into smaller parts.
- Examples are sit-to-stand practice, stepping on/off variety height blocks, side stepping, balance exercises.
- Stretching regimen

These Physiotherapy sessions allowed progression and correction of exercises and an opportunity to monitor response to exercise. Patients felt supported to continue an individualised home exercise programme (HEP) which they were able to take away and continue independently.

Following the six month period both patients had attended six one to one exercise sessions and were continuing independently with a HEP. All patients commented on how much they enjoyed the sessions and the confidence it gave them to continue independently and could feel an increase in their activity levels. One also reported a significant weight loss.

Both patients showed improvement in distance walked in their endurance test (the six minute walk), with increased walking distance of 12% and 43% in six minutes. Their muscle strength measurements remained the same. Neither patient experienced any adverse advents during or following their sessions.

Unfortunately, follow-up support in their local community was lacking for both patients and some of these gains were lost after one year.

In 2014 the protocol for commencing ERT has changed to allow only the first three infusions being given in Hospital before continuing into the community. At Queen Elizabeth Hospital we are continuing with Specialist Physiotherapy input / exercise during these three infusions.

Our experience has reinforced the importance of ensuring appropriate individualised advice and exercise programmes early in order to prevent deconditioning as a result of regular physical activity. There is also the need for local community Physiotherapist to maintain advice and input once they are no longer under the regular review of the IMD centre.

Patient One was diagnosed with Pompe Disease at the age of 47. Very active at school, was part of the

running team and enjoyed team sports. Last three years noticed waddling walk, now needing both hands on a bannister to climb stairs, short of breath on exertion and uses wheeled walker for balance when walking. (Ultrasound of the diaphragm showed equal movements, but had mild nocturnal hypoxia. Lung function tests identified global weakness of respiratory muscles and sleep studies found low perfusion.)

Patient 2 was diagnosed at the age of 40. Symptoms were fatigue and shortness of breath on minimal exertion. Presented with a waddling gait and unable to rise from a chair without using arms. Patient had suffered a recent bereavement along with the diagnosis and was struggling with this loss and low in mood.

Pompe news in brief

Abdullah wins Gold at London Youth Games. By Saam Amerat

At this year's London Youth Games Hackney excelled in the swimming again! Abdullah brought home 4 medals. Gold because Hackney won overall, Silver for individual 25 meter race, and Gold in two relay races.

Last year Hackney's came out on top in the swimming events, so this year is Hackney's second year of triumph.

Abdullah was 8 years old in both years. Last year it was on the 4th of July his 8th birthday, and this year it was on 3rd of July the day before his 9th birthday.

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

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

AGSD

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