# An Exciting New Chapter in Down Syndrome Research

**THE NATIONAL INSTITUTES OF HEALTH RECENTLY PLEDGED $37 MILLION TO DOWN SYNDROME RESEARCH — AN INVESTMENT WITH THE POTENTIAL TO TRANSFORM MILLIONS OF LIVES, INCLUDING THOSE TOUCHED BY ALZHEIMER'S DISEASE.**

## *SCIENCE SNPs*

### THE BENEFITS OF EARLY MORNING LEARNING

Because children with Down syndrome experience significant problems with sleep and also with learning, researchers at Coventry University in the U.K. predicted that sleep-dependent memory consolidation in these children would be impaired relative to typically developing children. Using a task in which children were taught nicknames for animals with the aid of picture flashcards, they observed that typical children remembered more words following nighttime sleep, demonstrating the active role sleep plays in memory consolidation. In contrast, performance of children with Down syndrome did not significantly change following wake or sleep periods, suggesting disruption of sleep-related memory consolidation. However, children with Down syndrome who had initially trained in the morning performed better than those who trained in the evening. Results suggest that children with Down syndrome may benefit from learning more important or difficult information in the morning.

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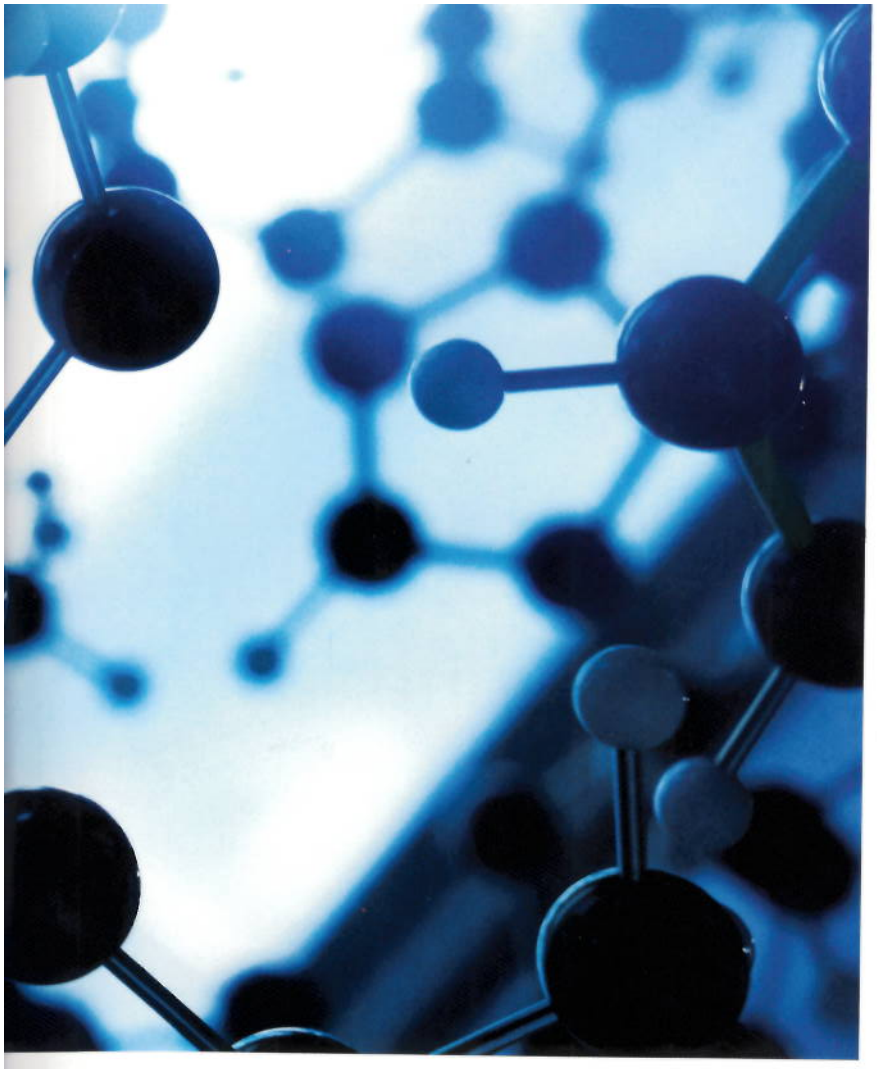
**THERE ARE TWO** main reasons the lifespan of people with Down syndrome has doubled over the last 30 years — medical advances and access to lifesaving procedures that people with Down syndrome were previously denied. But with this increased lifespan, new and critical challenges have emerged — especially that of Alzheimer's disease.

By age 40, all people with Down syndrome will have the brain pathology of Alzheimer's disease — plaques and tangles in the brain that can be picked up by a positron emission tomography (PET) scan of the brain. In addition, an estimated 50 to 70 percent will exhibit clinical and behavioral signs of Alzheimer's disease by age 60. Studying the development and progression of Alzheimer's in people with Down syndrome and understanding why some people with Down syndrome are protected from the dementia, despite having plaques and tangles in their brains, may lead to better diagnostic and treatment strategies that effectively put a stop to Alzheimer's-related dementia.

### BUILDING ON MORE THAN A DECADE OF RESEARCH

The estimated $37 million in funding reflects the largest single research grant awarded to study Down syndrome to date. A collaboration between the National Institute on Aging (NIA) and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) generated this groundbreaking grant. The NIA has a long history of studying diseases of aging, including Alzheimer's disease, while the NICHD funds research on Down syndrome and other pediatric genetic conditions.

"We now know what amyloid plaque and tau tangles look like in typical populations and in people who have a familial risk for Alzheimer's disease,"



# Champions of Progress

## THE NATIONAL INSTITUTES OF HEALTH (NIH) BIOMARKER OF ALZHEIMER'S DISEASE IN ADULTS WITH DOWN SYNDROME INITIATIVE — TWO WOMEN'S COLLABORATIO AND DEDICATION HELPED MAKE IT ALL POSSIBLE.

**MELISSA A. PARISI, M.D.,** Ph.D., is Chief of the Intellectual and Developmental Disabilities Branch of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD). A pediatric geneticist by training, Dr. Parisi earned a medical degree and a doctoral degree in developmental biology from Stanford University. After training i medical genetics, she was Assistant Professor at the University o Washington and Seattle Children's Hospital. Dr. Parisi was draw to the field of pediatric genetics by her desire to understand why some children have differences in growth and development and an interest in helping children with genetic conditions live full and fulfilling lives. She has dedicated her career to researching Down syndrome as well as other genetic conditions.

Laurie Ryan, Ph.D., is Chief of the Dementias of Aging Branch in the Division of Neuroscience at the National Institute on Aging (NIA). Dr. Ryan completed her doctorate in clinical

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said Laurie Ryan, Ph.D., Chief of the Dementias of Aging Branch in the Division of Neuroscience at the National Institute on Aging. "Because we're further along in biomarker discovery, applying these techniques to research Alzheimer's disease in people with Down syndrome is a natural next step."

### THE STUDIES AT A GLANCE

The NIA and NICHD will provide the estimated $37 million to two research teams over the next five years as part of a new project called the NIH Biomarkers of Alzheimer's Disease in Adults with Down Syndrome Initiative. Benjamin Handen, Ph.D., an investigator affiliated with the Department of Psychiatry at the University of Pittsburgh, and Nicole Schupf, Ph.D., an investigator affiliated with Columbia University Medical Center, will each lead a research team.

Through their research, Drs. Handen and Schupf hope to standardize research methodology and create a network of data and tissue samples that will inform future studies. They also hope to identify and track biomarkers that signal Alzheimer's disease development using information generated through the following:

- **PET scans,** which provide a clear picture of how organs and tissues in the body function. PET scans help detect cancer, infections, and changes in organ function, and the researchers will use these scans to track the levels of certain molecules in the brain and look for hallmarks of Alzheimer's disease, such as amyloid plaque deposits and tau tangles.
- **Magnetic resonance imaging (MRI),** which will be used to track the size, or volume, of the brain and brain function.
- **Blood tests** that will track biomarkers, such as cholesterol levels, and look for genes that may raise someone's risk of Alzheimer's disease or, conversely, offer protection against the accumulation of Alzheimer's disease-related amyloid plaque and tau tangles.

Because so many people with Down syndrome develop plaques and tangles in the brain without developing dementia, these researchers hope to identify blood biomarkers, including potentially genetic changes, that predict who will develop dementia among both the Down syndrome and the typical populations.

"Interestingly, typical people with Alzheimer's disease have developed many trisomy or Down syndrome-like cells throughout their bodies, including the blood and the brain. This mechanistic link between the two disorders has many important implications," said Huntington Potter, Ph.D., Director of Alzheimer's Research at the Linda Crnic Institute for Down Syndrome.

"Down syndrome offers a window into the study of Alzheimer's disease that is unparalleled," Dr. Potter added. "The NIH's recognition of the importance of investigating Alzheimer's disease in this special population represents a very big advancement in furthering the science behind Alzheimer's disease development."