



**Amish Research Clinic
Of the University of Maryland
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Holiday greetings from The Amish Research Clinic. The Amish Research Clinic has served the Amish community continuously since 1995. We have 10 ongoing studies that have enrolled more than 4,000 Amish volunteers. This year, the Clinic moved from the first floor of the Clinic for Special Children to a new and larger facility in Lancaster. Dr. Shuldiner and his staff would like to take this opportunity to thank all of our volunteers and the Amish community for their participation and support. This unique partnership in 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 ongoing research. Several of these studies continue to enroll volunteers. 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.

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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 even before it is diagnosed. Over the years, we have recruited more than 1,300 volunteers into this study. We have done extensive genetics research on the blood samples and have found a number of new genes that have increased our understanding of what causes diabetes. These discoveries will lead to better ways to identify people at risk for diabetes and also new and more effective ways to treat diabetes.

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 volunteered for this study. Although we are not recruiting any new volunteers, we are working very hard in the laboratory in Baltimore to analyze all of the information we have collected. We have already discovered a number of gene variants that influence heart disease and other medically relevant traits such as cholesterol and glucose levels.

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 275 subjects have participated in this study. If you are interested in participating in this study, please call or write.

Amish Family Calcification Study

Many thanks to the more than 1,000 subjects who have participated in the Amish Family Calcification Study. We have started looking at DNA from blood samples of study subjects and have just identified a variant in a gene that is related to having high levels of LDL (“bad”) cholesterol levels. About 12% of Amish have this high cholesterol gene variant. Those with the variant not only have higher levels of LDL-cholesterol, but they also have higher levels of calcium in the blood vessels of the heart. We will soon assess whether or not carriers of this variant might benefit from drug therapy. We are also working on making genetic testing for this variant available to the Amish community.





Amish Family Longevity Study

The goal of this study is to identify genes that allow for a long and healthy life and to understand what these genes do. In the current phase of the study we are determining whether any gene variants that we discover are related to the lifespan of study participants or to their health status. Many thanks to the 383 subjects who have participated in the Amish Family Longevity 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. We are also starting a new 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.

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 304 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 plan to start a new phase of this project in the near future.

Amish Family Hypertension Study

Hypertension or high blood pressure is a common disorder that predisposes people to heart and kidney disease and stroke. Understanding hypertension genes will help to treat and prevent heart and kidney disease and strokes in millions of people. Results from both the Amish Family Diabetes Study and the HAPI study show that a gene on chromosome 2 is important in regulating blood pressure. The unique characteristics of the Amish were instrumental in helping us identify this gene. 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. 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.





Amish Childhood Obesity Study

Obesity in children is a big problem in the general population because these children are more likely to develop diabetes, high blood pressure, high cholesterol, and heart disease later in life. In the Amish Children's Study, we have found that childhood obesity is much less common in the Amish. The investigators of the childhood obesity study are still looking at the exciting data from the accelerometers (the devices worn on the hip to measure physical activity over a week). We are attempting to compare the activity in Amish children with children from the Eastern Shore of Maryland, who are not Amish but live in a rural setting. We will bring our results in the next newsletter.

Women's Breast Health Study

Because our pilot study was so successful, we have been granted new funding to expand this study and recruit another 1,200 women over the next 3 years. The goal of this study is to identify the genes that influence the makeup of the female breast, in particular, the amount of dense tissue compared to fatty tissue (or what is referred to as breast density). Among breast cancer risk factors, breast density is one of the strongest but perhaps least understood. Identifying the genes that influence breast density may provide important insights into the prevention, control, and treatment of breast cancer. 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 or releasing your most recent mammogram (or x-ray of the breast to detect unsuspected breast cancer). 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. We thank all of the women and families who generously gave their time to participate in our pilot study.

Finally...

An exciting new finding from the HAPI Heart Study

Research in Europe has revealed a new gene variant that on average leads to an increase in body weight of about 3-6 lbs. The gene harboring the variant is called FTO. We looked at whether FTO variants had the same effect on body weight in the Amish as it has in people living in Europe and found - not so surprisingly, that it does. However, we found some new results using the accelerometer data (the devices worn on the hip to measure physical activity). In those Amish who are in the top 50% of physical activity level for their age and sex, the gene doesn't make any difference either way; people who engaged in high levels of physical activity with and without the variant weighed the same. Many people, especially non-Amish, are struggling with their weight. This finding means that at least one genetic variant (FTO) that contributes to overweight can be negated if one is physically active. Thanks to those who helped us make this important discovery.

