Examination of the earliest symptoms and biomarkers of FTLD MAPT carriers

https://neurodegenerationresearch.eu/survey/examination-of-the-earliest-symptoms-and-biomarkers-of-ftld-mapt-carriers/

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Contact information of lead PI
Country

USA

Title of project or programme

Examination of the earliest symptoms and biomarkers of FTLD MAPT carriers

Source of funding information

NIH (NIA)

Total sum awarded (Euro)

€ 2,270,164.22

Start date of award

21/09/2012

Total duration of award in years

1

The project/programme is most relevant to:

Alzheimer's disease & other dementias

Keywords

MAPT gene, Frontotemporal Dementia, tau mutation, mutation carrier, carrier status

Research Abstract

DESCRIPTION (provided by applicant): Discoveries about the genetic bases of frontotemporal lobar dementia (FTLD) have provided researchers with an unprecedented ability to characterize

the pre-symptomatic stages of this disease. This capability is critical to ascertaining the earliest clinical features of the disease and identifying biomarkers that can be used for early diagnosis and in treatment studies. The proposed project will examine individuals from a single family with a genetic mutation for FTLD who are approaching the age of disease onset, offering a unique and highly controlled environment in which to identify the earliest clinical features and biomarkers of this devastating disease. Specifically, this study proposes to follow the offspring generation of a large family with a known mutation in the MAPT (tau) gene to determine the earliest clinical features of FTLD and the variables which may modify disease onset and course. At least 90 members of the offspring generation of a single family will be offered participation in a longitudinal study examining cognition, behavior, psychiatric symptoms, and lifestyle features. Moreover, this proposal includes the use of state of the art neuroimaging tools including both structural and functional modalities, gray and white matter quantification, and regional and network analyses to detect the earliest signs of disease. Individuals will undergo genetic testing to determine carrier status, will be comprehensively characterized at baseline, and will be followed annually over the course of the study to examine change in clinical and imaging variables over time as a function of carrier status. Additionally, the proposed study will carefully characterize lifetime physical activity and alcohol and recreational drug use to determine if these lifestyle variables are early manifestations of disease and / or modify disease onset and course. Finally, biological specimens including cerebrospinal fluid and blood plasma will be longitudinally collected in an effort to characterize changes in brain protein levels that may provide early information regarding the onset and course of disease.

Lay Summary
PUBLIC HEALTH RELEVANCE: Improved characterization of FTLD at its earliest stages is a necessary step in facilitating earlier disease detection and developing disease modifying therapeutics that can be applied in advance of irrevocable neuropathological changes.

Further information available at:

Types:
Investments > €500k

Member States:
United States of America

Diseases:
Alzheimer's disease & other dementias

Years:
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