

Clinical, Pathological, Genetic and Biomarker Studies of Frontotemporal Dementia

<https://neurodegenerationresearch.eu/survey/clinical-pathological-genetic-and-biomarker-studies-of-frontotemporal-dementia/>

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Contact information of lead PI Country

Canada

Title of project or programme

Clinical, Pathological, Genetic and Biomarker Studies of Frontotemporal Dementia

Source of funding information

CIHR

Total sum awarded (Euro)

€ 1,050,344

Start date of award

01/04/2013

Total duration of award in years

5.0

The project/programme is most relevant to:

Neurodegenerative disease in general

Keywords

Research Abstract

Frontotemporal dementia (FTD) is a devastating disease that is characterized by profound changes in behavior, personality, language and movement. It is the second most common cause of dementia under the age of 70 years, most frequently having its onset between the ages of 40 and 65. Almost half of FTD patients have a positive family history, often with a very strong inheritance pattern. Unfortunately, there are still significant gaps in our understanding of

how the disease begins and progresses which have hampered attempts to develop accurate diagnostic tests and effective treatments. In 2006, we discovered that mutations in the progranulin gene cause 5-10% of all FTD and 20-25% of its familial forms. This spurred our efforts to better characterize the presymptomatic, prodromal and full-blown stages of FTD. Initial studies of subjects from our FTD families with progranulin mutations have demonstrated distinctive changes on neuropsychological testing and neuroimaging that precede the development of dementia by many years. This past year (2011), we discovered another gene (C9ORF72), in which mutations are an even more common cause of FTD and which are also the most common genetic cause of amyotrophic lateral sclerosis (ALS), linking these two neurodegenerative diseases. As a result of this research, we now have one of the largest and best-characterized collections of clinical, genetic, and neuropathological FTD materials of any center in the world. Continuing to investigate and follow our FTD and ALS subjects will allow us to define the earliest stages of these disorders. Combining this knowledge with our genetic and pathological data will assist us in developing new diagnostic markers and will set the stage for the development of novel and rational therapeutic approaches for these currently untreatable diseases.

Lay Summary

Further information available at:

Types:

Investments > €500k

Member States:

Canada

Diseases:

Neurodegenerative disease in general

Years:

2016

Database Categories:

N/A

Database Tags:

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