

# GWAS and HLA subtyping in autoimmune encephalitis

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Primary objectives: 1. To determine the gene frequencies of HLA-A, B, C, DR and DQ in patients with autoimmune encephalitis and compare this to the general Dutch population. 2. To determine genetic risk factors for the development of autoimmune...

<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruiting
<b>Health condition type</b>	Autoimmune disorders
<b>Study type</b>	Observational invasive

## Summary

### ID

NL-OMON49804

### Source

ToetsingOnline

### Brief title

GWAS and HLA in autoimmune encephalitis

### Condition

- Autoimmune disorders
- Central nervous system infections and inflammations

### Synonym

autoimmune encephalitis, Brain inflammation

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Erasmus MC, Universitair Medisch Centrum Rotterdam

**Source(s) of monetary or material Support:** Unrestricted vergoedingen voor onderwijs van Maarten Titulaer aan vakgenoten; het gaat om minimale logistieke kosten in het Erasmus MC. Het grootste gedeelte van de kosten wordt gedragen door de HLA afdeling van het LUMC

en door het GENERATE consortium in Duitsland.

## Intervention

**Keyword:** Autoimmune encephalitis, GWAS, HLA

## Outcome measures

### Primary outcome

1. Identification of a HLA allele or allelic combination associated in patients with autoimmune encephalitis.
2. Determination of genetic risk factors for the development of autoimmune encephalitis using Genome-Wide Association Studies (GWAS).

### Secondary outcome

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## Study description

### Background summary

Autoantibodies can cause severe encephalitis. A known genetic marker for autoimmune diseases is HLA. Specific HLA types are found for certain types of autoimmune encephalitis. Other genetic factors contributing to autoimmune encephalitis are unknown. Due to the scarcity of the disease and the relatively recent discovery of autoimmune encephalitis, the number of patients are small. International collaboration will lead to cohorts of sufficient size, including validation cohorts. This allows us to investigate which pathophysiological mechanisms and immunological pathways are relevant for autoimmune encephalitis.

### Study objective

Primary objectives:

1. To determine the gene frequencies of HLA-A, B, C, DR and DQ in patients with autoimmune encephalitis and compare this to the general Dutch population.
2. To determine genetic risk factors for the development of autoimmune encephalitis using GWAS.

Secondary objectives:

1. To determine the gene frequencies of HLA-A, B, C, DR and DQ in patients with autoimmune encephalitis and compare this to the general population within an international consortium.
2. To determine specific patient characteristics or disease characteristics associated with a common HLA subtype within autoimmune encephalitis, per antibody.

## **Study design**

Case-control study.

## **Study burden and risks**

One venepuncture will be performed, preferably in extension of a planned venepuncture. The venepuncture has negligible risks and burden. There is no direct benefit for the patient.

## **Contacts**

### **Public**

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### **Scientific**

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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 of 18 years and older.
- Diagnosis of autoimmune encephalitis (neuronal antibodies proven in serum or CSF).

## Exclusion criteria

- Patient and/or legal representative is withholding informed consent.
- Patient objects after initial informed consent.

## 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:	Recruiting
Start date (anticipated):	19-11-2020
Enrollment:	300
Type:	Actual

## Ethics review

Approved WMO  
Date: 10-11-2020  
Application type: First submission  
Review commission: METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

## 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	NL74462.078.20