

# Huntingtons disease

<https://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

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**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

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

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