ONWebDUALS: ONTology-based Web Database for Understanding Amyotrophic Lateral Sclerosis (Portugal)

https://neurodegenerationresearch.eu/survey/ontology-based-web-database-for-understanding-amyotrophic-lateral-sclerosis-portugal/

Title of study

ONTology-based Web Database for Understanding Amyotrophic Lateral Sclerosis (Portugal)

Acronym for cohort

ONWEBDUALS

Name of Principal Investigator - Title

Prof

Name of Principal Investigator - First name

Mamede

Name of Principal Investigator - Last name

de Carvalho

Address of institution -Institution

Instituto de Medicina Molecular

Address of institution - Street address

Avenida Professor Egas Moniz

Address of institution - City

Lisbon

Address of institution - Postcode

?1649-?028

Country

Portugal

Website

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

Motor neurone diseases

Q2a. In a single sentence what is the stated aim of the study? (Maximum 30 words)

To define specific factors that can possibly influence the onset and course of the disease taking into account the genotype-phenotype background.

Q2b. What distinguishes this case-control study from other studies?

Multi-center study that translates risk and prognostic factors from specific genotype-phenotype profiles into a disease-specific domain ontology applying sophisticated relevant bio-informatic methods for analysis

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

3

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

DOI: 10.1016/j.clinph.2016.02.011, DOI: 10.1007/s12576-016-0484-x, DOI: 10.1016/j.neucli.2016.08.001

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 case-control study 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?

Having Motor Neuron Disease, Revised El escorial criteria

Q4c. Study criteria: what are the exclusion criteria?

Having other neurological conditions

Q5a. What is the size of the cohort (i.e. how many participants have enrolled)?

1-1,000

Q5b. What is the expected number of control participants?

501-1,000

Q6a. Please describe what measures are used to characterise participants

Age, gender and ethnic background. Risk factors (trauma, surgery, exercise, smoking habits, lipids blood levels, stress level, place of birth and living, nutritional habits) medication taken, auto-immune diseases, diabetes and other medical disorders, cardiovascular events and other cardiovascular risk factors. Familial history (ALS, other neurodegenerative disorders, dementia, exercise, weight, cardiovascular events).

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

Yes. Age at disease-onset, Affected region at presentation, Symptoms at onset, Clinical signs, Functional measurement, Progression rate, Laboratory findings (thyroid function, CK, cholesterol, triglycerides, serum albumin, respiratory function tests including spirometry and maximal pressures). Survival (death or ? 23 hours ventilatory support).

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

No

If YES please specify
Q7. What is the study design?

Prospective cohort|Age|Sex

Q8. Are your cases matched by
Q9a. Does your study includes a specialised subset of control participants?

No

Q9b. If your study includes a specialised subset of control participants please describe
Q10a. Is data collection for this study

Data collection ongoing|Data analysis ongoing

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. Are data collected

Only through the study

Q12. Is there a system in place to enable re-contact with patients for future studies?

No

Q13a. Please give information on data stored in a database (1)

No

% Available
Q13a. Please give information on data stored in a database (2)

No

% Available
Q13a. Please give information on data stored in a database (3)

Database on spreadsheet (e.g. excel)

% Available

85

Q13a. Please give information on data stored in a database (4)

Database on paper

% Available

100

Q13a. Please give information on data stored in a database (5)

No

% Available
Please specify language used
% Available

100

Q13b. Please give information on how data is held as individual records

No

% Available
Q14a. Are 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 policy exists

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

No

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?

No

Types:
Case Control Studies, JPND Projects

Member States:
JPND, Portugal

Diseases:
Motor neurone diseases

Years:
2016

Database Categories:
N/A

Database Tags:
N/A