

# Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6

<https://neurodegenerationresearch.eu/survey/prospective-study-of-individuals-at-risk-for-spinocerebellar-ataxia-type-1-type-2-type-3-and-type-6/>

## **Title of cohort**

Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6

## **Acronym for cohort**

RISCA

## **Name of Principal Investigator - Title**

Prof

## **Name of Principal Investigator - First name**

Thomas

## **Name of Principal Investigator - Last name**

Klockgether

## **Address of institution -Institution**

University of Bonn

## **Address of institution - Street address**

Sigmund-Freud-Str. 25

## **Address of institution - City**

Bonn

## **Address of institution - Postcode**

53105

## **Country**

Germany

## **Website**

<http://www.ataxia-study-group.net>

**Contact email**

[klockgether@uni-bonn.de](mailto:klockgether@uni-bonn.de)

**Funding source**

**Q1a. Please indicate below if your cohort includes or expects to include, incidence of the following conditions?**

Spinocerebellar ataxia (SCA)

**Q1b. When are studies on the above condition(s) expected to become possible?**

Already Possible

**Q2a. In a single sentence what is the stated aim of the cohort?**

To enrol and study a cohort of asymptomatic gene carriers for spinocerebellar ataxia

**Q2b. What distinguishes this cohort from other population cohorts?**

**Q3a. i) Number of publications that involve use of your cohort to date**

0

**Q3a.ii) Please give up to three examples of studies to date (Principal Investigator, Institution, Title of Study)**

**Q3b. If data on research outputs are already available please paste the publication list/other data or provide a link to where these data are publicly available**

**Q3c. If no research has been done as yet, please explain in a few sentences what information (i.e. research findings) you expect will be gained from the population**

**Q4a. Study criteria: what is the age range of participants at recruitment? Age in years From:**

18

**Q4a. Study criteria: what is the age range of participants at recruitment? To:**

Until death

**Q4b. Study criteria: what are the inclusion criteria?**

First-degree relative of a patient with SCA1, SCA2, SCA3 or SCA6

**Q4c. Study criteria: what are the exclusion criteria?**

Manifested ataxia

**Q5. What is the size of the cohort (i.e. how many participants have enrolled)?**

1,000-5,000 participants

**Q6a. Please describe what measures are used to characterise participants**

Clinical scale, timed tests, MRI, electrophysiology

**Q6b. Are there additional measures for participants with a clinical disorder?**

**Q6c. Are there defined primary and secondary endpoints (e.g. defined health parameters)?**

Yes

**If yes please specify**

Manifestation of ataxia

**Q7. What is the study design (select all that apply)?**

Prospective cohort

**Q8. Are your cases matched by**

**Q9a. Does your study include a specialised subset of control participants?**

No

**Q9b. If your study includes a specialised subset of control participants please describe**

**Q10a. i) Please enter the data collection start date**

01/09/2008

**Q10a. ii) Please enter the data collection end date**

**Q10a. iii) Is data collection for this study**

Data collection ongoing| Closed to new patients

**Q10b. If data collection is ongoing, are there plans to continue the cohort study beyond the current projected end date?**

Yes - intend to apply for funding

**Q11. Is data collected**

**Other please specify here**

**Q12. Is there a system in place to enable re-contact with patients to ask about participation in future studies?**

**Q13a. Please give information on the format and availability of data stored in a database (1)**

% available

**Q13a. Please give information on the format and availability of data stored in a database (2)**

% available

**Q13a. Please give information on the format and availability of data stored in a database (3)**

% available

**Q13a. Please give information on the format and availability of data stored in a database (4)**

% available

**Other (please specify)**

% available

**Q13b. Please give information on the format and availability of data held as individual records (1)**

% available

**Q13b. Please give information on the format and availability of data held as individual records (2)**

% available

**Q13b. Please give information on the format and availability of data held as individual records (3)**

% available

**Q13b. Please give information on the format and availability of data held as individual records (4)**

% available

**Please specify language used**

**Q14a. Is data available to other groups?**

No

**Q14b. If data is available to other groups what is the access policy/mechanisms for access?**

**Q15. What data sharing policy is specified as a condition of use?**

No requirement to make data publicly available

**Q16a. Are tissues/samples/DNA available to other groups?**

Yes

**Q16b i) If yes, please describe below:**

**Q16b. ii) In what form are tissues/samples/DNA supplied?**

**Q16b. iii) Is the access policy/mechanism for obtaining samples the same as that for obtaining data (Q14 above)?**

**Q17. Is information on biological characteristics available to other groups?**

Yes, for all the cohort

**Number of Patients**

**% of total cohort**

**Types:**

Population Cohorts

**Member States:**

Germany

**Diseases:**

Spinocerebellar ataxia (SCA)

**Years:**

2016

**Database Categories:**

N/A

**Database Tags:**

N/A