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Newsletter Alpha1 Awareness

A Word from the Chairman

On the 18th of August our registration number came through from the Charity Commission and from that date we could call ourselves a Charity without the need to add other words to qualify it.

Since registration we have had some successes and some disappointments. Some active members have stood down because of ill health others have left for personal reasons. At the same time new members have joined us and brought their enthusiasm to get something done about the widespread lack of knowledge of AAT Deficiency.

One of our aims is to promote the detection and treatment of diseases resulting from AATD. One way of doing this is by lobbying our politicians both here in the UK and in the European Parliament. Often the response is 'these matters are being considered by this committee or that committee'.

In practice, these committees do listen to patient organisations. Large organisations can shout loud. We are not large but we can work with other continental Alpha groups to increase the level of our voice.

Alpha1 Awareness is now the UK member organisation of the Alfa Europe association which is the European Federation For Alpha1 Antitrypsin Deficiency. The new website is www.alphaeurope.eu and it promises to give much more information on health care in Europe and consequently in the UK. There are a few teething problems in showing the map with all the association members but these can be accessed via www.alfaeurope.org .

Apart from this, a long-term objective, we are trying to help Alphas here and now. We have a Parents Forum for those bringing children with Alpha1 related problems – mainly liver conditions. We also have a number of grants available to Alphas for a range of purposes (see our website for more information).

Alan Heywood-Jones

Meeting at ADAPT

In late October Alan Heywood-Jones, his wife Dawn and Stephen Mayhew met Professor Stockley at the ADAPT† Centre at the Queen Elizabeth Hospital in Birmingham. The purpose was to explain the objects of the charity and to discuss possible activities over the coming year. This is an important period for all lung-affected Alphas since the results of two major trials‡ are to be published in international medical journals. Possibly this will be the first step in the process whereby Prolastin becomes available in the UK for the slowing down of lung deterioration in some ZZ Alphas. The publication dates will probably be in the early part of 2009.

Professor Stockley explained some of the mechanisms of the progress of the disease. He talked about his hopes that regional centres could be created for the detection and treatment of the condition. He was optimistic that augmentation therapy would eventually be made available.

At the end of a convivial discussion the Professor expressed his continuing support for the work of Alpha1 Awareness.

†ADAPT is the centre for research into Antitrypsin Deficiency. Under the direction of Professor Stockley it has an international reputation for its work in genetic emphysema. Many English and Welsh patients go to the centre for advice and annual check-ups.

‡The two trials are the EXACTLE Trial (see article below) and the Roche Repair Trial which is an investigation of the means of repairing damaged alveoli in the lungs.

Report on the EXACTLE Trial

A recently-completed trial in three European countries suggests that Prolastin is effective in slowing the progression of emphysema, Professor Robert Stockley, MD, of the University of Birmingham told a group of patients who were in the trial.

To the question "Does it work?" Professor Stockley replied, "It appears so." He was speaking at a meeting in Birmingham on the 9th of June.

The study has major significance in the UK because no augmentation therapy has ever been approved by the national licensing authority in Britain.

The trial involved patient groups in Britain, Denmark and Sweden. Some participants were given Prolastin infusions, some a placebo; neither the patients nor the medical professionals administering the infusions knew which patients were on placebo.

Participants were followed for 24 to 30 months between 2003 and 2006. The study results were measured by four different measures: CT scan, exacerbations, pulmonary function tests and a standard questionnaire.

Professor Stockley presented the results of the study at the American Thoracic Society annual conference in Toronto in May.

Some of the points presented at the June patient conference by Professor Stockley to summarize the Exacerbation and CT Scan as Lung Endpoint (EXACTLE) Trial:

- The rate of decline in Prolastin patients became clearly slower after about 12 months on augmentation.
- Hospitalisation of Prolastin patients appeared to be only one-third that of the placebo patients
- Most decrease in lung deterioration took place in the lower lung.
- All four methods of measurement showed a decreased rate of lung deterioration with Prolastin augmentation, although only one method showed clinical significance by conventional standards.

Professor Stockley said three medical journals are being considered for publication of the study. After publication, the Professor said the study will be brought to the attention of the National Institute for Clinical Excellence (NICE), the UK's licensing body for prescription drugs.

Prolastin is the augmentation therapy manufactured by Talecris Biotherapeutics

Do you belong to a Breathe Easy Group, a LEEP Group, or a Pulmonary Rehab Group, we would love to hear about it, share your stories with us, tell them about us if there are other Alphas in your group.
Remember anyone can become a member of the Charity they do not have to be an Alpha, they can be a family member, a friend, they can even be from the medical profession, if they are sympathetic to our aims, they can join us, it doesn't cost anything to be a member, you can find a membership form on our website.

Scottish Alpha 1 Patient Meeting
Edinburgh, 11 October 2008

This event was the first meeting of Alphas and friends to be held in Scotland. About forty people came from all over Scotland to a manorial hall in the grounds of Edinburgh University to listen to, and question, specialists working with Alphas.

The keynote speech was given by Professor Bill MacNee, Professor of Respiratory and Environmental Medicine, Royal Infirmary of Edinburgh. He described the mechanism by which the faulty AAT produced by the liver was trapped there and the consequent uncontrolled processes within the lungs which lead to emphysema. His talk was well received and lead to a number of questions some concerning screening. The Professor's view was that whole population screening would be too expensive. The audience liked his renaming of lung exacerbations as Lung Attacks – giving them the impact associated with heart attacks.

Doctor Claire Cotterill spoke about the work done by the Genetic Interest Group to involve patients and their families and carers in association with the Scottish Executive to try and influence health policies and improve health services.

Dr Andy Robson, Chief Clinical Respiratory Scientist, Royal Infirmary of Edinburgh, gave a light-hearted, but informative, presentation about the role of the Respiratory Function Laboratory in respiratory medicine and how it can help with initial diagnosis, ongoing treatment and assess effects of treatment in patients. He also spoke about the various tests available and how they are in the best interest of the patient.

Larry Warren, Chairman of Alfa Europe, talked about the European union of Alpha1 associations and its work to promote awareness of the condition, to promote research and to monitor therapy.

After a tea-break the second session started with a presentation by Dr Thomas Kohnlein, Hanover Medical School. He talked about present and future treatments including augmentation therapy both by infusion and inhalation. His graphs of lung function deterioration and survival rates seemed very persuasive. There followed an animated discussion between the Alphas present on the level of proof needed for a augmentation therapy to become available. Professor MacNee defended maintaining the highest of scientific standards.

Wendy Pollock, Senior Physiotherapist, Royal Infirmary of Edinburgh, in a talk that bubbled over with her enthusiasm for her work, explained that pulmonary rehabilitation is a 6 week exercise and education programme to help patients become physically fitter and reduces anxiety. Pulmonary rehabilitation can be useful prior to transplantation.

The head of the BLF (Scotland and NI),Richard Dietrich talked about how the BLF is supporting pulmonary rehabilitation by a Breathe Easy Active initiative, which involves physical trainers going to college and then developing classes for COPD patients.

The day ended with a short talk by Alan Heywood-Jones on the differences in charity law between England & Wales and Scotland. To operate in Scotland a charity needs to be registered with the Office of the Scottish Charity Register and that Alpha1 Awareness are looking at various options to do this. The consensus of the room was that people were in favour of Alan's proposal to set up a charity in Scotland based on the UK model.

Thanks were given to Andrew and Karen for organising the event. It was an informative and convivial day for all the participants.

The day ended with a gala dinner with lots of happy chatter filling the room.

News in Brief

Fundraising

Our fundraising efforts, in this our first year as a registered charity, have been very encouraging. We started with the London Marathon in April. Tracy Hill ran and donated a substantial amount to the charity. Tracy's husband is PiMZ. September saw the Chichester 10 kilometre run. David Hollands-Hurst and his daughter Camilla ran in memory of their mother-in-law and darling grandmother Dannie who sadly died earlier this year. They raised over a thousand pounds – a magnificent donation.

Christmas cards

We have been very successful in selling our own cards. There are still some available at £3 per pack of ten. There is a small charge for postage. A copy of the designs available are attached to this newsletter, along with an order form, if you would like to order cards or would like more information please contact Lin Daniels at secretary@alpha1awareness.org.uk or send your order with a cheque to the PO Box number at the end of this newsletter

Quiz

Lin organised a quiz at her church hall and raised £166. Every little helps.

Shop Online and Raise Funds

It is completely free 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, Woolworths, 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

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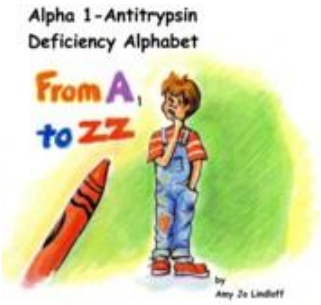
Alpha Children



If you have an Alpha child between the ages of four and eight then you may find the booklet *Alpha 1-Antitrypsin Deficiency Alphabet From A_1 to ZZ* a great help in explaining the condition to them. Despite the long title the booklet is easy to read and quite upbeat. It may also be helpful to non-Alpha children in explaining the illnesses of their older relatives. Contact Lin for a copy.

Over the last year we have heard of several babies and children being diagnosed with Alpha 1, parents of these children are obviously distressed, confused and concerned for their children when they are told of the diagnosis. To help these parents to understand more about Alpha 1 and to have contact with other parents and families in a similar position, we have set up a Parents Forum, it is possible to contact the forum via our website or by writing to the Parents Forum via the PO Box number, it is so important for these parents to know they are not alone.

There is an excellent article on our website, under "Alpha Children", written by a young woman, who was diagnosed as a child with Alpha 1, as was her sister, who had to have a liver transplant as a child. This is a story of encouragement and hope for all parents of Alpha children, as both these young women lead near normal adult lives (for those of you receiving this through the post we are attaching a copy of the article)

Panniculitis

Recently, two Alphas have been treated for this rare condition and this has lead to interest in this little known disease.

The following article is reproduced from the Alpha 1 Foundation Research Registry in the USA

Panniculitis in Alpha-1 Antitrypsin Deficiency

By James K. Stoller M.D., M.S.

Panniculitis an uncommon manifestation of Alpha 1 Antitrypsin Deficiency This brief paper will discuss the definition of panniculitis the variety of potential causes current understanding of the mechanism of panniculitis its signs and symptoms and available experience with treatment.

Panniculitis is an inflammation of the panniculitis, which is the fibro-fatty tissue layer that lies underneath the outermost or superficial layers of our skin. This layer of the skin resembles a honeycomb, with globules of fat separated by walls or septae In anatomic terms panniculitis is categorized as either being septal (involving the walls separating the fatty sections of the panniculitis) or lobular (affecting the fat globules or collections themselves).

Like most medical conditions, panniculitis can arise from many underlying causes. Alpha-1 Antitrypsin Deficiency is one of those causes. Among the other potential causes are a group of diseases known as connective tissue disease (which include conditions causing diffuse body inflammation, such as systemic lupus erythematosus and rheumatoid arthritis), underlying so-called lymphoproliferative diseases (like lymphoma), pancreatic disease, gout, kidney dysfunction, so-called atheroembolism (in which clots from blood vessels find their way to the fibro-fatty layer of the skin), and even adverse reactions to some drugs, including corticosteroids.

Panniculitis manifests as characteristically red nodular spots on the skin which may break down and ulcerate, causing an oily discharge. While these nodular blotches may occur anywhere on the body, common sites include the thighs and buttocks and areas subject to trauma. Conditions that may precipitate the development of such nodules include trauma (including rigorous exercise), intravenous injections, or cryosurgery on the skin (which is surgery involving freezing the skin). The lesions of panniculitis may go on to develop deep ulceration with tissue breakdown, called necrosis. Such necrotic nodules are usually painful to the touch.

Panniculitis is felt to be due to inflammation of the fibro-fatty layer of the skin, presumably mediated by unopposed protein breakdown. In Alpha-1related panniculitis, the mechanism of panniculitis resembles that believed to cause the development of emphysema, namely the unopposed breakdown of tissue by the absence of alpha-1 antitrypsin, allowing proteases within the body to affect structures underlying the skin (in the case of panniculitis) or the support matrix of the lung (in the case of emphysema).

Panniculitis is an uncommon complication of Alpha-1 Antitrypsin Deficiency. It was first described in a patient in France in 1972 by Dr Warter and colleagues. These physicians described a young woman with severe deficiency of alpha-1 antitrypsin who developed characteristic red nodular, painful skin ulcers. Since that original report, fewer than 50 cases of panniculitis in individuals with Alpha-1 Antitrypsin Deficiency have been reported in the medical literature establishing that panniculitis is a very uncommon complication of Alpha-1 Antitrypsin Deficiency. For example in the National Heart Lung and Blood Institute Registry of Individuals with Severe Deficiency of Alpha-1 Antitrypsin only a single participant reported having panniculitis In various reports specifically about panniculitis and Alpha-1 Antitrypsin Deficiency only 28 patients had been described through 1997 Tallying all reported cases through 2003 shows a total of 44 individuals with panniculitis complicating Alpha-1 Antitrypsin Deficiency described in the medical literature. Importantly, panniculitis in Alpha-1 Antitrypsin Deficiency can accompany various phenotypes (or genetic types of Alpha-1 Antitrypsin Deficiency), some with severe deficiency of serum levels of alpha-1 antitrypsin

(e.g., including PI*ZZ and Pl*SNull) and others with only mild deficiency (e.g., Pl*MZ and Pl*MS). In one series by Humbert and colleagues, of the 26 patients with panniculitis and Alpha-1 Antitrypsin Deficiency described, 62% were Pl*ZZ, 15% were PI*MZ, 8% were Pl*MS, and 4% were Pl*SNull; in the remaining 8%, the phenotype was not stated. The reported experience suggests that panniculitis occurs equally among men and women and that the mean age of onset is approximately 40 years old.

Various therapies have been tried and evaluated to treat panniculitis including corticosteroids antibiotics (including doxycycline and dapsone), full plasma exchange, and intravenous pooled human plasma alpha-1 antitrypsin (more popularly called augmentation therapy). Of these various treatments, augmentation therapy has been the most dramatically successful. Several reports describe resolution of panniculitis after as few as 3 doses of intravenous augmentation therapy. The dose of augmentation therapy for panniculitis is the same as that for established emphysema, 60 mg/kg once weekly.

In summary, panniculitis can be both an annoying and also potentially disabling complication of Alpha-1 Antitrypsin Deficiency. Panniculitis is thankfully very uncommon and is amenable to effective treatment with existing approaches for Alpha-1 Antitrypsin Deficiency, including augmentation therapy. Undoubtedly, the spectrum of treatment choices for panniculitis will grow along with ongoing research regarding optimal therapy of individuals with Alpha-1 Antitrypsin Deficiency and with the development of new treatment options.

Looking Ahead to 2009

2009 will be our first full year as a registered Charity and we are already looking at what we can do next year, to increase awareness of Alpha 1 and support you our members. We want to hear from you what you want to see from the Charity, remember we have small grants available for Alphas and their families, check out our website for more information or contact us at the PO Box number. Hopefully in the new year we will have some new literature in the form of leaflets available, we will let you know when these are available

London Marathon

We already have a young man raising money for us, Garry Stone is running in the London Marathon, in memory of his father who died recently, from Liver related Alpha 1, he has already set up his Justgiving site and is receiving sponsorship for his run. He will have the Alpha 1 Awareness Charity name on his running vest. We wish Garry very good luck in his training and on the day itself

Members Information Day

An Information Day is planned for the Spring/early Summer time, as yet we do not know where or when, but as soon as we have details of dates, times, place and speakers we will let you know. We look forward to seeing as many of you as possible at this day.

European Conference in Vienna

Now that the UK is a member of the Alpha Europe Federation, two people have been invited to the Conference in Vienna, next June, we are waiting for confirmation of the date.

Newsletter

We hope to produce a newsletter, quarterly, if there is anything you would like to see included, any questions you would like answered please contact us and we will do our best to include them in our forthcoming issues

We would like to wish all members of
Alpha 1 Awareness UK
A Very Merry Christmas
&
A Healthy, Happy and Prosperous 2009

Alpha 1 Awareness UK

Raising Awareness of Alpha 1 Antitrypsin Deficiency in the UK

Contact us at : Alpha 1
PO Box 2866
Eastville
Bristol BS5 5EE

email : secretary@alpha1.org.uk

visit our website : www.alpha1awareness.org.uk