CGPP Database

Title of the register

CGPP Database

Name of Principal Investigator

Title   MD, PhD

First name Jorge

Last name Sequeiros

Address of institution where award is held

Institution   CGPP and UnIGENe

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Country
Portugal
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1. Conditions included, or expected to be included, in the disease register
Alzheimer's disease and other dementias
Motor neurone diseases
Parkinson's disease
Huntington's disease
Spinocerebellar ataxia (SCA)
Neurodegenerative disease in general
2a. Stated aim of the cohort

Molecular diagnosis and genetic counseling of neurodegenerative disorders in a national reference center and to foster research

2b. Features distinguishing this register from other disease registers

Excellent clinical information from expert neurologists, connected to pedigree data and long-lasting knowledge on Portuguese families with neurodegenerative disorders. (2) Being a national reference diagnostic centre for these disorders, situated in a research institute.

3a. i) Number of publications that involve use of register to date

3a. ii) Up to three examples of studies to date (PI, Institution, Title of Study)

1. Name of PI

PI: Isabel Alonso, IBMC, Mutational spectrum and improvement in laboratory techniques applied to molecular diagnosis of neurological disorders

PI: Isabel Silveira, IBMC, Clinical Implications of genetic factors causing neurodegenerative diseases characterized by movement or cognitive dysfunction

PI: Paula Coutinho, IBMC, Survey of hereditary ataxias and spastic paraplegias in Portugal

4a. Study criteria: age range of participants

 Age in years from: Any age

4b. Study criteria: inclusion criteria

Suffering or having a family history of a neurodegenerative disorder with or without a molecular genetics diagnosis.

4c. Study criteria: exclusion criteria

None

5. Size of the register (i.e. number of patients enrolled)
More than 10,000 clinical cases

6a. Measures used to characterise participants

Personal, clinical and family data.

6b. Are there defined primary and secondary endpoints (e.g. defined health parameters)
No

7a. i) Is the register of fixed duration

1

7a. ii) Data collection start date

17-12-1997

7b. Stage of data collection/analysis for the register
Data collection ongoing

11. Limit on the number of studies that can be based on this set of patients

12a. Data stored in a database

Yes/No	% available
Y	100
N	
Y Access database	100
Y	100

12b. Data held as individual records

Yes/No	% available
Y	100
N	
Y	100
Y	

13a. Are data available to other groups

2

13b. Access policy/mechanisms for access if data are available to other groups
Apply to PI or co-ordinator at resource
14. Data sharing policy specified as a condition of use
No policy exists
15a. Are tissues/samples/DNA available to other groups

2

15b. i) Description of available tissues/samples/DNA
Living donors: DNA
Post-mortem donors: brain
15b. ii) Form available tissues/samples/DNA are supplied in
Secondary samples: plasma
Secondary samples: DNA
Secondary samples: cell lines derived from primary samples
15b iii) Is the access policy/mechanism for obtaining samples the same as that for obtaining data

1

16a. Is information on biological characteristics available to other group
16b. Is the access policy/mechanism for obtaining details of the characteristics the same as that for obtaining other data

2