

# Biomarkers of Alzheimers Disease in Adults with Down Syndrome

<https://www.neurodegenerationresearch.eu/survey/biomarkers-of-alzheimers-disease-in-adults-with-down-syndrome/>

## Principal Investigators

SCHUPF, NICOLE

## Institution

COLUMBIA UNIVERSITY HEALTH SCIENCES

## Contact information of lead PI

### Country

USA

## Title of project or programme

Biomarkers of Alzheimers Disease in Adults with Down Syndrome

## Source of funding information

NIH (NIA)

## Total sum awarded (Euro)

€ 17,760,066.97

## Start date of award

30/09/2015

## Total duration of award in years

2

## The project/programme is most relevant to:

Alzheimer's disease & other dementias

## Keywords

Acquired Cognitive Impairment... Aging... Alzheimer's Disease... Alzheimer's Disease including Alzheimer's Disease Related Dementias (AD/ADRD)... Brain Disorders... Clinical Research... Clinical Research - Extramural... Dementia... Diagnostic Radiology... Down Syndrome... Genetics... Intellectual and Developmental Disabilities (IDD)... Neurodegenerative... Neurosciences

### **Research Abstract**

? DESCRIPTION (provided by applicant): By age 40 years, individuals with Down syndrome (DS) show the neuropathological changes of Alzheimer's disease (AD) and have a high risk for dementia, but little is known about the biomarkers that may predict clinical onset or reflect disease progression. This study focuses on a longitudinal and multidisciplinary determination of key biomarkers that are likely to define this progression, including levels and rates of change in blood based biomarkers such as  $\beta$ -amyloid peptides, protein and lipid profiles, and measures of amyloid and tau concentration in cerebrospinal fluid, neuroimaging-based changes and genetic polymorphisms. Using a neurocognitive battery that we have developed and tested, systematic profiles of longitudinal stability and of decline will allow us to define dementia status, including Mild Cognitive Impairment in DS (MCI-DS), and characterize progression in clinical status. Previously generated protein, inflammatory and lipid signatures will be examined, as well as amyloid and tau profiles in cerebrospinal fluid (CSF). Imaging biomarkers will include structural MRI components and PET studies of brain amyloid uptake. Analysis of MRI imaging biomarkers will include longitudinal measures of atrophy, white matter abnormalities and intrinsic network connectivity paradigms. Amyloid positron tomography will delineate regional and whole brain uptake of amyloid. Polymorphisms in candidate genes for AD and related biomarkers will be studied as potential modifiers of risk and their relation to beta amyloid, proteomic, lipidomic and imaging biomarkers examined. Relationships among demographic, clinical, blood based and CSF biomarkers, imaging measures, and genetic variants will be examined to develop the most valid indicators of preclinical and early stages of AD. Importantly, the data and the biological samples will be archived and banked to establish a resource to be shared with other scientists. Collectively, our investigators have a combined clinical and research experience involving over 1500 patients (30% demented), over 850 banked blood samples, 500 DNA samples, and 50 imaging studies. Further, team investigators have previous experience with all methods that will be included in this new project. Thus, this application brings together a group of co-investigators with established expertise in studies of DS and makes available a combined cohort of 280 participants.

### **Lay Summary**

**PUBLIC HEALTH RELEVANCE** Individuals with Down syndrome have a high risk for Alzheimer's disease, but little is known about the biomarkers that characterize disease onset and progression or why some individuals develop dementia much earlier than others. This study focuses on a multidisciplinary determination of key risk as well as protective biomarkers that are likely to affect AD progression, including blood-based, CSF-based and imaging-based biomarkers, and polymorphisms in AD-related genes. Study of biomarkers for early dementia changes may yield critical data documenting the transition from normal aging to mild cognitive impairment to clinical dementia in individuals with DS. This can provide key insights into the pathways involved in AD progression and may allow for future therapeutic interventions before irreversible cognitive deterioration has occurred.

**Further information available at:**

**Types:**

Investments > €500k

**Member States:**

United States of America

**Diseases:**

Alzheimer's disease & other dementias

**Years:**

2016

**Database Categories:**

N/A

**Database Tags:**

N/A