The role of genetic factors in eosinophilic esophagitis

Published: 31-07-2013
Last updated: 22-04-2024

Primary:* To investigate associations between genomic variation and occurrence of eosinophilic esophagitis. Secondary:* To systematically collect DNA samples and biopsy specimens for future genetic analysis.* We aim to collect clinical data on all...

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Gastrointestinal inflammatory conditions
Study type	Observational invasive

Summary

ID

NL-OMON38986

Source
ToetsingOnline

Brief title
EoE Genetics

Condition

- Gastrointestinal inflammatory conditions
- Allergic conditions

Synonym
allergic esophagitis, Eosinophilic esophagitis

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Eosinophilic esophagitis, Genetics

Outcome measures

Primary outcome

Genetic variants associated with EoE.

Secondary outcome

-

Study description

Background summary

The pathophysiology of EoE remains partly unexplained. So far, only a few small genetic studies have been performed aiming to identify loci associated with EoE.9-13 These studies have shown associations with genes involved in the recruitment of eosinophils (CCL26), remodeling/fibrosis (TGFB1), atopy (TSLP) and epithelial barrier integrity (FLG) in small percentages of EoE patients.10-13 No replication studies have yet been performed to confirm these associations. No pathological mutations have been described in EoE patients. Identification of genes associated with EoE would extent our knowledge about the pathophysiology of EoE and could possibly contribute to the development of clinically used molecular analyses to predict EoE diagnosis and/or prognosis.

Study objective

Primary:
* To investigate associations between genomic variation and occurrence of eosinophilic esophagitis.

Secondary:
* To systematically collect DNA samples and biopsy specimens for future genetic analysis.
* We aim to collect clinical data on all EoE patients participating in this study.

Study design

Prospective, observational study.

Study burden and risks

The risks associated with participation are negligible. Blood withdrawal is routinely performed, with rarely any serious complications. Possible complications include bleeding/hematoma, thrombophlebitis and infection. Esophageal biopsies are taken regularly during upper endoscopy. A rare but potentially severe risk of a biopsy is a perforation. In most cases this perforation can be treated expectatively or endoscopically. In a minority of cases, surgery has to be performed to close the perforation. Another risk of an esophageal biopsy is bleeding, which can be treated endoscopically.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1100 DD
NL
Scientific
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1100 DD
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

* Previous clinical diagnosis of EoE confirmed by histopathology
* Written informed consent
* Age *18 years

Exclusion criteria

* Severe anemia (hindering safe vena puncture)

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):	31-07-2013
Enrollment:	300
Type:	Actual

Ethics review

Approved WMO

Date:	31-07-2013
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	NL45292.018.13