Amish Research Clinic
Of the University of Maryland
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Holiday greetings from The Amish Research Clinic. The Clinic has served the Amish community continuously since 1995. We have more than 10 ongoing studies that have enrolled more than 4,200 Amish volunteers. Dr. Shuldiner and his staff would like to take this opportunity to thank all of our volunteers and the Amish community for their partnership and support. Together, our research has resulted in new discoveries and understandings of the causes of a number of diseases and at the same time has benefited thousands of Amish volunteers by providing free medical evaluations and screenings.

In this newsletter, we provide updates on progress of our research. Several of these studies continue to enroll volunteers – studies on heart disease, diabetes, obesity, weak bones, high blood pressure, flu vaccine, and breast health – just to name a few. New studies are now being planned.

For most of these studies, you do not need to have the disorder to participate. Volunteering provides a number of health benefits including free medical evaluations and screening for a number of common diseases and disorders. It also provides the opportunity to contribute to new knowledge, which may help millions of people with the diseases 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. We even pay you for your time and effort. If you are interested in participating in any of our studies, please call 717-392-4948 or write to learn more.

We wish you and your family a healthy and happy New Year.

Alan Shuldiner, M.D.
Elizabeth Streeten, M.D.
Dan McBride, Ph.D.
Braxton Mitchell, Jr., Ph.D.
Richard Horenstein, M.D.
Soren Snitker, M.D., Ph.D.
John Sorkin, M.D.
Wendy Post, M.D.
Julie Douglas, Ph.D.
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Mary-Claire Roghmann, M.D.
Mary Morrissey, R.N.
Theresa Roomet, R.N.
Donna Trubiano, R.N.
Sue Shaub, R.N.
Nancy Weitzel, L.P.N.
Yvonne Rohrer, R.N.
Patrick Donnelly, R.D.C.S.
MaryAnn Drolet
Rhea Cosentino
Sylvia Newcomer
Sharon Rago

The Sitosterol Study

The study seeks to understand how levels of plant sterols in the blood differ between participants with one or zero copies of an altered sterol transporter gene. To date, over 300 Amish volunteers have provided blood samples. Our findings suggest that individuals with one copy of the changed sterol transporter gene are not at increased risk for heart disease. We have started a new phase of this project in which we are looking at how plant sterols in the diet affect health. To date, 4 Amish volunteers have participated in the new phase.

Women's Breast Health Study

The goal of this study is to identify the genes that influence the makeup of the female breast. The density of the breast is determined by how much gland tissue and fatty tissue is present. Increased breast density is a strong but poorly understood breast cancer risk factor. Identifying the genes that influence breast density may provide important insights into the prevention, control, and treatment of breast cancer. To date, more than 1,000 Amish women have participated in this study. We plan to enroll another 400+ women between now and the fall of 2009, when recruitment is scheduled to end. This study involves answering questions about your medical, reproductive, and family history, and medication use, providing us with samples of your blood, and visiting our clinic for a routine mammogram (x-ray) or providing us with your most recent mammogram. If you would be willing to participate in this study, please contact us in writing or by telephone (717) 392-4948 at the Amish Research Clinic. Please note that (unlike in previous years) you are not required to have a sister in order to be eligible for the study. We thank all of the women and families who have generously given their time to participate in our study.

Amish Family Osteoporosis Study

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 that are likely to contain genes that influence bone health.

Amish Sunshine Study

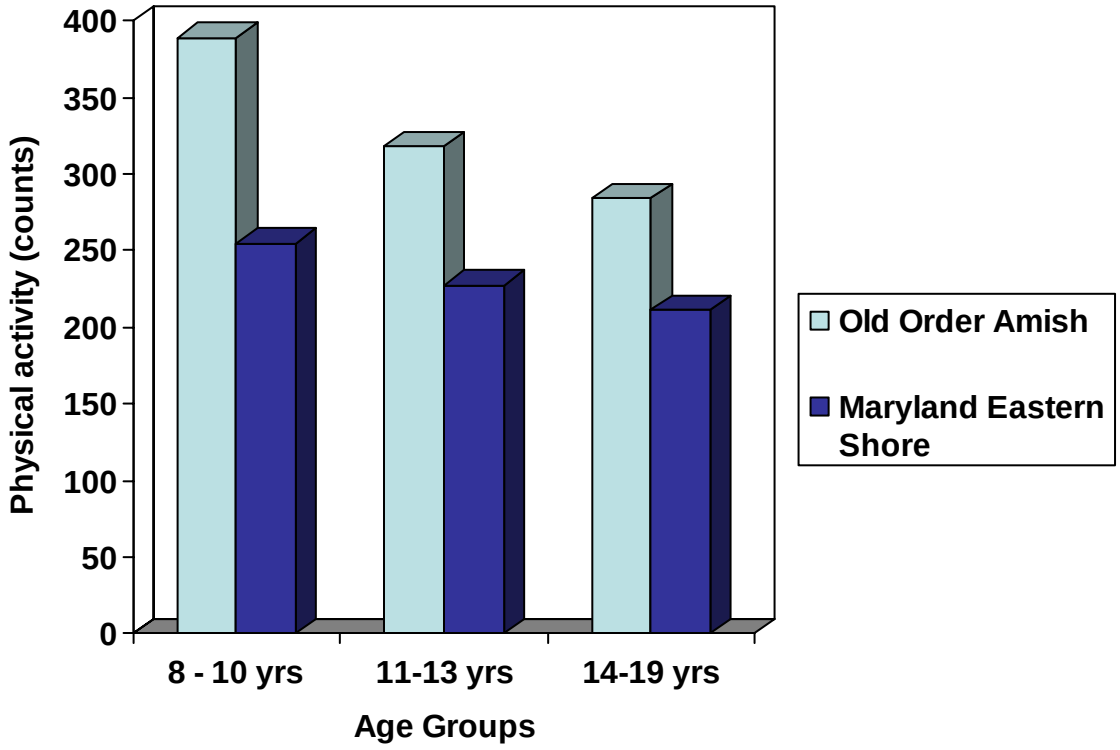
We have completed a small study, "The Sunshine Study" in which we are evaluating differences in how well people make Vitamin D in their bodies after a sunlamp exposure to the skin. We plan to continue the Sunshine Study after we get additional research funding to support it.

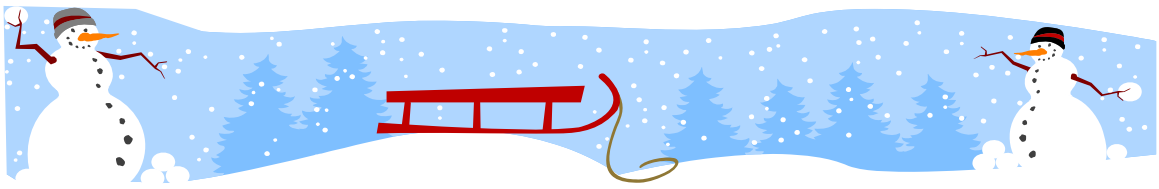
Amish Children's Physical Activity Study

We have compared physical activity as measured in the Amish Children's Study with physical activity in a group of children from the Eastern Shore of Maryland, who are not Amish but live in a similar rural setting. For these studies we used accelerometers, a type of device worn on the hip to measure physical activity over a week.

We found that Amish children are generally more active than Eastern Shore children, especially in the youngest age group we studied (see the figure below).

We also found that Amish children were less frequently overweight than Eastern Shore children. It is not healthy for children (or adults) to be overweight. This study suggests that Eastern Shore children should be more physically active, like the Amish children, so they can stay slim and healthy.







The PAPI Study

The goal of this study is to understand why some people do not respond to commonly used medications to prevent heart attacks including aspirin and clopidogrel (Plavix). This 8-day study involves taking clopidogrel and aspirin and having blood samples tested to see how well the medications prevent the blood from clotting. PAPI participants also get free testing for heart problems, high cholesterol, liver, kidney, and thyroid problems, and monetary compensation for their time and effort. To date, more than 400 subjects have participated in this study. This study is still open to enrollment. If you are interested in participating, please call or write.

A new and exciting finding is that a variation in a common gene called CYP2C19 is a major predictor of response to clopidogrel. Approximately 15-20% of the population carries the variation that predicts a poor response. This is an important finding because people with this variation might benefit more from a different medication to prevent heart attacks.

The HAPI Heart Study

This study was designed to better understand how genes interact with common lifestyle factors like stress and diet to contribute to heart disease. It was very successful – nearly 900 Amish community members volunteered for this study. We continue to analyze all of the information we accumulated and have made a number of very exciting discoveries. For example, we determined that the obesity-promoting effects of the FTO gene can be over-ridden by high levels of physical activity. This finding has important implications since it suggests that even if a person is born with genetic susceptibility to obesity, a healthy lifestyle can be very beneficial. These discoveries will lead to new approaches to improve health and prevent disease in the Amish and other populations as well.

Flu Vaccine Study

Older people are especially susceptible to getting the flu and are at greater risk of dying when they get it. The flu vaccine can help to prevent the flu. However, it is less effective in older people. The new Flu Vaccine Study aims to find out why some older people respond well to the flu vaccine while others do not. The study involves giving the flu shot to Amish people over the age of 65 and drawing a blood sample before and after the shot to measure the response. Volunteers will receive the same flu shot that is recommended to all older people at no charge. If you are interested in participating, please call or write us!



Amish Family Calcification Study

Many thanks to the more than 1,000 people who have participated in the Amish Family Calcification Study. In the Amish, as in other population, the amount of LDL ('bad") cholesterol in the blood is correlated with the amount of calcium in the blood vessels of the heart. By analyzing DNA isolated from the blood, we have now identified several gene variants that are also associated with how much calcium people have in the blood vessels of the heart. One of these gene variants appears to increase calcium in the heart vessels by increasing levels of LDL cholesterol, and a second gene variant appears to increase calcium levels through a pathway that we don't yet understand. Finally, we have identified a third gene variant, not found in any other population but present in a small number of Amish, that appears to decrease levels of calcium in the heart vessels, possibly by decreasing levels of fats in the blood (see more details in the New Findings in 2008 section below).

The Amish Family Diabetes Study

Diabetes is a very common disease in which the level of sugar in the blood is elevated. This can lead to eye, kidney, nerve, and blood vessel problems. Over the years, we have recruited more than 1,300 volunteers into this study. Recently, we tested nearly 100,000 different genetic variations in blood samples from participants of the Diabetes Study and found a new gene, called GRB10, that may be involved in diabetes. We are also working with an international team from England, France, China, and Arizona to identify additional diabetes genes. We hope these discoveries will lead to better ways to identify people at risk for diabetes and also new and more effective ways to prevent and treat diabetes.

Amish Staph Study

Staph is a bacteria or germ commonly found in the nose. The vast majority of people who carry the Staph bacteria in their nose remain healthy; however, it can cause skin and other types of infections in some people. This study examines how common Staph is in the Amish, and if related people are more likely to have Staph bacteria in their nose. Identifying the genes that influence whether an individual carries the Staph bacteria may help prevent infections. The Amish Staph Study is currently enrolling participants. The study involves answering some health questions and having two nose swabs taken by trained research staff at least one week apart. You do not need to visit the Amish Research Clinic to participate in the study. If you would be willing to participate in this study, please contact us at (717) 392-4948. Thank you to the 110 participants who have already enrolled in the Amish Staph Study. We are hoping to enroll a total of 400 participants.

The Amish Hypertension Study

Hypertension or high blood pressure is a common disorder that predisposes people to heart and kidney disease and stroke. Results from both the Amish Family Diabetes Study and the HAPI study show that a gene on chromosome 2 called STK39 is important in regulating blood pressure. This gene affects the amount of salt in the urine. This new finding may help us better understand how the body regulates blood pressure and may provide new targets for the treatment of high blood pressure. The unique characteristics of the Amish were instrumental in helping us identify this gene. We continue to look for additional genes that regulate blood pressure and to understand how these genes interact with other factors including diet, activity, tobacco use and stress.

New Findings in 2008

We discovered a variation in a gene called APOB. Although very uncommon in the general population, this variation is carried by 1 in 8 Amish people. Our studies indicate that those with the variation have much higher LDL (bad) cholesterol levels. Those with the variation are also more predisposed to having calcium in the arteries of the heart, a risk factor for heart attacks. Those who were found to have high-cholesterol were advised how to lower their cholesterol levels. We are now working on designing another study to test new approaches to prevent heart disease in those carrying this APOB gene variation.

We found a variation in another gene called APOC3. This variation is carried by 1 in 20 Amish people. As far as we know, the variation does not occur or is very rare in the general population. Those with the APOC3 variation have low levels of fat (triglycerides) in their blood and lower amounts of calcium in the arteries in the heart. Thus the variation seems to protect from heart disease. These findings are important because they suggest that decreasing APOC3 levels, even in people without the gene variation may protect them from heart disease.

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Finally, we have worked with other groups around the world to discover a number of new genes for cholesterol and glucose (sugar) levels in the blood. Working together with other groups allows us to discover things that none of us alone could have. It is also fun!