Annual Report

2018/2019

Association for Glycogen Storage Disease UK
www.agsd.org.uk

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AGSD UK
Mile House, Bridge End,
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Registered Charity no 1132271

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A New Trustee's View

I am writing to you as one of the relatively newer members of the AGSD-UK team, officially appointed to the board of trustees as of August 2018, to help in steering the charity into new, exciting and beneficial paths. Being appointed as trustee of a charity is a new role for myself, having not previously undertaken such a position. For a number of reasons however, when the chance arose to take up the position, it seemed like the perfect opportunity.

From a very early age, I was always surrounded by charity and voluntary work. Both my parents have spent many years either working in the charity sector, or volunteering on trustee boards both locally and further afield. The idea of giving back to the community is something that has been deeply embedded into our family culture. This was something I am very keen to continue and have since regularly sought to raise money for charity, recently completing various different fitness and sporting events. Then, when the opportunity came up to join the board of trustees at AGSD-UK and take a more direct role in a charity, it was one that seemed like the perfect next step for me.

My background is one involved in nutrition, physiology, medicine and more recently genetics, obtaining undergraduate, masters and a Ph.D in research themes across these fields. I am currently a research scientist working at Queen Mary University of London, researching how environmental exposures interact with our underlying genetics to shape our bodies physiological traits.

Having a background in nutrition, physiology and genetics meant I became quickly intrigued by GSDs, and the underlying reason for why it occurs. It is this that really got me interested in supporting AGSD-UK as a charity, and the AGSD-UK community, hoping I can use my knowledge and experience to give back to this community.

I have now been part of the charity for just over year. It has been an exciting and developmental period for both myself and the charity. Two things have struck me most significantly of all since joining the board. Firstly, it very quickly became apparent to me just how much of a close knit group the AGSD community is. And secondly, it is clear that the area of scientific development looking into therapeutics and/or cures for the spectrum of glycogen storage diseases, is one that is rapidly developing and is thus truly exciting. Both of these factors quickly became clear when I went to Orlando, Florida for the WORLDSymposium in January earlier this year. To see the extent of research, the wide array of organisations, institutions and communities actively working and supporting the disease was extremely impressive. Knowing some of the 'break-through' scientific developments that are now being trialled in drug development, and seeing some of these integrated into research looking to cure GSDs is very exhilarating and something that we all hope will progress quickly.

The next year promises to be a year of real development for the charity and one I am very much looking forward to being a part of and helping to support.

Robert Seaborne, Trustee

Recent AGSD UK publications

Chair's Report

Welcome to the AGSD-UK 2018-19 Annual Report. The trustee board are once again pleased to update our GSD community and all of our key stakeholders on the work of the charity during our 2018-19 financial year. As always it was a very busy time for the trustee directors, staff and volunteers.

The trustee board met three times in June 2018, October 2018 and January 2019. In addition a trustee training day was held in June 2018. A number of sub-committee meetings were also held throughout the year.

Allan Muir, who had been running the day-to-day operations of AGSD-UK since 2010, left the organisation in September 2019 to pursue his own business interests. Allan who has been closely involved with the charity for over 20 years, has helped AGSD-UK to develop and grow over that time and provided invaluable support to the UK and international Pompe community. The trustees would like to place on record their thanks and gratitude for Allan's many years of dedication to the UK GSD community.

Our new website was formally launched in January 2019 and I am sure you will agree it is a significant improvement when compared to our old clunky text-based site. The launch would not have been possible without a massive amount of input and effort from our McArdle co-ordinator, Andrew Wakelin.

Unfortunately, during the year two of our trustee directors resigned their positions. Heneage Legge-Bourke our Honorary Treasurer stood down in February 2019 and Mike Porter our Chairman announced he would be standing down as Chairman after the January 2019 trustee board; following five years' service as Chairman and more than six years as a trustee. Mike informed the trustee board that he had set himself a comprehensive set of personal objectives for the charity to achieve during his time as chairman, all of which have been achieved. These included:

- Stabilising and improving the financial position of the charity
- Strengthening the trustee board

- Developing and implementing a new website
- Putting in place a comprehensive set of policies and procedures
- Implementing a secure IT system
- Appointing a Specialist Care Advisor for the Pompe community
- Implementing a succession planning process for key roles

Mike feels extremely proud and privileged to have been part of the team taking the charity through the third phase of its development, building on the great work by his predecessor Andrew Wakelin and his trustee board and Mike is confident that AGSD-UK will continue to go from strength to strength. With no obvious replacement for Mike, Nick offered to stand in as Acting Chair, temporarily. Nick personally believes though that the role is most suited to a non-working person, (possibly someone working part-time); so if you would be interested in being a trustee and/or Chairperson, please make yourself known to the trustees.

One of the key outcomes of the implemented succession plan was that the trustee board identified that, in order to lead the charity through its next growth phase; it would require an experienced Chief Executive Officer to be identified and appointed. A strategic objective for the new CEO is to seek to further enhance the range of services provided to beneficiaries, via successfully bidding for larger tranches of funding. To that end the trustees, with the help of some senior HR support; initiated a UK-wide search to identify potential suitable candidates. We received over ninety applications and, following a robust selection process; Neil Bradbury was appointed CEO in August 2019. A primary focus for the coming year will be to embed Neil into the organisation, having him meet all of our stakeholder groups and to support him in delivering year 1 of our 3 year business plan.

One of the major successes of our Specialist Care Advisor role has been to provide practical and emotional support to those GSD sufferers going through the government mandated benefits move from Disability Living Allowance (DLA) to Personal Independence Payment (PIP). The process for claimants is very challenging and anxiety inducing and I am pleased to report that Jane Lewthwaite has supported 66 PIP submissions and around a further 40 submissions for Employment Support Allowance and Carers Allowance.

Jane has also been involved in 3 PIP appeals, all of which have been successful. Providing advocacy work such as this for the UK GSD community is invaluable in meeting the needs of our sufferers and their families/careers.

As well as hosting our annual AGSD-UK conference in Wyboston and a number of regional meetings, the charity was represented at the European Orphan Drug Congress in Barcelona by Ailsa Arthur, Jane Lewthwaite, Jason McMillan and Allan Muir, by Rob Seaborne and Allan Muir at the World Symposium on Lysosomal Storage Disorders in Florida and by Jane Lewthwaite at the Genetic Alliance Annual Meeting in London. As previously reported attendance at such events contributes to our store of knowledge regarding emerging developments in the field of GSD diseases as well as supporting the 'vision' of AGSD-UK in being the primary and central focus for information and support, driving positive change for all people affected by Glycogen Storage Disease.

Finally the trustees would like to wish our longest serving trustee, Jayesh Pindolia, a speedy recovery from a life threatening illness that has incapacitated him since January 2019 and hope to see him soon at future trustee meetings.

Mike Porter – until January 2019
Nick Jones – Acting chair from January 2019

Mike Porter & Nick Jones

Aims, Mission & Vision

AGSD-UK provides support and help for individuals and families affected by an ultra-rare condition known as Glycogen Storage Disease (GSD). Glycogen is a stored form of glucose, which is used as a fuel for energy. Glycogen Storage Disease occurs when there is an absence or deficiency of an enzyme needed to produce or break down glycogen. GSD primarily affects the liver and/or the muscles, including the heart.

The charity gives support to those with all GSD Types. There are currently 16 known Glycogen Storage Diseases; most have sub types and some have tiny numbers of people affected. Incidence, number diagnosed and age of diagnosis vary enormously.

AGSD-UK aims to help patients and their families affected by Glycogen Storage Diseases in a number of ways.

- Act as a patient and family contact and support group
- Provide information on request by phone or email
- Hold GSD-specific workshops
- Carry out, promote and/or support research and surveys into GSD
- Publish a newsletter for all stakeholders
- Create information and resource materials
- AGSD-UK organise an Annual Conference which usually takes place over two days in the autumn and is attended by people with GSDs, their families and many medical professionals, laboratory scientists and researchers

Mission Statement

Supporting people affected by GSD to live positively with their condition.

Vision Statement

To become the primary and central focus for information and support, and driving positive change, for people affected by GSD.

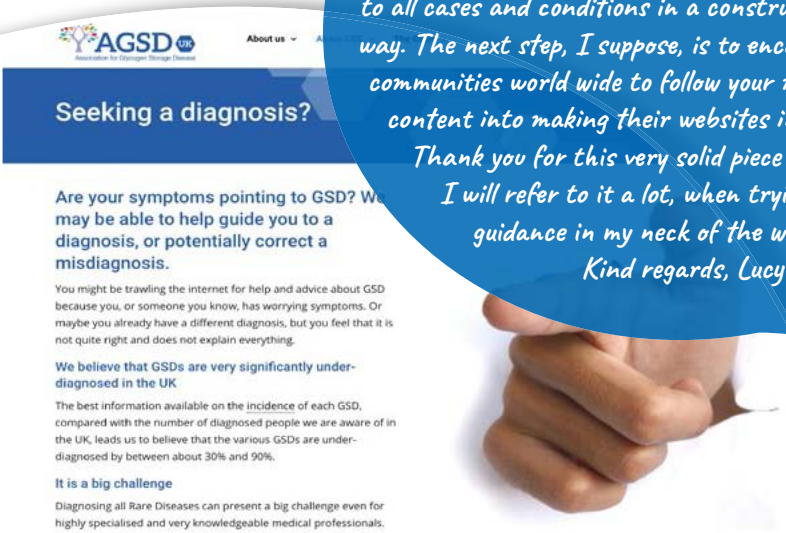
A New Website

Andrew Wakelin, GSD5 co-ordinator, led the development of our new website and delivered a complete redesign and restructure for the website. He gave several months of work as a volunteer.

Currently we are getting an average of **2,900** visitors per month, with over **7,500** page views. **35%** of users are from the UK, **29%** from the US and the rest from all around the world. Smartphone usage is growing, with now **47%** of users on that platform, another **47%** on desktop computers and **6%** on laptops.

In the most recent month, apart from the homepage, the most visited page was our news story on a ***Netflix*** documentary mentioning muscle GSDs.

I have spent quite a long, but very interesting time in discovering your new website, and clicking on all the propositions, and my personal conclusion is that it is GREAT! It is very easy to navigate, couldn't fault the content, seems to respond to all cases and conditions in a constructive and helpful way. The next step, I suppose, is to encourage the Pompe communities world wide to follow your model and depth of content into making their websites in their languages !! Thank you for this very solid piece of work. I know I will refer to it a lot, when trying to provide guidance in my neck of the woods.
Kind regards, Lucy



Regional Meetings

Regional meetings provide an opportunity to meet and make new friendships, share information, and connect which is vital for our GSD community in the UK.

The Pompe Support Team enabled four regional meetings for International Pompe Day in April.

The Cori Action team held a social event in May. Note ***Cori the Cat***, centre stage in the photo, who is a new mascot for Cori disease. All children with GSD3 were sent a mascot toy and comic about Cori's adventures. Very positive feedback was received from children who are very isolated.

Cori the Cat at the GSD3 Get Together, March 2019

Ensuring Good Governance

We have 7 trustees with a range of skills but in particular; experience of glycogen storage disease and knowledge of the disease as a prime carer or with a family member, management, marketing, finances and IT. Where necessary they seek advice from experts. All trustees are ambassadors for the charity and act in the charity's best interest at all times to meet the aims and objectives of the charity and to ensure it is of public benefit.

A full Induction Day and briefing for the Trustee Board by the staff followed by a lengthy discussion which formed the basis of our new 3 year Business Plan was held. Trustees were able to consider the feedback and comments gathered from our 2018 Stakeholder survey. Our volunteer survey in 2018 told us our volunteers felt valued and they had gained a great deal of satisfaction from their work but needed more briefing and information to feel better supported.

Trustee Ailsa Arthur prepared and sent an AGSD–UK awareness raising pack to 114 hospitals in the UK who offer services to people with a GSD. We continue to need to extend our reach.

Trustees take full responsibility for managing the finances including monitoring their performance with good budgetary and financial controls in place to keep funds safe. We have agreed a strategic plan for income generation and with the recruitment of our first CEO believe we can be much more successful. We appreciate how important it is to manage our limited resources very carefully and efficiently and with reasonable skill and care.

We are of course accountable first to the Charity Commission and Companies House but also as an employer of staff to have relevant policies in place to ensure we keep up to date with current laws such as for pensions, employment and health and safety. All 25 of our policies were reviewed and a refreshed Safeguarding Policy was adopted by Trustees. Jane Lewthwaite is our Safeguarding Officer who conducts the annual review of DBS check requirements in the organisation for all staff and volunteers.

We are also accountable to our membership, especially at the annual conference and AGM, and to ensure the greatest transparency we provide regular newsletters, hold meetings and keep our new website up to date.

As a charity, with staff who support people with serious health impairments, which includes many of our volunteers, we recognise the need to provide continuing support and care to them all.

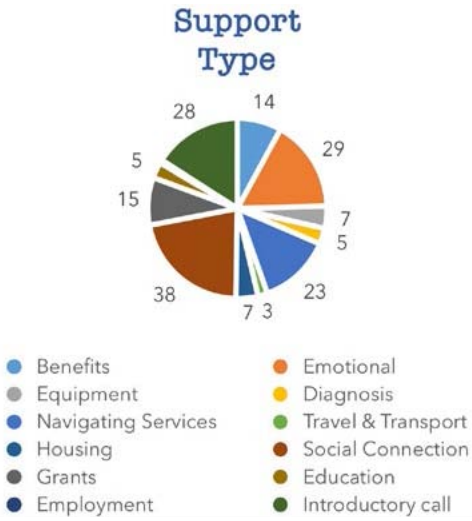
Jane Guy, OBE
Trustee & Secretary to the Board

SCA Report

The Pompe Specialist Care Advisor is tasked with these aims:

- To make a positive difference to the lives of people affected by Pompe disease through providing non-clinical advice, support and information to patients, parents and care-givers.
- To enable people affected by Pompe Disease to access a range of non-clinical services with the object of enhancing and improving their quality of life.
- To provide advice and support for all referred individuals and families on a broad range of quality of life issues including; benefits, housing, finance, aids, mobility and travel, grants, counselling, access, safety, education and employment.
- To ensure people who need the service are not excluded.

Most support is provided via phone, email and Facebook with some home or hospitals visits.



Enabling representation and building a voice:

The Care Advisor promotes opportunities for raising awareness of the patient voice and of GSD as a rare condition.

The Pompe Support Team is vital in providing speakers for events, Industry meetings and conferences. In March the team had one-day training in Health Economics provided by Francis Pang.

Jane Lewthwaite, SCA (Pmope)

Grants

- Nine individual £100 grants from Campbell Burns Metabolic Trust
- Eight In-house individual grants total £1,630
- £560 towards a family holiday from the Sandcastle Trust
- A £500 grant towards white goods R.E.A.C.T
- Two applications made to Merlin's Magic Wand for trips
- One application made to Short Breaks for children

Projects

Family Days

Hospitals are invited to collaborate. SCA arranges the day, including food, entertainment, craft, promotion and volunteers. During the event the priority is to promote AGSD UK and gather registrations. Every individual is followed up with phone calls and information such as leaflets or tailored advice. News items are submitted for the website. Feedback is gathered. Funded by a grant from St James Place.

- Great Ormond Street Children's Hospital
- Evelina Children's Hospital
- Birmingham Children's Hospital

International Pompe Day April 15th

A key aim was to deliver regional events for people with GSD2. For the last four years PST has been supported to deliver a weekend social but this is always in the Midlands and attended by more or less the same small group.

Events were planned in Stoke [a weekend social run by Ben Parker], Swansea, Southampton, London and Bolton. The latter two were cancelled but all the other events were well attended and successful, including new faces.

GSD Kids Camp

September 2018 Kids Camp was provided by Over the Wall and funded by Shire Pharmaceuticals. Benefits for children were extensive. Over the Wall provide a very high quality experience with attendant medical staff. However, their administration had issues and three GSD families who wished to attend missed the event.

"Since the GSD camp my kids have improved 100% with diet compliance; they have understood everything."
Azra

McArdle Group Report

We now have 320 people diagnosed with McArdle disease in the UK, of which about 240 attend the national McArdle disease clinic in London. We support the clinic in a number of ways. The clinic is now organised so that there can be a one hour patient group meeting around lunchtime. This is especially helpful for newly diagnosed patients to meet those with experience of the condition. Our members participated in a number of trials in London and Copenhagen.

In the summer of 2018 we ran our walking course in the Brecon Beacons National Park for the first time, having previously been in the Snowdonia and Pembrokeshire National Parks. Both the main course and the Children & Parents event were located in self-catering accommodation near Hay on Wye. Several of the walks were highlights, including a circuit over Hay Bluff, one that included going behind the cascading water of a waterfall, and a large group summited Pen y Fan, the highest mountain in southern Britain at 2,907 ft. A remarkable achievement for a disparate group of people with a muscle GSD. Both events were great successes with participants coming from England, Scotland, Wales, Ireland, France, Germany, Belgium, the USA and New Zealand.

We supported the continuing development of the international body for muscle GSDs – IamGSD, including helping to host an international workshop on the future of nutrition for McArdle disease, held in London in November. We also supported the German self-help group SHG at their annual conference, including running a few hours practical workshop on the lines of our walking courses.

Andrew Wakelin

McArdle Group Co-ordinator

McArdle Group on Hay-Bluff

Conference 2018

Our conference was held at the Wyboston Lakes Centre near Milton Keynes and was well attended. We welcomed a total of 180 delegates including 49 affected by GSD members. Our crèche was provided by the Bulwell Toy Library for the second year running.

We had colourful displays of the AGSD UK Teens Project, AGSD UK Kids Camp, Hope the GSD2 Sloth Mascot, Mangar Camel Floor Lifting Device event and with Accuvein.

Feedback on the success of the conference remains fairly consistent over the year. The primary reason people attend the conference is for social contact, making friends and networking. Secondly, delegates want to be updated on the latest research, new learning and improving knowledge. Mentioned by some, and also very important, are the contacts they make with medical professionals. Interestingly a lot of people mentioned a wide variety of benefits.

Thanks to the Committee for organising... Good venue, well organised, great food

Lovely to see everybody – good social support network

Updated information on GSD

So many nuggets – all useful

Feedback comments from conference grouped, and in order of frequency;

1. **Social:** meeting friends, renewing friendships, making new contacts.
2. **New research and updates**
3. **Increased knowledge**
4. **Contact with medical professionals**
5. **Personal development & coping:** hope

Excellent all round

Well organized and good content

Financial Report

2018/19 was a challenging year for all and AGSD UK was no exception. We are proud of the fact that the ranges of our services continued to grow and we almost doubled our unrestricted reserves over the year.

Due to an anomaly with our major institutional funders paying us on a calendar basis rather than a financial year, little income has been received in 2018/19 but the sums are now in AGSD UKs account for 2019, making a healthy picture better.

Fundraising and conference income did slip slightly and the board have developed a business plan to help reverse this, including the appointment of a new CEO and a business plan with fundraising strategies.

2018/19 has laid further foundations for the growth of the charity in the coming years. With the advent of improved diagnosis, AGSD UK is poised to welcome more people with FGSDs into our supportive family.

TOTAL INCOME: £167,640

Restricted: £103,566

Unrestricted: £64,074

TOTAL EXPENDITURE: £228,035

Restricted: £161,080

Unrestricted: £66,955

Future Developments

The board of AGSD have set out a business plan outlining a number of strategic ideas for development. AGSD has always been an excellent advocate for patients with GSD, offering practical support for people with GSDs and their support mechanisms. Going forward we want to provide more services like the Specialist Care Advisor and build on the success of our services for children and families.

As genetic testing becomes more prevalent we want to bring the diagnosis time down for GSDs and as a result welcome more people into the AGSD UK family. We will be looking for more resources to do this. We need to diversify our funding base so that our services can continue regardless.

One of the great strengths of AGSD UK has always been its unity across the GSDs. At the start, the focus was on GSD 1 as that was pretty much the only GSD with any level of diagnosis. Now we have strength across most of the GSD types and there is a wealth of treatments and support in some areas. We have funds for most of the GSD types meaning that people can, and do, fundraise to support people with experience in specific types but we are keen to continue to support people with all of the GSDs, not forgetting the rarer and underserved type groups.

Our experience shows that many GSDs are underdiagnosed so the ultra rare GSD of today may become more well diagnosed as we raise awareness. The unique way in which AGSD works, with specific disease types and across the full spectrum is a real strength and something to be proud of.

Thank You

We take this opportunity to thank all our wonderful fundraisers.

We are grateful to all our supporters who have used the following events to raise sponsorship money:

- London Marathon
- London Vitality 10k run
- Prudential Ride London-Surrey cycling events
- Project JOGLE cycle from John O Groats to Land's End led by the daughter of member Joan Wright

We thank everyone who contributed by organising local events such as bake sales or book stalls and especially those who requested donations to AGSD-UK in lieu of birthday presents.

Major donors who support our work are generally biotechnology or specialist nutrition companies:

- Amicus Therapeutics
- Audentes Therapeutics
- Sanofi-Genzyme Ltd
- Spark Therapeutics
- Valerion Therapeutics
- Vitaflo International

Trusts and Foundations who supported us and our members throughout the year:

- St James Place Foundation
- Campbell Burns Metabolic Trust
- Buttle UK
- Sandcastle Trust

We thank all of those who volunteer their time to AGSD-UK. Together the value of their donated services was an amazing £62,320
AGSD-UK could not exist without the wonderful work of our team of GSD Co-ordinators who we would also like to thank sincerely.
Donations from the public raised £19,018

AGSD UK
Association for Glycogen Storage Disease

valerion
Spark THERAPEUTICS
SANOFI GENZYME
Amicus Therapeutics
Vitaflo Innovation in Nutrition
AUDENTES THERAPEUTICS