Triplet repeat polymorphisms as modifiers of health and disease

https://www.neurodegenerationresearch.eu/survey/triplet-repeat-polymorphisms-as-modifiers-of-health-and-disease/

Question
Name of Fellow

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Related
Institution
Funder

ZonMw

Contact information of fellow
Country

The Netherlands

Title of project/programme

Triplet repeat polymorphisms as modifiers of health and disease

Source of funding information

ZonMw

Total sum awarded (Euro)

€ 250,000

Start date of award

01/01/15

Total duration of award in years

4.0

The project/programme is most relevant to:

Huntington's disease

Keywords

Triplet repeat polymorphisms | polyglutamine diseases | Huntington disease | depression | dementia

Research Abstract
Diseases such as dementia, depression and diabetes are among the leading causes of disability and exert a dramatic burden on society’s social, economic and health care systems. In order to understand their pathophysiology and devise more effective therapies it is essential to elucidate their genetic basis. However, to date genetic association studies have only identified a small fraction of the genetic determinants, possibly because of the focus on single-nucleotide polymorphisms and consequently neglect of other important genomic variations, especially DNA repeat expansions. Expanded DNA repeats above a certain threshold are associated with many hereditary neurological disorders, the most common of which are polyglutamine diseases caused by intronic triplet (cytosine-adenine-guanine (CAG)) repeat expansions leading to a range of cognitive, psychiatric, motor and metabolic abnormalities. Emerging findings, including our own pilot data, suggest that even CAG repeat length variations in the normal range

Types:
Fellowships

Member States:
Netherlands

Diseases:
Huntington's disease

Years:
2016

Database Categories:
N/A

Database Tags:
N/A