A genomewide association study of neurodegeneration in a characterized and genetically defined population

https://www.neurodegenerationresearch.eu/survey/title-of-pia-genomewide-association-study-of-neurodegeneration-in-a-characterized-and-genetically-defined-population/

Title of project or programme

Title of PI	A genomewide association study of neurodegeneration in a characterized and genetically defined population

Principal Investigators of project/programme grant

Title	Forname	Surname	Institution	Country
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Source of funding information

Health Research Board

Total sum awarded (Euro)

1693469

Start date of award

01-12-2007

Total duration of award in months

60

The project/programme is most relevant to

- Motor neurone diseases

Keywords

neurodegeneration; amyotrophic lateral sclerosis; clinical phenotype; frontotemporal dementia; genetic susceptibility

Research abstract in English

Amyotrophic lateral sclerosis (ALS) is a rapidly progressive, fatal neurodegenerative disorder of unknown etiology. Sporadic ALS is a complex genetic disease, in which ethnic/genetic background and environmental risks interact leading to the process of neurodegeneration. My group has identified a novel series of loss of function mutations in ANG, a gene not hitherto associated with neurodegeneration. Although originally considered as a pure motor degeneration, it is now recognized that considerable overlap exists between ALS and other neurodegenerative diseases, particularly frontotemporal dementia(FTD). Despite recent advances, common pathways shared between ALS and other neurodegenerative disease remain to be fully elucidated. The principal research hypothesis underling this project is that novel genes for neurodegeneration can be identified by phenotype/genotype characterization of a genetically homogenous population. This unique project will collect clinical phenotypes of ALS in the Irish population that will be stratified according to strict clinical and neuropsychological criteria. This clinical dataset will be complemented by an associated genetic resource that will permit detailed genetic characterization by whole genome association. This process will allow detailed phenotype genotype correlation, the later generation of endophenotypes within ALS kindreds and thus dissection of common pathways involved in the process of neurodegeneration. This work is likely to lead to the discovery of novel genes associated with neurdegeneration, and new pathways that can be targeted for therapeutic intervention.

In which category does this research fall?

- Basic research

Lay Summary