

# Powerful Study of Rare Variants in Alzheimer's disease

<https://neurodegenerationresearch.eu/survey/powerful-study-of-rare-variants-in-alzheimers-disease/>

**Name of Fellow**

Dr Rebecca Sims

**Institution****Funder**

Alzheimer's Society

**Contact information of fellow****Country**

United Kingdom

**Title of project/programme**

Powerful Study of Rare Variants in Alzheimer's disease

**Source of funding information**

Alzheimer's Society

**Total sum awarded (Euro)**

€ 271,160

**Start date of award**

01/04/12

**Total duration of award in years**

3.0

**The project/programme is most relevant to:**

Alzheimer's disease & other dementias

**Keywords****Research Abstract**

Identifying and studying genes that contribute to Alzheimer's disease (AD) development will help us to understand the causes of AD, providing the basis for developing new treatments. Common variation in nine novel genes are known to increase susceptibility to AD. However, these variants only account for roughly 32% of the genetic aetiology of the disease. Rare

variation is thought to account for a proportion of the missing heritability. Next generation sequencing (NGS) allows the identification and analysis of rare variants (RV). However, NGS remains expensive and thus does not allow investigators to utilise suitably powered samples to comprehensively test RV across the genome for association with disease. This fellowship will investigate the role of RV in AD using a powerful case-control study. It will utilise a novel exome chip, designed using 12,000 exome sequences and the 1000 genomes project, to assay 300,000 genetic variants. It will develop new skill sets across genetics, bioinformatics and biostatistics to develop novel algorithms and programs to analyse RV. Thus, identifying novel loci that increase risk for AD which can be comprehensively investigated through cost effective, targeted sequencing and genotyping efforts, further elucidating the genetic architecture of AD.

**Types:**

Fellowships

**Member States:**

United Kingdom

**Diseases:**

Alzheimer's disease & other dementias

**Years:**

2016

**Database Categories:**

N/A

**Database Tags:**

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