The role of microbiome in urea cycle defect disorders

No registrations found.

Ethical review	Not applicable
Status	Pending
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON25258

Source
NTR

Brief title
UCD

Health condition

UCD; Urea cycle defect; ureumcyclusdefect; PKU; Pheylketonuria; fenylketonurie; microbiome; microbioom; gut bacteria; darmbacterien

Sponsors and support

Primary sponsor: AMC
Source(s) of monetary or material Support: ESN (erfelijke stofwisselingsziekten Nederlands taalgebied).

Intervention

Outcome measures

Primary outcome

differences in the abundance of bacterial strains between healthy volunteers and UCD patients and between PKU and UCD patients

Secondary outcome

differences in the abundance of ammonia producing bacterial strains (e.g. Clostridium species) between healthy volunteers and UCD patients and between PKU and UCD patients

Study description

Background summary

Urea cycle disorders (UCDs) are a group of rare inherited metabolic diseases causing hyperammonemic encephalopathy. Despite intensive dietary and pharmacological therapy, outcome is poor in subset of UCD patients. Because of very low protein tolerance, UCD patients follow a strict low protein diet. Both the composition of the diet, as well as the amino acid supplementation these patients use could change gut microbial composition, with a potential negative effect on metabolic control.

Hypothesis:

In urea cycle defect patients the protein-restricted diet, in combination with essential amino acid supplementation, results in a different gut microbial composition compared to healthy individuals on a normal diet. Selection of ammonia producing bacteria can negatively influence metabolic regulation in UCD patients.

Objective:

To detect differences between microbiome composition of UCD patients, healthy controls and phenylketonuria (PKU) patients. PKU patients are included to differentiate between the effect of a low protein diet and the UCD itself on microbial composition)

Study objective

In urea cycle defect patients the protein-restricted diet, in combination with essential amino acid supplementation, results in a different gut microbial composition compared to healthy individuals on a normal diet. Selection of ammonia producing bacteria can negatively influence metabolic regulation in UCD patients

Study design

single measurement

Intervention

one day food diary

and
a one time stool (feces) sample

Contacts

Public
C. Timmer
Amsterdam
The Netherlands
Scientific
C. Timmer
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Eligibility criteria

Inclusion criteria

Patients:

- age 18 years and over.

- confirmed diagnosis of a urea cycle defect or phenylketonuria (enzymatic or mutation analysis and amino acid spectrum)

-treatment with a low protein diet (natural protein intake 0.8 gram/kg/day or lower) and the use of amino acid supplements

Healthy subjects:

-age 18 and over.

Exclusion criteria

Patients:

-known non-adherence to the diet

-known incompliance with taking the amino acid supplements

-use of antibiotics three months prior to sample collection

-other severe conditions that may influence gut microbiome composition (e.g. liver cirrhosis, renal failure, inflammatory bowel disease)

Healthy subjects:

-illnesses known to influence microbiome composition (e.g. liver disease, bowel disease, diarrhea)

-medication use known to influence microbiome

Study design

Design

Study type:	Observational non invasive
Intervention model:	Parallel
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-03-2017
Enrollment:	45
Type:	Anticipated

Ethics review

Not applicable

Application type:	Not applicable

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
NTR-new	NL6106
NTR-old	NTR6447
Other	NL61031 : ABR nummer

Study results