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

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- 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),...

Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON28601

Bron
NTR

Verkorte titel
CHDR1838 / LTI-OBS-001

Aandoening

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

Ondersteuning

Primaire sponsor: Lysosomal Therapeutics Incorperated
Overige ondersteuning: Lysosomal Therapeutics Incorperated

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

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

Toelichting onderzoek

Achtergrond van het onderzoek

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)

Doel van het onderzoek

- 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).

Onderzoeksopzet

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

Contactpersonen

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

• 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.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

• N.A.

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Anders
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	15-10-2018
Aantal proefpersonen:	350
Type:	Verwachte startdatum

Voornemen beschikbaar stellen Individuele Patiënten Data (IPD)

Wordt de data na het onderzoek gedeeld: Nee

Ethische beoordeling

Positief advies

Datum:	07-05-2019
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

ID: 46323
Bron: ToetsingOnline
Titel:

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

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

Resultaten