Alpha-1 UK Support Group Newsletter

Issue 13
Autumn 2014

Welcome

To our Autumn 2014 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

The Alpha-1 Alliance Campaign goes from Strength to Strength

Alpha-1 Alliance Clinical Advisory Board

The Alpha-1 Alliance campaign to establish a Highly Specialised Service in the NHS to enable Alpha-1 patients to be treated by expert clinicians in dedicated hospitals is now well underway.

The Alpha-1 community now have unprecedented levels of support in Parliament and an application is currently with Government officials on how an NHS Alpha-1 service should be organised.

The Alpha-1 Alliance campaign began in 2012 as a coalition of patients groups and expert respiratory consultants. In 2013 the Alliance delivered a petition to 10 Downing Street signed by over 2000 people calling on the Prime Minister to act.

Later that year the Alliance launched a ground breaking report at a seminar in Parliament detailing the true extent of the unmet medical need of the Alpha-1 community and made key recommendations to the Government on how an Alpha-1 Highly Specialised Service should be organised in the NHS.

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Alpha-1 Alliance launches policy report in Parliament

The seminar was attended by patients, MPs, expert clinicians and government officials who debated the findings of the report and how best the Government can support the Alpha-1 community.

Chair of the event, Mark Pawsey MP, the Conservative Member for Rugby, said: "It is crucial that we raise the profile of Alpha-1 amongst Parliamentarians to help them understand how patients are affected and the benefits of access to new services."

Earlier this year the campaign went from strength to strength with Alliance representatives briefing numerous high-profile MPs on the needs of the Alpha-1 community including the Secretary of State for Health, Jeremy Hunt MP and his shadow in opposition, Andy Burnham MP. Karen North. Alpha-1 Alliance Executive Member, said: "Meeting the Minister was an important milestone and Alpha-1 is now firmly on the political agenda."

More recently in June, the Alliance convened a committee of renowned Alpha-1 expert clinicians who developed detailed recommendations for an Alpha-1 service.

The recommendations formed the basis of the Alpha-1 Alliance application to the Government. The application is now in a multi-stage review process and the Alliance is committed to keeping the pressure on the Government to ensure a successful outcome.

"Meeting the Minister was an important milestone and Alpha-1 is now firmly on the political agenda."

England – Watch this space

In Scotland, significant progress is also being made with the official Alliance campaign launching in March this year and an event bringing together Scottish patients, politicians and clinicians being scheduled for November. Scotland – your invitation to attend Parliament to tell your story to MSPs will be coming soon. Please get involved and visit the Alliance website for further details on all campaign activities:

www.alpha-1-alliance.org.uk

Continuous support by and engagement of the wider Alpha-1 patient and clinical community is absolutely critical in order to make specialised services for Alphas a reality without the passionate - engagement of Alpha-1 UK Support Group in this campaign, the Alpha-1 Alliance would not have come this far.

Alpha-1 Alliance briefs Shadow Health Secretary on the need for a national service.

Alpha 1 Alliance

Campaigning for better services and treatment for Alpha-1 patients in the UK

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Alpha-1 Alliance Campaign Timeline

	Alpha-1 Alliance's application for highly specialised Alpha-1 service endorsed by CRG	12/07/2014
	MSPs commit to supporting our campaign for a highly specialised Alpha-1 service in Scotland	25/06/2014
	Dr Ravi Mahadeva steps down as Chair of the Alliance	29/05/2014
	Letters from the Cabinet Secretary for Health on Alpha-1	20/05/2014
	Formal submission for a highly specialised Alpha-1 Service	28/04/2014
	Alpha-1 Alliance meets Andrew Lansley MP to discuss need for a national service	25/03/2014
	Alpha-1 Alliance supports audit of Alpha-1 patients in Scotland	03/03/2014
	Alpha-1 Alliance joins Rare Disease UK	21/01/2014
	Alpha-1 Alliance launches policy report in Parliament	12/12/2013
	Alpha-1 Alliance briefs Shadow Health Secretary on the need for a national service	14/11/2013
	Alpha-1 Alliance present at patient group conference	14/09/2013
	Question on Alpha-1 in the House of Lords	14/08/2013
	Alpha-1 Alliance attends meetings with Parliamentarians	08/07/2013
	Meeting with NHS England to discuss service application	17/01/2013

27/06/2014	Alpha-1 Alliance secures wide clinical expert support for national service at Advisory Board	
12/06/2014	Minister answers Parliamentary Question on Alpha-1 Antitrypsin Deficiency	
29/05/2014	The Alpha-1 Community meets with Jeremy Hunt MP, Secretary of State for Health	
19/05/2014	Alpha-1 Alliance briefs MSPs in the Scottish Parliament	
27/03/2014	Minister answers Parliamentary Questions on Alpha-1	
25/03/2014	Alpha-1 Alliance in discussions with NHS England	
01/02/2014	Early Day Motion on Alpha-1 tabled in Parliament	
20/12/2013	Alpha-1 Alliance informs clinical community	
01/12/2013	Alpha-1 Alliance attends Genetic Alliance UK HST workshop	
15/10/2013	Policy report to be launched in Parliament	
15/08/2013	Department of Health responds to Alpha-1 Alliance e-petition	
31/07/2013	Visit to Downing Street	
04/07/2013	Alpha-1 Alliance's e-petition closes with over 2000 signatures	
12/12/2012	Alpha-1 patient survey closes	

Please visit the Alliance website for full news reports **www.alpha-1-alliance.org.uk/news/**

The Future of ADAPT

Professor Robert Stockley

The Future of ADAPT

ADAPT (Antitrypsin Deficiency Assessment and Programme for Treatment) has run for 18 years now and started as a research programme to understand the nature and progression of health in alpha-1 -antitrypsin deficiency (AATD) with particular emphasis on the lung. The programme was underwritten initially by Bayer Biologicals who manufactured Prolastin. Talecris and more recently Grifols continued this funding as their contribution to the AATD field of education and research.

This industry/academia collaboration was an innovative approach and has been copied in other branches of medicine in recent years. ADAPT resulted in over 200 publications and review articles as well as training about 30 young doctors in the identification and managements of the disease. The funding allowed ADAPT to see all patients and relatives referred by their health care workers (or themselves), to educate and inform patients and their families of the implications of AATD, optimise their current treatment and support local teams in their management. The programme evolved from a research one towards an assessment and management programme establishing the template for how AATD patients should be managed in expert centres.

The programme was supported by other independent research grants and became the most extensive and comprehensive database of AATD worldwide with over 1,000 Pi Z patients followed annually. This data enabled the natural history to be documented extensively and established the importance of other measures such as health status, chest infections, gas transfer and CT scan in monitoring patients rather than the conventional FEV_1. Indeed the programme established and validated CT as the most specific measure of the lung disease and its' progression. This has now become a recognised outcome measure for clinical trials and together with our knowledge of patient characteristics, has led to the development and delivery of several important clinical trials.

The NHS has never funded any aspect of the ADAPT programme and the recent sudden decision by Grifols to cease funding of ADAPT has led to other challenges. The NIHR (National Institute for Health Research) has funded the establishment of a network of academic centres to enable the principles of ADAPT and its detailed assessment and follow up to be available in other centres. Several grants from the AoF (Alpha-1 Foundation) have enabled us to continue following many patients with "normal" lung function as well as mild to moderate disease. CSL Behring has provided some funding to support clinical help and data analysis. However, we are aware that this restricts the number of patients who we can now see and monitor as the core funding for staff, travel and routine tests has become severely limited.

Nevertheless, within these constraints we are not abandoning the AATD community and will be working to secure the necessary NHS funding for our unit and the establishment of other centres that will provide/continue to provide the expert service AATD deserves. Meanwhile, we continue to see new patient referrals but have to limit our follow-up testing.

Patient Community

Being a patient under ADAPT means that you meet other patients. There is an alpha-1 "community" in which patients support each other in friendship, sharing information, supporting research etc and the strength of the patient group is central to our long-term aims.

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

Alpha-1 Foundation & Association Merger

Gordon Cadwgan and Henry Moehring

The Boards of Directors of the Alpha-1 Foundation and Alpha-1 Association USA announced on January 17th this year that talks were advancing for a proposed merger of the Alpha-1 Association within the Alpha-1 Foundation.

The two Boards, led by Foundation Chair Gordon Cadwgan and Association Chair Henry Moehring, recommended the merger of the Alpha-1 Association into the Alpha-1 Foundation, unifying the Alpha-1 community's voice. In recent months, a task force named by the Boards designed a plan that incorporates each organization's mission, values and operations.

The two organizations have collaborated since the Foundation was created in 1995. As the organizations are increasingly being called upon to provide leadership to Alphas around the world, and Alpha-1 detection is increasing, the Boards determined a joint effort could more effectively serve the Alpha-1 community.

The proposed plan will strengthen advocacy efforts, expand educational opportunities, provide for greater efforts in detection and support, and expand our ability to fund critical research, Cadwgan and Moehring said in a letter to members of the Alpha-1 Association.

All of the Association's programs, including support, education and advocacy, will continue under the Alpha-1 Foundation after the proposed merger. Support groups will remain intact, continuing their essential part in the community.

The proposed merger would further strengthen the Foundation's ability to continue funding research programs to seek new therapies and ultimately a cure for Alpha-1

The proposed merger would further strengthen the Foundation's ability to continue funding research programs to seek new therapies and ultimately a cure for Alpha-1. In 2013, the Foundation increased its in-cycle research grants by $200,000, and on January 13th announced a new research funding opportunity, the Gordon L. Snider Scholar Award, to encourage outstanding young investigators to pursue a career in Alpha-1 research.

Going forward as one organization, we will "position ourselves for future success and best serve the Alpha-1 community," Cadwgan and Moehring say.

On April 16th Association Chair Henry Moehring announced the membership of the Alpha-1 Association approved the organization's merger into the Alpha-1 Foundation by an overwhelming margin. The vote, with 426 ballots submitted, was 413 in favour and just 13 opposed, or a 97 percent favourable vote.
On Thursday 5th June, in advance of the Alpha-1 National Education Conference in Kansas, the respective boards of the Alpha-1 Foundation and Alpha-1 Association met and overwhelmingly voted in favour of finalizing the proposed merger of the two organizations.

In ratifying the merger, new Board Vice Chair Henry Moehring, formerly chair of the Alpha-1 Association, said, "We are honouring the work of people who came before, some still with us and some, unfortunately, not. The merger is an affirmation of our deep commitment to serving the Alpha community."

ALPHA-1 FOUNDATION

Social Gatherings, Information Days & Mini Meets

Bridgend, Wales September 2013

It was a very special annual event held in Bridgend, Wales where we commemorated the 50th Anniversary of the discovery of Alpha 1 Antitrypsin Deficiency.

We had some great speakers which included our Patron

Professor Robert Stockley
Head of the research team ADAPT the leading UK Research Centre for Alpha-1 based at the Queen Elizabeth Hospital Birmingham.

Dr Bibek Goopta
Research Fellow Berbeck Universaty of London who trained with Professor Lomas at Cambridge and talked about work that is being done to tease out the molecular mechanisms behind Alpha-1, and to look at how these may be addressed by small molecules and/or stem cell strategies.

Jamie Holyer
Secretariat for the Alpha1 Alliance who are campaigning for the establishment of nationally commissioned highly specialised services for Alpha-1 to ensure equal access for patients to optimal integrated clinical care for this complex disease.

Henry Moehring
Vice Chairman of the Alpha1 Foundation in the USA.

All our speakers were presented with traditional welsh gifts to thank them. As it was the 50th anniversary members had surprised the trustees and committee with gifts and a celebration cake for all their hard work, Pewter Welsh love spoons for the Ladies and Pens, key rings and Mugs for the Gentlemen the cake was enjoyed by everyone with their afternoon tea. The children that

attended were presented with gifts for being so well behaved, they were a joy to have.

A 100 people 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 our speakers, there was the usual Auction, Prize Draw and Tombola during the day and music and much merriment in the evening. It's always so nice to meet up with old friends and new each year and it's sad to say goodbye on the Sunday

morning but everyone agrees that as usual it's been a fabulous weekend and can't wait for the next one.

This year the Annual Social Gathering is at the Best Western plus, Bentley Hotel and Spa in Lincoln on 20th September 2014, any one interested in booking should contact:

John Mugford

infoalpha1uk@googlemail.com

Dundee, Scotland March 2014

The event commenced with a buffet lunch and an opportunity to say hello to everyone and browse the groups merchandise. Our Secretary Karen North then officially welcomed everyone and gave an introduction to the groups history, achievements since we were founded in 1997 and introduced the Trustees & Committee Members.

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The first guest speaker of the day was Jamie Holyer who spoke about the Alpha-1 Alliance who are campaigning for a national service in Scotland, Wales and England for the Alpha-1 community. He explained that whilst we have the expertise in the UK we don't have an organised national service or access to augmentation therapy. Before a medicine can be made available in the NHS 2 things need to happen, the first is that the drug needs a licence and the second is the cost of the medicine and the associated

healthcare needs to be funded. An augmentation drug manufacturer applied for a licence for their product to the EMA (European Medicines Agency) in December 2013 and it is anticipated that a licence will be granted sometime in 2015. Jamie explained that the campaign is necessary to secure support and investment from the Scottish Government and NHS for a specialised centre for Alpha-1 patients to include paediatric, lung, liver, and skin care and also augmentation therapy and other forthcoming medicines. In order to achieve this it is really important that the alpha-1 community speaks with one voice, that the clinicians and patient representatives are united and give one clear message to government and the NHS about what is required. This is why the Alliance has been formed to bring these groups together so that we can campaign with a single message.

Unfortunately Professor MacNee was taken ill and could not attend the meeting and Andrew Deans very kindly stepped in. Andrew spoke initially about the genetics of alpha-1, how the lung disease occurs and that we should take antibiotics as soon as possible when we have a chest infection. There was also mention of the vast amount of research being conducted on the liver to try and find the cure. We then looked at the incidence rates of Alpha-1 throughout Europe and into the statistics of incidence in Scotland by region. There is an audit currently underway to support the Alpha-1 Alliance campaign with the statistics required to support an application for access to services and treatment.. Andrew reinforced that he can provide the data to support a service but we have to actively support the campaign and the activities of the Alliance.

Phil Rowsby very kindly gave a presentation about how the Butterfly Trust supports Cystic Fibrosis sufferers in Scotland and encouraged involvement not only to support the campaign but also the activities of the UK Support Groups activities here in Scotland. The key message is "the baby that cries loudest gets fed first". After the coffee break Andrew Deans delivered a well-received presentation on "Inhalers!! Confused? There is no need to be". It was a practical session which commenced with everyone using a respiratory device which measured the speed at which you used your inhalers. Interestingly not all inhalers require the same technique when being used. In principal aerosol inhalers should be inhaled "gently" and non-aerosol should be used "forcefully" although it should be noted that with new inhalers being released the technique required may differ slightly. Trainer whistles are available from your practice nurses which will tell you if your technique is good … the better you can use your inhalers the more drug will reach your lungs rather than your stomach.

Jean Driscoll is a Physiotherapist in Dundee and delivers pulmonary rehab courses locally. She gave an interactive session on the benefits of exercise and discussed practical techniques to deal with breathlessness.

The day finished with a tour of the RRS Discovery followed by a dinner in the evening which gave a fantastic opportunity to get to know each other better.

Devon Mini Meet July 2014

Whenever our members get the opportunity they meet up for a Mini Meet. Norah Oliver who was at the Meet with her husband Bob said "We had a wonderful few days away and it has recharged our batteries for awhile. Thanks to Carol and Joe Lyons for joining us.

We also met the 'nutters' from Exeter in Sidmouth!! Only joking, Joy and John Hehir, you know we love you really. It was great also to meet Julie and Rob Dore for the first time. Although this horrible alpha is part of our life, through this support group we have met so many lovely people who we now think of as friends who otherwise we would never have met"

Members Stories - Linda Cooke

Linda Cooke

I was always what they called a "chesty child ", every cold went straight to my chest and I had lots of chest infections, my Dad use to make cough medicine from sliced onions, vinegar and brown sugar….it would have to left overnight to ferment as my Dad would say before you started taking it.

In the 60's like most teenagers I started smoking...it was considered very grown up and coming from a family of smokers, it just seemed the normal thing to do. Obviously once I started smoking my chest infections got worse and I usually ended up with Bronchitis in the winter, but it never occurred to me to stop smoking, well that was until I had my first baby...my son was about two when my Mum and I decided to give up smoking together as support for each other.

At 40 years of age I went for a company medical, I had no idea how my life was going to change course that day...

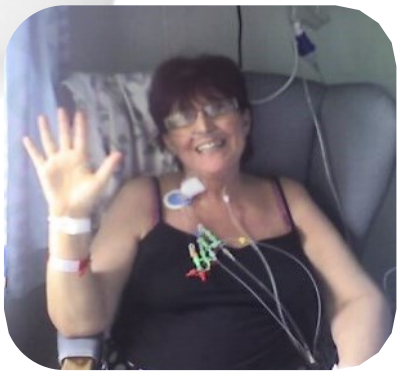
For over 11 years I didn't smoke and my health improved greatly. Unfortunately my marriage broke down around this time, I was in my early 30's and foolishly I started smoking again, of course all the old chest infections and bronchitis came flooding back but this time even worse than before.

I carried on smoking, madness I know but hindsight is a wonderful thing. I tried to give up many times and failed miserably. I knew I was unfit because I couldn't walk and talk, I would get too out of breath I tried to get fitter by cycling to work, only to arrive there looking like I was having some sort of a seizure, gasping for breath.

At 40 years of age I went for a company medical, I had no idea how my life was going to change course that day...after a day of tests which included lung function and chest x-ray the consultant asked if I had ever worked in a dusty environment as in his opinion the x-ray and lung function results showed I had Emphysema ...well I was shocked, in my ignorance I thought only old people got Emphysema.

I went to see my GP who refuted the diagnosis, in fact he said it was rubbish and that I had Asthma and nothing more and promptly prescribed an inhaler, I knew people that had Asthma and were smokers, they seemed ok so I carried on smoking for another year, but that winter I had the worst bronchitis I had ever had and asked my GP for a second opinion, it was at that second opinion where they confirmed that I did indeed have Emphysema, I had heard of a genetic form of Emphysema, and as my Grandfather had died from it I ask the consultant if I might have this genetic problem and could I be tested for it but he refused and said it was highly unlikely that I had it.. I knew I had to stop smoking, it was easier once I knew the alternative prospects.

By then I had changed GP's and my new Dr had a personal friend who was a chest consultant and she referred me to him. The new Consultant received a letter from Professor Stockley saying that he and his team had started a national register for the condition Alpha- 1 Antitrypsin Deficiency which was a genetic condition at the ADAPT research centre Queen Elizabeth hospital in Birmingham in the UK.



Four days after transplant

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My consultant asked if I would like to take part to see if I might have Alpha 1, of course I said yes.

The blood test kit came and was duly done and sent back and the result was I was a ZZ Alpha. In due course my Mum, Dad and both brothers were tested too...they were all MZ . My ex husband and my son were next to be tested and they were found to be MZ (at the time my daughter was too young to be tested)

I was referred to ADAPT and had my annual check ups there, they were very thorough and all the staff were just lovely...once my Daughter reached the right age she was tested and sadly found to be ZZ like me, but I hasten to add both my children now 42 and 33 years of age are both well.

10 days after transplant

My health deteriorated over the coming years and in 2002 I took early retirement on the grounds of ill health. Soon after that I was on oxygen 24/7 and a stair lift fitted in my home, and life became more difficult..
A few years before this I had been referred to Brompton hospital for a possible double

Time for home two and half weeks after transplant

lung transplant but they eventually turned me down saying I was at the time too well.

It was at a 6 monthly check up at my local hospital that I was approached by a new consultant who asked me if I would like to be referred to Papworth hospital in Cambridge for a double lung transplant by now as well as Emphysema I had been diagnosed with Sarcoidosis, Pulmonary Fibrosis and some Bronchiectasis and so I said yes to the referral and so it was in October 2008 that I started a 6 month long assessment.

In April 2009 I was told I had been accepted and placed on the waiting list, and incredibly 9 days later I got ‘ that call ‘ from Papworth.

It was surreal travelling to hospital that day it was all happening so quickly, but I didn't have any doubts in my mind that it was the right thing for me to do, a transplant is not for everyone, many Alphas will never need one and some choose not to have one, it’s a very personal decision, but my quality of life had become so poor I felt I had nothing to loose.

The transplant was a great success and in just over two and a half weeks later I was back home ready to start my new and fantastic life. My gratitude to my donor and her family knows no bounds...the word ‘Thank you’ just isn't a big enough word to use.

Sadly two years ago I was diagnosed with Chronic rejection...but I'm still here, fighting my corner and I haven't given up on the hope of a re-transplant.

I joined the Alpha 1 UK Support Group in the year 2000 and I am now a committee member and trustee.

My first holiday abroad in ten years, taken a year after transplant

From this group I have learnt more about this condition than any Doctor or books could have taught me, I've made so many wonderful friends who have supported me and propped me up through some of my darker days and rejoiced with me when I got new lungs. We are like one big family and feel each others pain and share in each others triumphs.

Members Stories - Lindsay Jarrett

Lindsay Jarrett

For me the biggest thing was diagnosis...or lack of it to be precise. I was diagnosed with asthma at the age of 13 following years of Winters blighted by repeated chest infections and pneumonia.

I joined the police force as an alternative to the armed forces. Armed forces was no longer an option with asthma. The police physical was easy enough although the distance running always caused me to be more breathless.

I was an athletic child despite 'asthma.' I had played football and rock climbed from young. I ran for my school and for the police as a sprinter. As I got older I found that I was developing allergies and sensitivities to multiple things. My breathing was worsening and flare ups were common. Hospital stays became the norm from my 20's onwards. However, in between these bouts of 'asthma' I was a strong and surprisingly fit individual. I also became extremely sensitive to a variety of foods and was a very thin young woman. I was diagnosed with coeliac disease at 30.

My breathing was worsening and flare ups were common. Hospital stays became the norm from my 20's onwards

By this point I had two daughters. Pregnancy proved to be a breathless experience but then I believe it is for most women! In my mid thirties following the birth of my third daughter, I noticed an obvious decline in my breathing. I was struggling to walk on inclines, lift shopping and walk at the same time and running was almost impossible. I thought I was merely out of condition. I started running with a 'runners' pram' and the wee one inside. I made repeated appointments with the GP to be told my asthma was hormonal and that pregnancy could be the answer. Despite the fact that 3 previous pregnancies had not improved my 'asthma' I thought it was worth a last shot. My son Rory was born. I spent almost the entire pregnancy in hospital under the watchful eye of a respiratory consultant. A caesarian section was booked on the proviso that I would not cope with the efforts of labour.

A student doctor said to me "I think I know what is wrong with you." I could have cried with relief. He performed a simple blood test and bingo Alpha-1

Despite birth control methods I found I was pregnant for the fifth time. Termination was not an option but following Findlay's birth I opted for sterilization. I spent this last pregnancy exceptionally weak. Both mentally and physically I felt done. When Findlay was 4 months old a student doctor sat by my bed and said to me "I think I know what is wrong with you." I could have cried with relief. He performed a simple blood test and bingo Alpha-1. I was not mad. I was sick. I was also angry. So, I am a ZZ Alpha who is liver and lung affected.

Oxygen running out...constant beeping to remind me!

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By this point I required perambulatory oxygen as my lung capacity was so poor. My consultant then told me that I required a double lung and liver transplant. That was such a shock. I had not even had time to understand my condition never mind this news. He was telling me I may not survive. My life would be shortened. All I could think about was not seeing my babies grow. It was horrendous.

My marriage broke down and I guess so did I for around 4 months. I was encouraged to attend a hospice as a day patient which I did where I received counselling 'to accept the inevitable.' I was preparing to die.

As time went on my mental strength resumed as did a fight within me. I was not lying down to this!

I was determined to find a way to carry on with my love of sports. The doctors were nervous to say the least and

pulmonary rehabilitation was as good as it got. I felt frustrated. I visited the local climbing wall in my wheelchair. Too weak to carry my own oxygen, I looked longingly at the walls and wanted so desperately to be climbing. I looked at the mountains and knew that I would never ever experience them again. It hurt.

My consultant then told me that I required a double lung and liver transplant. That was such a shock.

Every day I went to the wall. Bit by bit the staff lifted me and encouraged me to try and find ways of extending my oxygen tubing enabling me to have a go. I was tired and breathless but happy. Soon I went to the gym. Trying to build much needed muscle I lifted weights whilst attached to oxygen. I found 4L on constant was what I needed so I naughtily used the back up cylinder from the house as this was all I had to provide me with what I needed. At first this was painful. I was sick. I coughed a lot. It worked like chest physio which was a bonus!
I persevered and pushed through all the pain barriers and breathlessness.

A year on I was a sponsored disabled athlete for three of the largest companies in outdoor extreme sports.

More importantly I was ready to tackle the rock outside!

So, here I am now. I have 22% lung capacity but incredible function. I am positive. I am happy. I rock climb to raise funds and awareness for Alpha-1. People stop to look at this crazy woman hanging off rock attached to oxygen!

Above all I am physically strong and stand a really good chance of a successful transplant.

My official diagnosis is Alpha-1 Antitrypsin Deficiency, Bronchiecstasis, Aspergillosis and asthma plus latex fruit syndrome, coeliac disease and numerous allergies resulting in anaphylaxis.

Lindsay is rock climbing up and down the UK from April to September 2014 to heighten awareness of this condition and to give talks along the way.

Lindsay's Lungs Live Life to the Max

Visit Lindsay's Blog:

ljalpha.tumblr.com/

Fundraising Page:

justgiving.com/LindsayJarrett/

Hello from Australia

Jenni Nankervis

My name is Jenni Nankervis and I have been a volunteer for the Alpha-1 Association of Australia since August 2009.

About the Alpha-1 Association of Australia

The Alpha-1 Association of Australia (AAA) was established on 15 June 2005. It is non-profit Health Promotion Charity, endorsed as such by the Australian Taxation Office.

The AAA was established by Steven Knowles, following recognition of the absence of a central body of information and contacts for those affected by and working with Alpha-1 Antitrypsin Deficiency. We also have an online discussion group, Alpha1-ANZ, which has been established since 9 May 2002. This group is open to Australian or New Zealand residents who are affected by or have an interest in Alpha-1.

The AAA is a relatively small and very quiet group compared to the UK support group, with membership reaching only 146, on our discussion group. Since the fairly recent creation of our Facebook pages we are finding new alphas coming forward to join.

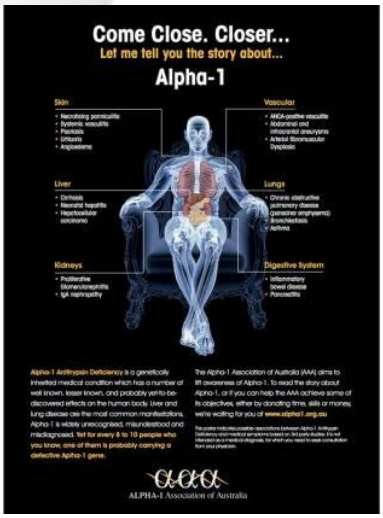
Presently, funds are obtained for the AAA via small functions or events organised by our members. For example, one of our members recently received a cheque from her workplace for a walk she did along with some of her colleagues. Another member Jules, did her Rocky River Run in Rockhampton, Queensland on the 25 May and I will be doing The Melbourne Run walk in July to raise awareness, but to also some funds for the AAA. We have also held sausage sizzles and a raffle, which raised a little money and of course much awareness.

Hopefully soon someone will see the benefit in sponsoring us, so that the AAA can help Australian Alphas more effectively. Regardless of not having current sponsorship, we will keep heading forwards trying to raise awareness of this disorder so that more alphas are diagnosed earlier.

The AAA have achieved some important milestones. For instance, we have been able to have November declared as ALPHA-1 AWARENESS MONTH throughout Australia, which I personally believe will be a great leap forward in raising awareness. We chose November to align ourselves with the Alpha-1 Foundation in the USA, as we think it important to be united with alpha groups around the world.

Most recently, we have aligned ourselves with a group called Rare Voices, Australia. Rare Voices is the unified voice for all Australians living with a rare disease. Many of the groups who have joined Rare Voices have diseases or disorders that are genetic like ours. By working together under the same umbrella, Rare Voices hopes to help raise awareness here in Australia of rare diseases. It is still early days however, this united voice will be helpful in lobbying governments for a National Rare Disease Plan.

The AAA have produced a new eye catching poster. This poster is very informative about the conditions associated with alpha-1.



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Steven and I attended the Alpha-1 Patient Congress in Barcelona in April, 2013 as representatives of the AAA, along with your Karen North and Bev Burroughs, who were of course representing your Alpha-1 UK Support Group. In 2011 we wrote an

Alpha-1 Patient Conference Barcelona

Information booklet and our first poster which can be viewed on our website home page,

Our plan was to print and distribute these to all medical practices around Australia, unfortunately, without appropriate funding this is yet to occur.

We are trying to obtain Newborn Screening and one of our members is currently working on this.

Steven has made much progress on starting up an Alpha-1 Australian Registry and hopefully this will be completed in the near future.

Because of Australia's great size, the AAA are unable to have annual catch ups like you have in the UK, but thankfully we can keep in touch via Facebook support groups, our discussion group and of course via telephone.

I must close by saying that I value the friendship I have with the Alpha-1 UK Support Group, my first contact point when I was first diagnosed.

Bill and I also have such fond memories of the weekend spent with our UK alpha family in Bridgend, Wales in September 2012.

Alpha-1 UK Support Group Meeting Wales 2012

ALPHA-1 Association of Australia

www.alpha1.org.au

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 you support is valued and very much appreciated.

Nicola De Freitas Company Cake Sale Jersey

Kevin Terry Rocking Charity Fundraiser for Research Sussex

Leo Stockley World Bellyboard Championship Cornwall

Mark Ley Lloyds Bank Cardiff Half Marathon

Fiona Rumsby 10K Winter Wolf Run Leicestershire

Charlotte, Bobby and Ted Goode Cake Sale Lancashire

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Mandy Thompson Charity Live Bands Gig Bathgate

Debbie Jayne Plymouth Half Marathon

Diana Penly Brighton BM 10K Marathon

Lindsay Jarrett UK Rock Climbing Marathon

Katy Skillicorn & Deborah Nightingale Bag Sale Dorset

Fay and Mike Whittaker Car Boot Sale Fife

Nichola Griffiths Worcester Half Marathon

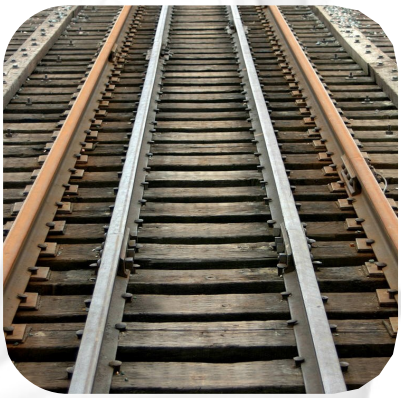
MTB Solicitors Belfast Marathon

Mel Brolly Facebook Auctions for Alpha-1 Cornwall

Continued from Page 15

Evie and Erin Loom Bracelets Cornwall

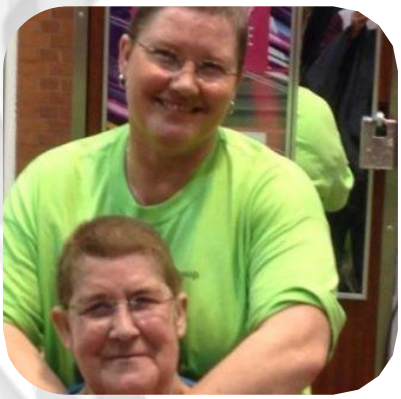
Harry Baxendale Manchester 26 Mile Cycle Ride



Unipart Rail Dress Down Day Cheshire

Charlotte, Bobby & Ted Sand Art Stall Lancashire

Jim & Emma Hunt Summer Fun Day High Wycombe



Pauline Hammons & Julieann Moore Headshave Rhyl

Jasmine Clarke Christening Fundraiser Manchester

Lydia Clarke-McCulloch 10K Run York

Continued from Page 16

Mick & Diane Stobart	Donation
Mrs D Simpson	Donation in memory of her husband who sadly passed away in December 2013
Eveline Jelstrom	Donation in memory of her mother who sadly passed away in February 2014
Elizabeth Booty	Donation in memory of her husband Alan who sadly passed away in March 2014
Emma Hunt	Donation in memory of her brother Paul Smoker who sadly passed away in March 2014
Mrs V Wood	Donation in memory of her son Paul who sadly passed away in April 2014

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

easyfundraising .org.uk

easyfundaising is a free fundraising service where you can raise funds for your charity when you shop online.

easyfundraising works with over 2000 of the UK's best-known retailers including many popular names such as Amazon, M&S, Argos, John Lewis and Vodafone. Just use the links on the easyfundraising site to visit the retailer you want to shop with and when you buy something, up to 15% of the purchase price is donated to our charity – at no additional cost or effort to you! It doesn't cost a penny extra to shop and raise funds for your cause, and as many retailers now give extra discounts when you buy online, you can even save money

easysearch .org.uk

easysearch 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 then receive a percentage of this revenue and pass on a large portion to the cause for which you are searching and supporting.

Please visit our Website for more details: -

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:

infoalpha1uk@googlemail.com

Healthcare Professionals welcome

Alpha-1 Antitrypsin Deficiency and the Liver

(Courtesy of the ADAPT team)

Many patients who come to ADAPT are surprised when we explain to them that the vast majority of alpha-1 antitrypsin is actually made in the liver. In addition, people with alpha-1 antitrypsin deficiency do make a normal amount of alpha-1 antitrypsin protein but the problem arises because the alpha-1 antitrypsin produced is the wrong shape. In people who do not have alpha-1 antitrypsin deficiency, the normal alpha-1 antitrypsin protein is made in the liver, as shown in figure 1. It then enters the bloodstream and travels to the lung to protect it from developing emphysema. The problem for people with alpha-1 antitrypsin deficiency is that the alpha-1 antitrypsin gene is abnormal. As genes are the body's 'instructions' for making proteins, the alpha-1 antitrypsin protein is then made incorrectly. These abnormal alpha-1 antitrypsin proteins have a tendency to stick to each other and form a big chain, otherwise known as a polymer. The polymers are too big to get out of the liver cells and in to the blood stream, and this has 2 consequences. Firstly, most alpha-1 antitrypsin protein cannot get to the lung to protect it from emphysema, and secondly, the polymers that are trapped inside the liver cells can sometimes cause liver disease.

Several different types of liver conditions can be associated with alpha-1 antitrypsin deficiency. A Swedish study has been the most informative in terms of assessing the risk of developing childhood liver conditions. In the study a test was performed for alpha-1 antitrypsin deficiency in 200 000 infants born between 1972 and 1974. They found that 127 had the Z type of alpha-1 antitrypsin deficiency, and these subjects have been followed up every few years to check if they have developed liver or lung disease.

The study found that 14 babies (11%) developed jaundice shortly after birth (neonatal cholestasis) and a further 8 (6%) had other signs of liver disease.

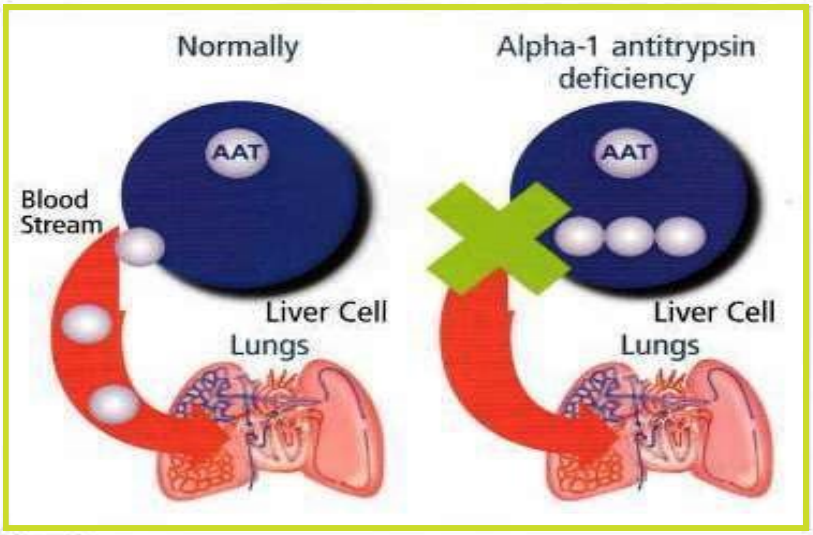


Figure 1

Of the 14 who developed jaundice, 3 went on to get liver cirrhosis between the ages of 2 & 4, but the rest of the babies who had jaundice or had other signs of liver disease got better.

Liver cirrhosis is a condition where the liver becomes permanently scarred and does not function well. Figure 2 shows a normal liver, which is smooth and shiny, and figure 3 shows a liver with cirrhosis that is rough and nodular. The liver has 2 main functions – to make things and to get rid of things from the body. The liver makes blood clotting factors, proteins, sugar and bile, which helps with digestion of fats.

The liver helps to get rid of hormones, waste products, alcohol and some drugs from the body. Therefore, if the liver is not working properly, patients can develop problems like bruising and bleeding (especially from the gut), swelling of the legs and tummy associated with low protein levels, and problems with the sugar level being too low.

Other symptoms can include the loss of body hair and the development of breasts & shrinking of the testicles in men (because the liver is not getting rid of hormones as it should). Itching and confusion caused by waste products being retained in the skin and the brain can also occur.

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You may recall that we sometimes ask you about these things in one of the questionnaires when you come to ADAPT.

Sometimes older adults can develop liver cirrhosis as well as children. The studies that look at this are not as informative as the Swedish study done in children. Many have centred around post-mortem findings, with 15-20% of people with alpha-1 antitrypsin deficiency who died in their 50s, having liver cirrhosis. This figure increases to 40-45% of patients in their 60s. However, this is not an accurate way of assessing the risk of developing liver disease in people who attend ADAPT.

The main reason for this is that post mortem studies are only done when the patient has died. Liver cirrhosis may have contributed to the death in many of these patients anyway, so the numbers seen at post-mortem may be falsely high. In addition, we know that only 1-2 % of adults who attend ADAPT

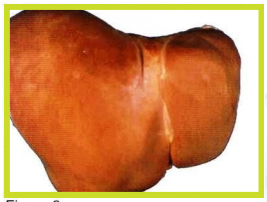

Figure 2

have clinical signs and symptoms of liver cirrhosis, and this may be a more accurate estimation for people with alpha-1 antitrypsin deficiency who are alive now.

At ADAPT, we check liver function blood tests at each visit. We have found that one particular blood test called gamma-glutamyl transferase (GGT) is abnormal in around 27% of patients, despite only 1-2% having liver cirrhosis. We know that drinking alcohol can cause this blood test to become abnormal, and we may have recommended that you reduce your alcohol intake if we have found this abnormality in your blood tests previously.
However, we have also done some new research over the last year and discovered that this blood test also relates to the severity of lung disease, and it may be lung disease that has caused this abnormality in some patients. We measure 3 other liver blood tests at each visit to ADAPT, and these tend to be abnormal in a much smaller proportion of patients (2-5%).
In order to reduce the risk of getting liver cirrhosis, it is recommended that people with alpha-1 antitrypsin deficiency drink no more alcohol than the government recommends. This is currently 14 units per week for women and 21 units per week for men, with one unit corresponding to a small glass of wine, half a pint of beer or a single measure of spirits. If somebody with alpha-1 antitrypsin deficiency has liver cirrhosis, it is best not to drink alcohol at all, and vaccinations against the other liver diseases, hepatitis B and C, are recommended.

Some of the symptoms caused by liver cirrhosis can be treated with various types of medication, but the only 'cure' is a liver transplant. Incidentally, having a liver transplant will also 'cure' alpha-1 antitrypsin deficiency, because the new liver will have normal genes, which will be instructions to make normal alpha-1 antitrypsin proteins, which will not form polymers, and will be able to get into the blood stream to get to the lungs, to protect them from emphysema. However, a liver transplant is a huge operation, and there is a shortage of donated organs. In addition, having a liver transplant will not repair any damage that may have occurred in the lung prior to the surgery. Therefore, liver transplant cannot, unfortunately, provide a 'cure' for alpha-1 antitrypsin deficiency for everyone.

Figure 3

There are, however, new medications being developed to try to stop the abnormal alpha-1 antitrypsin proteins joining together to form the polymers that cause the underlying problems. Work is also taking place at ADAPT, to insert a normal 'M' gene into the DNA of patients with alpha-1 antitrypsin deficiency. If this can eventually be made successful in liver cells, the correct 'instructions' will be available and the normal alpha-1 antitrypsin protein should be made, providing a cure for alpha-1 antitrypsin deficiency.

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Birmingham.
B15 2WB

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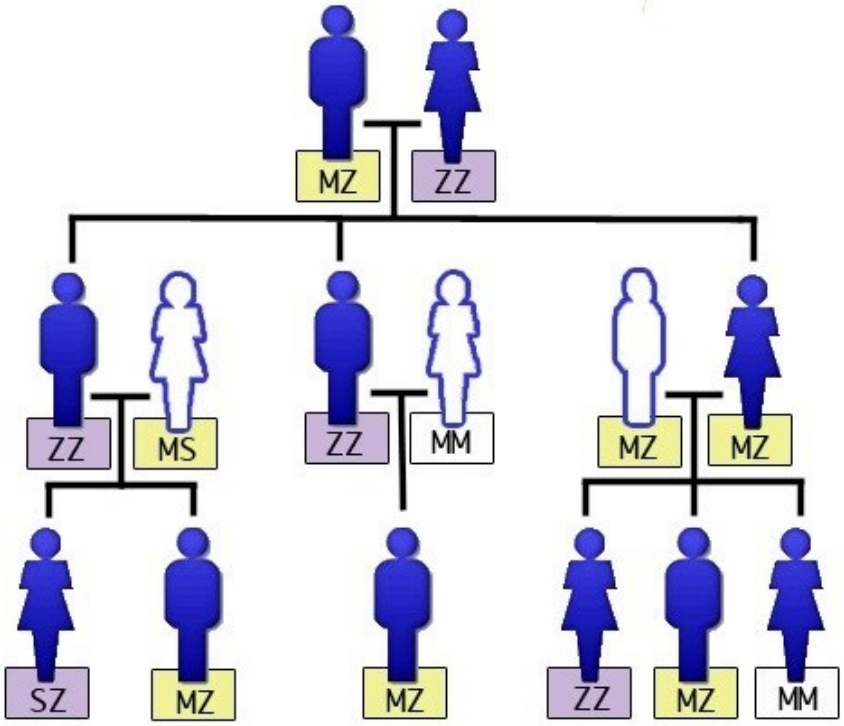
Alpha-1 Antitrypsin Deficiency Inheritance



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 | Chris Torrance Vice Chairman Treasurer Trustee | Karen North Secretary Project Co-ordinator Trustee | Linda Cooke Liaison Officer Trustee | Bev Burroughs Admin Support Trustee | Ray Overton Admin Support Trustee

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 Jemma Coad has accepted the position of Parent Support in addition to her roll as Fundraising and Awareness Coordinator.

We are pleased to announce that Mel Brolly and Fay Whittaker joined the committee as Fundraising and Awareness Co-ordinators. We very much look forward to working with Mel and Fay who will be a valuable asset to our group.

Our Patrons

Professor Robert A Stockley, MD, D.SC., F.R.C.P. Professor in Medicine

Professor of Medicine at the University Hospital Birmingham and Director of Research and Development for the University Hospital Birmingham NHS Foundation Trust.

Professor William MacNee MBChB MD FRCP(G) FRCP

William MacNee is 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 | CSL Behring Biotherapies for Life™

ALPHA-1 FOUNDATION | British Lung Foundation | KAMADA High Quality Pharmaceuticals | Alpha-1 Canada | Children's Liver Disease Foundation fighting childhood liver disease

In Memorium

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

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

by Joe Lyons

Pauline Adams
09/11/2013

Frances Barnsley
24/12/2013

Nathan Heavens
10/03/2014

Alan Booty
17/03/2014

Paul Smoker
22/03/2014

Paul Anthony Wood
12/04/2014

Sally Duff
20/04/2014

Steph Whitbread
24/06/2014

Rob Mckenna
21/07/2014

Donald Fraser
31/07/2014

Supporting alphas, their families, carers and friends since 1997

Alpha-1 UK Support Group
50 Wenning Lane
Emerson Valley
Milton Keynes
infoalpha1uk@googlemail.com

We are a registered Charity

England and Wales (1146330)

Scotland (SC043177)

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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