

Are Down Syndrome and Alzheimer's Disease "Tangled" Together?



Frances Dillon (left) is participating in the Sanders-Brown Center on Aging Down syndrome and Aging Project with the support of her sister, Linda Perkins (right).

When the National Institute of Child Health and Human Development issued a call for proposals in the area of Down syndrome research, Elizabeth Head, Ph.D., and Frederick Schmitt, Ph.D., wanted to come up with a truly innovative project. As investigators in the University of Kentucky Sanders-Brown Center on Aging, they were interested in taking a closer look at possible links between Down syndrome and Alzheimer's disease. Their idea intrigued NICHD, and the investigators were awarded a \$2.4 million grant for a five-year research study of aging in adults with Down syndrome. However, Head and Schmitt know the project cannot succeed without the help of research volunteers and their families.

It's been known for decades that the brains of adults with Down syndrome have characteristics similar to those of people with Alzheimer's disease. Most people with Down syndrome have an extra copy of chromosome 21, causing their brain to produce too many proteins that are linked to Alzheimer's. Both groups of people develop plaques and tangles that may signal dementia.

While other studies look for these



8:30 a.m.

Frances Dillon's appointment starts by reviewing the purpose of the important research and discussing the day's schedule with Roberta Davis, the Down syndrome research administrative coordinator at UK's Sanders-Brown Center on Aging.

proteins as they develop, the new approach Head, Schmitt and the UK research team are taking uses diffusion weighted imaging to examine the brain's fiber connections and to learn more about how the brain is wired. They will then try to link different proteins found in the blood to changes in brain connections, memory and thinking. Research has shown that virtually all individuals with Down syndrome have Alzheimer's changes in their brain by age 40. Interestingly, not all of them are clinically demented at that age—in fact, their risk for developing dementia doesn't go up dramatically until age 50.

"So they have a 10-year period of time where their brains have all these qualities, but they're still functioning well," explained Head, associate professor of Molecular and Biomedical Pharmacology. "What we hope to accomplish with this study is to learn how these people are doing this."

The same phenomenon occurs in the general population, as a number of people come to autopsy with full-blown Alzheimer's in their brains, but yet show no signs of dementia. At this point, it's unclear why these people with all the signs of the disease never develop it, but Schmitt and Head want to find the answer.

Successfully completing the five-year study will require the participation of 40 adults with Down syndrome who are 35

and older. Participants are asked to come to the Sanders-Brown Center on Aging every six months to give a blood sample, take tests that measure thinking skills and memory, and undergo a brain scan. They are also looking for 10 to 12 adults with Down syndrome who have already been diagnosed with dementia to come in for a single visit.

Volunteers are still needed. "People often don't quite understand the importance of research," said Schmitt, professor of Neurology. "Although not inevitable, people with Down syndrome are at a higher risk for Alzheimer's." Schmitt and Head are being contacted a lot more by family members who are not sure how to deal with signs of dementia in their relatives with Down syndrome. They are also partnering with the Down Syndrome Associations of Louisville and Cincinnati to identify potential volunteers, as well as the Kentucky Cabinet for Health and Family Services (Department for Behavioral Health, Developmental and Intellectual Disabilities), and the UK Human Development Institute.

When relatives of Frances Dillon, a 60-year-old with Down syndrome, first learned of the study, they immediately recognized the opportunity it presented. Linda Perkins, Dillon's sister, had learned that her sibling was predisposed to develop Alzheimer's, and wanted to know more about what to expect if it happens.

While Dillon has only completed two

visits to Sanders-Brown, Perkins can already see that Dillon will benefit from participating in the study. She knows she is going back for more sessions and is already excited about her next visit. A relative is showing early signs of Alzheimer's, so Dillon is familiar with the effects of dementia. After her trip to Sanders-Brown, Dillon showed a copy of her MRI scan to everyone she could, and boasted that the doctors told her she has no signs of dementia. "My brain is working great," she would say.

If anyone is on the fence about letting a loved one participate in the study, Perkins urges them to do it. "I think it's wonderful because the more information we have, then the better that Frances, and others, are going to be cared for," Perkins said. "All these other children and adults growing up with Down syndrome will have a better life because of what they are finding out."

Schmitt agrees with Perkins. "That's the real message: That there's hope for your family member, and that hope comes from research," he said. "The reason we're into this is because it's important to us. We want to benefit people. It's about getting an answer so we can do some good."

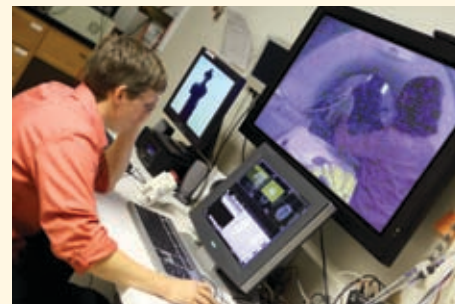
For more information, visit the website for the Down syndrome and Aging Project: <http://www.uky.edu/DSAging/>.



9:05 a.m.
Nadine Deehan and Dillon converse as Dillon's blood is drawn for a routine test and research purpose.



1:37 p.m.
David Powell, Ph.D., walks Dillon through a game to test her memory and cognitive skills.



2:03 p.m.
Dillon's brain scan will help researchers get a better idea of how the brain is put together, how it works and what changes may come about as she gets older. Over time, these scans will be compared to better understand Down syndrome and Alzheimer's disease.