Molecular basis of Parry-Romberg syndrome II

Published: 18-05-2017
Last updated: 12-04-2024

1. Detection of the gene causing Parry-Romberg syndrome. 2. Study of molecular and cellular mechanisms leading to the various manifestations of Parry-Romberg syndrome.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational invasive

Summary

ID

NL-OMON45270

Source
ToetsingOnline

Brief title
PRS II

Condition

- Congenital and hereditary disorders NEC

Synonym
progressive hemifacial atrophy; coupe de sabre; atrophia of the face

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: etiology, Parry-Romberg syndrome, progressive hemifacial atrophy

Outcome measures

Primary outcome

Detection of the gene causing Parry-Romberg syndrome using results of studies

of RNA in affected and non-affected skin.

Secondary outcome

Understanding of the molecular and cellular mechanisms leading to the various

manifestations of Parry-Romberg syndrome.

Study description

Background summary

Parry-Romberg syndrome (Romberg syndrome; progressive hemifacial atrophy) consists of slowly progressive atrophy of bone and the soft tissues of essentially half the face accompanied most frequently by contralateral Jacksonian epilepsy, trigeminal neuralgia, and ipsilateral changes in the eyes and hair. The consequences for the disorder for individual patients are significant due to the often enormous disfigurement it causes. There is at present no therapy.
We have limited proof that Parry-Romberg syndrome is a laminopathy in a mosaic state. In an earlier study in fibroblasts derived from affected facial region in 3 patients we were able to detect abnormal nuclear morphology (the hall mark of a laminopathy) which was absent in nuclei from leukocytes, indicating mosaicism, in three patients. Further molecular studies using next generation techniques searching for differences between variants in DNA from the affected area and leukocytes has been performed but the causative gene could not be detected. The best next way to detect the cause will be to perform studies on an RNA level, performed in fibroblasts from affected tissue and fibroblasts from non-affected tissue, and also compare in lymphocytes.

Study objective

1. Detection of the gene causing Parry-Romberg syndrome.
2. Study of molecular and cellular mechanisms leading to the various manifestations of Parry-Romberg syndrome.

Study design

observational study with invasive measurements

Study burden and risks

The risk of blood sampling is limited. The burden of a skin biopsy in a healthy adult with normal cognition is in general minimal if taken from a limb (unaffected tissue). The burden is higher if the biopsy is taken from affected tissue, i.e. the face. The site of the biopsy will be chosen together with the participant and likely will be in the anterior hairline as this is the least visible. The site will be locally anaesthetized. The biopsy (2 or 3 mm in diameter) does not need any stitching and will heal spontaneously. We realize this is a burden to the patients, but without biopsying this part of the skin it will not be possible to prove the hypothesis and find the causative gene. We have considered waiting for surgical procedures for patient care purposes in some patients, but it is at present uncertain whether these will be performed in the near future. Proving Parry-Romberg syndrome to be laminopathy and finding the gene would lead to a trial in order to decrease or stop progression. There is no benefit of participating in this study to the participants. There is a group benefit as the study should provide essential information needed for a future intervention study.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105AZ
NL
Scientific
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105AZ
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

diagnosis Parry-Romberg syndrome
Able to read and understand the information
16 years or older

Exclusion criteria

none

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	22-08-2017
Enrollment:	3
Type:	Actual

Ethics review

Approved WMO Date:	18-05-2017
Application type:	First submission
Review commission:	METC Amsterdam UMC

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	NL61303.018.17