UC San Diego Launches Unprecedented Down Syndrome Study

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To many, Down syndrome (DS) is a childhood condition. But improved health care means that individuals with DS now routinely reach age 50 or 60 years of age, sometimes beyond. However, if they live long enough, people with Down syndrome are almost certain to develop Alzheimer's disease (AD).

Risk estimates vary, but the National Down Syndrome Society says that nearly 25 percent of individuals with DS over the age of 35 show signs of Alzheimer's-type dementia, a percentage that dramatically increases with age. Almost all develop dementia by the age of 60.

"The more we learn about Down syndrome and Alzheimer's disease, the more we realize these conditions – one seen at birth, the other quite late in life – are two sides of the same coin," said William C. Mobley, MD, PhD, professor and chair of the Department of Neurosciences at UC San Diego School of Medicine. "Autopsies of DS and AD brains reveal virtually identical pathologies – the same telltale amyloid plaques and neurofibrillary tangles."

Under the auspices of the Alzheimer's Disease Cooperative Study (ADCS), based at the University of California, San Diego School of Medicine, a new clinical study called the Down Syndrome Biomarker Initiative (DSBI) was launched in March 2013. According to the study's director, Michael Rafii, MD, PhD – medical director of the ADCS – its aim is to discover indicators of Alzheimer's and study progression of the disease, with the ultimate goal of better understanding brain aging and AD in adults with Down syndrome.

The three-year pilot study has enrolled 12 participants, aged 30 to 60 years of age. Study participants will be screened for various biomarkers of AD, using tests that include three types of brain scans, retinal amyloid imaging and blood tests, among others.

"Findings to date using MRI and amyloid PET scans indicate that individuals with Down syndrome show the same brain patterns as those in the general population with the earliest stages of the memory-robbing disease, called prodromal AD," said Rafii. He added that indications of increased brain amyloid deposition – the insoluble protein aggregates found in the brains of patients with AD

that are thought to be an underlying cause of the disease – is similar in individuals with DS and those in the general population with AD.

People with amyloid deposition in the brain experience progressive cognitive deterioration. Brain atrophy – shrinking of the brain's hippocampus – caused by the amyloid buildup, affects routine functional abilities, ultimately leading to complete physical disability.

"By understanding the progression of the disease in people with Down syndrome and those in the general population, we hope discoveries can be made in each group that can be shared between both populations," said Rafii.

The design of the DSBI pilot study is patterned after the Alzheimer's Disease Neuroimaging Initiative (ADNI), which began in 2004 to establish neuroimaging and biomarker measures of AD. ADNI tracked the changes taking place in the brains of 800 older people, either free of symptoms or diagnosed with late-stage mild cognitive disorder and early Alzheimer's disease.

"Our aim is for the Down Syndrome Biomarker Initiative to mirror ADNI's successes," Rafii said. "ADNI has helped the international Alzheimer's research community learn significant lessons about the pathology and biomarkers of AD, which in turn has driven new ways of looking at the disease and new studies that we hope will lead to viable treatments. We are confident we can do the same thing for Down syndrome."

The 12-subject pilot study at UC San Diego is funded by Janssen Research & Development, LLC. The research is projected to expand into a five-year, 1,000-subject international study.

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