2016

University of Maryland School of Medicine Amish Research Clinic

Annual Newsletter

We hope that this newsletter finds you and your family healthy and happy.

This is our 20th anniversary! Since we began our research 20 years ago, we have enrolled over 6,000 Amish volunteers in our studies. Most of these volunteers have participated in more than one study. Our Wellness Study has recruited over 3,000 volunteers. For approximately half of these volunteers, this is the first study that they have done with us.. This study has provided these participants with screenings for heart disease, anemia, thyroid problems, liver disease, kidney disease, diabetes. abdominal aneurisms and osteoporosis.

This newsletter comes to you each year to keep you informed about our new studies and what we have learned from the studies that we have completed. Some of the diseases we study include diabetes, osteoporosis, high blood pressure, cholesterol abnormalities, breast density, celiac disease, longevity, seasonal affective disorder, obesity, heart disease and wellness. We currently have 10 active studies.

None of this work could be possible without our compassionate Amish volunteers and the Amish Community, who have provided their partnership and support.

Together, our research has resulted in new discoveries and it has helped us better understand the causes of a number of diseases. As described in this newsletter, the results of some of our studies have already had an impact on health in the Amish as well as in other populations.

Many of our studies continue to need volunteers. Thousands of Amish volunteers have benefited from participating because it provides a number of health benefits including free medical evaluations and screenings for a number of common diseases and disorders. Participating in research studies also provides the opportunity to contribute to new knowledge, which may help millions of people with the diseases that we study.

Some of the studies are conducted at our clinic in Lancaster and free transportation to and from the clinic is provided. Other studies are conducted right in your own home. Not only do you gain lots of knowledge about your health but with most studies, we even pay you for your time and effort. If you have any questions or you are interested in participating in any of our studies, please call 717-392-4948. You can also write us a note. Please make sure you include your address so we can get back to you.

Our Mission

The Amish Research Clinic contributes to improvements in healthcare through research. We serve as a resource for health information and knowledge to the Amish Community.

Our Staff

Alan Shuldiner, M.D.

Amber L. Beitelshees, Pharm.D, MPH

Andrew Goldberg, M.D.

Braxton Mitchell, Jr., Ph.D.

Christy Chang, Ph.D.

Coleen Damcot, Ph.D.

Elizabeth Streeten, M.D.

Elliot Hong, M.D.

Joshua Lewis, Ph.D.

Julie Douglas, Ph.D.

Mao Fu, Ph.D.

Michael Miller, M.D.

Nanette Steinle, M.D.

Nisa Maruthur, M.D.

Richard Horenstein, M.D.

Soren Snitker, M.D., Ph.D.

Teodor T. Postolache, M.D

Toni Pollin, Ph.D.

Mary Morrissey, R.N., BSN
Nurse Coordinator

Theresa Roomet, R.N., BSN
Research Nurse

Donna Trubiano, R.N.,
Research Nurse

Susan Shaub, R.N., BSN
Research Nurse

Yvonne Rohrer, R.N.,
Research Nurse

Patrick Donnelly, R.D.C.S.,
Sonographer

Maryann Drolet, BSMT,
ASCP, Research Specialist

Sylvia Newcomer, BSMT,
ASCP, Research Specialist

Elizabeth Zehr,
Clinic Assistant

Grace Redcay, Driver

Ongoing Studies

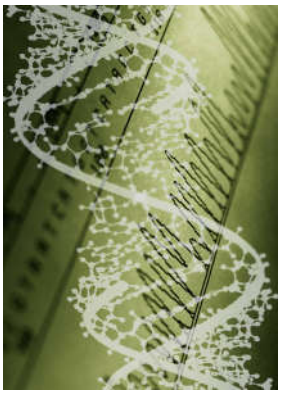
20 years ago the Amish Research Clinic started with 2 people. Now there are 23 employees at our Lancaster Clinic.

Wellness Study (active)

The Amish Wellness Study continues to recruit participants. This study offers all Amish adults basic wellness screening including tests of cholesterol, blood sugar, thyroid, bone strength, and heart health. We have found that high cholesterol and hypothyroidism (low thyroid function) are quite common in the Amish. Blood is also being collected and stored at the University of Maryland for research on genetic and non-genetic factors in health and disease. The research team hopes to visit all Amish households. Testing takes place in our "Wellmobile" housed in a large motor vehicle which visits each Church district. If we haven't visited your Church district yet, we will be there in the future. To date, nearly 3000 Amish individuals ages 18 and older have enrolled in the Wellness Study, which is funded by the University of Maryland Program for Personalized and Genomic Medicine. Thank you!

MiACT Study - Metabolic Impact of ApoC-III (active)

Based on our exciting finding that about 1 in 20 Amish people carry a gene change that helps them to clear dietary fat from their blood faster and may help prevent heart disease, we are conducting a study to learn more about this gene change called APOC3 R19X. People with this gene change make less of a substance in the body called ApoC-III. The new study is helping us to learn how ApoC-III works and whether lowering it in other people might be a useful way to prevent heart disease. We are comparing people with and without the gene change for how their fat is distributed in their bodies, how their bodies process dietary fat, cholesterol and sugar, and how fat and cholesterol move around in their bloodstream. So far over 100 people have enrolled in the study, which is funded by the National Institutes of Health.



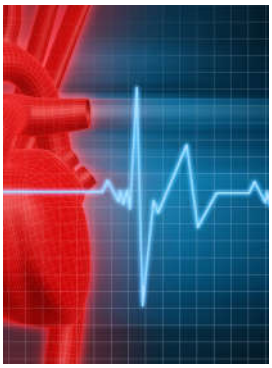
Ongoing Studies cont.

Pear 1 (active)

Cardiovascular disease is the leading cause of death in the United States with heart attacks (also known as myocardial infarction) being the most common form of this disease. When a person has a heart attack, aspirin is the most commonly given drug in order to help patients get better. Aspirin works by preventing blood clots, which is a common reason why people have heart attacks. While aspirin prevents the formation of blood clots in many people, some patients do not benefit (or get a reduced benefit) from taking this drug. This is known as aspirin resistance.

The reason why some people have aspirin resistance may be due to differences in their genes. In this study, we will evaluate how differences in the gene PEAR1 and other factors determine how well people respond to 3 different doses of aspirin. So far; we have enrolled 46 participants in this study.

The results of this study may be very important in helping doctors prescribe the best dose of aspirin based on the patient's genetic make up to prevent or treat a heart attack.



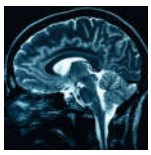
Osteoporosis Study (active)

This study was started in March 1997 and thanks to our many wonderful Amish participants, we are making great progress in studying genes that are important for bone health. We have found regions on chromosome 7 and 21 and variations in a gene called EIF2AK3, that are likely to influence bone health. This study remains active and in particular we are now studying the factors that cause some people to lose more bone after middle age while other people tend to be very slow "bone losers." If you have participated in the Wellness Study and your heel study indicated you may need further testing, you may call the clinic to arrange an appointment for a dexa scan. The dexa scan will be performed free of charge as part of the Osteoporosis Study.

Osteoporosis Pseudoglioma (OPPG) Study (active)

Osteoporosis pseudoglioma syndrome (OPPG) is a rare genetic disorder of weak bones (osteoporosis), blindness (from birth) and sometimes behavioral problems. Although OPPG is extremely rare in the general population (about 60 people with OPPG are known worldwide), many children with OPPG have been diagnosed in the Old Order Mennonite community in PA (15 children so far). OPPG can lead to multiple broken bones (fractures) of the upper and lower leg bones and back. Dr Streeten has been studying OPPG for 15 years, trying to find a new treatment that will help strengthen the bones in people with OPPG. Traditional medications used to treat osteoporosis can help in OPPG but do not totally prevent fractures. We completed a 6 month study of lithium, which was shown to strengthen the bones of mice with OPPG and it did strengthen the bones in children with OPPG but worsened behavioral problems in those who had behavior problems at baseline. We are now studying the quality of bones in OPPG with a special type of x-ray called pQCT, a painless procedure which gives detailed pictures of the bones to help us understand why the bones are so fragile in OPPG.

Amish Imaging and Mental Illness Study (active)



We continue our new study to learn more about mental health problems in the Amish community. The study also aims to find out more about brain differences that make it more likely someone will have mental health problems. This is a family study, so we would like several people from the same family to participate. This study has two parts. The first part involves completing a brief survey that asks about feelings and behaviors. We mail this survey to your home. For the second part of the study, participants travel by van with other family members to Baltimore, MD. In Baltimore, participants complete other study tasks and tests. We use a method that takes pictures of your brain; this is called magnetic resonance imaging (MRI). We can use these pictures to understand the brain circuits, or wirings, which are related to mental health. These brain wirings are often heritable, meaning that they run in families.

The study tasks in Baltimore, MD take about seven hours to complete. The van ride takes about two hours each way, so volunteers are gone for about 11 hours. Study participants are compensated for their time and effort. Amish adults and children aged 12 and above are eligible to participate. We enroll persons with and without histories of mental health problems.

If you would like to learn more about this study, please contact the Amish Research Clinic at (717-392-4948).

Genetics of Pain Study (active)

Pain is the primary reason that patients seek medical attention. Recent medical advances have dramatically increased life expectancy and, therefore, the number of people living with chronic diseases and chronic pain. More than 116 million Americans are chronically in pain, and they make more than 70 million visits to healthcare providers at a cost of more than $600 billion every year. Most of the patients have their pain for five or more years, causing decreased quality of life and increased stress for the entire family. Scientists believe that there is a link between our genes and how we sense pain, which is why some people require more pain medication than others after the same injury, or develop chronic pain after recovering from an illness. However, exactly which genes are involved in determining these differences are not known. The goal of this research project is to gain a better understanding of how our genes control pain sensing and why some people feel more pain than others. So far, 70 subjects have completed this study. We plan to recruit 100 participants and measure their response to pain from heat, cold and pressure. While the participants will experience some temporary pain from heat, cold and pressure produced by an instrument placed on their arms, they are not injured in any way. The study requires around 3 hours and participants are compensated for their time and effort. If you are interested in participating, please call our clinic @ 717-392-4948 and find out more information.

The Amish Family Diabetes Study (active)

The Diabetes Study was the very first study done by the Amish Research Clinic–**20 years ago!** The study began with Dr. Shuldiner and an Amish liason/ helper, Sadie Beiler (Joseph). Dr. Shuldiner brought equipment and processed blood in his car. We have come a long way since then. Diabetes remains a very common disease caused by increased sugar in the blood.. There are two major types of Diabetes. Type 1 occurs in children and the only treatment is insulin injectionssince the body cannot make its own. Type 2 diabetes is more common and generally occurs in overweight adults. Diet, weight loss, pills and sometimes insulin injections are needed to keep the blood sugar under control. Diabetes runs in families. The goal of this project is to identify genes involved in diabetes. Thanks to many of the Lancaster Amish , we have recruited over 1300 volunteers. One of the genes we identified in the Amish that is involved in type 2 diabetes, called GRB10 appears to also be important in other populations around the world. We continue to study which genes are involved in the development of Type 2 diabetes so that we can better identify people at risk for diabetes and also find new ways to prevent and treat it. Symptoms of diabetes may include fatigue, increased thirst, hunger and urination. If left untreated, diabetes can lead to eye, liver, kidney, nerve and blood vessel problems. If you or someone in your family is experiencing these symptoms and would like to be tested for diabetes, please call the clinic at 717-392-4948. All testing is free and usually done in your home.

CELEBRATING 20 YEARS!!

Our Amish Liaisons:

Mary Esch

Naomi Esh

Sadie Fisher

Sarah Fisher

Susie Fisher

Sarah Glick

Hanna King

Naomi Stoltzfoos

Barbie Stoltzfus

Fannie Stoltzfus

Katie Stoltzfus

Mary Stoltzfus

New Studies

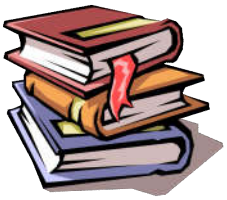
Omega 3 Study (active)

We recently began a new study to learn whether omega-3 fish oil can help to reduce the type of fat that causes health problems. Fat that accumulates in the gut appears to promote inflammation more than fat that accumulates in other areas of your body and this may put you at increased risk of heart disease. Omega-3 fish oil lowers blood fats (triglycerides) but we are not sure whether it also reduces inflammation of fat. To be eligible for this study, Amish men and women older than 18 years need to have a fasting triglyceride levels above 150 with an increased waistline (35 inches or more in women and 40 inches or more in men). If eligible, participants will be assigned to take either omega-3 or placebo capsules (4 each day). At the beginning and end of the 9 month study, pictures of your fat will be taken using a technique called magnetic resonance imaging (MRI). We will remove a small amount of fat from your belly by aspiration and blood and urine collection will be performed at the beginning and end of the study. All studies will be conducted in Lancaster and participants will be compensated for their time and effort. If you think you may be eligible for this study and are interested in participating, please call.

LP(a) (active)

Heart disease(CVD) is a leading cause of human morbidity and mortality in the world. A high cholesterol level in the blood is an important risk factor for heart disease. Recent evidence from large cohorts suggest that a particular protein that is associated with cholesterol, called Lipoprotein (a) [Lp(a)] is an important determinant of heart disease and stroke. . However, no practical method for pharmacologic lowering of Lp(a) levels is currently available. The objective of this study is to define how genes influence Lp(a). We are particularly interested in two regions on chromosomes 6 & 11. Participants of this study will be asked to provide a blood sample and also undergo other tests to look at blood vessels in the neck and brain using ultrasound and MRI. In last year, we have recruited 70 Amish subjects to this study. Identification of the genes that influence Lp(a) levels may lead to the design of novel therapeutic strategies to lower Lp(a) levels to prevent or treat heart disease in patients with diabetes.

Many of our studies continue to need volunteers.



2014 Publications can be viewed on this website…

http://medschool.umaryland.edu/endocrinology/publications.asp

Seasonal Affective Disorder Study (inactive)

Seasonal Affective Disorder (SAD) affects millions of Americans. People with SAD have low mood, low energy, gain weight, and feel sleepy through the winter. Decreased day length triggers SAD in some individuals and light therapy treats SAD. Some patients need medications or talk therapy for a full improvement. This is the first study of SAD in the Amish. Our findings reporting the frequency of SAD and heritability of the disorder, both lower than expected, have been published in the Journal of Affective Disorders . We have now an article in press in the Journal of Clinical Psychiatry suggesting that in Australian twins, but not Amish seasonal affective disorder, overlaps with bipolar disorder and schizophrenia. We are also submitting for publication an article documenting an association between adiponectin, a hormone implicated in weight regulation, and seasonality of mood We will be working on obtaining a grant to be able to study actual patients with SAD in the clinic in greater detail in the following years, to find out how we can predict who will better respond to light treatment. In the meantime, patients who experience problems with SAD or depression can call the clinic for a list of referrals to mental health professionals in the area.

We are no longer recruiting for these studies but analysis continues at the university's laboratory

Our ARC Family


Sue


Mary


Dr. Shuldiner


Donna


Patrick


Theresa


Yvonne


Nancy


Grace


Beth


Sylvia


Maryann

Some of the diseases we have studied include diabetes, osteoporosis, high blood pressure, cholesterol abnormalities, breast density, celiac disease, longevity, seasonal effective disorder, obesity, heart disease and wellness

Studies cont.

PPAR Study (inactive)

The purpose of the PPAR study is to determine why some people do not respond to pioglitazone, an approved drug commonly used to treat diabetes. We thank the around 30 Amish participants who participated in the study. We are in the process of analyzing the samples to find the genes responsible for individual differences in response. Once we finish the analyses we will return with more information in this newsletter. It is possible that we may resume recruitment in the future to study groups of people that are either very responsive or very non-responsive to pioglitazone. We are currently not recruiting for this study.

Zinc Study (inactive)

Based on increasing knowledge of genes that affect diabetes, we enrolled 60 participants in a study to evaluate the effect of a zinc supplement on blood sugar and insulin levels. The participants spent two mornings in the clinic and took a zinc supplement for 14 days in between. We found that some people improve their insulin levels in response to zinc while others do not. This work was recently published in a scientific journal. We will apply for funding for a larger and longer study using zinc to treat diabetes.

Breast Density Study (inactive)

Breast density refers to the amount of dense glandular tissue in the breast. It is measured by a routine mammogram or x-ray of the breast to detect unsuspected cancer. Dozens of studies over the past quarter of a century have suggested that dense breasts are more cancer prone. But no one knows exactly why. With the help of nearly 1,500 Amish women, we've been trying to answer that question by searching for the genes that affect density. We recently published an article, together with our colleagues in the US and abroad, describing over half a dozen genes that influence breast density in the Amish and other populations of European descent. Several of these genes are also associated with the risk of breast cancer.

Over the coming year, we plan to continue searching for new genes that influence breast density and studying the link between breast density and breast cancer. Meanwhile, we would like to remind all Amish women of the importance of getting a routine mammogram. The National Cancer Institute recommends that women over the age of 40 years have a mammogram every 1-2 years coupled with a breast exam by a doctor to improve the early detection of breast cancer. By doing so, a woman may reduce her risk of dying from breast cancer by about 17% (if she is 40 to 49 years old) and by about 30% (if she is 50 years or older). If you need assistance scheduling a mammogram, please call us at 717-392-4948.

Publication:

Nature Communications 5:5303Lindstrom S, Thompson DJ, Paterson AD, et al. (Oct 2014) Genome-wide association study identifies multiple loci associated with both mammographic density and breast cancer risk.

Our 20th Anniversary Edition!

University of Maryland
School of Medicine
Amish Research Clinic
1861 William Penn Way
Lancaster, PA 17601
717-392-4948

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