Whole genome sequencing in patients and families with dementia: building an open access UK resource

https://www.neurodegenerationresearch.eu/survey/whole-genome-sequencing-in-patients-and-families-with-dementia-building-an-open-access-uk-resource/

Principal Investigators

Professor Henry Houlden

Institution

University College London

Contact information of lead PI
Country

United Kingdom

Title of project or programme

Whole genome sequencing in patients and families with dementia: building an open access UK resource

Source of funding information

Alzheimer's Society

Total sum awarded (Euro)

€ 135,573

Start date of award

06/08/2016

Total duration of award in years

2

Keywords
Research Abstract
Aims: The Genomics England 100,000 genomes initiative represents an unprecedented opportunity for UK patients, clinicians and biomedical researchers. The project is only operational until the end of 2017 and patients have to be actively recruited by clinical teams. Alzheimer’s disease (AD), frontotemporal dementia (FTD) and rarer forms of dementia are highly heritable traits and genetically undefined patients are eligible disorders for genome

sequencing. We have to maximise this opportunity as the more dementia samples that are submitted and sequenced, the greater value this resource will be. We wish to obtain a specific genome sequencing nurse/coordinator to work with as many clinicians around the UK as possible, particularly when difficult for patients to travel or where relatives are distant. The aim is to consent and send blood for a total of 1200 individuals (400 trios/families) over 2 years. Methods: Families/trios will be recruited, consented and DNA will be extracted and sent to Genomics England for genome sequencing in AD, FTD and rare dementias. In addition blood will be taken for mRNA, plasma and serum and stored by Genomics England, along with DNA this can be openly requested for research. Outcomes: This open access resource will undoubtedly lead to improved diagnosis, the identification of new Mendelian genes/risk factors and the facilitation of biomarker and therapeutic pathway discovery. Results will be made available to the consultant and genome medicine centre where the patient is clinically seen and to the Genomics England Clinical Interpretation Partnership (GeCIP) for any UK clinician or scientist to access.

Further information available at:

Types:
Investments < €500k

Member States:
United Kingdom

Diseases:
N/A

Years:
2016

Database Categories:
N/A

Database Tags:
N/A