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Newsletter Alpha1 Awareness

A Word from the Chairman

The members of the AAW team wish our readers a happy, prosperous and, most importantly, a healthy New Year. The cold weather at the turn of the year has made going outside especially difficult for lung affected Alphas. Many are confined to the warmth of their homes and had to rely more than usual on their friends and family. This comes after similar 'stay indoors' advice given to at-risk Alphas because of the recent Swine Flu pandemic. Over Christmas I had the opportunity to speak to an Irish lung-affected Alpha who did contract this form of flu. This was a very worrying time for him and his family. Fortunately, he was treated promptly and after a few stays in hospital he is now recovering well.

The arrangements for the European Alpha1 Congress in London are progressing well. The major bookings have been made by the Alpha Europe Foundation and Lin and Dawn are now concentrating on checking all the details to make sure that the presentations and social events run smoothly. Meryl is hoping to hold a Lunch meeting in South Wales along the lines of the successful event in Cornwall. There are plans for a Mini InfoDay to be held later in the year in the North of England. The forum continues to be very active under the guidance of Neil.

The Department of Health have announced a scheme to make selected innovative medicines available on the NHS for a time-limited period, prior to a NICE appraisal. In July the Office of Life Sciences published the 'Life Sciences Blueprint' which announced a package of measures to help maintain a competitive life science sector. One of the initiatives announced in the blueprint was the introduction of an 'Innovation Pass'. While it is uncertain whether or not the scheme will assist competition, the scheme does pump £25 million into buying expensive medicines that have not yet received NICE approval. The initial medicines which will get an Innovation Pass are mainly for the treatment of terminal cases of cancer. This is not immediately of much use to Alphas but the introduction of the scheme does show that the Department of Health is responding to public concerns over NICE and how it regulates treatments for rare diseases. We can but hope that more is to follow and that augmentation therapy for AATD is included. Meanwhile, I have responded to the consultation paper for the Innovation Pass pilot and Dawn is talking to a pharmaceutical company about entering one of their plasma products into the scheme. See the article in this newsletter on Patient Advocacy in Europe.

Alan Heywood-Jones

ALPHA 1 LIVER DISEASE

While we know that the liver is the primary organ involved in the production of AAT protein and that it is problems with this production that lead to the AAT deficiency in the blood and liver disease in some individuals, we really have no answer to the questions regarding why some children and some adults get liver disease from Alpha1 while others do not.

About 2% of children born with Z-type Alpha1 develop liver failure requiring transplantation during the first two years of life. Most infants born with two Z genes have abnormal liver function tests during this same time period but for those who don't develop significant liver disease, these tests usually return to normal during childhood. Some investigators have suggested that there is a problem with the mechanism in the liver that handles accumulated proteins in the 2% who get severe liver injury and perhaps this work will lead to therapies to prevent this.

In adults, most individuals with Alpha1 will not develop clinically significant liver disease. If liver disease does occur, it is often very mild and chronic, remaining stable for years. However, there are a number of adults who develop liver disease suddenly and have a rapid downhill course that results in liver transplantation. As far as we know at present, there is nothing that distinguishes those with mild, chronic liver injury from those with rapidly progressive, sudden onset disease.

Studies have shown that most older individuals with Alpha1 (over 65 years old) have liver scaring that can be seen under the microscope, even though their liver function tests are entirely normal. This suggests that all or most individuals with Alpha1 have ongoing, low levels of liver injury throughout their lives. Fortunately, the liver can tolerate this type of injury and still perform its normal functions. Why the injury becomes too severe for the liver to tolerate in some individuals and not others is a major unanswered question at this time. It is believed that environmental factors and perhaps additional genetic factors may play roles in promoting such injury.

While it is clear that there is still a lot to learn, work is proceeding to use what we do know to develop new therapies for Alpha1. Since we know that both the deficiency in the blood that leads to lung disease, and the liver problems themselves are caused by the accumulation of abnormal protein within liver cells, scientists are trying to develop drugs that will cause the trapped AAT protein in liver cells to move out of these cells and into the blood. This would have the dual advantages of relieving the injury to liver cells and pushing this protein into the blood where it can bathe the tissues of the body and especially the lungs. Such a drug could represent a potential cure for this genetic condition.

In addition to therapies to treat or prevent Alpha1-related disease, work is moving forward on diagnostic tests that have the potential to detect mild or early liver injury more accurately than current methods.

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NEWS IN BRIEF

Donations & Research

As you will have read in our last Newsletter, we have been very fortunate in receiving some very substantial donations. These donations have given us the opportunity to give a donation to a research programme. After careful deliberation the board of Alpha 1 Awareness made the decision to give a donation of £1000 to the paediatric liver consultant, Dr Dino Hadzic at Kings College Hospital in London, who is carrying out research into the effect of Alpha 1 on the livers of diagnosed children who are referred to the hospital. It has pleased Alpha 1 Awareness immensely to be able to make this substantial donation and we are extremely grateful for the generosity of our donors.

However the good news does not end there, we are very, very grateful to the CEO of Alpha 1 Organisation in the USA, John Walsh, for agreeing on behalf of Alpha 1 Org, to match the £1000, donation to Dr Hadzic pound for pound, thus making the donation £2000 we send John and all at Alpha 1 in the USA our most sincere thanks.

We have held over some of the money which has been donated to AAW for research, to enable us to be able to make donations in the future, when we hear of other research projects.

Children's Books

Some of you will have seen the book, "A1-ZZ", copies of which we were sent from "Alpha Kids" in the USA. The book was written, by a Mum, in the USA, who wanted to explain Alpha 1 to her children. Using bright colourful illustrations and letters in alphabetical order, it explains about Alpha 1 and is aimed at children up to the age of around 10 years of age. The book is obviously aimed at children in the USA and in places uses slightly different phraseology, to what we would use hear in the UK. After discussion with members of "Alpha Kids" and the author of the book, they have agreed and given us permission to produce an "Anglicised" version of the book for children hear in the UK.

Production of this book will go ahead, in the next few months thanks once again to the kind and generous donations we have received, from marathon runners, personal donations and donations in memory of loved ones.

To everyone who in 2009 donated to us in anyway, by running marathons, holding fundraising events, receiving donations instead of gifts for significant birthdays or anniversaries or simply buying our Christmas Cards and notelets, our heartfelt thanks.

Membership

Our membership secretary, Meryl, reports that our membership is still growing steadily, which we are finding very encouraging.
Remember for every member we have join us, there will be family members who are also Alphas and so we are reaching more people all the time and continuing to spread the word and the support.
Can please remind you that if you change your email or postal address, please remember to let Meryl or Lin, our secretary, know in order that we can stay in touch and keep you up to date with all that is going on in the Alpha 1 world.

Forum

The online forum continues to go from strength to strength, with new Alphas, carriers and carers joining us every few days. Various topics have surfaced recently - not surprisingly, talk about Swine Flu and the winter weather have been high on the agenda!

We also had a few members going down with various bugs, illnesses and even brief hospital spells over the winter break - but luckily we were still able to keep in touch with each other, lending support and good wishes wherever we could! Such is the joy of modern hospitals that some even have internet access these days!

Remember, you'll find us at http://techno.demon.co.uk/a1aforum for the time being (until we move to a site with a more 'catchy' web-address!) Do come along and join us - if you're sitting on the fence, wondering what these internet forums are all about, just remember you'll be safe with us! And you're more than welcome to join up and 'lurk' for as long as it takes until you feel comfortable to start chatting.

See you there! That link again:
http://techno.demon.co.uk/a1aforum

Neil Jackson
Forum Administrator

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Alfa Europe Annual Congress 2010

This is the annual meeting of the member countries of the Alfa Europe Federation the highlight of which is also a day of presentations given by experts in the field of Alpha 1.

This year the Congress is being held in London. The event is organised and funded by the Alpha Europe Foundation who have selected the venue and the speakers. Alpha 1 Awareness UK are very privileged to have been asked to act as hosts for the Congress. You as a member of Alpha 1 Awareness UK are invited to attend along with your spouse, partner, carer and family.

The Congress will be held on Friday 9th and Saturday 10th July, at the Hilton Hotel, Kensington in London. . On the Friday evening a dinner will be held at the Hotel. This is an opportunity to meet other Alphas and talk about life as an Alpha in a convivial setting. Dinner, your overnight stay on Friday 9th July, breakfast and lunch on Saturday 10th July will be paid for by the European Foundation, as will a contribution to your travel costs. If you live over a certain distance, have young children with you or are on oxygen, it may be possible to have the second night, 10th July, paid for by the Foundation.

Saturday's day of presentations will begin around 9.30am. Invited speakers include Professor David Lomas from Cambridge University, Doctor Dino Hadzic from King's College Hospital Paediatric Liver Unit, John Walsh from the American Foundation, Melissa Hillier from the Genetic Interest Group and other speakers to be confirmed later. The event will close about 4pm.

We at Alpha1 Awareness hope that you and your family will be able to attend the Alpha1 Congress in London. I can speak from experience when I say how useful and interesting these meetings are, not only because it gives you a chance to hear at first hand experts in the field of Alpha 1, but also it is an opportunity to meet other Alphas and share experiences.

We will contact you as soon as the Alpha1 European Congress registration form is available, by post or email. This is planned for early March. However, it would greatly assist in forward planning if you were to tell us about your requirements (rooms, oxygen, childcare if needed, etc). Send the attached form as an email to congress@alpha1awareness.org.uk or by post to Alpha1, PO Box 2866, Eastville, BRISTOL BS5 5EE.

Lin Daniels
Secretary

Note: The spelling Alfa is not a mistake – the official name of the Federation uses this spelling; it is more understandable by the majority of European Alpha associations.

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Patient Advocacy Groups in Europe and the United Kingdom

This article is to introduce some of the organisations that help with the understanding and treatment of rare diseases including Alpha1 Antitrypsin Deficiency. The aims and mission statements of the groups often sound very similar. Better health care, access to expensive medicines, improved training for doctors are recurrent themes. To achieve these aims they hold conferences at which medical experts, health service planners and patient groups can talk about these issues and generally try to raise awareness. Also, under the general term ***advocacy***, they attempt to catch the attention of elected representatives – members of the European parliament as well as national MPs. The representatives can then vote to influence the national strategies for better health systems.

An example of what is happening at the European level is the EUROPLAN. This project collects and disseminates information concerning the EU Member States' national initiatives on rare diseases and information about their expectations of national plans for rare diseases. In addition, there will be information about best practices related to sharing personal experiences, collecting data and creating effective strategies to address rare diseases. The Department of Health has yet to respond fully to this project.

All of this may seem a long way from a satisfactory appointment system at your local hospital or a consultation with a doctor who knows more than the basics of AATD but these improvements will come eventually thanks to the work of these organisations. Visit their websites and find out more.

EURORDIS is a non-governmental patient-driven alliance of patient organisations representing more than 400 rare diseases patient organisations in over 40 countries. Their mission is to build a strong European community of patient organisations and people living with rare diseases, to be their voice at the European level, and to fight against the impact of rare diseases on their lives. Eurordis has great political influence in the European parliament. They are holding a congress in Krakow, Poland in May this year. www.eurordis.org.

EPPOSI is a European patient-led partnership between patients' organisations, science and industry. It was founded in 1994 for the exchange of information and discussion of policies in human health care in Europe.

Like Eurordis, EPPOSI has strong links with the decision makers in Brussels. www.epposi.org.

Orphanet is a useful source of information on rare diseases and aims to improve the quality of medical care for rare (orphan) diseases. www.orpha.net.

PPTA is the Plasma Protein Therapeutics Association and is the primary advocate for the world's leading source plasma collectors and producers of plasma-

based and recombinant biological therapeutics. The association supports patient groups with an interest in plasma based products – these include Haemophilia and Alpha1 Antitrypsin Deficiency. They are holding a congress in Berlin in March this year. www.pptaglobal.org.

ESHG is he European Society of Human Genetics and is a non-profit organisation. Its aims are to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all persons who share these aims, particularly those working in Europe. On their website you can find more on Third Annual European DNA Day Essay Contest for High School Students. Last year the first prize went to a student from a college in Portsmouth, Hampshire. The deadline for this year's essay is Sunday April 25, 2010. www.eshg.org.

PLUS is a new organisation whose aim is to facilitate the exchange of information towards the building of consensus views when possible among the organisations that represent regular users of plasma ,plasma proteins or plasma derived therapies. They are still building their website.

The **Genetic Interest Group** aims to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them. www.gig.org.uk.

Closely related is **Rare Diseases UK**. This is a joint initiative of the Genetic Interest Group and others, including Alpha1 Awareness UK, in response to unmet health care needs of hundreds and thousands of families who currently struggle to get access to integrated care and support from the NHS. www.raredisease.org.uk

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Rare Disease Day 28th February 2010

In the UK Rare Disease Day is being coordinated by the Genetic Interest Group (the UK alliance for all people affected by genetic conditions) and Rare Disease UK a group brought together to develop strategic planning for rare diseases in the UK. Alpha1 Awareness UK is a member of both of these organisations.
Rare diseases affect over 3.5 million people in the UK, that is 1 in 17 people. Collectively rare diseases are not rare at all, however NHS services remain patchy and poorly integrated meaning that families often struggle to get the support and information that they need.
Rare Disease UK and the Genetic Interest Group are working together to develop plans and strategies to help those living with rare disease and those that provide services and information for families affected by rare conditions.
Rare Disease Day is to raise awareness of these issues and is always on the last day of February - this year Sunday the 28th.
Meetings are being held in Wales, Scotland and Northern Ireland. Our Membership Secretary, Meryl, is attending the day at the Welsh Assembly, it would be lovely if any of our members in Scotland or Northern Ireland could attend their respect meetings
We are being urged to write to our local MPs, SMPs, Welsh Assembly members etc to help raise awareness of rare diseases, remember Alpha 1 falls into this category
To find out more visit http://www.raredisease.org.uk/rarediseaseday_2010.htm.

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It has not been a good start to the New Year, with the cold, snowy and icy conditions, we hope that you have all managed during this very difficult time, staying, healthy safe and warm. Let us all hope that Spring is just around the corner and we have seen the last of the snow and ice.

Please remember we love to hear from you and if you have something you would like us to include in our Newsletter or you would like to write something about your how Alpha 1 affects your life and how you cope from day to day; are you considering being assessed for transplant, we want to know and would love you to share it with our members. Remember sharing and caring is what we are all about

Alpha 1 Awareness UK

Raising Awareness of Alpha 1 Antitrypsin Deficiency in the UK

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