

Alpha-1 Nor'Easter a Support Group Newsletter (March 2010)

Serving the North Eastern US for the most part!

UPCOMING ALPHA EVENTS

(see "SUPPORT GROUP NEWS " for more info -Page 2)

1. Massachusetts: Thursday, April 8 7:00 PM Topic Explaining PFTs
2. Connecticut: Saturday, April 24. contact Jo Anne for more details
3. The Annual George Washington Bridge Walk, Saturday, May 8. Have fun and help raise money to find a cure.
4. The 19th Annual National Education Conference will be held on June 11-13, 2010 in Orlando, FL at the Hilton Walt Disney Resort. Take a vacation, bring the family it will be an awesome experience! I plan on attending

Saturday, May 8th

Come rain or come shine the **Third Annual George Washington Bridge Walk** is all set. Join John and Fred Walsh, Dr. Eden, Dr. Turino, Lori Tartell, me (Joe Reidy) and the Alpha community, all of whom are bringing family and friends to help raise money to find a cure. I recently heard that another of our favorites, Dr. Robert (Sandy) Sandhaus plans on walking!

Last year we collected \$17,000.00 and had an attendance of 115 people. Our goal this year is to realize \$25,000.00 in contributions and 150 participants. Remember these contributions are being doubled with a matching grant

(If you plan on coming you will need the Detailed Flyer, Directions, Registration Form and Pledge Form). If you receive the E-mail version of this newsletter I have already Emailed you that packet. If you need a packet just contact me (Joe) I can Email (easiest), FAX (easier) or send a hard copy (the only way for some). Joe Reidy, 66 Moore Ave., Waldwick, NJ, 07463 cell (201)264-8941 Email: JoeReidy@verizon.net

DATE	TOPIC	PLACE	CONTACT	
Thur 4/8	Explaining PFTs	Westgate Nursing Home, Needham, MA	SheilaFavaza	1 (978) 468-7704
Sat 4/24	???	Connecticut	Jo Anne Brailey	1 (860) 739-4331
Sat 5/8	A1F Fund Raiser	3rd Annual George Washington Bridge Walk	Lori Tartell	1 (212) 523-5471
6/11-13	19th Annual National Education Conf	Hilton Walt Disney Resort, Orlando, FL	Alpha-1 Assoc	1 (800) 521-3025
Sat 7/17		Connecticut	Jo Anne Brailey	1 (860) 739-4331
Sun 9/26	Educational Brunch	Metro NY	Lori Tartell	1 (212) 523-5471
Sat 10/16		Connecticut	Jo Anne Brailey	1 (860) 739-4331
Tues 12/7	Holiday Dinner	Metro NY	Lori Tartell	1 (212) 523-5471

We would hope and recommend that everyone receiving this notice is a member of the Alpha-1 Association (A1A) and has enrolled in the Alpha-1 Foundation (A1F) Registry. Likewise, if you don't receive the A1F Research Registry Update you are NOT part of the cure for Alpha-1 (shame! shame!). Even if you receive a copy of the Alpha-1 News you are NOT necessarily a member of A1A. Call them just to be sure. 1 (800) 521-3025 Another great publication is the Alpha1 to One magazine contact the Alpha-1 Foundation to get a copy. If interested contact me (Joe) as shown at the end or look at our phone list.

IMPORTANT PHONE NUMBERS: if all these numbers confuse you give me a call 201-444-7839

Alpha-1 Association (A1A)	1 (800) 521-3025	www.alpha1.org	Support
A1A Genetic Counselor	1 (800) 785-3177	courtesy of A1A	Family Concerns
Alpha-1 Foundation	1 (888) 825-7421	www.alphaOne.org	Research
Alpha-1 Coded Testing (ACT)	1 (877) 886-2383	AlphaOne@musc.edu	Free coded (anonymous) testing
Alpha-1 Registry	1 (877) 886-2383	http://www.musc.edu/	Be part of the cure
Baxter Healthcare	1 (800) 423-2090	www.Baxter.com/ Aralast.com	Aralast
Talecris Biotherapeutics	1 (800) 243-4153	www.Prolastin.com	Prolastin
CSL Behring	1-866-936-2472	www.Zemaira.com	Zemaira
Accredo	1 (866) 625-7421	www.Accredotx.com	Aralast/ Zemaira
AlphaNet	1 (800) 577-2638	www.AlphaNet.org	Prolastin/ Zemaira
Prolastin Direct	1 (800) 305-7881	www.ProlastinDirect.com/	Prolastin
Coram Healthcare	1 (866) 367-2174	www.CoramHealthcare.com	Aralast

WORLD WIDE WEB

Most of the following information was gathered at the ALPHA-1 MAILING LISTS. As with everything else in this meeting notice take it with a grain of salt. Remember We do edit some of these E-mail posts and We select which to print. There is much more on the lists. Some of which is very interesting. If you have any questions contact me <JoeReidy@Verizon.net >. Alpha-1 International Support Groups are email networks which Alphas use to rapidly exchange information, support, suggestions, questions, plans, activities; nearly anything that comes to our minds. Many of us have had to reduce our activities and contacts; the Alpha1 contacts are a great way to maintain contact with others. It is also a big psychological boost to know that we are not alone. Here is a list of the E-mail groups to which I (Joe) subscribe:

#	Mailing List	enrollment	owner(s)	TO JOIN: E-mail to:
1	Alpha-1 International	open to most interested in Alpha-1	Connie Storey	ALPHA-1-REQUEST@home.ease.lsoft.com
2	Alpha-1 LIVER	restricted mostly liver affected Alphas & family	Nancy Cropper	ALPHA-LIVER-REQUEST@home.ease.lsoft.com
3	Alpha-1 Lungs & Life	open to all interested in Alpha-1	Sally Turner	Alpha1_Lungs_and_Life-subscribe@yahoogroups.com

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Support Group News

NY/NJ Alpha-1 Support Group Meeting

We are working diligently for a successful walk. See page 1.

Massachusetts Support Group Meeting

DATE: Thursday, April 8, 2010 7pm to 9pm

PLACE: Wingate Nursing Home
589 Highland Ave.
Needham, MA

more info: Susan Binnall 617-916-9805 sbinnall@comcast.net

We are hoping to have someone speaks about PFTs...what the various tests tell you & how to interpret the results

Alpha Nutmeggers (CT) Support Group

Below is the schedule for Connecticut Nutmeggers Support Group meetings for 2010. They are all Saturdays.

April 24th July 17th October 16th

more info: Jo Anne Brailey 1 (860) 739-4331 iluv2sew@snet.net

Alpha-1 Association News

The 19th Annual National Education Conference

will be held on June 11-13, 2010 in Orlando, FL at the Hilton Walt Disney Resort. You can now make hotel reservations online. Registration forms for the conference will be available by the end of January. If you have any questions regarding scholarships to attend the conference, please contact Cathey Horsak at 877-346-3212 or via email at chorsak@alpha1.org.

ALPHA-1 PREVALENCE

From: Joe Reidy <JoeReidy@VERIZON.NET>

Date: March 17, 2010 6:05:19 PM EDT

Subject: [ALPHA-1] Happy St. Patrick's Day

May the wind be at your back. . .

On this fine St. Patrick's Day I came across this interesting article from the Irish Alpha One Foundation. I seems to support what Alpha Angel, Carole Bambrick, told me almost 20 years ago. She always maintained: "Alpha-1 Antitrypsin Deficiency was the scourge of the Irish"

Best to all, Joe with roots from County Clare

The National Alpha-1 Antitrypsin Deficiency Targeted Detection Programme

The Alpha One Foundation, based at RCSI Beaumont Hospital, was established in 2001 to promote research into and awareness of alpha-1 antitrypsin deficiency, to improve diagnosis and treatment and to improve the life expectancy and quality of life in people with Alpha-1. In 2004 Department of Health funding allowed the initiation of a national targeted detection programme and this screening programme is ongoing. We are proud to state this is the only national screening programme for Alpha-1 in the world. The primary objective of our targeted detection programme is to screen for Alpha-1 according to WHO guidelines in the following populations;

* Chronic obstructive pulmonary disease (COPD) * Poorly controlled non-responsive asthma * First degree relatives of known Alpha-1 individuals * Chronic liver disease * Individuals with reduced serum AAT

Recent research from our group has shown that there are 3,000 ZZ Alpha-1 cases and over 250,000 MZ carriers on the island of Ireland but less than 5% of these have been identified. It is worth noting that of the first 8 lung transplants in Ireland since the national

lung transplant programme was initiated, 4 were Alpha-1 patients. The continuing under-diagnosis of this condition is puzzling as it can be diagnosed by a simple blood test. A blood sample can be sent to our laboratory in Beaumont Hospital, and we offer GPs and hospitals a full testing service free of charge. However, thanks to the US Alpha-1 Foundation we can also provide finger-prick test kits, which are more convenient and less invasive than venous blood collection and particularly suited to screening of family members. Once collected the finger-prick sample can be stored at room temperature and sent via conventional post to us free of charge, allowing the patient to test themselves in their own home. Since inception in May 2004 the targeted detection programme has screened over 4,000 patients in the Irish population and identified 64 ZZ patients, 64 SZ patients, and over 600 MZ carriers (Figure 2).

As the national centre we are responsible for increasing awareness of AATD, providing information to patients and health professionals, and hosting educational seminars and conferences on AATD. A website www.alpha1.ie has been designed which is a vital tool for patients and health professionals alike and we have set up a national registry of AATD patients. We host an Alpha-1 National Patient Congress annually in Dublin, with speakers who provide clinical and patient perspectives and experiences. This congress allows us to disseminate information and results arising from this targeted detection programme at a national level amongst clinicians and patients alike. Taken together, these initiatives will hopefully reduce the length of time taken for accurate diagnosis of the condition and lead to the earlier identification of Alpha-1 patients.

In summary, AATD is more prevalent in Ireland than previously thought, with one of the highest incidences in the world. The advantages of early and accurate diagnosis of AATD are manifold and include (1) closer observation and management of affected individuals, especially regarding pulmonary and liver health, (2) family member testing, at least some of whom may have lung or liver complications, (3) aggressive smoking cessation efforts, which have been associated with lower rates of smoking among AAT-deficient individuals, and (4) consideration of occupational hazards and environment as exposures to some occupational dusts and vapors can accelerate pulmonary decline. Once identified, new Alpha-1 patients are offered a referral in our dedicated Alpha-1 clinic with Professor McElvaney. In addition, there is the opportunity to enroll in the AAT replacement therapy clinical trial for ZZ individuals, and to participate in our MZ family study which is attempting to fully clarify the risk of COPD in MZ individuals.

To conclude, the importance of an early diagnosis of AATD cannot be over-emphasised as the resulting appropriate medical follow-up and lifestyle changes can help prevent or at least postpone the development of AATD-related lung and liver disease.

INHALED ALPHA-1

From: John Mugford <john.mugford@googlemail.com>

Date: March 11, 2010 9:00:25 AM EST

Subject: Inhaled AAT Trials ADAPT Birmingham UK - Update

Hi All,

I promised to keep you updated on the inhaled AAT trials in the UK so here goes.

I attended a meeting on Tuesday 9th March where Professor Stockley gave a presentation which lasted about 1 Hour. The drug has been developed by the Biopharmaceutical Company Kamada (Israel) to treat patients with Alpha-1 Antitrypsin Deficiency.

The active ingredient in "Kamada-AAT for inhalation" is called Kamada-AAT, it is a type of protein called Alpha-1 Antitrypsin (AAT), it is the human form of this protein and in "Kamada-AAT for inhalation" the protein is taken via inhalation using a small nebuliser for 15min twice a day. The human AAT protein is taken from human

blood plasma sources that was collected in the USA and approved by US-FDA. The blood is processed and filtered according to strict standards in order to produce the human AAT protein used in "Kamada-AAT for inhalation". Over 90 patients have been treated with "Kamada-AAT for inhalation" in clinical research studies to date and over 70 others have received the same product by intravenous infusion.

This study is a randomised trial. Randomised trials are conducted to compare different treatments (in this study these are "Kamada-AAT for inhalation" and placebo). People who enter the trial are put into different groups and each group is given a different treatment. The results are compared to see if one is better. To try to make sure that the groups are the same to start with, each patient is put into a group by chance (randomly). In this trial, for every one person who is put into the group who will be given placebo, one person will be put into the group who will be given "Kamada-AAT for inhalation". This means that you will have a 50% (1 in 2) chance of receiving the "Kamada-AAT for inhalation" and a 50% (1 in 2) chance of receiving placebo.

This study is a double-blind trial, meaning that neither you nor your doctor will know which treatment group you are in (although if your doctor needs to find out he/she can do so). The study is being carried out in 10-12 hospitals across 5-6 countries and plans to include 220 patients of which 50 will be recruited at ADAPT. The trial will run for 12 months which would include approx 6 patient visits. There is also an electronic notebook which has to be completed each day which cannot be switched off and prompts you when it requires input. The data is then downloaded onto a central database.

I have signed up for the trial and should have my screening within the next 4 weeks.

If you require any further information I will try to answer your questions.

Best regards John L Mugford (PiZZ) Milton Keynes (UK)

From: Joe Reidy <JoeReidy@Verizon.net>
Date: March 12, 2010 9:11:07 PM EST
Subject: Re:Inhaled AAT Trials ADAPT Birmingham UK - Update
Hi John,

First of all thank you for participating in clinical trials. It is so essential in our struggle to find a cure.

Two questions come to mind:
1. What criteria are they using to judge effectiveness? FEV1? CT Scans? or ???
2. Was anything mentioned of a clinical trial using the Talecris product?
Thanks again, Joe from Northern New Jersey

From: John Mugford <john.mugford@googlemail.com>
Date: March 13, 2010 10:08:49 AM EST
Subject: Re:Inhaled AAT Trials ADAPT Birmingham UK - Update
Hi Joe,

I agree it is so important in our struggle to find a cure, this will be my second trial, the first was the Roche Repair 3 years ago which unfortunately was not a great success. I found out after the trial that I was on the drug and not the placebo.

1) - The following is an extract from my patient information sheet which explains the tests and examinations during the visits, a CT scan is carried out at the screening but only to confirm that we have emphysema. However, if a scan has been performed already it will not be necessary:

Visits to the Clinic During the Study*

You will also be required to visit the hospital six times to receive the study drug(s) and for a number of tests and examinations. Your first visit after starting study medication is known as Visit 4 and will take place 2 weeks after starting the study medication. You will need to come for the other visits (5 to 9) on days that are 4, 10, 20, 35 and 50 weeks after you start the study medication. The study staff will arrange these visits with you.

At visits 5 to 9, the following tests will be performed; *

- *Review of the medications you have taken since the previous visit* - * Review of any changes in your health that you may have experienced since the previous visit* - *Physical examination including blood pressure, breathing rate, heart rate temperature measurement.*

The following tests will be performed at visits 4, 5, 7 and 9 (not at visits 6 and 8)

- *Collection of approximately 20mln(about four teaspoonfuls) of blood to perform some tests these will include normal tests of you blood's chemistry as well as tests to check for

certain markers in your blood.* - *A urine sample to check for protein and blood in your urine* - *Pulmonary Function Tests*

*An ECG test will be performed at Visit 4 and Visit 9. An additional collection of approximately 5ml (about one teaspoonful) of blood to perform some tests on your blood to test for antibodies that are specific for AAT (if you have not been treated with the AAT protein before) will be taken at Visit 7

At the Visit 9 (the visit after your last dose of study medication) the following tests will also be carried out: *

- *Collection of approximately 5ml (about one teaspoonful) of blood to perform some tests on your blood to test for antibodies that are specific for AAT (if you have not been treated with the AAT protein before).*

*It is advised that you should avoid taking the following medications for the periods of time before the Pulmonary Function test, Your study doctor can discuss this with you in further detail: *

- *bronchodilators (long acting should be avoided 12hours before the test, and short acting 4 hours before the test.* - *theophyllin (should be avoided 12 hours before the test)* - *antihistamines 12 hours before the test.*

*You will be asked to bring all study medication vials (including empty vials) to your visits, at visit 9 you should also bring the nebuliser and inhalation device and return it to your study doctor.

2) - I said to Professor Stockley during the meeting that I had heard that Talecris were starting inhaled trials in Europe soon, to which he replied "no, they have no plans"***

If you need any further information please let me know.
Take care John L Mugford (PiZZ) Milton Keynes (UK)

VITAMIN D

From: Dutt's Logger <pdlogger@WINDSTREAM.NET>
Date: March 21, 2010 8:47:11 AM EDT
Subject: [ALPHA-1] FYI: "D" GOOD FOR YOU 3/21/2010

Vitamin D helps fend off flu, asthma attacks: By Howard Wolinsky
In a study of Japanese schoolchildren, vitamin D supplements taken during the winter and early spring helped prevent seasonal flu and asthma attacks.

The idea for the study, study chief Dr. Mitsuyoshi Urashima, told Reuters Health, came from an earlier study looking at whether vitamin D could help prevent the bone-thinning disease osteoporosis. The researchers in that study noticed that people taking vitamin D were three times less likely to report cold and flu symptoms.

This led Urashima, of Jikei University School of Medicine, Tokyo, and colleagues to randomly assign a group of 6- to 15-year-old children to take vitamin D3 supplements (1,200 international units daily) or inactive placebo during a cold and flu season.

Vitamin D3, or cholecalciferol, is more readily absorbed by the body and more potent than vitamin D2, or ergocalciferol, the form often found in multivitamins.

During the study, conducted between December 2008 and March 2009, 31 of 167 children taking placebo caught influenza A, the most common form of the virus, compared with only 18 of 167 taking vitamin D.

The vitamin D group was 58 percent less likely to catch influenza A, the researchers report in the American Journal of Clinical Nutrition.

Vitamin D also appeared to suppress asthma attacks in children with a history of asthma. Two children taking vitamin D had asthma attacks during the study, compared to 12 children taking placebo. Urashima admitted to being a bit surprised by this finding and hopes to confirm it in a randomized trial targeting children with asthma.

Dr. Adit Ginde, of University of Colorado Denver School of Medicine, who was not involved in the study, told Reuters Health: "This is the first time a study has been done that rigorously shows that vitamin D supplementation can reduce a type of influenza in a dedicated clinical trial." Ginde and colleagues published a study a year ago showing that asthmatics with lower vitamin D levels were at five times the risk for colds and flu.

In the Japanese study, vitamin D supplementation did not prevent influenza type B, which tends to appear later in the flu season than the "A" flu variety.

Ginde said there is no solid explanation for why vitamin D prevented influenza A and not influenza B. "The immune system fights different viruses in different ways. This finding needs to be explored in more detail," Ginde said.

Based on the current study, giving kids vitamin D

supplements during the winter may help reduce cases of influenza A, the researchers conclude. Urashima suggests that children could take 1,200 IU per day starting in September to prevent flu and asthma attacks during the flu season, but best for parents to check with their pediatrician first.

SOURCE: American Journal of Clinical Nutrition, online Mar 10, 2010.

OXYGEN

From: Dutt's Logger <pdlogger@WINDSTREAM.NET>

Date: March 2, 2010 2:14:18 PM EST

Subject: FYI: OXYGEN CONSERVERS- NOT ACCURATE 3/2/2010

FYI: Give it to me straight boss; right outta da E-bottle. PeterD

ethiopianreview.com | March 2nd, 2010 at 12:53 am
By Amy Norton

Devices that help lung disease patients have oxygen therapy on-the-go may not always perform consistently — and may in some cases provide users with inadequate oxygen when they are active, a new study suggests.

The concern, say researchers, is that patients and doctors may interpret any resulting activity limitations as a sign that the lung disease is worsening, when it could instead be a shortcoming of the oxygen device.

The devices in question, known as oxygen conservers, are used mainly by people with chronic obstructive pulmonary disease (COPD), a group of lung diseases that includes emphysema and chronic bronchitis.

Oxygen conservers attach to the portable oxygen cylinders that many COPD patients carry because they need supplemental oxygen as they walk, climb stairs or perform other daily activities. The conservers are designed to dole out a set oxygen dose each time a person inhales; this allows the cylinders' oxygen supply to last longer than it would if the oxygen flow were continuous.

In the new study, researchers at Case Western Reserve University in Cleveland, Ohio, tested four of what they describe as the most widely used oxygen conservers on the market. (They would not identify the specific brands.)

The researchers first bench tested each conserver to gauge performance, then had 13 COPD patients use each device in random order, at rest and during treadmill walking tests carried out over several weeks.

For comparison, the patients were also evaluated while breathing standard room air and while a wall unit supplied the room with 2 liters of oxygen per minute — the dose each patient required.

Overall, the study found, the conservers' performance varied from product to product. And none of the devices consistently performed up to technical expectations, according to findings published in the American Journal of Respiratory and Critical Care Medicine.

One conserver was no better than breathing standard room air when it came to maintaining patients' blood oxygen levels during rest or exercise, senior researcher Dr. Edward Regis McFadden, Jr. told Reuters Health.

The other three conservers performed better, he said, but patients' oxygen levels dipped when they walked on a treadmill compared with their levels at rest. In contrast, patients' oxygen levels were maintained when the room was supplied with oxygen.

As a group, the conservers' actual oxygen doses differed from what was expected based on bench testing. Two devices consistently delivered less than the expected amount of oxygen during rest and exercise, according to the researchers.

Moreover, they found, the amount of walking each study participant accomplished varied according to which conserver he or she was using.

The problem, according to McFadden, is in the technical aspects of the conservers.

The devices, he said, are unable to keep up with a person's breathing, matching each breath with a consistent dose of oxygen. All of the devices in this study showed "suboptimal activation with breathing," the researchers write.

The findings highlight an "extremely important" issue for patients and doctors to be aware of, said Dr. Barry Make, co-director of the COPD program at National Jewish Health in Denver. Make was not involved in the current study.

At National Jewish, a leading hospital for respiratory diseases, patients prescribed an oxygen conserver undergo testing to

make sure the device provides enough oxygen at rest and during activity, Make said in an interview. A sensor placed on the finger gauges the concentration of oxygen in the blood.

The current findings emphasize the importance of such testing, according to Make. He also pointed out that patients' oxygen needs change over time, and that he and his colleagues routinely reassess how well patients' oxygen conservers are working — at least yearly.

Make and McFadden both said that COPD patients with any concerns about their oxygen conservers' performance should talk with their doctors.

But McFadden and his colleagues also argue that their findings highlight a need for "uniform performance standards" for oxygen conservers — set, McFadden said, by government regulators, based on advice from professional medical organizations.

As it stands, oxygen conservers do not have to go through clinical testing — that is, studied in actual patients — before going on the market.

All conservers, McFadden noted, are considered to be "variations on a basic theme." So they fall under a Food and Drug Administration process that gives clearance to devices based on bench testing and a comparison to existing products.

That is based on the idea that they are all the same, and they will work the same," McFadden said. But the current findings, he said, indicate that "engineering equivalency" does not mean equivalent performance when people use the conservers.

One company that makes the conservers, Elyria, Ohio-based Invacare Corp, said that there are roughly 40 conserver models on the market, and the performance of four cannot be generalized to the entire market.

There is a large, and growing, body of peer-reviewed studies that demonstrate that conserving devices are able to meet the clinical needs of patients at rest, during exercise and even while sleeping," Joseph Lewarski, vice-president of Invacare's Respiratory Group, said in a written response to questions from Reuters Health.

Heather Anusbigian, marketing administrator for Inovo/CHAD Therapeutics, which makes another device, said that "the findings point out the need for further studies pertaining to the efficacy of oxygen conserving devices."

In their own studies, Inovo/CHAD "reached different conclusions than those found in the Case Western Reserve study," she told Reuters Health by email. "Most oxygen conserving device users are able to achieve adequate oxygen saturation levels either during rest or activity." Field testing, she said, "clearly indicates that these devices are working."

Asked about the idea of setting "uniform performance standards" for the devices, Invacare's Lewarski said that "we do and would continue to comply with all regulatory requirements applied to the technologies we develop."

However, Lewarski added, "the challenge associated with standardizing all technical specifications related to oxygen conservers is objectively and scientifically determining what standards are ideal for all devices and patients."

Invacare provided some of the funding for the study. (American Journal of Respiratory and Critical Care Medicine)

GENETIC DISCRIMINATION

From: Dutt's Logger <pdlogger@WINDSTREAM.NET>

Date: March 25, 2010 2:32:09 PM EDT

Subject: [ALPHA-1] FYI: DISCRIMINATION 3/26/2010

Research from Columbia University in the area of genetic diseases published

Research findings, 'Views of discrimination among individuals confronting genetic disease,' are discussed in a new report. According to recent research from the United States, "Though the US passed the Genetic Information Non-Discrimination Act, many questions remain of how individuals confronting genetic disease view and experience possible discrimination.

We interviewed, for 2 hours each, 64 individuals who had, or were at risk for, Huntington's Disease, breast cancer, or Alpha-1 antitrypsin deficiency."

"Discrimination can be implicit, indirect and subtle, rather than explicit, direct and overt; and be hard to prove. Patients may be treated 'differently' and unfairly, raising questions of how to define 'discrimination', and 'appropriate accommodation'.

Patients were often unclear and wary about legislation. Fears and experiences of discrimination can shape testing, treatment, and

disclosure. Discrimination can be subjective, and take various forms. Searches for only objective evidence of it may be inherently difficult.

Providers need to be aware of, and prepared to address, subtle and indirect discrimination; ambiguities, confusion and potential limitations concerning current legislation; and needs for education about these laws," wrote R. Klitzman and colleagues, Columbia University (see also Genetic Diseases).

The researchers concluded: "Policies are needed to prevent discrimination in life, long-term care, and disability insurance, not covered by GINA."

Klitzman and colleagues published their study in the Journal of Genetic Counseling (Views of discrimination among individuals confronting genetic disease. Journal of Genetic Counseling, 2010;19(1):68-83).

For additional information, contact R. Klitzman, College of Physicians and Surgeons, Mailman School of Public Health, Columbia University, Unit 15, 1051 Riverside Drive, New York City, NY 10032 USA.

Publisher contact information for the Journal of Genetic Counseling is: Springer, 233 Spring Street, New York, NY 10013, USA.

AUGMENTATION THERAPY

From: Dutt's Logger <pdlogger@WINDSTREAM.NET>

Date: March 24, 2010 4:29:07 PM EDT

Subject: [ALPHA-1] FYI: INHALE 3/24/2010

FDA letter encourages Kamada

The FDA says the company will receive a response on approval of its first product for the US market by July 1.

Kamada Ltd. (TASE: KMDA) which develops drugs on the basis of human plasma, mainly for respiratory conditions, received encouraging news from the US Food and Drug Administration (FDA) today. The FDA has informed Kamada by letter that it that it will receive a final response on approval of its ATT product, in the version administered intravenously, by July 1.

The letter gives no information about actual approval of the product for sale in the US, but it indicates that the answer will be positive. Kamada's share price rose 5.6% on the news, giving the company a market cap of NIS 703 million.

Alpha-1 Q and A I think I found this at the Alpha-1 Foundation web site but I can't find it anymore.

Q: I am a 40 year old female smoker, just diagnosed with mild emphysema. I am quitting smoking, of course! I asked my docs to test for Alpha-1. They reluctantly agreed.

My dad has pulmonary fibrosis. I also have a new intolerance to alcohol. It seems to make me quite sick now. Although I am not a drinker, I would like the occasional drink! I already have multiple health issues, autonomic dysfunction and coagulation abnormalities, and I am currently being tested for autoimmune illnesses. So, it will take a while before this gets sorted out.

The Alpha1 test results just came and I am a carrier—which I thought meant I was healthy. The Alpha-1 level is 26.8mm, phenotype is PiMZ, genotype was not performed. So, if I am just a carrier, why do I already have emphysema and could this be related to the alcohol intolerance?

What exactly does it mean to be a carrier? My doctor wrote that I am a carrier but my blood level is normal. Then why do I have these problems? And do I need treatment? And should I have my liver investigated further?

A: It sounds like you've had an extensive workup at an academic center. Unfortunately, it also sounds like you are suffering from the conditions you've been diagnosed with. You were certainly correct to request the Alpha-1 testing.

The results present three possibilities.

The first, and the one that needs to be evaluated first, is that the results are incorrect. With coagulation abnormalities and the MZ results, it is important to be sure the result is correct. There is a rare Alpha-1 abnormality, the Mpittsburgh phenotype, that leads to a normal level with coagulation problems. It can sometimes be confused with the M phenotype. If you were PiMpittsburghZ, this is a severely deficient phenotype and should be treated.

It might be wise to send a blood sample to a national reference laboratory such as the [Alpha-1 Detection Lab at the University of Florida](#), with a note about the possible results.

The second possibility is that the results are correct (PiMZ) and that the combination of this mild deficiency and your smoking has led to your early-onset emphysema. People with an MZ phenotype have been shown to have about a 2.3 times increased risk of getting emphysema if they smoke, compared with the general population of smokers. In this case, smoking cessation and avoiding environmental risk factors is the treatment of choice.

The third possibility is the PiMZ phenotype is correct and it has absolutely nothing to do with your early-onset emphysema. There are some people with entirely normal Alpha-1 genes who get early-onset emphysema. We assume that there are other genes involved that have yet to be identified in these people.

Singular, Tylenol are OK for Alpha-1 kids

Q: My daughter has Alpha-1 and was diagnosed with asthma when she was three. Her pediatrician put her on Singular as a preventative. Is Singular safe for children that have Alpha-1? I am worried about its long-term effects. Also, I have always heard that Tylenol is unsafe for Alpha-1 children. Is this true?

A: There are no known problems with Singular in Alpha-1 kids. Tylenol is OK when taken as directed. Make sure that the recommended dose is not exceeded. There are much worse risks in children from using aspirin than Tylenol, even in Alpha-1 kids.

For family testing, genotype or phenotype is better than just testing levels

Q: My father recently was diagnosed as a carrier of Alpha-1 and his doctor recommended my sister and I be tested. We had the test and my level was 104 and hers was 107. The normal range provided on the test was 90-200.

Should we consider having any additional testing done? Or are the levels considered normal and no further testing needed?

A: For family testing, where the goal is to discover whether siblings or offspring might be carrying one or two genes for Alpha-1 Antitrypsin Deficiency, we usually recommend checking a genotype or phenotype, rather than just a level.

The alpha-1 antitrypsin levels that you and your sister were given could mean that you have either two normal genes for Alpha-1 or one abnormal gene and one normal one.

Why would you want to know if you have a single abnormal gene?

Because there is a slightly increased risk of developing lung or liver disease even with one abnormal gene. Perhaps of greater importance, if you have children or are planning on having children, you may want to know so you will be able to determine the probability of passing an abnormal gene on to them.

For more information, see [our web page on testing](#) or call the Alpha-1 Research Registry Program at the Medical University of South Carolina toll-free at 1-877-886-2383.

Lung, liver symptoms call for Alpha-1 testing

Q: I am 51 years old, a female and have been diagnosed with COPD about five years ago. I found this to be extremely odd as I have NEVER smoked in my life. However, I was raised in a home where both parents were chain smokers.

For the past two years I have had elevated liver enzymes, with no explanation. I felt intuitively that the two were somehow connected.

I do not drink alcohol and have never used illegal drugs. Except for the asthma/COPD and some osteoporosis, I am currently OK. My question is, how do I bring this up to my doctor and am I better off seeing an allergy/asthma specialist that I met a while ago? Any guidance would be greatly appreciated.

A: All diagnosed Alphas should consider seeing a pulmonary or allergy specialist with experience in Alpha-1.

But first, you should be tested for Alpha-1. You can ask your current doctor to order an Alpha-1 blood test, based on your symptoms, all of them good reasons to be tested. If for any reason your doctor is reluctant to do the testing, you can see the asthma/allergy specialist you mentioned.

You can also get free and confidential testing by a finger stick done in your home through the Foundation's Coded Testing Study. For information, see [our web page on testing](#) or call the Alpha-1 Research Registry Program at the Medical University of South Carolina toll-free at 1-877-886-2383.

Thank you,

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Alpha-1 Nor'Easter (March 2010) BE PART OF THE CURE!



The James P Mara Center for Lung Disease at St. Lukes/Roosevelt Hospital Center in conjunction with your NY/NJ Alpha-1 Support Group
INVITES YOU, your family and friends to:
THE THIRD ANNUAL ALPHA-1 GEORGE WASHINGTON BRIDGE WALK
Saturday, May 8 (the day before Mother's Day) at 9:30 AM
(More details on page 1)

The NY/NJ Alpha-1 Support Group (not this newsletter) is endorsed by the:

Alpha-1 Association



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