Exome sequencing in dementias – more than a family business

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Alzheimer's Society

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United Kingdom

Title of project/programme

Exome sequencing in dementias - more than a family business

Source of funding information

Alzheimer's Society

Total sum awarded (Euro)

€ 542,300

Start date of award

01/08/15

Total duration of award in years

4.0

The project/programme is most relevant to:

Alzheimer's disease & other dementias

Keywords
Research Abstract

Background: The development and application of next generation sequencing technologies has allowed for an exponential increase in the number of findings of genetic causes associated with Mendelian diseases. One general and consistent finding has been the extent of previously unrecognized allelic heterogeinity and genetic pleomorphism associated with neurological

diseases.

Hypothesis: I hypothesize that by studying rare familial forms of dementia I will identify genetic factors that also play a role in Alzheimer’s disease (AD).

Specific Aims: 1) To identify the genes and mutations causing rare forms of dementias; 2) to assess the role of these newly identified genes in AD; 3) to integrate these results with available genetic and functional data to better characterize the pathogenicity of variants, and 4) to clarify the molecular associations between different forms of dementia.

Study Design: Exome sequencing will be performed in families presenting with rare forms of dementias. The genes identified will then be tested for association with AD using publicly available sequencing and genotyping data. The results will be integrated in order to improve the interpretation of variants found in dementia cases and clarify the molecular relationships between the different clinical entities.

Relevance: This strategy will allow the identification of genetic causes of disease in different families, which will have a direct impact in clinical management. Understanding the full spectrum of genetic factors involved in dementia will give us information on pathobiological events and will lead to the identification of potential therapeutic targets.

Types:
Fellowships

Member States:
United Kingdom

Diseases:
Alzheimer's disease & other dementias

Years:
2016

Database Categories:
N/A

Database Tags:
N/A