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Finding out the genetic cause of Juvenile Myoclonic Epilepsy

Submission date
20/11/2017

Recruitment status
Recruiting

Registration date
18/12/2017

Overall study status
Ongoing

Last Edited
08/05/2025

Condition category
Nervous System Diseases

[ ] Prospectively registered
[ ] Protocol
[ ] Statistical analysis plan
[ ] Results
[ ] Individual participant data
[X] Record updated in last year

Plain English summary of protocol

Background and study aims
Epilepsy is a common neurological disorder affecting 1% of the population. There are over 30 types of epilepsy, some common, some rare. Most epilepsies arise in childhood and have a genetic cause. Approximately 40% of patients have the common forms of Genetic Generalised Epilepsy (GGE), and the commonest GGE is “Juvenile Myoclonic Epilepsy” or JME. There is overwhelming evidence that JME is caused by changes in genetic code. These changes are likely to be found in more than just one gene and there may be more than one type of change. In order to find these changes we need to study a large number of people with JME and compare their genetic code with people who do not have epilepsy. Finding the causes of JME will lead to a better understanding of its cause, new treatments, and tailoring of treatments according to a person's genetic make-up. The aim of this study is to find the genetic cause for JME by comparing the genetic code in JME patients with that in people who do not have epilepsy, using clues from their electroencephalograph or brainwave test that is used to help diagnose epilepsy.

Who can participate?
Patients aged 6 to 50 years old who have a diagnosis of Juvenile Myoclonic Epilepsy.

What does the study involve?
Participants provide a single blood sample, along with permission to collect clinical data about their diagnosis and a copy of their clinical EEG.

What are the possible benefits and risks of participating?
There is no direct benefit or risk to the research participants but the results from this study may help other people with epilepsy or brain impairments in the future. Participants may experience discomfort when providing the blood samples.

Where is the study run from?
King’s College London (UK)

When is the study starting and how long is it expected to run for?
July 2015 to September 2026

Who is funding the study?
Canadian Institutes of Health Research (Canada)

Who is the main contact?
1. Professor Deb Pal
2. Sylvine Lalnunhlimi

Study website
http://www.childhood-epilepsy.org

Contact information

Type(s)
Scientific

Contact name
Prof Deb Pal

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Type(s)
Public

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Additional identifiers

EudraCT/CTIS number

IRAS number

199351

ClinicalTrials.gov number
NCT03400371

Secondary identifying numbers
CIHR ID: MOP-142405, IRAS Project ID: 199351

Study information

Scientific Title
Biology of Juvenile Myoclonic Epilepsy

Acronym
BIOJUME

Study objectives
1. JME is associated with variation in GABAA receptor genes
2. JME is associated with molecular networks of ion-channels
3. Endophenotypes of JME will increase power to localise disease-associated genes

Ethics approval required
Old ethics approval format

Ethics approval(s)
Approved 08/12/2016, South Central - Oxford C NHS Research Ethics Committee (Level 3, Block B Lewins Mead, Bristol, BS1 2NT, United Kingdom; +44 (0)20 7104 8049; nrescommittee.southcentral-oxfordc@nhs.net), ref: 16/SC/0266

Study design
Observational cross-sectional study

Primary study design
Observational

Secondary study design
Cross sectional study

Study setting(s)
Hospital

Study type(s)
Screening

Participant information sheet
Not available in web format, please use the contact details to request a patient information sheet

Health condition(s) or problem(s) studied
People with a diagnosis of Juvenile Myoclonic Epilepsy

Interventions
Participation includes one visit for one blood draw per recruited patient. 10-20ml peripheral venous blood is taken from the antecubital fossa. The DNA from the blood sample is then extracted and resequenced for analysis.

Intervention Type
Biological/Vaccine

Phase
Not Applicable

Drug/device/biological/vaccine name(s)
Not provided at time of registration

Primary outcome measure
Association between SNP marker and phenotype is measured using genomewide DNA markers at a single timepoint

Secondary outcome measures
Brain network ictogenicity is measured using quantitative EEG data at a single timepoint

Overall study start date
01/07/2015

Completion date
30/09/2026

Eligibility

Key inclusion criteria
Current inclusion criteria as of 08/05/2025:
1. Diagnosis of Juvenile Myoclonic Epilepsy in accordance with Consensus criteria
2. Age of myoclonus onset 6-25 years
3. Seizures comprising predominant or exclusive early morning myoclonus of upper extremities
4. EEG interictal generalized spikes and/or polyspike and waves with normal background
5. Current age 6-50 years

Previous inclusion criteria:
1. Diagnosis of Juvenile Myoclonic Epilepsy in accordance with Consensus criteria
2. Age of myoclonus onset 10-25 years
3. Seizures comprising predominant or exclusive early morning myoclonus of upper extremities
4. EEG interictal generalized spikes and/or polyspike and waves with normal background
5. Current age 10-40 years

Participant type(s)
Patient

Age group
Mixed

Lower age limit
6 Years

Upper age limit
50 Years

Sex
Both

Target number of participants
2,000

Key exclusion criteria
1. Myoclonus only associated with carbamazepine or lamotrigine therapy
2. EEG showing predominant focal interictal epileptiform discharges or abnormal background
3. Any evidence of progressive or symptomatic myoclonus epilepsy or focal seizures
4. Global learning disability
5. Dysmorphic syndrome
6. Unable to provide informed consent

Date of first enrolment
13/07/2017

Date of final enrolment
30/06/2026

Locations

Countries of recruitment
Canada

Czech Republic

Denmark

England

Estonia

Germany

Italy

Malaysia

Sweden

United Kingdom

United States of America

Wales

Study participating centre
King's College Hospital
London
United Kingdom
SE5 9RS

Study participating centre
College of Medicine
Swansea
United Kingdom
SA2 8PP

Study participating centre
Cardiff University
Cardiff
United Kingdom
CF10 3AT

Study participating centre
Charles University
Prague
Czech Republic
116 36

Study participating centre
Danish National Epilepsy Centre
Dianalund
Denmark
4293

Study participating centre
Tallinn Children's Hospital
Tallinn
Estonia
13419

Study participating centre
Vestre Viken Health Trust, Oslo
Drammen
Norway
3004

Study participating centre
Italian League Against Epilepsy
Rome
Italy
00198

Study participating centre
Hospital for Sick Kids
Toronto
Canada
M5G 1X8

Study participating centre
Nationwide Children's Hospital
Columbus, Ohio
United States of America
43215

Study participating centre
Odense University Hospital
Odense
Denmark
5000

Study participating centre
University of Malaysia
Kuala Lumpur
Malaysia
50603

Study participating centre

Karolinska University Hospital
Stockholm
Sweden
171 64

Study participating centre
Marburg University Hospital
Marburg
Germany
35043

Study participating centre
Sapienza University of Rome
Rome
Italy
00185

Study participating centre
The Newcastle upon Tyne Hospitals NHS Foundation Trust
Freeman Hospital
Freeman Road
High Heaton
Newcastle upon Tyne
United Kingdom
NE7 7DN

Study participating centre
Hasbro Children's Hospital, Brown University
Rhode Island
United States of America
02903

Study participating centre
Airedale NHS Foundation Trust
Airedale General Hospital
Skipton Road
Steeton
Keighley
United Kingdom
BD20 6TD

Study participating centre
Bradford Teaching Hospitals NHS Foundation Trust
Bradford Royal Infirmary
Duckworth Lane
Bradford
United Kingdom
BD9 6RJ

Study participating centre
University of Wales and Llandough Hospital NHS Trust
Heath Park
Cardiff
United Kingdom
CF14 4XW

Study participating centre
Darent Valley Hospital
Darenth Wood Road
Dartford
United Kingdom
DA2 8DA

Study participating centre
Guys and St Thomas' NHS Foundation Trust
London
United Kingdom
SE1 7EH

Study participating centre
Leeds Teaching Hospitals NHS Trust
St. James's University Hospital
Beckett Street
Leeds
United Kingdom
LS9 7TF

Study participating centre
Queen Elizabeth University Hospital
1345 Govan Road

Glasgow
United Kingdom
G51 4TF

Sponsor information

Organisation
King’s College London

Sponsor details
Director of Research Management and Innovation
Room 1.1 Hodgkin Building
London
England
United Kingdom
SE1 1UL

Sponsor type
University/education

Website
https://www.kcl.ac.uk/index.aspx

Organisation
King's College Hospital NHS Trust

Sponsor details
Denmark Hill
Brixton
London
England
United Kingdom
SE5 9RS

Sponsor type
Hospital/treatment centre

Website
https://www.kch.nhs.uk/

Funder(s)

Funder type

Government

Funder Name
Canadian Institutes of Health Research

Alternative Name(s)
Instituts de Recherche en Santé du Canada, Canadian Institutes of Health Research (CIHR), CIHR_IRSC, Canadian Institutes of Health Research | Ottawa ON, CIHR, IRSC

Funding Body Type
Government organisation

Funding Body Subtype
National government

Location
Canada

Results and Publications

Publication and dissemination plan
Planned publication in a high-impact peer reviewed journal.

Intention to publish date
30/09/2027

Individual participant data (IPD) sharing plan
Initially, study data will be used for project as detailed and then made available to a limited number of collaborators for other research projects. After this, the data is then expected to be put into a publically available repository or available upon request.

IPD sharing plan summary
Data sharing statement to be made available at a later date

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
HRA research summary			28/06/2023	No	No