


Hundreds of Families Enrolled in Genetics Study

We are pleased and grateful to report that more families have generously agreed to join the AD Genetics Study this year, and our unique community of volunteers is continuing to grow.


As of March 2007, more than 1,000 families had inquired about participation in the AD Genetics Study, and of these, 856 families were eligible for participation based on their family history.

Researchers have begun to collect samples and information from 702 eligible families in the study at the National Cell Repository for AD (NCRAD).

We are implementing new study goals, including re-evaluating families every three years and collecting more detailed information through a standardized questionnaire. We're pleased that even after we meet the original goal of enrolling 1,000 families, we will be able to continue recruitment of new families to this important research study.

Much of the study's success depends on the families who participate. We thank you and your family members for your commitment to this important project. Your participation is invaluable to the many researchers looking to find the genes that increase the risk of late-life AD. 

Progress in the Laboratory

As recruitment continues, scientists are moving forward with the information and samples that have already been collected. Last year, samples from 362 families were sent to the Center for Inherited Disease Research (CIDR) at NIH for genotyping. The genotype data has been completed and is being analyzed. Combined with the family history and other information, this data will give us more specific places to look in the genome for genes associated with AD. 

In the News:

Scientists Find New Genetic Clue to AD

Analyzing DNA samples from 6,000 volunteers, many from families like yours, an international team of researchers uncovered a new gene, known as "SORL1," that appears to be related to late-onset Alzheimer's disease (AD). Variations in the SORL1 gene may be a factor in the development of AD.

"We do not fully understand what causes Alzheimer's disease, but we know that genetic factors can play a role," said NIA director Richard J. Hodes, M.D. "Scientists have previously identified three genes, variants of which can cause early onset Alzheimer's, and one that increases risk for the late onset form. This discovery provides a new genetic clue about the late onset forms of this very complex disease. Further research is needed to determine what role this gene plays in Alzheimer's."

Richard Mayeux, M.D., of Columbia University (who also serves as the lead researcher for the AD Genetics Study), Lindsay Farrer, Ph.D., of Boston University, and Peter St. George-Hyslop, M.D., of the University of Toronto, led the study, which involved 14 collaborating institutions in North America, Europe and Asia, and 6,000 individuals who donated blood for genetic typing. The work was funded by NIH's National Institute on Aging (NIA) and National Human Genome Research Institute (NHGRI), as well as by 18 other international public and private organizations.

To start, the scientists combed two large data sets of genetic information from Northern European and Caribbean Hispanic families in which more than one person has Alzheimer's disease. They were soon able to see that many of the families with Alzheimer's had variations in the SORL1 gene but not consistently in any of the other six genes. *(continued, over)*

New Genetic Clue *(continued)*

They then expanded their search to genetic data sets from families of Caucasian, African American, and Israeli Arab heritage for changes in the SORL1 gene. Again, they found the same association between SORL1 variations and Alzheimer's disease.

Examining blood cells from people with and without Alzheimer's, the researchers found less than half the level of SORL1 protein in people with Alzheimer's compared to people without the disease.

"Involving 6,000 people, this is one of the largest studies ever done on the genetics of Alzheimer's disease," notes Dr. Creighton Phelps, who oversees NIA's Alzheimer's Disease Centers program. "The ethnic diversity of this study, the number of different samples and finding the gene association in so many people suggests a real link between SORL1 and Alzheimer's, one of the first new clues we have had in some time." ❧

Do you know other families like yours?

Please encourage any who may meet the eligibility criteria for this study to contact NCRAD at **1-800-526-2839** or e-mail alzstudy@iupui.edu.

Free Resources

For more info on Alzheimer's disease, contact:

- **National Institute on Aging's Alzheimer's Disease Education and Referral Center**
Phone: 1-800-438-4380
Web: www.alzheimers.nia.nih.gov
- **Alzheimer's Association**
Phone: 1-800-272-3900
Web: www.alz.org

The *National Institute on Aging*, the *Alzheimer's Association*, and *Alzheimer's Disease Centers (ADCs)*, coordinated by the *Columbia University ADC*, are working together to conduct the AD Genetics Study.

For more information, contact the National Cell Repository for Alzheimer's Disease (NCRAD) at 1-800-526-2839, e-mail alzstudy@iupui.edu, or visit the Web site at www.ncrad.org

Changes? Updates? Let Us Know!

As time goes by, it's likely that you and your family members will have new information to add to your history and health information that may be important for the AD Genetics Study.

Study sites have started to re-contact participating families to ensure that our information is up to date with any changes in health or memory, and to collect any missing pieces necessary to complete the family for enrollment in the study. While sites have identified 856 eligible families, only 403 families have been fully completed with all the necessary family members participating. We need complete information about all the identified family members to make the family useful for genetic studies.

Please expect a phone call from your study site (or you can contact them). Follow-up evaluations may include the following:

- 1) an update of family history information;
- 2) cognitive tests and other questionnaires;
- 3) a neurological evaluation; and
- 4) a repeat blood draw (if necessary).

If we are not able to see family members in person, we would like to check in and conduct some questionnaires over the phone .

Please let your study site coordinator know if your contact information changes. *We appreciate your help!* ❧

Study Contact Information

For further information on these or any other issues related to the AD Genetics Study, contact:

STUDY SITE COORDINATOR

[NAME]

[PHONE]

[E-MAIL]

