

...to our Autumn 2016 Newsletter

A BIG thank you to all our members for their loyalty and support over the years and a very special welcome to all our new members - we hope you enjoy being part of our group!

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We would like to thank everyone who has contributed to this issue of our annual Newsletter. We hope you find this newsletter informative and are inspired by the Members Stories' and the examples of fundraising for our group, and enjoy the quiz and wordsearch. This Newsletter also pays tribute to our friends who passed away in the last year, and who we remember with deep affection and respect.

Current status of access to intravenous alpha-1 antitrypsin augmentation therapy

by Dr Sandra Nestler-Parr

A number of intravenous alpha-1 antitrypsin (AAT) augmentation therapy products, sourced and purified from pooled human plasma, have regulatory approval, also referred to as a "licence" or "marketing authorisation", in various countries (see Table).

Table. Commercially available AAT products

Product	Manufacturer	Regulatory approval						
Alfalastin	LFB (France)	France						
Aralast NP	Baxter (USA)	USA						
Glassia	Kamada (Israel)	USA, Brazil						
Prolastin (Prolastina Pulmolast)	Grifols (Spain)	Austria, Belgium, Denmark, Finland, Germany, Greece, Ireland, Italy, Netherlands, Norway, Po- land, Portugal, Spain, Sweden, Switzerland						
Prolastin C	Grifols (Spain)	USA, Canada, Argentina, Colombia						
Trypsone	Grifols (Spain)	Spain, Argentina, Brazil, Chile, Mexico						
Zemaira (Respreeza)	CSL Behring (USA)	USA, EU, Brazil						

Current status of access to intravenous alpha-1 antitrypsin augmentation therapy (continued)

However, regulatory approval is not equivalent with patient access to the therapy. In order for any medicine to be universally available to all patients in a country, it needs to be paid for, or "reimbursed", by the country's public health care provider (in the UK this is the NHS). As is the case with many other medicines, several AAT augmentation therapy products have a licence in many countries, but they are not paid for by many public health care providers, as illustrated in the map.

Availability of AAT augmentation therapy in Europe (as of 31 Aug 2016)



One or several AAT augmentation therapy product are:

- Reimbursed
- Reimbursed partially or with conditions
- Not reimbursed

AAT augmentation therapy in the UK

Until last year, no AAT augmentation therapy product was licensed in the UK. In August 2015, CSL Behring's Respreeza[®] received a marketing authorisation by the European Medicines Agency. However, as with all prescription medicines in the UK, patients can only receive the drug if the NHS agrees to pay for it.

The process of formally assessing whether medicines should receive reimbursement by the NHS is conducted separately in each UK nation by dedicated Health Technology Appraisal (HTA) agencies that issue guidance on the use of medicines in the respective national health services. The responsible agencies are the National Institute for Health and Care Excellence (NICE) in England, the Scottish Medicines Consortium (SMC) in Scotland, and the All Wales Medicines Strategy Group (AWMSG) in Wales. There is no separate HTA agency in Northern Ireland, where NICE guidance is usually adopted after checking it is applicable locally.

Within the last year, Respreeza[®] has been the subject of appraisal for reimbursement by all UK HTA agencies. All agencies have different, formal processes for the review of medicines, but they all allow for patient groups to input their views on the disease and the medicine under consideration as part of the appraisal process. The Alpha-1 UK Support Group has therefore represented the Alpha-1 community in the formal review process of Respreeza[®] in England, Scotland and Wales to support the case for the medicine to be made available to all patients who need it. The next sections give an overview of the charity's engagement with the different HTA bodies and the status of access to Respreeza[®] in all UK nations.

England

The HTA process at NICE consists of several stages and starts with a "scoping" exercise, where NICE decides if and how they will assess a new medicine. That decision is based on an application from the drug manufacturer and consultation of NICE with different stakeholders, including clinicians and patient groups.

As part of the scoping exercise, our group submitted a detailed dossier to NICE in August 2015, particularly highlighting: the high degree of unmet need that Alphas



Current status of access to intravenous alpha-1 antitrypsin augmentation therapy (continued)

experience in England due to the absence of a diseasemodifying therapy; how Alpha-1 impacts all domains of patients' lives, their families and carers; how access to Respreeza[®] could improve the quality of life for those patients who are most likely to benefit from the therapy; that inequality currently exists across Europe with some countries offering augmentation therapy whilst others don't; and that the Alpha-1 community strongly supports the availability of Respreeza[®] on the NHS.

Two of our Trustees, Karen O'Hara (previously North) and Sandra Nestler-Parr, attended a "scoping meeting" at NICE in October 2015 and reiterated that Alpha-1 is a complex disease that requires consideration by NICE through a HTA process, which is specially designed for medicines that treat rare and very rare conditions. The decision from NICE as to whether and through which HTA process Respreeza[®] should be assessed is still pending, and we continue to liaise with NICE, NHS England and CSL Behring.

Scotland

The appraisal of medicines for rare diseases in Scotland includes a formal Patient and Clinician Engagement (PACE) process, designed to capture the added value of a new medicine from the patients' and clinicians' perspective. Earlier this year, the SMC invited our charity to participate in this process. We submitted a detailed dossier to the SMC which highlights similar points to those described above for our NICE submission. We drew heavily on the evidence we had previously systematically collected in our national patient survey from Alphas across Scotland, which focussed on the burden of living with the disease and how it affects



every-day life. In June this year, Jane Purves, an Alpha-1 patient from Scotland, and Sandra Nestler-Parr, one of our Trustees, attended the PACE meeting in Glasgow. Jane gave a very personal and moving account of her life with Alpha-1, and Sandra focussed on discussing how the clinical trial results of Respreeza[®] translate into clinical benefits for patients in real life.

Following the PACE meeting, Sandra travelled to Glasgow again in July to observe the SMC's Committee meeting where all information and evidence submitted for Respreeza[®] were discussed. SMC informed us in August that, sadly, the Committee decided not to recommend Respreeza[®] for use by NHS Scotland, as the therapy is not considered by SMC to provide value for money. The SMC stated in their official assessment report that neither the health benefits of Respreeza[®] nor the justification of the treatment cost in relation to its benefits have been sufficiently demonstrated by the manufacturer to gain approval for reimbursement. We are continuing to liaise with CLS Behring to discuss alternative options for making the therapy available for patients who are likely to benefit from it.

Wales

At the time of writing, AWMSG is in the process of assessing Respreeza[®] for the use in Wales. Our charity has produced a patient group submission to AWMSG in early September 2016, providing the patient perspective of living with Alpha-1 and the benefits we expect from patient access to augmentation therapy. AWMSG will make a decision on whether or not to reimburse Respreeza[®] in November.



In summary, we are very disappointed that Respreeza[®] has not been recommended for use in Scotland. The charity continues to engage in all relevant processes and do whatever it can to achieve a better outcome for England and Wales, and to explore alternative routes for patient access to AAT augmentation therapy in the UK.

An update on recent advances in the development of novel treatment approaches for alpha-1 antitrypsin deficiency

by Dr Sandra Nestler-Parr

Inhaled alpha-1 antitrypsin

In March 2016, Kamada Inc., an Israeli plasma-derived protein therapeutics company focused on developing treatments for rare diseases, announced the submission of a Marketing Authorisation Application (MAA) with the European Medicines Agency for the company's proprietary, inhaled alpha-1 antitrypsin (AAT) therapy as a treatment for AATD. The application is based upon results from a Phase 2/3 randomised, placebo-controlled clinical trial that evaluated the safety and efficacy of Kamada's inhaled AAT in 168 patients. Study results demonstrated a reduced decline or improvement in lung function parameters in patients who received inhaled AAT compared to patients on placebo.

"The submission of the MAA for inhaled alpha-1 antitrypsin to treat AATD is a major step toward bringing another treatment to AATD patients. This study is the first study ever that shows inhaled AAT's ability to reduce the decline in FEV_1 in a patient population suffering from frequent exacerbations of dyspnea and coughing. "I am looking forward to the regulatory authorities' approval for the benefit of AATD patients," stated Dr Jan Stolk from the Department of Pulmonology at Leiden University, Netherlands, and Principal Investigator of the trial.

These positive results were further supported by a more recent Phase 2 clinical trial of Kamada's inhaled AAT that assessed the levels of the inhaled AAT in the lung, its biological activity and capacity to protect the lungs from inflammatory damage. The study results are promising and suggest that inhaled AAT is the most effective mode of treatment for reaching the primary sites of potential lung injury, and restoring AAT inhibitory capacity. Kamada will need to confirm these findings in a larger, definitive clinical study.

RNA inhibitor-based therapies

In the late 1990s, a biological process was discovered, whereby small RNA molecules trigger the suppression, or "silencing", of gene expression, thereby regulating the synthesis of proteins in the cell. This natural cellular process is called RNA interference, or RNAi. Following its discovery, it became quickly evident that RNAi has enormous potential for the development of a new class of medicines, resulting in a Nobel Prize for its discoverers in 2006.



Schematic. RNAi-based therapies block the production of faulty proteins

An update on recent advances in the development of novel treatment approaches for alpha-1 antitrypsin deficiency (continued)

RNAi technology is based on blocking the expression of faulty genes that would, otherwise, facilitate the synthesis of proteins that cause disease. The mechanism of action of RNAi-based therapies is illustrated in a simple schematic. In contrast to the RNAi approach, many traditional therapies target proteins, thereby interfering at a later stage of the molecular disease process. Several biotech companies are currently developing RNAi-based therapies for a variety of conditions. Two US-based companies are currently working on the development of RNAi medicines for Alpha-1. These therapies aim to reduce the production of inflammatory Z-AAT protein (=faulty AAT) in the liver of patients with the PiZZ genotype. Mutant Z-AAT protein polymerizes in the liver and is believed to cause Alpha-1 associated progressive liver disease.

The continuing progress in the development of effective RNAi therapies for Alpha-1 is promising, and recent developments are summarized below.

Arrowhead Pharmaceuticals

The California-based biotech company has recently changed its name from Arrowhead Research to Arrowhead Pharmaceuticals. In last year's Newsletter we reported about Arrowhead's ongoing Phase 1 clinical trial, in which their RNAi drug candidate ARC-AAT is being administered to humans for the first time. The Phase 1 study is designed to evaluate the safety, tolerability and pharmacokinetics of ARC-AAT and its effect on AAT levels in blood circulation. This multicentre study has been conducted in several countries; the only UK trial site is Queen Elizabeth Hospital in Birmingham. Recruitment for this Phase 1 trial is still ongoing, and key inclusion criteria are a PiZZ phenotype and an FEV₁ of at least 60% predicted. We are liaising with Arrowhead for the publication of an advert for this trial on our group's website. We are currently awaiting ethics approval which we expect in late October this year, so please keep checking our website for news and more detailed information on this trial and the opportunity to participate in it.

In a press release dated 6th September 2016, Arrowhead announced the upcoming initiation of a Phase 2 clinical study of ARC-AAT, aimed at assessing safety and tolerability and to determine the effect of multiple doses of ARC-AAT on levels of alpha-1 antitrypsin in circulation and in the liver, as evidenced by changes in liver biopsy in patients with AATD. Arrowhead's Chief Medical Officer and Head of Research & Development, Dr Bruce Given, commented: "There remains no medical treatment for the liver disease associated with AATD, which is increasingly being recognised by patients and physicians as a serious problem. Our Phase 2 study should give us, and the AATD community in general, the first insights into whether ARC-AAT can stop the progression of liver disease and possibly even allow the liver to recover and heal existing damage. This would be a significant breakthrough for patients."

Our charity welcomes this news, as it indicates that the ARC-AAT Phase 1 trial was successful in demonstrating good safety and tolerability results. According to Arrowhead's press release, the Phase 2 trial will be conducted at multiple centres in Canada, Ireland, and Sweden. Additional centres may be included in other countries, pending regulatory and ethics review. However, we understand from Arrowhead that there are currently no plans for a Phase 2 study centre in the UK.

For more information please visit Arrowhead's homepage at http://arrowheadpharma.com or the international clinical trials database at https:// clinicaltrials.gov/ct2/show/record/NCT02363946? term=arrowhead&rank=13.

Alnylam Pharmaceuticals

Alnylam, headquartered in Boston, is another US biotech company that focusses on the development of RNAibased therapies for an array of different diseases, including Alpha-1.

According to the international clinical trials database, Alnylam is currently conducting a Phase 1/2 trial of their RNAi drug candidate ALN-AAT which is also aimed at reducing the synthesis of mutant Z-AAT protein in the liver of Alphas. The purpose of this trial is to evaluate tolerability, the safety, pharmacokinetics and pharmacodynamics of subcutaneously administered ALN-AAT in healthy adults and PiZZ patients with liver This trial is not currently recruiting study disease. subjects, and we have no information on whether any UK centres are participating in this study. The estimated completion date is stated as January 2017.

For more information please visit Alnylam's website at http://www.alnylam.com/ or the international clinical trials database at https://clinicaltrials.gov/ct2/show/record/ NCT02503683?term=alnylam&view=results.

An update on recent advances in the development of novel treatment approaches for alpha-1 antitrypsin deficiency (continued)

Gene therapy

Gene therapy is the introduction of normal genetic material into cells to replace missing or defective genes in order to synthesize a beneficial protein and correct genetic disorders.

The concept of gene therapy has long represented an appealing potential treatment approach for genic diseases based on single gene mutations, such as Alpha-1. Various approaches to correct Alpha-1 through gene therapy approaches have so far been tested in cell and animal models, but have proven challenging in achieving successful correction of the PiZZ gene defect in practice.

A recently published study from researchers at the Center for Regenerative Medicine of Boston University and Boston Medical Center describes the successful delivery of a copy of the normal AAT gene into lung cells of experimental mice. Therapeutic AAT protein levels were observed within the lung epithelial lining fluid of the mice, resulting in reduced severity of experimental emphysema. The corrected gene persisted in mice lung cells for at least one year. "These results support direct transgene delivery to the lung as a potential alternative approach to achieve the goal of developing a gene therapy for AATD," explained corresponding author Dr Andrew Wilson.

(Refs.: Boston University Medical Center; Payne et al. *Multilineage transduction of resident lung cells in vivo by* AAV2/8 for α 1-antitrypsin gene therapy. Mol Ther Methods Clin Dev. (2016); 3:16042)

A Phase 1 clinical trial by Annapurna Therapeutics is being planned that uses an adeno-associated virus to act as the vehicle to carry the "good" AAT gene into lung cells. This vector cannot multiply and is made harmless by removing all its genes, which are replaced by a normal copy of the AAT gene. The vector with the gene is the experimental drug. This study aims to test the safety of the experimental drug and to examine the effects of adding a normal copy of the AAT gene to the lungs of affected individuals. This small study is not yet recruiting, is due to be conducted in the US and is stated to be completed in early 2018. For more information, see https://www.clinicaltrials.gov/ct2/show/record/ NCT02168686?

term=alpha+1+antitrypsin+deficiency&rank=14.

Alpha-1 Antitrypsin Deficiency – an inherited risk of disease

by Professor David Parr and Dr Beatriz Lara, University Hospital Coventry

A guide to genetic testing

When people get diagnosed with alpha-1 antitrypsin deficiency (AATD) they naturally have a lot of questions about the condition and, when they see us in our AATD clinic, it is not uncommon for them to ask questions like "How did I get it?", "What do the results of my genetics test mean?" and "How is my health likely to be affected by this?". This kind of question doesn't come as a surprise because AATD is a complicated condition and it is understandable that patients may need to have information repeated or may want to hear it from more than one doctor, since most doctors have slightly different ways of explaining things. However, hearing this kind of question from patients attending a specialist clinic is occasionally a cause for concern when it becomes clear that the test for AATD may have been

performed without their prior knowledge or without an explanation of why the test was being done and what the answer may mean. Of course, these issues are important for any clinical test but they are even more important with testing for AATD, because it is a genetic test.

AATD is an inherited condition, which means that it may be passed on in families through the genes; each person has two copies of the AAT gene, one inherited from their father and the other from their mother. There are approximately 100 variations of the AAT gene and it is generally the case that both copies of the gene have to be an abnormal variant in order to cause a significant risk to health. When both AAT genes are abnormal, there are many potential health effects that can lead to various lung and liver problems (as well as other rarer

Alpha-1 Antitrypsin Deficiency – an inherited risk of disease (continued)

conditions). On top of this, the influence of the environment and personal habits, such as smoking and drinking, play major roles in determining whether and to what extent an individual person experiences ill-health, if at all.

All of these influences taken together create a complicated and wide range of potential scenarios! It takes time and specialist expertise to be able to provide patients with an understandable and accurate explanation of the commonest scenarios, but the time to do this is *before* the genetic test for AATD is performed; this is called 'pre-genetic test counselling'. Once the test results come back, it is possible to give more precise information about what the test result means, and this explanation may need to include the findings of other tests such as breathing tests and scans; this is called 'post-genetic test counselling'.

The terms 'pre-genetic test counselling' and 'postgenetic test counselling' apply to genetic testing in general, not just to AATD testing. They are just part of the best practice standards that are applied to the use of genetic testing and which should be followed by all doctors, whether they are GPs or hospital doctors, and by companies that provide what is known as 'Direct to Consumer' (DTC) testing.

In the UK, there are several organisations responsible for providing guidance to health care professionals and the public on the use of genetic tests. The guidance on best practice for doctors and advice for users of health services provide a good framework and lists the principles that should be adhered to when undertaking genetic testing. Some of this information is quite complex and the details differ according to whether the genetic testing is being done for someone with symptoms of 'disease' or for someone who is well but is related to an affected patient. However, there are some common themes and principles that should be followed in order to avoid confusion and worry.

In any case, patients should always speak to their GP or their hospital doctor about the possibility of genetic testing if they think they may need it. Best practice includes obtaining a referral from a GP, or specialist doctor if applicable, for genetic testing to be carried out. If the doctor thinks genetic testing may be appropriate, they should usually refer their patient for genetic counselling as well. It is also important to consider all implications of having a test for a genetic disease: some people may, for example, experience difficulties in finding or renewing insurance cover (for travel, private health or other insurances) after having had a genetic test, irrespective of the outcome.

The use of postal test kits for genetic conditions, where testing kits may be sent through the post to family members of known patients (often called 'presymptomatic testing'), should also follow the best practice guidelines for genetic testing. However, the logistics of using postal test kits clearly present an obstacle to being able to adhere to best practice standards, particularly the ability to provide direct face-to -face genetic counselling. Leaving the counselling to a family member with AATD, even if they have a good understanding of how the condition affects them personally, or searching for information about the disease on the internet, is not a substitute for professional genetic counselling. The use of postal test kits for genetic testing is therefore not recommended by the NHS.

We encourage anyone wishing to learn more about genetic testing to read the information available on the websites below, using the links provided. The specialist NHS clinics for AATD patients that are now running in Birmingham, Cambridge, Coventry, and London should also be able to provide advice and guidance by telephone or email. However, since there is no substitute for face-to-face discussion in a clinic appointment with an expert, some patients may prefer to be referred by their GP for an outpatient consultation. If you would like to do this, you simply have to ask your GP to make a referral through 'Choose and Book' to the expert clinic of your choice – often it takes only one appointment to put minds at rest.

Suggested links:

NHS UK Genetic Testing Network: http://ukgtn.nhs.uk/

NHS Choices – Genetic testing and counselling: http:// www.nhs.uk/Conditions/Genetics/Pages/genetic-testingand-counselling.aspx

Genetic Alliance – Benefits and risks of genetic testing: http://www.geneticalliance.org.uk/information/servicesand-testing/benefits-and-risks-of-genetic-testing/

Alpha-1 Antitrypsin Deficiency – an inherited risk of disease (continued)

Some more detail on the genetic aspects of Alpha-1

Apha-1 antitrypsin (AAT) is a protein that is made predominantly by the liver and, from here, it is transported into the blood stream for distribution throughout the body. The level of AAT in the blood is dependent primarily on an individual's genetic makeup. When both genes are the normal 'M' genes, there is usually an increase in production and blood levels of AAT at times of physical stress, such as infections. This process is thought to be essential for the protection of the tissues against uncontrolled inflammatory damage. For reasons that are still unclear, the lung appears to be particularly susceptible when this protection from AAT is missing or reduced. The liver produces several grams of AAT every day. Under specific circumstances, for instance during infections or exposure to inhaled toxins, the amount of AAT produced can be greatly increased to protect the organs against damage.

Different Alpha-1 genes and genotypes

Every individual receives one copy of the AAT gene from each of their parents. The gene carries the "building plan" for the production of AAT. Several variants of the AAT gene exist, and they are named with different capital letters to differentiate them. The most common variant of the gene is called M. This gene contains the information to produce normal AAT protein. An individual who inherited a normal M variant from both their father and their mother will have the genotype MM. Every AAT molecule that is produced in an MM individual has a normal structure and function. The MM molecules are transferred from the liver into the blood stream, and the blood concentration of AAT will be normal.

Other variants of the AAT gene produce proteins with molecular structures that are different from the normal M protein. For instance, the AAT protein of the S gene variant has a molecular structure that makes it prone to bind to other S molecules; this process is called "polymerisation" and can produce long chains of S molecules. As a consequence of polymerisation, part of that S protein is retained in the liver, thereby reducing the number of AAT molecules that are transferred into the blood stream. Consequently, individuals carrying one S variant and one M variant (genotype MS) have a slightly reduced concentration of functional AAT protein in the blood stream; although the normal M protein is transferred into the blood, a lot of the S protein gets retained in the liver.

Individuals carrying two S variants (genotype SS) have lower AAT blood levels than individuals with the MS genotype, because all of their AAT proteins will be the S type which polymerises and is retained in the liver.

Despite its abnormal molecule structure, the S type protein keeps its function, so the protein is able to protect the lungs against toxins and other stresses. Individuals with MS or SS genotypes are not at risk of developing any AATD-related lung disease, because their AAT blood levels are only slightly reduced, and the circulating AAT protein functions correctly.

On the other hand, the Z variant of the AAT gene produces a protein with a strong binding affinity to other



Risk of lung disease from blood AAT levels associated with common AATD phenotypes

(Modified from D. Waldrop)

Alpha-1 Antitrypsin Deficiency – an inherited risk of disease (continued)

Z molecules, forming polymers of Z protein that accumulate in the liver. These Z polymers can damage the liver cells causing chronic liver disease. A small number of Z molecules do get transferred to the blood stream, however, the abnormal structure of the Z variant makes the protein biologically inactive, resulting in a significantly reduced capacity to protect the lungs from damaging agents. Consequently, individuals with two Z genes (genotype ZZ) have very low levels of AAT protein in the blood and are more susceptible to develop AATD-associated health problems.

The combination of the two most common mutations of the AAT gene, S and Z, (genotype SZ) produces an intermediate situation. S and Z molecules can form polymers that are mostly retained in the liver, but S protein is not as functionally inactive as abnormal Z.

The scientific community considers that a blood concentration of 11micromolar or 0.5g/L AAT is sufficient to protect the lungs from damage. Only individuals with very low levels, such as the ZZ genotype and a small proportion of SZ, have blood levels that are below this supposed "protective threshold" and are considered to have clinically severe AATD.

How is alpha-1 antitrypsin deficiency diagnosed?

First of all, the amount of AAT protein circulating in the blood stream is measured: a normal concentration in a routine blood test excludes the possibility of deficiency, so no further tests are needed.

In people who have a low level of AAT in the blood, the next tests are aimed at identifying the type of AAT protein that circulates in the blood or which AAT genes the person has inherited. This can be done using two different approaches:

- Identification of the type of AAT protein (called 'phenotyping'): AAT protein is isolated from a person's blood and analysed with a technique that separates all of the blood proteins. This technique is called protein electrophoresis.
- Identification of the gene variant (called 'genotyping'): the genetic material (DNA) of an individual is analysed to identify which gene variants they have inherited.

Who needs to be tested using phenotyping and genotyping?

The World Health Organization (WHO) recommends that all patients who have COPD and all close relatives of a patient with AATD should have a test to check the level of AAT in their blood. This means that the first degree relatives (the parents, brothers, sisters and children) of patients known to have the SZ or ZZ genotype ought to be tested. All people who are found to have a low level of AAT in their blood should then have either a phenotyping or genotyping test, depending on specific circumstances, such as the capability of the testing laboratory and individual patient features.

At the end of the day, it is the right of every individual to decide on what they feel is best for them, but in order to make a rationale decision they need clear and reliable information. This is particularly important when the medical issues are so varied and complicated as they are in AATD, and the NHS AATD clinics that are now running across England are there to provide the necessary support.

Update on centres of excellence for routine clinical management and research in AATD in the UK

by the Board of Trustees

Historically, teams of researchers based in Professor Stockley's group in Birmingham and in Professor Lomas' group in Cambridge (now London) have placed the UK on the world map of AATD research. Over the last decades, research from these UK centres have contributed significantly to the current body of knowledge about AATD, the underlying mechanisms of AATD-associated lung and liver disease and its

progression, and to the development and testing of novel therapeutic strategies to treat AATD.

These centres continue to thrive and, in addition, many of the clinicians who received their training and have undertaken AATD research under Professors Stockley and Lomas have since moved to other locations and are establishing specialist AATD services. There are other

Update on centres of excellence for routine clinical management and research in AATD in the UK (continued)

established centres of excellence in respiratory medicine that are also seeing AATD patients.

In line with our ongoing campaign for better and more equitable patient access to expertise in the diagnosis and continuous multidisciplinary management of AATD across the UK, the charity is undertaking an audit of all tertiary hospitals in the UK and of all pharmaceutical companies with an interest in AATD, with the aim of gaining a better understanding of:

- the geographical distribution of clinical expertise and interest in AATD across the UK;
- how this translates to specialist clinical services available to AATD patients; and
- which research projects are ongoing and what novel therapeutic approaches are in development.

So far, we have reached out to hospitals in Birmingham, Cambridge, Coventry, Edinburgh, Leicester, London, Nottingham, Manchester and Southampton. All of these centres have either participated in a recently concluded National Institute for Health Research (NIHR) study aimed at in-depth characterisation of a group of patients with severe AATD, or they were already known to us as providing specialist NHS clinics for AATD. We are in the process of contacting other centres around the country, including those with specialist paediatric expertise and transplant centres.

Once we have received detailed feedback from all centres, we will finalise our "UK Alpha-1 Map" that will be published on our group website and will provide detailed information about the clinical and research activities available to Alpha-1 patients across the country, as well as contact details for each of the centres.

Here is a preliminary summary from the responses we have received so far:

Centres with specialist multidisciplinary NHS clinics for AATD are currently run at Birmingham (Queen Elizabeth Hospital; http://www.uhb.nhs.uk/alpha-1-antitrypsindeficiency-aatd-clinic.htm), Cambridge (Addenbrooke's Hospital; http://www.cuh.org.uk/addenbrookes/services/ clinical/respiratory_medicine/

respiratory_medicine_index.html), Coventry (University

Hospital Coventry; http://www.uhcw.nhs.uk/our-services/ a-z-of-services?sID=124) and London (Royal Free Hospital, https://www.royalfree.nhs.uk/services/staff-a-z/ dr-john-hurst/). All these centres offer a 'one stop' service for AATD-associated liver and lung disease, genetic testing and counselling, and they have access to other specialties as required (e.g. paediatrics, radiology, dermatology, transplantation etc.).

All other centres see AATD patients within specialist COPD clinics and/or have established links with the above mentioned centres that provide dedicated AATD clinics, for transition of care or shared care where appropriate. All centres offer lung function testing (respiratory physiology).

Southampton is in the process of establishing a specialist AATD clinic with a multidisciplinary team, and Leicester is considering the establishment of a dedicated AATD clinic in due course; both centres will keep us updated.

The majority of these centres also have an interest in doing research. There are two main types of clinical studies that patients can participate in:

- interventional studies (often called 'drug trials'), most commonly conducted on behalf of pharmaceutical companies, in which the safety and efficacy of new treatments are assessed over a period of time; and
- observational studies, in which the disease itself is studied. This includes, for example, a lot of the research that has been performed at ADAPT over the last two decades, where many different measurements of patients are made repeatedly over long periods of time to better understand how the disease progresses and how the changes can be best measured.

Anyone who is interested in taking part in clinical research should contact their specialist for details. Our group is liaising with clinical centres and industry to create a comprehensive list of ongoing AATD research activities in the UK which will be published on our website in due course.

The Alpha-1 UK Support Group's commitment to raise awareness of AATD and gain support from a broad range of stakeholders continues...

by the Board of Trustees

Within the last year, we have continued to raise awareness of Alpha-1 and strengthen the public profile of our charity through: formal engagement with Health Technology Appraisal agencies across the UK to support access to augmentation therapy (for details please see the article on pages 1-3); continued discussions with NHS England on our ongoing campaign to establish a nationally commissioned Specialised NHS Service for AATD; and our continuing engagement with politicians who have taken a special interest in AATD and have supported our cause for several years.

In last year's newsletter, we reported about our work with Mark Pawsey, MP for Rugby, who has been a longstanding supporter of constituents with AATD and the wider Alpha-1 community. Last year, Mr Pawsey visited University Hospital Coventry to learn about the West Midlands NHS Alpha-1 Antitrypsin Deficiency Service that was established in 2015 in Coventry (at University Hospitals Coventry and Warwickshire) and Birmingham (at Queen Elizabeth Hospital). At these clinics, Alpha-1 patients have access to a comprehensive team of experts so that all aspects of their condition can be addressed and they have the time to discuss what matters to them. This is particularly important if patients have had to travel long distances to visit the clinics.

We are delighted that Mr Pawsey returned to University Hospital Coventry for a recent follow-up visit on 23rd August, where he met with several Alpha-1 patients and clinicians of the multidisciplinary AATD team. Mr Pawsey commented: "It was a good opportunity to meet with Dr Parr and patients who have Alpha-1 Antitrypsin Deficiency and learn what progress has been made on treating this condition."

Mr Pawsey continued: "I first had my attention drawn to AATD by one of my constituents and I have taken an interest in the condition, including arranging a meeting with Dr Parr and the Health Secretary, Jeremy Hunt, during the last Parliament to raise his awareness of Alpha-1 and how it can be treated. I hope that we can continue to build on the progress that has been made."

Professor Parr from University Hospital Coventry added: "Mark is already known as a champion of the cause for patients with AATD and we're very grateful that he took time to visit us to learn more about patients' experiences and our rapidly developing service. We've had great feedback from our patients, some of whom travel from far afield, and it is clear that we are providing the kind of comprehensive clinical care that they have been seeking for many years."



L-R: Karen O'Hara (Patient & Chair, Alpha-1 UK Support Group), Michael Bartlett (Patient), Susan Townsend (Ward Sister), Robert Dore (Patient), Prof David Parr (Consultant Respiratory Physician), Mark Pawsey MP, Dr Beatriz Lara (Consultant Respiratory Physician)

Mr Pawsey reassured the patients and clinicians of his continuing commitment to support the Alpha-1 community and, following his visit, he released a very positive account of the meeting on his website (https:// www.markpawsey.org.uk/news/midlands-expertise-

treating-rare-respiratory-disorder-praised-mark-pawseymp). The Coventry Observer also reported about the meeting, (http://rugbyobserver.co.uk/view-edition/? ed i=85&ed u=/Rugby/2016/09/08&ed p=40, page 12).

This is a very promising example of how engagement with politicians can help us to have a voice and draw attention to Alpha-1 and patients' needs. We would like to encourage Alpha-1 patients to speak to their MPs about their disease and highlight the unmet need Alphas experience across the UK. Politicians need to understand the importance of improving patient access to expert clinical care and beneficial treatments, particularly in view of the current absence of specific treatment options for AATD in the UK.

14th Annual Social and Information Day, Lincoln 2015

Lincoln, September 2015

Our 14th annual gathering was held in Lincoln again at the same venue as 2014 at the Bentley Hotel Leisure Club and Spa.

The event is one of the largest gatherings of Alphas and their families in the UK where Alphas can meet and socialise with fellow Alphas and be updated by leading experts in the field of Alpha-1 Antitrypsin Deficiency.

We had some great speakers and we thank them all for sparing the time to attend our event:

Professor Rob Stockley is well know to us as the Director of ADAPT, the AATD research programme based at Queen Elizabeth Hospital Birmingham.

He provided an update on the research activities at ADAPT.

Professor David Parr has in the past worked with Prof Stockley at QE Hospital Birmingham. For the last



...Prof Parr (I) with his Alpha-1 team

10 years he has been a consultant at University Hospitals Coventry & Warwickshire, where he is the Clinical Director of Cardio Respiratory Services. He is the Lead Clinician in the specialist NHS Alpha-1 Antitrypsin Deficiency clinics at Coventry. He updated us on the West Midlands NHS AATD Service.

Peter Leone Vice President of Arrowhead Pharmaceuticals also attended our meeting in Lincoln last year and spoke to us about their research programme on AATD in relation to the liver. His first



...Peter Leone, Arrowhead Pharmaceuticals

experience of British Bingo was obviously enjoyed so much he's joining us again this year in Cheltenham.

Dr Surekha Frith, Head of Medical Affairs UK & Ireland at CSL Behring, gave an update on CSL's efforts to obtain reimbursement for their Alpha-1 Proteinase (A1-PI) Augmentation Therapy (Respreeza).

The 15th Annual Social Gathering will be held in Cheltenham at the Jury's Inn on the weekend of Saturday 24th September 2016. You can come for the weekend or just as a day visitor on the Saturday.

We look forward to seeing you



...Jill and Jackie

...four delegates who just could not wait to get their hands on the latest Newsletter.

...Norah, Elaine and

John with Carol



....catching up - Joe and Bob



Connecting Our Community



Gonny Gutierrez, Alpha-1 Global Director

More than two years after the launch of Alpha-1 Global in October 2013, much continues to happen in the Alpha-1 world at large! 2014 was a year of networking with existing Alpha-1 patient organisations, key Alpha-1 researchers and physicians, and laying the foundation for the program and the building of a designated Alpha-1 Global website. 2015 served as the year to "get together" again with the 5th Alpha-1 Global Patient Congress and 2nd Research Conference on Alpha-1 Antitrypsin, in Barga, Italy.

After the Patient Congress, the Global Steering Committee decided to tackle the following activities for the remainder of the year:

Central/Eastern European Conference

On October 16-17, Alpha-1 Global's Frank Willersinn and Gonny Gutierrez participated as speakers during the second Central/Eastern European Network Conference on Alpha-1 Antitrypsin in Warsaw, Poland, organized by Dr. Joanna Chorostowska, who had joined us in Barga. Attendance resulted in networking with Alpha-1 physicians from the following 9 countries: Poland, Czech Republic, Bulgaria, Romania, Estonia, Ukraine, Belarus, Lithuania, and Russia.

Nordic Pilot

Since the 5th Patient Congress, the leaders of the Alpha -1 groups in Norway, Sweden, and Denmark have done an amazing work. The "Nordic Pilot" serves as a prototype for building a strategic collaboration between regional Alpha-1 communities as together they seek improved access to care in their countries.

Leaders from the three countries are in close communication with each other and met late 2015, in

Oslo for an Alpha-1 Global Advocacy Toolkit conducted by Karen O'Hara, and to discuss next steps toward their goals, which includes continued networking within their own countries. Additionally, a symposium on augmentation therapy for key physicians in Denmark was held on April 12, 2016. Several experts from the European Alpha-1 community contributed to this strategic meeting, which resulted in close collaboration with the Danish Lung Association and Rare Diseases Denmark. taking concerted steps towards recommending augmentation therapy reimbursement in Denmark.

As a collaborative effort, the Nordic countries are preparing to conduct an in-depth Alpha-1 Questionnaire later this year, similar to the one conducted as part of the Alpha-1 Campaign in the UK. Steen Bengtsson, from The Danish National Centre for Social Science (SFI), has agreed to conduct this project and provide analysis and reporting of the outcomes for use in outreach to local authorities..

Latin American Summit

As follow-up from the 5th Patient Congress, Alpha-1 Global conducted a Latin American (LATAM) Summit on December 4-5, 2015, which was attended by more than 40 representatives from 9 different countries. Excitement and interest to work together in Latin America was expressed among the patient representatives from: Chile, Panama, Costa Rica, Uruguay, Venezuela, Ecuador, Colombia, Argentina and Brazil. Dra. Alejandra Rey, M.D., a pulmonologist from Montevideo, Uruguay, who was present in Barga last April, is spearheading a





Connecting Our Community

(continued)

committee of Alpha-1 physicians from the region to amend the **Standards of Care for Alpha-1 in Latin America.** The first meeting for this purpose took place after the Alpha-1 Patient Summit in Buenos Aires, and several more are scheduled throughout 2016 to accomplish this important task.

Carlos Cambon, one of Alpha-1 Global's newest Steering Committee members, serves as the representative for South America and continues to work closely with Alpha-1 patients, leaders and physicians in the region.

Patient Organisation Launches

Alpha-1 Global was able to assist with the following Patient Organisations launching in Europe: the Netherlands, Switzerland and Poland.

After the LATAM Alpha-1 Global Summit, Argentina launched an official patient organisation and Chile is starting a designated Facebook page for Alphas in their country.

In both Europe and Latin America, online meetings for Alpha-1 leaders in the region will be scheduled regularly in order to strategically move the mission forward.

Alpha-1 Global Advocacy

As we continue to collaborate as a global community on increasing awareness, detection and access to care for Alphas around the world, advocacy training remains high on our list.

a) EU Alpha-1 Standards of Care

In 2011, the European Policy Recommendations for Alpha-1 Standards of Care were crafted by an expert group of Alpha-1 leaders and physicians in collaboration with Rohde Public Policy in Brussels, Belgium. In 2016, the RAPID trial results are included in an updated document, in order to serve as an integral part to a new and improved outreach on a European and national level.

b) European Advocacy Training

On April 27, 2016, Alpha-1 Global conducted Advocacy Training for European country representatives in Milan, Italy. Each attending country shared their successes and goals and used the training and the toolkit to prepare their strategy for the coming months. A concerted effort will be made to include the updated European Recommendations as part of the ongoing patient advocacy strategy in Europe.

c) Advocacy Toolkit Translation

Thus far, we have translated and distributed the Advocacy Toolkit into the following languages: Spanish, German, French, Portuguese, Romanian and Italian. Additional translations will be made available as the need arises.

Additional Global Efforts

a) Alpha-1 Global Website

In our efforts to serve a global community with possibly limited understanding of English, we want to create a less overwhelming first impression on our website, by using less text and emphasizing a simplified user navigation concept. We believe that this will help to engage our audience more effectively, increase access to electronic tools and collateral, and show intentionality in establishing international relations. Additionally, we will include language translation tools.

b) Alpha-1 Global eNewsletter

The eNewsletter allows us to regularly connect with our global community with relevant and current Alpha-1 information. The issues are published every other month.

c) Alpha-1 Global Patient Congress in 2017

Another key element to building a collaborative global network is providing education and interaction between patients, physicians and researchers in the area of Alpha-1. Per the request of all three "communities" involved, we will continue to organize our biennial Patient Congress & International Research Conference on Alpha-1 Antitrypsin. The events will take place on **April 5-8, 2017**, at the InterContinental Hotel in Lisbon, Portugal.

www.alpha-1global.org



Member Stories - Mark Bradford

"Mark wrote the following as an update of his situation back in June 2016. Many of you know that he received a call for double lung transplant on 1 September but passed away soon after. The tributes that have appeared from family and friends have shown how respected, liked and loved Mark was by all who knew him. He was a friend to us and supported us with humour and shared experience amidst his own considerable struggles. One photo caption just said "always smiling". Perhaps the ultimate accolade to Mark's commitment to the cause was that fact that he received his call for transplant on the evening when he had been recording an article with the BBC for their South East News encouraging organ donation."

Since my single lung transplant in 2014 I have had several setbacks, unfortunately resulting in me requiring a double lung transplant. I still believe I am the luckiest person around and hope my story will inspire you and not worry you.

I was only considered for a single lung transplant due to an operation I had a few years earlier sticking the lining of my right lung to the chest wall to stop fluid entering the pleural space. One of the possible complications post-transplant is hyperinflation, a build-up of pressure in the untransplanted lung causing that lung to expand. This started to happen about two months after transplant. I remember going to the Alpha one meeting in Lincoln and feeling fantastic and a month later I was in A&E being transferred to Papworth. Papworth also discovered that the main bronchia had partially collapsed in the transplanted lung, this could have been caused by the hyper inflated lung pushing on where the surgeon clamped the bronchia during surgery, it's another risk you take.

The consultants decided to design a stent to re-open the airway, they knew of a company in France that made these specialist stents but it would take about a month to come. The stent arrived just before Christmas I think it was about the 20th December 2014, they fitted it and all seemed great I could breathe again and I could go home for Christmas Day. On the 27th December though the stent moved and was stuck in between the two lungs it was so painful: imagine having a metal tube scraping up and down your airway. My local hospital transferred me back to Papworth they tried to replace the stent on New Year's Day but it slipped again after a few hours. My consultant tried several other types of stent to see if they would hold but due to the position of the collapse nothing worked. The surgeons and consultants decided to make a 3D print of the airway to see if that would give them

any ideas, from this they thought a T-shaped stent would be the answer. So a tube the length of my bronchia with an airway to the right and left at the bottom to hold it in place was designed.

In September 2015 I picked up an infection in my transplanted lung which resulted in my transplanted lung collapsing and leaving only the top lobe working. I was back on oxygen 24/7 and in a wheelchair again. I spoke with my consultant and he pointed out that having another stent now was pointless as the lung was no longer working. They would put a valve in my left lung to release some pressure to make breathing easier. I asked about re transplant which he felt would only be an option if I had the right lung removed as well, also finding a surgeon to do the operation might be difficult. I agreed with him to go ahead with the pre-transplant test and then speak with the surgeons to plead my case. The consultant also advised me that having the valve fitted to reduce the pressure in my left lung could affect my chances of being accepted on the transplant list. I have left the lung hyper-inflated rather than take that chance.

I met with the head surgeon and explained my situation and how I felt. He explained the risks involved and the high risk of bleeding when they took out the stuck down lung. I accepted the risks and asked him to consider me for transplant. About a week later I received the call that two surgeons had agreed to undertake the operation, the head surgeon and one other. I have now been on the transplant list for six months; hopefully the next phone call will be "the one".



Member Stories - Fay Whittaker



I was always a sickly child. I was diagnosed with asthma and had inhalers. I always remember when I was about eleven years old being chased around in the consultant's room because he wanted to take bloods; I was petrified. In 1974 I didn't know what Alpha-1 Antitrypsin Deficiency was.

My mother was always so breathless and physical challenges became very difficult. There was a very large black oxygen cylinder with a tube and mask which my mum was connected to all the time. I had three older siblings who had all flown the nest. My father was a long distance lorry driver and was only home at weekends. He was a hard working man and, in those days, he had to work to put food on the table. I took over the care of my mother by doing the washing, shopping, cleaning and cooking. I was 13 at the time but I didn't feel any remorse, as my mother and I had a great relationship. But my school work suffered as a result of missing so much school.

I married young and went on to have two kids now aged 33 and 31, both are MZ., and alpha-1 carriers My mother passed away at the age of 53 years, that was in 1984. I was devastated. It was just two weeks before my 21st birthday, and I was pregnant with twins, but due to all of the stress I lost one of the twins. It was then that my marriage started to develop problems but I carried on for the sake of my children. Years after this I got divorced and started to get my life back on track only to have it shattered by alpha-1 again.

My brothers were twins; James was found to be PiZZ, and got ill, yet his twin John was fine PiMM and healthy. James was married with no children and he lived a good distance from us, but his marriage also broke down when he was diagnosed with alpha-1. For the next few years I tried to help him not only by getting him rehoused. In between caring for him I went to university and became a fitness instructor, after which I found full time employment. I loved my life. I was still on inhalers at this time and, apart from the odd chest infection, I was pretty good, but then I started to deteriorate catching infection after infection. I then found I couldn't keep up with all the keep fit classes I did everyday. Eventually I had to cut down, then it came to the point I couldn't finish a class. Back and forth to the doctors I would go. During this time I was about to marry my second husband Michael, and everything was great. Then, boom! After collapsing at work I was admitted to hospital and was told that I had alpha-1 antitrypsin deficiency. I learned I am PiZZ, just like my mother and brother . I was 36 years of age. I couldn't tell Michael I felt so low. Eventually I told him to leave. He couldn't understand why, but I did, I knew what my future held, I had seen what both my mother and brother went through and I just couldn't put him through that. But he stuck it out and we were married a year later. That was 16 years ago.

My health started to deteriorate quite quickly. My brother was being assessed for a single lung transplant (which happened in 2002 and he looked fantastic afterwards). I was getting worse, and I retired on medical grounds in 2001. I felt so anxious, lost all my confidence, I started to go into a deep depression, I was 38 years old. My husband was great, he took it on the chin, and he helped me get through this very difficult time.

In the mean time, my brother's health was getting worse and he had lung reduction surgery, but his recovery was not good. At one point we were both admitted to the same ward at the same time because of chest infections. The years passed and we were both getting worse. My husband stopped working to be my full time carer. My brother James contracted pneumonia, and sadly passed away in my arms in 2010 aged 55.

Another low blow, I thought that was it - how the hell was I going to get through this now, no one understands how I feel. My brother and I had that connection, we could talk to each other, we were there for each other. We were alphas.

I got stronger mentally, I wish I could say physically but I knew that would never happen. I went on to take part in two clinical trials of new treatments for alpha-1.

What does my future hold now? Well, in 2014, I became a Committee Member of the Alpha-1 UK Support Group as one of the Fundraiser and Awareness Coordinators. This is a charity I hold close to my heart. I also signed a document twelve years ago to leave my body to science.

I am a fighter, but it's been hard, its hard on all alphas, that is why I try to educate people about Alpha-1 Antitrypsin Deficiency. Amongst other activities I hold an information/charity booth at my local hospital in Kirkcaldy Fife. I hope I can continue to provide as much information and advice to others for a long time yet.

Member Stories - Norah Oliver



My life as an Alpha...part two.

As we have so many new members and as I am, I think, the longest standing member, I have been asked to write the next part of my alpha journey in the hope it may help some of you.

Although I had been diagnosed with asthma when I was 40 my alpha diagnosis wasn't until I was 53 and I can still remember the shock and fear of being told I had this unheard of illness ...so I do understand the fears and concerns you are all going through.

I felt I had been given a death sentence and, after doing some research, I joined this support group. This was the best thing I ever did. Here I found out more about alpha-1 than any doctor could ever tell me. Plus, just as importantly, I found understanding, advice and friendship. These friendships have become a big part of my life.

So, my life to date, as I approach the grand old age of 70! Something I never thought I would. This last year has brought many changes to my life. S adly some of my best alpha friends lost their long fight. Also I have had the most challenging year to my health since my initial diagnosis. Since becoming ill with chest infections and influenza in March, sadly I have lost a fair bit of lung function and I am now receiving palliative care and getting the best possible help to keep me well.

The support I get from my family is wonderful and this is so very important for us alphas. Plus, it's so easy to forget how hard it is for them too.

My husband Bob is my rock and our daughter Lisa and my wonderful sisters are my best friends.

The two grandchildren I have give me so much joy and happiness and being with them gives me reason to carry on and enjoy life.

Yes, life gets hard and being human I get depressed at times. Breathing is difficult and it is so frustrating not being able to do things I used to do. However, I know how lucky I am, so I am trying so hard to be positive and I intend to be around for a few more years yet.

To all new alphas, the best advice I can give as an 'old' one is "be positive and live life well but to the full". A cure WILL come in the not to distant future....

Member Stories - Lee Paterson

Last year Cora Paterson wrote a wonderfully candid article about the challenges and details of being the spouse of someone diagnosed with alpha-1. I am sure we all found Cora's honesty incredibly helpful. I had asked for an update on life in the Paterson household and Lee's condition but their world has changed....

I am sure many of you know by now that at the end of July, after 8 false alarms, Lee was called to Harefield Hospital and received a successful double lung transplant. He was sitting up within 4 days and the physiotherapists were so impressed and optimistic about his progress that they put an exercise bike in his room! Nothing like throwing down the gauntlet.

A successful lung transplant never ceases to amaze. Lee has gone from severe breathlessness and immobility to saying "I feel wonderful", whilst still having drains, cannula, PIC lines, ECG tabs and the rest. We wish Lee a full and speedy recovery and look forward to seeing him at future events.



Member Stories - Anonymous

"A family study is warranted, smoking should be strongly discouraged." These were the words my sister shared with me after her test result for alpha-1 antitrypsin deficiency came back from the lab. She wasn't sure what it all meant but encouraged me to get tested too. We had both stopped smoking years ago but had been passive smokers for most of our youth as our father was a heavy smoker.

After searching the internet and finding out as much information as I could I asked my doctor to test me and my husband. I wondered if this could be an answer to the asthma, bronchitis, chest infections and allergies that had been part of my life and that of my children and grandchildren.

Several weeks later we got the results. I was PiSZ and my husband was PiMM. The same warning was on my results, "Smoking should be strongly discouraged, a family study is warranted, combined alpha-1 antitrypsin deficiency associated with early onset emphysema". I then joined several online support groups.

Armed with information I asked my three adult children if they would be tested. They all said they would get around to it next time they saw the doctor.

The only family member who wanted to be tested at the time was my 17 year old granddaughter Annie who had lots of chest infections. When Annie's results came back she was a PiMZ and had the warning on her result – "*Smoking should be strongly discouraged*".

Annie's mother Kara decided to get herself tested and was confused when her result came back as normal, PiMM. Kara knew that being a PiMM was not possible as she would have inherited one faulty gene from me, either an S or a Z (assuming that my test results had been correct).

She queried the doctors nurse, who had given her the test result, and was told that "she should be happy she didn't have an alpha-1 gene".

Kara asked her doctor to get the lab to check her result again as she strongly suspected a mistake had been made. The lab was adamant this was the correct test result. Kara was asked if there was a possibility she may have been adopted. These suggestions and the uncertainty of not knowing the correct alpha-1 status caused distress to me and my daughter. Although we reside outside of the UK, my husband is a UK citizen. So I asked for help from the Alpha-1 UK Support Group in trying to solve our family problem. I was put into contact with an expert centre that arranged for me, my daughter and granddaughter to be re-tested. In addition, three other family members were also tested.

When the results arrived we found that Kara and Annie are both PiMZ but the results from the other family members were not what we expected.

Our son is PiIS and two others tested are PiMI - the I ("eye") gene must have come from my husband who had previously been informed that he was PiMM.

I asked the lab for information on the "I" gene and learned that it is a rare gene variant which has not been well studied as yet. However, it seems that it is similar to the S gene.

We were all relieved to get my daughter's results clarified and surprised to find that the lab had found the rare "I" gene in our family.

The test result for our son came with the information – "Lung disease risk, if present, is confined to current or ex -smokers. Clinical judgement must apply to the management of their lung disease, if present in PilS heterozygotes". As our son is an ex-smoker with bouts of chest infections, this is timely information.

A Family study is warranted – When a family knows that it is affected by alpha-1, healthy life-style choices can be consciously made, especially in regard to smoking. Future generations can be educated about the disease and its potential impact at an early stage.

Member Interview - Keith White by Andrew Willis

In July 2016, nine months after his successful double lung transplant in Queen Elizabeth Hospital Birmingham, Keith White (KW) was interviewed by Andy Willis (AW) about his alpha-1 journey.

AW: Keith, start by telling us a little bit about yourself. Where you grew up, your family and so on.

KW: I was born in Dagenham in Essex. Sixth of a large family. I was a poorly child, having asthma. I spent much of my early life in hospital or boarding school. In fact, I did not go to a normal school at all – it was all boarding schools from the age of 7. I was first at school in St Catherine's Home in the Isle of Wight, then Meeth in Surrey and my final one for 5 years was Lingfield Hospital School in Surrey because, at the age of 11, I developed epileptic fits and I was getting treated for that. Another school I was going to couldn't look after me with the epilepsy plus the asthma. So I went to an epilepsy school which was Lingfield Hospital School. I left there aged 16.



AW: When you left school you worked where? In Dagenham or on the Isle of Wight or...?

KW: Barking, near Dagenham. I was a cabinet maker's apprentice – that was the theory. But I did not get on with that very well. I did not like the money. Mum was taking too much of it so I left there. I went on to various other jobs: repairing telephone cables, in a factory, making dustbins, all around the Barking area – jobs. Then I went to college in Leatherhead to do woodwork. I finished the course but did not do much work-wise with

woodwork. It was only really when I got on the buses in 1984 that I got a steady job. I was doing that for 20 years until they medically retired me in 2004.

AW: So when did you start getting breathing symptoms?

KW: Well, I had always had asthma and I took up jogging in the 80s. I applied to get into the London Marathon a few times and, eventually, I got in the 1988 marathon and, while training for that, I realized I wasn't recovering very well and realized it wasn't asthma – it was something else. So I went to the doctor's at Northwick Park. He wasn't very good, and he gave me the same spirometry tests and was always just telling me to come back in 6 months. It was only when I went to Central Middlesex, I think it was Dr Mack and he discovered I had alpha-1 antitrypsin deficiency.

AW: So when were you diagnosed, the early 1990s?

KW: 1995.

AW: And when did you have to retire?

KW: 2004.

AW: So you managed to keep working for a good time.

KW: I was having a lot of breathing difficulties. I spent a lot of time off work with infections and various problems I had walking. You might think a bus driver doesn't walk a lot but there was the walking to and from taking over your bus. And I think it might have been my earlier career in bus driving because I don't think the fumes in the garage helped a lot. I was working in the basement of Victoria Garage and if you can imagine 15 to 20 buses and minibuses all starting up at the same time in the morning you could get a lot of fumes and there wasn't much air getting in there.

AW: What about the rest of your family? Were there others that had respiratory issues?

KW: Yes. My sister who was older than me, Doreen. She was not actually diagnosed with AATD but she passed away in 1996 and she had breathing problems for quite a while; but it was never diagnosed officially. I always thought it was AATD after I had been diagnosed. She couldn't walk very far. She was put on the transplant list but unfortunately while she was waiting she passed away. She didn't get the chance that I've got.

Member Interview - Keith White (continued)

AW: So when did you go on oxygen?

KW: It was about 8 or 9 years ago. Just after we moved here so say 9 years on oxygen. I was on 4 litres per minute at rest before the transplant.

AW: Now we are both in the Alpha-1 UK Support Group. When did you join that?

KW: I think I joined it the year after I was diagnosed. About 1996.

AW: So that was in the very early days of the group because it was founded in the mid-90s.

KW: The first meeting we went to was Swindon. It was near the station in the railway museum.

AW: ...and do you remember who was there?

KW: John Doyle was there. A lady called Jill, Ray who still comes with the two boys, John and Joy, Linda Cooke might have been there. And the man in Bristol who died...

AW: Right so you have been involved in it right from the beginning. How did the patient group help you?

KW: Just general support. Talking to other people with the condition. Because even your family, although they are there for you they don't really know what you have gone through and what you are going through at the time with breathing. I don't think anyone can take in – if you can't breath – you've got to have it to appreciate what it's like. I don't think most people understand it, it is hard to explain.

AW: Over the years, did you have a good GP?

KW: I have been pretty lucky with GPs to be honest. Even as a kid I had the same GP when I was at home. I remember Dr Brady and she lived just up the road, or her surgery was just up the road, and she was always very helpful. She would ask after me even when I was away at boarding school.

AW: And when do you first remember transplant being mentioned as an option?

KW: 2006 at the Royal Brompton. I went there for assessment. In fact, I had lung reduction surgery in 2000 at Brompton which wasn't very successful. It is in some cases but it wasn't really effective for me. What annoyed me is that it was not explained to me that if, later on in life, I wanted to think about transplant it might affect that. I wasn't told that at the time. I just assumed it was another surgery that would help me at the time. Then, when it came to wanting a transplant when I went to Harefield in 2007, they refused to do it. One of the reasons was, after the lung reduction surgery, it would be too dangerous. I went to Papworth for a second opinion, more in hope than anything, and they said no as well. So at that stage I just thought I would have to live with it.

AW: Then Birmingham came along.

KW: Well, I was going to Birmingham for the ADAPT research trials they were doing. While I was there we discussed transplantation and I told them I had already been turned down by Harefield and Papworth. They asked me if I was aware that there was a transplant team at Birmingham and I said no I didn't so I thought I would see how I'd get on there. So I went there for assessment as well.



AW: So that was more recently.

KW: Yes that was 2 or 3 years ago. I went in there for 4 days just to have the assessment and a week or so later they informed me that I could go on the list. So that was it. I was well up for that. The way I saw it, it was transplant or nothing. I wanted quality of life. That was the only option open for me. I jumped at the chance.

AW: So when the time came was your first call the call or did you have a couple of fails?

KW: My seventh call was the one that gave me the lungs. I was in various stages of being called. Once I was in hospital with an infection, they wouldn't do it then. A couple of times I had been on my way to Birmingham and they telephoned and said to go home because the

Member Interview - Keith White (continued)

lungs were unsuitable. I'd got as far as having a shower and cleaning up with the gown on – just waiting with the family. Then they came along and cancelled it and didn't give a reason. I didn't let it get me down – I just got on with my life. The last call, the 7^{th,} I was in Watford General with another infection but the Transplant Coordinator said it was such a good match. So he talked with the surgeon, and he talked with the anaesthetist, and they said "let's go for it". So, I was ambulanced from Watford General to Birmingham QE which I can't remember anything about. Two weeks before the operation I can't remember anything.

AW: And how long before you came round and started to become aware that you could breath a lot better?

KW: About 2 weeks?

AW: So you were out of it for a couple of weeks. And then how long before you were out of bed?

KW: There was always a problem getting me out of bed. I love my bed. Even when I had bad lungs – I used to hate the physios coming to drag me out of bed – or even to sit on the side of the bed - it was quite a while – about 3 weeks – getting me to walk short distances up the corridor – I didn't really enjoy it. It had to be done. I had fluids in my chest and I had to go into surgery again to have a drain inserted or two. I had two drains at first. That was in quite a while. They pierced my fatty acid pipe/tube and that was leaking into my chest so my lungs were restricted from expanding properly. They had to drain it out so my lungs could expand. A big problem.

AW: So how long were you in hospital altogether then?

KW: Nearly 2 months.

AW: And when you came out what could you do – having had a lung transplant 2 months before – what was the difference?

KW: The difference was no oxygen, no stairlift. I could walk upstairs unaided without oxygen. I still have problems with my legs aching - various body parts aching. It's only recently that my legs have stopped aching from walking and I'm talking the last couple of weeks. So it took a good 9 months before I could walk without any aches or pains.

AW: So your transplant date was October...

KW: 10th October 2015

AW: So it has taken you 9 months - but you are in pretty good form now and this morning you were out playing golf and very well too. So looking back for you so far it has been pretty successful process.

KW: Yes I won't mention the diabetes or osteoporosis!

AW: You have got other issues but you are living with them.

KW: Yes I would rather be as I am now than how I was before. I would be puffing and panting. No way would I be playing golf. It was just a dream playing golf – and now it is a reality. That's the change.

AW: And you were saying earlier that you have written to your donor family. That's a brave thing to do. You have got your emotions and they have got their emotions – what made you do that?

KW: Well, with the help of Claire, my daughter. She is the one with the English to put it in writing properly. The sentiments are still the same but she put it in a way -1couldn't put it to paper - but what can you say to someone who has given you another chance of life. It's like they have taken a bullet for you. You can't thank them enough can you. So I wanted to put it the best way I could. Their loss is not wasted.

AW: And hopefully at some stage you will get a reply. I am sure they will appreciate being thanked and knowing that you are doing well. Thank you Keith for telling us your story. Have you got anything else about transplant or alpha-1 or the patient group or - come September there is a conference – tell us a little about the alpha-1 patient conferences.

KW: Well it's a time we all get together – people we have met via Facebook or email and we chat and talk about our own problems. Hopefully, I can be a bit of an encouragement to others to go for transplant if they get the chance. Unfortunately for some it does not work out but, for me, it is worth the risk. But that is only a risk you can contemplate for yourself. But there was no question of me not going for it. I wanted the quality of life – not quantity. If the worst happened at least I'd gone for it. That's the way I saw it anyway. I didn't like being a burden...they would tell you different – but everything revolved around me. How long we could go out for. Where we visited – what we did when we were out – I didn't want that – I wanted normal family life.

AW: Well Keith thank you very much for that. It has been very interesting hearing your story.

In Memory of Jane Hill by her husband David



We celebrate the remarkable life of Jane: inspirational, brave, courageous, stoic, strong, happy, funny, laughing, never complaining, always positive, determined, the embodiment of fortitude, an absolute delight to know, beautiful, independent, amazing, kind, caring, delightful, a wonderful example, a lovely patient, a joy to look after, unselfish. The list could and does go on, but she would be so embarrassed if we dwelt on it for too long – she was always so self-effacing.

Jane came into the world and made her presence felt immediately, the doctors had to operate to save her, even having her baptised in theatre without any family present. Things got a little bit quieter after that with the usual childhood scrapes. She had a happy upbringing in York. School proved to be 'a bit of a challenge', she was a bit of a rebel in her teenage years and didn't achieve what could be described as 'her full academic potential'.

Following college Jane went to work with her dad, Peter, who was running the independent family footwear retail business, based in York. It was here that she came into her own, helping him select and buy the ranges of merchandise with a flair for fashion, quality, and style that she maintained throughout her business and personal life. The wardrobes at home are a testament to that! Father and daughter always worked well together and enjoyed each other's company.

It was shoe retailing that brought Jane and I together. I was also running our independent family footwear business started by my great-grandfather. Both the businesses were members of the Independent Footwear Retailers Association, who held an annual conference and we first met at the Windermere 1986 conference.

Roughly six weeks after we first met, I was in York for the weekend – well just a Sunday actually as we both worked Saturday every week – and I asked Jane to marry me! She said yes – thankfully – we got officially engaged on her birthday that year and married the following year.

Jane moved South and started work in our business, initially as a branch manager in our Deal branch and eventually back to Folkestone where she took over the buying duties for the whole business. She maintained the quality of merchandise we were able to offer the consumers and her usual high standards.

She was so, so proud of our two lovely boys. Being a mum came completely naturally to her and she managed to juggle child care, work and all that goes with them. Married life continued with happy times, holidays in York, Scotland, Spain and Switzerland – where we first noticed her shortness of breath and thought it due to the altitude.



Chronic Fatigue Syndrome was the first of a long line of illnesses to afflict the family and Jane in particular. She really was quite poorly for a large part of the last 12 years with a badly infected tonsil and lymph node requiring surgery to remove them, her gall stones and

In Memory of Jane Hill (continued)

subsequent gall bladder removal were probably the most distressing as she was in so much pain with that condition. It was after the gall bladder surgery that it was discovered that she had the Alpha-1 Antitrypsin Deficiency that led eventually to the double lung transplant in 2011.

Most of you are aware of the immense struggle that Jane had following that incredible operation on Palm Sunday 2011. She suffered numerous other issues following the transplant; but we were warned that it wasn't a cure. She spent months in hospital, underwent further procedures and treatments, felt dreadful - but this is where that original list of characteristics I recited at the start really came to the fore – her will to get better and get home was stronger than ever and without that surgery we would have lost her much earlier.

Jane very quietly and gently managed to inspire us all in the family, but also many others. Her bravery and courage in dealing with the numerous hurdles that were placed in front of her, was astounding. She never complained and was grateful, truly and deeply grateful, for every extra day she had been granted.

Life became increasingly difficult with the mobility, eating and eventually breathing difficulties – however, and in true Jane fashion, she took it all in her stride – or perhaps I should say wheelchair tracks – she just got on with things and coped as only she could. She always said she was lucky to be happy in her own company and when I was at work she would cuddle Tilly – who never left her side – and spend countless hours on her beloved cross-stitch, such a talent and the patience to produce such beautiful pieces which are treasured by those who have received them.

We were both so proud of all that the boys, Matthew and Joshua, have achieved under the extreme pressures that such a serious illness in the family inevitably brings. She was so happy to see both the boys with such lovely girlfriends – Katie and Eliza have been simply wonderful in supporting Matt & Josh through all of Jane's trials and we were particularly blessed that they were both able to be with us over Christmas. Thank you to you both - you have been fantastic.

She had many friends that she had made in Harefield Hospital before, during and after her transplant both among fellow patients but also the staff. She always said she felt safe at Harefield when she had to stay. She also had many friends made through the Alpha-1 UK Support Group that gave her huge comfort in the days before, during and after her transplant.

We were fortunate to have had the time to say all the things we needed to say to each other as a family – there are no regrets that we didn't say something to each other or tell everyone that we loved them. We had a wonderful Christmas Day and she left us peacefully and serenely on her own terms, as she had always wanted to do. She was so very tired and ready to go, she needed the rest of eternal peace.



In memory of Paul Hodson by his wife Lyt

I first met Paul at Secondary School when we were 11. We were in the same class but did not share the same bunch of friends and we only spoke in passing.

A few years later, two marriages under my belt and 3 children, my boys decided to put me on Facebook.

One of the first people to say "Hi" to me was Kim (Brumpton). She was in the year below me at school, but I remembered her well and we started chatting and meeting up when she was back in Lincolnshire.

One day she said she was going to get Paul, her brother, on Facebook. "You remember him don't you?" she said. Of course I did and I said, "Yes, get him on here, I'll chat to him".



The rest, you could say, was history. BUT I did know that Paul had Alpha-1 Antitrypsin Deficiency, and I knew what the effects were because Kim had told me. Did I have any doubts when I met Paul? I can truthfully say, at the time, I had none. I was so happy with Paul, he quickly became my world.

We went on our first date on 11th July 2010 and we were engaged on the 10th October the same year. Paul was insistent that we should be married exactly a year to the day of our first date, which we were. Not bad going for a man that had avoided marriage up to then. My boys and my grandsons thought the world of Paul. He was intelligent and kind and would do anything for them. Not always happily, he could be quite grumpy sometimes, but he still did it. He never said "your children/grandchildren", it was always "our".

Paul would struggle quite a lot sometimes with his breathing and could spend countless days in bed but, after a few rounds of antibiotics and steroids, he would be up and about again.

To say he was a hoarder was an understatement. I went from a clean and tidy house and garden to something resembling Steptoe and Son, inside and outside. He loved to spend his time tinkering away mending Dysons, washing machines, lawn mowers, strimmers. You name it, I had it in my house, yep, my house. But he wanted to feel useful. He hated the fact that he couldn't work, it affected his pride and dignity and he hated anyone thinking he was weak.

One of my main bugbears was that he carried on smoking. He insisted it opened up his airways and he felt that he didn't have any other vices (hoarding not counted of course!) and so he continued smoking.

Last year in 2015, around about our fourth wedding anniversary, Paul began to feel unwell. We, as a family thought, "Oh, here we go again". He had at least four lots of antibiotics and steroids over the following weeks and he never seemed to pick up. On 11th September he rallied himself round and we went to the Alpha-1 UK Support Groups annual meeting in Lincoln which wasn't far from us. We spent the day with our lovely Alpha family and chatted for quite a while afterwards. In the evening we went to a fake music festival because Paul absolutely loved music. He was a whizz at quizzes.

On the Sunday, he stayed in bed. On Monday he stayed in bed. On Tuesday he stayed in bed. Only getting up for drinks and toileting but we still chatted as normal. On Tuesday evening I asked him if it was time for the ambulance and he insisted that it wasn't.

We woke up on Wednesday morning and he asked me to phone for the ambulance. Thank goodness I thought, "now they can sort him out".

I went to work because it is a regular thing for Paul to go into hospital and come out again three or four days later. I had a telephone call around 3 o'clock in the afternoon to say Paul was quite poorly and to come in. I headed for the hospital and Paul was in ICU. They told me they were monitoring him closely, he was quite

In memory of Paul Hodson (continued)

poorly but he had improved a little over the last couple of hours.

We were chatting and filling in his form for what his meals would be the following day. I told him about my day and he said he was starting to feel better. He was on his nebuliser and they kept taking his bloods. Suddenly, he became quite disorientated and said he felt light-headed and weak. They laid him down, took some blood and he nodded off.

A few minutes later the doctor came and had a word with me. They were really sorry but there was nothing they could do. I was so shocked and I thought, "no, Paul always rallies round and I know he'll be home in a couple of days".

Life is short, live it. Look after each other, love each other and never, ever, go to sleep without saying, "I love you".

The hospital staff were so lovely. They brought a bed in so that I could lie next to Paul, they pulled the curtains around and monitored him from the work station to give us some privacy. Before he went to sleep, Paul said "I love you", and I am so pleased he did because they were the last words he ever spoke. I lay with my darling man and told him all the things that I needed to tell him. I'm not ashamed to say I begged him to fight and not let go, but I don't think he could hear me. At 10 o'clock that evening he gave a big sigh and that was it, my darling's last breath. We found out that Paul had left his illness for so long, and just kept self-medicating without seeking medical advice or sputum testing, that he had developed pneumonia and septicaemia had set in.

The days and weeks after are a haze. I felt like my heart was broken and nothing would mend it. My lovely boys kept me going, Chris came up from Surrey and organised and sorted everything. Steve lived with us and he kept me going each day. My youngest son Nick hubby down and his Chris came from Scarborough. Family and friends rallied round, for that I was so very grateful. The boys, with tears streaming down their faces, were pallbearers for their "Pops". Grandson Sam had tears streaming when he placed an orange flower on his "Gramp's" coffin.

How am I now? Meeting milestones. The first of everything is extremely hard. Managing, because I have my family to think about. You carry on, you go to work, you laugh with people and it looks like things are fine and they are to a certain extent. In reality there is a BIG hole in my life. My heart is broken. My soul-mate, best friend, confidante and rock is no longer here. I don't cry all the time now, but when I do, I do it alone. My grief is personal and it keeps me sane to let it out, but I don't need to bring anyone else down with it.

So my lovely friends I would like to say to you. "Life is short, live it. Look after each other, love each other and never, ever, go to sleep without saying, "I love you".

... and a tribute to Paul Hodson by his sister Kim Brumpton

Paul was the head of our family. He set an example to his siblings of how to be respectful, how to treat people fairly, he encouraged you to strive and achieve your goals and he never demoralised anyone, he never lost his temper even when life situations and illness got to him, he was a true gentleman. He was my protector, my mentor, my best friend and the best brother anyone could wish for. He was a very loving, caring and intelligent man and always strove to help anyone less fortunate than himself. He had a natural ability when it came to repairing anything electrical or mechanical. He was a problem solver and had great leadership qualities, to the extent he progressed from Trainee to Area Manager in a short time while combatting alpha 1, asthma, eczema and hay fever. He loved motorbikes, puzzles, reading, music and karting and was always game to try anything new like flying and gliding which he did in his teenage years. He was the rope that bound this family together and we are at loss without him.

In memory of Linda Cooke by her close friend Bev Burroughs



30th October 1950 – 21st January 2016

R.I.P. Linda Cooke You can now breathe easy

Linda peacefully passed away in hospital surrounded by her family and friend on 21 January after a long and courageously fought battle with Alpha-1 and subsequent rejection following a double lung transplant in 2009.

Linda was always there, in person, on the end of the phone, by email and Facebook, to offer support and friendship to all who came within her sphere, no matter what walk of life they were from, and no matter how ill she herself was feeling....

Linda was always one of the first to welcome new members, who often had just been diagnosed, and had found the Alpha-1 UK Support Group whilst researching Alpha-1 on the internet. She would offer words of comfort, support and encouragement and somehow she always knew what to say to alleviate their fears of this little known illness. When a member was listed for a transplant, she would offer them love and support, and when they were lucky enough to get "the call" she was also there to support their family through this difficult time.

But it wasn't just within the Alpha-1 community that she was like this. To all her family and friends, she was a tower of strength, and was always there for them. Your problem was her problem, she would laugh with you, cry with you, celebrate your successes and give support in times of need. But for all this the centre of her world was her son and daughter, Andrew and Helen. Her world revolved around them and I know that she was happy in the knowledge that when the time came for her to leave this life, they would have Louise and Michael there to support them as well as family and friends.

As her daughter Helen said at her service:

"She always said to us how lucky she was to have us as her kids but, in actual fact, it was us that were the lucky ones to have her. In the last few days of her life mum said she had been blessed to have such a loving family and such caring friends. Sadly, I'm not sure mum ever really realised how special she was. She was always so surprised and humbled by the well wishes and messages of love she received off others, especially when she was poorly, but looking round the congregation, at how sad people are that you're no longer with us mum, I hope you see just how much of a difference you made to all of our lives. You were truly an angel on this earth and taken far too soon."

Linda is now at peace, up there in Heaven, in the loving arms of her Mum and Dad, and chattering away ten to the dozen with Jane Hill, Chris Torrance, John Doyle and all the Alphas that have been taken too soon.

Linda posted this picture on her Facebook page in March 2015. She is second from left and Bev in the middle. She wrote: "Blast from the past....this is Bev and I and some girls from work helping out at the wedding of a work colleague ...This was taken about 20 years ago...Bev and I have been friends for almost 30 years ...thank you Bev Burroughs for your love, loyalty and friendship....you are the dearest of souls xxx"



Poetry Please by Joe Lyons



HOPE

Hope's word of four letters that can mean a lot Especially if that's all you've got Fate always changing day by day Surmounting difficulties come what may If lucks behind you and why shouldn't it be Tomorrow will bring happiness for all to see

UNTIL THE SUN APPEARS

Standing on a rock watching below Waves crashing, breaking as they go A bird swoops so fast only to see It's breakfast escaping in the sea Boats buffeted waves so rough With hulls of steel or fibreglass tough The wind it howls into the night Until the sun appears to show it's alright.

Poetry Please (continued)

THE GROUP

It hardly seems anytime since I became aware Of a group of people with a friendship that's so rare Friends who understand the problems we with this illness can befall How we deal with life making some problems seem nothing at all

Each day that passes by enjoying laughter sometimes pain We read with real interest how life's treated all of us again Welcoming new people with compassion and kindness Doesn't matter who you are, we treat all with open mindness

New friends and old meet once a year in different places Doesn't matter how you get there we still have a few spaces Friend or stranger the welcome always will be with smiles If you've travelled from afar or only a few miles Join and help us enjoy the life we live All we ask is for the many smiles and laughter you can give

Alpha-1 Quiz

The answers to the following questions are all words in the Wordsearch on the next page. Knowing some of the main words used by the medics can help our understanding and help make our appointments more thorough, both for us and for clinicians. The answers can be found on page 36.

- 1. The East Anglian hospital where the NHS alpha-1 clinic is based (12)
- 2. The part of the lung where oxygen and carbon dioxide exchange takes place (6)
- 3. An American medical research company particularly involved in AATD liver issues (9)
- 4. Another word for replacement therapy (12)
- 5. Inhaling this opens up the airways; salbutamol is an example (14)
- 6. Scarring of the liver due to continuous long term liver damage (9)
- 7. The initials for a generic name for many lung diseases; far too general for our liking. (4)
- 8. Dr Parr is the alpha-1 consultant at this specialist alpha-1 NHS clinic (8)
- 9. They produce Respreeza—an inhaled replacement therapy. (3-7)
- 10. It makes us what we are and backwards (3)
- 11. AATD leads to the destruction of lung tissue by this. (anagram of 'east seal') (8)
- 12. The European Medicines Agency (3)
- 13. Forced Expiratory Volume (3)
- 14. A medical technology for fixing a condition by modifying human DNA (11)
- 15. A plasma products company beginning with 'G' they produce Prolastin (7)
- 16. When cells contain two different alleles of a gene. For AATD this could be MZ (12)
- 17. AATD principally affects these two organs (these words appear seperately, 5 and 4)
- 18. They approve medicines in the UK (4)
- 19. Ply more (anag) a large molecule formed by linking many smaller molecules (7)
- 20. The replacement therapy provided by Grifols (9)
- 21. The hospital at which the Birmingham NHS alpha-1 clinic is based (2)
- 22. The condition where acid from the stomach leaks up into the gullet (6)
- 23. The north London hospital where the NHS alpha-1 clinic is based (5-4)
- 24. Prednisolone is one of these types of medicine (7)
- 25. Initials for (t)ransfer factor for the (l)ung for (c)arbon m(o)noxide (4)
- 26. The invaders after whom alpha-1 is sometimes known (6)

Alpha-1 Wordsearch

Find the answers to the questions of the quiz on page 29.

													N. 1999		
Ρ	R	D	Е	Ν	Y	D	0	С	U	Μ	G	А	Ρ	С	G
Ρ	Е	L	А	S	т	А	S	Е	I.	F	N	н	R	0	н
Ο	F	W	U	А	R	R	G	М	G	R	I.	F	О	L	S
L	L	N	G	U	Ο	R	V	А	S	R	R	V	L	н	E
Y	U	С	М	В	С	0	Ρ	D	С	т	н	Ρ	А	G	К
М	х	Q	E	G	L	W	Q	Ν	T	С	E	G	S	E	0
E	R	А	N	N	т	н	N	А	R	н	В	Ν	т	N	0
R	Е	D	т	I	F	E	V	N	R	С	L	U	I	E	R
S	т	U	А	к	0	А	А	S	н	D	S	L	N	т	В
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н	Е	т	Е	R	0	Z	Y	G	0	т	Е	R	А	Y	K

Fundraising and Awareness

A Big Thank You to everyone involved in fundraising activities and for donations to the group. This year supporters have been active as ever - jumped out of airplanes, zipwired at 100mph, organized quizzes and musical events, organized sales and auctions and taken part in sporting events and competitions.

Through your activities we are able to continue funding our programmes of providing support and education for patients, families, carers and friends who are affected directly or indirectly by Alpha-1 Antitrypsin Deficiency. We aim to do this by

- · growing a social network for patients
- providing discussion groups focusing on how better to cope with their condition aiming towards improving quality of life
- advancing education, understanding and awareness of the condition, in particular among medical professionals, including information relating to genetic implications, treatment, and lifestyle choices.
- supporting research and campaigning for better access to treatment for Alpha-1 patients.

We know there is so much more we can and need to do to promote better knowledge and understanding of Alpha-1 Antitrypsin Deficiency but we are limited by the funds we receive, so your support is valued and very much appreciated.



Helen Brackley from Sussex did the Voodoo Zipline in Vegas. She says: "It was terrifying, but worth every moment, knowing the money I raised was going to a cause that a very special woman I know suffers with. I'm going back to Vegas February 2017 and plan to raise more by doing something else terrifying at the Stratosphere called the Sky Jump. Terrified, but I know it all goes to a fantastic cause, and if the money I help raise goes to helping solve this disease, I'll do whatever it takes."

Wendy Howlett wrote telling about the fundraising Observation quiz trail they did at a caravan rally in Wiltshire:

"My husband John and I ran a caravan rally in a primary school in Corsham, Wiltshire, to coincide with the Centre's Observation competition. The questions are set out by our Competitions officer, we have to walk round the local town or village and try to decipher the clues that have been set.



We ran the usual raffle on the rally and a lottery bonus ball draw. As a lot of people have never heard of Alpha-1 Antitrypsin Deficiency I thought that it would be a good way of bringing this condition to the notice of others.

I chose the Alpha-1 charity as my sister was diagnosed January/February 2015 and was not well enough to be given a liver transplant (although she was sent to Kings College Hospital in London and it was here that the hospital decided that her heart would not stand the operation). She eventually passed away in November 2015."

All my family have now been tested and at least one sister has the ZZ gene, three other siblings have the MZ gene and my self has the PiZ gene.

Fundraising and Awareness (continued)



06/06/2015 - Marjorie Hayward (grandmother of Jensen Kay) ran a Jamaican Reggae Day Fundraiser at Bolton Lever-Hulme Rotary



25/06/2016 - Sandy Brown Afternoon Tea Friends and Family in memory of her sister Beryl Saunders Southampton



1-3/07/2016 , Sandy Brown and sister Fleet Carnival Soft Toy Stall Southampton



11/07/2015 - Fay Whittaker Car Boot Sale Fife



16/10/2015 - Janiney Sporiney Continuous Crafting



18/09/2015 - Karen Skalvoll Oslo Marathon 3k and 10k Alpha Warriors





Chris Underwood

In memory of his alpha stepdad Paul Hodson aged 55 who sadly passed away on the 16th Sept 2015

15/05/2016 - Skydiving from 13,000ft in Brackley 10/04/2016 - Zipline -1 mile, 500ft high, 100mph in Wales 16/04/2016 - Abseiling down the Spinnaker Tower in Portsmouth

THANK YOU FOR THE MUSIC

A MUSICAL REVUE AND BUFFET SUPPER AT 7.30PM DUNSCAR GOLF CLUB

BL7 9QY

In aid of Alpha1, Diabetes and Rotary charities

3RD JUNE 2016 7.30PM

TICKETS:- £12.50p

SHINE CHILDREN'S CHOIR, AKT, SURESH AND MAINBASE

> 23/06/2015 - Ruth Kay Rotary Club Show Bolton

Fundraising and Awareness (continued)



13/03/2016 - Tanya Jones Surrey Half Marathon 2016

"Well that's it ... Cried from about mile 10 as I was thinking about Alpha-1 and how crap it is ...any way, I beat you today Alpha-1 at 2 hours 10 mins at the half marathon... Plus my company will match all I raised .."



Mel Brolly continues to run a fundraising Facebook page. Check it out at Auctions for Alpha-1

Donations

Diane & Mick Stobart

Monthly donations

Donations in Memory	
Ruth Champion	In memory of husband Brian Champion
Sally Watt	In memory of Paul Hodson
Lyt Hodson	In memory of Paul Hodson
Ken Macfarlane	In memory of June Macfarlane
Karen Adams/Andrea Ellis	In memory of mum Beryl
Brian Saunders	In memory of Beryl Saunders

Raising Funds - How You Can Help

Perhaps you could help raise funds to enable us to continue our work? Whether £5 or £500, all donations will be put to good use, providing information, equipment and support for all Alpha-1 patients.

In addition we aim to promote better awareness and understanding of the condition throughout the medical profession, support research and campaign for better services and treatment for Alpha-1 patients in the UK. **Please visit our Website for more information: www.alpha1.org.uk**

JustGiving[®]

You will raise more for Alpha-1 UK Support Group on **JustGiving**. It's easy (and completely free) to set up a fundraising page for your favourite charity. It only takes 60 seconds to get up and running.

You can write out your personal fundraising story, add photos and even video and colour to your page. Best of all, it's all incredibly simple to do giving you the best tools to make it easy to ask friends to sponsor you.

If you are a UK tax payer our charity can also claim back via Gift Aid the basic rate tax already paid on donations by the donor. This means we can claim back from the government on your behalf 25p for every £1 donated, boosting the value of the donation by a quarter.

You can also use your mobile to send a donation. Text "ALPH10 £amount to donate" to 70070 to donate to Alpha-1 UK Support Group.

JustTextGiving is powered by Vodafone.





easyfundraising.org.uk is a great way to raise money for our charity just by shopping online.

1. Start at easyfundraising

Let's say you want to buy a pair of shoes from John Lewis.

Instead of going to johnlewis.com directly, you first go to easyfund-raising.org.uk.

2. Make a purchase

From the easyfundraising website, click through to John Lewis to make your purchase. This tells John Lewis you came from easyfundraising. The price of the shoes is exactly the same.

3. Get a donation

After you buy your shoes, John Lewis will give you a cash

reward that you can turn into a donation for your good cause. easyfundraising collect this and send it on at no extra cost.

4. Get the easyfundraising Donation Reminder

You can skip steps 1 and 2 with the easyfundraising Donation Reminder. Just click the Reminder when you shop to receive any eligible donations. You'll never forget a free donation again!



easysearch.org.uk is a free search engine that enables you to raise funds for the good cause of your choice whenever you search the Web. It costs nothing - easysearch is completely free.

How does it work?

If easysearch is used as the search engine for a web search, easysearch will donate half a penny to the Alpha-1 UK Support Group for every search you make This is an easy way to raise money, so please use easysearch.



Alpha-1 UK Support Group Merchandise

On our website we have a selection of Alpha-1 merchandise available for purchase, including T-Shirts, wristbands, trolley key rings, badges and christmas cards.

Alpha-1 information packs, booklets and posters are also available at no cost. Please e-mail us with your full name and address at:

Info@alpha1.org.uk

We are happy to supply our materials to healthcare professionals.





CHRISTMAS CARDS for 2016

Here is our 2016 selection of Christmas Cards. Please order either in our online Shop: alpha1.org.uk or by email: info@alpha1.org.uk The cost is £3.75 for a pack of 10 (140mm/5" square).











Snow Santa

Gold Tree Twelve Days of Christmas

Santa's Reindeer

Star Christmas Wishes

Answers to Quiz on page 29

- 1. Addenbrookes
- 2. Airsac
- 3. Arrowhead
- 4. Augmentation
- 5. Bronchodilator
- 6. Cirrhosis
- 7. COPD
- 8. Coventry
- 9. CSL Behring
- 10. DNA
- 11. Elastase
- 12. EMA
- 13. FEV
- 14. Gene Therapy
- 15. Grifols
- 16. Heterozygote
- 17. Liver & Lung
- 18. NICE
- 19. Polymer
- 20. Prolastin
- 21. QE
- 22. Reflux
- 23. Royal Free
- 24. Steroid
- 25. TLCO
- 26. Viking

Trustees & Committee



Karen O'Hara Chairman Trustee, Treasurer



John Mugford Vice Chairman, Trustee



Bev Burroughs *Trustee,* Admin Support



Jemma Coad Trustee, Fundraising Awareness Co-ordinator Parent Support



Dr Sandra Nestler-Parr *Trustee, Strategy and External Relations*



ane Purves *Trustee, Lead for Scotland*



Mel Brolly Fundraising Awareness Co-ordinator



Graham Dalton Scottish Representative



s Fay Whittaker rt Scottish Representative







Professor Robert A Stockley

Professor of Medicine at University Hospital Birmingham, Director of Lung Immunobiochemical Research Laboratory



Professor William MacNee

Professor of Respiratory and Environmental Medicine at University of Edinburgh, Honorary Consultant Physician at Lothian Health

Supporters



In Memorium

All Alpha friends that we have lost have left their mark on our lives, and it was a privilege to have known them.

Reflections by Joe Lyons

For the people who've gone before us, your fight was not in vain Our thoughts and prayers are with you, we tried to ease your pain We know you were the bravest, the best that you could be And even then you smoothed a path for someone just like me

In life we all need heroes who would fight and be strong You are all classed amongst them, even though you've gone In our thoughts you're always there, we'd think of what you'd do Even when life is a struggle we strive to be as good as you

Life takes so many heroes before we can get it right Our thoughts are always with them throughout the day and night So pause just for a moment let your mind free to take stock Be thankful in that moment remembering what you've got



Notes



Supporting alphas, their families, carers and friends since 1997

Alpha-1 UK Support Group 50 Wenning Lane Emerson Valley Milton Keynes MK4 2JF

We are a registered Charity

England and Wales (1146330)

Scotland (SC043177)

www.alpha1.org.uk

info@alpha1.org.uk



alpha1uksupportgroup

Who are we?

The Alpha-1 UK Support Group is a not for profit organisation and registered charity founded in 1997 by those diagnosed with the genetic condition Alpha-1 Antitrypsin Deficiency who are dedicated to help, advise and support fellow sufferers, their families, carers and friends.

Mission Statement

- To provide support and education for patients, families, carers and friends who are affected directly or indirectly by Alpha-1 Antitrypsin Deficiency.
- To grow a social network for patients, by providing discussion groups focusing on how better to cope with their condition, aiming towards improving quality of life.
- To advance education, understanding and awareness of the condition, in particular among medical professionals, including information relating to genetic implications, treatment, and lifestyle choices.
- To support research and campaign for better access to treatment for Alpha-1 patients.

What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin Deficiency also known as Alpha-1, A1AD or AATD is an inherited, genetic condition that is passed on from generation to generation. As the name suggests it is a deficiency of Alpha-1 antitrypsin (AAT) in the bloodstream. AAT is an enzyme produced in the liver to help protect the tissues of the body during infections. The low level of AAT in the blood occurs because the AAT is abnormal and cannot be released from the liver at the normal rate. This leads to a build up of abnormal AAT in the liver that can cause liver disease and a decrease of AAT in the blood can lead to lung disease.



alpha1uk

