

Screening test for Fabry disease in patients receiving haemodialysis in England

Submission date 25/03/2021	Recruitment status No longer recruiting	<input checked="" type="checkbox"/> Prospectively registered <input checked="" type="checkbox"/> Protocol
Registration date 26/05/2021	Overall study status Completed	<input type="checkbox"/> Statistical analysis plan <input checked="" type="checkbox"/> Results
Last Edited 17/04/2025	Condition category Nutritional, Metabolic, Endocrine	<input type="checkbox"/> Individual participant data

Plain English summary of protocol

Background and study aims

Fabry disease is a rare (X-linked) genetic disorder that can affect many parts of the body including the kidneys and result in premature death. A recent large genetic screening study of newborns reported a higher incidence of Fabry disease in males, showing that Fabry disease is more frequent than previously expected. The diagnosis of Fabry disease is frequently delayed by around 14 years in males and 19 years in females. In the UK screening is not routinely done but is available in the forms of a dried blood spot test which measures enzyme activity. A female who has part of the Fabry genes (carriers) may have normal to low enzyme activity, so an additional blood test (plasma Lyso-GB3) and genetic testing are required. The aim of this study is to find out how many of the patients receiving haemodialysis in the West Midlands have Fabry disease, especially in those who are not known to have a cause of their kidney failure. This will also lead to further testing of relatives of identified patients (cascade screening), which in turn might help with an earlier diagnosis of Fabry disease and an earlier start of enzyme replacement treatment. Moreover, the identified patients in the haemodialysis population may benefit from enzyme replacement therapy for their heart disorders (i.e. coronary artery disease, cardiac failure and death).

Who can participate?

Patients receiving haemodialysis under the care of six kidney units in the Midlands in the UK

What does the study involve?

Participants' blood will be taken during routine dialysis session at a single timepoint. The blood will be dried on a card and sent for testing. In addition, participants will be asked to complete a questionnaire looking for any symptoms suggestive of Fabry disease. The study team will also collect participants' relevant demographics and clinical data. The study will run for 12 months. All participants who test negative for Fabry disease will be notified by letter. No further visits or follow-up will be required unless the participant's Fabry test is positive. The local kidney consultant involved in this study will inform the participants of the new diagnosis of Fabry disease and refer to the specialist Fabry disease clinic at the Queen Elizabeth Hospital in Birmingham. Any new cases of Fabry disease identified by the study will continue to receive specialist service at the Queen Elizabeth Hospital Birmingham as part of their routine NHS care. The specialist clinic will offer testing for family and relatives as the condition can be inherited.

Treatment in the form of enzyme replacement is available and may be offered if deemed suitable.

What are the possible benefits and risks of participating?

All new cases of Fabry disease diagnosed during the study will be informed by their local kidney doctors. All cases will also be referred promptly for further counselling and review at the specialist Fabry Disease Clinic at the Queen Elizabeth Hospital Birmingham in order to support participants with the new diagnosis. All participants will also be offered further screening for their relatives. As blood samples are sampled on dialysis, participants will not have extra pain or discomfort. As Fabry disease is a genetic disorder, if participants are diagnosed to have Fabry disease during the study, it will have significant implications for participants' themselves as well as their relatives. Such clinical implications are fully explained in the Patient Information Sheet.

Where is the study run from?

University Hospitals Birmingham NHS Trust (UK)

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

February 2020 to December 2023

Who is funding the study?

Sanofi-Genzyme (USA)

Who is the main contact?

Prof. Indranil Dasgupta

indranil.dasgupta@uhb.nhs.uk

Contact information

Type(s)

Scientific

Contact name

Prof Indranil Dasgupta

Contact details

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Additional identifiers

EudraCT/CTIS number

Nil known

IRAS number

ClinicalTrials.gov number

Nil known

Secondary identifying numbers

Nil known

Study information

Scientific Title

Screening for Fabry disease in the haemodialysis population

Acronym

SoFAH

Study objectives

To estimate the prevalence of Fabry disease in a large haemodialysis population in the UK.

Ethics approval required

Old ethics approval format

Ethics approval(s)

Approved 15/03/2022, East of England - Essex Research Ethics Committee (The Old Chapel, Royal Standard Place, Nottingham, NG1 6FS, UK; +44 2071048227; essex.rec@hra.nhs.uk), ref: 22/EE/0026

Study design

Cross-sectional epidemiological screening study

Primary study design

Observational

Secondary study design

Epidemiological study

Study setting(s)

Hospital

Study type(s)

Screening

Participant information sheet

Not available in web format, please use contact details to request a participant information sheet

Health condition(s) or problem(s) studied

Fabry disease

Interventions

All eligible patients are given a patient information sheet (PIS) and consent form by the research nurse during one of their haemodialysis sessions. All potential participants will be given the

opportunity to ask questions regarding the study after receiving the PIS, following which the consent will be taken at the next dialysis session 24 to 72 hours later. Written informed consent is obtained by a research nurse prior to the participant undergoing procedures that are specifically for the purposes of the study. In the case of participants who cannot read or write or require translators, the study will allow a witness to sign on a participant's behalf (in the case of problems with reading or writing), allow a witness to date the form on behalf of the participant and allow a hospital or personal interpreter.

Participants' blood will be taken during routine dialysis session at a single timepoint. The blood will be dried on a card and sent for testing to the Archimed Laboratories in Vienna, Austria. In addition, participants will be asked to complete a questionnaire looking for any symptoms suggestive of Fabry disease. The study team will also collect participants' relevant demographics and clinical data. The study will run for approximately 6 months. All participants who were tested negative for Fabry disease will be notified via letter. No further visits or follow-up will be required unless the participant's Fabry test is positive. The local kidney consultant involved in this study will inform the participants of the new diagnosis of Fabry disease and refer them to the specialist Fabry disease clinic at the Queen Elizabeth Hospital in Birmingham. Any new cases of Fabry disease identified by the study will continue to receive specialist service at Queen Elizabeth Hospital Birmingham as part of their routine NHS care. The specialist clinic will offer testing for family and relatives as the condition can be inherited. Treatment in the form of enzyme replacement is available and may be offered if deemed suitable.

Intervention Type

Other

Primary outcome measure

Prevalence of Fabry disease as defined by dried blood spot alfa-galactosidase A (GLA) enzyme activity, Lyso-Gb3 level and genetic mutation of GLA analysis at a single timepoint

Secondary outcome measures

1. Clinical characteristics of new cases of Fabry disease identified by the study, including age, gender, ethnicity, duration of dialysis, cardiovascular history, previous renal diagnosis and previous renal biopsy report, measured using review of medical records at the time of study recruitment
2. Fabry disease symptoms measured using a questionnaire designed by the SoFAH study which consists of six questions at the time of study recruitment (single timepoint)
3. Quality of life measured using EQ5D-5L at the time of study recruitment (single timepoint)

Overall study start date

01/02/2020

Completion date

31/12/2023

Eligibility

Key inclusion criteria

1. Patients receiving haemodialysis under the care of the six participating renal units
2. Aged 18 years and above
3. Capable of giving informed consent

Participant type(s)

Patient

Age group

Adult

Lower age limit

18 Years

Sex

Both

Target number of participants

2200

Key exclusion criteria

Does not meet inclusion criteria

Date of first enrolment

04/08/2022

Date of final enrolment

31/07/2023

Locations**Countries of recruitment**

England

United Kingdom

Study participating centre**Birmingham Heartlands Hospital**

University Hospitals Birmingham NHS Trust

Bordesley Green East

Birmingham

United Kingdom

B9 5SS

Study participating centre**Queen Elizabeth Hospital Birmingham**

University Hospitals Birmingham NHS Trust

Mindelsohn Way

Birmingham

United Kingdom

B15 2TH

Study participating centre
University Hospital Coventry and Warwickshire
Clifford Bridge Rd
Coventry
United Kingdom
CV2 2DX

Study participating centre
New Cross Hospital
Royal Wolverhampton NHS Trust
12, Corporate Services Centre
Wolverhampton
United Kingdom
WV10 0QP

Study participating centre
Royal Stoke University Hospital
Newcastle Rd
Stoke-on-Trent
United Kingdom
ST4 6QG

Study participating centre
Shrewsbury and Telford Hospital NHS Trust
Mytton Oak Rd
Shrewsbury
United Kingdom
SY3 8XQ

Study participating centre
Russells Hall Hospital
Dudley Group NHS Trust
Pensnett Rd
Dudley
United Kingdom
DY1 2HQ

Sponsor information

Organisation

University Hospitals Birmingham NHS Foundation Trust

Sponsor details

Queen Elizabeth Hospital Birmingham
Mindelsohn Way
Birmingham
England
United Kingdom
B15 2TH
+44 (0)1213713702
joanne.plumb@uhb.nhs.uk

Sponsor type

Hospital/treatment centre

Website

<http://www.uhb.nhs.uk/>

ROR

<https://ror.org/014ja3n03>

Funder(s)**Funder type**

Industry

Funder Name

Sanofi Genzyme

Alternative Name(s)

Genzyme Corporation, Genzyme Corp.

Funding Body Type

Private sector organisation

Funding Body Subtype

For-profit companies (industry)

Location

United States of America

Results and Publications

Publication and dissemination plan

The findings of this study will be reported at appropriate conferences and the aim is to publish them in a relevant open access journal. A summary report will be produced adherent to the funder's guidelines at the completion of the project. Participants in the study will also be informed of the study outcome via a letter.

Intention to publish date

31/12/2024

Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are/will be available upon request from Indranil Dasgupta (Indranil.dasgupta@uhb.nhs.uk). Anonymised patient-level data will be available after the publication of the primary paper of the study for up to 5 years. A data-sharing agreement with the sponsor trust will be required. Data will be shared with researchers interested in this area. As this is an industry-funded study, the data will also be shared with the funders. Consent will be obtained along with the consent for the study. Data will be pseudoanonymised.

IPD sharing plan summary

Available on request

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Protocol file	version V1.0	18/02/2021	26/05/2021	No	No
HRA research summary			28/06/2023	No	No
Preprint results	version 1.5	25/03/2025	17/04/2025	No	No
Protocol file	version 1.3	27/09/2022	17/04/2025	No	No