Genetic screening in Parkinson*s Disease in order to identify patients who can participate in clinical trials with new targeted therapies

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* Genotyping of the full GBA1 gene in people with Parkinson*s Disease, assessed as wildtype (GBA-) or containing a mutation (GBA+); the specific mutation will be recorded as well. * Assessing the presence of 7 known PD-causing mutations in the LRRK2…

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

Summary

ID

NL-OMON45734

Source
ToetsingOnline

Brief title
GBA1 and LRRK2 screening

Condition

- Movement disorders (incl parkinsonism)

Synonym
movement disorder, Parkinson's disease

Research involving
Human

Sponsors and support

Primary sponsor: Centre for Human Drug Research

Source(s) of monetary or material Support: Centre for Human Drug Research,Clinical Research Organisation,Lysosomal Therapeutics Inc

Intervention

Keyword: GBA1 gene, LRRK2 gene, Parkinson's Disease

Outcome measures

Primary outcome

Sequence of the GBA1 gene

Presence of 7 specific mutations in the LRRK2 gene

Secondary outcome

Database of genotyped PD patients, for future research on PD

Study description

Background summary

For the upcoming Phase 1B study (CHDR1710), investigating a possible first-in-class disease modifying drug, 28 Parkinson*s disease patients with a GBA1 mutation (PD-GBA+) are needed. This is a mutation that occurs in approximately 5-10% of PD patients. There is no way to phenotypically differentiate between PD patients with and without a GBA1 mutation. In order to identify these patients, a large-scale screening is needed.
Another gene which is known to be involved in the Parkinson*s disease process is the LRRK2 gene. This gene is also a possible target for novel treatments, currently being investigated. In order to perform proof-of-concept or efficacy studies of such treatments, a database of genotyped PD patients is important in order to be able to efficiently enroll a relevant subject population.

Study objective

* Genotyping of the full GBA1 gene in people with Parkinson*s Disease, assessed as wildtype (GBA-) or containing a mutation (GBA+); the specific mutation will be recorded as well.
* Assessing the presence of 7 known PD-causing mutations in the LRRK2 gene in people with Parkinson*s Disease, assessed as wildtype (LRRK2-) or containing a mutation (LRRK2+); the specific mutation will be recorded as well.
* Storage of DNA, obtained through saliva, for possible further assessments of

genes related to Parkinson*s Disease in the future.

Study design

The screening of the GBA1 and LRRK2 gene in people with Parkinson*s disease will take place by means of saliva sampling. This can be provided at home through a special saliva kit, which can be returned by mail. Patients will be approached by their treating neurologist by letter with a referral to CHDR if they wish to participate. After the patient has contacted CHDR, a letter with additional information and a saliva kit will be sent to their home. The received saliva will be genotyped at GenomeScan laboratory. Patients with a GBA1 mutation will be contacted to participate in the planned Phase 1B study (CHDR1710). This design will allow a large-scale, non-invasive screening of PD patients with a low patient burden.

Study burden and risks

This study concerns genetic screening of Parkinson*s patients to identify those with a GBA1 or LRRK2 mutation. 28 Parkinson*s patients are needed the planned Phase 1B study (CHDR1710) where a possible first-in-class disease modifying drug LTI-291 will be investigated. Similarly, selective LRRK2 kinase inhibitors are being developed and several are about to enter the clinical phase of drug development. To obtain these 28 Parkinson's patients with a GBA1 mutation and to identify patients with a LRRK2 mutation, Parkinson's patients will be approached to donate a saliva sample (an at home kit) to screen the GBA1 and LRRK2 genes. This is a low risk procedure and the burden is minimal.

Contacts

Public
Centre for Human Drug Research

Lysosomal Therapeutics Inc (LTI) 19 Blackstone St
Cambridge MA 02139
NL
Scientific
Centre for Human Drug Research

Lysosomal Therapeutics Inc (LTI) 19 Blackstone St
Cambridge MA 02139
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

1. Signed informed consent prior to any study-mandated procedure;
2. Diagnosis of Parkinson*s Disease, diagnosed by a neurologist;
3. Has the ability to communicate well with the Investigator in the Dutch language and willing to comply with the study restrictions.

Exclusion criteria

N/A

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Other

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	12-04-2017

Enrollment:	1000
Type:	Actual

Ethics review

Approved WMO Date:	04-04-2017
Application type:	First submission
Review commission:	BEBO: Stichting Beoordeling Ethiek Bio-Medisch Onderzoek (Assen)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

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

ID: 23101
Source: Nationaal Trial Register
Title:

In other registers

Register	ID
CCMO	NL61137.056.17