ESMI: European Spinocerebellar Ataxia Type 3/Machado-Joseph Disease Initiative

https://www.neurodegenerationresearch.eu/survey/esmi-european-spinocerebellar-ataxia-type-3machado-joseph-disease-initiative/

Principal Investigators

T. Klockgether, P.Giunti, M.Lima, LP de Almeida, O.Riess, B van de Warrenburg

Institution

Multiple

Contact information of lead PI
Country

Germany|United Kingdom|Portugal|Netherlands

Title of project or programme

ESMI: European Spinocerebellar Ataxia Type 3/Machado-Joseph Disease Initiative

Source of funding information

JPND-JPcofuND

Total sum awarded (Euro)

€ 1,557,261

Start date of award

01/01/2016

Total duration of award in years

3.0

The project/programme is most relevant to:

Spinocerebellar ataxia (SCA)

Keywords
Research Abstract

Spinocerebellar ataxia type 3/Machado-Joseph disease (SCA3) is the most common familial ataxia. Although the gene mutation causing SCA3 is known, there is no treatment. However, as there is an advanced understanding of the mechanisms underlying SCA3, new therapeutic approaches are being developed. To enable drug trials, the availability of large cohorts of people who carry the mutation is mandatory. ESMI will bring together 8 cohorts comprising

more than 800 subjects. We will integrate the existing data in a common database and apply standardized and quality-controlled assessment protocols. A major part of our initiative will be the development of new disease markers. The expected results have immediate relevance for application in clinical research and in routine health care. Bringing together existing cohorts will greatly facilitate the enrolment of participants in drug trials. The development of disease markers will allow for proof of concept studies with a biomarker out- come that require smaller numbers of participants than conventional trials. The data on the long- term evolution of the disease will inform statisticians who design clinical trials in SCA3. Lastly, our research will have a direct impact on health care, as novel instruments can be used to further improve the clinical management of ataxia patients.

Lay Summary
Further information available at:

Types:
Investments > €500k, JPND Projects

Member States:
Germany, JPND, Netherlands, Portugal, United Kingdom

Diseases:
Spinocerebellar ataxia (SCA)

Years:
2016

Database Categories:
N/A

Database Tags:
N/A