Some new studies, and ways to cut infections

Letter from the Director

WELCOME to this edition of the Research Registry Newsletter. We have continued to list the active studies enrolling in the Alpha-1 Community on the Registry website at www.alphaoneregistry.org. If you have not visited recently, please take a look. Our website is one way to keep you updated between newsletters.

In this edition you will learn about some of the new studies in Alpha-1 and lung disease. These include a study for people with NTM lung infections, and the first bronchoscopic lung volume reduction study in the U.S. that includes Alphas. We also review some good health habits that may reduce lung infections and exacerbations, results from completed Alpha-1 research studies, advice for caregivers, and more.

Thank you for being a member of the Research Registry. Because of you, we are able to learn more about Alpha-1 and work toward better treatments through research. Without you, this research would not be possible.

A growing number of Clinical Resource Centers (CRCs) are joining the CRC Research Registry Network. We are excited to introduce two new sites to you in this newsletter. If you visit a participating CRC site, please consider being part of this important project.

One of the important initiatives for the coming year will be an emphasis on the family. We estimate that only 10% of people with Alpha-1 have been diagnosed. Alpha-1 is a genetic disease, so your family members should be offered testing. Remember, it’s all in the family. The Alpha-1 Coded Testing (ACT) study offers free and confidential Alpha-1 testing that you can do from home. Please let your relatives know about this easy way to be tested. Our genetic counselor is here to help with any questions and we invite anyone with abnormal Alpha-1 genes to join the Registry. Family testing is an important way to help the Alpha-1 community and those closest to you.

Since our last newsletter, the MZ Carrier Genomics Study has completed enrollment. The goal of this study was to better understand why some MZs develop lung disease while many other MZs do not. Stay tuned, and we look forward to sharing the findings with you after the data analysis is complete.

The Alpha-1 team at MUSC is pleased to have two students assisting our coordinators with the ACT study and Alpha-1 research. You heard about the importance of exercise from Alysha Carlos in the last edition. In this edition Alison Garbarini tells you why brushing your teeth may be good for your lungs.

Lastly, we invite you to mark your calendars for Oct. 29, 2016. The Alpha-1 Foundation and the MUSC Alpha-1 team will be hosting a Regional Education Day here in sunny Charleston, South Carolina. This will be a great opportunity to learn from Alpha-1 experts, meet us in person, and socialize with other Alphas. Bring your families and your questions.

Sincerely,
Charlie Strange,
MD,
Director, Alpha-1 Foundation
Research Registry and ACT Study
Two new sites help to build new CRC Research Registry

By Deirdre Walker
Registry Coordinator

Research over the past five decades has greatly advanced our understanding of Alpha-1 Antitrypsin Deficiency. Advances include understanding the genetics of Alpha-1, creating treatments and learning more about emphysema and liver disease related to Alpha-1. It is our hope that we can continue to grow the Alpha-1 Research Registry and continue helping researchers find better treatments and a cure for Alpha-1. Thanks to you and people like you in the Research Registry, we have successfully enrolled participants in over 75 studies.

As you may have heard, a new endeavor of the Alpha-1 Foundation is the Alpha-1 Clinical Resource Center (CRC) Research Registry.

The aim of this project is to improve the current registry by increasing the amount and quality of patient information that is gathered, acquiring lung function tests and collecting biological samples from willing participants. The CRC Research Registry will make this information available in a new format to investigators and others to further the understanding of Alpha-1.

Would you like to be part of our new, improved CRC Research Registry? Here's how you can.

This new Registry’s more specific format for data requires you to visit a CRC and see an Alpha-1 doctor to join. The visit will also include meeting with a study coordinator to go over the consent form, collection of medical information, and reporting family history and other information with online questionnaires. The questionnaires will ask you about exposure to cigarette smoke, other dust and fumes, as well as your symptoms and quality of life. The questionnaires take about 30 minutes to complete. You will be offered the opportunity to donate a blood sample to a biorepository where blood and other biological materials are stored for the purpose of use in future research.

This is why we believe the new CRC Research Registry will be even more useful to Alpha-1 research than the current Research Registry: Clinical data obtained in a medical office, rather than patient-reported data, offers advantages in better understanding.

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You may contact the Alpha-1 Foundation Research Registry staff by email, at alphaone@musc.edu for additional assistance in locating resources related to Alpha-1 research, to obtain information about current research activities, to participate in the Research Network or Registry, or to receive Foundation publications.
Welcome to Provincetown

Andrew Wilson, MD, center, and the team from the Alpha-1 Center at Boston University and Boston Medical Center, celebrate completing the “Escape to the Cape” bike trek. The team took part in the annual bike trek as a fundraiser for the Alpha-1 Foundation’s research programs.

the Alpha-1 population and disease progression in years to come.

Those who join the CRC Research Registry will receive invitations to participate in Alpha-1 research and receive our twice-a-year newsletter, exactly the same way that current Registry members do. No member of either Registry is ever required to participate in any study, and participants’ names and addresses are never given out to any researchers. Before you begin any study, a complete explanation of the research will be presented to you so that you can make an informed decision about whether or not to participate.

The Alpha-1 CRC Research Registry is hosted at the Medical University of South Carolina (MUSC). To date MUSC has enrolled 32 participants in the CRC Registry. We are hoping to create a multi-site national CRC network, so Alphas around the country can join the CRC Research Registry at a site convenient to them. More new CRC sites are expected to join this CRC Research Registry network every month.

We are excited to announce that Boston University and The Ohio State University are the first to partner as CRC Registry sites and are now enrolling participants.

Boston University,
Boston Medical Center

Andrew Wilson, MD, and his co-director, Darrell Kotton, MD, at the Alpha-1 Center at Boston University and Boston Medical Center, have been working together on a project to develop a gene therapy for Alpha-1 since 2004. The emergence of a kind of stem cell (called induced pluripotent stem cells or “iPSCs”) as a powerful research tool has allowed Wilson and Kotton to study Alpha-1 in a dish using stem cells from Alpha-1 patients. These researchers are able to coax these cells to turn into liver or lung cells.

Darrell Kotton, MD

Continued on page 4
the organs most commonly affected by Alpha-1), and in doing so reproduce key disease features in the lab. This allows them to gain a better understanding of how the disease works and the effects of possible treatments.

In addition to the iPSC studies, their Alpha-1 Center is also involved in clinical studies like the Alpha-1 Foundation Liver Study (enrolling 22 patients to better understand Alpha-1 liver disease).

The Alpha-1 Center clinic opened in 2012 and has now seen over 50 patients with Alpha-1. Both Wilson and Kotton are heavily involved in the Alpha-1 patient community and regularly interact with patient support groups in the region. The Boston CRC integrates patient care, basic science stem cell research and clinical studies in a Center with a deep interest in caring for Alphas and their families. You can meet their clinical team and contact the Alpha-1 Center at Boston Medical Center by visiting www.alpha-1center.org or by calling 617-638-7680. Mark Dodge, study coordinator at BU/BMC leads the center’s clinical research efforts and can be reached by email (mdodge1@bu.edu) or by phone at 617-414-2968.

Ohio State University
Wexner Medical Center

The Wexner Medical Center at The Ohio State University (OSU) has provided services to the Alpha-1 patient community in Ohio since the early 1980s. The OSU program was a major recruitment center for the first Alpha-1 Registry created by the National Heart, Lung, and Blood Institute for its longitudinal study of Alpha-1 from 1989-96.

The center’s directors have played historic roles in studies related to therapy for Alpha-1 patients over the years. James Gadek, MD (retired), the first to recruit Alpha-1 patients to Ohio State, was the first physician to test Alpha-1 augmentation therapy as a feasible treatment for Alpha-1 lung disease. Mark Wewers, MD, his OSU successor, has carried on that tradition, providing clinical service and advice to Alphas at Ohio State for three decades.

While at the Pulmonary Branch of the National Institutes of Health (NIH) in the mid-1980s, Wewers led the pivotal study of augmentation therapy under the direction of Ronald Crystal, MD, chief of the Pulmonary Branch. This study led to the U.S. Food and Drug Administration (FDA) approval of augmentation therapy in December of 1987.

Wewers is an advocate for Alpha-1 patients who have to make difficult decisions regarding how to approach this challenging condition, including issues such as the Alpha’s own prognosis, family risks and the economic issues complicating therapy options.

The OSU Alpha-1 Research Registry Center is located at the Martha Morehouse Medical Plaza in Columbus, Ohio, and provides support for the Center work within the Pulmonary Clinical Trials Office. This Center also offers state-of-the-art outpatient lung function testing and convenient parking. If you would like to visit The Ohio State University to enroll in the Alpha-1 CRC Research Registry, contact Karen Martin at karen.martin@osumc.edu or call 614-366-2934.

You can learn more about the Alpha-1 CRC Research Registry by visiting our website, alphaneregistry.org.

Mark Wewers, MD, in Ohio State lab with researcher Anasuya Sarkar, PhD.
Some of you may remember being invited last October to answer a survey from the University of Florida about exercise and pulmonary rehabilitation among Alphas. We received over 500 responses—thank you!

Pulmonary rehab is a structured and monitored exercise program designed to help people with long-term lung disease. Pulmonary rehab also uses nutritional counseling, education and breathing techniques, in addition to exercise training, to accomplish three main goals:

- Help with your shortness of breath
- Improve your quality of life
- Improve your day-to-day activity level

Nearly everyone agrees that exercise is good for you. You may have read an article in our last Registry Update about why you shouldn’t give up on exercise.

Exercise improves shortness of breath, helps to lower blood pressure, reduces anxiety and improves sleep. Exercise also improves blood circulation, which makes it easier for the body to absorb oxygen. But despite the numerous benefits of exercise, many people with COPD remain physically inactive. This is likely due to shortness of breath, leading to inactivity, which leads to physical decline in function, which can affect nearly all systems of the body. This vicious cycle can be broken!

According to the American Thoracic Society, pulmonary rehab plays an essential role in the management of chronic lung disease. It helps to break the cycle of less and less physical activity.

Yet, the majority of Alphas who responded to the survey (63 percent) had never participated in pulmonary rehab.

So why have so many Alphas with COPD not participated in pulmonary rehab? The survey asked just that. Seventy-three percent of Alphas who reported never participating said their physician never recommended pulmonary rehab for them.

Of the Alphas who had participated in pulmonary rehab, almost 90 percent said the program was beneficial. The majority said it improved their quality of life, and many said it helped them to feel safer while exercising. Others liked that pulmonary rehab allowed them to meet new people and learn more about their lung disease.

Although most Alphas had not participated in a formal pulmonary rehab program, 68 percent of survey respondents reported they took part in physical exercise. Most of these active Alphas are exercising two or three days a week. Walking, weight lifting, and biking are the most popular ways to get some exercise.

If you have Alpha-1 and COPD, talk to your doctor about whether a pulmonary rehab program could help you. Exercise is important for everyone. Consider what activities you enjoy, and discuss a safe exercise plan with your doctor to attain or maintain the many health benefits of being active and fit.
New approaches to lung volume reduction

Shrinking overinflated lungs without surgery

By Danielle Woodford
MUSC Research Coordinator

Emphysema is characterized by holes in the lungs that trap air, making the lung hyperinflated (larger than normal). This hyperinflated lung interferes with the function of healthy areas of the lung and also interferes with the diaphragm, the key muscle used to breathe. The result is shortness of breath when you exercise, sometimes even when you just walk across a room.

Surgical lung volume reduction (LVRS) is one treatment used for emphysema; it can improve lung function and exercise performance. Surgical reduction is done either through sternotomy (cutting through the breastbone), or by thoracoscopy (small incisions between the ribs). The surgery removes up to 30% of the lungs. The procedure is associated with a 5% mortality (death) rate and about 25% of patients don’t see improvement. For those reasons, in recent years interest has focused on alternative ways of reducing lung volume.

Some Alpha-1 patients are familiar with bronchoscopy, a technique that uses a scope placed down the back of the throat under light sedation to reach inside the lung. Bronchoscopy is a less invasive and much safer procedure than surgery and may be performed for several reasons in patients with lung disease.

Bronchoscopic lung volume reduction (BLVR) uses small medical devices or methods in the lung to reduce lung volume during a bronchoscopy procedure.

There are several possible benefits unique to BLVR. For example, BLVR can be used to treat lower lobe emphysema. It also may be possible to treat patients with BLVR whose emphysema is complicated by additional diseases that would otherwise have too many risks for surgery.

No BLVR technique is currently available for clinical use in the United States, but BLVR technologies are being evaluated in clinical trials. Three recent trials have published their results:

The REVOLERS bronchoscopic coil clinical trial was done in France. One hundred patients, 71 men and 29 women (mean age 62 years), with severe emphysema were included. Those selected for treatment had a bronchoscopic procedure that inserted coils into the damaged lung to reduce the volume of the damaged areas. Those in the control group received usual emphysema care. After six months of follow-up, patients who received coils showed improved exercise capacity. On the standard 6-minute walk test, 18 patients (36%) in the coil group had improved by at least 54 meters (177 feet), versus nine patients (18%) in the control group. Quality of life measurements were also improved. Similar findings were recently released in the RENEW study in the United States.

The Step-up study was done in seven countries outside of the U.S. to study the effectiveness of treating upper lobe emphysema with a bronchoscopic technique that uses vapor (steam) to cause scarring of hyper-inflated lung tissue, which can counteract the hyperinflation. In this study, 154 patients were screened and 70 were enrolled. Participants were randomized 2:1 to receive treatment or be a control. A total of 46 patients received bronchoscopic vapor treatment and 24 received usual emphysema treatment. After six months, those in the vapor treatment group showed average improvement in lung function of 14.7%, when compared to the control group, and quality of life measurements were improved.

The STELVIO endobronchial valve trial was completed in The Netherlands. This study used Endobronchial Valve (EBV) therapy, where tiny one-way valves were placed in the lungs to block airflow to diseased regions. A total of 68 patients were randomly assigned to the EBV group (34) or the control group (34). The treatment group received EBV therapy and the control group received usual emphysema care. After six months, the treatment group had a 17% increase in their lung function score and their 6-minute walk distance increased by an average of 61 meters (200 feet). Quality of life measurements were improved.

A new research study of BLVR has opened to people with advanced emphysema who have Alpha-1 in the United States.
Periodontal disease may complicate Alpha-1

By Alison Garbarini
MUSC Research Assistant

The struggle with recurring respiratory infections is all too familiar for people with lung symptoms associated with Alpha-1 Antitrypsin Deficiency (Alpha-1). Another struggle, that often goes unnoticed, is that of periodontal disease.

Periodontal disease is a long-term infection of the gum tissues and other supportive structures that surround the teeth, creating chronic inflammation. Symptoms range from bleeding gums and bad breath to loose teeth. According to the Centers for Disease Control and Prevention (CDC), about half of all American adults have some form of periodontal disease.

What does this mean for Alphas?

The Alpha-1 Research Registry asked about the frequency of periodontal disease in a survey in 2014. Our hypothesis was that Alphas might have more periodontal disease than people with normal Alpha-1 levels. We found that very few people in the Alpha-1 community reported having any periodontal disease.

Unfortunately, we do not think this can be true, if half of all Americans have periodontal disease when examined.

We asked these questions because many respiratory diseases, such as bacterial pneumonia and bronchitis, are related to aspiration (inhaling) of unhealthy oral bacteria into the lungs. The bacteria that cause pneumonia most often come from our mouth.

Most of us handle small numbers of bacteria by our natural defenses. However, if the number of bacteria is high, or the type of bacteria is unusual because of periodontal disease, then the risk of pneumonia and bronchitis increases. Aspiration of oral secretions is common even in healthy people and is worse while sleeping. However, if you have chronic obstructive pulmonary disease (COPD) as many Alphas do, any form of periodontal disease may cause more frequent and/or severe exacerbations.

To avoid the chances of your mouth becoming a reservoir for unhealthy bacteria, understand that the mouth is the gateway to the rest of the body. Periodontal disease can be a systemic illness that is worse when someone is sick. Periodontal disease also occurs more frequently when people have other illnesses.

Chronic periodontal disease may go completely unnoticed for years because there may be no symptoms until the teeth begin to shift and become loose. In addition, many Alphas use medications, including anticholinergic inhalers such as ipratropium or tiotropium, which can cause dry mouth, making periodontal infections more difficult to combat.

If you are an Alpha-1 patient, you should see your dentist regularly and ask if you have periodontal disease. If you do, treating this common disease may have a positive impact on your lung health as well. Treatment will often require more frequent appointments and possibly deep scaling and root planing.

The best, simplest and cheapest way to keep your teeth and gums healthy is the advice your dentist (and probably your mother) told you: Flossing daily and brushing two times per day.

New approaches to lung volume reduction – continued

The EMPROVE study is designed to determine if one-way valves, placed inside emphysematous airways, will improve lung function and symptoms of treated patients.

This study opened in March 2013 as a randomized study (meaning that some patients get the valves and some get supportive care without the valves), and Alpha-1 patients were initially excluded because of a lack of information about the potential benefits of BLVR in patients who have Alpha-1. Since then, the study sponsor, Spiration, Inc. has decided to add an Alpha-1 arm to the EMPROVE study and recently received Food and Drug Administration (FDA) approval for this treatment arm.

Thirty Alpha-1 patients can be enrolled. The Alpha-1 patients who meet eligibility criteria will all receive the valve treatment and will be followed for five years. The trial is recruiting across the U.S. and Canada. If you are interested in this study you can locate the enrolling site closest to you by visiting www.clinicaltrials.gov and searching for EMPROVE or NCT01812447; or call the Registry staff at 1-877-886-2383.
How the DNA and Tissue Bank helps Alphas

By Joanna Nolte
University of Florida Clinical Programs Coordinator
and Kimberly Brown
Genetic Counselor

The Alpha-1 Foundation’s DNA and Tissue Bank is located at the University of Florida. It is a collection of biological material such as DNA and tissue samples, and serves as a unique resource for Alpha-1 researchers.

The Alpha-1 DNA and Tissue Bank is a biorepository (biobank) for storing DNA and tissue to be used in research. Biobanks have become increasingly important in recent decades for use by investigators around the world. Without a large biobank, no researcher has access to sufficient Alpha-1 DNA to conduct studies that could lead to a cure.

Mark Brantly, MD, founded the DNA and Tissue Bank in 2002 and he runs it today with sub-investigator Farshid Rouhani.

The Alpha-1 biobank accepts Alpha-1 blood samples from people with suspected rare or null Alpha-1 alleles after their first round of testing. Joining the biobank requires the recommendation of a healthcare provider and the informed consent of the participant who submits a sample.

Any scientist who wants to use samples from the Alpha-1 biobank must make a request for sample access, which is then reviewed by the Tissue Bank Advisory Committee.

Each sample in the biobank is stored under a unique research code and only linked to participant information in a secure and confidential database. When samples are released for use in approved research, it is without any personally identifying information, guaranteeing that researchers do not know who donated the samples. All findings of research on samples must be reported to the biobank and the remaining sample destroyed. Many important studies have been possible because of samples in the Alpha-1 biobank.

Some Alphas are familiar with the Alpha-1 DNA and Tissue Bank because advanced Alpha-1 testing was recommended for them to clarify suspected rare genotypes. The biobank performs DNA sequence analysis of the Alpha-1 gene for all samples received. This detailed test is needed to detect, and then better understand, rare Alpha-1 alleles. Sequencing is not necessary for people with more common genotypes, including the alleles M, S, Z, F and I, which are easily defined by less costly and labor intensive tests.

Farshid Rouhani, left, and Mark Brantly, MD.

Participation in the Alpha-1 DNA and Tissue Bank is recommended as a next step for participants in the Alpha-1 Coded Testing (ACT) Study who receive a result of a suspected rare genotype.

Participation may also be recommended by a doctor if a patient’s Alpha-1 results seem inconclusive after common, first-tier tests are complete.

Over 150 rare Alpha-1 alleles have been recognized. Identifying a rare allele – and knowing which rare allele it is – can be significant for a patient’s medical management and appropriate family testing. It also helps better expand our Alpha-1 body of knowledge. Our biobank is also a resource for investigators who need samples to research other rare diseases. Because the samples in the DNA and Tissue Bank are not exclusively Alpha-1 samples, there are many samples with normal or common Alpha-1 genotypes, in addition to the wealth of rare genotype samples. More than 2,500 people have donated their DNA or tissue to the bank.

The samples that people can reasonably donate to research are most often limited to blood, which contains DNA. However, should you undergo an organ transplant, the biobank is able to accept the organ removed from your body, regardless of your Alpha-1 genotype. All such procedures must be set up in advance of your transplant surgery. This allows for arrangements to be put in place for our biobank to receive your lungs or liver when they are removed and replaced. The biobank is not able to accept post-mortem (after death) tissue samples at this time.

We know that many Alphas are happy to help however they can. While the Alpha-1 DNA and Tissue Bank is not a project that most people can personally contribute to, it is a project that indirectly benefits all Alphas and is good to know about. For those who are invited to participate in the biobank, we appreciate you doing so and hope you have found this to be beneficial.

Other Alpha-1 biorepositories, affiliated with the Clinical Resource Center (CRC) Research Registry or Alpha-1 research institutions, may be more general repositories for other people to donate samples for research uses in the future.
Study seeks people with Nontuberculous Mycobacterial infections

By Laura Schwarz
ACT Coordinator

The Alpha-1 Foundation Research Registry is recruiting for the CONVERT clinical trial, a study for adult patients with Nontuberculous Mycobacterial (NTM) lung infections caused by Mycobacterium avium Complex (MAC). If you have been diagnosed with NTM due to MAC infection, and haven’t improved since beginning antibiotics, this may be a study for you. Insmed Incorporated designed the CONVERT study to evaluate an investigational drug (Liposomal Amikacin for Inhalation) for patients who have experienced no improvement with previous treatments.

NTM can cause chronic lung infections. Nontuberculous Mycobacterium are naturally occurring pathogens that affect tens of thousands of people every year in the United States. The bacteria are widely found in the environment, including soil and tap water. Statistics have shown a link between NTM and Alpha-1 Antitrypsin Deficiency, COPD and bronchiectasis.

Coughing, with or without sputum, and sometimes with blood, night sweats, fever, shortness of breath and chest pain are some of the symptoms of NTM lung disease. Treatment usually involves a minimum of three antibiotics for at least 18 months taken orally, by inhalation, or intravenously.

The two main objectives of the CONVERT study are:

- To determine if and how well the study medication works, when taken once daily in addition to your current medicine, at achieving three lab tests in a row that do not show MAC.
- To determine if and how well the study medication works, when taken once daily in addition to your current medicine, at increasing the distance you can walk in a 6-minute period.

Do you qualify for the CONVERT study?
You must meet these (and other) eligibility criteria:
- Be 18 years of age or older
- Be diagnosed with MAC NTM lung infection with evidence of lung disease
- Continue to be positive for MAC on sputum culture while following a multi-drug treatment regimen for a minimum duration of six months which is either ongoing or was stopped no more than 12 months ago
- Have a MAC lung infection documented by at least two positive cultures with at least one obtained within six months.
- Not be pregnant, trying to become pregnant, or nursing a baby
- Not have cystic fibrosis, active tuberculosis, or a history of lung transplant

This study involves 150 sites spread throughout the U.S. You can visit www.clinicaltrials.gov and search for NCT02334004 to locate a site near you. The Alpha-1 Research Registry staff is also available to discuss the CONVERT study with you. The Registry is not participating as a study site itself. Travel reimbursement is available for this study. Participating in the study is completely voluntary and your decision will not affect your participation in the Alpha-1 Research Registry in any way. (This is true of every research study.) An invitation to the CONVERT study is enclosed with this newsletter for those who are interested in participating.

COPD Foundation launches online community

The COPD Foundation recently launched an online global community for people with Nontuberculous Mycobacterial lung disease or bronchiectasis (www.BronchandNTM360social.org). Their new site includes features such as an activity feed, user messaging, questions and answers, and more.
Caregivers need to care for themselves, too

By Kimberly Brown
Genetic Counselor

Caregivers are an essential part of our Alpha-1 families and community.

Caregivers are often a family member such as a spouse, parent or child, but can also be a friend or other significant person. Caregiver roles have many variations and change over time based on family circumstances, the health of the Alpha and the relationship between Alpha and caregiver.

Caregivers perform many roles: attending medical appointments, helping with medical decisions, negotiating with insurance companies, paying bills, financial planning, managing medications, home care, legal work, long-term planning, making sure the Alpha uses prescribed oxygen, even sometimes giving the Alpha his or her augmentation infusions. A caregiver is a companion, support person and advocate.

With a genetic condition like Alpha-1, the caregiver may face their own risks (if they are a blood relative) or take on the responsibility for talking to the Alpha’s relatives about the diagnosis and family testing.

Caregivers must adjust to the health issues of Alpha-1 in both their own life and the life of their loved one. They may have to learn new skills, balance old and new goals and find themselves dealing with uncomfortable and conflicting feelings. Caregiving itself is an act of love – yet with an array of new responsibilities and unexpected circumstances.

It’s no wonder that caregivers are often stressed.

It is common for caregivers to worry about their own future or the future of their loved one. A caregiver can be angry or resentful – toward the person who has become chronically sick; toward roles the caregiver may not have volunteered for; or toward others who don’t have caregiving responsibilities in their lives.

Caregivers often feel guilty for being able to enjoy things that their loved one cannot, or for not being the “perfect” caregiver. It’s common for caregivers to grieve for the loss of dreams and expectations, or potential loss of a loved one. An Alpha’s inability to work anymore due to illness, and a loss of income while the cost of medical care and medications increases, can be an added burden.

Studies of caregivers for people with chronic illness consistently find higher levels of depression and mental health problems among caregivers than among non-caregiving peers. Caregivers are less likely to do things to care for themselves, such as exercise, recreation and maintaining their own medical care. It is important for caregivers to be aware of common stressors and reactions, as well as strategies to combat caregiving stress.

You must keep in mind: You will be a better caregiver if your own needs are met and you enjoy a better quality of life.

When someone we love gets sick, we know how important emotional support, good nutrition, and proper rest are for them. Exercise has many physical and mental health benefits. These same activities are just as important for the caregiver. Caregivers need social engagement and support, too.

It’s essential to maintain hobbies and friendships. Don’t go it alone. Learn to accept help when family and friends offer. Take a few minutes to yourself each day to just breathe and relax. If you find that your feelings are mostly negative on more days than not, consider asking a doctor or mental health provider for help. Using these resources is a sign of strength, not weakness!

Communicate with your Alpha openly and honestly about hopes, dreams, fears and plans for the future. The two of you can become care partners in the face of Alpha-1! Separate the illness from the person and do things you both enjoy together. Remember that illness and caregiving can be opportunities for connecting in new ways.

Caregivers should learn as much as possible about Alpha-1 and the Alpha’s medical care plan. There will be fewer surprises and you will be better prepared for whatever comes your way. Caregivers may find new purpose by raising Alpha-1 awareness, spearheading family testing and getting involved in Alpha-1 support groups.

The Alpha-1 Foundation Caregivers Virtual Support Group is dedicated to providing support, education and information for Alpha-1 caregivers. Attending Alpha-1 Foundation Education Days and local Alpha-1 Support Group events are other great ways for Alpha-1 caregivers to get involved and make new friends who understand Alpha-1. Many local hospitals and churches also offer caregiver support groups. For more information about Alpha-1 Support Groups, call Barbee Bennington, Support Group Coordinator, at 1-855-351-6610. To learn more about Alpha-1 genetics and family resources, call Kimberly Brown, Genetic Counselor, at 1-800-785-3177.
Obesity, Alpha-1 and exacerbations

Too much body fat hampers your breathing

By Tatsiana Beiko, MD
MUSC Clinical Instructor

Obesity is a risk factor for advanced liver disease in adults with Alpha-1 Antitrypsin Deficiency (Alpha-1). If you can avoid excess weight gain, you can help to prevent fatty liver disease, which can damage the Alpha-1 liver.

Most people with severe Alpha-1 have lung disease, commonly COPD. While weight loss is a feared complication of advanced COPD, avoiding excessive weight is also important to help you breathe more easily without the burden of carrying around a lot of excess pounds.

Alphas with lung disease in the QUANTUM-1 research study who were overweight or obese had more exacerbations than people with normal weight. This is important because exacerbations are closely linked with loss of lung function, quality of life and mortality in Alphas. (An exacerbation is a flare-up or worsening of your lung disease, such as a chest cold or pneumonia.)

The worldwide prevalence of obesity has doubled in the past 30 years, which has brought more attention to the relationship between obesity and lung disease, especially COPD and Alpha-1.

Interestingly, the prevalence of obesity is higher among people with COPD, compared to people without a COPD diagnosis. This may be due to lower activity levels, because shortness of breath discourages people from getting the exercise they need. People with COPD who are overweight or obese are at increased risk of developing heart disease, diabetes, and sleep apnea.

In general, obese people have more shortness of breath, worse health-related quality of life, more medication use and greater limitations in their physical activity, regardless of the degree of lung function impairment.

Obesity is not simply increased body mass, but a complex metabolic condition that influences many aspects of the human body.

Obesity is defined by the Body Mass Index (BMI) scale, which uses a formula to compare a person’s weight to height. A BMI of 30 or greater indicates obesity; 25-29.9 is overweight; 18.5-24.9 is normal or healthy weight; and below 18.5 is underweight, according to the U.S. Centers for Disease Control and Prevention (CDC).

However, the BMI scale does not differentiate between fat and muscle mass. Some people have a normal body mass but have reduced muscle mass compensated for by excessive body fat. This is especially true in early Alpha-1 lung disease when breathlessness limits exercise but not food intake. Therapies, such as pulmonary rehabilitation classes, for promoting fat loss but preserving muscle mass should be implemented for those who would benefit from BMI reduction or body composition improvement. Remember that being underweight, which can occur in advanced lung disease, also carries health risks.

Proper diet and exercise are very important for all Alphas and help to achieve or maintain healthy body weight and composition. Studies of large patient groups in the United States and abroad confirm a relationship between dietary intake and stability of lung function or preventing COPD. It is best to choose a well-balanced diet rich in nutrients and anti-oxidants.

Specifically, increased intake of antioxidants, including vitamins C, E, beta-carotenes and nutritious foods like olive oil were found to correlate with better lung function. Alphas should also engage in regular exercise aimed at aerobic, endurance and strength training.

The Step Forward Study sponsored by AlphaNet was recently conducted to study the effects of diet and exercise on the health of Alphas. The study has completed enrollment and the results are being analyzed for publication. We are looking forward to the results to learn more about the roles of obesity and exercise in Alpha-1 and COPD.

Many people struggle with body weight. It is a great idea to review your body mass, body composition, dietary needs and safe exercise with your healthcare provider, especially if you are experiencing unintentional weight loss or weight gain. Your healthcare provider can help you understand your ideal weight and develop a personalized nutrition and exercise plan to achieve your best health.
## Calendar 2016

### Building Friends for a Cure Events

<table>
<thead>
<tr>
<th>Date</th>
<th>Event Name</th>
<th>City, State</th>
<th>Contact &amp; Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aug. 27</td>
<td>Prairie Walk for a Cure</td>
<td>Campton Hills, IL</td>
<td>Tom Corron <a href="mailto:corron@live.com">corron@live.com</a></td>
</tr>
<tr>
<td>Sept. 3-5</td>
<td>Everesting for Alpha-1</td>
<td>Worthington, MA</td>
<td>Tom Pease</td>
</tr>
<tr>
<td>Sept. 23-25</td>
<td>Escape to the Cape</td>
<td>Cape Cod, MA</td>
<td>Angela McBride <a href="mailto:amcbride@alpha1.org">amcbride@alpha1.org</a></td>
</tr>
<tr>
<td>Oct. 2</td>
<td>Step Forward for Alpha-1 Indiana</td>
<td>Gas City, IN</td>
<td>Kim Dunham <a href="mailto:k.dunham@tlcngnt.com">k.dunham@tlcngnt.com</a></td>
</tr>
<tr>
<td>Oct. 29</td>
<td>50's Friends for a Cure Dance</td>
<td>Shoemakersville, PA</td>
<td>Larry &amp; Marian Hoffman <a href="mailto:allalfaalphaalphagroup@gmail.com">allalfaalphaalphagroup@gmail.com</a></td>
</tr>
<tr>
<td>November</td>
<td>Alpha-1 Virtual Walk</td>
<td>Anywhere</td>
<td>Angela McBride <a href="mailto:amcbride@alpha1.org">amcbride@alpha1.org</a></td>
</tr>
</tbody>
</table>

For more information about Building Friends for a Cure, contact Angela McBride, (877) 228-7321, ext. 233.

### Education Days

<table>
<thead>
<tr>
<th>Date</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sept. 10</td>
<td>Pittsburgh, PA</td>
</tr>
<tr>
<td>Oct. 29</td>
<td>Charleston, SC</td>
</tr>
</tbody>
</table>

For more information about Education Days, contact Kim Caraballo, (877) 228-7321, ext. 323.

### Virtual Support Group Calls

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sept. 27</td>
<td>Timely Topics Forum</td>
<td>Open enrollment</td>
</tr>
</tbody>
</table>

To participate: At 9 pm Eastern, dial: **1-800-920-7487**.
When prompted, enter the code: **9335 9985#**
For more information about events, contact us at **1-877-228-7321**.

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## Alpha-1 Foundation

The Alpha-1 Foundation is committed to finding a cure for Alpha-1 Antitrypsin Deficiency and to improving the lives of people affected by Alpha-1 worldwide. The Foundation has invested nearly $60 million to support Alpha-1 Antitrypsin Deficiency research at 103 institutions in North America, Europe, the Middle East and Australia.

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## AlphaNet

AlphaNet, Inc. is a not-for-profit organization that provides a comprehensive disease management and prevention program to improve the lives of people with Alpha-1 Antitrypsin Deficiency. AlphaNet also oversees and sponsors clinical trials involving Alpha-1 therapies.

The Registry Update is funded by unrestricted educational grants from: **AlphaNet; Baxalta, now part of Shire; CSL Behring; Grifols**