Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6 (SCA1, SCA2, SCA3, SCA6)

https://neurodegenerationresearch.eu/de/cohort/prospective-study-of-individuals-at-risk-for-spinocerebellar-ataxia-type-1-type-2-type-3-and-type-6-sca1-sca2-sca3-sca6/

Cohort Acronym
RISCA

Cohort type
Rare neurodegenerative condition

Disease
Spinocerebellar ataxia (SCA)

Participant type
At-risk diagnosis

Profile
Recruitment Period
Sample size at start or planned sample size if still recruiting 300
Estimated Current Sample Size 0 to 4,999
Age at Recruitment >18
Gender Male and Female
Abstract

The aim of RISCA is to answer the following questions:

1. What is the incidence of disease manifestation in mutation carriers?
2. Which clinical signs precede the onset of manifest ataxia in SCA1, SCA2, SCA3 and SCA6?
3. What are the prevalence and incidence of preceding signs?
4. Are the prevalence and incidence of preceding signs affected by genotype, gender, age, estimated time until disease manifestation and repeat length?
5. Does the presence of certain preceding signs predict the manifestation of ataxia?
6. Are there MRI alterations that precede the onset of ataxia?

Study participants are followed at 24 months intervals over six years and than at irregular intervals. At each visit, study participants are asked in a structured interview for a number of predefined clinical signs that potentially precede the onset of ataxia.

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Contact details

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Funders (Core support) E-Rare program

Variables Collected

Brain related measures:
Cognitive function, Mental health
Funtional rating:
Individual physiological, Individual psychological
Anthropometric:
Height, Weight
Physical:
N/A
Biological samples:
Blood, Urine
Genotyping:
N/A
Brain imaging:
N/A
Brain banking:
N/A
Lifestyle:
Alcohol, Physical activity, Smoking
Socio-economic:
N/A
Health service utilisation:
N/A