Patient Registry of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS)

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To generate a large, international Friedreich*s ataxia patient database (including healthy controls), alongside an integrated clinical and natural history database, which is linked to a biological samples and imaging repository, hereby creating a...

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

Summary

ID

NL-OMON57202

Source
ToetsingOnline

Brief title
EFACTS

Condition

- Movement disorders (incl parkinsonism)

Synonym
Friedreich Ataxia

Research involving
Human

Sponsors and support

Primary sponsor: RWTH Aachen University

Source(s) of monetary or material Support: Friedreich Ataxia Research Alliance

Intervention

Keyword: biomarkers, longitudinal cohort

Outcome measures

Primary outcome

Build and study a large cohort of Friedreich ataxia patients, to develop and discover new clinical, motor, imaging and biochemical markers and to infer a model of disease evolution in Friedreich Ataxia.

Secondary outcome

x

Study description

Background summary

Friedreich*s ataxia (FA) is the most frequent early-onset autosomal recessive hereditary ataxia. The GAA-repeat expansion leads to a progressive neurodegenerative disease, characterized by progressive (sensory) ataxia, spasticity and sensomotor polyneuropathy. Hypertrophic cardiomyopathy and diabetes mellitus often occur in association with the disease. Patients gradually lose coordination and are frequently wheelchair bound as adolescents. There is no disease modifying therapy and average life expectancy ranges from 40 to 50 years, with many patients dying prematurely of cardiomyopathy. Friedreich*s ataxia is a rare disease, leaving single hospitals to have limited options for clinical or genetic research. By coordinating their efforts, EFACTS gathers vital information about the progression of Friedreich*s ataxia which can be delivered to patients to give them a better knowledge of their prognosis and the development of their condition.

Study objective

To generate a large, international Friedreich*s ataxia patient database (including healthy controls), alongside an integrated clinical and natural history database, which is linked to a biological samples and imaging repository, hereby creating a large trial-ready cohort for possible future

intervention studies.

Study design

Longitudinal cohort study with annual visits.

Study burden and risks

The burden for participants consists of annual visits, including a neurological examination, several questionnaires taking a blood sample. Optional are undergoing a MRI or cardiac evaluation. All measurements are without significant side effects (*negligible risk*).
These type of natural history cohorts are important for increasing understanding of the disease and provide viable information for future trials with possible disease-modifying therapies.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adults (18-64 years)

Inclusion criteria

Genetic conformation of Friedreich Ataxia

Exclusion criteria

no genetically confirmed diagnosis of Friedreich*s ataxia (except control participants)
acute or ongoing severe and unstable medical or other conditions that would interfere with the conduct and assessments of the study

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Other

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	11-01-2024
Enrollment:	25
Type:	Anticipated

Ethics review

Approved WMO

Date:	24-12-2024
Application type:	First submission

Review commission: CMO regio Arnhem-Nijmegen (Nijmegen)

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
ClinicalTrials.gov	NCT02069509
CCMO	NL86151.091.24