

GlysteN

Glycogen Storage News

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AGSD-UK Development 2011

Building on sound foundations

As you will no doubt be aware, 2010 saw a number of significant changes to the structure and governance of the AGSD-UK that have allowed us to move more confidently into the future. We had realised that the charity demands were becoming too great for the current volunteer staff, and that we needed a professional team to handle the increasing workload. Under the leadership of our Chairman, Andrew Wakelin, the charity was converted into a company limited by guarantee; this provides protection to all the membership as well as its governing body, and allows the trustees to develop the AGSD-UK in ways that were not available to them in the past. Given our new status we have a number of further developments to announce and some exciting events and projects to tell you about.

Our London conference attracted nearly 200 people in November, showing that there is increasing interest in the AGSD-UK as the essential forum for GSD patients, families and professionals.



AGSD-UK Office



From January 1st 2011 the AGSD-UK occupied a new office in Hampshire situated between Southampton, Portsmouth and Winchester. Initially we are sharing office space with an established graphic design company. Eventually, within the first year we hope, we plan to take the lease of a small office within the same centre, so our postal address and phone number should not change. Our new contact details are given on the back page

New Image

Our office-sharing arrangement with the Paul Martin Design Company (www.pmdc.co.uk) will give us access to all of their graphic design services. We are already considering how we might improve the image of the charity through our publications as well as the Internet, so this close relationship will be very useful and will hopefully continue to benefit the charity over subsequent years.

Staff

Shortly after our annual conference in November I was successful in my application to take a staff role within the AGSD-UK as its Development Director. This role takes on a wide range of responsibilities to develop the charity in an organised but pragmatic manner, it is a huge challenge and requires close liaison with the trustees and the volunteer GSD Type Coordinators, newsletter editors and fundraisers.

We are currently in the process of recruiting a part-time Administrator, based at our new premises. The Administrator will take over much of the charity's day-to-day administration as well as helping with event organisation – particularly our family conference.

The funding from Genzyme for our Family Support Nurse has recently been renewed so Joan Fletcher continues to work on our behalf for Pompe families from St Mary's Hospital, Manchester.

Steve Saunders was employed in November as our part-time fundraising consultant. Steve has a long history of fundraising and he has already organised a number of events for us: for example "Walking the London Marathon" and "Abseiling down Guy's Hospital Tower". See his articles on fundraising in this newsletter and on the website. Steve will also be helping with other fundraising events and grant applications.



Projects and Activities

In order to provide an improved service to the GSD community we have a number of projects in mind for which we will require help from our membership.

We want to improve our GSD information leaflets and this will require the latest understanding of the symptoms, treatments and the types of support required by patients, families and carers. We want to understand the requirements of the GSD community and how they are being met by the NHS, social Services and other statutory bodies. We also want to understand the geographical variability of services so that we can campaign to raise the level of care to the highest standard possible.

Part of this process will involve questionnaires or interviews to survey the GSD community and perform an audit of services; this will be sent to patients, families, carers and health professionals. To give us a head start you may like to write to me with any ideas of topics that may benefit from further investigation.

Fundraising

Developing the charity will only be possible if we are able to generate funds to support our costs. Steve's organised events should easily cover his own costs and provide additional funds for the charity. Lucca Venditto has put a tremendous amount of effort into organising an AGSD-UK cycling event taking place in September. We are hopeful that his "Pompey to Pompeii Cycle Challenge" will raise over £60,000 net for the charity.

We receive many donations from supporters throughout the year and for those we are extremely grateful. We would like to encourage everyone with an interest in GSD to think of the charity throughout the year because they are our greatest resource. Many of our supporters are our expert families who have a great knowledge of the impact of GSDs and can tell stories to help others understand the condition and also reach out to potential donors. Fundraising should ideally have the joint aims of raising both GSD awareness and financial donations.

Steve Saunders can offer advice if you would like to hold a small fundraising event in your home or your locality. A coffee morning can raise £100 by charging 20 people £5 for a drink and a piece of cake; multiply that by 200 of our members and we could potentially raise £20,000 in one day!

February 28th 2011 is Rare Disease Day, so why not plan a small event like a short organised walk followed by a bowl of soup for a fiver...

Volunteering

Whilst the professional team and trustees will coordinate the core business of the charity, we must not be allowed to lose sight of the fact that there are many stakeholders (patients, families, carers, healthcare professionals, teachers etc) who benefit from and contribute to the work of the AGSD-UK. So whilst we will try to keep you informed of our work through our print and on-line publications, we would encourage all members to consider how they might make a contribution. For example:

- Write letters or personal stories for our newsletters
- Suggest topics for investigation
- Give feedback to the staff or trustees about the work of the charity
- Take part in one of our fundraising activities
- Hold your own fundraising events
- Advertise our organised events to your family, friends and colleagues
- Volunteer your services to the charity

2011 is set to be another difficult year for many of us and chasing charitable donations will be hugely challenging in this climate of diminishing funds. So please help us in our mission to improve the support to families living with a GSD.

Best Wishes for 2011

Allan Muir, Development Director



The National Alliance for people with rare diseases & all who support them

New Rare Disease report highlights experiences of patients and families living with rare conditions

A new Rare Disease UK report highlights a number of worrying issues experienced by patients and families affected by rare conditions. The report, "Experiences of Rare Diseases: An insight from patients and families", deals with the views and experiences of 600 patients and families affected by over 100 different rare conditions on a wide range of topics ranging from research to diagnosis, and access to care, information, support and treatment.

Although some patients and families indicated positive experiences of timely diagnosis and good quality care and support from the NHS, unfortunately this is not the case for the majority of patients and families with rare diseases.

There are over 6000 recognised rare conditions each with different symptoms and prognoses, yet the publication highlights how patients and families frequently face common problems. Some of the problems highlighted by the report include:

- Significant delays in diagnosis.
- Patients being misdiagnosed (often multiple times).
- Patients having to visit multiple specialists before receiving an accurate diagnosis.
- Difficulties in accessing information and support.
- Fragmented and poorly coordinated care.
- Patients and families having to attend multiple hospital appointments long distances from home.
- Problems during transition from paediatric to adult services.
- A lack of effective treatments.
- Inconsistencies in access to medicines.
- A lack of information and opportunities to be involved in research.

Rare Disease UK is the national alliance for all those with an interest in rare diseases brought together to campaign for a strategy for rare diseases.

The Chair of Rare Disease UK, Alastair Kent said:

"Many people living with rare diseases and their families have to go through years of medical tests and procedures before an accurate diagnosis can be made. Not only is it often a battle to get an accurate diagnosis, patients and families then struggle to find out the medical impact of a condition and how to manage it, on top of having to cope with day-to-day life without adequate support.

"The results of RDUK's survey hammer home the need for a co-ordinated UK-wide strategy for the diagnosis, treatment and research of rare diseases. This is a hugely important issue that needs to be tackled; patients and families should not have to face an inequitable level of care from the NHS because they have the misfortune of their condition being rare."

A copy of the report is available at: www.raredisease.org.uk/documents/RDUK-Family-Report.pdf

For information, or a copy of the report, please contact Stephen Nutt: stephen@raredisease.org.uk 020 7704 3141.

Register your place at the Rare Disease Day Parliamentary receptions

Please note: due to security regulations and to help us with planning you must register in advance to attend the receptions.

To register please send your name(s), address and contact email/telephone number to:

Scottish Parliament event – 22nd February 6:30pm – 8:30pm
scotland@raredisease.org.uk, Natalie Frankish - 0131 651 4805

Westminster event – 28th February 4pm-6pm
assistant@geneticalliance.org.uk, Paula Barbosa – 020 7704 3141

Northern Ireland Assembly event – 3rd March 12pm-2pm
stephen@raredisease.org.uk, Stephen Nutt - 020 7704 3141

Welsh Assembly event – 16th March 12pm-2pm
cope@cf.ac.uk, Buddug Cope - 02920 687 646

We encourage attendance from all with an interest in rare diseases including patients and families, patient organisations, clinicians and health professionals, researchers, members of the pharmaceutical industry etc.



AGSD-UK Conference Reports

For the 26th AGSD-UK Conference held at the Royal Pharmaceutical Society GB on 20th and 21st November 2010

“A most successful conference with some 140 family members and 60 professionals”

Type I and III Workshop Report by Sue DelMar

Saturday 20th November

Some sixty patients and professionals came to the Type I and III workshop which is by far the most we have ever had.

Dr Priya Kishnani, Duke University USA gave an update on the Management Guidelines for Type III for the USA which were been published this summer*. There is a summary in the USA AGSD conference 2010 report.

Notes on Dr Kishnani's presentation to Types I and III Workshop after her update

- Presently there is no newborn screen available for GSD I or GSD III, but research is occurring in this area. Early blood spot diagnosis should decrease the risk of developmental delay from unrecognized hypoglycaemia and improve long term prognosis
- A high protein diet (3 g/kg) is needed for type III GSD as it has been associated with decreased CK concentrations and improved muscle strength.
- Exercise is beneficial for both type I and type III GSD. In type I GSD, exercise is the only way weight loss can occur safely. In type III GSD, moderate exercise improves bone density and strength. Maximal exertion (sprinting, maximal weight lifting) is not recommended in GSD III.
- Vitamin D deficiency is almost universal in GSD I and common in GSD III. It is critical to maintain normal vitamin D levels since people with GSD I and III are at risk for osteoporosis (There is a summary of Dr Ann Boney's presentation on Vitamin D in the USA AGSD conference 2010 report.)

*I HAVE A COPY OF THE COMPLETE DOCUMENT IN CASE ANYONE WOULD LIKE TO READ IT
(Sue DelMar gsdaddresses@btinternet.com)

Vitaflo latest update

Representatives from Vitaflo International reported on the latest developments with Glycosade. These details can also be found in the USA AGSD conference 2010 report given by Dr Weinstein, University of Florida.

We are very grateful to Vitaflo International for their generous donation of £2000 towards the conference.

Report by Dr Elaine Murphy, Charles Dent Metabolic Clinic

Dr Elaine Murphy from the Charles Dent Adult Metabolic Unit, National Hospital for Neurology and Neurosurgery, London presented two cases of adult patients with glycogen storage disease who were having problems controlling their blood sugar levels.

The first adult patient had GSD Ia and was found to have a high glucose level at a routine outpatient visit. Further testing of glucose levels throughout the day (by the patient using a home glucometer) showed that all blood glucose levels were high throughout the day (15-20 mmol/L), even before meals, and therefore the patient had diabetes. There was also a strong history of diabetes in other older members of the patient's family. A dietary history revealed that the patient was drinking up to five 500ml bottles of lucozade a day in order to 'keep up blood sugar levels'. The patient was not taking any regular cornstarch during the day.

We advised the patient to stop all lucozade (and other sugary drinks) and gave some dietary advice regarding longer-acting carbohydrates to have with meals. Blood glucose levels improved considerably over the next few days (to 7-9 mmol/L).

The second adult patient had GSD VI and was very well. A few times a week the patient noticed that a couple of hours after breakfast blood glucose levels were a little low (around 2.5 mmol/L) and the patient felt hungry and a bit shaky. Otherwise blood glucose levels throughout the day were normal. A dietary history revealed that the patient was having a bottle of lucozade for breakfast. It was felt that the lucozade was increasing blood glucose levels rapidly and that the body's normal response to this (an increase in insulin levels – the hormone that returns high blood glucose levels to normal) was a little over-exuberant leading to a subsequent drop in blood glucose levels below normal. This can happen to people without any medical problems (young slim women are said to be particularly susceptible) and is called 'reactive' hypoglycaemia. The patient was again advised to stop drinking lucozade (and other similar drinks) and to have a more balanced breakfast to prevent this problem.

Type I and III Workshop Report continued

Learning points: Lucozade and other similar sugary drinks contain very short acting sugars and have very little role in the regular long term management of GSDs. They result in a very fast increase in blood sugar levels which may rise well above normal levels. These drinks should only be used in the very short term if hypoglycaemia (a very low blood sugar level with symptoms) is imminent. Otherwise a good diet and regular cornstarch as recommended by your GSD doctor / dietitian is a more appropriate way to maintain blood glucose levels within normal limits (not too low and not too high!). Remember that a glucometer can be used to check for high as well as low blood glucose levels. As a general rule if your blood glucose level is in double figures (>10mmol/L) two hours or more after your previous meal then this may be too high and you should talk to your local GSD team for further advice. This is particularly important if you have a family history of diabetes as you may be more susceptible.

Conference in Milan

Professor Smit, University of Groningen Holland, reported on the conference in Milan in October where many doctors and GSD Association presidents and representatives met to discuss future collaboration between the various groups.

There will be a follow up meeting in Spain in June 2011 at which representatives from AGSD(UK) hope to attend.

An Update on Gene Therapy , Dr. David Weinstein, University of Florida

Patients with GSD types 0, I, III, VI, and IX are now doing well, and the prognosis for children and adults with these conditions is outstanding. At the University of Florida, 32 children have now been born to mothers with these conditions, and 2 patients with GSD III are now grandparents. While there have been tremendous advances over the past 2 decades, the ultimate goal is to cure the disease. Several techniques are being investigated at the University of Florida and at Duke University aimed at curing the disease:

- Traditional gene therapy using a viral vector to insert the gene
- Use of non-viral carriers to insert the gene
- Infusion of normal mature liver cells
- Use of non-embryonic stem cells to regenerate the liver with normal cells

Gene therapy work for GSD is also being performed in France, Italy, and Israel.

Dr. Weinstein showed slides of both affected mice and puppies compared with their normal siblings demonstrating the severity of the disease. Gene therapy was first successfully performed in mice with GSD Ia by Dr. Janice Chou at NIH who demonstrated normalization of growth and laboratory studies following gene therapy in mice. The studies were subsequently performed again in mice by Dr. Dwight Koeberl at Duke University. Since people physiologically are different from mice, studies were then transitioned to dogs in 2005.

GSD type Ia occurs naturally in Maltese dogs. Without treatment, all of the dogs die. Even with medical therapy, no dog with the disease had survived for more than 4 weeks when this work started.

Update from the University of Florida:

Dr. Weinstein began his presentation on the University of Florida dogs by emphasizing that this is a huge team effort with over 20 investigators and 80 students involved in the project. In addition to the GSD research team run by Dr. Weinstein and Laurie Fiske, there is a gene therapy team run by Dr. Thomas Conlon, and the veterinary team run by Dr. Andrew Specht and Maggie Struck.

Gene therapy was performed on a dog with GSD type Ia (Dulce) in Florida for the first time on September 11, 2007. Within 2 weeks, the duration of fasting markedly increased, and visually the dog improved with decreased hepatomegaly, increased energy, and improved growth. The effect of gene therapy waned so a second gene therapy was performed in January 2008. Following this treatment on Dulce the lactate concentration normalized, and she was able to fast for 9 hours without development of hypoglycemia. All glucose support was stopped at 6 months of age, and Dulce had no problems clinically while weaning off therapy. A liver biopsy 6 months after the 2nd gene therapy dose demonstrated 7% activity. Dulce remained off of therapy for 19 months (21 months following the last gene therapy treatment), but elevated lactates subsequently developed when she went into heat. As a result, low dose therapy was restarted, and Dulce had a third gene therapy treatment in February 2010. Dulce continues to do well overall at 3 years of age. She has no adenomas or evidence of complications.

Type I and III Workshop, continued



New puppies Tucker and Jasmine were born April 9, 2010. Gene therapy was performed at 3 6 hours of life using a modified vector, and the response was even more dramatic. By 2 weeks of age, puppies were able to fast for 6 hours (baseline 45-60 minutes), and glucose support discontinued at 3 weeks. A second gene therapy treatment was performed at 8 weeks, and the dogs are doing well at 7 months of age.



Tucker with his Mother

Next Goal: Gene Therapy in an Older Dog with GSD (already selected and called Ginger)

Since most children and adults will be treated outside of the newborn period, therapy has recently been attempted in an older dog which was medically treated for 2-1/2 months. Without the gene therapy, the dog struggled to maintain her blood sugars, and rapid decompensations occurred. Poor growth also occurred demonstrating the impact of the gene therapy. It is too early to assess the response to this treatment.



Future of Gene Therapy

- Gene therapy appears to be a promising therapy for the future in GSD Ia
- Gene therapy has been successfully performed by Dr. Chou in mice with GSD Ib
- Gene therapy trial in humans with GSD II in Florida has been approved by FDA
- Gene therapy trials beginning for dogs with GSD III in Florida and at Duke

Sunday 21st November

Renal Problems which can occur in Type I patients, Professor Smit

Enlarged kidneys can lead to hyperfiltration which means that the blood flow passing through the kidney is increased.

This makes for increased pressure on the kidney and ultimately may cause Glomerulosclerosis.

The filter system in the kidneys acts like a sieve. Glomerulosclerosis occurs when the sieve is damaged ie its holes get larger. This allows larger protein molecules to pass through into the urine which causes proteinuria. Initially, this can be detected by testing for albumin in the urine. There can also be a problem with tubular dysfunction after filtration which can lead to the molecules not being reabsorbed from the urine. This problem in absorption may lead to an increase in calcium and a decrease in citrates causing hypercalciuria and hypocitraturia.

The treatment for these is as follows:

Allopurinol will reduce the level of Uric Acid and thereby reduce the risk of crystals of Uric Acid. Prescribing Citrates will increase the amount of Citrate in the urine and prevent from Calcium stones by preventing the stone crystallisation.

An ACE Inhibitor will (*angiotensin-converting enzyme inhibitor*)

- *Reduce (Micro) Albuminuria
- * Prevent an increase in Macroalbuminuria
- * Maintain Renal Function

An ACE Inhibitor enlarges the diameter of the blood vessels in the outflow tract of the glomeruli and therefore decrease the pressure of the kidney filter system as a whole. It also contributes in the prevention of the development of glomerulo-fibrosis

NOTES

- ACE inhibitors are generally prescribed (if indicated) to children from age 3 years on.
- Stop ACE Inhibitors before and during pregnancy

The test for Kidney function: **GFR Test (Glomerular Filtration Rate Test)**

Professor Smit also commented on the fact that with gene therapy not all liver cells are "cured", but in the experience with mice and dig models so far about 20%. For the maintenance of normal blood glucose concentration this appears to be sufficient. But the question remains to solved whether this percentage will prevent the "not-cured" liver cells from developing long-term complications like liver adenomas.

At the tail-end of November the 2010 AGSD-UK Conference finally revisited London after many years touring the country. The setting of the RPS was chosen to tie-in with the 'Pompe Steps Forward' Symposium for healthcare professionals, organised by Genzyme (producer of Pompe ERT Myozyme/Lumizyme) which ran just prior to the Conference. Turnout was the best to date, reflecting in terms of the Pompe section of the audience what has been an eventful year.

The scope and scheduling were ambitious, with perhaps the widest range of speakers and topics I've seen so far, including John Crowley – subject of *Extraordinary Measures* – as an after-dinner speaker, and the welcome introduction of patient presentations. The following is organised under three sections: Research/Corporate, General, and Advocacy in order to provide ease of reference.

Research and Corporate

To start with, an **Update on the Genzyme Pompe Programme, which was given by Dr Khazal Paradis** – Senior Vice-President of Clinical Research at Genzyme Europe. Dr Paradis may be familiar to some of the Pompe Community, having been closely involved with the approval process of Myozyme in Europe.

He gave some brief statistics on the global reach of Myozyme today – 1,400 patients enrolled in 46 countries worldwide (only half of which grant charitable access to the drug however.) Some update as to the manufacturing of Myozyme was outlined: the drug is currently produced from two 4,000 litre bioreactors in Geel, Belgium, though in the pipeline is a third reactor which will be fully functional next year, and the possibility of a further two reactors – all to cope with increased global demand.

Of special interest was his discussion of the development of neo-rhGAA – a second generation ERT that by increasing Mannose-6 phosphate moieties in ERT has been shown to dramatically increase glycogen breakdown in Pompe mice. He suggested that neo-rhGAA can be up to five times as effective at breaking down glycogen as Myozyme, though he stressed that effectiveness and tolerance in terms of human subjects was as yet unknown.

Jayne Gershkowitz, Director of Patient Advocacy and Public Policy at Amicus Therapeutics, gave an update on **Pharmacological Chaperone Therapy for Pompe Disease**. Those unfamiliar with Pharmacological or Enzyme Chaperone Therapy (ECT) may like to consult my report from the 2008 conference in issue 15 of the Bulletin. The programme for a Pompe ECT (AT2220) at Amicus has met with mixed fortunes over the past year. Unfortunately in May 2010 the decision was undertaken not to pursue AT2220 as a standalone therapy. This was due to the aborted US-based preclinical trial in which several subjects experienced adverse effects such as increased muscle weakness, resulting in the trial being put on hold by the FDA.

Dr Barry Byrne, of the Powell Gene Therapy Center at the University of Florida, delivered an Update on Gene Therapy for Pompe.

The development of Gene Therapy has been slow and beset with setbacks, yet Dr Byrne outlined some positive developments of late – including a trial of GT in tandem with ERT which was granted approval in August 2010. The aim is to study effects upon respiration, which Dr Byrne described as being the most pressing issue for Pompe patients. A US-based Phase I/II clinical trial is now being proposed. This multidisciplinary use of GT in tandem with ERT is the direction GT for Pompe would seem to be heading.

For a comprehensive description of this trial, now recruiting, I would refer you to the recorded AMDA Webinar at www.amda-pompe.org/index.php/main/webinar/.

Dr Brian Bigger, from the MPS Stem Cell Research Group based at the University of Manchester, spoke on Antibodies to Delivered Enzyme in Pompe and related LSDs. He highlighted the fact that antibody production can often be an issue in the treatment of LSDs, but that various methods are being developed in order to counteract this. Whilst he emphasised that most of the current data surrounding this issue comes from patients with MPSI, II and IV, the findings so far should also be applicable to Pompe.

Dr Nina Raben, Staff Scientist at the National Institute of Health (NIH) in Maryland, USA delivered a talk on Autophagy and Pompe Disease. Autophagy is the process of cell-component degradation through the lysosomal machinery – with this in mind she quoted cytologist and biochemist Christian de Duve - who first referred to lysosomes as 'suicide bags'.

Yet despite this dramatic tag, autophagy plays a standard role in cell growth, development and homeostasis, helping to recycle worn-out or toxic cellular products. It can also function as an inbuilt survival mechanism, kicking in when the body experiences a deficiency of nutrients. In such circumstances the cell can effectively consume its own reserves in order to survive.

Pompe Workshop, continued

Dr Raben also noted that cardiac muscle responds much better to ERT than skeletal muscle and that the pathology of glycogen build-up in the latter was in general less well documented. Crucially, ERT will not be successful in restoring skeletal muscle fibres which have already significantly degraded. Trials in Pompe mice have also confirmed that skeletal muscle resistance to ERT is associated with the presence of large areas of undigested autophagic material. Autophagy then would seem to play a key role in the depletion of skeletal muscle in particular for Pompe patients.

For a detailed discussion of the role of autophagy in Pompe disease, I would again refer you to the excellent AMDA Webinar at: www.amda-pompe.org/index.php/main/webinar/.

Dr Linda Van Den Berg, from the Pompe Research Team at Erasmus Medical Centre in Rotterdam, gave a talk on **Osteoporosis and Effects of Training in Pompe Disease**. She began by outlining the fact that bone is a living tissue which is subject to continual remodelling. Bone diseases therefore result from an imbalance in the regulation of such remodelling. Milder cases of bone loss tissue loss may be classified as osteopenia, and more severe cases as osteoporosis.

Aside from family history, lifestyle factors such as calcium deficiency, lack of sunlight and a sedentary lifestyle are often associated with osteoporosis. The latter in particular may be more prevalent in patients with conditions such as Pompe than in the average population.

Where possible, exercise can reduce risk of conditions such as osteoporosis by increasing muscle function and fibre size, alongside benefiting heart and lungs. A further study on exercise training for Pompe patients is due to start in January 2011, looking into endurance, strength and core stability. Preliminary results are expected by the end of 2011.

Dr Nicolai Preisler, from the Neuromuscular Research Unit at the National University Hospital in Copenhagen, delivered a talk on **Exercise Tolerance in Pompe Disease**. He focused on aerobic forms of exercise, specifically cycling, which he stated would be an effective option for many Pompe patients as it could be carried out at home (on exercise bikes presumably.) Benefits of exercise he listed included reduced risk of cardiovascular disease, Type II diabetes, high blood pressure and stroke.

However the capacity of Pompe patients for exercise is often limited by depleted skeletal muscle and lack of 'fuel', as Dr Preisler termed it - though he added that reduced ventilatory capacity is 'not a contraindicator for capacity to exercise'. He laid out that the most important part of a training programme is in the individual tailoring of it to the needs and abilities of each patient. Duration and frequency of training should be carefully considered, along with the degree of restitution (resting) time required. He pointed out that it is always 'important to listen to the signals your body is sending you'. Patients should always be aware of excessive muscle pain in particular.

But he concluded by stating that 'it is always safe to exercise' (given, I would've thought, that the above is taken into account) and that regular exercise could lead to increased glycogen clearance. This is however as yet unconfirmed, though Dr Preisler will be conducting a study into exercise for Pompe patients in 2011.



General

After dinner on the Saturday the audience was addressed in a motivational speech by **John Crowley**, President and CEO of Amicus Therapeutics and subject of the recent CBS-Harrison Ford movie *Extraordinary Measures*. John delivered a highly eloquent and often candid talk, reflecting on the history behind the story of the film, which involves the search for a 'cure' for his two Pompe-afflicted children, Megan and Patrick. Those who have seen the film or read the previous issue (17) of the Pompe Bulletin will already be familiar with the details.

It was rewarding however to receive a first-hand account, with John reflecting on many of the challenges that were faced both personally and professionally. Despite the fact that the Novazyme ERT that John helped pioneer was not ready for commercial development and as such was not further pursued, he stressed that the efforts made fed into and were worth the eventual result in terms of Pompe ERT and that the world of start-ups for rare diseases required of its protagonists vision, tenacity and a certain amount of obstinacy in the face of often seemingly insurmountable obstacles.

John remains optimistic and upbeat about the future for Pompe treatment, and he echoed some of the speakers earlier in the day by stressing that a complementary approach involving a range of newly emerging therapies would be most likely to characterise the future Pompe landscape.

Pompe Workshop, Continued

Patient Advocacy and Support

Sam Murduck gave a frank and moving talk on the Sunday morning which, for me and others I spoke to, was one of the highlights of the weekend. Sam, 32, was diagnosed with Pompe earlier in 2010 after a not-atypical experience of being misdiagnosed (with Limb-Girdle Muscular Dystrophy.) Having had a normal and fairly sporty childhood she first experienced symptoms of muscle weakness in 2001. She emphasised a phenomenon that may be all too familiar to sufferers of progressive conditions such as Pompe - that 'you don't really notice it at first, because you naturally adjust and adapt.'

After being told by doctors that her problems were merely 'growing pains' or related to stress, she had various tests which proved inconclusive before finding that things took a turn for the worse around the time of her father's death in 2005. She went blind for 10 minutes, and the shock of the experience led her to revisit her doctor - at which point she received the misdiagnosis, before finally being diagnosed with Pompe.

Familiar signs and symptoms she discussed were difficulty with walking, lack of strength in legs and lower body, stiffness in neck and shoulders, being often fatigued and waking up tired and groggy, often with a severe headache (arising from poor respiratory function during sleep.) These latter symptoms were however relieved by night-time ventilation which she now uses routinely.

She went on to expand upon the 'emotional rollercoaster' she described in the Bulletin, discussing how she was initially 'paralysed by fear', that she often cried and felt generally miserable. She also experienced anger that people had not believed in the truth of her symptoms prior to her diagnosis. She described how she also often felt alone at first, but added that through discovering the existence of a Pompe community, via events such as the AGSD-UK conference and services such as GSDNet, she has felt far better informed and connected.

She stressed that whilst she could be stubborn in terms of her own independence, 'accepting help from other people has been important'. She perfectly described the grey area that Pompe patients can find they lie in in terms of other people's perception – often appearing fairly normal upon first glance, yet beneath the surface experiencing a set of very real symptoms. Those with disabled parking badges for example may be well aware of this problem.

Sam has been very proactive in terms of investigating the range of possible services on offer. As well as receiving medical support in the form of ERT, night-time ventilation, neurological and respiratory consultation, she has visited her GP, seen an OT and dietician (who sadly was not at all knowledgeable about the condition and resultantly of little use) and has also appeared in both the above-mentioned article and a video for the benefit medical students at Southampton University.

Inspiringly, Sam still works full-time as an Occupational Therapist which, whilst often exhausting, is undoubtedly rewarding for her. Ironically however her employer, the NHS (National Health Service) has often been less than understanding of her condition. This only reinforces the fact that better awareness of rare conditions is required in the general workplace. She touched on the future and her desire to become a parent. Sam remains optimistic.

Following Sam was Cori (GSD Type III) patient **Robert Wood**. Like Sam he relayed how he ostensibly looks normal, but that 'for GSD patients things are often not so simple as they appear.'

Rob was diagnosed at 6 months of age, having presented with an enlarged liver and bright orange nose, and was originally given a life-expectancy of just 3 years. He described receiving 'fantastic care' at Great Ormond Street Hospital under the auspices of Dr Phil Lee, who will be familiar to some Pompe patients in the UK.

His childhood regime was complex, involving daily monitoring of blood sugars and night-time feeds every four hours. He also underwent a high carbohydrate diet. He was unable to walk freely until 3 years of age. As he progressed he continued to be seen at GOSH. His liver remained oversized and he was prescribed 50g of cornstarch daily. His bone and muscle growth was delayed and he was unable to maintain prolonged physical activity.

However despite these limitations Rob has gone on to achieve a BSc in Business Management and now works full-time as an Account Manager for Manchester United Football Club, nonetheless. He emphasised that 'people with GSDs can still achieve many things in life. We can be proud of who we are.'

Pompe Workshop, continued

Dr Yvette Easthope-Mowatt, Paediatric Clinical Psychologist, Robert Jones & Agnes Hunt Hospital, Oswestry, UK addressed the subject of "Emotional Support".

Yvette was invited to speak because, from conversations with families, we are becoming increasingly aware that our NHS is poorly equipped to offer the emotional support patients and families require outside of the clinic. This is particularly so in the period immediately after being given the news that they or their child have a rare genetic condition; a condition that may be progressive or require complex management.

Alexandra Crampton, Senior Policy and Campaigns Officer at the **Muscular Dystrophy Campaign** gave a general talk on the organisation. The MDC is becoming a very effective political lobbyist on behalf of all neuromuscular conditions and for Pompe in particular in the case of the Mackie Report - which urges that the Scottish Government reassess its position on the current access to Myozyme for Pompe patients north of the border. The organisation is also responsible in part for the formation of the All-Party Parliamentary Group for Muscular Dystrophy which meets regularly and currently encompasses 66 Members of Parliament.



Alexandra discussed how many of the achievements of the MDC are attributable to its several regional 'muscle groups', which alongside campaigning for better health and social care also help to provide services such as support of families through an Advocacy Service, linking-up patients with similar conditions, and obtaining grants towards specialist equipment. There are now 15 groups nationwide with over 700 members.

However some of the statistics she outlined give pause for thought on the progress still to be achieved with respect to neuromuscular diseases. For instance over 50% of people with NMDs do not as yet receive a correct and prompt diagnosis, 25% do not currently see a specialist, and over 50% feel their GP does not properly understand their condition.

The MDC organises regional meetings within the UK, (See their website for details), Pompe families are encouraged to attend these to discuss local services for neuromuscular conditions. If there is sufficient interest the AGSD-UK may organise separate meetings for GSD-specific discussions afterwards.



Katie Bainbridge gave an excellent account of the progress towards screening for Pompe disease. This was much the same as my report in the recent Pompe Bulletin (Issue 17, September 2010), available on our Pompe website. So I won't repeat it here.

And following was **Pat Roberts**, Executive Director of the **Save Babies Through Screening Foundation UK**, who gave an eloquent talk on the activities of her organisation. The Foundation was established in 2008 in order to lobby for expansion of the newborn screening programme to include a range of rare inherited diseases, including Pompe. To put some perspective on the issue, there are over 600 inherited metabolic diseases alone.

As I wrote in my report from Great Ormond Street in the last issue of the Pompe Bulletin, the current situation in the UK with regard to screening is sorely inadequate. Currently only five conditions are routinely screened for at birth. This compares to between 48-60 conditions in various US states. In fact as Pat pointed out, the UK is today roughly where the US was ten years ago.

She emphasised that the Foundation has opted for a collaborative as opposed to oppositional stance towards government on this issue. The objectives she has been pursuing are those of raising awareness and building evidence bases. She discussed PANS: the UK Patient Advocacy for Newborn Screening Working Group - a voluntary affiliation which includes her own organisation alongside the MPS Society, Myelin Project, the AGSD-UK and others. The aims of PANS include identifying, researching, reviewing and agreeing on diseases that could meet criteria for acceptance onto UK newborn screening programmes.

So to sum up, the 2010 Conference was certainly, in terms of numbers, the most successful to date; reflecting in part a high-profile year for Pompe. It was great to meet new patients who I had not known of before, many appearing at the conference for the first time. As several people said throughout the weekend, events such as these provide a focal point for the Pompe community and a unique chance to directly share experiences and information.

We welcomed several members who had not attended the conference before - Aidan Drew, Yasar Ayub, and all the way from Canada, Stacey Reason. Also attending were Sioned Williams, John Clarke, Rob Gray, Andrew Wakelin and Norbert Eckelmann from Germany; plus of course some very welcome family members and partners.

Stacey and Andrew gave a presentation on the Walk over Wales (WoW), based around a slide show but with lots of explanation and opportunities for questions. The various techniques used in achieving the walk of 210 miles were explained in detail, particularly addressing the issue of hills. The walk was a great success for the participants who all gained in technique and confidence. But it was also a success in inspiring people with the disease around the world. There is a report elsewhere in this newsletter and a great deal of information on the AGSD-UK web site, including the daily blog and photos.

Andrew explained that resulting from WoW, the four main walkers have decided to return to Wales in 2011 to run two one-week residential courses called "Walking with McArdle's". The aim is to help a wider group to learn how to walk safely and get the most out of it. Each course will take 8 participants. Again, details are elsewhere in this newsletter and on the web site.

WoW raised approximately £14,000 towards the objective of making a DVD of video clips about how to best live with McArdle Disease. The idea is to include all the tips and tricks that people have learned, and share them amongst the whole McArdle community. The content of the DVD is now being planned. Andrew asked that anyone who was interested in contributing or assisting to get in touch with him on 01597 860686 or type5@agsd.org.uk.

In a go-around of members we heard many fascinating stories, impossible to report in detail here. Standing out was Rob's account of being diagnosed by Dr Brian McArdle himself about 40 years ago, age 22. Rob has a brother and a sister also diagnosed with the disease. Stacey had been hit by a car whilst out running and over the long recovery had lost her aerobic conditioning. When she returned to exercise she experienced severe rhabdomyolysis and that led to her diagnosis of McArdle Disease.

We welcomed Dr Kathryn Wright to the workshop. Kathryn had been writing the "McArdle Disease Handbook" - an account of all the research and scientific papers on McArdle's explained in layman's language (or as close as it is possible to get!). Kathryn gave a fantastic presentation that she had prepared specially for the workshop. This covered all the main scientific areas and Kathryn explained key points. There were of course lots of questions and Kathryn was very patient in answering them, pointing out that she was a scientist rather than a doctor. Kathryn's book will be published by AGSD-UK early in 2011.

Dr Yvette Easthope-Mowatt from Oswestry contributed to the workshop. Yvette is a psychologist and has been working with Dr Quinlivan on the emotional aspects of neuromuscular disease, including McArdle's. She was able to provide some useful insights.

Andrew explained about the move of the UK McArdle Clinic from Oswestry to London. Although this had not been an objective, it should turn out to be for the best in the long run. The final clinic at Oswestry had been in August 2010 and it was hoped that the new clinic would be operational early in 2011. The major concern was retaining the expertise which had been built up at Oswestry, but Dr Ros Quinlivan was transferring to London and Dr John Buckley had expressed his interest in continuing to be involved. Some of the other staff might also still support the clinic. The hospital at Queen Square, WC1, is a major neurological centre and sees patients with many neuromuscular diseases. The facilities are excellent and there will be much greater support for the work of the clinic than had been the case at Oswestry. It is hoped that a research assistant will be appointed, and this will enable more clinical trials to be carried out. Travel to London will be easier for many, but of course harder for others. Andrew will work with Dr Quinlivan and the hospital to work out the best arrangements that we can for travel, directions, parking, low cost overnight accommodation, etc. Keep an eye on the website for details in due course.

It was a very busy conference and workshop. We all left recharged with ideas and determination to keep on top of our condition and grateful to the many people who continue to support us in so many ways.

Walk over Wales



Raises Awareness (and £14,000)

Surely you can't have missed the "Walk over Wales" for McArdle Disease (rightly abbreviated to "**WoW**")? A group of people with McArdle's walked 210 miles from the north Wales coast to Cardiff on the south Wales coast over 32 days from 2nd July to 2nd August 2010.



We made the ITV Wales News at 6pm on the day we finished, and had lots of local press coverage on the way down Wales. That tackled the first objective - to raise awareness of this rare muscle disease amongst the public, parents, youth leaders, teachers and doctors - so that more children are diagnosed at an early age and rescued from a painful, damaging and miserable time at school.

The next objective was to inspire other people with McArdle Disease so that they could be encouraged to become fitter, healthier, more active, have less episodes of serious injury and avoid stays in hospital. We did that through a daily blog and photo upload on the AGSD-UK web site (it is still there if you'd like to take a look). The blog was followed and commented on by hundreds of people around the



world and towards the end we had almost 400 "supporters" on a Facebook group.

The final objective was to raise money towards producing video/DVD information to help people with McArdle's learn how best to cope with their condition. We raised an amazing £14,000 through JustGiving.com.

Walk over Wales, continued

The walk was led by Andrew Wakelin, AGSD-UK Type V Coordinator, who has McArdle's Disease but is an experienced hill walker. The route was planned from his extensive experience of walking with McArdle's and took in the main peak in each of the mountain groups as we travelled south. We sometimes had to take a lower level alternative due to bad weather – it rained most of July and on both Snowdon and Cadair Idris we had 10 yard visibility and winds gusting 50 mph. We stayed at a variety of B&Bs and bunk houses and had a series of support drivers who picked us up at the end of each day and delivered us back to the same point the next morning.

We had walkers from Wales, USA (Dan Chambers), Canada (Stacey Reason) and Singapore (and Wilaims). Charlton Thear, age 13 from the Canary Islands, managed to get out of school for the last three days in South Wales. People with McArdle's struggle with pain and exhaustion to walk 100 yards up the gentlest of slopes. We learned a lot. But using the right techniques and building up fitness, by the end of the month those who started out hill-adverse, were actually doing extra hills for fun!

Thanks to our support drivers, everyone else who helped en-route and all our many WoW supporters in the UK and around the world who kept our spirits up, cheered us on our way and made donations to the cause. It was an experience of a lifetime.



Type V - Website Developments

We have made a number of improvements to the McArdle Disease information on the AGSD-UK web site. There is a revised structure and a few new pages.

McArdle Emergencies

It was a shock last year to hear of the death of Jessica Binder, age 31, in the USA. She had only recently been diagnosed and suffered a severe episode of rhabdomyolysis with myoglobinuria. Regrettably she delayed going to hospital for several days. She was suffering acute renal failure and put on 25 lbs in weight due to retained fluid. Once in hospital she was treated and put onto dialysis, but she unfortunately suffered a fatal heart attack a few days later before her systems had fully stabilised.

Please check out the new page on "McArdle Emergencies". This provides guidance for McArdle patients about the signs that indicate that they should seek medical help for a severe episode. It also alerts emergency doctors to the risks of a McArdle episode. And finally, it provides a lot of background information about such episodes and how you should expect them to be treated.

Guidance for the McArdle patient

If you have one or more of the signs below you should:

- Start drinking water, 500ml (1 pint) per hour.
- Get to hospital promptly.
- Take a urine sample with you, if possible.

Dark coloured urine
This is called myoglobinuria and appears as reddish tea to cola coloured urine. (However, if you have not done any unusual activity and have eaten a strongly coloured food such as beetroots (beets) there is probably nothing to worry about.)

Feeling very unwell after exercise
Feeling very unwell (perhaps with flu-like symptoms) after you have undertaken strenuous or unusual exercise/activity can be a sign of rhabdomyolysis (muscle breakdown).

Low volume of urine
Producing a very low volume of urine, or no urine at all, both constitute a medical emergency. You should go to hospital immediately as the complications can become life threatening.

If in doubt, telephone your McArdle Disease consultant or specialist nurse.

Information for the Emergency Doctor

Background: McArdle Disease (GSD Type V) [1] is a rare metabolic myopathy. Patients experience exercise intolerance and muscle cramps. They are at risk of rhabdomyolysis [2], which can lead to acute renal failure.

Presentation: Patients may appear well but have a muscle cramp, myoglobinuria or feel unwell/have fever after exercise. There may be an extreme muscle contracture with swelling and often severe pain.

Investigations: Urgent assessment for possible rhabdomyolysis. Creatine Kinase usually grossly raised. Consider urine analysis for myoglobinuria and full chemistry panel (including CK, glucose, calcium and bone profile, urea and electrolytes).

Management: Consider fluid bolus followed by IV saline at 2x maintenance and (unless diabetic) possibly 10% dextrose to keep blood glucose >3.5 mmol/L. Episodes require monitoring of urine output, CK and electrolyte status.

Complications: Oliguria can occur requiring prompt referral for haemodialysis. In rare cases compartment syndrome [3] has developed, requiring surgical referral.

Remember that this page is available so that you can refer to it when needed and bring it to the attention of an emergency doctor when the occasion arises. Some people have printed out the page to keep in their wallet or handbag.

Starting an exercise programme

This is another new page intended to be of assistance to people with McArdle Disease who are at a low level of aerobic conditioning and need to get started on a regular exercise regime. People at a low level of conditioning are much more prone to muscle contractures.

The objective is to slowly build up aerobic capacity whilst avoiding any anaerobic activity. When you exercise aerobically your body gets better at transporting oxygenated blood and blood-borne fuels to your muscles. More mitochondria grow within the muscle cells and these make it easier for you to exercise. This is the same effect that athletes use when they train, but it is particularly beneficial to those of us with McArdle's.

If you are not exercising regularly, why not have a look at these suggestions? They have been prepared with input from a number of people who have been very severely affected in the past and have returned to a good level of aerobic conditioning. This programme has already been used with success.

2010 AGSD USA Conference

Report by Sue DelMar

The 2010 US AGSD Conference was held at the Radisson Celebration Hotel in Orlando, Florida on the 9th October 2010. Some 400 people attended including many families and professionals. It was most efficiently organised by Drs David and Geraldine Weinstein and the University of Florida and included a fun run and a visit to Gator Park to see the alligators both of which were much enjoyed by all who took part.

Diagnostic and Management Guidelines (GL) for GSD Type III. Dr Priya Kishnani, Duke University

These consist of:

- Clinical practice GL to assist doctors and clinics
- Dietary GL
- Manuscript and publication GL
- Well baby care GL

These guidelines are needed as not all general practitioners and even some specialist doctors have seen a GSD patient. They will provide improved healthcare outcomes and greater availability of access and they will provide educational resources and facilitate more consistent treatment.

The guidelines have been compiled under the auspices of ACMG (American College of Medical Genetics) Dr Mike Watson, an ex-director of ACMG has co-ordinated the project

Experts in the field were chosen and each expert was given a section for research into diagnosis and treatment strategies. Meetings were held to consolidate and agree on the findings and the final results were documented and published in "Genetics in Medicine" during the summer of 2010 and will be updated every three years.

It is hoped that the GL for Type V and Type VII will be published in 2011 and those for Type I in 1-2 years. Type II GL were published in 2006 and need updating. They will predominantly apply to the United States. It may well be necessary to have different GL for Europe and the USA.

*Sue DelMar has a copy of the complete document if you would like to read it

Gene Replacement Therapy, Dr Barry Byrne, University of Florida and Dr Dwight Koebel, Duke University. Dr Barry Byrne showed photos of the Type I puppies that are having gene replacement therapy at the University of Florida. The oldest, Dulce, is now three years old. They are re-dosed at 20 weeks and calculations are needed for each dog to establish further re-dosages

Dr Dwight Koebel also showed photos of the GSD puppies at Duke University There are four of them. Lollipop, the oldest is 48 months, and doing well.

The first colony of GSD-IA dogs were treated at the NCSU College of veterinary Medicine for over 10 years. Almost two years ago they were moved to Duke University

He gave a brief description of the vector used in Gene Replacement Therapy.

The AAV vector is used to carry the gene into the liver (Adeno Assisted Virus)

Only 10% of normal glucose-6-phosphatase is needed to prevent hypoglycaemia so it is important that the correct amount of the gene is put in the liver. Too much would make the liver over react.

The dogs are injected at three days old. Untreated dogs die very quickly.

The vector actually works better as the dog gets older as the liver responds better

Vectors have to be varied to find the maximum benefit.

Cornstarch Update, Dr David Weinstein, University of Florida

In 2003 six versions of modified cornstarch were tested. Three showed no increase in fasting time and two were quite unpalatable. The sixth became known as Glycosade, made by Vitaflo International, and it was able to increase the fasting time by an hour in some patients. It was quickly approved in the UK but not in the USA and Canada

Initially it was made in batches of 1kg but as production increased it was produced in batches of 500kg. This sadly reduced the effectiveness of the starch.

Vitaflo has been taken over by Nestle but we are assured that this will make no difference to their research.

Vitaflo have reverted to producing the starch in small batches. A new International study on the efficacy of Glycosade is being created in London with doctors from USA, England, France and Italy taking part.

2010 AGSD USA Conference, continued

Coping with GSD, Dr Lisa Merlo, University of Florida

Dr Merlo gave a very cogent presentation on coping with GSD which she kindly sent me as a computer file and we hope that it will be presented at one of our future conferences.

Notes on Type I and III Workshop

- Type III patients need 25% of their diet to be proteins. High protein bars used by athletes are a good source
- Only some patients suffer with muscle deterioration
- Beta blockers not a good idea
- Because of delayed puberty always use bone age not chronological age
- When working out keep a check on sugar levels and take a glucose tablet if they are in the low 80's..
- Try to get teenagers to take responsibility for their cornstarch and medication. Always praise and encourage.
- In the USA patients should always carry cornstarch, glucose meter, snack, spare battery, glucose and water with them
- All patients should have the Medi-alert and emergency phone number on their mobile phone.
- Google ICE and have an ICE sticker on the phone.

It is hoped that glycogen in the future will be able to be measured by a marker in the urine.

Vitamin D, Dr Ann Boney, Duke University

Vitamin D is commonly referred to as the sunshine vitamin needed to build strong bones. However we now know it does far more than that. In addition to causing rickets vitamin D deficiency has been linked to autoimmune disease, cardiovascular disease, cancer, type II diabetes, infectious diseases and depression. In addition to sunshine which helps our body make vitamin D we can get it from tinned salmon with bones, milk, yoghurt and other vitamin D fortified foods such as orange juice and cereals. Having Glycogen Storage Disease may put you at a higher risk for developing vitamin deficiency. In a study at Duke, 16 out of 26 patients with GSD Type I (61.5%) had sub-optimal vitamin D level. It is important to know your vitamin D level. You can talk to your doctor about having a simple blood test called "Vitamin D 25 hydroxy". The sufficient range for vitamin D is 30-100ng/ml. If your level is less than 30ng/ml you can talk to your doctor or dietician about the best treatment option.

Sue Del Mar gsdaddresses@btinternet.com



Philip Maes*, Sue Del Mar, Abi Adewumi+, David Weinstein, Geraldine Weinstein and Justin Weinstein enjoying Brunch in Celebration, Florida, after the end of the conference.

*President of AFG France

+ Dental doctor researching the long term effect of cornstarch on teeth.

Fundraising Events and Ideas



WHY RUN WHEN YOU CAN WALK?

Gary Lineker says: "I am giving my full backing and support to this unique event and I hope that it is an enormous success!"

Walk the London Marathon on Saturday 9th and Sunday 10th April

Have you ever fancied running the marathon but know running is just not your thing? Well why don't you join us on 9th/10th April 2011 walking the entire route of the famous London Marathon, with full back up and support given.

The self led walk will be starting on Blackheath on the morning of Saturday 9th April, walking at a steady pace to make lunchtime at the Cutty Sark. After lunch we will then make our way to the half way point of the marathon at Tower Bridge.

For those that wish to, we have a list of hotels located around the Tower Bridge area to stay the night and get plenty of rest and relaxation in preparation for 13 miles on day two (your luggage will be carried for you).

On day two we will pick up where we left off and make our way around Canary Wharf before heading back west to Westminster and the finishing line at the Mall. After 2 days and 26.2 miles, we'll then gather with friends and family for celebration drinks and well deserved nibbles.

To take part in this event you need to be over 12 years old (under 18's must have a responsible adult taking part with them) and you don't need to have any walking experience, we'll send you a full training guide for that! If you're looking for a health kick, this is an easy and inexpensive way to get you fit and motivated. Don't wait, sign up now and start your walk training.

For all the details please call Steve on 07990 900374 or e-mail fundraising@agsd.org.uk

Join us for the first ever AGSD-UK fundraising abseil in the UK! SUNDAY 8TH MAY 2011

This is the big one! - At 500 feet high this is probably the biggest public abseil that you are likely to be able to do.

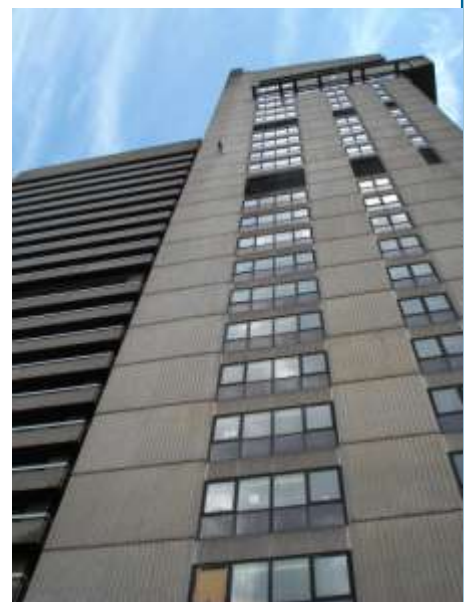
Take in the amazing views of London while abseiling down the side of this landmark building. But don't worry! You will be in the safe hands of our abseil experts "Over the Edge Adventures" who have many years experience of running these types of events.

Entry Fee is only £15 and all participants will need to raise a minimum of £175 of sponsor money to be able to take part. Places are strictly limited and bound to go quickly so "don't hang around" contact us now for your entry form.

In return we will send you full information about the day, how to come prepared and what to expect, as well as the sponsor forms so that you can get started on raising those vital funds.

Minimum age to take part is 16 years old and you do not need previous experience as all the training will be provided on the day. There will also be incentives for raising funds.

For entry forms and any queries please call Steve on 07990 900374 or e-mail fundraising@agsd.org.uk



Fundraising Events and Ideas, continued

EVER DONE A ZIP SLIDE?

If not then here's your chance!

We have an exhilarating Zip Slide event taking place at more than a dozen locations around the country during 2011 and you can take part.

These events will be taking place from a variety of different buildings and structures from various heights ranging from 17 metres to 50 metres high and from 140 metres to 300 metres long.

Venues are at Bristol, Leeds, Tyne Bridge, Newport, Glasgow, Manchester, and Milton Keynes and London so there is bound to be somewhere near to you.

e-mail Steve on fundraising@agsd.org.uk or call 07990 900374 for more information.



Want to do your own thing?... Can we help you?

If you have your own ideas or plans and want to raise funds for AGSD-UK then we may well be able to help!



You might be planning a sponsored head-shave, a fundraising dinner party or maybe a car boot sale to help de-clutter as well as raising some money. Whatever it is you would like to do please let us know as we can supply help and advice as well as things like official sponsor forms. If it is something particularly unusual it may be newsworthy so we can help with raising awareness as well through your local press or media.

If you fancy running one of the many marathons, 10K's, half marathons or fun runs that regularly take place around the UK then let us know and you can raise funds for our charity.

Do you want to do a parachute jump, trek the Inca Trail in Peru or even walk along the Great Wall of China? – Again we can help you realise these ambitions as well as raising vital funds to help AGSD-UK.

Just e-mail fundraising@agsd.org.uk and we are ready to help!

Alternatively give Steve a call on 07990 900374.

Pompey to Pompeii for Pompe

(and related GSDs)

A cycling challenge of 1400 miles from Portsmouth to Pompeii, Italy. September 2011

Are you, or do you know, a keen cyclist who is looking for a challenge this year? We are still recruiting fit cyclists to participate in the whole event or selected sections of the route. But if cycling isn't your bag you can help raise awareness of the ride locally and help our cyclists reach their sponsorship targets by visiting our Virgin Money Giving account:



Search <http://uk.VirginMoneyGiving.com> for "agsd"

The event will take place over three weeks, setting out on September 10th from Portsmouth. Day 1 will reach Hastings and on Day 2 we will cycle to Dover for a ferry to Calais. Thereafter we will try to follow the Pilgrim's Route (aka EuroVelo 5) from Canterbury to Rome and then on to Pompeii. We plan to have a detailed route worked out before the end of March.

The Route

There are 4 main stages in the 1400 mile route:

- The hills of the South Downs (150 miles)
- Rolling French country roads (500 miles)
- Crossing the Alps from France through Switzerland into Italy (250 miles)
- Roman roads from Milan to Rome and on to Pompeii (500 miles)

Accommodation

It is likely that the cyclists will sleep in tourist class hotels or hostels. The whole trip will take 3 weeks but it will be possible for cyclists to join for all or part of the route; preferably whole weeks in mainland Europe. We also hope to attract additional cyclists to join us for one of the sections in England:

- Portsmouth to Brighton – about 50 miles or
- Portsmouth to Hastings – 90 to 110 miles depending on route or
- Hastings to Dover – 50 miles

Fundraising Requirements

Participants must raise £1000 for each whole week or £250 for a single day (in England). We will give fundraising help and advice and we have already set up a webpage on Virgin Money Giving where riders can create their own fundraising page with the benefit of Gift Aid (<http://uk.virginmoneygiving.com> and search for AGSD).

We expect to have corporate sponsorship to cover most of the event costs so that all funds raised by riders will go directly to the charity.

A 4-page brochure has been produced as an aid to fundraising and corporate sponsorship and a website will go live very shortly.

For more information please contact the organising team:

Allan Muir allan.muir@agsd.org.uk
0300 123 2790 Daytime Mon to Thurs
0300 123 2799 Out of Hours

Luca Venditto luca@italianway.co.uk
01424 753173
07876 250220

For fundraising requirements and advice:

Steve Saunders fundraising@agsd.org.uk, Telephone: 07990 900374



Walking with McArdle's

We are organising two one-week holiday courses for people with McArdle Disease, to be run in July and August this year.

This is an opportunity to experience walking with other people who have McArdle Disease, to learn how to make walking safe and enjoyable. You can do this with a friendly and understanding group, in the beautiful Snowdonia National Park. We will stay at the Craflwyn Basecamp near Beddgelert, on a National Trust estate, in comfortable bunkroom accommodation. The group will self-cater with the occasional meal out.

The organisers undertook the 'Walk over Wales' in 2010. All have McArdle Disease and have been severely affected in the past. The Team will be led by Andrew Wakelin who knows the area pretty well.

What you will gain

We aim to teach you the best techniques for walking with McArdle's, so that you can gain in confidence. We will help you to improve your aerobic fitness and reduce the risk of muscle injury. You will gain confidence in managing your need for rests, without embarrassment. You are likely to improve your motivation to undertake regular exercise - with all its well proven physical and mental health benefits. We hope you will go home feeling confident that you can exercise safely and with the desire to expand your McArdle's boundaries, inspired by what you have achieved in Wales.

The programme

We will be running a series of relatively informal theory sessions plus there will be plenty of opportunity for discussions. The programme will include: aerobic V anaerobic, the aerobic to anaerobic continuum, second wind, the 6 second rule, starting on the flat, effect of terrain and surface, slow and steady, regular rests, headwinds, benefit of walking poles, correct use of walking poles, walking with non-McArdle people, route planning, eating on walks, hypoglycaemia, hydration, zig-zag technique, stepping up, bad pain versus good pain, etc.

The cost

You should budget for a total of approx £250 to cover the base camp accommodation, breakfast and lunch, evening meal, transport during the week - shared cost for petrol etc, and a few incidental costs.

Dates

Week 1:

Friday 22nd to Friday 29th July 2011

Week 2:

Monday 1st to Monday 8th August 2011



Contact

Some people are already booked from the UK and overseas. For further information please see the AGSD-UK web site. To register your interest in attending please email Andrew Wakelin at type5@agsd.org.uk

Sioned Williams mentions McArdle's on Radio 4

On the 20th of January "Woman's Hour" on BBC Radio 4 featured an interview with Sioned Williams, principal harpist with the BBC Symphony Orchestra. As many of you will know, Sioned has McArdle Disease.

The interview and recital was about the first public performance of the only new type of harp in 100 years. Sioned was playing a piece of music which had been specially commissioned for the launch of the harp.

Sioned managed to get in some comments about her McArdle Disease and how she copes with a very demanding job. It was a great piece of awareness raising for the disease.



Donations

We would like to thank everyone for their kindness and continued support

The following have made donations to AGSD(UK) during the past few months

Thomas for Jodi Venditto	Saunders Family
Joan, Neighbour of the Muirs	Sagar
Pratt for Jodi Venditto	Writtle Bell Ringers
Buriton events (Muir's home village)	Virginia Altrogge
John Lewis via P.Critchley	Lara Daly (Half Marathon)
Moyze family In memory of Sylvia Hawke	James Graham
Nearly new Shop, Arborfield	

Instead of sending Christmas cards by post

Allan and Barbara Muir (Type II)

Grants for the 2010 Conference

Genzyme (UK)
Amicus International Ltd
VitaFlo International Ltd
BioMarin Ltd

We are most grateful to them all



Could **You** be an **AGSD-UK** Trustee?

The AGSD-UK charity is governed by a Board of Trustees and we are keen to expand on their number.

Whilst many other people volunteer to help the charity, it is the trustees who have ultimate responsibility for directing the affairs of a charity. They have to ensure that it is solvent, well-run, compliant with regulations and delivering the charitable outcomes for the benefit of the public for which we have been set up. We need people with backgrounds in areas such as finance, administration, promotion, legal, IT, human resources, health care, medical research, etc.

Charity trustees come from all walks of life bringing abilities, knowledge and/or experience to the charity. Being a trustee can be rewarding and enjoyable and provide an opportunity to make a positive difference.

A strong personal commitment to our charity's aims is important. We meet four times per year, usually on a Saturday in London, and a variable amount of additional work may be required between meetings. Trustees are unpaid volunteers but are allowed to be reimbursed for reasonable expenses.

Over the last 18 months AGSD-UK has come a long way in converting to a company limited by guarantee, appointing a firm of accountants, establishing a Development Plan, employing a Development Director as our first regular employee and establishing a small office for the charity. These steps lay the ground work for the next few years to be very exciting and take the charity on in many ways.

If you would like to explore the idea of becoming a Trustee, or volunteering in any other way, please contact the Chairman, Andrew Wakelin on 01597 860686, or email chairman@agsd.org.uk.

Scientific Advisory Board

The AGSD-UK has a Scientific Advisory Board of medical and research professionals who are able to advise the Board of Trustees on policy issues and on funding applications. We are very grateful for their support.

Dr Helen Mundy MRCP

Metabolic Department, Guys and St Thomas NHS Foundation Trust, London, UK

Dr Philip Newsome MBChB BSc MRCP PhD

Liver and Hepatobiliary Unit, Queen Elizabeth Hospital, Birmingham, UK

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Contact Page



The AGSD (UK) Ltd is managed by a volunteer Board of Trustees elected by its members at the Annual General Meeting held each year as part of the Annual Conference. The current members of the board are listed below. If you would like to volunteer to help the charity in any capacity or would like to consider becoming a Trustee, please get in touch with the Chairman or contact the office.

Trustees

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Andrew Wakelin - type5@agsd.org.uk

Treasurer and Type I Coordinator

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AGSD-UK Staff

The following staff are employed by AGSD-UK, on a part time basis.

Development Director and Type II Co-ordinator

Allan Muir - allan.muir@agsd.org.uk

Family Support Officer

Joan Fletcher, FCO/CNS - joan.fletcher@pompe.org.uk

Telephone 0161 701 2601

Joan is employed by the NHS under a grant from Genzyme Ltd to support Pompe families

Fundraiser

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Volunteer appointments

The following are volunteer appointments of the AGSD-UK but are not Trustees.

Type IX Co-ordinator

Christine Evans - type9@agsd.org.uk

Newsletter Editor

Kate Phillips - editor@agsd.org.uk

If you have any articles for the newsletter please let me know.

We are always keen to hear from our members!

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