Alpha-1 UK Support Group
Newsletter

Issue 14
Autumn 2015

Welcome

To our Autumn 2015 Newsletter
A BIG thank you to all our members for your 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

Inside This Issue

West Midlands NHS Alpha-1 Antitrypsin Deficiency Service provided specialist clinics by multi-disciplinary expert team

by Dr David Parr, University Hospitals Coventry and Warwickshire

Most of you will be aware that the Alpha-1 Alliance conducted a national survey of Alpha-1 patients and their families in 2012 which sought to identify the unmet need that patients currently experience in their day-to-day care in the UK. The result of the survey provided conclusive evidence that many of you experience difficulties in getting access to both clinical experts with detailed knowledge and experience in Alpha-1 and comprehensive NHS services that optimally manage your condition. The survey also identified that patients feel they would significantly benefit if they were seen at centres which were able to provide expertise in all aspects of Alpha-1: in other words, you wanted to have access to specialists in lung disease, liver disease and the skin complications of Alpha-1, as well as being able to obtain advice on the genetic aspects of the condition.

Following on from this survey, came the Alpha-1 Alliance campaign to raise awareness of Alpha-1 in the English and Scottish Parliaments and to provide the Department of Health with a case for setting up nationally commissioned specialist Alpha-1 services. As is often the case when politicians are involved, the response was not a cause for celebration – earlier this year we were informed that the officials took the view they would not support national Alpha-1 services until a specific therapy for Alpha-1 (such as augmentation therapy) is available! Hopefully, this will soon be the case (for more details, see page 5) at which point the Alpha-1 Alliance can re-apply for the establishment of nationally funded specialist Alpha-1 services.

However, undeterred, it was decided that steps should be taken to provide patients with the kind of service that they had said they wanted. Consequently, in the West Midlands, specialist multi disciplinary clinics (with lung, liver and skin specialists) have recently been established which are currently running in Coventry at the University Hospitals Coventry and Warwickshire. Queen Elizabeth Hospital Birmingham is also planning to run these NHS clinics in due course (see pages 2 and 3 for more details on these clinics in both centres). The West Midland NHS Alpha-1 Service will provide everything the Alpha-1 Alliance has been calling for, except that the funding for these clinics is not provided by the NHS from a national but a local budget.

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You may wonder what services these clinics provide, what value they can add to your care, how they differ from the care you are currently receiving in the NHS, or what the differences are between being seen at NHS clinics and being seen at an Alpha-1 research programme (such as ADAPT in Birmingham). For that to become clear, it is important to first look at the status of Alpha-1 care that has been available in the UK so far.

Alpha-1 Alliance meets Health Secretary to discuss national specialist NHS Alpha-1 services (from left to right: Dr David Parr, Karen North, Health Secretary Jeremy Hunt, Margaret Millar, Mark Pawsey MP)

Patients with Alpha-1 have always been able to obtain treatment within the NHS by GPs, Consultants and other health care professionals. However, the main reason for the significant differences in the level of care and management provided to patients locally by the NHS is that Alpha-1 is a rare disease and, consequently, not many doctors have been able to gain a lot of knowledge or experience with the condition. Health care workers do the best for patients they can under the circumstances but, like with many other rare diseases, optimal care for Alpha-1 is best provided in specialist NHS centres by a team of multi disciplinary experts with the necessary experience that only comes from frequent encounters with affected patients.

It has been possible for some patients to attend centres at which Alpha-1 research is undertaken, such as Queen Elizabeth Hospital in Birmingham, where patients have the opportunity to enrol in the ADAPT research programme. Research programmes and patient registries like ADAPT have not only significantly contributed to the progress made in understanding Alpha-1 and developing novel therapies for different aspects of the disease, they have also provided advice, support as well as some practical help in the delivery of care at patients' local hospitals.

Although it is important for the development of novel therapies that patients continue to participate in clinical research, there are many reasons why attending a research programme is not a substitute for clinical management of Alpha-1 patients in NHS-funded and NHS-run specialist clinics, and this is why the West Midlands NHS Alpha-1 clinics have been established.

Some of the differences and advantages that the West Midlands NHS Alpha-1 Service provides to Alpha-1 patients, compared to being seen only by a local doctor and/or in research programmes, are:

- Patients have access to teams of specialists with expertise in different clinical areas of Alpha-1 (lung, liver, skin, joint, genetics, children etc.).
- NHS specialists can prescribe treatments and give advice to non-specialists on prescribing treatments through the NHS.
- The NHS provides consistent funding for these clinics, based on clinical needs.
- Patients can be referred to the West Midlands NHS Alpha-1 clinics by their GP or local specialist from all over England (for more detail, see below).
- All doctors and other health care workers across the NHS have access to the patient's clinical information, test results, medication etc.
- Patients can obtain specialist care from a team that knows them, 'around the clock'.
- There is continuity of care across all NHS services that the patient requires.
- The NHS 'Advice and Guidance Service' allows GPs to directly contact Alpha-1 specialists from the West Midland service or anywhere else in the country in order to obtain help with managing their patients (thereby obtaining prompt advice that could potentially avoid unnecessary hospital attendance).
- The NHS keeps comprehensive medical records for the purpose of day-to-day clinical management (as opposed to research programmes that collect clinical data primarily for the purpose of answering specific research questions).

As mentioned above, the West Midlands Alpha-1 NHS Service is currently providing specialist multi-disciplinary clinics in Coventry at the University Hospitals Coventry and Warwickshire (UHCW). The team consists of David Parr (consultant chest physician), Esther Unit (consultant liver physician), Andy Ilchyshyn (consultant skin physician) and Dimitri Grammatopoulos (Clinical Biochemist) as well as respiratory nurses (Sue Townsend and Charlotte Beddow), a physiotherapist (Morag Clarke) and a team of respiratory physiologists, headed by Joanna Shakespeare. The regular involvement of a Dietician and Psychologist are also planned. All members of the team are experienced in Alpha-1 – for example, David Parr was a member of the ADAPT research team for several years, is an internationally recognised expert in the use of CT in clinical trials for Alpha-1 and has extensively published and advised pharmaceutical companies as well as regulatory agencies in this field,

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and he is a strong supporter of the Alpha-1 Alliance's campaign; Joanna Shakespeare has worked at ADAPT in Birmingham for many years before joining the Coventry team.

The team at UHCW will be joined in October by an additional Consultant Chest Physician, Dr Beatriz Lara, who has specialised in Alpha-1 for many years. Beatriz previously worked in Spain where she was in charge of the Spanish Alpha-1 registry. She has also extensively published in the field of Alpha-1. Beatriz will be a particularly important addition to the UHCW team, as she is the only chest physician in the UK who has extensive experience of treating patients with augmentation therapy outside the setting of a clinical trial, as this treatment has been routinely available in Spain for many years. The clinics at UHCW have been set up to provide patients with enough time for a thorough clinical assessment and the opportunity to discuss their condition, treatment options and other issues with the specialist team. Our promise at UHCW is that every patient will be seen by consultant physicians only, rather than by junior doctors.

A similarly run specialist NHS clinic will also be established in Birmingham at the University Hospital (UHB) although, at the time of going to press, the final details of this service have yet to be agreed.

Patients from across England who would like to attend the specialist Alpha-1 clinics at UHCW in Coventry can do so by asking their GPs for a referral which can be made through the NHS 'Choose and Book' system (which is not a favourite amongst many GPs, since it is quite a laborious system, but this is planned to be the only method for all hospital referrals in future). Alternatively, they can ask their GP to make a referral in writing to a named consultant at UHCW or UHB, since this traditional route of referral is still allowed

Some of the team members running the specialist NHS Alpha-1 clinics in Coventry (from left to right: Dr David Parr, Charlotte Beddow, Dr Esther Unitt, Sue Townsend and Dr Andy Ilchyshyn).

(details of the UHCW Alpha-1 service can be found on the UHCW website link: (http://www.uhcw.nhs.uk/our-services/a-z-of-services?sID=124)

– currently, Alpha-1 patients are being seen in a clinic entitled Complex Airways Disease Clinic, which is held on Wednesday mornings).

Despite attempts to avoid a muddle, there appears to be continued confusion about what these developments mean for patients who have either previously been involved in research or would like to become involved in the future. There is a clear distinction between these specialist NHS clinics and research activity. In summary, patients who attend the NHS clinics will have their clinical care managed by specialists who will obtain the necessary tests and investigations required for accurate assessment and who will be able to prescribe treatment or give treatment advice to patients' local doctors (i.e. in the same way that all specialist NHS clinics run). In addition, all NHS patients will be offered the opportunity to participate in research which may include giving consent for their NHS clinical investigations to be used for research purposes (this too is in keeping with what is now expected by the Department of Health of all NHS services - it will also avoid duplication and wasted time); the decision on whether or not to participate in research will have no influence on the care provided in the NHS clinics and will, of course, be optional and without any prejudice to the delivery of NHS care. However, patients attending for research purposes alone should not see their participation in research as a substitute for specialist clinical care, since treatment decisions relating to routine clinical care cannot be undertaken as part of research activity. Patients who have attended the research programme in Birmingham (ADAPT) should also be aware that, whilst the programme will continue, there will need to be a greater focus on specific research questions, based on more targeted funding. This means that routine follow up of all patients may not be possible and the ADAPT office will be sending out an explanatory letter in due course. The only blurring of the distinction between the NHS clinics and research activity may be that the same specialists providing NHS care may also be undertaking some of the research activity and that some of the clinical tests obtained in order to guide treatment on the NHS may be used for research purposes, provided that the patient has given their consent for this to be done.

The provision of the West Midlands NHS Alpha-1 clinics in the way outlined above will mean that Alpha-1 patients will, for the first time, be able to obtain specialist care from multi-disciplinary teams of experts which have the ability to manage all aspects of the condition. In addition, these NHS clinics will provide a link into research for those patients wishing to participate.

Arrowhead develops novel gene-silencing therapy for liver disease in Alpha-1 – Phase I clinical trial underway *by Dr Sandra Nestler-Parr, Trustee*

Alpha-1 Antitrypsin Deficiency (Alpha-1) got its name when it was learned that reduced blood levels of alpha-1 antitrypsin (AAT) were associated with premature lung disease. It was soon understood that the vast majority of patients who have Alpha-1 (Alphas) produce normal amounts of AAT but the protein fails to fold properly and only a fraction of the AAT is able to escape the liver (where most alpha-1 antitrypsin is produced) into the blood stream. This leads to a build-up of the protein in the liver that can lead to serious problems.

Liver disease caused by Alpha-1 can be apparent from birth or may become apparent in toddlers. In rare cases this type of liver disease requires liver transplant to prevent death. In adults, Alpha-1 is more commonly diagnosed when a patient presents with lung disease. While there are some reports of adults with Alpha-1 receiving liver transplants or dying from liver disease, the risk of liver disease in adults is not very well understood. Many in the medical community believe that adult liver disease in the Alpha-1 population is underdiagnosed. Interest in liver complications from Alpha-1 is growing because of improving medical care for lung disease and the development of new potential treatments for the liver.

Research is advancing to more fully understand how to reduce the amount of abnormal protein the liver produces. In animal models of Alpha-1, reducing liver production of the abnormal AAT protein allows for reversal of disease changes in the liver (but this doesn't necessarily address any Alpha-1 related lung problems). Therapies intended to reduce abnormal AAT protein in humans are now being developed by several companies and are beginning to be tested in humans. In some of these initial investigational studies Alphas who participate do not have to be showing signs of liver disease. If shown to be safe in humans, long-term studies in adults will determine in due course if these therapies halt or reverse the liver disease caused by Alpha-1. Once that is shown in adults, it is hoped that studies in children can begin.

Alpha-1 UK Support Group is now working to get the word out about the often hidden liver disease in adult Alphas and about the opportunity to participate in clinical studies of new therapies. To that end, we have recently met with Arrowhead Research Corporation, a biotech company based in the US that is developing a potential new treatment for liver disease associated with Alpha-1.

Arrowhead announced in July that they received regulatory permission in the United Kingdom to proceed with Part B of their Phase 1 study of ARC-AAT, their innovative RNAi-based drug candidate for the treatment of Alpha-1 liver disease. The Phase 1 study is currently enrolling patients at a single center in Australia. Arrowhead may soon begin recruiting patients at additional sites and expects to complete enrollment for this Phase I trial by the end of 2015.

Arrowhead's Phase 1 trial of ARC-AAT is the first time ARC-AAT has been administered to humans. The study is designed to evaluate the safety, tolerability and pharmacokinetics of ARC-AAT and its effect on circulating AAT levels.

The study consists of two parts; Part A in healthy volunteers, which has been completed, and Part B is being being conducted in Alphas with PiZZ genotype. According to the study protocol, dosing in patients begins at the highest dose level used in healthy volunteers and then continued dose escalation may proceed. The study evaluates participants for 28 days following dosing, with additional follow-up if needed every 2 weeks until AAT plasma levels return to baseline.

According to the Arrowhead website, ARC-AAT, which was granted orphan drug designation in the United States, employs a novel unlocked nucleobase analog (UNA) containing RNAi trigger molecule designed for systemic (intravenous) delivery using the Dynamic Polyconjugate™ delivery system. ARC-AAT is effective at reducing the liver production of the mutant AAT (Z-AAT) protein in animals. Reduction of liver production of the inflammatory Z-AAT protein, which is believed to be a cause of progressive liver disease in Alpha-1 patients, may be able to halt the progression of liver disease and potentially allow repair of Alpha-1 related liver injury in patients. With success in the ongoing Phase 1 trial, Arrowhead anticipates studies in patients with evident liver disease in a multi-dose Phase 2 study.

Arrowhead and the Alpha-1 UK Support Group have agreed to exchange relevant information on a regular basis, and Arrowhead will keep us abreast of any new developments. We will keep you updated on any news from Arrowhead and other companies relating to any ongoing and prospective clinical trials.

European Medicines Agency recommends approval of CSL Behring's Alpha-1 augmentation therapy product

CSL Behring Receives Positive CHMP Opinion for Respreeza® (human alpha-1 proteinase inhibitor) as Maintenance Treatment for Patients with Severe Alpha-1 Antitrypsin Deficiency in Europe

CSL Behring announced 26th June 2015 that the European Medicines Agency (EMA) Committee for Medicinal Products for Human Use (CHMP) has recommended granting marketing authorisation for Respreeza®, a highly purified alpha-1 protein derived from human plasma, indicated to treat patients with severe (e.g. genotypes PiZZ, PiZ(null), Pi(null,null), PiSZ) alpha-1 antitrypsin deficiency (AATD).

Patients are to be under optimal pharmacologic and non pharmacologic treatment and show evidence of progressive lung disease (e.g. lower forced expiratory volume per second (FEV1) predicted, impaired walking capacity or increased number of exacerbations).

AATD is a hereditary condition marked by a lack of the alpha-1 antitrypsin protein, whose main function is to protect the lungs from inflammation. Respreeza® replaces the protein that these patients are missing and raises the alpha-1 antitrypsin levels in their blood, which can help to protect the lungs from damage due to inflammation.

"Respreeza® replaces the protein that these patients are missing and raises the alpha-1 antitrypsin levels in their blood"

"CSL Behring continues to make strides towards fulfilling our promise to improve the lives of our patients. This positive opinion from CHMP brings us closer to providing Respreeza® as a new treatment option to the AATD community in Europe," said Lutz Bonacker, Senior Vice President & General Manager Commercial Operations, Europe, CSL Behring.

The CHMP positive opinion will be transmitted to the European Commission (EC) to start the EC decision-making process. The EC may then grant a centralised European marketing authorisation for Respreeza® as a maintenance treatment to slow the progression of emphysema in adults with documented severe alpha-1 proteinase inhibitor deficiency. The CHMP positive opinion is based upon data from CSL Behring's RAPID(randomised, placebo-controlled trial of augmentation therapy in alpha1 proteinase inhibitor deficiency) study. According to findings of the study, patients with AATD treated with alpha-1 proteinase inhibitor therapy exhibited a lower annual rate of lung density decline compared to placebo, when measured using chest computed tomography, at full inspiration. This demonstrated that Respreeza® significantly slows the progression of emphysema in these critically ill patients.

"This demonstrated that Respreeza® significantly slows the progression of emphysema in these critically ill patients"

Receiving a positive opinion from CHMP is a significant achievement but the next challenge will be ensuring access to treatment in member countries, such as the UK and Ireland, where decisions regarding usage are subject to pricing and reimbursement.

Respreeza® will be indicated for maintenance treatment, and to slow the progression of emphysema in adults with documented severe alpha-1 proteinase inhibitor deficiency (e.g. genotypes PiZZ, PiZ(null), Pi(null,null), PiSZ). Patients are to be under optimal pharmacologic and non-pharmacologic treatment and show evidence of progressive lung disease (e.g. lower forced expiratory volume per second (FEV1) predicted, impaired walking capacity or increased number of exacerbations) as evaluated by a healthcare professional experienced in the treatment of alpha-1 proteinase inhibitor deficiency. Respreeza® is contraindicated in patients with hypersensitivity to the active substance or to any of the excipients and IgA deficient patients with known antibodies against IgA, due to the risk of severe hypersensitivity and anaphylactic reactions.

Source: CSL Press Release 29/06/15

Lancet Publishes RAPID Trial Results Showing Effectiveness of Alpha-1 Augmentation Therapy

The Alpha-1 Foundation on 28th May 2015 congratulated the investigators and sponsor of the RAPID Trial, demonstrating the effectiveness of augmentation therapy in slowing emphysema due to Alpha-1 Antitrypsin Deficiency, on the publication of the trial in The Lancet, one of the world's oldest and most prestigious medical journals.

When the results were first announced at the international conference of the American Thoracic Society (ATS) in 2013, lead author Kenneth Chapman, MD, director of the Asthma and Airway Centre of the University Health Network in Toronto, Canada, called the trial "the most rigorous evidence to date that augmentation therapy slows the progression of emphysema in patients with Alpha-1 Antitrypsin Deficiency. The effect of A1-PI seen in this trial was both clinically and statistically significant, finally confirming its benefit in preventing the loss of lung tissue in patients with this potentially debilitating disease."

The Alpha-1 Foundation hailed the trial results and The Lancet publication. "We congratulate CSL on this landmark clinical trial, which provides the strongest evidence yet that augmentation therapy preserves the lung tissue of individuals with Alpha-1-related lung disease," said John Walsh, Foundation president and CEO. "We hope these results will support Alpha-1 communities around the world in their efforts to win access to therapy."

Walsh said, "The Foundation has communicated its support for acceptance of the RAPID data as proof of efficacy of augmentation therapy to the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA the European equivalent of the FDA), and has continued a dialogue with both agencies to urge acceptance of these findings."

CSL Behring sponsored the trial, which randomly assigned 180 Alpha-1 patients to receive either the augmentation product Zemaira (known as Respreeza in Europe) or a placebo for a two-year period, followed by a two year of open-label extension study in which all subjects were offered augmentation therapy. "We applaud CSL for its commitment of time and effort to conduct this multinational, multi-year study." said Walsh.

The Lancet article says that an interim analysis of data from the two-year extension trial suggest that early treatment with augmentation therapy shows persistent efficacy in patients with Alpha-1 and emphysema. In addition, when patients who had been receiving a placebo in the original two-year trial switched to treatment with augmentation therapy, their lung density decline (which was more rapid than the treated group in the main study) slowed to the same rate as the treatment group. "These findings should encourage early introduction of augmentation therapy [in patients with emphysema due to Alpha-1] and should stimulate further research into optimum dosing," says the Lancet article.

Robert Sandhaus, MD, PhD, clinical director of the Alpha-1 Foundation and medical director of AlphaNet, was a co-author of the Lancet article.

The RAPID Trial is the first well-powered randomized, placebo-controlled trial to use CT scan lung density as the primary outcome measure. CT scans are currently considered the most sensitive measure of emphysema detection. The trial was conducted at 28 sites in the United States, Europe, Canada, Australia and Russia.

The multi-center, multi-national trial randomized patients with homozygous Alpha-1 (ZZ) to receive either Alpha-1 antitrypsin augmentation therapy intravenously at 60 mg/kg weekly or a placebo over two years. CT scan lung density was measured at baseline, three months, one and two years. Secondary endpoints included spirometry, changes in exercise capacity and the rate of pulmonary exacerbations over two years.

The annual rate of lung density loss was significantly less in augmentation-treated patients (-1.45 +/- 0.24 units vs. -2.19 +/-0.25 units; p = 0.017, one-sided). Secondary outcome variables and adverse events were not significantly different between groups, according to the article.

Source: Alpha-1 Foundation 28/05/2015

5th Alpha-1 Global Patient Conference Italy

Report by Andrew Willis

5th Alpha-1 Global Patient Congress in Barga, Italy

It was a privilege to attend this year's Biennial Alpha-1 Global Conference in Barga near Lucca in Italy. Parallel sessions were held for patient and clinical/scientific audiences over the three-day congress. The Alpha -1 UK Support Group was represented by Jemma Coad, Sandra Nestler-Parr, Karen North and I. Jemma was the official photographer for our team, Karen both presenting as well as being heavily involved in the organisation of the congress, Sandra represented our group in discussions with clinicians, researchers and industry, and I took notes of the sessions.

I had previously attended an Alpha-1 Patient Conference in Zurich in 2005. Compared with the Zurich conference 10 years ago, which only about 50 delegates from Europe and very few clinicians attended, this year's congress in Barga is testament to the amazing growth in the size of Alpha-1 patient groups across the world, the growth in clinical interest and research along with the growth in general awareness of the disease. The patient conference in 2005 was sponsored by just a single pharmaceutical, whereas Barga 2015 was sponsored by Baxter, Grifols and CSL Behring.

The aims of the congress were to:

- bring together representatives of the Alpha-1 community to discuss the status of Alpha-1 awareness, diagnosis and research and around the world;
- continue to build a collaborative and effective international network of Alpha-1 organizations and patients; and
- develop strategies to strengthen advocacy for
- Alpha-1 awareness, detection and access to care.

It was a real pleasure and very informative to mix with such a variety of people from different backgrounds and with a diverse range of professional interest and/or personal experience of AATD. The conference offered plenty opportunity for networking and mingling with other participants. To give you a flavor of the variety of congress participants - some of the most memorable conversations I had with:

- the single delegate from Lebanon who may be the only diagnosed Alpha in Lebanon and was eventually diagnosed through written communication by Professor Charlie Strange, an Alpha-1 expert in the US;
- Jim and Val from New Zealand who emigrated from the UK 40 years ago;
- Dennis, a retired town mayor, from Sacramento, USA;
- Peter Leone from Arrowhead, a biotech company currently developing an innovative therapy that targets Alpha-1 related liver disease and who provided information on their research to patients in a special session;
- Bill and Jenni Nankervis from Melbourne, Australia, who are members of the Alpha-1 UK Patient Support Group;

"Bring together representatives of the Alpha-1 community to discuss the status of Alpha-1 awareness, diagnosis and research around the world"

- Professor Bruno Christ of Leipzig University Hospital, Germany, who is undertaking innovative liver research into Alpha-1;
- a delegate from Romania whose son is an Alpha and who attended with a respiratory consultant - together, they are hoping to start a national patient group soon;
- Gonny Gutierrez, Alpha-1 Global Director, USA, who presented at our patient meeting in Lincoln last year. She did a great job organizing such a brilliant conference in Barga;

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- CSL Behring staff including Sebastian Soluch who is a Market Access Manager based in the UK. Market access deals with the challenges of getting health care therapies reimbursed by health insurances and payers (e.g. the NHS in the UK).

The key points that I personally took home from this interesting conference were:

- the ever more extensive research being undertaken in Alpha-1, both in liver and lung disease;

- the increase in collaboration and networking between patient communities, health care professionals, researchers and pharma/ biotech companies;

Hillegonda (Gonny) Gutierrez Director of Alpha-1 Global

- the increasing depth of knowledge of the disease and available treatment options by AATD patients;

- the increasing impact of disease awareness campaigns run by patient groups;

- Sandra Nestler-Parr from our group summarized the needs of the UK Alpha-1 community as follows, and this probably applies globally:

- "The UK needs a government-mandated specialist NHS service for the diagnosis and treatment of Alpha-1. The service needs to be delivered by multi-disciplinary teams (MDTs) of Alpha-1 experts who are integrated into the existing NHS care pathway for Alphas. These specialist services need to work in a coordinated fashion and to agreed national standards. These MDTs should include healthcare professionals that can cover all aspects required for optimal clinical management of Alpha-1. There needs to be better access to specialist Alpha-1 care as well as improved equality of access to adequate services across the country."

One topic which was frequently and tantalizingly mentioned throughout the conference was an Advocacy Toolkit for patient groups which would provide a set of generic guidelines for any country to set up an advocacy programme to lobby governments and other official bodies for the improvement of aspects of AATD diagnosis and care. It would have been good to have some trial training sessions with it at the Barga congress, but perhaps it has not yet been far enough developed.

The session topics covered a large variety of subjects including clinical aspects of Alpha-1, latest news in Alpha-1 research into novel therapies, patients support, advocacy and the importance of patient groups. Below is a brief summary of each of the sessions presented at the patient conference. If you would like more detailed information about any of the individual sessions, please contact John Mugford or myself.

"Alpha-1 Global Patient Community Updates"

In the first session a status report was provided from representatives of each of the global areas: Australia and New Zealand, Canada, USA, South America, Central Europe and Western Europe.

Karen North Vice Chairman Alpha-1 UK Support Group

"European Medicines Agency"

This session included a technical but very interesting talk by Dr Laura Fregonese of the European Medical Agency (EMA) about the scientific and regulatory pathways for new treatments for rare diseases, a very pertinent topic regarding the currently anticipated EMA license for CSL's augmentation therapy product (see page 5 of this newsletter).

Karen North from our group gave a presentation in the second session, entitled *Case Study: UK Campaign, Advocacy and Government Relations* in which she spoke about the rationale for a patient-led national campaign, the underlying campaign strategy and achievements of our campaign in the UK, including our national patient survey and policy reports for England and Scotland.

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Karen concluded by listing some of the challenges for such campaigns, such as the rarity of Alpha-1, national health budget restrictions, competition for funding from other rare diseases and changes in political priorities.

"Liver Disease – Therapies on the Horizon"

This session began with an excellent summary of the functions of the liver presented by Dr Jeffrey Teckman, USA. In essence, the liver produces bile which is delivered into the intestine along the bile duct in order to help digestion, remove waste from blood, process

Professor Robert Stockley

nutrients from food, remove drugs and toxins from blood, and produce proteins including alpha-1 antitrypsin.

The second speaker in this session was Professor Bruno Christ from University Hospital Leipzig, Germany. He is developing a novel treatment approach for Alpha-1 that involves transplantation of heathy hepatocytes (liver cells) into the liver. The idea behind this approach is that the healthy hepatocytes can function alongside unhealthy liver cells. Stem cells from patients are harvested and turned into healthy hepatocytes before being transplanted into the patients' liver. Currently, the survival time of transplanted hepatocytes is unfortunately limited. Professor Christ stated that, at present, it is not known whether liver therapy in any way affects Alpha-1 related lung disease.

"The UK needs a government-mandated service for the diagnosis and treatment of AATD".

"Lung Disease – Current and Future Therapies"

The first speaker in this session was Professor Robert (Sandy) Sandhaus, Medical Director of Alphanet in the US. Dr Matthias Griese from Munich, Germany, spoke on the four Alpha-1 inhaled studies which have been run so far. The next presentation was by Jonathan Edelman of CSL Behring, USA, entitled *The Effect of Intravenous Alpha-1 on Lung Structure*. The final presentation in this session was given by Professor Jan Stolk from Leiden, Netherlands, on *Future of Gene and Stem Cell Therapy*.

"Research Registry Updates":

The session began with an overview by Dr Kenneth Chapman from Canada, of the functions of Alpha-1 patient registries which include:

- facilitation of research by bringing patients to researchers together;
- recording the progression of disease in Alpha-1 patients;
- recording information on patients' treatment regimens; and
- facilitation of national or regional standardised approaches to managing the disease.

The drawbacks of patient registries are that they are not necessarily set up to deliver clinical care. Another big issue is the question of who should provide the necessary financial support to run patient registries – the pharm/biotech companies developing new therapies, health care providers such as the NHS or others? Professor Rob Stockley reported on the UK registry (ADAPT), Charlie Strange on the US registry and Dr Ilaria Ferrarotti on the Italian Alpha-1 patient registry.

Dr Kenneth Chapman

"Combined Session with Researchers and Patients"

The session provided an overview of the most exciting research news in Alpha-1 and underlined that Alpha-1 is now thoroughly on the medical research map – this was also reflected by the calibre of speakers in this session.

- Adam Wanner, Alpha-1 Foundation, USA, discussed amongst other things whether animal models are necessary for human Alpha-1 research.

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- Professor Robert Stockley, Director of ADAPT, UK, emphasized how better patient/GP interaction leads to better health outcomes: so, know your disease!
- Dr Stolk, Netherlands, spoke on recent advances in CT scanning technology.
- Professor Ed Silverman, USA, spoke on his recent findings that the risk of lung disease in heterozygous patients i.e. MZs is increased. This is a relatively new subject as it has been thought until recently that MZs are not at greater risk of lung disease than normal COPD patients.

"Gene therapy is probably 10 years away for lung disease"

- Dr Matthias Griese, University of Munich, Germany, who conducts research in cell based therapies, spoke on inhaled Alpha-1 therapies and emphasized that it has been shown to be safe and, in a trial in cystic fibrosis patients, was shown to have a positive treatment effect.

Andrew Willis, Dr Sandra Nestle-Parr, Karen North, Jemma Coad

- Dr Ron Crystal, Cornell University, USA, spoke about a particular gene therapy technique which has been found in animal experiments to be safe and raise serum antitrypsin levels.
- Professor Sandy Sandhaus, USA, spoke on alpha-1 augmentation therapy and the challenges of defining meaningful and sensitive primary study endpoints in order to assess in gold-standard clinical trials whether the therapy works.
- Dr Mark Forschag, Glaxo SmithKline, USA, summarised the challenges that both regulators and the pharmaceutical industry face in regards to new treatments for rare diseases. He also emphasised that, in addition to protecting the lungs from oxidative stress, alpha-1 antitrypsin has many other important functions in the body that are moving into the focus of clinical research.
- Professor David Lomas, UCL, London, provided an overview of his research into addressing the problem of alpha-1 polymers that are retained in the liver of Alphas by trying to create a drug that can break up these polymers.
- Dr Mark Brantly, University of Florida, USA, spoke about the different technical approaches that are currently under investigation to create a gene therapy that can fix Alpha-1 related lung disease. Early results suggest that some of the techniques that use viruses as alpha-1 trypsin 'gene production systems', are safe and promising. However, Dr Brantly emphasised that a commercial gene therapy for Alpha-1 lung disease is still quite a few years away from becoming available.

"Partnership and Networks to Strengthen the Alpha-1 Message"

The main presentation of this session was delivered by Johan Prevot of the International Patient Organization for Patient Immunodeficiencies (IPOPI) who provided a case study from his organisation where the focus on partnering with other stakeholders has related in improvement of care for patients, an example from which the Alpha-1 community can learn. Frank Willersinn of Alpha-1

Dr Sandra Nestle-Parr and Jemma Coad

Global spoke on the importance of forming partnerships with other rare disease groups, stakeholder collaboration and sharing of patient stories.

"Patient Advocacy"

This session was also delivered by Johan Prevot of IPOPI who talked about the importance of pan-European advocacy, but he also stressed that advocacy needs to be targeted at a national level in order to achieve changes. Johan presented a case study of IPOPI's international campaign which has resulted in improvements for patients in many countries. The campaign included running skills building workshops to train patients on how to effectively engage with politicians and other stakeholders.

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Johan emphasised the importance of underpinning any campaign with good clinical data in order to convince key opinion leaders to implement proposed changes to clinical care. A well-run global campaign can make an impact nationally, empower local organizations and improve access to correct diagnoses and clinical care.

John Walsh President COPD Foundation

"Global Patient Community: Building a Sustainable Organisation"

Johan Prevot of IPOPI also led this session and provided a summary of the key points that any patient community should focus on in order to successfully establish sustainable and successful patient organisations.

"Global Communication, Campaigns and Resource Sharing"

Shane Fitch of Alpha-1 Global emphasised the value of working together and sharing knowledge and resources. This includes translation and adaptation of materials as well as running and attending global conferences. Her top tips on how to enhance global communication included better engagement through social media and wider interest groups, improving a creative digital presence as websites per se may be on the way out whilst Twitter and Facebook are taking over, engagement with national and global umbrella organisations. Shane also reminded the audience to be realistic about the cost of new therapies and the importance of creating guidelines for standards of care and increasing patient advocacy activities.

"Meet the Experts"

This session provided an opportunity for patient delegates to ask the experts questions. The experts on the panel were Dr Kenneth Chapman, Professor Bruno Christ, Luciano Corda, Dr Laura Fregonese Professor Robert Sandhaus, Shane Fitch, Frank Willersinn and John Walsh. The question and answer session included but were not limited to issues relating to:

- the regulatory approval process for new drugs;
- criteria for clinical trials to improve chances of getting licence and reimbursement;
- the problem of new patients not knowing that Alpha-1 patient registries exist;
- the interconnectedness of liver or lung disease in Alphas and how this relates to therapeutic approaches;
- the desired achievement in Alpha-1 in the next years.

Keywords

If you want to undertake your own research and extend your knowledge, then here are some relevant keywords that you could use in a google search: COMP (Committee for Orphan Medicinal Products), orphan medicine, clinical trial, primary endpoint, regulatory

Sandy Sandhaus, and John Walsh

procedures, quality/safety/efficacy, therapeutic indication/side affects/risk management, Alpha-1 Alliance, NHS services, multi-disciplinary team (clinical), public policy, null/null, hepatocytes, fibrosis/cirrhosis, bile duct, MM/ZZ/MZ, gene therapy, elastase, augmentation therapy, recombinant, bronchodilator, gene therapy, retinoids, Alpha-1 International Register (AIR), dose-ranging study, EXACTLE study, RAPID study, Severe Combined Immuno-Deficiency (SCID), child screening, pharmaco-economics, health economics, cost benefit analysis, cost-utility analysis, health technology assessment (HTA), standards of care.

Thank you again for allowing us to represent you at this interesting international event.

Dr Sandra Nestle-Parr, Andrew Willis, Karen North, Jemma Coad

Hillegonda (Gonny) Gutierrez
Director of Alpha-1 Global

About Alpha-1 Global

The mission of Alpha-1 Global is to build and maintain a collaborative global network of Alpha-1 organizations and patients to increase awareness, detection, and access to care around the world. Alphas worldwide experience many of the same problems: little to no awareness of their condition; long delays in getting the correct diagnosis, all the while experiencing deteriorating health; inadequate clinical care by physicians unfamiliar with the standard of care for lung and liver disease caused by Alpha-1; and, in many cases, limited or no access to augmentation therapy (the only specific therapy for Alpha-1).

The various challenges of obtaining the appropriate medical care vary in severity by country. Many countries have already faced these hurdles, and have made progress in the areas of awareness, detection, health management, and access to therapy and reimbursement. While countries have different governance structures and

health care systems which demand a unique approach to advocacy, much can be learned from the experience of Alphas who have faced these same challenges and have won some hard fought battles.

5th Alpha-1 Global Congress

In collaboration with AIR, the European Alpha-1 International Registry, the Foundation convened the 2nd Biennial International Research Conference on Alpha-1 Antitrypsin and the 5th Global Patient Congress in Barga, Italy, on April 9-10, 2015. Both events combined attracted more than 200 individuals from 26 countries.

All components of the Patient Congress were organized by

Congress Hall Barga Italy

Alpha-1 Global, and the program was focused on avenues in which the global community can collaborate and learn from each other as it pertains to education and access to care and therapy. Presentations were given by renowned Alpha-1 scientists, clinicians and experts, who spoke on the status of the latest research, the regulatory framework for licensing therapies for rare disease in Europe, and the status of patient registries.

Physicians, researchers and Alpha-1 patients emphasized the need of creating strong collaboration across patient, clinician and the scientific communities. Close relationships between all groups will progress the areas of earlier diagnosis, disease management, and the role of patient registries.
The need for building sustainable organizations to strengthen the Alpha-1 message and reinforce patient advocacy, was one of the most talked about topics during the Patient Congress. To reach this goal, the audience agreed to develop patient networking and communication strategies focused on raising awareness, detection and access to care.

Global Congress Action Items

The following action items are put into place:

1) Design a comprehensive Global Awareness and Screening Campaign for 2016. Mrs. Cristina Barbiero from J.W. Thompson Marketing Agency (Health division) in Rome/Milan has offered to assist with the design of this Global Campaign. Initial discussions with the Global Steering Committee are taking place before the end of July 2015.
2) Assist the Nordic countries (Denmark, Sweden and Norway) with a unified approach towards (re)educating their medical community and policy makers. Alpha-1 Global will use this “Nordic Pilot” to build a campaign template for other countries.

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2) Karen North, our UK Steering Committee member, will be able to assist as necessary, since she is a forerunner in the UK and Scottish Alpha-1 campaigns.
3) Latin American Alpha-1/COPD Summit. On December 4-5, prior to the Latin America Respiratory Congress in Buenos Aires, Argentina, from the 6th-10th, we will conduct a regional Summit for Alpha-1/COPD doctors and patient leadership to discuss a networking and advocacy strategy specifically for this region of the world. Spanish and Portuguese translation will be provided.
4) In April 2017, we will organize a next Patient Congress, combined with an International Research Conference on Alpha-1 Antitrypsin.

Group Photo Barga Italy

Alpha-1 Global Website

The website continues to serve as a centralized Alpha-1 Global communication platform for the Alpha-1 community worldwide. With the help of a designated communications manager, we have established the following communication flow to keep Alpha-1 partners informed, engaged and motivated to pursue the Alpha-1 Global mission:

- Provide current news related to research, medical and global community
- E-news (bi-monthly)
- A designated Alpha-1 Global FB page (forthcoming in August)
- Twitter (forthcoming in August)

Alpha-1 Global Advocacy

The Alpha-1 advocacy toolkit has been developed, which can be adapted for use by Alpha-1 communities in their own language and context. This toolkit will be made available in August as downloadable resource on our website. Other advocacy resources at this point include:

- The policy reports summarizing national Alpha-1 patient surveys in England and Scotland, utilised by the UK patients for their national campaign
- The Standards of Care guidelines for Alpha-1 (produced by CSL Europe) and Expert Recommendations, Alpha-1 in the European Community
- USA Advocacy Training Video

Additional advocacy tools will be provided as they become available.

Global Steering Committee

The Global Steering Committee, currently consisting of 8 dedicated Alpha-1 leaders (from Belgium, Spain, Portugal, Denmark, UK, Canada, USA, and Australia) who all have greatly contributed to building a solid program infrastructure and cohesive network of leaders. The 4th face to face Steering Committee meeting is scheduled to take place on July 23, preceding the National Education Conference in Orange County, California.

Alpha-1 Global Representation

Our network of countries is steadily growing. Due to enthusiastic participation of our steering committee members and network partners, Alpha-1 Global will have an active role

in a variety of upcoming meetings.

The most key events forthcoming are:

- The International Patient Advisory Committee during ERS Congress where we have been invited to participate. Apart from this important meeting, ERS is a valuable place for networking with other organisations and industry partners
- The 2nd Central-Eastern European Alpha-1 Antitrypsin Network

Conference in Warsaw, Poland, in October 2015. Representatives from Poland, Slovakia, Czech Republic, Hungary, Bulgaria, Romania, Ukraine, Belarus, Lithuania, Russia and possibly other countries will attend this 150 people gathering. Dr. Frank Willersinn and Gonny Gutierrez will participate in the conference program.

alpha-1 GLOBAL

www.alpha-1global.org

Alpha Parent and Children Day Bristol Zoo *Party In The Park* *Report by Jemma Coad*

Our first ever Alpha-1 UK Support Group children's day (Party In The Park) for families affected by Alpha-1 was held at Bristol Zoo on Sunday, 2nd August. The event offered an opportunity for Alpha-1 families from all over the country to meet and share their experiences of living with Alpha-1 and/or having children who are affected by the disease. I am delighted to say that the day was a great success!

Thirteen Alpha-1 families from across the UK met together for the first time. The day was a great way for children and parents to meet and see that they are not alone.

The day started off by meeting in the Bristol Zoo Garden Room which we hired for the day. I met and spoke to the families, it was lovely to put faces to the people I had previously spoken to, either on the phone or via our charity's social media sites. Whilst having coffee, we all shared our Alpha-1 experiences and how the condition had affected us as a family.

At mid-morning some families did the ZooRopia experience whilst others went off to explore the zoo. ZooRopia is a five metres high adventure ropes course that gives visitors a unique opportunity to swing alongside some of the most popular animals at the zoo.

At noon we all met again at the Garden Room for a buffet lunch. All families mixed well whilst the children ate, talked and played together.

After lunch we all explore the zoo - some went in groups others went with their families. We got to see the whole of the zoo and many animals close-up, which included lions, seals, penguins and much more.

After a good look around we headed back to the Garden Room where we had a close animal encounter: the children sat on the floor ready to meet the animals that Linda and Emma, two staff from the zoo, came to show us. Linda and Emma talked about the different animals that the children got to hold which included stick in-

sects and cockroaches. There were also lizards and snakes but they couldn't be held as they didn't like to be handled by lots of people. The children loved this experience and asked lots of questions, and they were not scared at all holding the animals - unlike the parents!

Then a perfect day came to an end. Everyone had a fantastic time and gained so much out of meeting other Alpha-1 families that had the same or similar experiences as them, the children got to play with each other and realised that they are not alone and there are other children out there who are also Alphas. Everyone was keen to make the *Alpha-1 Parent and Children Day* an annual event. It was a truly fantastic day.

This is what some parents had to say about the day:

Hodges family, Somerset:

"Myself, my husband and three children were invited to the "Alpha-1 Party in the Park" by Jemma, a friend that I have made on the Alpha-1 UK Support Group. We all jumped at the opportunity to meet others that may understand what we are going through. We have only just been made aware that this condition exists after our daughter was diagnosed with it this year, then consequently one of our sons and my husband.

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The whole day was fantastic, we felt welcome and everyone was so friendly. We made some new friends there that don't live to far from us so we will be arranging to see them in the near future. It was so good to hear other people talking about Alpha-1.

No-one we know knows anything about this disease, neither do the doctors half the time, so hearing people practically mimicking what we were saying with their experience was amazing. The zoo experience with the buffet lunch and the animal encounter was brilliant. The kids loved it, it was such great value for money and we got a free parking spots which was even better!

Jemma did a fantastic job in organising the event and made us feel at home and at ease. The day went very well, was very well organised and the goody bag the kids got was ever so thoughtful and they very much appreciated it. The whole experience has been a blessing for us. We needed this so we can begin to understand what lies ahead. I'm truly hoping that this will become a yearly thing, if not, more often would be fab! Thank you so much for organising such a fun filled day and informative day."

Gregory family, Manchester:

"We truly loved the Party in the Park get-together. It was amazing to meet new friends and family's going through what we are going through with Alpha-1. Eva adored playing with the other children. Jemma had organised it so well and the day was fantastic. Really hoping this becomes an annual thing as we loved every minute of it."

Curran family, Somerset:

"The organisation was slick. Our directions were clear and we were shown into the conference facilities, which were very grand. The fresh coffee was an excellent arrival refresher after our lengthy drive. As others arrived, we found it interesting to compare perspectives on the Alpha-1 symptoms and we learnt more on a how wide spread they were. We found a couple who take their son to the same consultant that we go to. The zoo exploration then began and we returned for lunch. The food was excellent and very generous - one of the best buffet lunches we have had.

More zoo exploration and then a hands-on animal session and individually chosen toy packs for the children (a fantastic touch Jemma). Overall, fantastic value and we took away a great deal of shared knowledge."

Edwards family, Cambridgeshire:

"Ben's condition is so rare that meeting others who have Alpha -1 is difficult, so having a get-together like Party in the Park (zoo) was a great idea. It was lovely to talk to other families in our position, and Ben and Oliver made some new friends. All in all a truly fantastic day."

Gardiner family, Dorset:

"We had a lovely day at Bristol Zoo, the weather was perfect and the company was great. It was really good to have the opportunity to talk face to face with other Alpha-1 families and to people who understood what it is like living under the shadow of Alpha-1. It was lovely that our special children were allowed a special treat and for them to realise that there are other children just like them."

Hunt family, Buckinghamshire:

"Thanks again Jemma Louise Coad for a fabulous day. It was the best day to finish a perfect weekend away with my family. Lovely to meet a few people whose lives are/have been affected by Alpha-1 and I feel blessed to be part of a great group of people."

13th Annual Social Gathering/Information Day

Lincoln September 2014

The Alpha-1 UK Support Group 13th Annual Social Gathering was held on Saturday, 20th September 2014, in the historic city of Lincoln 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 Robert Stockley
Director of ADAPT, the UK Alpha-1 Patient Registry and research programme based at the Queen Elizabeth Hospital Birmingham.

Professor Nedim Hadzic
Consultant Paediatric Hepatologist at the Paediatric Centre for Hepatology, Gastroenterology and Nutrition King's College Hospital London. Professor Hadzic's main clinical research interests are biliary atresia, Alpha-1 antitrypsin deficiency, primary immune deficiencies, liver-based metabolic conditions and liver transplantation.

Ms Sarah Jones
Clinical Research Physiotherapist for the Biomedical Research Unit at Royal Brompton and Harefield NHS Foundation Trust. Sarah's main clinical areas of interest are in post-hospitalisation pulmonary rehabilitation.

Jamie Holyer
former Secretariat of the Alpha-1 Alliance who are campaigning for NHS Alpha-1 expert centres and better access to treatment for Alpha-1 patients.

Lindsay Jarrett (Patient)
Alpha-1 patient with severe lung disease. Lindsay is rock climbing up and down the UK from April to September 2014 to heighten awareness of Alpha-1 and to give talks along the way, Lindsay said "I am attached to oxygen but do not let this stop me, I train five times a week to strengthen myself for this 'marathon' I have set myself."

Hillegonda Gutierrez (Gonny)
Alpha-1 Foundation USA Global Director - Gonny spoke about how the Alpha-1 community worldwide can collaborate to further its cause.

About 100 participants attended the meeting on the Saturday with the majority staying at the hotel and making a weekend of it...it's always a popular event.

As well as presentations by our guest speakers, there was the usual Auction, Prize Draw and Tombola during the day with music and much merriment in the evening. It's always so nice to meet up with old and new friends each year and it's sad to say goodbye on the Sunday morning. However, everyone agreed that, as usual it's been a fabulous weekend and can't wait for the next one.

The 14th Annual Social Gathering will again be held in Lincoln at the Bentley Hotel Leisure Club and Spa on the 12th September 2015.

We look forward to seeing you

Members Stories - Cora Paterson (Caregiver)

Cora Paterson

I consider myself a wife, not a carer, but over the last 8 years my role has metamorphosed into the carer one. I stopped using aerosol deodorants 8 years ago as they would get on my husband's chest and have him reaching for his inhaler. Over time aerosol cans have been replaced with wipes or trigger sprays in my cleaning cupboard. In the last 2 years we moved our bedroom down - stairs as my husband was struggling with the stairs.

Every morning I deal with our children, and unlike in most marriages, my husband is unable to support me when the children are upstairs playing up, getting washed and dressed because he struggles to get up the stairs. When he has made the trip he will lean on the wall for several minutes gathering his breath.

My husband drives and I don't, and the children's school is a couple of miles away, so he drives us. Going from the house to where he parked the car (he tries to park as close as possible) again leaves him struggling for breath, so he has to sit for a couple of minutes to catch his breath.

Our routine after the school run depends on whether I am working or not. If I am not working and it is time to clean, I often use a damp cloth to dust and try and keep dust and anything else that could irritate his lungs at bay. I can't use air freshener sprays, sticks or plugins. I do occasionally burn scented candles. I open the windows to try and air the house so I have planted my garden with Jasmine, Honeysuckle, English Lavender and Roses in the hope that their sweet scent enters the house. If I am working I'll do washing and fold the washing before work. I do my shopping before I start work and have to ensure only to get the essentials, as my husband has to take the bags into the house. Carrying too much can cause a loss of his bowel control. While at work my husband collects our youngest from nursery which is approximately 300 yards from our home, but he has to drive as he struggles to walk this far and would have to stop several times for about 5 minutes to get his breath along the way. I rely on my mother in law to collect and look after the other children whilst I am at work.

I have to do all the cooking nowadays, my husband used to love cooking but this is another thing he has come to struggle with. Our oven is a fan oven and when opened the steam takes away his breath and he reaches for his inhaler. So he now rarely cooks to avoid struggling for breath.

We are both not yet 40 years old, and my husband often feels useless and depressed because he is unable to do so much that a person of our age should be able to do. He often lashes out verbally because of how low and useless he feels which, in turn, has a negative effect on my confidence, self-esteem and often has me feeling low. However, as a caregiver and mother I have to carry on because too many people rely on me to keep everything going. I only get support online from Alpha-1 UK Support Group. My husband has only just (after 8 years of suffering) applied for a blue badge and to go on our County Council's disability register. He still doesn't claim DLA or PIP and has not applied as he believes his claim will be rejected and he is left feeling like a fraud.

Lee Paterson (Right)

On a very personal note, intimacy is also greatly affected. Yes, we have the leaflets about positions but they don't help to cope with the guilt of having to stop making love because you see your loved one struggling to breathe.

Members Stories - Norah Oliver

Norah Oliver

My life as an Alpha-1

Where to start ..well I guess the best place is the beginning ...

I was the youngest child of a large family and lived in the country where my Dad was a shepherd, all the houses we lived in were 'tied' houses, meaning that they belonged to the farmer who my Dad worked for. The houses were usually cold, damp and no heating apart from a log fire in the kitchen. I remember the water streaming from the windows in the winter. Although I personally didn'tsuffer side effects from this then, my sister Ivy did and because she had severe asthma she was sent to live by the sea in Hayling Island to be looked after by nuns for nine months. It wasn't a nice experience for her but she did come back seemingly clear of asthma. She has since found out she is PiMZ.

"At eighteen I started smoking as it was the 'in' thing to do! Something I will always regret."

My problems started to occur later. At school I couldn't run and even jumping over the 'pummel horse' in athletics would leave me breathless.. At eighteen I started smoking as it was the 'in' thing to do! Something I will always regret.. Well life carried on and at that time I was living in Newquay in Cornwall and I vividly remember walking up the hill there to work. It must have taken me forever as I was constantly stopping to catch my breath, I just thought this was ' normal'. Aren't we naïve when we are 23! Anyhow my marriage broke down which caused more stress. Eventually I met and married Bob and we were lucky to have our daughter Lisa. I probably should add here that when Lisa was early eight she was diagnosed with leukaemia, I virtually lived in Great Ormond St hospital with her for two years and when she was asleep I would go to the parents room and smoke endless cigarettes, along with the other mums and dads...just to help us live with the stress and worry. Thankfully Lisa recovered and life went on.

About this time I started to get repeated illnesses and endless chest infections and bronchitis. One such episode left me so ill that I was referred to a respiratory specialist, who after getting me to breathe into the spirometer told me I had 25 percent lung function, also that I had asthma and emphysema. I was 40 years old and had already stopped smoking when I was so ill.

The next 13 years my life I carried on with the usual inhalers. At this time I was working in a shoe shop. A job I loved and had great friends there. However it was getting harder and harder to work, which involved constantly climbing up ladders for stock. Without my friends helping me out I doubt I would have managed it as long as I did.

One day in May 1999 I suddenly collapsed at home and was taken to hospital. There I was seen by the same specialist I had seen 13 years earlier. Whilst in hospital I had the dreaded blood gases done for the first time by needle in the wrist ..oh how that hurt!!

Me with my sisters Dolly and Ivy at the Wales group meeting

Unbeknown to me a test was carried out for Alpha1. On my next visit to see the specialist he dropped the Bombshell, I had a genetic lung disease called Alpha 1 and would probably need oxygen 16 hours a day. Needless to say I was devastated. Plus my first thought was, has my daughter inherited this as I was PiZZ… she turned out to be PiMZ.

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The first thing I did when I got home was to look on the Internet ..a fatal thing to do! I read the average lifetime age was 55 and as I was 53, I remember saying to Bob "I've only got two years to live". That year was the worst ever for me as I was rushed into hospital so many times unable to breathe. On one such occasion I was unconscious when Bob found me and I remembered nothing until I came to in the ambulance. About this time I was put in touch with an alpha called

With Bob and Family on London Bridge

Robin who lived about 12 miles away and he became a good friend and was a mine of information on Alpha-1. Sadly he is no longer with us. I joined the apha1 Support group which included Robin and only about six others. One of them being our dear departed John Doyle, who was one of the founder members.

I was referred to the ADAPT project and received very good annual assessments for 13 years. My sister Dolly was also found to have the ZZ gene.

As I loved my job I tried to go back but as I was working with the general public especially children, I ended up having repeated chest infections and sadly had to retire on health grounds. When I was about 60 years old I was referred to Papworth for lung transplant. As it turned out I was turned down for two reasons. One being I was too well and the other that because of my other illnesses it was unlikely I would even survive surgery.

So from getting the original diagnosis 16 years ago I have remained relatively stable. Although I have been in hospital a few times with pneumonia and chest infections I know how lucky I am. Yes life is hard and getter harder for me as I get older, I am now nearly 69 so a long way from that dreaded 55 years!!

Getting dressed now is a struggle and any exertion makes me breathless...but I still only use oxygen on a ambulatory basis. Getting ready to go out exhausts me and sometimes it's easier just to not go. When people see me they say "you look so well', but they have no idea the effort it has taken me. However, I know that many of my alpha friends are struggling much more than me and I am in constant awe of how they cope.

"If you are unlucky enough to be diagnosed as being Alpha-1, please remember it doesn't mean a death sentence"

Since joining the Support group it has gone from strength to strength and I have made so many wonderful friends here. Without their support and my husband Bobs, whose life changed too when I became ill, I know the depression which anyone gets living with a chronic illness would be much worse.

My story is of an ordinary person, I haven't achieved any major feats or done anything spectacular.

Enjoying a stroll at Rutland Water

I am just a normal person who was born with abnormal genes!

I hope my story is a story of hope rather than despair .. If you are unlucky enough to be diagnosed as being Alpha-1, please remember it doesn't mean a death sentence. So much is happening in genetic research now and I truly believe a cure will be found in the not too distant future. Not for me maybe, In the meantime I love life, have a wonderful family and husband and am truly grateful for every day and hope to be here for a few years yet!.....

Members Stories - Anonymous

Anonymous

I am an alpha.
Am I surprised?, No
Is there anger, ? Yes
Why I am not surprised?

Leading up to December 2011, I was ill and it was getting worse. It was diagnosed with idiopathic membranous glomerulonephritis (kidney disease) and treated with ACE inhibitors. Thankfully this condition has stayed in remission so far. But over that terrible Christmas, a cough and a chest infection also developed. The cough was so explosive that it caused an umbilical hernia that required an operation. After months of taking different antibiotics, the bacterial infection was eventually eliminated; yet the cough continued. I easily became breathless and that, too, remained.

My GP referred me to a distinguished chest specialist who was briefed on my medical history, including details of childhood asthma and hay fever. I told him that my father had chronic chest problems all of his life, and my brother had asthma and serious chest problems as a child.

The specialist initially diagnosed background asthma, which is what my GP has diagnosed.

He put me on a course of medication for 6 weeks that was different from that prescribed by my GP. Initially, he observed that the coughing in particular might be due to night-time reflux, as I suffer from hiatus hernia and GORD.

A follow-up was due two months later. Spirometric tests were performed but no check for asthma reversibility.
Having already had chest and lung x-rays, the specialist announced his Conclusions as:

1) Background asthma that the medication had moderated; my chest he said was fine.

2) After conducting research, he was convinced that the cough symptoms were caused by GORD/reflux and he was going to prescribe an alternative, more effective treatment.

3) He also thought that the cough might be exacerbated by the ACE inhibitor that I was prescribed for renal problems and hypertension. Again, he was going to change the medication to an ARB.

4) Finally the spirometry results were normal, indeed above predicted for my age and BMI. The specialist said this result was a predicator of long-life so I should be pleased!

"The specialist initially diagnosed background asthma as had my GP"

I disagreed with points two and three. I knew the symptoms of night-time reflux from experience and these were not apparent. He said I might not know.

The cough developed 5 months before being prescribed an ACE inhibitor and I did not believe this was contributory. The specialist was however convinced of his diagnosis and signed me off. On a deeper level I privately thought he was wrong.

In all this reasonable discussion between adults, the previous points made about my father and brother was forgotten! His qualified opinion clearly overruled my own experiences and the new medication was prescribed with disastrous results.

The new medication for reflux upset my system and gave me acute stomach problems and diarrhoea. Within three months I was back on the original medication. With agreement from my GP, we had not changed from ACE inhibitors because previous changes to blood-pressure medicines had caused me several upsets. In any case, neither I nor my doctor felt that this factor was contributory to my condition.

Since the beginning of 2013, my GP tried various medications for the cough and breathlessness all to no avail.

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About mid-2014, I gave up the asthma medication as it was having no - effect.

Over Christmas 2014, the cough got worse and the breathlessness increased.
I refused to be put antibiotics as sputum tests revealed there was no evidence of a bacterial chest infection. In desperation I told my GP that that the asthma medication had had no - effect and we must now look at other causes.

I took an initial spirometry test followed by a second after a dosage of Medihaler to test for asthma reversibility. The results showed moderate COPD with the Medihaler proving no benefit.

I was offered a steroid combination Medihaler yet I refused this, just in case I had a viral infection and this was a one-off result. I get on with my GP very well and we talk things over.

Six weeks later a follow-up test gave a worse result. In a previous life I worked in the pharmaceutical industry and had studied applied biology. I therefore suggested an alpha-1-antitrypsin deficiency test. I remembered the genetic connection to COPD and had explored some research papers on the internet to refresh my knowledge.

"One of my brothers, plus a blood-cousin on my mother's side had chest problems. My father had serious chest illnesses all of his life"

My GP agreed and the test was done. Four days later, I was advised that my serum alpha-1 antitrypsin level was below the reference range and my serum was to be phenotyped. Six long weeks later, I was told that the result was two defective alleles, so I am homozygote. This means that both of my parents were at least carriers!

Whilst I had waited for the phenotype results I did some familial research as well as, medical research. I remembered that although my mother had no chest problems (other than two bouts of pneumonia before I was born), her youngest sister died early from chronic chest problems. Also one of my brothers, and a blood-cousin on my mother's side had chest problems. Again, my father had serious chest illnesses all of his life; but I knew nothing about his family who were long dead in another country.

"When will specialists start to really listen to their patients and step outside of their comfort zones and test for Alpha-1"

So with the low serum alpha-1 antitrypsin result and prior to receiving the results of the phenotyping, I guessed at-least to be a carrier. There was a risk that it could be worse and so it has proved – no surprise! My anger is that this reputable chest specialist had given me the all-clear, seemingly taking no-account of the familial history and the other factors that I had told him about. My GP is blameless and had little option but to agree with his respected opinion. In any case, she has been most helpful throughout my many unfortunate illnesses.

Sadly three and a half years has been lost whilst not having received the correct diagnosis and I understand from the support group website that this is far from uncommon with Alpha-1. And all because a specialist has fixed ideas and will not listen to the patient?

This one-sided approach is contrary to basic professional patient - doctor relationship.

The defective phenotype result has had far reaching implications. My daughter who is eight months pregnant and lives 400 miles away has had to be tested and we are currently awaiting the results. The baby will probably arrive before she gets the results, and the baby and her husband may also need to be tested.

Since my last doctors visit, I have again been referred to a COPD specialist. The name is not yet known; but if it is the previous consultant I might ask for someone different.

Although usually quite amiable, I am now so ill that I do not trust myself what to say to this specialist about my feelings. Living alone and with my health deteriorating, over the past few years I have tended to withdraw from friends and social activities; so this is no joke as I now feel vulnerable and isolated.

I fully agree with the support group's mission and message. It is 2015, so when will specialists start to really listen to their patients and step outside of their comfort zones and test for Alpha-1 at the first opportunity?

Name withheld

Members Stories - Mark Bradford

Mark Bradford

I have written this in the hope it will help other people understand the choices you may have to make with end stage lung disease.

<u>I am not intending to provide medical advice</u> - this is my personal journey and the decisions I made surrounding pre-transplant lung surgery and why I made them…

I have seen a lot of interest regarding being listed for organ transplantation or having Video Assisted Thoracic Surgery (VATS) followed by talc sticking the lining of your lung together (pleurodesis).

This for me is a three year journey, after a very bad spell of infections I started to collect fluid in the cavity of my right lung, (plura). To remove the fluid they put in a chest drain. This is done by placing a needle and tube into the plura, and the fluid then drains off into a bottle.

"the consultant then spelled out my options pleurodesis or transplant"

This is all done under local anesthetic and you are awake the whole time, it's uncomfortable but bearable. Unfortunately for me, this kept happening so they asked if they could look inside my lungs using the VATS procedure. They discovered the lining of my lungs were badly scared and most probably would never heal enough to stop fluid entering into the plura. In all I had seventeen lung drains and one permanent drain (which blocked after two weeks) fitted. This all took place over the period of a year.

The consultant then spelled out my options, pleurodsis or lung transplantation. I could be listed for a double lung transplant now or have the lining of my lungs stuck together, and the chances are I could only have a single lung transplant later if my lungs kept on deteriorating.

"the consultant said in his opinion my lungs were "past the tipping point" and they were eventually going to pack up"

As I had a liver transplant three years earlier and the new liver was producing the correct alpha-1 antitrypsin protein, they could not be sure. At around this time they started me on a long term course of Azithromycin, an antibiotic 3 times a week. This was to help prevent further infections and damage to my lungs.

My choice was the pleurodsis route,as I felt, at that moment in time, my lungs were bad but I could get by. And I could still have a single lung transplant later on in life if it became necessary. I had to weigh up the fact that, statistically, a Single lung transplant doesn't last as long as a double lung transplant. But what if my lungs stopped deteriorating after pleurodsis I might never need a lung transplant? The consultant said that, in his opinion, my lungs were "past the tipping point" and they were eventually going to pack up but he would not put a time scale to this.

I found this to be a big operation - much bigger than I expected, and it took me about a month to get over.

Under general anesthetic you are operated on in an operating theatre. All the fluid is drained from the plura and talc is introduced to irritate the lung wall and chest lining causing them to fuse together.

The operation took around 1 hour. When I came to, I had two drains coming out of my chest as I had a lot of bleeding. After a couple of days the drains were removed, and I left hospital after a week. I remember one of the worse pains was in my right arm, and it was very painful to move my right arm for weeks afterwards. My oxygen saturation levels also dropped by a couple of points after pleurodsis.

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After about one month I managed to return to work, and I was mobile again, although with limitations. My ambulatory oxygen was increased from 2 to 4 litres per minute to keep above 90% when I walked. All seemed ok I thought: as I recover fully my oxygen saturation will improve and life will return to normal for me, and although I'm not very mobile, I still have an acceptable quality of life.

I did manage a good year without any chest infections, I was getting very tired at the end of a day's work but all in all it was a good year. I managed a holiday and thought, yes, I can live life like this.

The first signs that things were going wrong was fluid building up in my feet (edema) which would swell so much that I could not put shoes on. It got so painful at times that I couldn't even wiggle my toes. I would finish work, take my shoes off lay flat with my legs Elevated. In the middle of the night I would get the worse cramp - it really was an awful time. I had to stop working again, it reached a point were crossing the road was a challenge some days getting out of bed. My walking was down to about 100 yards on 4 litres of oxygen per minute.

Things were going wrong fast. My consultant then told me that my lung function had started to drop again and the only option was a lung transplant. I did the transplant assessment and, sure enough, due to the pleurodsis I would only be considered for a single lung transplant. After two false alarms and 80 days on the transplant waiting list I received the gift of life from my wonderful donor and I feel good.

Overview

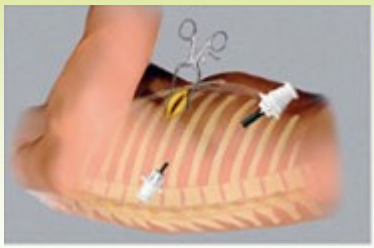
I have changed my mind from thinking, 'I made the wrong choice three years ago' to 'Maybe I got it right'. As I lie in my hospital bed 15 days post transplant feeling fantastic with my future ahead of me,

I must believe I made the right choice but only time can tell. If I have chronic rejection early, I clearly got it wrong. If in five years all is going well, then I was right. I gained one good year and put my body through a lot to get it. I also delayed the transplant for three years - do I take those years into account? One of the best things I did was to go on long - term antibiotics, this did keep me infection free. I most probably left it too late to be assessed for transplantation and let myself get too ill and gambled with my life.

I was lucky to get the transplant so fast, thanks to the wonderful Papworth Hospital for pulling out all the stops. People should consider whether they will be as lucky as me you - might be on the waiting list a long time with no quality of life or you may die whilst waiting for an organ. It must be each individual's choice - my advice is to be guided by your body and to listen to your family. I am guilty of believing I'm healthier than I actually was, but don't we all? Overall, I believe I was lucky and I got it right but only time will tell.

What is VATS (Video Assisted Thoracic Surgery)?

VATS is 'keyhole surgery' carried out in the chest and is a procedure where investigations can be performed to identify, diagnose and treat many types of chest and lung problems.

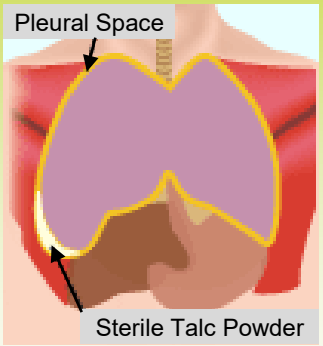


The procedure is performed under a general anaesthetic where several (typically 1 to 3) small incisions are made between the ribs on the relevant side of the chest. One incision is for the insertion of a small camera into the chest and the remaining incisions for the instruments to enable the surgeon to take biopsies, remove fluid or remove any abnormal tissue.

What is Pleurodesis

Pleurodesis is a procedure that causes the membranes around the lungs to stick together and prevents the buildup of fluid in the space between the membranes (pleural space). Pleurodesis is done in cases of severe recurrent pleural effusions (outpourings of fluid around the lungs) to prevent the reaccumulation of the fluid.

During pleurodesis, an irritant is instilled inside the pleural space in order to create inflammation that tacks the two pleura together.



This procedure thereby permanently obliterates the space between the pleura and prevents the re-accumulation of fluid.

Fundraising and Awareness

A Big Thank You to everyone involved in fundraising activities and donations for our group. Through your kind support 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. Growing a social network for patients, by 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.

Jim & Emma, Kate & Annabelle Hunt - Summer Fun Day, High Wycombe, Jul 2014

Michaelah Telfer - Total Warrior Challenge, Lake District, Aug 2014

George Henderson - Hell on Humber Challenge, Aug 2014

Karen Skalvoll - KK Mila 10K Run, Oslo, Sep 2014

Alice Langley - Static Line Parachute Jump, Sep 2014

Fay Whittaker - Dunfermline Round Table Beer/Fest, Sep 2014

Continued from Page 24

Unipart Rail - Dress Down Day, Cheshire, Sep 2014

Leo Stockley - Banger Rally, Cornwall/Croatia, Oct 2014

Charlotte Quantick—Great West Run, Exeter, Oct 2014

Tracy & Eric Barnett - Stroud Half Marathon, Oct 2014

Fay & Mike Whittaker - Fife Arms Fundraiser, Scotland, Nov 2014

Tina Cooke - 60th Birthday Fundraiser, Cheshire, Dec 2014

Christine Sylvester - Christmas Concert, Dec 2014

Walk-A-thon Fundraiser

Laura, Vicky & Stuart Heavens - 13 Mile Walk, Mar 2015

Emma Hunt - Silverstone Half Marathon, Mar 2015

Continued from Page 25

Karen Skalvoll - Tough Mudder, London, May 2015

Maria Reid & Erin Coad - Zip Wire, Cornwall, Jun 2015

Helena Pangalos, Tanya Jones & Team - Grand Union Challenge, London, Jun 15

Donations

Mick & Diane Stobart	Donation
Claire Adam	Donation in memory of her Mother-in-Law who sadly passed away in July 2014
Janice Fraser	Donation in memory of her husband Donald who sadly passed away in July 2014
L Burgess	Donation in memory of his wife Sheila who sadly passed away in July 2014
Nicola Stewart	Donation in memory of her Mother who sadly passed away in August 2014
Brian Wray	Donation in memory of his wife Margaret who sadly passed away in September 2014
Keegan Wilson	Donation in memory of his Mother Pamela Oliver who sadly passed away in September 2014
Fairview Homes	Donation - Stephen Brown (Farview Homes) on behalf of Danny Chappel's Alpha-1 Daughter Poppy October 2014
Pat Barriball	Donation in memory of her Husband Ivor who sadly passed away in October2014

Continued from Page 26

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.

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 govern-ment on your behalf 25p for every £1 donated, boosting the value of the donation by a quarter.

easyfundraising.org.uk

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

How does it work?

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 easyfundraising.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

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?

The search engines easysearch work with generate revenues from advertising goods and services. They receive a percentage of this revenue and pass on a large portion to the cause for which you are searching and supporting. The final amount per search can vary, however generally works out around 0.5p. The average user generates approximately £20 per year for their cause.

Please visit our Website for more information: -

www.alpha1.org.uk

Alpha-1 UK Support Group Merchandise

On our website we have a selection of Alpha-1 merchandise available including T-Shirts, Wristbands, Pedometers, Trolley Key Rings, Badges, Mouse Mats, Shopping Bags and Christmas Cards.

Alpha-1 Information Packs, Booklets and Posters are also available at no cost to you please e-mail us with your full name and address to:

Info@alpha1.org.uk

Healthcare Professionals welcome

Pulmonary Function Tests Alpha-1 Patients

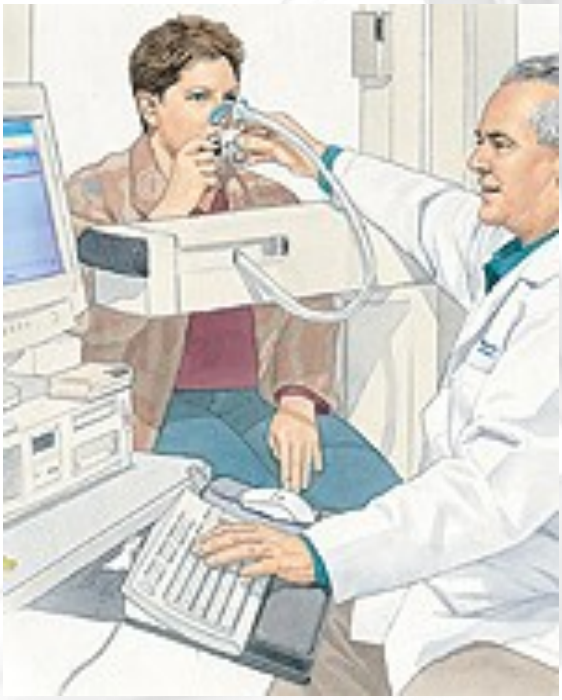
Courtesy of the ADAPT team

"Blow, blow, blow, just a little bit longer and a big deep breath in" are all phrases commonly heard on the entering the Lung Investigation Unit Laboratory at the University Hospital Birmingham. But what exactly is all this huffing puffing all about?

Well, here we enter the wonderful world of breathing tests (or torture as some patients refer to it as), which for ADAPT patients essentially involves four different types and about an hour or two of time.

Upon your first visit to the laboratory as part of the ADAPT project is the longest can with tests being performed without any medication in your system and then repeated following administration of bronchodilator medication which opens up the airways within the lung so that we measure the optimal lung function. On subsequent visits such medications are giving first and all tests are performed when this has taken effect in order for a true comparison of lung function between visits to be made without any day to day variability or any external factors affecting the results.

These tests usually start off gently (honestly) with some normal breathing, a steady blow out, a full breath in and a steady full blow out. This test is measuring the maximum amount of air in litres that can be inhaled or exhaled from the lungs and is known as relaxed vital capacities. This helps determine how big your lungs are by the combining the results with another test performed later on (lung volume test).



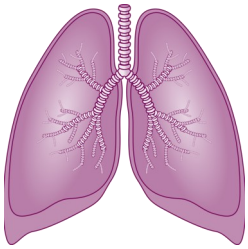
The tests are repeated until consistent results are obtained which adhere to specific acceptability criteria, believe us the physiologists are not so cruel as to make you repeat them over and over again there is always a reason!

Then onto something a little bit more fun/strenuous (don't quote me on that).

The next test is called spirometry and involves normal breathing, a full deep breath in and a fast and hard blow out for as long as you can, ideally until you are empty or pass out, which ever one comes first (only joking). This allows us to measure the volume of air which can be expired in the first second of a forced manoeuvre (FEV1), and the amount air expired overall (FVC) and the speed of which the air comes out (Peak Expiratory Flow).

This test measures how open your airways are by calculating how much air you blow out in 1 second divided into how much air you can blow out all together. When the airways are narrowed as happens in Alpha-1 Antitrypsin deficiency (A1ATD), it takes longer to empty the lungs leading to a reduced measurement.

Now on to lung volumes, an altogether more gentle affair (no really it is!).



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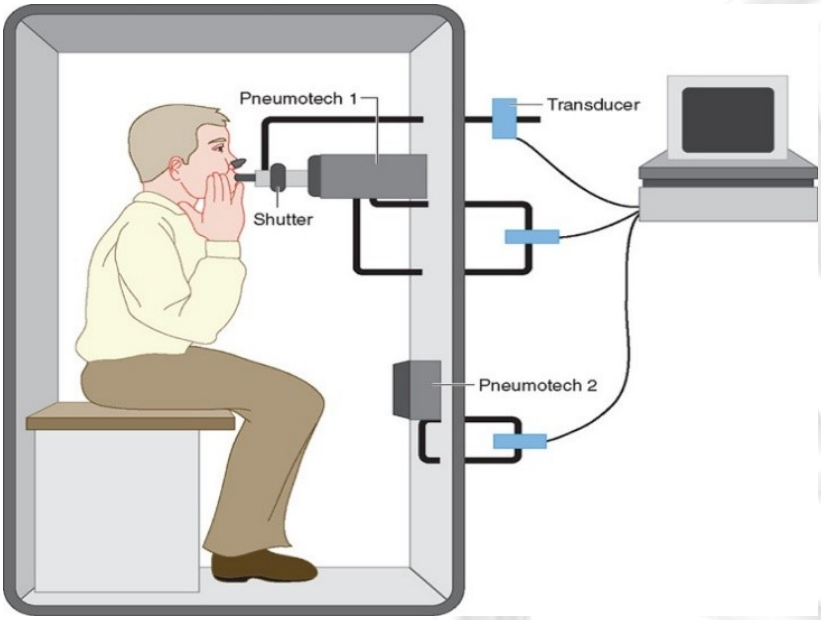
This allows the estimation of total lung capacity as air always remains in the lung even when we have maximally expired in order to prevent lung collapse.

This test involves steadily breathing a specific gas mixture through a mouthpiece for around 2-4 minutes and maximally breathing in and out a couple of times. By comparing the concentrations of gases in the mix before you start breathing and those at the end of the test and with a weird and wonderful calculation it is determined how big your lungs are. In A1ATD lung volumes are normally raised and this test can indicate whether there are areas of the lung which aren't ventilated correctly, where gas is trapped in bullae and if the lungs themselves are bigger than normal.

Onto the home straight now with what is known as the gas transfer test. This measures the ability of a person to transfer oxygen from the air into the lungs and then into the blood. This is a short test that involves normal breathing, a breath out until empty, a full breath in then a breath hold, finished with a blow out at the end. In A1ATD, this will show a reduced measurement due to the destruction of the tissue in the lungs that takes part in the oxygen transfer process.

The final test (at which patients breath a sigh of relief when the word 'final' is spoken) involves a technique known as body plethysmography (eh?!) commonly referred to by the physiologists here as 'body box' a term known to strike fear into the hearts of many a patient. We're sure some ADAPT patients remember the era of the old body box often described as doctor who's tardis which remains in the department but is soon to disappear – possibly to make an appearance on BBC1!!!



The newer plethysmograph is a clear Perspex box which it is commonly said to smell of furniture polish. This test requires normal breathing mostly, with five seconds of breathing against a closed shutter, a full breath in and steady blow out until empty. This test also measures the size of the lungs and is commonly used in A1ATD as it gives a more accurate reading of lung size. The result from this test is compared to the earlier lung volume test to give additional information about how well the lungs are ventilated i.e. how well air moves in and out of them. In A1ATD, there may be areas where air is trapped inside the lungs and this cannot be measured completely by the first test (bullae).

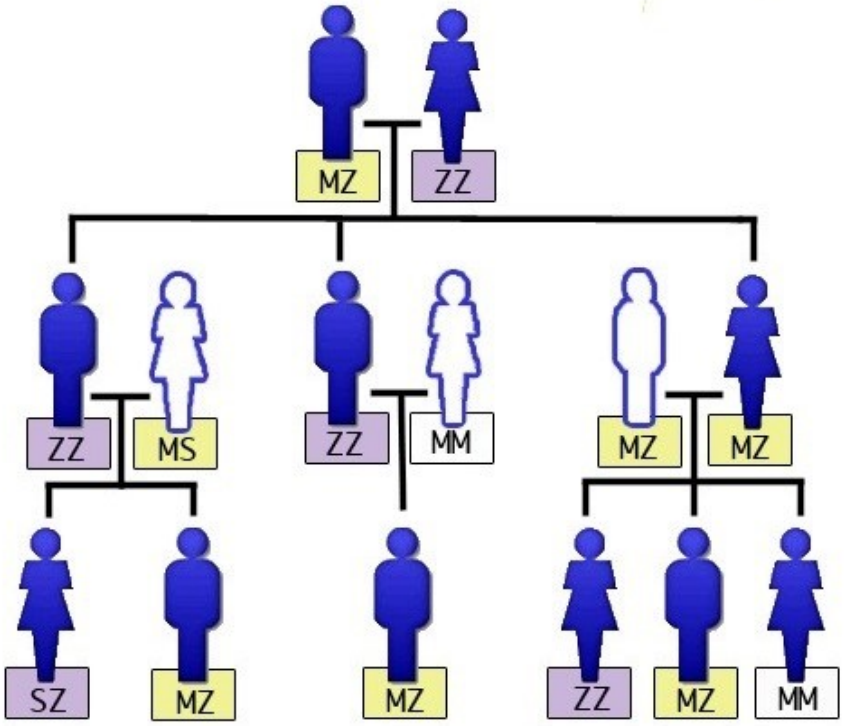
During all the tests, the respiratory physiologist (the person who says blow, blow, blow) will coach you through with words of encouragement with the aim of achieving consistent test measurements and pair of rosy well worked cheeks at the end. These measurements allow the ADAPT team to monitor your condition from year to year.

ADAPT Project
Lung Function and Sleep Dept.
Office 4, Outpatients,
Ground Floor
Queen Elizabeth Hospital
Mindelsohn Way
Edgbaston
Birmingham.
B15 2WB

Tel no: 0121 371 3885

ADAPT

Alpha-1 Antitrypsin Deficiency Inheritance



Affected by Alpha-1	Carrier of Alpha-1 Gene	No Alpha-1

How Will It Affect My Family?

If you have been diagnosed with one of the most severe genetic combinations, it does mean that your children will have inherited at least one faulty gene, because that is all you have to pass on. Unless your partner also has a faulty gene, your children will be carriers i.e. they will have one normal gene and one faulty gene. This is why many partners have blood tests themselves, to rule out a second faulty gene. So, if you are ZZ or other serious combination, you will automatically pass on one faulty gene. If your partner is also ZZ the children will be ZZ.

If your partner has just one faulty gene e.g. MZ, then the children could be either ZZ or MZ depending on which gene they have inherited from them. If you are an MZ carrier and your partner is the same, then the children could be ZZ, if they are unfortunate enough to inherit both faulty genes. Alternatively, they could be MZ (or ZM, basically the same). On the other hand, they could be lucky and inherit both normal genes (MM).

It therefore follows that an MZ carrier partnered with someone who has normal MM genes, would have children who are either MZ carriers or completely normal, with MM genes.

Carriers of A1AD have less AAT than “normal” people but they do usually have enough in their bloodstream to prevent serious problems. It isn’t really understood why but all Alpha-1 patients, including some carriers, do seem to be more susceptible to picking up colds and ‘flu, resulting in secondary chest infections. Therefore, it is just as important for carriers to look after themselves and it would be sensible to avoid smoking, and drinking to excess.

Trustees, Committee, Patrons and Supporters

Trustees and Committee

John Mugford
Chairman
Trustee

Karen North
Vice Chairman
Trustee
Treasurer

Linda Cooke
Trustee
Liaison Officer

Bev Burroughs
Trustee
Admin Support

Dr Sandra Nestler-Parr
Trustee
Strategy and External Relations

Andrew Willis
Trustee
Admin Support

Sioned Lewis
Admin Support

Jemma Coad
Fundraising Awareness Co-ordinator
Parent Support

Charlotte Goode
Fundraising Awareness Co-ordinator

Mel Brolly
Fundraising Awareness Co-ordinator

Fay Whittaker
Fundraising Awareness Co-ordinator

Announcements

We are pleased to announce that Dr Sandra Nestler-Parr and Andrew Willis have accepted the position of Trustee of our Charity. We very much look forward to working with Sandra and Andrew who will be a valuable asset to our charity.

Our Patrons

Robert A. Stockley MD, D.Sc ., F.R.C.P., F.E.R.S.

Professor in Medicine and Chair of the ERS Strategy on AATD

Professor William MacNee MBChB MD FRCP(G) FRCP

Professor of Respiratory and Environmental Medicine at the University of Edinburgh and Honorary Consultant Physician at NHS Lothian Scotland.

Our Supporters

ADAPT

Alpha 1 Alliance

The InterNet Factory

British Liver Trust

Alpha-1 Canada

CSL Behring
Biotherapies for Life™

Almirall

Alpha-1 Foundation
Research for a Cure

Boehringer Ingelheim

KAMADA
High Quality Pharmaceuticals

British Lung Foundation

Chiesi

Children's Liver Disease Foundation
fighting childhood liver disease

In memory of the Alpha friends we have lost, they have all 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

Pauline Hammons
18/12/2014

Thomas Calvin
09/02/2015

Pauline Bradford
02/03/2015

Christine Torrance
07/04/1946 - 11/06/2015

Christine (Chris) passed away, peacefully at home in York aged 69 after courageously fighting a long term illness caused by the genetic condition Alpha1 Antitrypsin Deficiency. Chris is survived by two adult sons Stuart and Michael, Chris's third son Grahame was tragically killed in a road accident. Chris drew some comfort in her grief from knowing that several of Grahame's organs were able to be used for transplant to save other people's lives.

Chris had been a member of the Alpha-1 UK Support Group for over 14 years and at one time took ownership of the group as well as being the treasurer, committee member and trustee. As Chris's health declined she sadly had to hand over ownership but remained treasurer until just recently.

Chris was a very talented lady, very artistic and she designed and produced the group informative leaflets. She was a great artist and loved to paint and produced some lovely pieces. One of which was auctioned to raise money for the support group.
Chris could turn her hand to most things, she loved to decorate her home, she loved gardening, cooking and made wonderful celebration cakes. One of Chris's big passions was tennis and everyone knew that when it was Wimbledon fortnight it was lock down in the Torrance household, Chris loved to watch it and considered those two weeks her holiday .

Chris was a no nonsense northern lass who didn't like a fuss but we hope she will indulge us while we make one last fuss to say how much she was loved by all who knew her and how very much we will miss her.

The world may change from year to year, And friends from day to day, But sweet memories of you Will never fade away.

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

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.

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