# Relation between enzyme replacement therapy and progression of brain lesions in Fabry disease

Submission date 27/01/2015	<b>Recruitment status</b> No longer recruiting	[X] Prospectively registered [_] Protocol
Registration date 10/02/2015	<b>Overall study status</b> Completed	<ul> <li>[] Statistical analysis plan</li> <li>[X] Results</li> </ul>
Last Edited 21/06/2019	<b>Condition category</b> Nutritional, Metabolic, Endocrine	Individual participant data

### Plain English summary of protocol

Background and study aims

Fabry disease is an inherited disorder that results from the build-up of a particular type of fat (globotriaosylceramide) in the body's cells. Beginning in childhood, this build-up causes signs and symptoms that affect many parts of the body including the brain. In the brain, small groups of dead cells clump together in the white matter and are known as white-matter lesions. These lesions may lead to a high risk of early dementia, stroke or death. Little is known about the development of white-matter lesions and how they relate to other factors (e.g., age, sex or smoking) or how they are affected by treatment of Fabry disease with ERT. Salford Royal NHS Foundation Trust (UK) has a database of patients with Fabry disease in the northwest of England. The aim in this study is to look in detail at the relation between ERT and progression of brain lesions in Fabry disease so as to understand how the incidence and burden of the lesions change over time.

Who can participate?

Adults with Fabry disease who have had two MRI scans, 1 year apart

What does the study involve?

The size of the white-matter lesions will be measured over time and this information will be used alongside details of age, sex, risk factors for stroke/heart disease and treatment with ERT.

What are the possible benefits and risks of participating? There are no known benefits or risks to participants taking part in this study

Where is the study run from? Salford Royal NHS Foundation Trust (UK)

When is the study starting and how long is it expected to run for? February 2015 to December 2015

Who is funding the study? Investigator initiated and funded (UK) Who is the main contact? Mrs Sharon Hulme sharon.hulme@manchester.ac.uk

### **Contact information**

**Type(s)** Public

**Contact name** Mrs Sharon Hulme

**Contact details** 

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### Type(s)

Scientific

**Contact name** Dr Craig Smith

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## Additional identifiers

EudraCT/CTIS number Nil known

**IRAS number** 

**ClinicalTrials.gov number** NCT00196742

**Secondary identifying numbers** Nil known

## Study information

### Scientific Title

Magnetic resonance imaging of effect of enzyme replacement therapy on progression of cerebral white-matter lesions in Fabry disease: an observational study

### **Study objectives**

1. Does treatment of Fabry disease with enzyme replacement therapy (ERT) affect the build up and progression of white matter lesions?

2. White matter lesions can increase the risk of stroke and dementia and it is important to assess if treatment with ERT increases this risk

#### Ethics approval required

Old ethics approval format

### Ethics approval(s)

NRES Committee West Midlands - South Birmingham, 19/02/2015, ref: 15/WM/0064

**Study design** Observational study

**Primary study design** Observational

### Secondary study design

**Study setting(s)** Hospital

#### Study type(s) Other

Participant information sheet

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

Fabry disease

#### Interventions

Retrospective analysis of a database and serial magnetic resonance imaging (MRI) scans to assess:

- 1. Progression of white matter lesions in patients with Fabry disease
- 2. Relation between disease progression and treatment with ERT

Intervention Type Biological/Vaccine

#### **Phase** Not Applicable

Primary outcome measure

Prevalence and burden of white matter lesions over time: MRI scans at baseline and at 2 years will be compared for evidence of white-matter lesions using a visual severity rating scale

### Secondary outcome measures

Progression of white matter lesions: MRI scans at baseline and at 2 years will be compared for evidence of white-matter lesions using a visual severity rating scale

### Overall study start date

15/02/2015

### **Completion date**

31/12/2016

## Eligibility

### Key inclusion criteria

- 1. Confirmed diagnosis of Fabry disease
- 2. Age at least 18 years old
- 3. Being followed up at Salford Royal NHS Foundation Trust (UK)
- 4. Registered in Fabry disease registry
- 5. At least two serial MRI brain scans (1 year apart)

### Participant type(s)

Patient

**Age group** Adult

#### **Lower age limit** 18 Years

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Sex

Both

**Target number of participants** 100 data records

# **Total final enrolment** 149

### Key exclusion criteria

New patient
 No serial MRI scans
 MRI scans of insufficient quality for analysis

# Date of first enrolment 15/02/2015

Date of final enrolment 31/12/2015

### Locations

**Countries of recruitment** England

United Kingdom

**Study participating centre Salford Royal NHS Foundation Trust** Clinical Sciences Building Stott lane Salford United Kingdom M6 8HD

### Sponsor information

**Organisation** University of Manchester

**Sponsor details** Room 3.53 Simon Building Oxford Road Manchester England United Kingdom M13 9PL

**Sponsor type** University/education

ROR https://ror.org/027m9bs27

## Funder(s)

Funder type Not defined

**Funder Name** Investigator initiated and funded (UK)

### **Results and Publications**

#### **Publication and dissemination plan** Planned publication.

### Intention to publish date

31/12/2018

### Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are/will be available upon request from Dr Ana Jovanovic Ana.Jovanovic@srft.nhs.uk

### IPD sharing plan summary

Available on request

### Study outputs

Output type	<b>Details</b> results	Date created	Date added	Peer reviewed?	Patient-facing?
Results article		09/10/2018	14/06/2019	Yes	Νο
HRA research summary			28/06/2023	No	No