Molecular pathway analysis of Amyotrophic Lateral Sclerosis by genome-wide expression profiling of human blood and skin fibroblasts.

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Neuromuscular disorders
Study type	Observational invasive

Summary

ID

NL-OMON31437

Source
ToetsingOnline

Brief title
Biomarkers in ALS

Condition

- Neuromuscular disorders

Synonym
Amyotrophic Lateral Sclerosis (ALS), motor neuron disease

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht

Source(s) of monetary or material Support: Vidi toekenning Prof LH van den Berg,Prinses Beatrix Fonds (aanvraag loopt)

Intervention

Keyword: ALS, biomarkers, blood, fibroblasts

Outcome measures

Primary outcome

Expression profiles will be observed specific for ALS and/or disease progression. Findings can be translated into diagnostic tools. The genetic study will identify DNA variants related to ALS specific expression profiles leading to new insights into the genetic basis of disease susceptibility and progression.

Secondary outcome

nvt

Study description

Background summary

ALS is an adult-onset, disabling and fatal disease characterized by progressive degeneration of motor neurons in brain and spinal cord. No cure is available for ALS and the median survival is 3 years. There is no definitive diagnostic test for ALS available and other conditions can resemble ALS clinically. The diagnostic delay is more than one year and misdiagnosis is common. In approximately 10% of patients, ALS occurs in families. Familial ALS is clinically and pathological indistinguishable from sporadic ALS. The pathogenesis of ALS is unknown but there is convincing evidence that several molecular pathways play a role.

Study objective

The two objectives of our proposal are: (i) to identify specific gene expression profiles in blood and fibroblasts from patients with ALS or ALS-mimic in order to improve diagnosis and measure disease progression; (ii)

the identification of genetic determinants linked to disease-associated expression profiles.

Study design

First, gene expression profiling is performed in ALS patients and ALS-mimic patients leading to specific patterns (functional biomarkers). The ALS-mimic patients will subsequently determine the sensitivity and specificity for ALS. Molecular pathways involved in riluzole treatment will be identified by comparing gene expression before and after treatment. Any findings will be confirmed in biopsy material. Lastly, when specific profiles are observed we will genotype SNPs in differentially expressed genes to identify the genetic basis for the functional biomarkers.

Study burden and risks

Risks in participating in this study is associated with the risk performing a venapunction. This is a rather save and frequent performed exercise with uncommon complications. The procedure is not time consuming.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

- Age > 18 years
- Diagnosis of ALS or other ALS-mimic disorder
- No cognitive impairement

Exclusion criteria

- Diagnosis unknown

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):	01-09-2006
Enrollment:	2000
Type:	Actual

Ethics review

Approved WMO Date:	25-07-2006
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)
Approved WMO Date:	11-03-2008
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)

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	NL11662.041.06