ISRCTN35040926 https://doi.org/10.1186/ISRCTN35040926

Neurology and enzyme therapy in MODY8

Submission date	Recruitment status	[_] Prospectively registered
21/11/2007	No longer recruiting	[_] Protocol
Registration date	**Overall study status**	[_] Statistical analysis plan
04/12/2007	Completed	[X] Results
Last Edited	**Condition category**	[_] Individual participant data
10/06/2021	Nutritional, Metabolic, Endocrine	

Plain English summary of protocol
Not provided at time of registration

Contact information

Type(s)
Scientific

Contact name
Prof Pal Njolstad

Contact details
Section for Paediatrics
Department of Clinical Medicine
Haukeland University Hospital
Bergen
Norway
5021
+47 55 97 51 53
pal.njolstad@pedi.uib.no

Additional identifiers

EudraCT/CTIS number

IRAS number

ClinicalTrials.gov number

Secondary identifying numbers
N/A

Study information

Scientific Title
Neurology and enzyme therapy in MODY8

Study objectives
The MODY8 syndrome is a monogenically inherited syndrome of diabetes and pancreatic exocrine dysfunction due to single-base deletion mutations in the Carboxyl-Ester Lipase (CEL) gene Variable Number of Tandem Repeats (VNTR), registered in OMIM as MODY8 or DPED.

Hypotheses:
That pancreatic enzyme substitution therapy will:
1. Ameliorate exocrine function as reflected by fecal fat excretion and fat-soluble vitamin status
2. Improve glycemic control as measured by HbA1c
3. Improve neuropathology in patients with the MODY8 syndrome

Ethics approval required
Old ethics approval format

Ethics approval(s)
The Regional Committee for Research Ethics of Western Norway (REK Vest), approved on 11 November 2004, (ref: REK Vest 209.04)

Study design
Open, non-randomized, single-center, interventional study.

Primary study design
Interventional

Secondary study design
Non randomised controlled trial

Study setting(s)
Not specified

Study type(s)
Treatment

Participant information sheet

Health condition(s) or problem(s) studied
MODY8 syndrome

Interventions
All participants initially received one Creon enterocapsule (Solvay Pharmaceuticals, Germany) containing 10,000 units lipase, 18,000 units amylase, 600 units protease three times daily, orally at meals. If the clinical effect was unsatisfactory based on patient symptoms, the dose was first increased to 1-2 capsules 3-4 times daily, and if the clinical effect was still unsatisfactory, the medication was changed to Creon Forte, taken orally with meals, 1-2 capsules per meal.

Intervention Type
Drug

Phase
Not Specified

Drug/device/biological/vaccine name(s)
Creon enterocapsule and Creon Forte,Creon enterocapsule and Creon Forte

Primary outcome measure
1. Fecal elastase-1, assessed at 0, 6, 12 and 30 months
2. Fecal fat excretion, assessed at 0, 12 and 30 months
3. HbA1c (blood test), assessed at 0, 6, 12 and 30 months
4. Vitamins A, D and E (blood test), assessed at 0, 6, 12 and 30 months
5. Creatinine (blood test), assessed at 0, 6, 12 and 30 months
6. Total calcium (blood test), assessed at 0, 6, 12 and 30 months
7. Total High Density Lipoprotein (HDL) and Low Density Lipoprotein (LDL) cholesterols (blood test), assessed at 0, 6, 12 and 30 months
8. Triglycerides (blood test), assessed at 0, 6, 12 and 30 months
9. C-peptide (blood test), assessed at 0, 6, 12 and 30 months
10. Bone mass density (age-matched Z-scores), assessed at 0, 12 and 30 months
11. Visual evoked potential, assessed at 0 and 18 months
12. Sensory evoked potential, assessed at 0 and 18 months
13. Nerve conduction velocity, assessed at 0 and 18 months

Secondary outcome measures
No secondary outcome measures

Overall study start date
01/09/2004

Completion date
30/06/2007

Eligibility

Key inclusion criteria
Participants should:
1. Be a carrier of a single-nucleotide deletion mutation in the CEL VNTR
2. Have diabetes by the World Health Organization criteria
3. Have exocrine dysfunction defined by fecal elastase <200 micrograms/ml in two consecutive tests
4. Patients of both sexes and all ages should be included

Participant type(s)
Patient

Age group
Not Specified

Sex
Both

Target number of participants
16

Total final enrolment
9

Key exclusion criteria
1. Ongoing treatment with pancreatic enzyme supplements
2. Inability to attend clinical examinations and other necessary investigations for geographical reasons
3. Side effects of medication (strong stomach ache)

Date of first enrolment
01/09/2004

Date of final enrolment
30/06/2007

Locations

Countries of recruitment
Norway

Study participating centre
Section for Paediatrics
Bergen
Norway
5021

Sponsor information

Organisation
University of Bergen (Norway)

Sponsor details
Faculty of Medicine
Post Box 7804
Bergen
Norway
5020
+47 55 58 20 86
post@medfa.uib.no

Sponsor type
University/education

Website
http://www.uib.no/info/english/

ROR
https://ror.org/03zga2b32

Funder(s)

Funder type
University/education

Funder Name
Haukeland University Hospital, Innovest, University of Bergen (Norway)

Funder Name
The Norwegian Research Council (FUGE Program)

Results and Publications

Publication and dissemination plan
Not provided at time of registration

Intention to publish date

Individual participant data (IPD) sharing plan

IPD sharing plan summary
Not provided at time of registration

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Results article		01/09/2008	10/06/2021	Yes	No