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Screening test for Fabry disease in patients receiving haemodialysis in England

Submission date
25/03/2021

Recruitment status
No longer recruiting

Registration date
26/05/2021

Overall study status
Completed

Last Edited
17/04/2025

Condition category
Nutritional, Metabolic, Endocrine

[X] Prospectively registered
[X] Protocol
[_] Statistical analysis plan
[X] Results
[_] 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

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Additional identifiers

Clinical Trials Information System (CTIS)
Nil known

ClinicalTrials.gov (NCT)
Nil known

Protocol serial number
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

Study type(s)
Screening

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(s)
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

Key secondary outcome(s))
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)

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

Healthy volunteers allowed
No

Age group
Adult

Lower age limit
18 years

Sex
All

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
United Kingdom

England

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

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

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?
HRA research summary			28/06/2023	No	No
Participant information sheet	Participant information sheet	11/11/2025	11/11/2025	No	Yes
Preprint results	version 1.5	25/03/2025	17/04/2025	No	No
Protocol file	version V1.0	18/02/2021	26/05/2021	No	No
Protocol file	version 1.3	27/09/2022	17/04/2025	No	No