Identification of molecular and genetic mechanisms in familial Lewy Body Dementia: a novel approach

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Country

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Identification of molecular and genetic mechanisms in familial Lewy Body Dementia: a novel approach

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ZonMw

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4.0

The project/programme is most relevant to:

Alzheimer's disease & other dementias

Keywords
Research Abstract

Dementia with Lewy Bodies (DLB) is the second most common dementia after Alzheimer's Disease in the elderly. Recognizing DLB remains challenging due to the highly variable clinical

presentation (varying from cognitive fluctuations, parkinsonism, hallucinations, to sleep disorders) and considerable overlap with AD and Parkinson's Disease (PD). The early diagnosis is further hampered by the lack of cerebrospinal fluid (CSF) and genetic biomarkers. As a consequence, DLB is diagnosed on average 3 years after symptom-onset, a year later than AD. Differentiation from other dementias is clinically relevant, because of a high sensitivity for cholinesterase inhibitor therapy and to avoid the detrimental effects of antipsychotics in DLB.

Aggregation of dementia and PD in families with DLB suggests that genetic susceptibility plays an important role. However, the genetic and molecular mechanisms are still poorly understood. Whole exome sequencing (WES) is a novel genetic technique used to selectively sequence the coding regions of the genome, which has proven successful in identifying functional variation in small families and in genetical and phenotypical heterogeneous disorders.

Main objective: to identify novel biomarkers to improve the early diagnosis of DLB.

Aims:
1. to identify proteins that potentially serve as novel biomarker in DLB using brain tissue (Lewy bodies) and cerebrospinal fluid proteomics in patients with familial and pathological proven DLB.

2. to identify novel genetic factors in familial DLB using whole exome sequencing in several families with (pathological proven) DLB.

Lay Summary
Further information available at:

Types:
Investments > €500k

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Netherlands

Diseases:
Alzheimer's disease & other dementias

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2016

Database Categories:
N/A

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