

# Investigating genes in patients with polymyositis and dermatomyositis

|                                        |                                                       |                                                                                                                         |
|----------------------------------------|-------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------|
| <b>Submission date</b><br>10/08/2010   | <b>Recruitment status</b><br>No longer recruiting     | <input type="checkbox"/> Prospectively registered<br><input type="checkbox"/> Protocol                                  |
| <b>Registration date</b><br>21/09/2010 | <b>Overall study status</b><br>Ongoing                | <input type="checkbox"/> Statistical analysis plan<br><input type="checkbox"/> Results                                  |
| <b>Last Edited</b><br>23/01/2026       | <b>Condition category</b><br>Musculoskeletal Diseases | <input type="checkbox"/> Individual participant data<br><input checked="" type="checkbox"/> Record updated in last year |

## Plain English summary of protocol

### Background and study aims

Polymyositis (PM), dermatomyositis (DM) and inclusion body myositis (IBM) belong to a group of inflammatory muscle disorders, of unknown cause, that are characterised by skeletal muscle inflammation and progressive muscular weakness, which can be debilitating and chronic in nature (occasionally fatal). The current treatment options for these conditions are steroids and various other immunosuppressive drugs. However, these are usually only partially effective at reducing symptoms, and their toxic side effects also limit their usefulness. In order to develop more specific and therefore more effective treatments for myositis, it is important to understand the exact mechanisms that cause the disease in the first instance. The aim of this study is to identify genes that are associated with the development and clinical characteristics of inflammatory muscle diseases. By understanding the genetic cause of the diseases, it should be possible to design specific drugs for treating the conditions in the future.

### Who can participate?

Patients aged 18 or over with PM, DM or IBM.

### What does the study involve?

Participants are asked to give 20 ml of blood. These blood samples, along with the patient's clinical details, are then be sent to the Centre for Integrated Genomic Medical Research (CIGMR) at The University of Manchester, where the genetic analysis takes place.

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

Not provided at time of registration

### Where is the study run from?

Manchester University and Salford Royal NHS Foundation Trust (UK)

### When is the study starting and how long is it expected to run for?

January 2000 to December 2030 (updated 04/02/2021, previously: January 2020)

### Who is funding the study?

Manchester University and Salford Royal NHS Foundation Trust (UK)

Who is the main contact?

Mr Paul New

paul.new2@nca.nhs.uk

## Contact information

### Type(s)

Scientific

### Contact name

Mr Paul New

### Contact details

Northern Care Alliance NHS Foundation Trust

Salford Royal Hospital

Clinical Science Building Room B213

Manchester

United Kingdom

M6 8HD

+44 (0)161 2064295

paul.new2@nca.nhs.uk

### Type(s)

Scientific

### Contact name

Prof Hector Chinoy

### Contact details

Centre for Musculoskeletal Research

Division of Musculoskeletal & Dermatological Sciences

The University of Manchester School of Biological Sciences

2.810 Stopford Building

Oxford Road

Manchester

United Kingdom

M13 9PT

+44 (0)161 275 1676

hector.chinoy@manchester.ac.uk

## Additional identifiers

ClinicalTrials.gov (NCT)

NCT01171573

Protocol serial number

7996

## Study information

**Scientific Title**

Identification of disease susceptibility genes associated with development and clinical characteristics of primary inflammatory muscle diseases, polymyositis (PM), dermatomyositis (DM) and inclusion body myositis (IBM)

**Acronym**

UKMYONET

**Study objectives**

Polymyositis (PM), dermatomyositis (DM) and inclusion body myositis (IBM) belong to a group of inflammatory muscle disorders, of unknown cause, that are characterised by skeletal muscle inflammation and progressive muscular weakness, which can be debilitating and chronic in nature (occasionally fatal). The current treatment options for these conditions are steroids and various other immunosuppressive drugs. However, these are usually only partially effective at reducing symptoms, and their toxic side effects also limit their usefulness.

In order to develop more specific treatments for myositis in the future (and therefore more effective), it is important to understand the exact mechanisms that cause the disease in the first instance. In other similar inflammatory diseases such as rheumatoid arthritis (RA) and systemic lupus erythematosus (SLE), it is known that changes to the Human Leukocyte Antigen (HLA), as well as certain inflammatory cytokines, are involved in both the development and expression of the disease.

As many of the inflammatory mechanisms that cause damage in PM, DM and IBM are similar to those in RA and SLE, it seems likely that similar genetic factors will also be involved in the development and expression of PM, DM and IBM. In order to understand the genetic aspects /causes of myositis, and ultimately develop more effective treatment therapies in the future, patients with PM, DM or IBM, will be asked to give 20 ml of blood. These blood samples, along with the patient's clinical details, will then be sent to the Centre for Integrated Genomic Medical Research (CIGMR), at The University of Manchester, where all of the genetic analyses will take place. By understanding the genetic cause of the disease, it should be possible to design specific drugs for treating the condition in the future.

**Ethics approval required**

Old ethics approval format

**Ethics approval(s)**

Approved 04/05/1999, NRES committee North West Haydock (HRA NRES centre Manchester 3rd Floor Barlow House 4 Minshull Street Manchester M1 3DZ, Manchester, M1 3DZ, United Kingdom; +44 (0)161 6257827; nrescommittee.northwest-haydock@nhs.net), ref: 98/8/086

**Study design**

Interventional clinical laboratory study

**Primary study design**

Interventional

**Study type(s)**

Screening

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

Polymyositis, dermatomyositis and inclusion body myositis

### **Interventions**

Venepuncture, 20 ml of blood collected in EDTA tubes and sent off for genetic and antibody analysis. Genetic analysis is taking place on these samples as an ongoing process and will continue to do so until sufficient numbers have been collected for a conformation Genome Wide Association Scan (GWAS), possibly 2020.

### **Intervention Type**

Genetic

### **Primary outcome(s)**

To identify any disease susceptibility genes associated with development and clinical characteristics, measured once conformation GWAS performed (possibly 2020)

### **Key secondary outcome(s)**

No secondary outcome measures

### **Completion date**

31/12/2030

## **Eligibility**

### **Key inclusion criteria**

1. Skin lesions of (DM):
  - 1.1. Heliotrope rash (violaceous rash and on upper eyelids)
  - 1.2. Gottron's sign (violaceous keratotic macules on extensor aspect of finger joints)
  - 1.3. Violaceous slightly raised rash over elbows/knees
2. Proximal muscle weakness (PM, DM and IBM)
3. Elevated plasma muscle enzymes
4. Myalgia, at rest or with contraction
5. Myopathic changes on electromyogram (EMG)
6. Anti Jo1 Ab
7. Nondestructive arthritis
8. Systemic inflammatory signs (fever, erythrocyte sedimentation rate [ESR] greater than 20, elevated C-reactive protein [CRP], weight loss)
9. Myositic changes on muscle biopsy
10. Additional patients with proven Inclusion Body Myositis (IBM)
11. Male and female, lower age limit of 18 years

### **Participant type(s)**

Patient

### **Healthy volunteers allowed**

No

### **Age group**

Mixed

### **Lower age limit**

18 years

**Upper age limit**

100 years

**Sex**

All

**Total final enrolment**

1947

**Key exclusion criteria**

1. Below the age of 18 years
2. Myositis secondary to:
  - 2.1. Alcohol or drug abuse
  - 2.2. Non-abusive drug ingestion (e.g with statins, fibrates etc), or
  - 2.3. A recent viral illnesses
3. Unable to give consent due to diminished mental capacity or inability to speak sufficient English
4. Unwilling to give blood samples

**Date of first enrolment**

06/01/2000

**Date of final enrolment**

26/08/2020

## **Locations**

**Countries of recruitment**

United Kingdom

England

**Study participating centre**

**Rheumatic Diseases Centre**

Northern Care Alliance NHS Foundation Trust

Salford Royal Hospital

Stott Lane

Salford

England

M6 8HD

## **Sponsor information**

**Organisation**

Manchester University (UK)

**ROR**

<https://ror.org/027m9bs27>

**Funder(s)****Funder type**

University/education

**Funder Name**

University of Manchester

**Alternative Name(s)**

University of Manchester in United Kingdom, University of Manchester UK, The University of Manchester, UoM

**Funding Body Type**

Government organisation

**Funding Body Subtype**

Universities (academic only)

**Location**

United Kingdom

**Funder Name**

Salford Royal NHS Foundation Trust

**Alternative Name(s)**

SRFT

**Funding Body Type**

Government organisation

**Funding Body Subtype**

Local government

**Location**

United Kingdom

**Results and Publications**

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

### **IPD sharing plan summary**

Not provided at time of registration