# Molecular insights in inherited dementia disorders – Cerebrospinal fluid biomarkers and stem cell derived neuron pathology

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**Country**

Denmark

**Title of project or programme**

Molecular insights in inherited dementia disorders - Cerebrospinal fluid biomarkers and stem cell derived neuron pathology

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Lundbeckfonden

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€ 67,264

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3

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**Research Abstract**

Nearly 40 million people are affected by dementia worldwide. The number is estimated to double every 20 years, thus becoming a global socioeconomic and medical problem. Alzheimer’s disease (AD) is the most prevalent cause of dementia in elderly whereas frontotemporal dementia (FTD) is the most common cause of early onset dementia. The diagnosis of AD and FTD is based on history, neurological examination, neuropsychological

evaluation, brain imaging, blood and cerebrospinal fluid (CSF) biomarkers. However, it is difficult to distinguish AD and FTD from one another. Patients are often misdiagnosed and consequently mis-medicated. For a more accurate, fast and less costly diagnostic set-up a new battery of more specific biomarkers is needed. Rare genetic forms of diseases have been extremely instrumental in elucidating disease mechanisms and generating treatments for many common disorders. As partner in the project “Patient-specific stem cell-derived models for Alzheimer’s disease” funded by the Danish National Advanced Technology Foundation we have access to stem cells reprogrammed from fibroblasts from individuals with those rare familial forms of AD and FTD. Investigation of CSF from patients with familial AD and FTD by unbiased proteomics will shed light upon new potential biomarkers for use in the clinic. Additionally, we will study and cross validate biomarkers by immunoassays in both neurons and CSF from and compare findings to sporadic forms and other familial dementia disorders. The exact molecular mechanisms responsible for triggering the cellular pathology seen in AD and FTD are not yet known and we will investigate endosome fusion, microtubule transport, tau and Aß pathology in the neurons by applying well-established and novel biochemical assays. This project will be the first to investigate biomarkers in patients with familial AD and FTD using unbiased proteomics and patient-specific fibroblast-derived neurons.

**Further information available at:**

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Denmark

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