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Newsletter Alpha1Awareness

CELEBRATING 50 YEARS OF ALPHA 1

1963–2013

A Word from the Chairman

For the Information Day on the 11th May 2013 entitled 50 years of Alpha-1 we have been fortunate in the speakers that have agreed to come and describe their work. There will be short talks by Professors Sabina Janciauskiene, Robert Stockley and David Lomas. Doctor Ravi Mahadeva will talk about our efforts to persuade the DoH, the NHS and the wider medical community for a specialised service for alpha-1. Doctor Tamir Rashid (a member of Prof Lomas' research team) will describe his ground-breaking results in correcting the genetic mutation by using stem cells. This was widely reported in October 2011 and formed the theme of our meeting last year in Bristol.
Away from the medical subjects we shall have a talk by Monica Fletcher who is the chair of the European Lung Foundation which is based in Sheffield, on her work with respiratory patients and the educational programme being developed. John Walsh, co-founder and CEO of the influential Alpha-1 Foundation, is making the journey from America to talk about patient empowerment.
We are lucky to have such an array of eminent speakers but let us not forget that the day is essentially for Alphas and families. Meeting other Alphas and talking about personal experiences is just as important as learning a little about advances both in medical science and in healthcare.

Here is reminder about the e-petition for a nationally commissioned specialised for alpha-1 antitrypsin deficiency. The petition has now attracted almost 800 votes but we need many more to be able to show that Alpha-1is a concern for the general public. Ask to your friends, neighbours and work colleagues to sign the petition – it does not take long and the benefits could be enormous!

Alan Heywood-Jone

50 Years of Alpha-1

In 1963 a paper written by the Swedish researchers Carl-Bertil Laurell and Sten Eriksson was published in the Scandinavian Journal of Clinical and Laboratory Investigation. They described the first emphysemic patients with what was to become known as alpha-1 antitrypsin deficiency. Now, 50 years on, we can look back and see the marvellous science that this discovery initiated. It is also a time to look forward to better treatments for alpha-1 deficient patients.

The 50th anniversary year is also the year for each European Union member state to publish its national plan or strategy on rare diseases. The Department of Health published for consultation the UK's first-ever action plan to enable earlier diagnosis and better care for those with rare conditions. While many such plans may be short on detail for some rare diseases they promise an era of better treatment for alpha-1 deficient patients.

In the meantime the entire Alpha community is marking the Jubilee Year with events celebrating the work of researchers and clinicians and looking forward to a future of better healthcare for alpha patients. In April there will be two overlapping events in Barcelona, Spain; the *Fourth International Alpha-1 Patient Congress* and an *International Research Conference on Alpha-1 Antitrypsin*.

Here in the UK we are marking the jubilee year by holding a *50 Years Information Day* in Bristol, England. Alpha patients and their families will be able to learn more about the ongoing work in fighting this devastating disease. (see later in the Newsletter for further details of this event)

Alpha-1 is a genetic condition. It is a rare genetic condition. It can lead to emphysema, liver cirrhosis, panniculitis and vasculitis. There is some evidence that it may be a contributory factor in other more common diseases such as diabetes, asthma and glomerulonephritis. In other words it spans many separate medical specialised subject areas. If alpha patients are rare then clinicians with the expertise to understand all these conditions are even rarer.

In the past, a patient with a diagnosed condition was given a standard treatment: the 'one pill fits all' approach. As we move into an age of in which healthcare is tailored to the individual patient we shall need more experts. These experts will need to understand vast amounts of new knowledge coming from the research laboratories and doctors 'working at the bedside'. They will have to balance the narrowness of medical speciality with a holistic approach to each patient. In practice this will mean that medical care will be a team effort. The much talked about Centres of Expertise are expected to follow this model and be multidisciplinary in diagnosis, treatment and monitoring of patient care.

Medical training universities and learned societies should be at the forefront of these changes. Organisations such as the British Thoracic Society and the European Respiratory Society can use their educational programmes and ability to influence change to produce the next generation of experts sensitive to the increasing complexity of medical treatments and the more demanding needs of patients.

It would be wrong to see medical care as simply an issue between doctor and patient. Society at large has a say in public health, research and treatment and the

resources that we devote to these areas. Public policy has slowly shifted from narrow party politics to European consensus on how to provide a high standard of care equal across all our countries. Patient organisations have been at the forefront in this gentle revolution. They are no longer content to accept wisdom from above – they wish to make a full contribution to the decision making process that vitally affects their members.

Alan Heywood-Jones

For the events in Barcelona: http://alpha-1barcelona2013.org/
For the event in Bristol: http://www.alpha1awareness.org.uk/
For the 50th Year e-visitors book: http://50yearsofalpha1.org/

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NEWS IN BRIEF

There has been a lot of discussion going on within Europe as well as here in the UK about Alpha 1 and how sufferers are treated, as you will see in the two following articles

The Wider Picture

There are obvious inequalities between the member states of the European Union when it comes to the diagnosis, treatment and care of Alpha-1 patients. The European Federation of Alpha patient organisations has been pressing for action by the European Commission and the European Parliament to redress this imbalance.

One step along this road is the publication of the ***Alpha-1 in the European Union - Expert Recommendations***. The objective of the report is to identify the issues facing Alpha-1 patients and give concrete recommendations that will help the European Institutions and Member States to choose the best way forward.

The working party that prepared the report was called by Christopher Fjellner MEP (Sweden) and chaired by his colleague Carl Schlyter MEP (Sweden). It was my job as president of Alpha Europe Federation to explain, very briefly, the structure and aims of the organisation and also to introduced the speakers.

Robert Stockley is a Professor of Medicine at the University of Birmingham, Director of the ADAPT programme and co-founder and current chairman of AIR (Alpha-1 International Registry). He briefly touched on the history of the discovery of AATD before talking about our growing knowledge of the pathologies that stem from the genetic condition, Augmentation therapy was mentioned as was the difficulty of using standard double-blind powered trials to collect evidence for its effectiveness. Professor Stockley then referred to recent trials using CT scans to measure lung density and how these provide evidence in favour of augmentation therapy.

Next was John Walsh. John is co-founder, president and CEO of the Alpha-1 Foundation. (The Foundation is the largest funder of Alpha-1 research in the world.) He is a former chairman of the National Health Council (USA). He is on the Director's Council of the FDA (Food and Drug Administration in USA) and he has been awarded the FDA's Commissioners Special Citation for accelerating orphan drug therapies.

The presentation went through the stages of the growth of the foundation – starting from a group of patients on some of the early trials to an effective lobbying organisation in Congress.

Next we heard from two Italians. Mario Ciuffini is vice-president of the Italian national association of Alpha-1 patients. He is a member of GARD (Global Alliance for Rare Diseases) Europe and a member of the Italian Commission for the Early Diagnosis of Rare Diseases. Mario is a film-maker and he spoke with passion on how his Alpha-1 emphysema affected his working-life and his family. This was followed by Maurizio Luisetti, Professor of Respiratory Medicine at the University of Pavia, co-ordinator of the Italian Alpha-1 registry and Director of the Centre for the Diagnosis of Alpha-1. The professor spoke briefly about the structure health care system in Italy as it affected Alphas.

Carl Schlyter invited the attendees to an open discussion points from which went into the report.

The printed report is being distributed to European Members of Parliament from the office of Mr Fjellner.

Alan Heywood-Jones

Extract relevant to the UK

> Alpha-1 patients struggle seven years on average before receiving an accurate diagnosis, as Alpha-1 is often misdiagnosed as "regular" smoking-induced chronic obstructive pulmonary disease (COPD) or asthma. In addition, several Member States do not recognise the specificity of Alpha-1 and bundle it together with regular COPD, without recognising its specificities, genetic origin and the fact that it is caused by the lack of a natural protective protein as is also the case in other rare diseases, such as Fabry's disease.
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> A case in point is the United Kingdom, where the disease is not widely recognised as a rare disease even though the country is home to the biggest physician-led patients' registry in Europe. NICE, the National Institute for Health and Clinical Excellence, does not differentiate between Alpha-1 and smoking-induced COPD or asthma. As a result, patients are not granted access to optimum treatment and often receive minimal information about their condition and how to prevent its life-threatening consequences. Poor awareness results in late diagnosis or no diagnosis at all, which far too often leads to irreversible, serious lung damage.

Extract – some of the specific recommendations

- Member States and the EU must ensure appropriate recognition of Alpha-1 as a rare condition and Alpha-1 related emphysema as a specific ultra-rare disease.

- Member States must raise awareness of Alpha-1 in the medical community and the general public in order to ensure a timely diagnosis that will increase the chances of preventing irreversible tissue damage.
- Member States must prevent and put an end to health inequalities affecting patients suffering from Alpha-1 and other rare diseases.

Recommendations Paper - Executive Summary.

1. Member States and the EU must ensure appropriate recognition of Alpha-1 as a rare condition and Alpha-1 related emphysema as a specific ultra-rare disease.
2. Member States must raise awareness of Alpha-1 in the medical community and the general public in order to ensure a timely and fast diagnosis that will increase the chances of preventing irreversible tissue damage.
3. Member States must prevent and put an end to health inequalities affecting patients suffering from Alpha-1 and other rare diseases.
4. The EU must ensure that all Member States respect the EU definition of rare diseases.
5. Future EU and national policies with a relevance to rare and ultra-rare diseases should respect the spirit and the letter of existing EU policies addressing these issues.
6. EU Member States should ensure that policies and legislation in the field of rare diseases are not jeopardised by cost containment measures. Such measures should not have a negative impact on areas where long-term investments are needed to make a difference, such as public health.
7. Each EU Member State should develop and implement ambitious national plans or strategies on rare diseases, as recommended by the Council of the European Union's recommendations on action in the field of rare diseases.
8. Member States should ensure that Alpha-1 patients can access the treatments they need, notably when implementing the Cross-Border Healthcare Directive.
9. The EU should work towards better standardisation of treatments and devices supporting breathing to ensure that patients can enjoy their freedom of circulation.
10. The EU should develop an ambitious strategy on information to patients so that all patients can make informed choices about their treatment options.
11. Member States should ensure that the optimal guidelines for the treatment of Alpha-1 are implemented in order to reduce the need for lung transplants and thereby contribute to increasing the availability of lungs for transplantation.
12. Patients should be given the possibility to decide with their physician whether and when they should undergo organ transplantation.
13. Member States, national HTA* experts and policy makers must acknowledge the reality of clinical research on therapies for rare and ultra-rare conditions and accept alternative evidence validated by experts. Gold

standard randomised, placebo-controlled and double-blind clinical trials with a sufficient number of patients are impossible to conduct and unethical. Physicians treating Alpha-1 patients should be asked about effectiveness when a therapy is being assessed.

14. The EU and Member States must provide support to Alpha-1 expert groups, including academic and patients' groups, in order to pool expertise and build on it.
15. The EU and Member States should support the creation and the management of Alpha-1 patient registries and seek the advice of Alpha-1 experts who are already running them.

* HTA Health Technology Assessment produces research information about the effectiveness, costs and broader impact of healthcare treatments and tests for those who plan, provide or receive care in the NHS. HTA is used by the Department of Health and NICE.

Photograph of some of the members of the working party. From left to right: Giovanni Asta, Prof Jacques Hutsebaut, Alan Heywood-Jones, Mario Ciuffini, Bo Jonas Sigedal (assistant to Mr Fjellner), John Walsh, Carl Schlyter, Prof Rob Stockley, Prof Maurizio Luisetti.

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Patient Organisation focus: Alpha Europe Federation

The following item has appeared in the online Newsletter of the European Lung Foundation it is an interview with our Chairman, Alan Heywood-Jones, in his role as President of the Alpha European Federation

The Alpha Europe Federation aims to provide a network for patient organisations in Europe who work together to improve understanding of Alpha-1. This year, the organisation will be marking the 50th anniversary of the discovery of alpha-1 antitrypsin deficiency.

In the interview, Chairman of the federation, Alan Heywood-Jones, tells us how he became involved in the organisation, what he is most proud of and the changes he'd like to see in the management of Alpha-1 in the future.

Can you tell us a bit about the Alpha

Europe Federation?

The Alpha Europe Federation started in 2004 when patient organisations across Europe joined together to work collaboratively and share information. The Federation now acts to support individual members and to lobby as one voice at a European level.

How did you become involved in the organisation?

I became aware of alpha-1 after my wife, Dawn, was diagnosed with the condition. We had moved to Spain in 2000 to help Dawn's health but we weren't aware at this time that she had alpha-1. It wasn't until a doctor in Spain diagnosed the condition that we knew what we were dealing with. After this, we registered the charity, Alpha-1 Awareness UK in 2008 and 2009, I was elected President of the Alpha Europe Federation.

What are your big milestones this year?

This year, Alpha-1 is celebrating its 50th anniversary and our activities will focus on raising the profile of the condition across Europe.

How will you be marking the 50th anniversary of the discovery of alpha-1?

In 1963, a paper published in the Scandinavian Journal of Clinical and Laboratory Investigation described the first emphysemic patients, with what was to become known as alpha-1 antitrypsin deficiency.

A key part of the year's events will be two overlapping events in April in Barcelona, Spain; the Fourth International Alpha-1 Patient Congress and an International Research Conference on Alpha-1 Antitrypsin. Representatives from each patient organisation in Europe will attend the Congress.

What are your key challenges?
The two biggest challenges we face with alpha-1 are discrepancies with treatments between countries and the need for early diagnosis.

There are varying standards across Europe for treating alpha-1. Augmentation therapy is the key treatment to help prevent and slow the progression of the disease. The treatment is available in the USA and some countries in Europe, but it is not uniformly licenced as it is an expensive way to treat the condition.

The second challenge is the need for primary care doctors to be aware of the condition and carry out the appropriate tests to diagnose the condition. It is thought that only 10% of people who actually live with alpha-1 are diagnosed with the condition. As with other rare diseases, identification of the condition is a key challenge.

What changes would you like to see in the future?

As we address the above challenges, we would like to see a gold standard of treatment across Europe and improved diagnosis of the condition.

Additionally, in the long term, we hope to find a cure for alpha-1. There are several research teams taking steps towards genetic correction of the condition and we hope that this

research will ultimately discover a cure for alpha-1.

What are you most proud of in your organisation?

I am most proud of the publication, Alpha-1 in the European Union Expert Recommendations. The report provided a common understanding of Alpha-1 as a rare disease. It was initiated and chaired by Members of the European Parliament and was a significant milestone in raising the profile of
alpha-1.

Is there anything else you'd like to share with our readers?

The 50th anniversary year is also the year for each European Union member state to publish its national plan or strategy on rare diseases. While many such plans may be short on detail for many rare diseases they promise an era of better treatment for alpha-1 deficient patients.

Alan Heywood-Jones is the chairman of the Alpha Europe Federation. The Federation consists of Alpha patients' associations from thirteen European countries. The aim is to provide a European information and resources network for patient support groups and linked associations, health professionals, institutions and industry who wish to improve and extend their knowledge of alpha-1 antitrypsin deficiency. Alan is also the chairman of the charity Alpha-1 Awareness UK.

50 Years of Alpha 1- come and celebrate with us

We would love you to join us, to help celebrate Alpha 1's 50th birthday, at a Celebration Information Day on Saturday 11th May, at the Aztec Hotel Bristol, our speakers will include our patrons Professors Sabina Janciauskiene and David Lomas, also Professor Robert Stockley, from ADAPT, Doctor Ravi Mahadeva from Cambridge and the Alpha 1 Alliance UK, and another good friend of Alpha 1 Awareness UK, from the Alpha 1 Foundation in USA, it's founder and CEO John Walsh. Other speakers will be Monica Fletcher from the European Lung Foundation and Dr Tamir Rashid, whose research work with Prof Lomas, lead to the ground-breaking results in correcting the genetic mutation by using stem cells and of course you will have the opportunity to meet up with friends old and new, we all know how much Alphas love a chance to get together and chat! It will I am sure be a great day and we would love to see as many of you as possible.
Following the success last year we are again offering a creche facility for children aged 0- 11 years, I have contacted the same agency we employed last year so we know the children will have a great time and be well looked after.
There is no charge for the day and you will be able to claim a contribution to your travel costs. Included in the day will be coffee on arrival, buffet lunch and tea at the end of the afternoon.
A booking form, details of the hotel and directions are included with this Newsletter, it would be useful for us to have your forms back as soon as possible but certainly no later than 11th April, **however if you are going to be**

using the creche facility, please return you form to me by 10th March as I need to confirm with the agency how many children there will be in order that they can organise the correct number of staff for the children.

Congratulations

Congratulations to our Patron Prof Davis Lomas who took up a new post as Dean of the Faculty of Medical Sciences and Chair of Medicine at University College London, on 1st January 2013. He will of course continue with his passion of researching and moving towards a cure for Alpha 1.

Alpha-1 Alliance UK

Work is still on going with the Alpha 1 Alliance UK and it is hoped that we will be able to send you a full report on their most recent meeting, shortly, in the meantime, the online e-petition to commission a Specialised Service for alpha-1 antitrypsin deficiency, is still active, there is 1000 signatures so far, but many more are needed. Go to http://epetitions.direct.gov.uk/petitions/39732 to add your signature to the petition. Your family, friends, neighbours and work colleagues can also sign the petition and by doing so they will be helping you.

Online Forum, Twitter & Facebook

The original Alpha-1 Awareness Web Forum is still online at http://techno.demon.co.uk/a1aforum and the place where you may write freely in the knowledge that the only people who can read what you write are other members of the Alpha-1 Awareness UK charity or approved carers or medical professionals.

Our re-energised Twitter account, which is @alpha1uk (see its website-page at www.twitter.com/alpha1uk), continues to gather followers, it is a way to get word out to people about alpha-1 antitrypsin deficiency, the charity and related health issues. It enables us to keep contact with other groups and professionals and hopefully helps them to support our cause, as well as announcing timely news or providing links back to important features on our main website or Facebook Page.

As well as our main charity website at www.alpha1awareness.org.uk, we introduced you in our last Newsletter to our a new Facebook Page at www.facebook.com/Alpha1AwarenessUK (you'll find quicklinks to this page from the top-right corner of our main website, if that's easier for you). This Facebook Page is the public face of the charity on Facebook, and it contains general news items about Alpha-1, our charity's activities in the UK and those of our members (particularly people who may fundraising for us; the new page links to and promote their various Justgiving or Facebook pages especially), as with our Twitter page we are building followers. This new Facebook Page is not the same as the 'closed' Facebook Group on which you may freely post comments on matters relating to Alpha-1 and how it affects

your lives. You can find this Group on the slightly different link www.facebook.com/groups/Alpha1AwarenessUK (note the addition of the word 'groups' in the middle there). There are links to this Group from our new Facebook Page too, so you should be able to find it easily. The Facebook Group is loosely restricted in terms of access – each new joiner is approved by one of the Charity's administrators, having checked that person's own Facebook page to see whether they have a logical reason to be a part of our Group - but bear in mind there is limited amount of checking we can do on Facebook, if a person is not already a charity-member. There are people on our Facebook Group who are members of other Alpha groups or charities both in the UK and around the world.

Fundraising

10K Run 2013

I am pleased to report that we now have 6 runners signed up to do the British 10K Run on 14th July in London, it is great news that all our allocated places have been fill for this year. I am sorry that we have had to disappoint people who wanted to take part.

If you still want to take part in the run and raise funds for Alpha 1 Awareness UK, you can register as an individual, by going to www.thebritish10klondon.co.uk and raise money in the usual, we will give you running vests sponsor forms etc.

South West Kayak Challenge

The Kayak Challenge is still on course for summer this year and we are posting regular updates on our Facebook page, you can follow the training progress by visiting http://www.southwestkayakcharitychallenge.co.uk/

Corporate Sponsorship

As you are aware from our previous Newsletters, in 2012 we had our first corporate sponsorship, with Proact, this sponsorship was made possible by Alan Wooler (one of our Board members) who works for the company and did some excellent negotiating with the company to achieve this, we were extremely grateful to Alan for securing this for us and for the money that the company raised for our Charity.
2012 was the first year that Proact had taken on sponsorship of Charities (they chose 3 last year) and have decided this year that they will support 2 Charities and the wonderful news for Alpha 1 Awareness UK is one of the 2 Charities they will support during 2013, we are delighted about this and are very, very grateful to Alan for the hard work he has put into securing this for us.

Alan reports that in December a charity ball and auction was held so far we do not know what money Alpha 1 Awareness with receive as Proact are still waiting for one of the auction buyers to pay their money at which point the total will be made fully public, what ever to total sum is, will be split between the 3 Charities Proact supported in 2012. Alan was asked at the Ball to give a short speech about Alpha 1 and Alpha 1

Awareness, as the father of a very young PiZZ sufferer, Alan was able to talk, with passion, experience and knowledge, he was nervous about speaking in public like this, but obviously did an excellent job, as he told us that after his speech there was a great deal of awareness and a number of people approached Alan and his wife Emma after his speech wanting to know more.

To make sure no one forgot who Alpha 1 Awareness are, Alan and Emma were allowed to put one of our brand new lapel badges on ever other napkin on each table. By the end of the evening, only 12 were still remaining of the 52 that were put out and you could see that people were wearing them. Alan has since seen some one wearing the badge on his work suit. Alan was also very pleased when the representative from the Sheffield Children's NHS Foundation Trust (one of the other Charities), who spoke immediately after Alan, supported Alpha 1Awareness as he plugged us during his speech, by telling people to wear our pins and help get our name out there by simply remembering us.

Promotional Merchandise

Along with our t-shirts, polo-shirts and sweatshirts, we now have other items with the Charity name on them, they are
Trolley Coins - £1.50
Lapel Badges - £1
Shopping Bags - £2.50

All profits from the items will go to the Charity

Alan Wooler has put in a lot of work to get these items and is working on other items, which we hope to introduce this year.

Alan is setting up an online shop where you can purchase the goods, details of this shop will be put on our Facebook page, Twitter feed and Forum, as soon as it is set up, details will also appear in our next Newsletter. In the meantime you can obtain the items by emailing me at secretary@alpha1awareness.org.uk or Alan at alanw@alpha1awareness.org.uk

In the t-shirts and polo shirts we have a choice of black or white and the Charity logo is embroidered on them. The sizing is small, medium, large & ex large, and the cost is £11 for polo shirt and £8-50 for t-shirt

polo shirts

t-shirts

We do also have sweat shirts available they are the same sizes as the above and cost £12 each


Sweatshirt

We are asked a lot about insurance cover and always recommend that people that you look at www.geneticalliance.org.uk/insurance , if you do not have access to the internet, you can contact : Genetic Alliance UK, Unit 4D, Leroy House, 436 Essex Road, London N1 3 QP and request the leaflet about insurance for people with genetic conditions.

Insurance

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Shop Online and Raise Funds

Don't forget when shopping online to register with **easyfundraising.org.uk** and it won't cost you a penny more to shop and raise funds in this way. In fact you could even SAVE MONEY as many retailers give exclusive discounts, special offers and even 'e-vouchers' when you shop through the easyfundraising site.

Easyfundraising is a shopping directory featuring over 600 trusted online stores, including: Asda, Tesco , Argos, Amazon, the Body Shop, NEXT, Debenhams, John Lewis, Toys'R'Us, HMV, Virgin, iTunes, CD WOW, Marks and Spencer, Currys, Dixons, Staples, PLAY.COM, Pets at Home, Choices Direct, WH Smith, The AA, RAC, Direct Line, Churchill, The Carphone Warehouse, Ticketmaster and over 600 others...

Register for **Alpha 1 Awareness UK** and just by doing your shopping online you will be helping raise funds for the Charity.

Also you can set up www.easysearch.org.uk, as your search engine page and register Alpha 1 Awareness as your chosen cause then every time you search a webpage you raise money for the Charity.

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