REGISTRY - an observational study of the European Huntington's Disease Network (EHDN)

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Registry is the core study of the European Huntington*s disease Network (EHDN). The aim of the Registry study is to collect prospective data on the phenotypical characteristics of Huntington's disease (HD) mutation carriers regardless of...

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Movement disorders (incl parkinsonism)
Study type	Observational invasive

Summary

ID

NL-OMON38177

Source
ToetsingOnline

Brief title
Registry

Condition

- Movement disorders (incl parkinsonism)

Synonym
Huntington's disease

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W,High Q Foundation;New York;VS

Intervention

Keyword: European Network, Huntington's disease, Registry

Outcome measures

Primary outcome

There is no fixed end of study as 'Registry' will take place during visits at

the out-patient clinic of the department of Neurology or nursing home. The

study is imbedded in the network that provides a platform for communication and

exchange of views on HD and a basis for research.

Secondary outcome

n.v.t.

Study description

Background summary

Huntington*s disease (HD) is an autosomal dominant neurodegenerative disorder that results from an unstable expansion of the trinucleotide repeat CAG in the HD gene IT-15. HD has a prevalence of 5-10 per 100,000 in the general population. The clinical features of HD usually emerge in adulthood (mean age of 40 years) with a movement disorder, cognitive dysfunction and psychiatric symptoms. The course of HD is relentless, leading to functional disability and death over a period of 10-30 years. With genetic testing (following genetic counseling) it is possible to predict that a person will develop HD a long time before clinical symptoms and signs develop. To date, there is no treatment that has been shown to alter the progression of the disease. Benefical effects have been reported when applied in model systems of HD but the predictive value of these results for patients are unknown. As HD is a rare disease, extensive cooperation is essential to be able to include the number of participants required for conclusion well powered studies.

Study objective

Registry is the core study of the European Huntington*s disease Network (EHDN). The aim of the Registry study is to collect prospective data on the phenotypical characteristics of Huntington's disease (HD) mutation carriers

regardless of whether they display clinical symptoms and signs of HD and of individuals who are part of an HD family (irrespective of their mutation carrier status), in order
•to obtain natural history data on a wide spectrum of HD patients, HD mutation carriers and individuals who are part of an HD family
•to relate phenotypical characteristics with genetic factors and biomarkers
•to expedite identification and recruitment of participants for clinical trials
•to plan for future research studies (observational and interventional trials aimed at better symptom control or aimed at slowing or postponing the onset and progression of HD).

Study design

The registration will take place once a year within regular visits at the out-patient clinic of the department of Neurology or nursing home. Participant evaluation is carried out clinically using the Unified Huntington Disease Rating Scale (UHDRS 1999) and supplementary optional questionnaires. Furthermore participants are asked to donate biosamples (blood and urine) for studies to identify genetic modifiers of HD and to establish and validate biological markers tracking the progressive course of HD; in this context a family history is requested as well in order to understand the relationships of clinical data sets and biosamples from related donors. In addition, non-mutation carrying family members of participants are asked to consider donating biosamples to serve as controls. The biosamples are stored in a central repository, BioRep, Milan.
Participant data are entered electronically via internet-based technology after creating a unique pseudonym for each individual, based on unchanging information. The pseudonym is a nine-figure number created by a secure one-way algorithm. The identifying data are never stored electronically. An investigator is only allowed to see their own patients* data. The whole database is saved in the portal. Central Coordination is allowed to view all data of all centres for plausibility checks, quality control and monitoring.

Study burden and risks

Since Registry is an observational study, participants do not undergo specific risks by participating. Their burden is limited to a minimum as the evaluations occur within the ambulant care of the Neurology department on a regular basis.
Participants will receive no immediate benefit from participation in this study. The only potential benefit is a better understanding of HD and the possibility that the information obtained in this study lead to potential treatments and to plan future research studies of experimental drugs aimed at slowing disease progression or postponing the onset of HD.

Contacts

Public
Leids Universitair Medisch Centrum

Postbus 9600
2300 RC Leiden
NL
Scientific
Leids Universitair Medisch Centrum

Postbus 9600
2300 RC Leiden
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

.Individuals with clinical features of HD with a confirmatory family history of HD or with DNA testing results demonstrating the presence of the HD mutation (i.e. a CAG repeat expansion within the HD gene >35 on larger allele)
•Individuals without clinical features of HD with DNA testing result demonstrating presence of the HD mutation (i.e. CAG repeat expansion within the HD gene >35 on larger allele)
•First-degree relatives (i.e. parents, siblings, or children) of individuals with HD
•Second-degree relatives (i.e. grandparents and grandchildren) of participating individuals with HD
•Family members of participating individuals from category 1 or 2 who are know not to carry

the HD mutation (e.g., spouses)

Exclusion criteria

• Subjects who are unable to understand the study protocol or unable to give informed consent, and have no legal representative.
•Participants with choreic movement disorder other than HD.

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	17-07-2006
Enrollment:	800
Type:	Actual

Ethics review

Approved WMO Date:	07-07-2006
Application type:	First submission
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)
Approved WMO Date:	03-02-2012

Application type:	Amendment
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)
Approved WMO Date:	14-02-2012
Application type:	Amendment
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)
Approved WMO Date:	08-03-2012
Application type:	Amendment
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL12224.058.06