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Newsletter Alpha1 Awareness

A Word from the Chairman

Last year Alpha 1 Awareness hosted the Fifth European Alpha1 Congress in London. Apart from the opportunity to meet and talk to other Alphas many of you appreciated the quality of the presentations made by the expert medical speakers. On a smaller scale we held a lunch for Alphas in Newcastle. It was delightful to see so many children at this event and we now need to replenish our stocks of Alphie Teddy Bears.

If 2010 was a year of achievement and consolidation, then 2011 will be a year of change and challenge. All charities are finding it difficult in the current economic climate to raise funds to support their work. We are fortunate in having members willing to go to extraordinary lengths to raise money. This Spring we have one member running in the London Marathon and another entering a Kayak and Cycle race in Devon. We wish them great success in these events.

To help Alphas and their families we are continuing our programme of regional events. Liz McKenna and Dawn are completing the plans to hold an Information Day in Scotland in early Summer. More lunches are planned for North Wales and Central/East England.

We recognise that we need to engage more with the caring medical professions to inform them about alpha1 antitrypsin deficiency and its causes, cures and diagnosis. For this reason we will continue to support the FIND-A1 project. By now you should have received a number of leaflets explaining this project. I hope that you will find time to pass these on to your GP and local hospital.

Alan Heywood-Jones

The more it changes the more it is the same

Last July, the Government set out its ambitious plans for the reform of the NHS in the White Paper *Equity and excellence: Liberating the NHS*. It was claimed that the plans had a simple aim: "to deliver health outcomes for patients which are among the best in the world, harnessing the knowledge, innovation and creativity of patients, communities and frontline staff in order to do so".

There are many changes proposed in the White Paper but most of the Press interest centred on the handing of budgets over to doctors – GP Commissioning. There was widespread scepticism that this hand out of £80 billion was the best way of shaking the NHS up so that it actually suits patients instead of managers. The response from medical professional bodies was more positive.

Between July and October 2010 the Government consulted with health professionals, medical institutions, patients' organisations, etc. on how best to implement the proposed changes. The results of the consultation process are now published along with the government's response.

Many of the criticisms of the details of the plans receive the response 'the Government disagrees' and this is followed by a justification along the lines of 'we have done our homework'. However, the true value of the consultation process is seen in the modifications to the original proposals. Some of these are:

- allow a longer period for completing the reforms
- create a clearer introduction of GP commissioning
- increase transparency in commissioning - GP consortia to have a published constitution
- extend councils' scrutiny powers to cover all NHS-funded services (the original proposal to merge local authorities' scrutiny functions into the health and wellbeing board is now seen to be flawed)
- phase the timetable for giving local authorities responsibility for commissioning NHS complaints advocacy services
- give GP consortia a stronger role in supporting the NHS.

In short, the changes will come slower than promised and there will be more tweaks round the edges. More cynical opinion among policy makers is that this is likely to generate a greater deficit, burning more money, not less. The crisis will grow, and the Government will have to intervene to support GP consortia with health boards or hubs, creating new bureaucracies. How this will affect the post-code lottery is open for debate.

The United Kingdom is in the middle range of expenditure on a health service but, by international standards, patient satisfaction levels are low. More worryingly, clinical outcomes are disappointing across a range of indicators from stroke and cancer care to preventable deaths. Many of us hoped that the position of patients with rare conditions such as alpha1 antitrypsin deficiency would improve when the Government published its National Plan or Strategy for Rare Diseases. The White Paper makes no mention of the Government's commitment to publish such a National Plan by 2013. When it does arrive the National Plan for Rare Diseases will be yet another tweak to modernised NHS – last week Number 10 sent a memo to all departments instructing them to use the word "modernisation" instead of "reform".

National Plan for Rare Diseases

In November Dawn and I attended the Europlan conference in Manchester. Dawn represented the Charity, I was there on behalf of the Alfa Europe Federation. The event was organised by Genetic Alliance UK as part of the Europlan project which aims to assist European Member States develop their National Strategies for Rare Diseases.

Patients with Rare Diseases (RD) have medical needs but often they have consequential social needs. The Nation Plans are supposed to satisfy all the needs arising from Rare Diseases. In those countries where the health needs and the social needs are handled by separate organisations it is possible that RD patients will not receive adequate care. Even in countries with a holistic approach to the patient and his needs there are variations in regional provision of services.

With many points to discuss and a number of stakeholders (patient organisations, doctors and pharmaceutical representatives) the conference was divided into a number of workshops.

One of the issues brought up in the Governance Workshop was the problem of identifying rare diseases. At present most countries use the International Classification of Diseases (ICD 10) or some variation of this. One problem with ICD10 is that many of the rare diseases do not have a code and they are often grouped in with other more common diseases. Alpha1 antitrypsin deficiency is coded E88.0 along with bisalbuminaemia! The result is that it is difficult to compile statistics on the diagnosis and treatment of rare diseases and evaluate how effective the plan is. The new version, ICD11, will allow for rare diseases to be codified and it will be adaptable so that new diseases may be added. Unfortunately, ICD11 will not be available for a few years.

Another issue mentioned in the Patient Empowerment Workshop was the divide between health care and social care. To simplify the position, people in the social care system are ignorant of the rare disease or condition and how it may affect the patient's life or ability to work. Conversely, the medical staff were not trained to advise on all the various forms of help available in the social care system. The hope is that when Centres of Expertise for rare conditions are established they will be multidisciplinary and include social welfare assistance. Inclusion of patient representatives is recognised at the highest political levels but the practical problems of including such people in the planning and operation of health care provision have not been resolved.

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Everyone at Alpha1 Awareness UK is delighted that Alastair Kent, Director of Genetic Alliance UK has been awarded an OBE in the 2011 New Years Honours List. The award is for his work over twenty years in healthcare, helping families affected by genetic conditions.

Alan Heywood-Jones

JP was her husband's carer until his death in 2009, she has kindly agreed to share her story with us, it will, I am sure, make you all stop and think. Thank you to J for her courage to challenge the authorities and for sharing your story with us

A Winter's Tale

This is the story of Edward, my husband, and of his fight against alpha1 antitrypsin deficiency and how he and I fought against incompetence and ignorance within the health service.

Ed was about 35 years old when he began to show signs of untypical breathlessness. This was diagnosed as asthma and he remained under the care of his GP for the next two decades. During this time the bronchial attacks become worse but the GP refused to refer Ed to a consultant. In desperation, I contacted NHS Direct and I was told to ring the hospital and demand an appointment. I did this and managed to arrange an appointment for 48 hours later. The hospital consultant carried out various tests which all came back indicating something else was going on. He said he had suspicions that Ed may have another condition and that he wanted to carry out one more test. When the results came back they showed that his suspicions were correct and that Ed was suffering from alpha1 antitrypsin deficiency.

We were told that the condition was a rare genetic disease that would affect the lungs, liver and kidneys[i] and that the family should be tested. The consultant admitted to a lack of knowledge and this prevented him from giving us more information. The clinical appointments continued but no more information was forthcoming and so I researched the condition myself. I contacted the Alpha 1 Foundation in the USA and they sent me a lot of information about the condition. I took these to the next appointment at which I received assurances that they would be studied. (It has been admitted recently that they were never read.)

In 2001 Ed was referred to the Brompton Hospital for a lung-and-heart transplant but on assessment was deemed not to be a suitable candidate. The medical notes showed that Alpha sufferers tended not to cope well after such surgery[ii]. We were told that Ed would not be called for a further appointment.

In 2002 the consultant retired and Ed's care and treatment was passed to a new consultant, Dr X. This was when the breakdown in communications started to occur. Over the next five years much correspondence went back and forth between the GP and Dr X regarding Ed's treatment and care. Included in these letters were requests from the GP asking the consultant to furnish the family with more information about the condition as he did not have the knowledge to give it himself. In addition, I was also constantly nagging the GP and Dr X for the information.

The letters show that the GP had written several notes to the Dr X requesting that our sons be tested. The tests had been done in 2005 but Dr X failed to notice this and have the results sent to the GP. At my first joint meeting with the Dr X and the Primary Care Trust in February 2010 they confirmed that our sons test results were available and showed them to be MZ carriers they also showed that Ed was PiZZ. The results had been sitting on the hospital computer system for 5 years – no one would admit to having known about them.

From about 2006 Ed's legs started to show signs of what we believed to be Panniculitis. He suffered, swelling, very red (turning black in the later stages) raised nodules leaking an oily substance. The sores were very painful and made it impossible for him to get about. Request after request for a skin biopsy to confirm or rule out the condition again fell upon deaf ears.

After three years and many requests for a diagnosis, a home visit was eventually made by a dermatologist. My comments that Panniculitis can be caused by alpha1 antitrypsin deficiency went unheard or was just ignored. In just four minutes and without a proper examination, the specialist announced "I don't think so" and promptly left.

By this time Ed was completely housebound and on oxygen 24 hours a day and this meant he was not able to eat properly. He was also suffering from severe bladder and digestive problems but were told that this was due to *corparbanally*. Later the hospital stated that this was just another GP mistake for *cor pulmonale*[iii]. During this period I requested that Ed's antibiotic be changed from Amoxicillin to something else as it clearly was not helping. This request was refused.

By now I had found Alpha1 Awareness and my knowledge of the condition was growing. I asked Dr X for a referral to ADAPT so that we could gain more information about the condition in order to make Ed more comfortable. I was told that this was not available: had I known more at the time I would have challenged this.

Ed died in September 2009 at the age of 65.

This is not a story about personal loss so I will continue by telling about my efforts to stop Alphas having to go through the same experiences. The starting point was a meeting between me, my legal representative and the PCT and Dr X. The latter's opening position was "I'm sorry but there was a breakdown in communication". This allowed us to ask where, how and why the breakdown occurred. We did not receive satisfactory answers to these questions even though the medical records clearly showed incompetence or worse. Nor did we receive an answer to why Dr X did not feel that he had a duty to research the condition; a condition about which, by his own admission, he knew little. The meeting closed with Dr X saying that he would get answers and we would reconvene at a later date. This second meeting was held three months later but the message was the same – a breakdown in communication. At a third meeting in late October 2010 I reacted to this line by demanding a meeting with the Chief Executive and Medical Director of the PCT.

The joint meeting was held in January this year. Dr X and the Chief Executive clearly admitted that the breakdown in communications was down to Dr X and that he had failed in his duty as a consultant to provide a reasonable standard of treatment and care to Ed and to his family. They also agreed to put in place improved procedures. For instance, they will test for the deficiency in adults between 30 and 60 who present with unexplained COPD. They guaranteed better education, advice and support for diagnosed Alphas and an option of referral to ADAPT in Birmingham.

Even after Ed's passing COPD is still listed as the main cause of death and alpha1 antitrypsin deficiency as a secondary condition. I am now in the process of challenging this in order to have the death certificate amended, a minor point but one that we as a family feel the need to have put right.

JP

[i] Although AAT is synthesised in small amounts in the kidney there is very little research showing this to be a concern for most Alphas.

[ii] There are no differences in the long-term prognosis according to the individual indications. See Levine, Anzueto, Peters et al., *Medium term functional results of single-lung transplantation for endstage obstructive lung disease* , Am. J. Respir. Crit. Care Med. (1994). In lay terms, Alphas do no worse than others when it comes to lung transplants.

[iii] Cor pulmonale is the enlargement of the right ventricle of the heart as a response to increased resistance as the small blood vessels in the lungs become very stiff and rigid.

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Rob McKenna, joined the Charity back in the summer of 2010, he is on the Transplant list and has agreed to keep us in touch with how things progress for him in his waiting time, here is the second part of Rob's diary.

My Diary *I hope everyone has had a great holiday over the festive season and kept bug free. Me - I am just hoping this year will be the one when I get my transplant as the waiting and the hearing of no news is quite a strain but I must try to keep up beat and as positive as I can. I have not heard any news from the medical team at Freemans Hospital and it is a bit concerning. I thought I would be informed of some sort of progress. It is really strange wanting the transplant operation so much, and yet, at the back of your mind someone else has to loose their life to improve and save my life. I have had my good and bad days and more so because of this bad weather. Liz tries her best to keep me going and it is not easy for both of us. I guess I am fortunate and must always remember at least I have the privilege and knowledge of knowing I am on the transplant list. I just wish that there were less days that I suffer with my breathing. I know the staff at Freemans Hospital are great caring people and anyone going to Freemans Hospital will be very well cared for. If any one else is on the list I would love to compare notes or if any one has questions about assessment I would be happy to help. Until the next news letter stay safe and stay healthy!*

Take care
Rob

NEWS IN BRIEF

Here we are at the beginning of a New Year and all of us at Alpha 1 Awareness UK wish all our members and their families a very Happy and Healthy 2011. We have begun looking forward to 2011, as Alan, our Chairman, has mentioned at the start of the Newsletter, we look forward to meeting up with many of you, at our Information Day in Scotland and at our Informal Lunches, in various parts of the country. When dates are finalised they will be put on to our website and in our next Newsletter.

Also looking ahead to later in the year we have several people taking part in various Marathons and sponsored events to raise money and also raise awareness of Alpha 1.

Kayak & Cycling Chris Brown-Martin and friends are taking part in a kayak and cycle one day event, covering the 30 or so miles, between Hatherleigh, Okehampton and Bideford, this event is planned for around 19th March, originally there were around 8 people taking part, there are now over 20 taking part.
http://www.justgiving.com/Kayak-and-cycle-Event2

London Marathon Emma Dent is taking part as the first ever person to run solely for Alpha 1 Awareness UK, Emma took up the Silver Bond place that we were offered in this year's marathon.

Berlin Marathon Phil Sharpe who raised money for us in the Paris Marathon in 2009, is this year taking part in the Berlin Marathon, for our Charity.

Kilamanjaro Climb Emma Price, whose young sister is an Alpha sufferer, is taking part in the Kilamanjaro climb in June.

To all these brilliant people doing so much to raise money for Alpha 1 Awareness and to raise the profile of Alpha 1, we send our heartfelt thanks, and wish them all very good luck in their events, we look forward to getting photos of them taking part in their events.

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** and just by doing your shopping online you will be helping raise funds for the Charity.
You can also use **easysearch.org.uk** as your search engine, each time you search for a topic, you raise money.

Please share with us any plans you have for raising funds, be it a special birthday or anniversary, a coffee morning with your friends, a Quiz Night, getting the children to do a sponsored silence, we would love to hear. Thank you

Congratulations to Emma and Alan Wooler, parents of Tyler, who by setting up a Justgiving page have raised over £900 for the Charity, we are so grateful to them for all they to raise money and awareness. Thank you so much from everyone at Alpha1AwarenessUK
http://www.justgiving.com/AlanWooler-Alpha1

Membership

Membership of the Charity continues to grow and we are reaching many Alphas and their families all over the country and still we hear the old cry "our doctor knows nothing about Alpha 1". All members are now automatically sent copies of all our Information Booklets, we hope this helps information get out to GPs and other medical professionals.

We have had news from one of our members Ray Overton, he had a lung transplant 6 months ago, he is doing well and is hoping to return to work, this is wonderful news and we send Ray our very best wishes.

FIND-a1

This is a new initiative to raise awareness of Alpha 1 among the medical profession, you will be receiving copies of the FIND leaflet to take to your GP, Health Centres and hospital Consultants, the next time you attend. With your help we are getting the message about Alpha 1 out to the wider community. Thank you

Don't forget our online forum, lets you chat to other Alphas, it can be found at http://techno.demon.co.uk/a1aforum, it would be great to see you there.

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If you have changed your postal or email address and wish to continue receiving communications from us please will you contact us with your new details. We are getting a large number of email addresses bounce when we try to send out emails to a number of members. Thank you

If you wish to contribute in any way to future editions of the Newsletter, please contact at the address below.

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