Research into the progression of Parkinson's disease in patients with a mutation in the GBA1 gene, based on medical history and genetic analyses

No registrations found.

Ethical review	Positive opinion
Status	Recruiting
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON28601

Source
Nationaal Trial Register

Brief title
CHDR1838 / LTI-OBS-001

Health condition

Parkinson’s disease and the GBA-PD subset of patients

Sponsors and support

Primary sponsor: Lysosomal Therapeutics Incorperated
Source(s) of monetary or material Support: Lysosomal Therapeutics Incorperated

Intervention

Outcome measures

Primary outcome

Based on multiple parameters, the following compound measurements are constructed:

- Parkinson's disease - Progression Rate Inventory (pD-pRI)
- GBA1 genotype and PD associated SNPs
- Parkinson's Risk Score (PRS)
- Rate of disease progression as rated by a clinical expert: Slow, Intermediate, Fast

The endpoints are the analyses of the associations between:
- genetic factors ând phenotypic factor
- genetic factors and the rate of disease progression as rated by a Movement Disorder Neurologist
- genetic factors and a data-driven algorithm based on phenotypic characteristics

Secondary outcome

N.A.

Study description

Background summary

This protocol is a follow-up on the previous study CHDR1707 (Toetsing online number: NL61137.056.17), titled
“Genetic screening in Parkinson’s Disease in order to identify patients who can participate in clinical trials with new
targeted therapies.” In this previous study, Parkinson’s patients throughout the Netherlands were genetically
screened for presence of mutations in the GBA1 gene and LRRK2 gene. In approximately 15% of all screened
patients, a mutation was found in the GBA1 gene. The current protocol aims to further characterize this subgroup
of patients with a GBA1 mutation, based on phenotype, as assessed by medical history, and on genotype, as
assessed by Parkinson’s disease related Single Nucleotide Polymorphism (SNP) analysis. The goal of this study
is to exploratively investigate whether clinical and genetic factors may contribute to the rate of clinical progression
in patients with Parkinson’s disease associated with a GBA1 mutation in the gene encoding GCase (GBA-PD)

Study objective

- To determine the relationship between phenotypic and genetic characteristics of GBA-PD patients. Phenotypic characteristics will be obtained by patient dossier review. Genetic characteristics of the GBA1 gene and a panel of SNP’s (gene markers), previously associated with risk or progression of idiopathic Parkinson’s disease, will be assessed.

- To determine the correlation between genetic characteristics (as described above) and the estimated disease progression rating (Fast, Intermediate, Slow) by a Movement Disorders Neurologist, based on retrospective phenotypic characteristics (as described above).
o Inter-rater correlation between Movement Disorders Neurologists will be determined in their rating of disease progression.
- To determine the correlation between genetic characteristics (as described above) and a data-driven algorithm based on phenotypic characteristics (as described above).

Study design

N.A. Observational study of hospital records and genotype records of PD patients (of CHDR1707 study)

Contacts

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Eligibility criteria

Inclusion criteria

• Signed informed consent prior to any study-mandated procedure.
• Minimum age of 18 years.
• Clinical diagnosis of Parkinson’s disease at least 6 months prior to screening, confirmed by a Movement Disorder’s Neurologist.
• Mutation(s) in the glucocerebrosidase GBA1 gene. Reference Appendix A for a list of GBA1 mutations.

Exclusion criteria

• N.A.

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	15-10-2018
Enrollment:	350
Type:	Anticipated

IPD sharing statement

Plan to share IPD: No

Ethics review

Positive opinion

Date:	07-05-2019
Application type:	First submission

Study registrations

Followed up by the following (possibly more current) registration

ID: 46323
Bron: ToetsingOnline
Titel:

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

No registrations found.

In other registers

Register	ID
NTR-new	NL7717
CCMO	NL67297.056.18
OMON	NL-OMON46323

Study results