POMPE SUPPORT NETWORK POST

Issue 1 December 6th, 2019

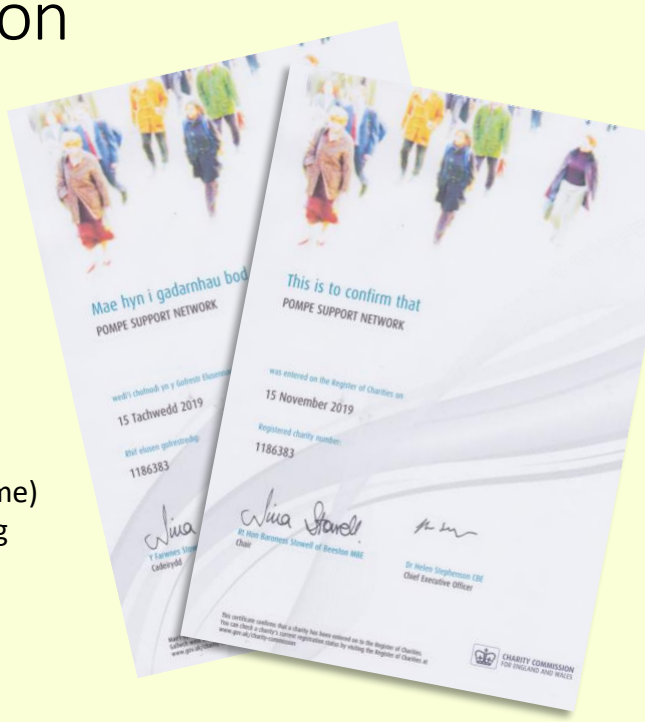
First past the post for Pompe

Welcome to our first issue of our **Pompe Post**, a community newsletter from the Pompe Support Network that will bring regular stories and news of activities by the community, research developments from academia, NHS and industry, and any other items that we, the Pompe Community, feel are appropriate.

If you have concerns, stories, or you spot interesting articles of interest, please let us know and we'll try to include them in future editions.

Charity Commission recognition

The Pompe Support Network was given charitable status on 15th November this year by the Charity Commission of England and Wales. This launched the organisation as an official body representing the UK Pompe community. Many of our activities will have a UK focus, but we welcome full and active participation in our network from the global Pompe community.



One major aim of the Pompe Support Network is to continue as robust participants in the Pompe Model; a collaboration with other patient organisations, healthcare professionals, academic researchers and major players in the orphan drug industry. This model successfully led to the development of Myozyme (Lumizyme) in 2006 and we will be fully involved in shaping research and drug improvements for Pompe disease in the years ahead.

Saved by the Pompe Model

By Allan and Barbara Muir, 6th December 2019

Exactly 30 years ago today, our son Jamie was born. He was a few weeks premature and remained in hospital for ten days until his bilirubin levels returned to normal. Returning home and hugely relieved after that initial worry, we enjoyed a joyful Christmas, proudly presenting our very special family member to relatives and friends.

Jamie's first two years continued to create parental anxiety, however. He was a floppy child, took forever to shake off chesty coughs, and slept with a snore. His snoring became more worrying as it developed into sleep apnoea; his breathing would pause for a moment until it restarted with a loud grunt. Trips to his local paediatrician became more frequent and more fraught, but without ever receiving a diagnosis of the problem. That was until one fateful day we shall never forget. Barbara checked on him one morning to find him blue and floppy in his cot; his breathing had stopped, and his pulse was faint. Despite her panic, Barbara gave light CPR and mouth-to-mouth resuscitation to his delicate, limp body. Thankfully, his breathing restarted.

The paramedics arrived very quickly and took our precious child to the intensive care unit in Portsmouth where he remained sedated and fully ventilated for three days. Those were three very long days during which we held our breath imagining what might result from such a prolonged period of oxygen starvation for our little boy.

Thankfully, on the third day, as the sedation lifted, he gave his first whisper of "mummy" and his recovery began. After being threatened with a tracheostomy we transferred him to Great Ormond Street Hospital (GOSH), London, where he stayed for two weeks, endured a tonsillectomy and adenoidectomy which totally and surprisingly cured his sleep apnoea. Finally, we received the long-awaited and shocking diagnosis - **Pompe Disease.**

That was 1991; when very little was known about Pompe disease; a little Black Book of Neurology told us not to expect him to live beyond his twentieth year. We were devistated.

Now as we celebrate ten years passing beyond that predicted date, and look proudly at a young man in good health, enjoying life to the full and following a promising career in the creative film industry, we have many people to thank for getting us all to this point.

In short, we must give our heartfelt thanks to the "Pompe Model", it was created in the late 1990s by the drive and foresight of people like our own Kevin O'Donnell, the medical research teams in the Netherlands and USA, and by industry leaders willing to work with patient organisations as trusted partners in the quest for the first safe and effective treatment for Pompe disease. We sincerely hope that by working together, the Pompe Model will continue to deliver such incredible life-changing advances for the Pompe community.

Advent of the Pompe Model

Dr Kevin O'Donnell

Allan and Barbara's story reminded me of my very first Pompe meeting, back in 1994. There were a few of us who had recently lost children to infantile Pompe disease. There were adults who were already wheelchair/ventilator dependent. And there was Allan and Barbara Muir, with a young Jamie. I found myself standing in their shoes: "What is ahead for Jamie?"

Things seemed bleak then – there was no treatment for Pompe and, seemingly, no prospect of one. After all, why would anyone be interested in a disease that no one had ever heard of? A Pompe diagnosis was a very lonely experience. And yet, just 5 years later, this picture had changed dramatically – clinical trials were underway, carried out by not one, but two different companies. Trials that led to the approval of Myozyme in 2006. How did that change come about?

Part of the answer is that we were not alone after all. There were Pompe patients and organisations all over the world – and the growing power of the internet helped us to find each other and work together. We learned of, and supported, the pioneering work of Drs Arnold Reuser and Ans van der Ploeg on enzyme replacement therapy. Pharmaceutical companies started to get interested. We found that together, we

were strong; strong enough to make the dream of a treatment for Pompe into a reality. In 1999, the International Pompe Association (IPA) was formed, giving solid form to what was, by then, a worldwide Pompe community.

Along the way, we saw that we had created a new way of bringing about medical progress – the Pompe Model. This involved patients, researchers and industry working together, as equal partners. It hasn't always been easy – but it has been very successful. In October, I spoke at the AMDA conference to celebrate its 20th anniversary. 20 years of remarkable progress, through the Pompe model, with the hope and expectation of more to come.

The progress over the last 26 years has transformed the prospects for people diagnosed with Pompe. Although he won't like being written about, that transformation is personified in Jamie's story (sorry mate, your dad put me up to it).

The Pompe Support Network will work to ensure that the UK Pompe community continues to play a full part in – and benefit from – the Pompe model that we helped to create.

Business news

Astellas Pharma acquires Audentes Therapeutics

You may have been following the development of a gene replacement therapy for Pompe disease by this San Francisco based company over the past few years. Audentes are due to begin clinical trials of AT845 in the Spring next year. Study centres have not yet been announced, so keep an eye on our website for details as they emerge.

The following is taken from the company's press release issued earlier this week:

> Astellas Pharma Inc. and Audentes Therapeutics, Inc. today announced that they have entered into a definitive agreement for Astellas to acquire Audentes at a price of US$60.00 per share in cash, representing a total equity value of approximately US$3 billion.
>
> "Recent scientific and technological advances in genetic medicine have advanced the potential to deliver unprecedented and sustained value to patients, and even to curing diseases with a single intervention," said Kenji Yasukawa, President and CEO, Astellas. By joining together with Audentes' talented team, we are establishing a leading position in the field of gene therapy with the goal of addressing the unmet needs of patients living with serious, rare diseases."
>
> "We are very pleased to enter into this merger agreement with Astellas," stated Matthew R. Patterson, Chairman and Chief Executive Officer of Audentes. "With its focus on innovative science and a global network of research, development and commercialization resources, we believe that operating as part of the Astellas organization optimally positions us to advance our pipeline programs and serve our patients."

Clinical trials

To stay in touch with the locations of current clinical trials for Pompe, and what criteria each has for taking part, we have developed a small spreadsheet on our website under the Research section. We'll update the spreadsheet as new studies are introduced and as old ones close. Currently it includes:

- **COMET, Sanofi Genzyme**: To compare the efficacy and safety of ERTs neoGAA and Myozyme. Patients with LOPD not previously treated with Myozyme
- **VAL-1221, Valerion Therapeutics**: Adult ambulatory and ventilator-free Patients.
- **Mini-COMET, Sanofi-Genzyme**: IOPD Patients Previously treated with Myozyme
- **PROPEL, Amicus Therapeutics**: To assess the efficacy and safety of ATB200 Co-administered With Oral AT2221 in Adult LOPD compared with Myozyme/Placebo
- **SPK-3006, Spark Therapeutics**: Dose-escalation study to evaluate the safety, tolerability and efficacy of a single IV Infusion of SPK-3006 Gene Transfer in adults with LOPD. Not yet open
- **Fortis, Audentes Therapeutics**: To evaluate the safety and preliminary efficacy of AT845 Gene Transfer Therapy in patients with LOPD. Not yet open

You'll find a link to our downloadable spreadsheet here: https://pompe.uk/news/research.

If you're interested in joining one of the studies, you should first discuss it with your consultant at the Highly Specialised LSD Centre you attend. You should be able to transfer to another centre if yours is not offering the trial you would like to join.

Neonatal screening for Pompe disease

Next Generation Sequencing in Newborn Screening in the United Kingdom National Health Service
Int. J. Neonatal Screen. 2019, 5(4), 40; https://doi.org/10.3390/ijns5040040

The above paper presents a very promising technology for the future using next-generation genetic screening (NGS) for rare diseases, but will it help or hinder getting Pompe and other LSDs onto the panel of conditions screened in the UK? Another article in the British Medical Journal (BMJ) questions recent media reports and government statements raising hope of such technologies:

Will genome testing of healthy babies save lives?
BMJ 2019; 367 doi: https://doi.org/10.1136/bmj.l6449 (Published 11 November 2019)

This BMJ paper concludes with:

> *Hancock's suggestion that this scheme is going to be life-saving is misleading. Aside from the ethical and data concerns about testing healthy babies' genomes, the technology doesn't seem to be ready for the NHS and could lead to more harm than good. Any screening programme of this kind would need to adhere to the criteria of the National Screening Committee, including that the benefit gained by individuals would outweigh any harms such as overdiagnosis, overtreatment, false positive results, false reassurance, uncertain findings, and complications*

Whilst we would agree with the reservations of the BMJ report, we would urge the National Screening Committee to expand the panel of screened conditions to include Pompe disease and other rare disorders that have an effective and approved therapy.

Fundraising

The Pompe Support Network is, like most small charities, dependant on generous donations from the public and charitable grants from organisations. We have an area on our website which we will develop to make giving mostly painless, although we also have places in endurance sport events where pain is all part of the fun.

Donations

Please visit our website to learn more about how we can accept donations. Currently we are limited to those listed below, but we will shortly be adding more fundraising opportunities once we receive our HMRC authority to claim Gift-Aid. Expect to see additional opportunities through Virgin Money Giving and Facebook, very soon, for example.

Give as you Live is a neat way of supporting the Pompe Support Network as you shop online, without costing you a penny. Many online companies support giving in this way and the sums can mount up to be significant over time.

Click on this Give as you Live link and you'll be set up in no time to generate donations from your online purchases.

PayPal Donations: Pompe Support Network can receive donations through PayPal using your own account or using a debit or credit card using this link to our PayPal page.

eBay for Charity

We're now able to receive donations through eBay trading. So if you're having a clear-out, please think of Pompe Support Network! You can add us to your charity favourites here:
https://www.charity.ebay.co.uk/charity/Pompe-Support-Network/3832401

Ride London-Surrey 100

16 August 2020

Pompe Support Network has taken five places in this annual cycling event; riding 100 miles from the Olympic park in Stratford, through London and around the Surrey Hills, including Leith Hill and Box Hill, before returning along the Mall to the finish line. Charity places are expensive, so we ask all our team members to pledge sponsorship of £500 or more.

Please contact Pompe Support Network if you would like a charity place or if you have won a ballot place and would like to join our team.

Sign up for future Pompe Posts

Please join the Pompe Support Network by clicking on the button at the foot of our webpages to ensure you receive future updates.

Also please remember to “Like” our Facebook page; that helps us to notify you of any important developments of interest to the Pompe community.