Letter from the Director

By Charlie Strange, MD
Professor of Pulmonary, Critical Care, Allergy and Sleep Medicine
Medical University of South Carolina

Dear Registry Members,

WELCOME to this issue of the Alpha-1 Foundation Research Registry Newsletter. This is a special edition that is being packaged with Alpha-1-to-One to highlight the Registry, an internal program of the Alpha-1 Foundation housed at the Medical University of South Carolina.

For those of you who have received the Registry Newsletter for years, thank you for your participation in Registry studies. For anyone who doesn’t see the Registry newsletter regularly, please consider participation in the Registry if you are a carrier of a deficient alpha-1 gene or severely deficient in alpha-1 antitrypsin (Alpha-1).

Participation in the Registry requires a signed research consent form. You can get one with an email to alphanet@musc.edu or a toll-free telephone call to 1-877-866-2383. The consent form details your involvement with the Registry. The Registry was established to invite those with Alpha-1 to participate in Alpha-1 research. There are many reasons that someone may not qualify for a particular research study. By targeting research invitations to those who meet study details, we advance the mission of the Alpha-1 Foundation to find a cure.

Please keep in mind that much of the research in Alpha-1 is done through surveys that take time, but no financial commitment.

The Medical University of South Carolina is going through some major transformations. Last month the university received the Clinical & Transitional Science Award (CTSA) from the National Institutes of Health. This $20 million award provides research support for databases, electronic surveys, and a training platform for the researchers of tomorrow. Each of these should help the Registry develop.

I invite you to participate in discussions highlighted by Rebecca McClure’s article in this edition of the Newsletter about the ultimate conversion to a paperless electronic registry, for those who desire this option.

2008 was a busy year for the Registry, with eight studies begun. It often takes years from the time a study begins to publication in a medical journal. Therefore, the first of these studies is only now being submitted for publication. We track these studies after the Registry has been used for study recruitment, and will give you the results of the research in future editions. Please note the appreciation given to Registry members by Dawn McGee and Kristin Holm in their articles in this edition.

In the last issue of the Registry Newsletter, we asked for your help in further developing the Clinical Resource Centers (CRC) of the Alpha-1 Foundation. Names of physicians at these centers are given to individuals who call the Registry with requests to find a local physician. The response to this effort has been good; but there are still states without a CRC that we would like to see filled. The map for Clinical Resource Centers is located under the tab for Healthcare Providers at the Alpha-1 Foundation website, www.alphanet.org. We believe that these centers will be important to help enroll individuals in large Alpha-1 research studies in the future. By any measure, the first meeting of CRC members earlier this spring was a success. If your physician is knowledgeable, has an interest in Alpha-1 and might consider becoming a CRC, we would be interested in speaking to him or her.

Lastly, we welcome Jeff Teckman, MD, and his featured CRC in this edition of the newsletter. Jeff has been at the forefront of pediatric liver disease in Alpha-1 for many years. He is a strong advocate of Alpha-1 research and a wonderful physician and educator. Please thank him when you see him for his contribution to this edition. As always, we welcome your ideas for human interest stories, research topics, and directions for the Registry. Please introduce yourself if our paths should cross.

Sincerely,

Charlie Strange, MD
Director, Alpha-1 Foundation
Research Registry
What family and persona the health of an Alpha?

By Kristen Holm, PhD
National Jewish Health, Denver, CO

You can help Alphas cope with COPD by answering an easy questionnaire

AN IMPORTANT STUDY OF ADJUSTMENT TO LUNG DISEASE among Alphas has been underway for the past year and a half and will continue through next January. We have had an excellent response to this study so far and we need your continued support for the final round of data collection, which will occur in January 2010.

The study focuses on health and quality of life among people with Alpha-1 Antithrypsin Deficiency (Alpha-1) who have developed COPD (chronic obstructive pulmonary disease, which includes emphysema or chronic bronchitis) or chronic asthma.

In the first stage of this study, we mailed a questionnaire to members of the Alpha-1 Research Registry with COPD. This questionnaire asked Alphas about their health and quality of life, including symptoms of lung disease and how lung disease affects normal life activities. It also asked about social support, including support from family members.

Two different research teams—one in South Carolina and one in Denver developed the questionnaire. More than 600 people completed and returned this questionnaire. (By returning the questionnaire, these people helped not just one, but both of the research teams that developed it)

While more than half of the respondents said their health was good to excellent, 44 percent said that their health was fair or poor. An important goal of this study is to learn more about who does well over time and why.

We mailed a follow-up questionnaire to everyone who had completed the first one. Four hundred and fifty people returned this questionnaire.

We will mail the final round of questionnaires to everyone who completed the original questionnaire. The follow-up questionnaires are extremely valuable because they provide information about how aspects of peoples' lives have changed or stayed the same over time.

Research such as this project, which track changes over time, can help us learn some of the important facts about people’s lives that are related to improvements in health over time, and which are linked with

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You may contact the Alpha-1 Foundation Research Registry staff by email, at alphagene@musc.edu for additional assistance in locating resources related to AAT Deficiency research, to obtain information about current research activities, to participate in the Research Network or Registry, or to receive Foundation publications.
declines in health over time.

This information will help us to develop new ways of helping Alphas in the future. So if you get a questionnaire in the mail next January, please complete and return it!

Thanks to everyone who has participated in this study. You have all been very generous with sharing your time and your experiences living with lung disease. We appreciate the information that you have provided, and we will use it to help other Alphas in the future.

Are you OK with email from the Alpha-1 Research Registry?

By Rebecca McClure, MLIS
Alpha-1 Research Registry Coordinator

THE INTERNET, World Wide Web, email, mailing lists, discussion boards, texting...

Yes, we've noticed the explosion of online communication in recent years, here at the Alpha-1 Foundation Research Registry. Many businesses use these new forms of communication to cut paperwork and make it easier for their customers to get in touch with them. Here at the Registry, we would like to do the same. Our goal is to have an electronic Registry one day, where we could easily update records, store responses to surveys for future reference, and save time, postage, and trees by reducing our mailings.

We have been asking people for their email addresses since our first application was approved by the Medical University of South Carolina (MUSC) Institutional Review Board in March of 2001. But we have never used those email addresses, for several reasons.

Our concern for the opinions of Registry participants is perhaps the most important reason:

- Are you comfortable with receiving emails from the Research Registry?
- If we send you an email to tell you about a research opportunity, are you comfortable with that?
- How about if we ask you to contact us to update your record - or even let you update your own record online?
- What if we ask you to participate in an online survey?

Another reason we have not rushed into online communication is that not everyone gives us an email address. We want to include as many people as possible in everything we do. The more people who participate, the more likely we are to succeed in our ultimate goal of finding a cure for Alpha-1. We do not want to miss anyone simply because they don't have a computer or an email account. We also don't want to exclude anyone who has decided that it is too risky.

The security of online communication is a legitimate reason for caution, so we work hard to protect participants' confidentiality.

The Registry database is housed on a server maintained by MUSC's IT staff, who keep the server's virus protection, software, and operating systems up-to-date and in compliance with HIPAA Security Regulations. This server itself is protected by the university's electronic firewall, which prevents access from outside the MUSC network, except through a Virtual Private Network (VPN) connection.

A VPN allows computers to connect over the Internet through a "tunnel" that connects just the two computers involved. All VPN messages are encrypted, or coded, so that no one can snoop on the network. This VPN connection is what allows Registry Coordinators to connect to the Registry at Education Days, since the Registry's study protocol requires that no copy of the Registry can ever be transferred from the server to another computer or laptop.

Accessing the Registry at an Education Day requires four passwords, and in the office at MUSC, it still requires two. Each time a Coordinator accesses the Registry, the server logs who is accessing it and at what time, so that there is a record of every use of the Registry. Each Coordinator has a unique user name and password that is not shared with others. If anyone in the Alpha-1 Research Registry is uncomfortable with our procedures, he or she may unenroll at any time. (This has always been true.)

But security is not the only concern. It's already a challenge to keep participants' mailing addresses up to date. Many Registry members have moved and never updated us with their new address. With each new mailing, we discover a few more. There are many electronic services available now that can help to find addresses, and a recent audit of the Registry recommended we use one of these to try to locate these missing participants.

Imagine how hard it is to keep email addresses (which are changed much more easily and often) up to date!

Luckily, we have expert supervision. The Alpha-1 Foundation Registry Oversight Committee, which includes Alpha-1 patients and is chaired by Jim Stocks, MD, must approve all new initiatives in the Registry. This committee takes seriously Registry members' opinions, as we all do.

We invite all members to contact us with their thoughts, questions, and concerns about how you would like the Registry to proceed. We hope that through discussion and collaboration, we can update the Registry for the benefit of the entire Alpha-1 community, including Alphas and researchers. Please send your comments to alphaone@musc.edu or give us a toll-free call at 1-877-886-2383.

We need your opinion on emailing our members
Four generations of an Alpha-Family
By Laura Schwarz
ACT Study Coordinator

Wow!
When Helen Nichols participates in a research survey, she goes full throttle!
Last fall, we mailed a survey headed by Friedrich Kueppers, MD, of Temple University Hospital in Philadelphia, to Research Registry members with the ZZ phenotype, requesting one simple piece of information:

**How many children did your grandparents have?**

We here at the Registry like Nichols' pictorial response to this question so much, we want to share it with you. Here are some of her photos, revealing many members of her family, past and present.

All the photos are numbered. Take a look at them as we run through the family.

Nichols herself (see #10) lives an active life in Billings, Montana. After earning a degree in Vocational Rehabilitation, she became a crisis counselor for teenage runaways. She married a firefighter, and they had two sons (#12 and #13). She became the activities leader at a nursing home, and then drove a school bus for 17 years before becoming disabled from lung disease due to her Alpha-1 Antitrypsin Deficiency.

Nichols was diagnosed with the ZZ phenotype in 1993, after learning that her brother tested positive for Alpha-1 at the University of Utah in 1992.

As you can tell by reading the descriptions of the pictures, she comes from one of those families where Alpha-1 seemed to run rampant. Her mother's genotype was ZZ and her father's was SZ.

It's interesting to note the longevity in her family. Most family members lived well past age 70. Her mother, (#6) a ZZ non-smoker, lived to be 87; her mother's father (#5) lived to be 79.

Most of the men on both sides of Nichols' family were smokers, which accounts for the emphysema diagnosis they were given before Alpha-1 testing began. Many were farmers, a difficult vocation in itself. Imagine anyone with severe Alpha-1 working from dawn to dusk in a Montana winter's below-zero temperatures!

Nichols has always believed that if her brother (#11) had had the advantage of early diagnosis, he may have made different choices in lifestyle and occupation.

Some factors which contributed to his lung disease were smoking and outdoor work in the oil fields in extreme cold, dust, and wind. He was also a truck driver for a time and probably breathed diesel fumes, another risk factor for lung disease. His death at age 42 was not entirely due to the genetic hand he'd been dealt.

When her father (#3) was diagnosed in 1970 at National Jewish Hospital in Denver, someone suggested testing the rest of the family. That didn't happen, however, because of the distance to Denver. Today, fingerstick testing at home has made family testing much easier.

When she looks back at her genealogical research, the phrase "cataarrhal jaundice" from a death certificate speaks hauntingly to Nichols and her sister from an earlier time. Their mother had two sisters who died within a week of birth with what was described as "a swollen abdomen."

Nichols has been an Alpha-1 Association support group leader in Montana for seven years. Her group consists of anywhere from five to 15 members meeting quarterly.

She says her most rewarding experience was when she staffed (by herself) an Alpha-1 booth at the Montana State Fair in Billings. She had borrowed a set of plastic lungs from the Montana Lung Association,
which attracted children to her booth. This brought their parents to the booth, so she was able to explain Alpha-1 to them, using the plastic lungs to demonstrate where it shows up. She manned her booth for 12 hours a day — 11 am to 11 pm — for 10 straight days!

Mary Heberlee, (#9) Nichols’ sister, lives in Pierce, Colorado, a small town about 70 miles north of Denver. She was diagnosed as an SZ in 1993. Luckily, she has no symptoms due to her Alpha-1.

Heberlee participates in 10K runs to keep healthy. Retired from the army after 22 years of service, she currently works part-time as a dental hygienist at Sunrise Community Health Center in Greeley, CO. She has come across quite a few people with Alpha-1 due to her work at the health center. She enjoys talking with them about the condition and encouraging them to have their relatives tested.

Heberlee remembers at seven years old when her grandfather was diagnosed with emphysema and how her family spent time learning the spelling and meaning of this strange word. The doctor said it probably came from using a product called Seresan in his work as a farmer. He would mix this powdery substance with the grain to help precipitate germination. There was no mention of smoking being the cause of his lung disease!

Both sisters want to make the point that Alphas can live long lives when they make the right lifestyle choices – stopping smoking, working and living in clean air, and taking care of themselves with regular exercise and choosing nutritious food.

They also want to promote the importance of Alpha-1 testing to everyone in the medical field — especially to make primary care physicians more familiar with Alpha-1.

Their key point: Everyone with a family history or with lung or liver symptoms needs to be tested sooner rather than later. Testing gives people time to change negative lifestyle habits early, allowing a long and healthy life.

Four generations of Alpha-1 in a Montana family
1. Paternal grandfather, farmer who smoked all his adult life and died of emphysema at 74.
2. Paternal grandmother, non-smoker, died at 78 of emphysema.
The children:
10. is Helen Nichols and
9. is her sister, Mary Heberlee.
3. Father, a farmer, diagnosed SZ, smoked 50 years, died at 64 of emphysema and asthma.
4. Maternal grandfather, farmer, smoked all his adult life, died at 79 of ruptured lung abscess.
5. Maternal grandmother, non-smoker, died at 35 of uterine cancer.
7. Mother’s brother, diagnosed ZZ, smoked 20 years, died at 71 of emphysema.
10. Helen Nichols, ZZ, smoked 13 years, has asthma and emphysema.
6. Mother of Helen and Mary, ZZ, a non-smoker, on oxygen at 76 and died at 87.
9. Mary Heberlee, SZ, non-smoker, no symptoms, participates in 10K runs.
11. Brother, smoked 17 years, worked outdoors in oil fields, had asthma and emphysema, died at 42.
12. and 13. are sons of Helen Nichols, both with allergies, one with asthma.
14. Brother’s son, an MZ.
SAINT LOUIS UNIVERSITY AND CARDINAL GLENNON CHILDREN'S MEDICAL CENTER in St. Louis, MO, have been known for a focus on liver diseases for many years.

Jeffrey Teckman, MD, Director of Pediatric Gastroenterology and Hepatology at Saint Louis University and Cardinal Glennon Children's Medical Center, has been studying liver damage in Alpha-1 since 1994.

His research examines how liver cells are damaged by the alpha-1 protein, and how that damage can be blocked. He and his colleagues have used biochemical studies, as well as tests of promising drugs in humans, to try to develop new treatments for Alpha-1 liver disease.

Teckman is also Associate Professor of Pediatrics and Biochemistry and Molecular Biology.

Teckman also serves on the Steering Committee of the Cholestatic Liver Disease Consortium (CLIC), now part of CHiLDREN, the Childhood Liver Disease Research and Education Network.

This is a study involving various liver diseases and children's hospitals all over the US. The study is funded by the National Institutes of Health and by the Alpha-1 Foundation.

The groundbreaking study will follow several hundred children with Alpha-1 from all over the nation for at least the next 10 years. The goal is to better understand Alpha-1 liver disease and to lay a foundation for new treatments.

Teckman enrolled the first patient in this study, a nine-year-old girl with Alpha-1, in November of 2007. Teckman has been involved in many aspects of the Alpha-1 community. His service includes the Board of Directors of the Alpha-1 Association, the Medical and Scientific Advisory Committee (MASAC) of the Alpha-1 Foundation, the Educational Materials Working Group of the Alpha-1 Foundation, and helping to plan several national patient education days.

Pediatric patients at Saint Louis University are seen through Cardinal Glennon Children's Medical Center. Cardinal Glennon serves patients with all types of financial support, public and private.

Cardinal Glennon has a liver transplant program which includes patients from all over the country.

The team involved in Alpha-1 at Saint Louis University and Cardinal Glennon includes researchers Nancy Marcus, PhD, Keith Blomenkamp, BS, research coordinator Vikki Kocelia, RN, clinical nurses Kim Killebrew, RN and Laura Hotle, RPNP, office staff and GI procedure nurses.

Children with Alpha-1 who suffer from asthma or other lung-related conditions are supported by Cardinal Glennon's pediatric lung specialists, who are experienced in rare lung diseases in children and in the coordination of multi-disciplinary care.

Adult patients with liver disease are served by the Saint Louis University Liver Center at Saint Louis University Hospital. This facility includes clinical care for many liver conditions, including liver transplant, as well as liver research. Liver studies at the Liver Center include specialized drug trials, National Institutes of Health sponsored multi-center research, biochemical studies of the liver, diagnosis and treatment of viral hepatitis, and liver cancer research.

Care is also available for Alphas with pulmonary symptoms or lung disease.

THE ALPHA-1 TEAM at St. Louis University and Cardinal Glennon Children's Medical Center, an Alpha-1 Foundation Clinical Resource Center: Thomas Foyle, MD, Jeffrey Teckman, MD, Erica Blackmon, Laura Hotle, RPNP, Kim Killebrew, RN, Keith Blomenkamp, BS, Emily Hermann, and Nancy Marcus, PhD.

Contact information:
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Emily Hermann, 314-577-5547
Nurse Coordinator, Kim Killebrew, RN, 314-268-2700 ext. 6114

Contact information for ADULT liver patients:
Phone (314) 977-6150 (for patient appointments)
Adult GI and Hepatology Clinical Research Unit
Phone (314) 977-9400 (for information regarding clinical trials)
Can stem cells be used to grow a new, healthy liver?

By Jeffrey H. Teckman, MD
Director, Gastroenterology and Hepatology, Cardinal Glennon Children’s Medical Center, St. Louis, MO

Q: I have a son who has developed liver disease due to Alpha-1. I am told he will need a transplant eventually. I am wondering if there are any studies being done that could "cure" Alpha-1.

There have been so many stories lately about stem cells from a healthy sibling’s umbilical cord curing genetic disorders. Is that a possibility for Alpha-1 patients?

A: Unfortunately, at present we have no “cure” for Alpha-1.

There is a lot of research going on. We are testing potential treatments in test tubes in the lab, in animals, and gathering information from patients through activities like the registry and the "CLIC" study.

In the past there have been trials of experimental treatments for the liver disease in humans, although there are no such tests are going on now. Using techniques to "regrow" the liver with stem cells or special techniques called "gene therapy" are being studied, but so far they are not shown to be effective or safe in humans. Many researchers are working on questions like this, so if we continue to support research we may have better options in the future.

Q: I am 37 years old and I have Alpha-1 (I do not know the genotype; I was diagnosed from the level of alpha-1 in my blood). Recently, my doctor found out that the level of platelets in blood was too low (around 100,000 instead of at least 150,000). Could this be related to Alpha-1 Antitrypsin Deficiency – in particular with liver-associated complications? To what extent should I worry about this low level of platelets?

A: Platelets are small blood cells which help with blood clotting.

If the liver becomes damaged, from Alpha-1 or from any cause, then scar tissue can build up in the liver. If the scar tissue becomes extensive, this is called "cirrhosis." Cirrhosis can become so severe that blood flow through the liver becomes partly blocked.

Since all of the blood from the intestines and some internal organs goes through the liver on the way to the heart, any blockage in that flow can be a problem. Sometimes the blood can "back up" into the spleen (another organ in the abdomen) or into the intestines. This is called "portal hypertension." Portal hypertension can result in swelling of the spleen ("splenomegaly"), bleeding in the stomach or intestines, or fluid accumulation in the abdominal cavity ("ascites").

Another possible result of portal hypertension is that platelets will get stuck in the spleen. There are still plenty in the person's body, but they are not out free in the circulation and so they come up low on the blood count result. Since I don’t know your specific situation, I don’t know if this is what is going on in this case, or if there is another reason. However, this is a common problem in various kinds of liver disease. I suggest you discuss this with your doctor.

Q: What is the role of nutrition in alpha-1 and liver disease?

A: Proper nutrition is important in all aspects of health, including lung and liver disease. For most people there is no special "Alpha-1 diet." It is important to eat a diet moderately low in fat and salt and rich in vegetables, fruits, lean meats, fish, etc. Abstaining from alcohol consumption is important for people with any kind of liver disease.

In some situations when people have very severe liver problems, doctors restrict salt and protein. This is very individual and people should talk to their doctors on a case-by-case basis. It is also important to keep a healthy weight. Obesity is bad for the liver. It is also important, if lung or liver disease is severe, to try to keep from losing too much weight. Some people will get supplemental feedings, or other special arrangements when, for example, they are waiting for an organ transplant.
We're Building Friends to match

By Angela McBride
Director of Development
Alpha-1 Foundation

THE ALPHA-1 FOUNDATION hosts the First Annual "Alpha-1 Walk & Run Miami" Nov. 8.

We expect to have about 150 Alphas, friends and family, community leaders and healthcare providers attending.

We're inviting everyone to visit the Alpha-1 Foundation offices Saturday, Nov. 7, followed by a "Meet the Doc" session to hear about our current research projects. Visitors can register early for the event and enjoy hors d'oeuvres.

Then next day – Sunday, Nov. 8 – we begin our Walk and Run in Crandon Park, Key Biscayne, FL. For information, contact Angela McBride at 888-825-7421, Ext. 233 or amcbride@alphaone.org

This is a new event for the Alpha-1 Foundation's Building Friends for a Cure Program, which since 2005 has been raising a steadily increasing amount of funds for research and programs.

This year, we have a unique opportunity to double every dollar donated – up to a million dollars.

Our Million Dollar Match is made possible by a matching grant from Talecris Biotherapeutics. All administrative and fundraising costs are being covered by a portion of AlphaNet's contribution to the Foundation, so 100 percent of donations will be restricted to research programs (and doubled by our matching grant).

And Building Friends for a Cure is helping us to raise that million to be matched.

Team Alpha-1, under the Building Friends for a Cure umbrella, had its biggest year ever in 2008 at the ALA of Massachusetts Autumn Bike Trek – the "Escape to the Cape." The team raised more than $70,000 and earned the first "Silver Spoke Award" for having the most riders.

This year, the goal was to top themselves. The 2009 "Escape to the Cape" bike trek was held Oct. 2-4, as this issue of the Registry Update was going to press.

But don't think you need to match that huge success. We encourage you to start small – especially if the whole idea of fundraising seems a bit intimidating.

You might join our Firstgiving online fundraising campaign at http://www.firstgiving.com/alpha-1foundation. The website makes it easy to create your own personal web page.

For ideas, a few examples of Firstgiving pages:

David Moody, an Alpha from Kingsport, TN, planned to run in the Eastman 10K Road Race Sept. 12 this year. He got a checkup because he was having chest pains during training runs, and his doctor said they were due to his asthma and Alpha-1, and "No more running!" Walking is much better exercise for David, the doctor said.

So David just entered the 3K walk at the same event, which has many participants, too. His page: www.firstgiving.com/roberdtimothymoody

Kimmie Tulip and Roger Greene celebrated their wedding by welcoming donations to Alpha-1 research and programs, in honor of Roger's father, Roger S. Greene Sr., who died of emphysema.


In June, Tom and Liz Corron and Iowa support group leader Peg Iverson organized "Get the Scoop on Alpha-1," an ice cream social in Des Moines, IA. Their page: http://www.firstgiving.com/scoop

The goal of the Alpha-1 Foundation's Building Friends for a Cure Program is to build stronger links between the organization and the Alpha-1 community using social events to increase awareness and raise funds for Alpha-1 research and programs. For information, contact Angela McBride at 888-825-7421, Ext. 233 or amcbride@alphaone.org.

Team Alpha-1 was created to encourage participation of Alphas in athletic events to promote awareness and raise funds in support of research and programs. The sponsors are Baxter Healthcare, CSL Behring and Talecris Biotherapeutics.

GET THE SCOOP ON ALPHA-1 – This bright, simple little graphic helped Tom and Liz Corron and Iowa support group leader Peg Iverson get attention for their June ice cream social, "Get the Scoop on Alpha-1," in Des Moines, IA.

See the Building Friends for a Cure 2009 Calendar of Upcoming Events on page 12.
for a Cure, and helping $1 million

alpha - 1
walk and run
Miami

5 K walk & run, November 8, 2009, Crandon Park, Key Biscayne

RICHARD AND SARAH JOHNSON of Jacksonville, FL, and their children will be among the families attending the First Annual Alpha-1 Walk & Run Miami Nov. 8. Grace, 3, and Lucas, 1½, both have Alpha-1, though only Lucas has symptoms.
Health care reform that covers all, protects Alphas

By Miriam O'Day
Senior Director, Public Policy, Alpha-1 Foundation

Health care reform remains a top priority for the Obama administration.

While Congress has yet to present a single plan for comprehensive health reform, there are several bills working their way through the process in both the House and the Senate. All of these bills attempt to define standards for health benefit plans; list services that must be offered to individuals; establish subsidies for lower income households; and create some type of mandate for coverage.

The Alpha-1 Foundation and Alpha-1 Association both believe that it is very important for the concerns of Alphas to be heard while these bills are developed. The organizations are unable to take a firm position now, because no single piece of legislation has emerged. We have expressed the concerns of the Alpha-1 community by working with coalitions to establish principles of health reform that we believe will help those living with rare chronic disorders. For Alphas, access to augmentation therapy and transplantation are critical concerns.

In the health care reform debate, we have pressed Congress to support the development of new therapies for rare disorders; provide special concessions for review by rare disorder experts in the use of Comparative Effectiveness Research; and promoted better access to allied health professionals such as respiratory therapists.

By working with the National Health Council (NHC), the Genetic Alliance, and the Plasma Users Coalition, we have been able to identify many important principles that we believe should guide the development of health reform.

Along with the NHC, we believe that health reform should cover everyone, curb costs responsibly, abolish exclusions of pre-existing conditions, eliminate lifetime caps, and ensure access to long-term and end-of-life care.

We also support the Genetic Alliance's principles. These are to create universal access to optimal care, realign financial incentives to center on the health of people, coordinate health care delivery, empower patients by creating an individual sense of ownership and responsibility for their health, link research and healthcare systems, and increase research focus on quality of life and health outcomes.

The principles of the Plasma Users Coalition (PUC) have a particular significance for Alphas. The high cost of augmentation therapy makes affordability very important, particularly limiting out-of-pocket and co-pay costs to set dollar amounts and not percentages, as is so common with private insurance now.

PUC principles also include access to specialists with knowledge of the diagnosis, treatment, and management of rare diseases; access to therapies as decided upon by physicians and their patients; and support of comparative effectiveness research which goes beyond cost effectiveness studies.

Health care reform legislation will very likely be passed and signed into law before the end of the year. There is still time to contact your legislators to express the need to bear in mind chronic rare conditions and the special needs of Alphas when considering health reform.

Let your voice be heard while the debate is still in process!
Learning more about the "S" gene:  
the PiSS Genotype Study

By Dawn McGee, MS, CGC  
Program Director, Alpha-1 Association Genetic Counseling Program,  
Medical University of South Carolina

A variety of different genes are involved in Alpha-1, which is one thing that makes studying the condition difficult.
Most people know about the PiZZ genotype. The "Z" gene is the one we most often hear about, when someone has the severe deficiency.  
Much less is known about diseases associated with the "S" gene, however.

Three genotypes that carry an "S" gene have been studied.

THE PISS GENOTYPE has been shown to produce very low levels of alpha-1 antitrypsin (AAT) in the blood and is associated with chronic obstructive pulmonary disease (COPD). Some people with PiSS choose to receive augmentation therapy. The PiMS genotype is slightly more common than the PiMZ genotype and has blood levels of AAT very close to normal. Although some studies have suggested that the PiMS genotype increases the risk for asthma, we can't draw that conclusion yet.

The PiSS genotype has been the least studied of all the "S" genotypes.

Only studies performed through large groups of people can make any statement about the PiSS genotype. In the United States, it is estimated that approximately 1 in 1058 individuals have the PiSS genotype.

We were able to telephone 19 people who joined the Registry or were found to have PiSS through the Alpha Coded Testing (ACT) Study. Participants from the ACT study identified as having the PiMM genotype were matched to the study participants for comparison.

The aims of the study were to investigate features of the PiSS genotype, such as possible lung and liver symptoms, other diagnoses, and medication and allergy histories. Since the AAT concentration associated with the PiSS genotype is similar to the concentration associated with the PiMZ “carrier” genotype, the current understanding is that the associated risks for these two genotypes are similar.

This study involved answering a questionnaire over the telephone. The questionnaire asked questions about lung and liver symptoms, smoking history, medication use, race and ethnicity, other diagnosed conditions, allergies, and surgical history. There were also some unique questions, including ones about vocation and artistic experience, pregnancy history, and three-generational family history.

We asked questions about vocation, artistic experience, and psychiatric diagnosis because a recent published study discussed psychiatric diagnoses in combination with artistic experience in individuals with at least one S gene for Alpha-1.

We also asked about pregnancy history in family members, specifically about miscarriages and stillbirths, to investigate whether the PiSS genotype is associated with a decrease in live births. This study provides additional information about the clinical description of the PiSS genotype and is the largest case series reported to date.

Our research study suggests there may be lung symptoms associated with the PiSS genotype. However, there was also a high degree of lung diagnoses in the control population. While the study provided further information about the PiSS genotype, it also indicated that further research needs to be done with larger numbers of people in order to fully understand this rare genotype.

The research team sincerely thanks all the participants of the ACT Study and the Alpha-1 Research Registry that graciously took the time to answer our questions and participate in this study. It is your participation that leads us to a better understanding of Alpha-1.

A recent published study discussed psychiatric diagnoses in combination with artistic experience in individuals with at least one S gene for Alpha-1.
Upcoming Events, Conferences and Education Days
For the most up-to-date listings, check our website at www.alphafone.org.

### Building Friends for a Cure 2009 Calendar of Upcoming Events

<table>
<thead>
<tr>
<th>When?</th>
<th>What?</th>
<th>Where?</th>
<th>Who?</th>
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</thead>
<tbody>
<tr>
<td>Ongoing</td>
<td>&quot;In Blue&quot; A Cross-Stitch Angel</td>
<td>Holland, MI</td>
<td>Ken Walkley <a href="http://www.firstgiving.com/kenwalkley">www.firstgiving.com/kenwalkley</a></td>
</tr>
<tr>
<td>Nov. 8</td>
<td>Alpha-1 Walk and Run Miami</td>
<td>Miami, FL</td>
<td>Angela McBride <a href="mailto:amcbride@alphafone.org">amcbride@alphafone.org</a></td>
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<tr>
<td>Nov. 14</td>
<td>Swirl for Alpha-1</td>
<td>Newbury Park, CA</td>
<td>Karen Erickson <a href="mailto:kerickso@amgen.com">kerickso@amgen.com</a></td>
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### 2009 National Education Programs

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
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<tbody>
<tr>
<td>Nov. 14</td>
<td><strong>Alpha-1 Education Day</strong></td>
<td>Ann Arbor, MI</td>
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<tr>
<td>June 10-13, 2010</td>
<td><strong>2010-19th Annual National Education Conference</strong></td>
<td>Lake Buena Vista, FL</td>
</tr>
</tbody>
</table>

The Alpha-1 National Education Series is co-sponsored by the Alpha-1 Foundation and the Alpha-1 Association and is made possible by unrestricted education grants from AlphaNet, Baxter BioTherapeutics, Centric Health Resources, CSL Behring, and Talecris Biotherapeutics. For information on attending or exhibiting at an education program, contact Marlene Erven at 1-800-521-3025 or email [mserven@alphafone.org](mailto:mserven@alphafone.org). For information on Building Friends for a Cure events, contact Angela McBride at 1-888-925-7421, Ext. 233 or [amcbride@alphafone.org](mailto:amcbride@alphafone.org).

*Commitments and dates are subject to change.

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**Alpha-1 Foundation**

The Alpha-1 Foundation is a not-for-profit organization dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1). The Foundation has invested $35 million to support Alpha-1 Antitrypsin (AAT) research and programs in nearly 70 institutions in North America and Europe.

**Alpha-1 Association**

The Alpha-1 Association is a member-based not-for-profit organization founded in 1991 to identify those affected by Alpha-1 Antitrypsin Deficiency and to improve the quality of their lives through support, education and advocacy. The Association has a network of 75 volunteer-led support groups around the U.S.

**AlphaNet**

AlphaNet, Inc. is a unique disease management organization. Through its medical and operations staff, AlphaNet provides a wide range of integrated support services to individuals with Alpha-1 Antitrypsin Deficiency who require augmentation therapy, oversees and sponsors clinical trials involving Alpha-1 therapies, and makes available a comprehensive disease management and prevention program to improve the quality of life of those affected by Alpha-1.