

Huntingtons disease

<https://www.neurodegenerationresearch.eu/survey/huntingtons-disease/>

Title of the register

Huntingtons disease

Name of Principal Investigator - Title

Prof

Name of Principal Investigator - First name

Evzen

Name of Principal Investigator - Last name

Ruzicka

Address of institution -Institution

Dept. of Neurology and Center of Clinical Neuroscience, Charles University in Prague

Address of institution - Street address

Katerinska 30

Address of institution - City

Prague

Address of institution - Postcode

12000

Country

Czech Republic

Website

<http://www.neuro.lf1.cuni.cz/>

Contact email

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

Huntington's disease|Parkinson's disease|Neurodegenerative disease in general

Q2. In a single sentence, what is the stated aim of your register?

concentrate patients with rare diseases, provide and improve specialised healthcare, to have a source of suitable subject for cooperation in clinical trials and research

Q2b. What distinguishes this register from other disease registers?

e.g. Huntington's disease register is the only one in our country.

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

50

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

Jan Motlik, Czech Academy Science, Institute of Animal Physiology and Genetics-PIGMOD Center, , Transgenic minipig for Huntington's disease (TgHD)-funded by CHDI, Czech-Norwegian Research Programme 7F14308, National Programme of Sustainability LO1609|Radoslav Matej, First Faculty of Medicine, Charles University in Prague and Thomayer Hospital in Prague, Diagnostic markers and pathophysiological mechanisms of atypical parkinsonian syndromes|Petr Dusek, Dept. of Neurology and Center of Clinical Neuroscience, Charles University in Prague, Biomarkers of progression and treatment response in neurodegenerative disorders

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?

all recent publications are available in <https://www.ncbi.nlm.nih.gov/pubmed>

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 register

Q4a. Study criteria: what is the age range of participants? Age in years: from

15

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until death

Q4b. Study criteria: what are the inclusion criteria?

clinically possible or probable diagnosis, results of genetic testing

Q4c. Study criteria: what are the exclusion criteria?

unclear diagnosis, advanced stage

Q5. What is the size of the register (i.e. how many patients have been enrolled)?

501-1000 clinical cases

Q6a. Please describe what measures are used to characterise participants

Yes

Q6b. Are there defined primary and secondary endpoints (e.g. defined health parameters)?

occurrence of a disease, symptom, sign or laboratory abnormality, clinical trial

If YES, please describe

Q7a. i) Is the register of fixed duration?

No

Q7a. ii) Please enter the data collection start date

02/01/1998

Q7a. iii) Please enter the data collection end date

Q7b. Could you provide some information about the data collection for this register?

Data collection ongoing|Data analysis ongoing

Q8. Funding of the register - How is the register funded?

Huntington's disease REGISTRY and ENROLL database - CHDI

Q8. Funding of the register - Is this funding expected to continue

yes

Q8. Funding of the register - If so, for how long (months)?

for many years

Q9. Could you provide information about data sweeping? - How many data sweeps have taken place?

Q9. Could you provide information about data sweeping? - When was the most recent data sweep?

Q9. Could you provide information about data sweeping? - When is the next data sweep?

Q9. Could you provide information about data sweeping? - How many more data sweeps are planned on current funding? e.g 0,1,2.....

Q9. Could you provide information about data sweeping? -How many more data sweeps are planned in total (with funding and with funding yet to be secured) e.g. 0,1,2...

Q10. Is the clinical (phenotypic) information that is held in the register from patients and other participants such as family members:

Routinely collected as medical records

Q11. Is there a limit on the number of studies that can be based on this set of patients?

Yes

If YES, please give details

depends on characterd study

Q12a. Please give information on the format and availability of data stored in a database (1)

Data summarised in database

% Available

Q12a. Please give information on the format and availability of data stored in a database (2)

No

% Available

Q12a. Please give information on the format and availability of data stored in a database (3)

No

% Available

Q12a. Please give information on the format and availability of data stored in a database (4)

Database on paper

% Available

Q12a. Please give information on the format and availability of data stored in a database (5)

% Available

Please specify language used

Czech language

Q12b. Please give information on how data is held as individual records (1)

Data is held as individual records

% Available

Q12b. Please give information on how data is held as individual records (2)

Data is web-based

% Available

Q12b. Please give information on how data is held as individual records (3)

Data held on computer based records

% Available

Q12b. Please give information on how data is held as individual records (4)

Data held on cards

% Available

Please specify language used

Czech language

Q13a. Is data available to other groups?

No

Q13b. If data is available to other groups what is the access policy/mechanisms for access?

Q14. What data sharing policy is specified as a condition of use?

Data made publicly available after a specified time point

Q15a. Are tissues/samples/DNA available to other groups?

No

Q15b. i) If yes, please describe below:

Q15b. ii) In what form are tissues/samples/DNA supplied?

Primary Samples: Stabilised samples (frozen or fixed)|Secondary samples: DNA|Secondary samples: RNA|Secondary samples: other (Skin Biopsy)

Q15b. iii) Is the access policy/mechanism for obtaining samples the same as that for obtaining data (Q13b above)?

Yes

Q16a. Is information on biological characteristics available to other groups?

No

Number of patients

% of total cohort

Q16b. If yes, is the access policy/mechanism for obtaining samples the same as that for obtaining data (Q13b above)?

No

Types:

Disease Registers

Member States:

Czech Republic

Diseases:

Huntington's disease, Neurodegenerative disease in general, Parkinson's disease & PD-related disorders

Years:

2016

Database Categories:

N/A

Database Tags:

N/A