

# Identifying genetic determinants of outcome in multiple sclerosis

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<b>Registration date</b> 04/05/2023	<b>Overall study status</b> Ongoing	<input type="checkbox"/> Statistical analysis plan <input type="checkbox"/> Results
<b>Last Edited</b> 20/03/2026	<b>Condition category</b> Nervous System Diseases	<input type="checkbox"/> Individual participant data <input checked="" type="checkbox"/> Record updated in last year

## Plain English summary of protocol

### Background and study aims

Multiple sclerosis (MS) is what scientists call an “autoimmune” disease. In such diseases the immune system is faulty and mistakenly attacks part of the body. In MS, this faulty attack is directed against the central nervous system (CNS); that is against the brain and the spinal cord. The damage caused by MS results in increasingly severe neurological disability that is known as disease “progression”. The rate at which this irreversible damage develops varies greatly between patients. Some become markedly disabled very quickly, while others develop little or no disability even after years. This study will examine the DNA code from people with MS in order to identify genetic factors that influence how quickly progression develops in the disease. That is, to identify changes in the DNA sequence that influence the outcome of the disease.

### Who can participate?

People with MS who have had their disability recorded at least once using a clinical assessment called the Extended Disability Status Scale (EDSS). The EDSS is frequently measured in MS research studies and is now also often measured as part of routine clinical care.

### What does the study involve?

The study involves the genetic analysis of DNA donated by participants. The DNA will be extracted either from venous blood or saliva collected from participants. The analysis of the DNA will be undertaken in the laboratory. The genetic data emerging from the laboratory analysis will be correlated with the demographic and clinical details collected about the participants.

### What are the possible benefits and risks of participating?

This is an observational study in which the only intervention is the collection of venous blood. This collection will be undertaken by professional appropriately trained staff. The collection of venous blood is a routine procedure with no meaningful risk. The study will also involve the sharing of data about participants between the researchers involved in the study. All data sharing in the study will be undertaken in accordance with best practice and legal requirements such as GDPR. There are no benefits to participants from taking part in the study other than the knowledge that they are helping to bring effective treatments closer for future generations of patients.

Where is the study run from?  
The University of Cambridge (UK)

When is the study starting and how long is it expected to run for?  
September 2026 to September 2030

Who is funding the study?  
Funding for the study is not yet confirmed

Who is the main contact?  
Prof. Stephen Sawcer, sjs1016@cam.ac.uk

## Contact information

### Type(s)

Principal investigator

### Contact name

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**Additional identifiers**

**Clinical Trials Information System (CTIS)**

Nil known

**Integrated Research Application System (IRAS)**

324856

**Protocol serial number**

A096498, IRAS 324856

**Study information**

**Scientific Title**

Progression in Multiple Sclerosis

**Acronym**

PIMS

**Study objectives**

The primary aim of this study is to identify genetic variants influencing the clinical course of multiple sclerosis

**Ethics approval required**

Old ethics approval format

**Ethics approval(s)**

Not provided at time of registration

**Study design**

Genome wide association screen

**Primary study design**

Observational

**Study type(s)**

Other

**Health condition(s) or problem(s) studied**

Multiple sclerosis

**Interventions**

The researchers will undertake array-based genome-wide genotyping in DNA extracted from patients' venous blood or saliva

**Intervention Type**

Genetic

**Primary outcome(s)**

Disability measured using the Extended Disability Status Scale (EDSS) and age-corrected to generate the Age-Related Multiple Sclerosis Severity (ARMSS) Score. All available measures of the ARMSS will be considered in the analysis as observed at any timepoint in the disease course.

**Key secondary outcome(s)**

Time to event measures including time to EDSS 6.0 and time to confirmed disability worsening. These variables will be assessed in the sub-group of patients with EDSS measured at multiple timepoints.

**Completion date**

30/09/2030

**Eligibility****Key inclusion criteria**

1. A locally confirmed diagnosis of multiple sclerosis
2. At least one measure of Extended Disability Severity Score (EDSS)
3. Able to give valid informed consent

**Participant type(s)**

Patient

**Healthy volunteers allowed**

No

**Age group**

Adult

**Sex**

All

**Total final enrolment**

0

**Key exclusion criteria**

1. Patients with Radiologically or Clinically Isolated Syndrome (RIS or CIS)
2. Comorbidities causing significant disability likely to have confounded the reliability of the EDSS
3. Inclusion in the discovery cohort of the recently completed first progression GWAS

**Date of first enrolment**

01/03/2026

**Date of final enrolment**

30/09/2028

## Locations

**Countries of recruitment**

United Kingdom

England

**Study participating centre****Addenbrookes**

Addenbrookes Hospital

Hills Road

Cambridge

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## Sponsor information

**Organisation**

Cambridge University Hospitals NHS Foundation Trust

**ROR**

<https://ror.org/04v54gj93>

## Funder(s)

**Funder type**

Other

**Funder Name**

Not yet confirmed

# Results and Publications

## **Individual participant data (IPD) sharing plan**

It is the researchers' intention to store the data generated in this study (demographic, clinical and genetic) in a genomics data repository such as the European Genome-Phenome Archive (EGA, <https://ega-archive.org/>). Anonymised data will be available when the results of the study are published by application to the Data Access Committee (DAC) via the repository. All participants will consent to data sharing with bona fide third-party researchers undertaking appropriate biomedical research.

## **IPD sharing plan summary**

Stored in publicly available repository